# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Associated Therapy |
|---|---|---|---|---|---|---|
| EGFR | EGFR exon 20 insertion | p.H773dup | c.2319_2320insCAC | POSITIVE | COSM12377 | ZEGFROVY™ (sunvozertinib) |

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

None Detected

### Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

### SNVs/Indels

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| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (1**4**4/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

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# Results: No Calls

### SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

### CNVs

None Detected

# Results: Negative Calls

### SNVs/Indels

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6937267 | NEGATIVE | del |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |

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|---|---|---|---|---|
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |

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|---|---|---|---|---|
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |

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|---|---|---|---|---|
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1delinsTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( | p.? | OMINDEL689 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2del CCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_472delGC AGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2_465-1del insTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1_598delin sTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 c.727_734+9delGT CACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.850_862+4delGACCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.465-2_465-1delAG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.465-4_479delGC AGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.465-1_465delin sTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.375-2_386delAC GTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| EGFR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

## QC Metrics

### Sample QC - RNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| Mapped Fusion Reads | 35289 | >=6000 | Passed |
| RNA Expression Ctrls Detected | 7 | >=5 | Passed |

### Sample QC - DNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| MAPD | 0.28 | [0-0.5] | Passed |
| Mapped Reads | 524728 | >=100000 | Passed |
| Mean AQ20 Read Length (bp) | 84 | >=64 | Passed |

## Sequencing Run Details

Assay Name: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Panel: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Started On: **15 FEB 2022 18:38**
Completed On: **30 APR 2025 04:44**
Operator:
Instrument: **ValkyrieTestRig**
Serial Number: **Val123**
Filter Chain:
Gene List:

## Reagents

| Consumable | Part # | Expiration Date | Lot # |
|---|---|---|---|
| PurificationKit | N/A | N/A | N/A |
| ChipKit | N/A | N/A | N/A |
| PanelKit | N/A | N/A | N/A |
| LibraryKit | N/A | N/A | N/A |
| BarcodeKit | N/A | N/A | N/A |
| TemplatingKit | N/A | N/A | N/A |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Kit Barcodes

| Sample Name | Assay Name | Nucleic Acid Type | Extraction Kit | Control Kit |
|---|---|---|---|---|
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | DNA | 100101850 | - |
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | RNA | 100101851 | - |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 37 |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 38 |
| NTC | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 39 |
| NTC | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 39 |

180 Oyster Point blvd
South San Francisco

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Associated Therapy |
|---|---|---|---|---|---|---|
| EGFR | EGFR exon 20 insertion | p.H773dup | c.2319_2320insCAC | POSITIVE | COSM12377 | ZEGFROVY™ (sunvozertinib) |

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| MET | MET exon 14 skipping (DNA) | p.? | c.3072_3082+7delTTTTCCAGAAGGTATATT | POSITIVE | COSM6937267 | del |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | POSITIVE |

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

# Cancer Mutations with Potential Clinical Significance (Level 3)

## SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| EGFR amplification | POSITIVE |

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

**Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0**

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Results: No Calls

## SNVs/Indels

None Detected

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

# Results: Negative Calls

## SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1delinsTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET c.2942-29_2942-28d elinsGG | p.? | OM3305 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( | p.? | OM3188 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( | p.? | COSM6057022 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2delCCCCA | p.? | OMINDEL1005 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_472delGCAGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2_465-1delinsTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGGCACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1_598delinsTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 c.727_734+9delGTCACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.850_862+4delGACCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-2_465-1delAG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.731_734+9delCCCTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 c.465-1_465delinsTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.375-2_386delACGTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# QC Metrics

### Sample QC - RNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| Mapped Fusion Reads | 35289 | >=6000 | Passed |
| RNA Expression Ctrls Detected | 7 | >=5 | Passed |

### Sample QC - DNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| MAPD | 0.28 | [0-0.5] | Passed |
| Mapped Reads | 524728 | >=100000 | Passed |
| Mean AQ20 Read Length (bp) | 84 | >=64 | Passed |

# Sequencing Run Details

Assay Name: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Panel: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Started On: **15 FEB 2022 18:38**
Completed On: **30 APR 2025 04:44**
Operator:
Instrument: **ValkyrieTestRig**
Serial Number: **Val123**
Filter Chain:
Gene List:

# Reagents

| Consumable | Part # | Expiration Date | Lot # |
|---|---|---|---|
| PurificationKit | N/A | N/A | N/A |
| ChipKit | N/A | N/A | N/A |
| PanelKit | N/A | N/A | N/A |
| LibraryKit | N/A | N/A | N/A |
| BarcodeKit | N/A | N/A | N/A |
| TemplatingKit | N/A | N/A | N/A |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Kit Barcodes

| Sample Name | Assay Name | Nucleic Acid Type | Extraction Kit | Control Kit |
|---|---|---|---|---|
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | DNA | 100101850 | - |
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | RNA | 100101851 | - |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 37 |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 38 |
| NTC | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 39 |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

None Detected

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

None Detected

### Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Results: No Calls

### SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

### CNVs

None Detected

# Results: Negative Calls

### SNVs/Indels

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773dup | COSM12377 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6937267 | NEGATIVE | del |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |

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|---|---|---|---|---|
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |

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|---|---|---|---|---|
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |

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|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |

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|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |

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|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |

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|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |

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|---|---|---|---|---|
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |

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|---|---|---|---|---|
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1delinsTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.598-1_598delinsTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-6_375-2delCCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-4_472delGCAGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.465-1_465delinsTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.850_862+4delGACCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-2_465-1delAG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 c.465-2_465-1delinsTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.727_734+9delGTCACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.375-2_386delAC GTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| EGFR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

## QC Metrics

### Sample QC - RNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| Mapped Fusion Reads | 35289 | >=6000 | Passed |
| RNA Expression Ctrls Detected | 7 | >=5 | Passed |

### Sample QC - DNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| MAPD | 0.28 | [0-0.5] | Passed |
| Mapped Reads | 524728 | >=100000 | Passed |
| Mean AQ20 Read Length (bp) | 84 | >=64 | Passed |

## Sequencing Run Details

Assay Name: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Panel: **Oncomine™ Dx Express Test - FFPE - RS v5.0.0**
Started On: **15 FEB 2022 18:38**
Completed On: **30 APR 2025 04:44**
Operator:
Instrument: **ValkyrieTestRig**
Serial Number: **Val123**
Filter Chain:
Gene List:

## Reagents

| Consumable | Part # | Expiration Date | Lot # |
|---|---|---|---|
| PurificationKit | N/A | N/A | N/A |
| ChipKit | N/A | N/A | N/A |
| PanelKit | N/A | N/A | N/A |
| LibraryKit | N/A | N/A | N/A |
| BarcodeKit | N/A | N/A | N/A |
| TemplatingKit | N/A | N/A | N/A |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Kit Barcodes

| Sample Name | Assay Name | Nucleic Acid Type | Extraction Kit | Control Kit |
|---|---|---|---|---|
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | DNA | 100101850 | - |
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | RNA | 100101851 | - |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 37 |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 38 |
| NTC | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 39 |
| NTC | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 39 |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Associated Therapy |
|---|---|---|---|---|---|---|
| EGFR | EGFR exon 20 insertion | p.H773dup | c.2319_2320insCAC | POSITIVE | COSM12377 | ZEGFROVY™ (sunvozertinib) |

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

None Detected

### Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

### SNVs/Indels

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

**Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0**

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Results: No Calls

## SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

# Results: Negative Calls

## SNVs/Indels

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6937267 | NEGATIVE | del |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1del insTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( | p.? | OMINDEL689 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2del CCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_472delGC AGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2_465-1del insTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1_598delin sTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 c.727_734+9delGT CACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.850_862+4delGACCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.465-2_465-1delAG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.465-4_479delGC AGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.465-1_465delin sTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.375-2_386delAC GTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| EGFR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

None Detected

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

None Detected

### Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

**Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0**

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Results: No Calls

## SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

# Results: Negative Calls

## SNVs/Indels

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773dup | COSM12377 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6937267 | NEGATIVE | del |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1del insTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.598-1_598delinsTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-6_375-2delCCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-4_472delGCAGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.465-1_465delinsTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.850_862+4delGACCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-2_465-1delAG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 c.465-2_465-1delinsTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.727_734+9delGTCACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.375-2_386delAC GTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| EGFR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

None Detected

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| MET | MET exon 14 skipping (DNA) | p.? | c.3072_3082+7delTTTTCCAGAAGGTATATT | POSITIVE | COSM6937267 | del |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | POSITIVE |

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

## SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| EGFR amplification | POSITIVE |

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

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# Results: No Calls

### SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

### CNVs

None Detected

# Results: Negative Calls

### SNVs/Indels

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773dup | COSM12377 | NEGATIVE | ins |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertio | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| n | | | | |
| ERBB2 exon 20 insertio n | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertio n | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertio | p.G778dup | COSM26681 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| n | | | | |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1del insTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2del CCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-4_479delGC AGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-2_465-1del AG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2_465-1del insTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.598-1_598delin sTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.727_734+9delGT CACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.375-2_386delACGTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.465-1_465delin | p.? | COSM6928792 | NEGATIVE | mnp |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| sTT | | | | |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.850_862+4delGA CCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.465-4_472delGC AGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |

# Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# QC Metrics

### Sample QC - RNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| Mapped Fusion Reads | 35289 | >=6000 | Passed |
| RNA Expression Ctrls Detected | 7 | >=5 | Passed |

### Sample QC - DNA

| Metric Name | Value | Reference Range | QC Status |
|---|---|---|---|
| MAPD | 0.28 | [0-0.5] | Passed |
| Mapped Reads | 524728 | >=100000 | Passed |
| Mean AQ20 Read Length (bp) | 84 | >=64 | Passed |

# Sequencing Run Details

| | | | |
|---|---|---|---|
| Assay Name: | **Oncomine™ Dx Express Test - FFPE - RS v5.0.0** | Panel: | **Oncomine™ Dx Express Test - FFPE - RS v5.0.0** |
| Started On: | **15 FEB 2022 18:38** | Completed On: | **30 APR 2025 04:44** |
| Operator: | | Instrument: | **ValkyrieTestRig** |
| Serial Number: | **Val123** | Filter Chain: | |
| Gene List: | | | |

# Reagents

| Consumable | Part # | Expiration Date | Lot # |
|---|---|---|---|
| PurificationKit | N/A | N/A | N/A |
| ChipKit | N/A | N/A | N/A |
| PanelKit | N/A | N/A | N/A |
| LibraryKit | N/A | N/A | N/A |
| BarcodeKit | N/A | N/A | N/A |
| TemplatingKit | N/A | N/A | N/A |

## Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Kit Barcodes

| Sample Name | Assay Name | Nucleic Acid Type | Extraction Kit | Control Kit |
|---|---|---|---|---|
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | DNA | 100101850 | - |
| FFPE01_D7_420 | ODxET FFPE RS v5.0.0 | RNA | 100101851 | - |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 37 |
| PositiveControl01 | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 38 |
| NTC | ODxET FFPE RS v5.0.0 | DNA | - | 01101903020165751724 0601101234567240A513 39 |
| NTC | ODxET FFPE RS v5.0.0 | RNA | - | 01101903020165751724 0601101234567240A513 39 |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

None Detected

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| MET | MET exon 14 skipping (DNA) | p.? | c.3072_3082+7delTTTTCCAGAAGGTATATT | POSITIVE | COSM6937267 | del |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | POSITIVE |

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

## Cancer Mutations with Potential Clinical Significance (Level 3)

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

## SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| EGFR amplification | POSITIVE |

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

**Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0**

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

# Results: No Calls

## SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 V592M | p.V592M | COSM6840954 | NO CALL | SNV |
| PIK3CA R108C | p.R108C | COSM6438095 | NO CALL | SNV |
| RET R912W | p.R912W | COSM3415038 | NO CALL | SNV |
| STK11 D194H | p.D194H | COSM5325765 | NO CALL | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

# Results: Negative Calls

## SNVs/Indels

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773dup | COSM12377 | NEGATIVE | ins |
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.A775_G776insV | MAN307 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertio n | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertio | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| n | | | | |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertio | p.G778dup | COSM26681 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| n | | | | |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1del insTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET c.2942-29_2942-28delinsGG | p.? | OM3305 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057022 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3188 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2del CCCCA | p.? | OMINDEL1005 | NEGATIVE | del |
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-4_479delGC AGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-2_465-1del AG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 c.465-2_465-1del insTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.598-1_598delin sTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 c.727_734+9delGT CACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delCAGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.375-2_386delACGTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| STK11 c.598-2_600delAGGCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10delTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.465-1_465delin | p.? | COSM6928792 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| sTT | | | | |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.850_862+4delGA CCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.731_734+9delCC CTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.465-4_472delGC AGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Sample | | Patient | |
|---|---|---|---|
| Sample Name: | **FFPE01_D7_420** | Patient Id: | **PID01** |
| Cancer Type: | **Non-Small Cell Lung Cancer** | Gender: | **Male** |
| Cancer Stage: | **Stage 0** | Date of Birth: | **30 MAR 1975** |
| Sample Type: | **FFPE** | Collection Date: | **22 FEB 2021** |
| %Cellularity: | **100** | Ordering Physician: | **Dr. Lee** |
| %Necrosis: | | Created On: | **02 APR 2025 21:29** |

## Companion Diagnostics (CDx) Biomarkers

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Associated Therapy |
|---|---|---|---|---|---|---|
| EGFR | EGFR exon 20 insertion | p.H773dup | c.2319_2320insCAC | POSITIVE | COSM12377 | ZEGFROVY™ (sunvozertinib) |

## Tumor Mutation Profiling Results: Summary

The results below provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms.

Results reported in this section are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Cancer Mutations with Evidence of Clinical Significance (Level 2)

### SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| MET | MET exon 14 skipping (DNA) | p.? | c.3072_3082+7delTTTTCCAGAAGGTATATT | POSITIVE | COSM6937267 | del |

### Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CCDC6-RET fusion | CCDC6(1) - RET(12) | Fusion | POSITIVE |

ⓘ Fusions include gene fusions and splice variants.

### CNVs

None Detected

# Cancer Mutations with Potential Clinical Significance (Level 3)

## SNVs/Indels

| Gene | Variant Name | AA Change | Nuc Change | Test Result | Variant ID | Variant Type |
|---|---|---|---|---|---|---|
| ALK | ALK R1231Q | p.R1231Q | c.3692G>A | POSITIVE | COSM5967491 | SNV |
| IDH2 | IDH2 R140W | p.R140W | c.418C>T | POSITIVE | COSM41877 | SNV |
| NTRK1 | NTRK1 G595R | p.G595R | c.1783G>A | POSITIVE | BT109 | SNV |
| TP53 | TP53 C275F | p.C275F | c.824G>T | POSITIVE | COSM10701 | SNV |

## Fusions

None Detected

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| EGFR amplification | POSITIVE |

## Intended Use

The Oncomine™ Dx Express Test is a qualitative *in vitro* diagnostic test that uses targeted next- generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as a companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1. Companion Diagnostic Indications**

| Tissue Type | Gene | Variant | Targeted Therapy |
|---|---|---|---|
| NSCLC | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

## Contraindication

There are no known contraindications.

## Assay Warnings and Limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.

2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.

3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.

4. Genomic findings listed in the Oncomine Dx Express Test results report under Cancer Mutations with Evidence of Clinical Significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.

5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.

6. The Oncomine Dx Express Test is for use with the Genexus Dx Integrated Sequencer and the Genexus Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.

7. The Oncomine Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.

8. The Oncomine Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.

9. The Oncomine Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

10. Although the Genexus Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.

12. The Oncomine Dx Express Test is for use with FFPE tumor specimens.

13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine Dx Express Test.

14. The effects of potential variations in FFPE specimen fixation have not been evaluated.

15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.

16. The Oncomine Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.

17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.

18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.

19. The performance of the Oncomine Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.

20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient’s tumor and should be followed by additional confirmatory testing.

21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.

22. The minimum tumor content thresholds for Oncomine Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.

23. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is less than 20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.

24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20-39%. Additional confirmatory testing with a clinically validated test is strongly recommended.

25. The presence of more than 70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.

26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.

27. The accuracy of the Oncomine Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.

28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).

29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

## Performance Characteristics

Please refer to the product label: www.thermofisher.com/ODxETDocs

## Test Description

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

| Alteration Type | Genes |
|---|---|
| Deletions, insertions, substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants (CNVs) from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Gene fusions and splice variants from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

The test output classifies variants into the following levels according to the cancer indication:

Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use
Level 2: Cancer Mutations with Evidence of Clinical Significance
Level 3: Cancer Mutations with Potential Clinical Significance

# Results: No Calls

## SNVs/Indels

None Detected

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ALK imbalance | ALK | RNAExonTiles | NO CALL |
| FGFR1 imbalance | FGFR1 | RNAExonTiles | NO CALL |
| FGFR2 imbalance | FGFR2 | RNAExonTiles | NO CALL |
| FGFR3 imbalance | FGFR3 | RNAExonTiles | NO CALL |
| NTRK1 imbalance | NTRK1 | RNAExonTiles | NO CALL |
| NTRK2 imbalance | NTRK2 | RNAExonTiles | NO CALL |
| NTRK3 imbalance | NTRK3 | RNAExonTiles | NO CALL |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

None Detected

# Results: Negative Calls

## SNVs/Indels

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AKT1 Q79K | p.Q79K | COSM159008 | NEGATIVE | SNV |
| AKT1 L52R | p.L52R | COSM93893 | NEGATIVE | SNV |
| AKT1 R76_C77insWPNTFIIR | p.R76_C77insWPNTFIIR | COSM5045024 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AKT1 P70_C77dup | p.P70_C77dup | COSM6568358 | NEGATIVE | ins |
| AKT1 P51L | p.P51L | COSM4468165 | NEGATIVE | SNV |
| AKT1 E17K | p.E17K | COSM33765 | NEGATIVE | SNV |
| AKT1 P51S | p.P51S | COSM5991558 | NEGATIVE | SNV |
| AKT1 C77F | p.C77F | COSM5027916 | NEGATIVE | SNV |
| AKT2 D324H | p.D324H | COSM474742 | NEGATIVE | SNV |
| AKT2 E17K | p.E17K | COSM159009 | NEGATIVE | SNV |
| AKT2 D324G | p.D324G | COSM474741 | NEGATIVE | SNV |
| AKT3 E17K | p.E17K | COSM224779 | NEGATIVE | SNV |
| AKT3 Q78K | p.Q78K | COSM242892 | NEGATIVE | SNV |
| AKT3 L51R | p.L51R | COSM309035 | NEGATIVE | SNV |
| ALK R1275Q | p.R1275Q | COSM28056 | NEGATIVE | SNV |
| ALK G1269A | p.G1269A | COSM1169707 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28062 | NEGATIVE | SNV |
| ALK F1245I | p.F1245I | COSM28492 | NEGATIVE | SNV |
| ALK R1209Q | p.R1209Q | COSM327056 | NEGATIVE | SNV |
| ALK S1206Y | p.S1206Y | COSM144251 | NEGATIVE | SNV |
| ALK G1202R | p.G1202R | COSM144250 | NEGATIVE | SNV |
| ALK L1198V | p.L1198V | OM3314 | NEGATIVE | SNV |
| ALK L1196Q | p.L1196Q | COSM1169447 | NEGATIVE | SNV |
| ALK R1192P | p.R1192P | OM3259 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28055 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK F1174L | p.F1174L | COSM28057 | NEGATIVE | SNV |
| ALK I1171S | p.I1171S | COSM4440758 | NEGATIVE | SNV |
| ALK C1156F | p.C1156F | BT144 | NEGATIVE | SNV |
| ALK L1152R | p.L1152R | COSM97185 | NEGATIVE | SNV |
| ALK T1151M | p.T1151M | COSM98478 | NEGATIVE | SNV |
| ALK E1129V | p.E1129V | BT117 | NEGATIVE | SNV |
| ALK G1123D | p.G1123D | OM3262 | NEGATIVE | SNV |
| ALK R1275L | p.R1275L | COSM28060 | NEGATIVE | SNV |
| ALK F1245C | p.F1245C | COSM28500 | NEGATIVE | SNV |
| ALK E1210K | p.E1210K | OM3161 | NEGATIVE | SNV |
| ALK D1203N | p.D1203N | COSM6191344 | NEGATIVE | SNV |
| ALK L1198F | p.L1198F | COSM97028 | NEGATIVE | SNV |
| ALK V1180L | p.V1180L | COSM4381101 | NEGATIVE | SNV |
| ALK F1174V | p.F1174V | COSM28054 | NEGATIVE | SNV |
| ALK I1171T | p.I1171T | COSM4381100 | NEGATIVE | SNV |
| ALK C1156Y | p.C1156Y | COSM99136 | NEGATIVE | SNV |
| ALK T1151dup | p.T1151dup | COSM144252 | NEGATIVE | ins |
| ALK T1151K | p.T1151K | OM3261 | NEGATIVE | SNV |
| ALK A1280V | p.A1280V | COSM6914238 | NEGATIVE | SNV |
| ALK F1245V | p.F1245V | COSM28499 | NEGATIVE | SNV |
| ALK S1206F | p.S1206F | COSM6909200 | NEGATIVE | SNV |
| ALK S1206C | p.S1206C | BT147 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK G1202del | p.G1202del | OMINDEL698 | NEGATIVE | del |
| ALK A1200V | p.A1200V | COSM317003 | NEGATIVE | SNV |
| ALK R1192Q | p.R1192Q | OM3372 | NEGATIVE | SNV |
| ALK F1174L | p.F1174L | COSM28061 | NEGATIVE | SNV |
| ALK F1174S | p.F1174S | COSM53063 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM28498 | NEGATIVE | SNV |
| ALK I1171H | p.I1171H | OM3163 | NEGATIVE | mnp |
| ALK C1156T | p.C1156T | OM3350 | NEGATIVE | mnp |
| ALK L1152V | p.L1152V | OM3513 | NEGATIVE | SNV |
| ALK G1123S | p.G1123S | COSM5045108 | NEGATIVE | SNV |
| ALK F1174I | p.F1174I | COSM28491 | NEGATIVE | SNV |
| ALK F1245L | p.F1245L | COSM28493 | NEGATIVE | SNV |
| ALK D1203A | p.D1203A | OM3313 | NEGATIVE | SNV |
| ALK I1171N | p.I1171N | COSM1169448 | NEGATIVE | mnp |
| ALK S1157F | p.S1157F | BT145 | NEGATIVE | SNV |
| ALK L1152P | p.L1152P | COSM1407659 | NEGATIVE | SNV |
| ALK T1151R | p.T1151R | OM3260 | NEGATIVE | SNV |
| ALK R1231W | p.R1231W | BT148 | NEGATIVE | SNV |
| ALK F1174C | p.F1174C | COSM28059 | NEGATIVE | SNV |
| ALK L1198P | p.L1198P | OM3162 | NEGATIVE | SNV |
| ALK Y1278S | p.Y1278S | COSM28058 | NEGATIVE | SNV |
| ALK G1128A | p.G1128A | COSM98475 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ALK I1268V | p.I1268V | BT149 | NEGATIVE | SNV |
| ALK L1196M | p.L1196M | COSM99137 | NEGATIVE | SNV |
| ALK E1154Q | p.E1154Q | OM3324 | NEGATIVE | SNV |
| AR W742L | p.W742L | COSM5944172 | NEGATIVE | SNV |
| AR H875Y | p.H875Y | COSM238555 | NEGATIVE | SNV |
| AR T878A | p.T878A | COSM236693 | NEGATIVE | SNV |
| AR L882I | p.L882I | COSM5945751 | NEGATIVE | SNV |
| AR E894K | p.E894K | COSM5945748 | NEGATIVE | SNV |
| AR V716M | p.V716M | COSM3094417 | NEGATIVE | SNV |
| AR F814V | p.F814V | COSM150793 | NEGATIVE | SNV |
| AR T878S | p.T878S | COSM5570418 | NEGATIVE | SNV |
| AR M896V | p.M896V | COSM5945749 | NEGATIVE | SNV |
| AR M896T | p.M896T | OM3321 | NEGATIVE | SNV |
| AR L702H | p.L702H | COSM238553 | NEGATIVE | SNV |
| AR V731M | p.V731M | COSM1468984 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5572266 | NEGATIVE | SNV |
| AR D891H | p.D891H | COSM5945767 | NEGATIVE | SNV |
| AR H875Q | p.H875Q | COSM5945766 | NEGATIVE | SNV |
| AR W742C | p.W742C | COSM5944171 | NEGATIVE | SNV |
| AR S889G | p.S889G | COSM5945753 | NEGATIVE | SNV |
| AR D880E | p.D880E | COSM5945750 | NEGATIVE | SNV |
| AR E898G | p.E898G | OM3322 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| AR F877L | p.F877L | COSM5946164 | NEGATIVE | SNV |
| AR F877L | p.F877L | COSM5945752 | NEGATIVE | SNV |
| ARAF S214T | p.S214T | COSM1558277 | NEGATIVE | SNV |
| ARAF S214F | p.S214F | COSM612884 | NEGATIVE | SNV |
| ARAF S214A | p.S214A | COSM1742787 | NEGATIVE | SNV |
| ARAF S214Y | p.S214Y | COSM5416443 | NEGATIVE | SNV |
| ARAF S214C | p.S214C | COSM5044705 | NEGATIVE | SNV |
| BRAF K601R | p.K601R | COSM1568586 | NEGATIVE | SNV |
| BRAF V600D | p.V600D | COSM477 | NEGATIVE | mnp |
| BRAF V600D | p.V600D | COSM308550 | NEGATIVE | mnp |
| BRAF D594_T599dup | p.D594_T599dup | COSM26504 | NEGATIVE | ins |
| BRAF T599_V600insETT | p.T599_V600insETT | COSM4172018 | NEGATIVE | ins |
| BRAF [T599R;V600G] | p.[T599R;V600G] | OM3547 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM474 | NEGATIVE | mnp |
| BRAF T599dup | p.T599dup | COSM144982 | NEGATIVE | ins |
| BRAF A598V | p.A598V | COSM21549 | NEGATIVE | SNV |
| BRAF L597S | p.L597S | COSM1126 | NEGATIVE | mnp |
| BRAF G596V | p.G596V | COSM9145962 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM468 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM253330 | NEGATIVE | SNV |
| BRAF D594A | p.D594A | COSM1583010 | NEGATIVE | SNV |
| BRAF [G593=;D594N] | p.[G593=;D594N] | COSM211600 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF N581T | p.N581T | COSM1699452 | NEGATIVE | SNV |
| BRAF G469L | p.G469L | COSM6056848 | NEGATIVE | mnp |
| BRAF G469E | p.G469E | COSM461 | NEGATIVE | SNV |
| BRAF G466A | p.G466A | COSM452 | NEGATIVE | SNV |
| BRAF G464V | p.G464V | COSM450 | NEGATIVE | SNV |
| BRAF I463S | p.I463S | COSM448 | NEGATIVE | SNV |
| BRAF K601I | p.K601I | COSM26491 | NEGATIVE | SNV |
| BRAF V600E | p.V600E | COSM476 | NEGATIVE | SNV |
| BRAF [T599I;V600K] | p.[T599I;V600K] | COSM1735763 | NEGATIVE | mnp |
| BRAF V600R | p.V600R | COSM1583011 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM33808 | NEGATIVE | SNV |
| BRAF A598T | p.A598T | COSM28505 | NEGATIVE | SNV |
| BRAF L597V | p.L597V | COSM470 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM53198 | NEGATIVE | SNV |
| BRAF D594V | p.D594V | COSM466 | NEGATIVE | SNV |
| BRAF D594N | p.D594N | COSM27639 | NEGATIVE | SNV |
| BRAF Y472C | p.Y472C | COSM1133046 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM458 | NEGATIVE | mnp |
| BRAF G466V | p.G466V | COSM451 | NEGATIVE | SNV |
| BRAF G464E | p.G464E | COSM449 | NEGATIVE | SNV |
| BRAF K601T | p.K601T | COSM3878760 | NEGATIVE | SNV |
| BRAF V600K | p.V600K | COSM473 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF V600E | p.V600E | COSM475 | NEGATIVE | mnp |
| BRAF V600L | p.V600L | COSM219798 | NEGATIVE | SNV |
| BRAF [T599=;V600R] | p.[T599=;V600R] | COSM1127 | NEGATIVE | mnp |
| BRAF V600A | p.V600A | COSM18443 | NEGATIVE | SNV |
| BRAF V600M | p.V600M | COSM1130 | NEGATIVE | SNV |
| BRAF T599I | p.T599I | COSM472 | NEGATIVE | SNV |
| BRAF L597P | p.L597P | COSM1448593 | NEGATIVE | SNV |
| BRAF G596C | p.G596C | COSM6936824 | NEGATIVE | SNV |
| BRAF F595S | p.F595S | COSM1123 | NEGATIVE | SNV |
| BRAF F595L | p.F595L | COSM21612 | NEGATIVE | SNV |
| BRAF D594E | p.D594E | COSM1121 | NEGATIVE | SNV |
| BRAF D594Y | p.D594Y | COSM6928839 | NEGATIVE | SNV |
| BRAF D594H | p.D594H | COSM144576 | NEGATIVE | SNV |
| BRAF N581I | p.N581I | COSM26460 | NEGATIVE | SNV |
| BRAF G469S | p.G469S | COSM27912 | NEGATIVE | mnp |
| BRAF G469S | p.G469S | COSM1113 | NEGATIVE | mnp |
| BRAF G469V | p.G469V | COSM459 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM457 | NEGATIVE | SNV |
| BRAF G469R | p.G469R | COSM455 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM1112 | NEGATIVE | SNV |
| BRAF G466R | p.G466R | COSM253328 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1448615 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| BRAF R462I | p.R462I | COSM447 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM6265 | NEGATIVE | SNV |
| BRAF K601N | p.K601N | COSM1132 | NEGATIVE | SNV |
| BRAF V600_K601delinsE | p.V600_K601delinsE | COSM1133 | NEGATIVE | del |
| BRAF T599_V600insEAT | p.T599_V600insEAT | COSM4166148 | NEGATIVE | ins |
| BRAF V600G | p.V600G | COSM6137 | NEGATIVE | SNV |
| BRAF T599dup | p.T599dup | COSM30730 | NEGATIVE | ins |
| BRAF T599R | p.T599R | COSM4172020 | NEGATIVE | SNV |
| BRAF L597Q | p.L597Q | COSM1125 | NEGATIVE | SNV |
| BRAF N581S | p.N581S | COSM462 | NEGATIVE | SNV |
| BRAF E586K | p.E586K | COSM463 | NEGATIVE | SNV |
| BRAF G466E | p.G466E | COSM453 | NEGATIVE | SNV |
| BRAF G464R | p.G464R | COSM1111 | NEGATIVE | SNV |
| BRAF D594G | p.D594G | COSM467 | NEGATIVE | SNV |
| BRAF G469A | p.G469A | COSM460 | NEGATIVE | SNV |
| BRAF L597R | p.L597R | COSM471 | NEGATIVE | SNV |
| BRAF K601E | p.K601E | COSM478 | NEGATIVE | SNV |
| BRAF G596D | p.G596D | COSM26506 | NEGATIVE | SNV |
| BRAF G596R | p.G596R | COSM469 | NEGATIVE | SNV |
| BRAF V600Q | p.V600Q | COSM249889 | NEGATIVE | mnp |
| CDK4 R24S | p.R24S | COSM3463914 | NEGATIVE | SNV |
| CDK4 R24H | p.R24H | COSM1989836 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CDK4 K22Q | p.K22Q | OM3153 | NEGATIVE | SNV |
| CDK4 R24L | p.R24L | COSM363684 | NEGATIVE | SNV |
| CDK4 R24C | p.R24C | COSM1677139 | NEGATIVE | SNV |
| CDK4 K22R | p.K22R | COSM232013 | NEGATIVE | SNV |
| CDK4 K22M | p.K22M | COSM3463915 | NEGATIVE | SNV |
| CHEK2 R523H | p.R523H | COSM1559596 | NEGATIVE | SNV |
| CHEK2 R523P | p.R523P | COSM1559595 | NEGATIVE | SNV |
| CHEK2 M381V | p.M381V | COSM1559601 | NEGATIVE | SNV |
| CHEK2 [S372=;K373E] | p.[S372=;K373E] | COSM1294063 | NEGATIVE | mnp |
| CTNNB1 D32A | p.D32A | COSM5690 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM22939 | NEGATIVE | SNV |
| CTNNB1 S33L | p.S33L | COSM6098 | NEGATIVE | mnp |
| CTNNB1 S33C | p.S33C | COSM5677 | NEGATIVE | SNV |
| CTNNB1 G34V | p.G34V | COSM5670 | NEGATIVE | SNV |
| CTNNB1 I35K | p.I35K | COSM28727 | NEGATIVE | mnp |
| CTNNB1 I35M | p.I35M | COSM28711 | NEGATIVE | SNV |
| CTNNB1 S37T | p.S37T | COSM5729 | NEGATIVE | SNV |
| CTNNB1 S37C | p.S37C | COSM5679 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5716 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6128 | NEGATIVE | del |
| CTNNB1 S45_G48del | p.S45_G48del | COSM5992 | NEGATIVE | del |
| CTNNB1 S45_S47del | p.S45_S47del | COSM1617582 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 L46_S47del | p.L46_S47del | COSM6129 | NEGATIVE | del |
| CTNNB1 S45A | p.S45A | COSM5685 | NEGATIVE | SNV |
| CTNNB1 L46_S47del | p.L46_S47del | COSM6132 | NEGATIVE | del |
| CTNNB1 K335T | p.K335T | COSM1725761 | NEGATIVE | SNV |
| CTNNB1 D32H | p.D32H | COSM5668 | NEGATIVE | SNV |
| CTNNB1 D32E | p.D32E | COSM5766 | NEGATIVE | SNV |
| CTNNB1 S33A | p.S33A | COSM5683 | NEGATIVE | SNV |
| CTNNB1 G34I | p.G34I | COSM1666841 | NEGATIVE | mnp |
| CTNNB1 I35V | p.I35V | COSM1566774 | NEGATIVE | SNV |
| CTNNB1 H36Y | p.H36Y | COSM5703 | NEGATIVE | SNV |
| CTNNB1 S37A | p.S37A | COSM5675 | NEGATIVE | SNV |
| CTNNB1 T41A | p.T41A | COSM5664 | NEGATIVE | SNV |
| CTNNB1 P44S | p.P44S | COSM5704 | NEGATIVE | SNV |
| CTNNB1 [S45E;L46=] | p.[S45E;L46=] | COSM86011 | NEGATIVE | mnp |
| CTNNB1 S45Y | p.S45Y | COSM5692 | NEGATIVE | SNV |
| CTNNB1 S45del | p.S45del | COSM6131 | NEGATIVE | del |
| CTNNB1 K335R | p.K335R | COSM5945941 | NEGATIVE | SNV |
| CTNNB1 D32Y | p.D32Y | COSM5661 | NEGATIVE | SNV |
| CTNNB1 D32G | p.D32G | COSM5681 | NEGATIVE | SNV |
| CTNNB1 S33N | p.S33N | COSM6099 | NEGATIVE | mnp |
| CTNNB1 S33Y | p.S33Y | COSM5673 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5684 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 G34A | p.G34A | COSM5988492 | NEGATIVE | SNV |
| CTNNB1 I35N | p.I35N | COSM5695 | NEGATIVE | SNV |
| CTNNB1 I35T | p.I35T | COSM13168 | NEGATIVE | SNV |
| CTNNB1 H36P | p.H36P | COSM5678 | NEGATIVE | SNV |
| CTNNB1 S37P | p.S37P | COSM5687 | NEGATIVE | SNV |
| CTNNB1 S37Y | p.S37Y | COSM5666 | NEGATIVE | SNV |
| CTNNB1 S37F | p.S37F | COSM5662 | NEGATIVE | SNV |
| CTNNB1 T41N | p.T41N | COSM5730 | NEGATIVE | SNV |
| CTNNB1 T41S | p.T41S | COSM5701 | NEGATIVE | SNV |
| CTNNB1 P44L | p.P44L | COSM5761 | NEGATIVE | SNV |
| CTNNB1 S45_S47delinsC | p.S45_S47delinsC | COSM28708 | NEGATIVE | del |
| CTNNB1 S45C | p.S45C | COSM5689 | NEGATIVE | SNV |
| CTNNB1 K335Q | p.K335Q | COSM4421324 | NEGATIVE | SNV |
| CTNNB1 P44A | p.P44A | COSM17661 | NEGATIVE | SNV |
| CTNNB1 D32N | p.D32N | COSM5672 | NEGATIVE | SNV |
| CTNNB1 D32V | p.D32V | COSM5691 | NEGATIVE | SNV |
| CTNNB1 S33P | p.S33P | COSM5682 | NEGATIVE | SNV |
| CTNNB1 H36R | p.H36R | COSM27378 | NEGATIVE | SNV |
| CTNNB1 I35S | p.I35S | COSM5674 | NEGATIVE | SNV |
| CTNNB1 T41P | p.T41P | COSM5688 | NEGATIVE | SNV |
| CTNNB1 T41I | p.T41I | COSM5676 | NEGATIVE | SNV |
| CTNNB1 S45P | p.S45P | COSM5663 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| CTNNB1 S33F | p.S33F | COSM5669 | NEGATIVE | SNV |
| CTNNB1 G34E | p.G34E | COSM5671 | NEGATIVE | SNV |
| CTNNB1 G34R | p.G34R | COSM5686 | NEGATIVE | SNV |
| CTNNB1 S45F | p.S45F | COSM5667 | NEGATIVE | SNV |
| CTNNB1 S45T | p.S45T | COSM5719 | NEGATIVE | SNV |
| CTNNB1 S33T | p.S33T | COSM27311 | NEGATIVE | SNV |
| CTNNB1 K335I | p.K335I | COSM17797 | NEGATIVE | SNV |
| EGFR A289D | p.A289D | COSM21685 | NEGATIVE | SNV |
| EGFR S464L | p.S464L | BT126 | NEGATIVE | SNV |
| EGFR G465R | p.G465R | COSM601581 | NEGATIVE | SNV |
| EGFR K467T | p.K467T | BT132 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236670 | NEGATIVE | SNV |
| EGFR G598V | p.G598V | COSM21690 | NEGATIVE | SNV |
| EGFR E709_T710delinsA | p.E709_T710delinsA | COSM85796 | NEGATIVE | del |
| EGFR E709_T710delinsG | p.E709_T710delinsG | COSM48981 | NEGATIVE | complex |
| EGFR E709A | p.E709A | COSM13427 | NEGATIVE | SNV |
| EGFR L718V | p.L718V | OM3316 | NEGATIVE | SNV |
| EGFR G719S | p.G719S | COSM6252 | NEGATIVE | SNV |
| EGFR S720P | p.S720P | COSM88729 | NEGATIVE | SNV |
| EGFR G724S | p.G724S | COSM13979 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM12423 | NEGATIVE | ins |
| EGFR exon 19 insertion | p.K745_E746insVPVAIK | COSM26444 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 insertion | p.K745_E746insTPVAIK | COSM255152 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.I744_A750delinsVK | COSM85798 | NEGATIVE | complex |
| EGFR I744T | p.I744T | COSM87245 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_A750del | COSM3734668 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_E749del | COSM26038 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746del | COSM18420 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K745_A750delinsT | COSM1190791 | NEGATIVE | del |
| EGFR [K745=;E746I;L747P] | p.[K745=;E746I;L747P] | OM3355 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.E746_T751del | COSM24869 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM13549 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsFPS | COSM6506513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM13550 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM13552 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750del | COSM6225 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752del | COSM133189 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751del | COSM12728 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsQ | COSM22999 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsRP | COSM12413 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM26680 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsIS | COSM133191 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsM | OMINDEL716 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133188 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_T751delinsIP | COSM51526 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsP | COSM6947327 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsL | COSM51502 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754del | COSM24970 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsA | COSM12678 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750delinsAP | COSM144207 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsAPS | COSM6924852 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM6968288 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM18426 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVS | COSM18427 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM133193 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM53205 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsVQ | COSM51524 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM12384 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM52935 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVS | COSM6980200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750del | COSM9179903 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_P753del | COSM255211 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsD | COSM6220 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM12421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM26441 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12422 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM22944 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsD | COSM6974307 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_P753delinsDT | COSM5340693 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM12382 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM1235296 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsAN | COSM85891 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_K754delinsANKG | COSM1667023 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsN | COSM51503 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM51501 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_S752delinsQ | COSM12403 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12420 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsP | COSM12383 | NEGATIVE | complex |
| EGFR L747P | p.L747P | COSM24267 | NEGATIVE | mnp |
| EGFR exon 19 deletion | p.L747_A750delinsS | COSM4170221 | NEGATIVE | del |
| EGFR L747S | p.L747S | COSM26704 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_P753delinsS | COSM12370 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_A755delinsSKG | COSM1667026 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A755delinsSMS | COSM6933365 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsF | OMINDEL951 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsPT | COSM5023004 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_E758delinsP | COSM26440 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.A750_I759delinsGS | COSM5023005 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.T751_E758del | COSM133207 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM133200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM22945 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsS | COSM1667027 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM24270 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.S752_I759del | COSM13556 | NEGATIVE | del |
| EGFR exon 19 deletion | p.S752_I759del | COSM6256 | NEGATIVE | del |
| EGFR exon 19 deletion | p.K754_I759del | COSM5271797 | NEGATIVE | del |
| EGFR K757R | p.K757R | COSM133588 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.? | COSM26720 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insTLA | COSM12425 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM12376 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGGTASV | OMINDEL1052 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | A767_V769dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insTSV | OMINDEL1055 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_P772dup | OMINDEL1053 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insRSV | OMINDEL952 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_N771dup | S768_N771dup | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM18429 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGSV | COSM1235344 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRVDNP | OMINDEL1056 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insCV | COSM12379 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.S768_V769delinsISW | OMINDEL1058 | NEGATIVE | complex |
| EGFR S768N | p.S768N | COSM12989 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_P772dup | OMINDEL1057 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_N771dup | COSM20885 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGVV | COSM18430 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768delinsRRGH | OMINDEL1059 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM6984779 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769dup | COSM6506514 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1066 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1063 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1065 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSTL | OMINDEL1064 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1072 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1073 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insKD | OMINDEL1067 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1068 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1071 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGQ | OMINDEL1070 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGG | MATNV07 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insMASVD | COSM28638 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_P772dup | COSM1651745 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGTD | OMINDEL1074 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.V769_D770insERG | COSM1651742 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | MATNV03 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsNNN | OMINDEL1077 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsNNPH | OMINDEL1078 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_1 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insNN | OMINDEL1085 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insTHN | OMINDEL1086 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHN | OMINDEL1084 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1082 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYNP | OMINDEL1083 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insH | OMINDEL1081 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGNPH | D770_N771insGNPH | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsASVH | OMINDEL1080 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGTH | COSM6983510 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM13558 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsGGT | OMINDEL1090 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770_N771delinsAGG | COSM12737 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771_P772dup | MAN123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | COSM6977296 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1108 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1106 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1103 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.H773_V774insGNPH | COSM51544 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPNP | OMINDEL1087 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1110 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1102 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insANP | OMINDEL1099 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1100 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insT | COSM5023008 | NEGATIVE | ins |
| EGFR N771T | p.N771T | OM3174 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_N771insQRG | COSM4970107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insP | OMINDEL1096 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1094 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM13004 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insVDSVDNP | OMINDEL953 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEG | OMINDEL1092 | NEGATIVE | ins |
| EGFR [D770G;N771H] | p.[D770G;N771H] | MATNV09 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771delinsERG | MATNV06 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1116 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1114 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1112 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1111 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insSVE | COSM1651743 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insSVE | D770_N771insSVE | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGT | COSM1238029 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | MATNV05 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGL | COSM48921 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGF | COSM655155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsHH | OMINDEL1118 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGD | OMINDEL1119 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsVH | COSM5023007 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1123 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insHH | COSM6931207 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insH | COSM1238031 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHPH | OMINDEL1122 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | p.H773_V774insPHPH_2 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insPHP | OMINDEL1136 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insRHP | OMINDEL1137 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1132 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTHP | COSM48923 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL1133 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insRH | COSM166390 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1126 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsTH | COSM22946 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1127 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsSTH | COSM6920147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1130 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1128 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSGH | COSM1651744 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772delinsSVDNR | COSM13554 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPP | OMINDEL955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKL | COSM6438147 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1138 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1145 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_C775dup | OMINDEL956 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1183 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1139 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insG | OMINDEL1143 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1142 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1147 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1150 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_V774dup | COSM6845098 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTH | OMINDEL1151 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insAH | COSM1238028 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGT | MATNV02 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772delinsHR | OMINDEL1153 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1154 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insR | OMINDEL1156 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insL | OMINDEL1155 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTP | COSM12388 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1158 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insGH | OMINDEL1157 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM22948 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insV | COSM255205 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsYNPY | OMINDEL1160 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1162 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsYPNPY | OMINDEL1163 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.C775_R776insPYVC | OMINDEL1164 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insY | OMINDEL1168 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | OMINDEL1165 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsPNPY | COSM1735761 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insTQPP | COSM3727813 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insHV | OMINDEL1169 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insQ | COSM131552 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V774_C775insPR | COSM4170223 | NEGATIVE | ins |
| EGFR L792V | p.L792V | OM3265 | NEGATIVE | SNV |
| EGFR L792P | p.L792P | COSM13190 | NEGATIVE | SNV |
| EGFR G796C | p.G796C | OM3318 | NEGATIVE | SNV |
| EGFR C797G | p.C797G | COSM6493936 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR L833W | p.L833W | COSM133204 | NEGATIVE | SNV |
| EGFR P848L | p.P848L | COSM22943 | NEGATIVE | SNV |
| EGFR L858K | p.L858K | COSM24268 | NEGATIVE | mnp |
| EGFR L858R | p.L858R | COSM6224 | NEGATIVE | SNV |
| EGFR L861Q | p.L861Q | COSM6213 | NEGATIVE | SNV |
| EGFR R108G | p.R108G | COSM1451536 | NEGATIVE | SNV |
| EGFR V292L | p.V292L | COSM43067 | NEGATIVE | SNV |
| EGFR S492R | p.S492R | COSM236671 | NEGATIVE | SNV |
| EGFR E709H | p.E709H | COSM12428 | NEGATIVE | mnp |
| EGFR L718M | p.L718M | COSM1716255 | NEGATIVE | SNV |
| EGFR G719D | p.G719D | COSM18425 | NEGATIVE | SNV |
| EGFR G721R | p.G721R | OM3325 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.I744_E749delinsLKR | COSM133186 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.K745_A750del | COSM221565 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_A750del | COSM6223 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_P753del | OMINDEL950 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM12385 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_E749del | COSM6218 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_S752delinsI | COSM133190 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsIP | COSM6966471 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsS | COSM26513 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsA | COSM12367 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsVP | COSM28623 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_S752delinsV | COSM674057 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVA | COSM12416 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751del | COSM12369 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsQ | COSM12419 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_A750delinsDP | COSM18428 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_A750delinsP | COSM4170220 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_T751delinsA | COSM23572 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.L747_P753delinsQ | COSM12387 | NEGATIVE | complex |
| EGFR L747V | p.L747V | OM3537 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_K754delinsST | COSM20883 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_A755del | COSM26718 | NEGATIVE | del |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM1667024 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsN | COSM96856 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.P753_I759del | COSM24269 | NEGATIVE | del |
| EGFR exon 20 insertion | p.Y764_V765insHH | OMINDEL1051 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_V769dup | COSM20884 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insSSV | OMINDEL1054 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.S768_D770dup | COSM13428 | NEGATIVE | ins |
| EGFR S768G | p.S768G | COSM1451595 | NEGATIVE | SNV |
| EGFR S768T | p.S768T | COSM291998 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insANV | COSM1651741 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR S768I | p.S768I | COSM6241 | NEGATIVE | SNV |
| EGFR [S768I;V769L] | p.[S768I;V769L] | OMINDEL1181 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.V769_D770insGTM | OMINDEL1061 | NEGATIVE | ins |
| EGFR [V769G;D770I] | p.[V769G;D770I] | OM3540 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM85795 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGE | OMINDEL1069 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGD | COSM22955 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTR | OMINDEL1075 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771dup | COSM13003 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_H773dup | COSM12381 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insY | COSM1238030 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsGY | COSM12427 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1101 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTPNP | OMINDEL1107 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_V774dup | OMINDEL1097 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGNP | OMINDEL1088 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1104 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insMATP | COSM26719 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insGV | OMINDEL1095 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770delinsEF | OMINDEL1091 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1115 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insYPNP | OMINDEL1117 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.D770_N771insGL | OMINDEL1182 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insG | COSM12378 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1121 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsGC | OMINDEL904 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsKPPH | OMINDEL1125 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | COSM12380 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1134 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKH | OMINDEL1124 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsRH | OMINDEL1129 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSH | COSM24434 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsSVDS | OMINDEL1140 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773_V774insAPH | OMINDEL1144 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insTTP | OMINDEL1146 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insT | OMINDEL1148 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771_P772insL | OMINDEL1149 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insNH | COSM5023006 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774dup | COSM12389 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773dup | OMINDEL1184 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insQ | OMINDEL1159 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1166 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsQY | OMINDEL1167 | NEGATIVE | ins |
| EGFR [H773L;V774M] | p.[H773L;V774M] | COSM6978402 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR T790M | p.T790M | COSM6240 | NEGATIVE | SNV |
| EGFR L792H | p.L792H | COSM6493934 | NEGATIVE | SNV |
| EGFR G796A | p.G796A | COSM27568 | NEGATIVE | SNV |
| EGFR L833V | p.L833V | COSM13424 | NEGATIVE | SNV |
| EGFR T854A | p.T854A | COSM28537 | NEGATIVE | SNV |
| EGFR L858M | p.L858M | COSM12366 | NEGATIVE | SNV |
| EGFR L861V | p.L861V | COSM24848 | NEGATIVE | SNV |
| EGFR R108K | p.R108K | COSM21683 | NEGATIVE | SNV |
| EGFR A289T | p.A289T | COSM21686 | NEGATIVE | SNV |
| EGFR A289V | p.A289V | COSM21687 | NEGATIVE | SNV |
| EGFR G465E | p.G465E | BT129 | NEGATIVE | SNV |
| EGFR I491M | p.I491M | BT135 | NEGATIVE | SNV |
| EGFR E709K | p.E709K | COSM12988 | NEGATIVE | SNV |
| EGFR E709Q | p.E709Q | COSM116882 | NEGATIVE | SNV |
| EGFR E709G | p.E709G | COSM13009 | NEGATIVE | SNV |
| EGFR E709V | p.E709V | COSM12371 | NEGATIVE | SNV |
| EGFR [L718=;G719C] | p.[L718=;G719C] | COSM18441 | NEGATIVE | mnp |
| EGFR G719C | p.G719C | COSM6253 | NEGATIVE | SNV |
| EGFR S720I | p.S720I | OM3317 | NEGATIVE | mnp |
| EGFR G721S | p.G721S | COSM22992 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_E749del | COSM28517 | NEGATIVE | del |
| EGFR exon 19 deletion | p.E746_T751delinsI | COSM13551 | NEGATIVE | complex |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 19 deletion | p.E746_A750delinsQP | COSM13557 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsV | COSM12386 | NEGATIVE | complex |
| EGFR E746G | p.E746G | COSM6849765 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.L747_S752del | COSM6255 | NEGATIVE | del |
| EGFR exon 19 deletion | p.L747_T751delinsS | COSM6210 | NEGATIVE | del |
| EGFR A750P | p.A750P | COSM6219 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.Y764_D770dup | COSM6962256 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.A767_S768insIA | COSM13559 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insGTV | OMINDEL1060 | NEGATIVE | ins |
| EGFR [S768I;V769L] | p.[S768I;V769L] | COSM85750 | NEGATIVE | mnp |
| EGFR V769M | p.V769M | COSM13425 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.V769_D770insM | OMINDEL1062 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.V769_D770insP | OMINDEL1076 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771delinsHNPH | OMINDEL1079 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.D770delinsGV | D770delinsGV | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insTRNP | OMINDEL1109 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insARV | OMINDEL1093 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insNPNP | OMINDEL1113 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsPH | MATNV04 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.P772_H773insGDP | OMINDEL954 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insGHP | OMINDEL1135 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsKG | OMINDEL1141 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771_P772insV | COSM6922328 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773_V774insPHPH | H773_V774insPHPH | NEGATIVE | complex |
| EGFR exon 20 insertion | p.H773delinsNPY | OMINDEL1161 | NEGATIVE | complex |
| EGFR T790A | p.T790A | COSM1272071 | NEGATIVE | SNV |
| EGFR L792Y | p.L792Y | OM3266 | NEGATIVE | mnp |
| EGFR L792R | p.L792R | OM3267 | NEGATIVE | SNV |
| EGFR G796S | p.G796S | COSM20891 | NEGATIVE | SNV |
| EGFR C797N | p.C797N | OM3543 | NEGATIVE | mnp |
| EGFR H835L | p.H835L | COSM6227 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM13553 | NEGATIVE | mnp |
| EGFR L861R | p.L861R | COSM12374 | NEGATIVE | SNV |
| EGFR C797S | p.C797S | COSM5945664 | NEGATIVE | SNV |
| EGFR G598A | p.G598A | COSM3412196 | NEGATIVE | SNV |
| EGFR R451C | p.R451C | COSM3266583 | NEGATIVE | SNV |
| EGFR E709_T710delinsD | p.E709_T710delinsD | COSM51525 | NEGATIVE | del |
| EGFR L718Q | p.L718Q | COSM6503269 | NEGATIVE | SNV |
| EGFR exon 19 deletion | p.E746_P753delinsLS | COSM13200 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.E746_T751delinsVP | COSM18421 | NEGATIVE | complex |
| EGFR exon 19 deletion | p.T751_I759delinsREA | COSM22956 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.A767_S768insYVM | COSM1651740 | NEGATIVE | ins |
| EGFR V769L | p.V769L | COSM6242 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.D770_P772dup | COSM6845099 | NEGATIVE | ins |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.P772_H773insSNP | OMINDEL1105 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.D770_N771insAPW | COSM20886 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.N771delinsGF | COSM18431 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.N771delinsSPH | OMINDEL1131 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.P772_H773insT | OMINDEL1152 | NEGATIVE | ins |
| EGFR exon 20 insertion | p.H773delinsRY | MATNV08 | NEGATIVE | ins |
| EGFR C797S | p.C797S | COSM6493937 | NEGATIVE | SNV |
| EGFR T854I | p.T854I | COSM87246 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM12429 | NEGATIVE | mnp |
| EGFR G721D | p.G721D | COSM22993 | NEGATIVE | SNV |
| EGFR L833F | p.L833F | COSM219794 | NEGATIVE | SNV |
| EGFR L858R | p.L858R | COSM133630 | NEGATIVE | mnp |
| EGFR exon 20 insertion | p.D770_N771insKH | D770_N771insKH_2 | NEGATIVE | ins |
| EGFR L858Q | p.L858Q | COSM29578 | NEGATIVE | SNV |
| EGFR L792F | p.L792F | COSM6493933 | NEGATIVE | SNV |
| EGFR R836C | p.R836C | COSM28604 | NEGATIVE | SNV |
| EGFR C775Y | p.C775Y | COSM22954 | NEGATIVE | SNV |
| EGFR exon 20 insertion | p.N771delinsGY | COSM53189 | NEGATIVE | complex |
| EGFR G796R | p.G796R | COSM6493935 | NEGATIVE | SNV |
| EGFR exon 19 insertion | p.I740_K745dup | COSM51504 | NEGATIVE | ins |
| EGFR exon 19 deletion | p.L747_S752delinsQH | COSM133196 | NEGATIVE | complex |
| EGFR G719A | p.G719A | COSM6239 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| EGFR exon 20 insertion | p.N771delinsRD | OMINDEL1120 | NEGATIVE | complex |
| EGFR exon 20 insertion | p.? | OMINDEL1050 | NEGATIVE | ins |
| EGFR L798I | p.L798I | OM3268 | NEGATIVE | SNV |
| EGFR T854S | p.T854S | COSM1235378 | NEGATIVE | SNV |
| ERBB2 G309A | p.G309A | COSM436496 | NEGATIVE | SNV |
| ERBB2 S310F | p.S310F | COSM48358 | NEGATIVE | SNV |
| ERBB2 C311R | p.C311R | COSM35763 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM6503262 | NEGATIVE | mnp |
| ERBB2 L755_T759del | p.L755_T759del | COSM7345568 | NEGATIVE | del |
| ERBB2 L755A | p.L755A | COSM6906940 | NEGATIVE | mnp |
| ERBB2 D769N | p.D769N | COSM1302747 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM20959 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.E770_A771insGIRD | COSM7449692 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.Y772_A775dup | COSM12558 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insTVMA | MAN302 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insSVMA | OMINDEL821 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insVVMA | OMINDEL822 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774delinsWLV | OMINDEL1178 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.A775_G776insVAG | OMINDEL817 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN307 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN309 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN308 | NEGATIVE | ins |
| ERBB2 G776C | p.G776C | COSM303938 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM20895 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsAVGC | COSM6907033 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM12553 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777insL | COSM6438151 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_S779dup | COSM681 | NEGATIVE | ins |
| ERBB2 V777M | p.V777M | COSM14064 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12555 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778dup | COSM26681 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM12556 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_P780dup | COSM6865893 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insCPG | OMINDEL1180 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPG | COSM7449770 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G778_S779insLPS | COSM5802314 | NEGATIVE | ins |
| ERBB2 V842E | p.V842E | COSM6005745 | NEGATIVE | SNV |
| ERBB2 R896H | p.R896H | COSM119971 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 S310Y | p.S310Y | COSM94225 | NEGATIVE | SNV |
| ERBB2 G660D | p.G660D | COSM4681497 | NEGATIVE | SNV |
| ERBB2 L755M | p.L755M | COSM1205571 | NEGATIVE | SNV |
| ERBB2 I767M | p.I767M | COSM51317 | NEGATIVE | SNV |
| ERBB2 D769Y | p.D769Y | COSM1251412 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insAVMA | OMINDEL816 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.M774dup | OMINDEL823 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insC | OMINDEL825 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.A775_G776insV | MAN300 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsIC | OMINDEL826 | NEGATIVE | complex |
| ERBB2 G776R | p.G776R | OM3421 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G778_S779insCVG | OMINDEL1179 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM85995 | NEGATIVE | ins |
| ERBB2 V777A | p.V777A | COSM35496 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.V777_G778insCG | COSM303939 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.V777_G778insC | OMINDEL827 | NEGATIVE | ins |
| ERBB2 V842I | p.V842I | COSM14065 | NEGATIVE | SNV |
| ERBB2 E321G | p.E321G | COSM35764 | NEGATIVE | SNV |
| ERBB2 L755_T759del | p.L755_T759del | COSM5029269 | NEGATIVE | del |
| ERBB2 L755P | p.L755P | COSM683 | NEGATIVE | mnp |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 L755W | p.L755W | COSM436499 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.Y772_V773insMMAY | OMINDEL820 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776_V777delinsSVA | G776_V777delinsSVA_3 | NEGATIVE | complex |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM12554 | NEGATIVE | complex |
| ERBB2 G776S | p.G776S | COSM685 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsVC | COSM5235349 | NEGATIVE | ins |
| ERBB2 G776V | p.G776V | COSM18609 | NEGATIVE | SNV |
| ERBB2 T798I | p.T798I | OM3210 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.G776delinsLC | COSM19875 | NEGATIVE | complex |
| ERBB2 D769H | p.D769H | COSM13170 | NEGATIVE | SNV |
| ERBB2 G309E | p.G309E | COSM94224 | NEGATIVE | SNV |
| ERBB2 V659E | p.V659E | COSM3724566 | NEGATIVE | mnp |
| ERBB2 L755S | p.L755S | COSM14060 | NEGATIVE | SNV |
| ERBB2 exon 20 insertion | p.A775_G776insYVMS | OMINDEL824 | NEGATIVE | ins |
| ERBB2 exon 20 insertion | p.G776delinsVV | COSM6977433 | NEGATIVE | complex |
| ERBB2 L869R | p.L869R | COSM249793 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM436500 | NEGATIVE | SNV |
| ERBB2 S310P | p.S310P | COSM1563863 | NEGATIVE | SNV |
| ERBB2 S310Y | p.S310Y | COSM6906440 | NEGATIVE | mnp |
| ERBB2 exon 20 insertion | p.A775_G776insVA | COSM6865891 | NEGATIVE | ins |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB2 T733I | p.T733I | COSM14059 | NEGATIVE | SNV |
| ERBB2 V777L | p.V777L | COSM14062 | NEGATIVE | SNV |
| ERBB2 R678Q | p.R678Q | COSM436498 | NEGATIVE | SNV |
| ERBB2 R868W | p.R868W | COSM289684 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.G778_P780dup | COSM303948 | NEGATIVE | ins |
| ERBB2 R896C | p.R896C | COSM14066 | NEGATIVE | SNV |
| ERBB2 exon 20 insertio n | p.G776delinsVC | COSM12552 | NEGATIVE | ins |
| ERBB3 M60L | p.M60L | COSM1606366 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM1299636 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM191840 | NEGATIVE | SNV |
| ERBB3 D297H | p.D297H | COSM1935392 | NEGATIVE | SNV |
| ERBB3 E332Q | p.E332Q | COSM4838033 | NEGATIVE | SNV |
| ERBB3 M60K | p.M60K | COSM254678 | NEGATIVE | SNV |
| ERBB3 M60R | p.M60R | COSM941484 | NEGATIVE | SNV |
| ERBB3 V104L | p.V104L | COSM160824 | NEGATIVE | SNV |
| ERBB3 D297Y | p.D297Y | COSM160822 | NEGATIVE | SNV |
| ERBB3 V104M | p.V104M | COSM172423 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM5413554 | NEGATIVE | SNV |
| ERBB3 D297N | p.D297N | COSM941489 | NEGATIVE | SNV |
| ERBB3 D297V | p.D297V | COSM941490 | NEGATIVE | SNV |
| ERBB3 M91I | p.M91I | COSM122890 | NEGATIVE | SNV |
| ERBB3 G284R | p.G284R | COSM941488 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| ERBB3 E332K | p.E332K | COSM254677 | NEGATIVE | SNV |
| ERBB3 R103G | p.R103G | COSM6907312 | NEGATIVE | SNV |
| ERBB4 R711C | p.R711C | COSM160827 | NEGATIVE | SNV |
| ERBB4 R711L | p.R711L | OM3150 | NEGATIVE | SNV |
| ESR1 K303R | p.K303R | COSM4745827 | NEGATIVE | SNV |
| ESR1 P535T | p.P535T | COSM5666097 | NEGATIVE | SNV |
| ESR1 Y537N | p.Y537N | COSM1074635 | NEGATIVE | SNV |
| ESR1 R555C | p.R555C | BT32 | NEGATIVE | SNV |
| ESR1 V392I | p.V392I | COSM1545070 | NEGATIVE | SNV |
| ESR1 L536R | p.L536R | COSM4774826 | NEGATIVE | SNV |
| ESR1 E380Q | p.E380Q | COSM3829320 | NEGATIVE | SNV |
| ESR1 P535H | p.P535H | BT24 | NEGATIVE | SNV |
| ESR1 L536Q | p.L536Q | COSM4766050 | NEGATIVE | mnp |
| ESR1 Y537C | p.Y537C | COSM1074637 | NEGATIVE | SNV |
| ESR1 S463P | p.S463P | COSM4771561 | NEGATIVE | SNV |
| ESR1 D538G | p.D538G | COSM94250 | NEGATIVE | SNV |
| ESR1 Y537S | p.Y537S | COSM1074639 | NEGATIVE | SNV |
| FGFR1 K687E | p.K687E | COSM35673 | NEGATIVE | SNV |
| FGFR1 K687N | p.K687N | COSM6578115 | NEGATIVE | SNV |
| FGFR1 K687M | p.K687M | COSM4771556 | NEGATIVE | SNV |
| FGFR2 K659M | p.K659M | COSM49175 | NEGATIVE | SNV |
| FGFR2 L617V | p.L617V | OM3291 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 E565A | p.E565A | OM3292 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36902 | NEGATIVE | SNV |
| FGFR2 N549D | p.N549D | COSM4604460 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM48382 | NEGATIVE | SNV |
| FGFR2 C382G | p.C382G | COSM6197913 | NEGATIVE | SNV |
| FGFR2 A315T | p.A315T | COSM30777 | NEGATIVE | SNV |
| FGFR2 P253R | p.P253R | COSM49170 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM49173 | NEGATIVE | SNV |
| FGFR2 K641R | p.K641R | COSM5010388 | NEGATIVE | SNV |
| FGFR2 V564F | p.V564F | OM3293 | NEGATIVE | SNV |
| FGFR2 N549S | p.N549S | COSM3665553 | NEGATIVE | SNV |
| FGFR2 H544Q | p.H544Q | COSM1474415 | NEGATIVE | SNV |
| FGFR2 C382R | p.C382R | COSM36906 | NEGATIVE | SNV |
| FGFR2 K310R | p.K310R | COSM36901 | NEGATIVE | SNV |
| FGFR2 V679I | p.V679I | COSM4822956 | NEGATIVE | SNV |
| FGFR2 L617M | p.L617M | OM3562 | NEGATIVE | SNV |
| FGFR2 G613V | p.G613V | COSM94318 | NEGATIVE | SNV |
| FGFR2 V564L | p.V564L | OM3564 | NEGATIVE | SNV |
| FGFR2 N549H | p.N549H | COSM250083 | NEGATIVE | SNV |
| FGFR2 C382Y | p.C382Y | COSM915493 | NEGATIVE | SNV |
| FGFR2 Y375C | p.Y375C | COSM36904 | NEGATIVE | SNV |
| FGFR2 A314D | p.A314D | COSM49171 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR2 P253S | p.P253S | COSM1346291 | NEGATIVE | SNV |
| FGFR2 S252W | p.S252W | COSM36903 | NEGATIVE | SNV |
| FGFR2 K659N | p.K659N | COSM683054 | NEGATIVE | SNV |
| FGFR2 E565G | p.E565G | OM3563 | NEGATIVE | SNV |
| FGFR2 V564I | p.V564I | OM3247 | NEGATIVE | SNV |
| FGFR2 I547V | p.I547V | COSM36913 | NEGATIVE | SNV |
| FGFR2 M391R | p.M391R | COSM36907 | NEGATIVE | SNV |
| FGFR2 P253L | p.P253L | COSM537801 | NEGATIVE | SNV |
| FGFR2 K659E | p.K659E | COSM36909 | NEGATIVE | SNV |
| FGFR2 V395D | p.V395D | COSM250081 | NEGATIVE | mnp |
| FGFR2 Y375H | p.Y375H | COSM1560916 | NEGATIVE | SNV |
| FGFR2 A648T | p.A648T | COSM29836 | NEGATIVE | SNV |
| FGFR2 N549K | p.N549K | COSM36912 | NEGATIVE | SNV |
| FGFR2 N549T | p.N549T | COSM6952624 | NEGATIVE | SNV |
| FGFR3 R248C | p.R248C | COSM714 | NEGATIVE | SNV |
| FGFR3 S249C | p.S249C | COSM715 | NEGATIVE | SNV |
| FGFR3 S371C | p.S371C | COSM17461 | NEGATIVE | SNV |
| FGFR3 G380E | p.G380E | COSM51545 | NEGATIVE | SNV |
| FGFR3 R399C | p.R399C | COSM296687 | NEGATIVE | SNV |
| FGFR3 D641G | p.D641G | COSM1428721 | NEGATIVE | SNV |
| FGFR3 K650R | p.K650R | OM3224 | NEGATIVE | SNV |
| FGFR3 V677I | p.V677I | COSM178247 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR3 G370D | p.G370D | COSM5966955 | NEGATIVE | SNV |
| FGFR3 A391T | p.A391T | OM3151 | NEGATIVE | SNV |
| FGFR3 V555M | p.V555M | COSM303914 | NEGATIVE | SNV |
| FGFR3 K650T | p.K650T | COSM731 | NEGATIVE | SNV |
| FGFR3 K715M | p.K715M | COSM732992 | NEGATIVE | SNV |
| FGFR3 A391V | p.A391V | COSM1737909 | NEGATIVE | SNV |
| FGFR3 K650E | p.K650E | COSM719 | NEGATIVE | SNV |
| FGFR3 K650M | p.K650M | COSM720 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM1428730 | NEGATIVE | SNV |
| FGFR3 S249T | p.S249T | COSM29431 | NEGATIVE | SNV |
| FGFR3 G380R | p.G380R | COSM24842 | NEGATIVE | SNV |
| FGFR3 K650N | p.K650N | COSM3993568 | NEGATIVE | SNV |
| FGFR3 A391E | p.A391E | COSM721 | NEGATIVE | SNV |
| FGFR3 K650Q | p.K650Q | COSM726 | NEGATIVE | SNV |
| FGFR3 R248H | p.R248H | COSM1757310 | NEGATIVE | SNV |
| FGFR3 V553M | p.V553M | OM3223 | NEGATIVE | SNV |
| FGFR3 Y373C | p.Y373C | COSM718 | NEGATIVE | SNV |
| FGFR3 G370C | p.G370C | COSM716 | NEGATIVE | SNV |
| FGFR3 D641N | p.D641N | COSM29438 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM3738141 | NEGATIVE | SNV |
| FGFR4 V550L | p.V550L | COSM1066172 | NEGATIVE | SNV |
| FGFR4 V550M | p.V550M | COSM6163 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FGFR4 V550E | p.V550E | COSM9215998 | NEGATIVE | SNV |
| FLT3 D839E | p.D839E | COSM5945287 | NEGATIVE | SNV |
| FLT3 I836del | p.I836del | COSM19836 | NEGATIVE | del |
| FLT3 I836T | p.I836T | COSM5879386 | NEGATIVE | SNV |
| FLT3 D835E | p.D835E | COSM788 | NEGATIVE | SNV |
| FLT3 D835F | p.D835F | COSM19844 | NEGATIVE | mnp |
| FLT3 D835A | p.D835A | COSM27650 | NEGATIVE | SNV |
| FLT3 D835_I836del | p.D835_I836del | COSM5879385 | NEGATIVE | del |
| FLT3 I836L | p.I836L | COSM19649 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3571 | NEGATIVE | mnp |
| FLT3 D835N | p.D835N | COSM789 | NEGATIVE | SNV |
| FLT3 D839H | p.D839H | COSM3399304 | NEGATIVE | SNV |
| FLT3 D839N | p.D839N | COSM6005768 | NEGATIVE | SNV |
| FLT3 D835_I836delinsV | p.D835_I836delinsV | COSM27651 | NEGATIVE | del |
| FLT3 I836D | p.I836D | OM3569 | NEGATIVE | mnp |
| FLT3 D835E | p.D835E | COSM787 | NEGATIVE | SNV |
| FLT3 D835V | p.D835V | COSM784 | NEGATIVE | SNV |
| FLT3 D835G | p.D835G | COSM1366188 | NEGATIVE | SNV |
| FLT3 D835H | p.D835H | COSM785 | NEGATIVE | SNV |
| FLT3 I836S | p.I836S | COSM19843 | NEGATIVE | SNV |
| FLT3 D835K | p.D835K | OM3570 | NEGATIVE | mnp |
| FLT3 D835L | p.D835L | OM3572 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| FLT3 D835Y | p.D835Y | COSM783 | NEGATIVE | SNV |
| FLT3 D839G | p.D839G | COSM1166729 | NEGATIVE | SNV |
| FLT3 D835del | p.D835del | COSM854 | NEGATIVE | del |
| GNAS R201S | p.R201S | COSM27899 | NEGATIVE | SNV |
| GNAS R201L | p.R201L | COSM99221 | NEGATIVE | SNV |
| GNAS Q227L | p.Q227L | COSM27888 | NEGATIVE | SNV |
| GNAS R201G | p.R201G | COSM1757313 | NEGATIVE | SNV |
| GNAS Q227E | p.Q227E | COSM4962873 | NEGATIVE | SNV |
| GNAS R201H | p.R201H | COSM27895 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM4168960 | NEGATIVE | SNV |
| GNAS Q227H | p.Q227H | COSM27900 | NEGATIVE | SNV |
| GNAS Q227K | p.Q227K | COSM28618 | NEGATIVE | SNV |
| GNAS R201C | p.R201C | COSM27887 | NEGATIVE | SNV |
| GNAS Q227R | p.Q227R | COSM27896 | NEGATIVE | SNV |
| HRAS Q61H | p.Q61H | COSM502 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM498 | NEGATIVE | SNV |
| HRAS Q61K | p.Q61K | COSM496 | NEGATIVE | SNV |
| HRAS G13C | p.G13C | COSM488 | NEGATIVE | SNV |
| HRAS G12A | p.G12A | COSM485 | NEGATIVE | SNV |
| HRAS G12C | p.G12C | COSM481 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM501 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM499 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| HRAS G13V | p.G13V | COSM489 | NEGATIVE | SNV |
| HRAS G12V | p.G12V | COSM483 | NEGATIVE | SNV |
| HRAS G12R | p.G12R | COSM482 | NEGATIVE | SNV |
| HRAS Q61P | p.Q61P | COSM500 | NEGATIVE | SNV |
| HRAS Q61R | p.Q61R | COSM52979 | NEGATIVE | mnp |
| HRAS G13D | p.G13D | COSM490 | NEGATIVE | SNV |
| HRAS G12D | p.G12D | COSM484 | NEGATIVE | SNV |
| HRAS G12N | p.G12N | COSM4517473 | NEGATIVE | mnp |
| HRAS Q61R | p.Q61R | COSM33695 | NEGATIVE | mnp |
| HRAS Q61H | p.Q61H | COSM503 | NEGATIVE | SNV |
| HRAS G13R | p.G13R | COSM486 | NEGATIVE | SNV |
| HRAS Q61L | p.Q61L | COSM52978 | NEGATIVE | mnp |
| HRAS G12S | p.G12S | COSM480 | NEGATIVE | SNV |
| HRAS G13S | p.G13S | COSM487 | NEGATIVE | SNV |
| HRAS Q61E | p.Q61E | COSM497 | NEGATIVE | SNV |
| IDH1 R132Q | p.R132Q | OM3221 | NEGATIVE | mnp |
| IDH1 R132V | p.R132V | COSM28751 | NEGATIVE | mnp |
| IDH1 R132G | p.R132G | COSM28749 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM86993 | NEGATIVE | mnp |
| IDH1 R132S | p.R132S | COSM1169216 | NEGATIVE | mnp |
| IDH1 R132Q | p.R132Q | OM3222 | NEGATIVE | mnp |
| IDH1 R132C | p.R132C | COSM28747 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| IDH1 G70D | p.G70D | COSM96531 | NEGATIVE | SNV |
| IDH1 S280F | p.S280F | OM3315 | NEGATIVE | SNV |
| IDH1 R132L | p.R132L | COSM28750 | NEGATIVE | SNV |
| IDH1 R132S | p.R132S | COSM28748 | NEGATIVE | SNV |
| IDH1 R132H | p.R132H | COSM28746 | NEGATIVE | SNV |
| IDH2 I319M | p.I319M | OM3319 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM34090 | NEGATIVE | SNV |
| IDH2 R172T | p.R172T | COSM1636997 | NEGATIVE | SNV |
| IDH2 R172W | p.R172W | COSM34039 | NEGATIVE | SNV |
| IDH2 R140G | p.R140G | COSM1737874 | NEGATIVE | SNV |
| IDH2 R172S | p.R172S | COSM133672 | NEGATIVE | SNV |
| IDH2 R172G | p.R172G | COSM33731 | NEGATIVE | SNV |
| IDH2 R172N | p.R172N | COSM256046 | NEGATIVE | mnp |
| IDH2 R172K | p.R172K | COSM33733 | NEGATIVE | SNV |
| IDH2 R172V | p.R172V | OM3327 | NEGATIVE | mnp |
| IDH2 R140Q | p.R140Q | COSM41590 | NEGATIVE | SNV |
| IDH2 R172M | p.R172M | COSM33732 | NEGATIVE | SNV |
| IDH2 Q316E | p.Q316E | OM3320 | NEGATIVE | SNV |
| IDH2 R140L | p.R140L | COSM41875 | NEGATIVE | SNV |
| KEAP1 c.1709-2A>G | p.? | COSM7088100 | NEGATIVE | SNV |
| KEAP1 c.1708_1708+1delinsTT | p.? | COSM6927565 | NEGATIVE | mnp |
| KEAP1 c.1532-1G>T | p.? | COSM6969960 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 c.1531+2T>G | p.? | OM3723 | NEGATIVE | SNV |
| KEAP1 R470C | p.R470C | COSM564847 | NEGATIVE | SNV |
| KEAP1 R415G | p.R415G | COSM6196677 | NEGATIVE | SNV |
| KEAP1 R320P | p.R320P | COSM346494 | NEGATIVE | SNV |
| KEAP1 c.640-1G>T | p.? | OM3725 | NEGATIVE | SNV |
| KEAP1 c.640-1G>A | p.? | COSM6972470 | NEGATIVE | SNV |
| KEAP1 V155F | p.V155F | COSM710187 | NEGATIVE | SNV |
| KEAP1 c.1708+1G>T | p.? | COSM7738757 | NEGATIVE | SNV |
| KEAP1 R470H | p.R470H | COSM1524064 | NEGATIVE | SNV |
| KEAP1 M409* | p.M409* | COSM6916353 | NEGATIVE | del |
| KEAP1 R320W | p.R320W | COSM1524062 | NEGATIVE | SNV |
| KEAP1 V167F | p.V167F | COSM710188 | NEGATIVE | SNV |
| KEAP1 c.1531+1G>T | p.? | COSM6956065 | NEGATIVE | SNV |
| KEAP1 D422N | p.D422N | COSM710198 | NEGATIVE | SNV |
| KEAP1 G333C | p.G333C | COSM1193323 | NEGATIVE | SNV |
| KEAP1 R320Q | p.R320Q | COSM710195 | NEGATIVE | SNV |
| KEAP1 S243C | p.S243C | COSM710191 | NEGATIVE | SNV |
| KEAP1 Y584* | p.Y584* | COSM6201646 | NEGATIVE | ins |
| KEAP1 c.1532-2A>G | p.? | COSM381459 | NEGATIVE | SNV |
| KEAP1 R470S | p.R470S | COSM564846 | NEGATIVE | SNV |
| KEAP1 c.640-1G>C | p.? | OM3726 | NEGATIVE | SNV |
| KEAP1 R320L | p.R320L | COSM2812614 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KEAP1 R554Q | p.R554Q | COSM1524066 | NEGATIVE | SNV |
| KEAP1 c.1708+2T>A | p.? | COSM6149754 | NEGATIVE | SNV |
| KEAP1 R470G | p.R470G | OM3724 | NEGATIVE | SNV |
| KEAP1 c.640-2A>C | p.? | OM3727 | NEGATIVE | SNV |
| KEAP1 c.1709-1G>T | p.? | COSM7724088 | NEGATIVE | SNV |
| KIT T417_D419delinsI | p.T417_D419delinsI | COSM1148 | NEGATIVE | complex |
| KIT D419_L421delinsH | p.D419_L421delinsH | COSM1578133 | NEGATIVE | complex |
| KIT D419_R420insFF | p.D419_R420insFF | COSM36334 | NEGATIVE | ins |
| KIT S501_A502dup | p.S501_A502dup | COSM5702501 | NEGATIVE | ins |
| KIT A502_Y503dup | p.A502_Y503dup | COSM1326 | NEGATIVE | ins |
| KIT A502_Y503insFA | p.A502_Y503insFA | COSM53306 | NEGATIVE | ins |
| KIT F522C | p.F522C | OM3237 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | COSM1160 | NEGATIVE | del |
| KIT exon 11 deletion | p.? | COSM1158 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_E554del | COSM1172 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsI | COSM1164 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_Q556delinsII | COSM1161 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIL | COSM6936468 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_P551delinsI | COSM6971267 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551_V555del | COSM1169 | NEGATIVE | del |
| KIT K550I | p.K550I | COSM1165 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.P551_V555delinsTL | COSM30550 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.P551_Y553delinsH | COSM6934157 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_M552delinsL | COSM1176 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V560delinsL | COSM27901 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557del | COSM1177 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM96881 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | OMINDEL902 | NEGATIVE | complex |
| KIT exon 11 deletion | p.M552_W557del | COSM1180 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553del | COSM1179 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554del | COSM1181 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556del | COSM1327 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsK | COSM36311 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Q556delinsK | COSM96868 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_Y553delinsN | COSM96863 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555delinsI | COSM1185 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V559delinsI | COSM3737059 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_W557del | COSM1187 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_K558del | COSM27909 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_E554del | COSM6923694 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559del | COSM1191 | NEGATIVE | del |
| KIT Y553N | p.Y553N | COSM133763 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E554_N564del | COSM96947 | NEGATIVE | del |
| KIT Y553C | p.Y553C | COSM4413463 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.E554_E562del | COSM36310 | NEGATIVE | del |
| KIT exon 11 deletion | p.E554_K558del | COSM1192 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_G565del | COSM37027 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_V560del | COSM1202 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1198 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_Q556del | COSM1201 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_N566delinsD | COSM96870 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557del | COSM36309 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM36315 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558del | COSM23418 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560delinsF | COSM1207 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_E562del | COSM36312 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_K558del | COSM1217 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_W557delinsR | COSM133642 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_K558delinsR | COSM28993 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsRG | COSM6954545 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V560delinsH | COSM1213 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559delinsH | COSM133641 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsQ | COSM6954755 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_V559delinsHT | COSM1214 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_V560del | COSM1223 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559del | COSM1218 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.W557_V559delinsI | COSM1222 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1219 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_V560delinsF | COSM1230 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsF | COSM1226 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_E562del | COSM18896 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558del | COSM6008996 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V560delinsC | COSM1232 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_V559delinsC | COSM1233 | NEGATIVE | del |
| KIT W557C | p.W557C | COSM133700 | NEGATIVE | SNV |
| KIT [W557C;K558T] | p.[W557C;K558T] | COSM28992 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560del | COSM1234 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_N564del | COSM1238 | NEGATIVE | del |
| KIT [W557C;K558P] | p.[W557C;K558P] | COSM21978 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_V560delinsL | COSM6940807 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V559_E561del | COSM1250 | NEGATIVE | del |
| KIT K558N | p.K558N | OM3238 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.K558_V560delinsI | COSM1241 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsM | COSM1242 | NEGATIVE | complex |
| KIT V559C | p.V559C | OM3239 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.K558delinsNP | COSM21976 | NEGATIVE | ins |
| KIT exon 11 deletion | p.K558_V559delinsN | COSM1244 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM1249 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT V559K | p.V559K | COSM96869 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_V560del | COSM1248 | NEGATIVE | del |
| KIT exon 11 deletion | p.V560del | COSM1247 | NEGATIVE | del |
| KIT V559P | p.V559P | COSM1272086 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.V559_N566delinsD | COSM6977976 | NEGATIVE | del |
| KIT V559A | p.V559A | COSM1255 | NEGATIVE | SNV |
| KIT V560D | p.V560D | COSM1257 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.E562del | COSM1263 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562_V569delinsD | COSM1267 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_T574del | COSM24839 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsT | COSM36299 | NEGATIVE | del |
| KIT exon 11 deletion | p.N566_P573del | COSM6948216 | NEGATIVE | del |
| KIT exon 11 deletion | p.N567_P573del | COSM1276 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_L576delinsCV | COSM1279 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_Q575del | COSM1281 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y570_L576del | COSM1285 | NEGATIVE | del |
| KIT Y570H | p.Y570H | COSM1430152 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P573_H580dup | COSM34126 | NEGATIVE | ins |
| KIT exon 11 insertion | p.P573_D579dup | COSM18668 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575del | COSM96875 | NEGATIVE | del |
| KIT exon 11 insertion | p.Q575_L576dup | COSM1292 | NEGATIVE | ins |
| KIT exon 11 deletion | p.Q575_P577del | COSM219110 | NEGATIVE | del |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM219110old | NEGATIVE | complex |
| KIT exon 11 deletion | p.L576del | COSM1289 | NEGATIVE | del |
| KIT exon 11 insertion | p.Y578_P585dup | COSM41632 | NEGATIVE | ins |
| KIT L576P | p.L576P | COSM1290 | NEGATIVE | SNV |
| KIT L576R | p.L576R | COSM5945006 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.P577_H580dup | COSM1296 | NEGATIVE | ins |
| KIT exon 11 deletion | p.P577_Y578del | COSM1291 | NEGATIVE | del |
| KIT exon 11 insertion | p.P577_H580dup | COSM18666 | NEGATIVE | ins |
| KIT exon 11 insertion | p.D579_H580insSYD | COSM96866 | NEGATIVE | ins |
| KIT exon 11 deletion | p.D579del | COSM1294 | NEGATIVE | del |
| KIT K642E | p.K642E | COSM1304 | NEGATIVE | SNV |
| KIT V654A | p.V654A | COSM12706 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM235682 | NEGATIVE | SNV |
| KIT N680K | p.N680K | COSM5352270 | NEGATIVE | SNV |
| KIT R796A | p.R796A | COSM1272091 | NEGATIVE | mnp |
| KIT D816I | p.D816I | COSM21979 | NEGATIVE | mnp |
| KIT D816Y | p.D816Y | COSM1310 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM5749965 | NEGATIVE | SNV |
| KIT D820Y | p.D820Y | COSM12710 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM12709 | NEGATIVE | SNV |
| KIT N822S | p.N822S | COSM33975 | NEGATIVE | SNV |
| KIT Y823N | p.Y823N | COSM22378 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT Y823C | p.Y823C | COSM18682 | NEGATIVE | SNV |
| KIT A829P | p.A829P | COSM13172 | NEGATIVE | SNV |
| KIT D419_R420del | p.D419_R420del | COSM1578132 | NEGATIVE | del |
| KIT D419del | p.D419del | COSM1600400 | NEGATIVE | complex |
| KIT S501_A502dup | p.S501_A502dup | COSM219786 | NEGATIVE | ins |
| KIT N505I | p.N505I | OM3236 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.? | OMINDEL901 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_E554delinsI | COSM6953101 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K550_W557delinsIR | COSM6977972 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P551del | COSM1171 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_Q556del | COSM1170 | NEGATIVE | del |
| KIT exon 11 deletion | p.P551_V555delinsL | COSM1174 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM6957862 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_V555del | COSM18827 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_K558del | COSM1182 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_W557delinsR | COSM133636 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E554delinsI | COSM6944981 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y553_V559delinsE | COSM96946 | NEGATIVE | complex |
| KIT exon 11 deletion | p.E554_V559del | OMINDEL701 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_I563del | COSM1196 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V559del | COSM1199 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_V560del | COSM1204 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q556_V560delinsTTF | COSM27904 | NEGATIVE | complex |
| KIT exon 11 deletion | p.Q556_I563delinsL | COSM1651755 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557del | COSM1220 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q556_E561delinsHH | COSM1208 | NEGATIVE | complex |
| KIT [W557F;K558P] | p.[W557F;K558P] | COSM1224 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.W557_K558delinsE | COSM24748 | NEGATIVE | complex |
| KIT [K558S;V559S] | p.[K558S;V559S] | COSM28637 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.K558_G565del | COSM1236 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V559del | COSM1235 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_G565delinsR | COSM1240 | NEGATIVE | del |
| KIT exon 11 deletion | p.V559_G565del | COSM6971633 | NEGATIVE | del |
| KIT K558N | p.K558N | COSM19216 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V559_I571del | COSM36301 | NEGATIVE | del |
| KIT V559D | p.V559D | COSM1252 | NEGATIVE | SNV |
| KIT V560A | p.V560A | COSM36302 | NEGATIVE | SNV |
| KIT V560G | p.V560G | COSM1260 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.I563_D572del | COSM1268 | NEGATIVE | del |
| KIT exon 11 deletion | p.N564_P573delinsTS | COSM22376 | NEGATIVE | complex |
| KIT exon 11 deletion | p.V569_D572del | COSM1282 | NEGATIVE | del |
| KIT exon 11 deletion | p.V569_L576delinsG | COSM1283 | NEGATIVE | complex |
| KIT exon 11 deletion | p.I571_L576del | COSM133754 | NEGATIVE | del |
| KIT exon 11 insertion | p.D572_D579dup | COSM308544 | NEGATIVE | ins |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Q575_L576del | COSM6987753 | NEGATIVE | del |
| KIT exon 11 deletion | p.Q575_P577delinsT | COSM96865 | NEGATIVE | complex |
| KIT exon 11 deletion | p.P577_D579del | COSM98379 | NEGATIVE | del |
| KIT exon 11 deletion | p.P577del | COSM1684822 | NEGATIVE | del |
| KIT Y578C | p.Y578C | COSM6503446 | NEGATIVE | SNV |
| KIT I653T | p.I653T | COSM19326 | NEGATIVE | SNV |
| KIT N655K | p.N655K | COSM4413464 | NEGATIVE | SNV |
| KIT H697Y | p.H697Y | COSM36053 | NEGATIVE | SNV |
| KIT R796G | p.R796G | COSM51381 | NEGATIVE | SNV |
| KIT D816N | p.D816N | COSM1313 | NEGATIVE | SNV |
| KIT D816F | p.D816F | COSM1312 | NEGATIVE | mnp |
| KIT D820N | p.D820N | COSM17947 | NEGATIVE | SNV |
| KIT D820V | p.D820V | COSM1317 | NEGATIVE | SNV |
| KIT N822I | p.N822I | COSM3604523 | NEGATIVE | SNV |
| KIT V825I | p.V825I | COSM19110 | NEGATIVE | SNV |
| KIT P838L | p.P838L | OM3240 | NEGATIVE | SNV |
| KIT T417_D419delinsRG | p.T417_D419delinsRG | COSM1149 | NEGATIVE | complex |
| KIT D419_R420insFL | p.D419_R420insFL | COSM36332 | NEGATIVE | ins |
| KIT exon 11 deletion | p.? | COSM1162 | NEGATIVE | del |
| KIT exon 11 deletion | p.K550_V555delinsQ | COSM6938892 | NEGATIVE | complex |
| KIT K550N | p.K550N | COSM1167 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.M552_Q556delinsL | COSM1178 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT exon 11 deletion | p.Y553_Q556del | COSM1189 | NEGATIVE | del |
| KIT exon 11 deletion | p.V555_E562del | COSM1194 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561del | COSM1332 | NEGATIVE | del |
| KIT exon 11 deletion | p.W557_E561delinsPS | COSM6922994 | NEGATIVE | complex |
| KIT W557R | p.W557R | COSM1216 | NEGATIVE | SNV |
| KIT [W557S;K558S] | p.[W557S;K558S] | COSM96948 | NEGATIVE | mnp |
| KIT W557S | p.W557S | COSM1227 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558delinsC | COSM96887 | NEGATIVE | complex |
| KIT exon 11 deletion | p.K558_Y570delinsN | COSM28991 | NEGATIVE | del |
| KIT exon 11 deletion | p.K558_V560delinsN | COSM27069 | NEGATIVE | del |
| KIT V559G | p.V559G | COSM1253 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V560_I563del | COSM18897 | NEGATIVE | del |
| KIT exon 11 deletion | p.E562del | COSM1262 | NEGATIVE | del |
| KIT exon 11 deletion | p.Y568_T574del | COSM1334 | NEGATIVE | del |
| KIT exon 11 insertion | p.T574_D579dup | OMINDEL702 | NEGATIVE | ins |
| KIT L576F | p.L576F | COSM33966 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.H580del | COSM1295 | NEGATIVE | del |
| KIT K642Q | p.K642Q | COSM96871 | NEGATIVE | SNV |
| KIT V654L | p.V654L | COSM5941904 | NEGATIVE | SNV |
| KIT N655T | p.N655T | OM3303 | NEGATIVE | SNV |
| KIT T670E | p.T670E | COSM19293 | NEGATIVE | mnp |
| KIT T670A | p.T670A | OM3304 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT R796K | p.R796K | COSM1600411 | NEGATIVE | SNV |
| KIT D816H | p.D816H | COSM1311 | NEGATIVE | SNV |
| KIT D816V | p.D816V | COSM1314 | NEGATIVE | SNV |
| KIT D816A | p.D816A | COSM24675 | NEGATIVE | SNV |
| KIT D816E | p.D816E | COSM19285 | NEGATIVE | SNV |
| KIT D820H | p.D820H | COSM22379 | NEGATIVE | SNV |
| KIT D820G | p.D820G | COSM1316 | NEGATIVE | SNV |
| KIT D820E | p.D820E | COSM19280 | NEGATIVE | SNV |
| KIT N822Y | p.N822Y | COSM19109 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1321 | NEGATIVE | SNV |
| KIT Y823D | p.Y823D | COSM18681 | NEGATIVE | SNV |
| KIT A829V | p.A829V | COSM133757 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.K558delinsNP | COSM1245 | NEGATIVE | complex |
| KIT D419del | p.D419del | COSM29014 | NEGATIVE | del |
| KIT K550R | p.K550R | COSM1166 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.W557_K558del | COSM6986076 | NEGATIVE | complex |
| KIT exon 11 deletion | p.W557_K558delinsQ | COSM6946777 | NEGATIVE | complex |
| KIT W557C | p.W557C | COSM19108 | NEGATIVE | SNV |
| KIT V560E | p.V560E | COSM1333 | NEGATIVE | mnp |
| KIT exon 11 insertion | p.I571_D579dup | COSM23417 | NEGATIVE | ins |
| KIT K642N | p.K642N | COSM4125155 | NEGATIVE | SNV |
| KIT N655S | p.N655S | OM3302 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KIT D816H | p.D816H | COSM1600404 | NEGATIVE | mnp |
| KIT D816G | p.D816G | COSM12711 | NEGATIVE | SNV |
| KIT N822H | p.N822H | COSM1318 | NEGATIVE | SNV |
| KIT N822K | p.N822K | COSM1322 | NEGATIVE | SNV |
| KIT D419_L421delinsF | p.D419_L421delinsF | COSM1152 | NEGATIVE | complex |
| KIT D820A | p.D820A | COSM133670 | NEGATIVE | SNV |
| KIT exon 11 insertion | p.Y578_H580dup | COSM28995 | NEGATIVE | ins |
| KIT T670I | p.T670I | COSM12708 | NEGATIVE | SNV |
| KIT W557G | p.W557G | COSM1221 | NEGATIVE | SNV |
| KIT exon 11 deletion | p.V569_L576del | COSM133635 | NEGATIVE | del |
| KIT exon 11 deletion | p.M552_E561delinsK | COSM96880 | NEGATIVE | del |
| KIT V559I | p.V559I | COSM1251 | NEGATIVE | SNV |
| KIT R796P | p.R796P | COSM1272092 | NEGATIVE | mnp |
| KIT exon 11 deletion | p.Q556_E561del | COSM1206 | NEGATIVE | del |
| KIT Y823H | p.Y823H | COSM1651649 | NEGATIVE | SNV |
| KIT V825A | p.V825A | COSM1323 | NEGATIVE | SNV |
| KRAS A146S | p.A146S | COSM5967494 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM19940 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM1168052 | NEGATIVE | mnp |
| KRAS Q61H | p.Q61H | COSM554 | NEGATIVE | SNV |
| KRAS Q61E | p.Q61E | COSM550 | NEGATIVE | SNV |
| KRAS A59E | p.A59E | COSM547 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G15S | p.G15S | COSM538 | NEGATIVE | SNV |
| KRAS G13D | p.G13D | COSM531 | NEGATIVE | mnp |
| KRAS G13D | p.G13D | COSM532 | NEGATIVE | SNV |
| KRAS G13N | p.G13N | COSM53283 | NEGATIVE | mnp |
| KRAS [G12=;G13C] | p.[G12=;G13C] | COSM87281 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM30566 | NEGATIVE | mnp |
| KRAS G12E | p.G12E | COSM519 | NEGATIVE | mnp |
| KRAS G12A | p.G12A | COSM522 | NEGATIVE | SNV |
| KRAS G12R | p.G12R | COSM518 | NEGATIVE | SNV |
| KRAS G12S | p.G12S | COSM517 | NEGATIVE | SNV |
| KRAS A146T | p.A146T | COSM19404 | NEGATIVE | SNV |
| KRAS Q61R | p.Q61R | COSM552 | NEGATIVE | SNV |
| KRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM87298 | NEGATIVE | mnp |
| KRAS G15D | p.G15D | COSM539 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM530 | NEGATIVE | mnp |
| KRAS G13I | p.G13I | COSM525 | NEGATIVE | mnp |
| KRAS G13C | p.G13C | COSM527 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM515 | NEGATIVE | mnp |
| KRAS G12L | p.G12L | COSM514 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM516 | NEGATIVE | SNV |
| KRAS A146V | p.A146V | COSM19900 | NEGATIVE | SNV |
| KRAS K117N | p.K117N | COSM28519 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS K117E | p.K117E | COSM6854421 | NEGATIVE | SNV |
| KRAS Q61L | p.Q61L | COSM553 | NEGATIVE | SNV |
| KRAS Q61P | p.Q61P | COSM551 | NEGATIVE | SNV |
| KRAS A59G | p.A59G | COSM28518 | NEGATIVE | SNV |
| KRAS [G13D;V14I] | p.[G13D;V14I] | COSM30568 | NEGATIVE | mnp |
| KRAS G13V | p.G13V | COSM534 | NEGATIVE | SNV |
| KRAS G13S | p.G13S | COSM528 | NEGATIVE | SNV |
| KRAS G12W | p.G12W | COSM36281 | NEGATIVE | mnp |
| KRAS G12C | p.G12C | COSM513 | NEGATIVE | mnp |
| KRAS G12F | p.G12F | COSM512 | NEGATIVE | mnp |
| KRAS A146P | p.A146P | COSM19905 | NEGATIVE | SNV |
| KRAS Q61K | p.Q61K | COSM549 | NEGATIVE | SNV |
| KRAS A59T | p.A59T | COSM546 | NEGATIVE | SNV |
| KRAS G13V | p.G13V | COSM12721 | NEGATIVE | mnp |
| KRAS G13E | p.G13E | COSM30567 | NEGATIVE | mnp |
| KRAS G13R | p.G13R | COSM529 | NEGATIVE | SNV |
| KRAS G12V | p.G12V | COSM520 | NEGATIVE | SNV |
| KRAS G12D | p.G12D | COSM14209 | NEGATIVE | mnp |
| KRAS G12Y | p.G12Y | COSM25081 | NEGATIVE | mnp |
| KRAS G12N | p.G12N | COSM13643 | NEGATIVE | mnp |
| KRAS K117R | p.K117R | COSM4696721 | NEGATIVE | SNV |
| KRAS G13A | p.G13A | COSM533 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| KRAS G13E | p.G13E | COSM87280 | NEGATIVE | mnp |
| KRAS G12I | p.G12I | COSM34144 | NEGATIVE | mnp |
| KRAS G12D | p.G12D | COSM521 | NEGATIVE | SNV |
| KRAS G13R | p.G13R | COSM526 | NEGATIVE | mnp |
| KRAS A59S | p.A59S | COSM1235389 | NEGATIVE | SNV |
| KRAS Q61H | p.Q61H | COSM555 | NEGATIVE | SNV |
| KRAS A146G | p.A146G | COSM1360828 | NEGATIVE | SNV |
| MAP2K1 F53I | p.F53I | COSM3503329 | NEGATIVE | SNV |
| MAP2K1 F53S | p.F53S | ClinVar13350 | NEGATIVE | SNV |
| MAP2K1 Q56P | p.Q56P | COSM1235481 | NEGATIVE | SNV |
| MAP2K1 K57M | p.K57M | MAN124 | NEGATIVE | SNV |
| MAP2K1 V60E | p.V60E | COSM2257209 | NEGATIVE | SNV |
| MAP2K1 P124S | p.P124S | COSM235614 | NEGATIVE | SNV |
| MAP2K1 G128V | p.G128V | COSM1517232 | NEGATIVE | SNV |
| MAP2K1 E203K | p.E203K | COSM232755 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM555604 | NEGATIVE | SNV |
| MAP2K1 F53C | p.F53C | COSM1562837 | NEGATIVE | SNV |
| MAP2K1 K57E | p.K57E | COSM5369532 | NEGATIVE | SNV |
| MAP2K1 V60M | p.V60M | OM3576 | NEGATIVE | SNV |
| MAP2K1 P124Q | p.P124Q | COSM1167912 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | COSM1235480 | NEGATIVE | SNV |
| MAP2K1 L215P | p.L215P | OM3252 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MAP2K1 F53V | p.F53V | COSM5077832 | NEGATIVE | SNV |
| MAP2K1 F53Y | p.F53Y | COSM5588244 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM1725008 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM5520914 | NEGATIVE | SNV |
| MAP2K1 L115P | p.L115P | OM3248 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM555601 | NEGATIVE | SNV |
| MAP2K1 C121S | p.C121S | COSM1315829 | NEGATIVE | SNV |
| MAP2K1 G128D | p.G128D | COSM1374186 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3249 | NEGATIVE | SNV |
| MAP2K1 E203V | p.E203V | COSM3386991 | NEGATIVE | SNV |
| MAP2K1 F53L | p.F53L | COSM2257208 | NEGATIVE | SNV |
| MAP2K1 K57Q | p.K57Q | OM3155 | NEGATIVE | SNV |
| MAP2K1 P124L | p.P124L | COSM1315861 | NEGATIVE | SNV |
| MAP2K1 V211D | p.V211D | OM3251 | NEGATIVE | SNV |
| MAP2K1 F129L | p.F129L | OM3250 | NEGATIVE | SNV |
| MAP2K1 K57T | p.K57T | COSM4756761 | NEGATIVE | SNV |
| MAP2K1 K57N | p.K57N | COSM1235478 | NEGATIVE | SNV |
| MAP2K2 V35M | p.V35M | COSM5855836 | NEGATIVE | SNV |
| MAP2K2 C125S | p.C125S | COSM5855814 | NEGATIVE | SNV |
| MAP2K2 N126D | p.N126D | COSM1731742 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL803 | NEGATIVE | del |
| MET c.2942-29_2942-28d elinsGG | p.? | OM3305 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL708 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL674 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM49015 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL709 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL676 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL710 | NEGATIVE | del |
| MET c.2942-20_2942-19delinsAGAG | p.? | OMINDEL804 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM29636 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL677 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL712 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL679 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL680 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL682 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL683 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL684 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL685 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OM3178 | NEGATIVE | SNV |
| MET Y1021S | p.Y1021S | COSM7450124 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL809 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL811 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL687 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL812 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL813 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL668 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL694 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL715 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028H | COSM6438141 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL696 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6108461 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6923013 | NEGATIVE | ins |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL815 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6947995 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3217 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6108460 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OM3186 | NEGATIVE | SNV |
| MET exon 14 skipping ( | p.? | OM3188 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET H1112Y | p.H1112Y | COSM696 | NEGATIVE | SNV |
| MET H1112L | p.H1112L | COSM698 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6980941 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL675 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL640 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL681 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6976260 | NEGATIVE | complex |
| MET Y1021F | p.Y1021F | COSM339515 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6977526 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL656 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL662 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL814 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028IfsTer19 | COSM24687 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6940825 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL802 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6056832 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6930439 | NEGATIVE | del |
| MET exon 14 skipping ( | p.? | COSM6057022 | NEGATIVE | del |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| DNA) | | | | |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL642 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6948341 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL808 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL655 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL654 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL688 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL689 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6965225 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL692 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL693 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL695 | NEGATIVE | complex |
| MET H1112R | p.H1112R | COSM703 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL706 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6056834 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6983140 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM744044 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.A1023IfsTer19 | OMINDEL810 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| MET exon 14 skipping ( DNA) | p.? | OMINDEL686 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL690 | NEGATIVE | complex |
| MET exon 14 skipping ( DNA) | p.? | COSM6911117 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL670 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL691 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3177 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.D1028Y | COSM6917314 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL697 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OM3185 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM6947399 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | OMINDEL658 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM29635 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.? | COSM6057025 | NEGATIVE | del |
| MET exon 14 skipping ( DNA) | p.D1028N | COSM6924587 | NEGATIVE | SNV |
| MET c.2942-21_2942-20delinsGC | p.? | COSM8631602 | NEGATIVE | mnp |
| MET exon 14 skipping ( DNA) | p.? | COSM6928344 | NEGATIVE | SNV |
| MET exon 14 skipping ( DNA) | p.? | COSM24688 | NEGATIVE | del |
| NRAS A146V | p.A146V | COSM4170228 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS A146T | p.A146T | COSM27174 | NEGATIVE | SNV |
| NRAS [Q61H;E62K] | p.[Q61H;E62K] | COSM26494 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM33693 | NEGATIVE | mnp |
| NRAS Q61L | p.Q61L | COSM12725 | NEGATIVE | mnp |
| NRAS Q61K | p.Q61K | COSM580 | NEGATIVE | SNV |
| NRAS G13V | p.G13V | COSM572 | NEGATIVE | mnp |
| NRAS G13D | p.G13D | COSM573 | NEGATIVE | SNV |
| NRAS G13R | p.G13R | COSM569 | NEGATIVE | SNV |
| NRAS G12A | p.G12A | COSM565 | NEGATIVE | SNV |
| NRAS G12C | p.G12C | COSM562 | NEGATIVE | SNV |
| NRAS K117N | p.K117N | MAN13 | NEGATIVE | SNV |
| NRAS Q61H | p.Q61H | COSM586 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM30646 | NEGATIVE | mnp |
| NRAS Q61R | p.Q61R | COSM579 | NEGATIVE | mnp |
| NRAS A59D | p.A59D | COSM253327 | NEGATIVE | SNV |
| NRAS G13Y | p.G13Y | COSM568 | NEGATIVE | mnp |
| NRAS G12E | p.G12E | COSM144577 | NEGATIVE | mnp |
| NRAS G12Y | p.G12Y | COSM560 | NEGATIVE | mnp |
| NRAS Q61H | p.Q61H | COSM585 | NEGATIVE | SNV |
| NRAS Q61L | p.Q61L | COSM583 | NEGATIVE | SNV |
| NRAS Q61E | p.Q61E | COSM581 | NEGATIVE | SNV |
| NRAS [G60=;Q61K] | p.[G60=;Q61K] | COSM12730 | NEGATIVE | mnp |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NRAS G13V | p.G13V | COSM574 | NEGATIVE | SNV |
| NRAS G13A | p.G13A | COSM575 | NEGATIVE | SNV |
| NRAS G13C | p.G13C | COSM570 | NEGATIVE | SNV |
| NRAS G12V | p.G12V | COSM566 | NEGATIVE | SNV |
| NRAS G12P | p.G12P | COSM559 | NEGATIVE | mnp |
| NRAS G12R | p.G12R | COSM561 | NEGATIVE | SNV |
| NRAS A59T | p.A59T | COSM578 | NEGATIVE | SNV |
| NRAS G13N | p.G13N | COSM24668 | NEGATIVE | mnp |
| NRAS G13S | p.G13S | COSM571 | NEGATIVE | SNV |
| NRAS G12N | p.G12N | COSM12723 | NEGATIVE | mnp |
| NRAS Q61P | p.Q61P | COSM582 | NEGATIVE | SNV |
| NRAS G12S | p.G12S | COSM563 | NEGATIVE | SNV |
| NRAS G12D | p.G12D | COSM564 | NEGATIVE | SNV |
| NRAS Q61R | p.Q61R | COSM584 | NEGATIVE | SNV |
| NRAS Q61K | p.Q61K | COSM53223 | NEGATIVE | mnp |
| NTRK1 F589L | p.F589L | BT106 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT108 | NEGATIVE | SNV |
| NTRK1 G667C | p.G667C | BT111 | NEGATIVE | SNV |
| NTRK1 V573M | p.V573M | BT104 | NEGATIVE | SNV |
| NTRK1 F589C | p.F589C | OM3255 | NEGATIVE | SNV |
| NTRK1 F589L | p.F589L | BT107 | NEGATIVE | SNV |
| NTRK1 G667S | p.G667S | BT112 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK1 G667A | p.G667A | OM3301 | NEGATIVE | SNV |
| NTRK1 G595R | p.G595R | BT110 | NEGATIVE | SNV |
| NTRK1 F589Y | p.F589Y | BT105 | NEGATIVE | SNV |
| NTRK2 V617M | p.V617M | OM3274 | NEGATIVE | mnp |
| NTRK2 F633L | p.F633L | OM3277 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3309 | NEGATIVE | SNV |
| NTRK2 G709A | p.G709A | OM3310 | NEGATIVE | SNV |
| NTRK2 Q612P | p.Q612P | OM3273 | NEGATIVE | SNV |
| NTRK2 F633Y | p.F633Y | OM3307 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3280 | NEGATIVE | SNV |
| NTRK2 F633I | p.F633I | OM3276 | NEGATIVE | SNV |
| NTRK2 F633C | p.F633C | OM3278 | NEGATIVE | SNV |
| NTRK2 G639R | p.G639R | OM3279 | NEGATIVE | SNV |
| NTRK2 R646K | p.R646K | OM3281 | NEGATIVE | SNV |
| NTRK2 Q612E | p.Q612E | OM3272 | NEGATIVE | SNV |
| NTRK2 F633L | p.F633L | OM3308 | NEGATIVE | SNV |
| NTRK2 V617D | p.V617D | OM3306 | NEGATIVE | SNV |
| NTRK2 V617G | p.V617G | OM3275 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3294 | NEGATIVE | mnp |
| NTRK3 G623R | p.G623R | OM3253 | NEGATIVE | SNV |
| NTRK3 F617Y | p.F617Y | OM3311 | NEGATIVE | SNV |
| NTRK3 G623R | p.G623R | COSM6951362 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| NTRK3 V601F | p.V601F | OM3312 | NEGATIVE | SNV |
| NTRK3 G696E | p.G696E | COSM5850004 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3297 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3299 | NEGATIVE | SNV |
| NTRK3 G696C | p.G696C | OM3295 | NEGATIVE | mnp |
| NTRK3 G696A | p.G696A | OM3296 | NEGATIVE | SNV |
| NTRK3 F617L | p.F617L | OM3298 | NEGATIVE | SNV |
| NTRK3 V601M | p.V601M | OM3300 | NEGATIVE | mnp |
| PDGFRA A530_I543dup | p.A530_I543dup | COSM7002381 | NEGATIVE | ins |
| PDGFRA V561D | p.V561D | COSM739 | NEGATIVE | SNV |
| PDGFRA D568N | p.D568N | OM3225 | NEGATIVE | SNV |
| PDGFRA H650Q | p.H650Q | OM3229 | NEGATIVE | SNV |
| PDGFRA N659S | p.N659S | COSM6960333 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM96895 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsN | COSM12411 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_D846delinsS | COSM12409 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsY | COSM96896 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_I843delinsV | COSM405285 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_D846delinsG | COSM12412 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.D842_H845delinsV | COSM12414 | NEGATIVE | complex |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.D842_D846delinsE | COSM12408 | NEGATIVE | complex |
| PDGFRA exon 18 deletion | p.I843_S847delinsT | COSM12407 | NEGATIVE | del |
| PDGFRA Y849S | p.Y849S | OM3234 | NEGATIVE | SNV |
| PDGFRA V561A | p.V561A | COSM1430077 | NEGATIVE | SNV |
| PDGFRA Q579R | p.Q579R | OM3226 | NEGATIVE | SNV |
| PDGFRA N659Y | p.N659Y | COSM22416 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842del | COSM12406 | NEGATIVE | del |
| PDGFRA exon 18 deletion | p.D842_S847delinsRI | OMINDEL900 | NEGATIVE | complex |
| PDGFRA D842Y | p.D842Y | COSM12396 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM12400 | NEGATIVE | del |
| PDGFRA Y849C | p.Y849C | COSM94952 | NEGATIVE | SNV |
| PDGFRA S566_E571delinsR | p.S566_E571delinsR | COSM12418 | NEGATIVE | del |
| PDGFRA H650Q | p.H650Q | OM3228 | NEGATIVE | SNV |
| PDGFRA N659H | p.N659H | COSM51516 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22415 | NEGATIVE | SNV |
| PDGFRA N659K | p.N659K | COSM22414 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.I843_D846del | COSM737 | NEGATIVE | del |
| PDGFRA D842I | p.D842I | COSM12398 | NEGATIVE | mnp |
| PDGFRA D842V | p.D842V | COSM736 | NEGATIVE | SNV |
| PDGFRA D842Y | p.D842Y | COSM12397 | NEGATIVE | mnp |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PDGFRA exon 18 deletion | p.I843del | COSM4969655 | NEGATIVE | del |
| PDGFRA G853D | p.G853D | OM3235 | NEGATIVE | SNV |
| PDGFRA W559_R560del | p.W559_R560del | OMINDEL700 | NEGATIVE | del |
| PDGFRA P577S | p.P577S | COSM6438084 | NEGATIVE | SNV |
| PDGFRA N659R | p.N659R | OM3230 | NEGATIVE | mnp |
| PDGFRA R841K | p.R841K | OM3233 | NEGATIVE | SNV |
| PDGFRA D842F | p.D842F | COSM29007 | NEGATIVE | mnp |
| PDGFRA exon 18 deletion | p.D842_S847delinsEA | COSM12415 | NEGATIVE | complex |
| PDGFRA V536E | p.V536E | COSM43059 | NEGATIVE | SNV |
| PDGFRA Y555C | p.Y555C | COSM133645 | NEGATIVE | SNV |
| PDGFRA D846Y | p.D846Y | COSM12399 | NEGATIVE | SNV |
| PDGFRA D842H | p.D842H | COSM734174 | NEGATIVE | SNV |
| PDGFRA N848K | p.N848K | COSM28052 | NEGATIVE | SNV |
| PDGFRA H845Y | p.H845Y | COSM96893 | NEGATIVE | SNV |
| PDGFRA exon 18 deletion | p.D842_M844del | COSM12401 | NEGATIVE | del |
| PIK3CA R38G | p.R38G | COSM40945 | NEGATIVE | SNV |
| PIK3CA E39K | p.E39K | COSM30625 | NEGATIVE | SNV |
| PIK3CA E81A | p.E81A | COSM5347052 | NEGATIVE | SNV |
| PIK3CA G106R | p.G106R | COSM86046 | NEGATIVE | SNV |
| PIK3CA R108H | p.R108H | COSM27497 | NEGATIVE | SNV |
| PIK3CA E109_I112delinsD | p.E109_I112delinsD | COSM445995 | NEGATIVE | del |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA K111N | p.K111N | COSM27505 | NEGATIVE | SNV |
| PIK3CA V344A | p.V344A | COSM86951 | NEGATIVE | SNV |
| PIK3CA N345T | p.N345T | COSM446002 | NEGATIVE | SNV |
| PIK3CA E418K | p.E418K | COSM12582 | NEGATIVE | SNV |
| PIK3CA D538G | p.D538G | COSM29123 | NEGATIVE | SNV |
| PIK3CA E542K | p.E542K | COSM760 | NEGATIVE | SNV |
| PIK3CA E542G | p.E542G | COSM761 | NEGATIVE | SNV |
| PIK3CA E545A | p.E545A | COSM12458 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM765 | NEGATIVE | SNV |
| PIK3CA Q546R | p.Q546R | COSM12459 | NEGATIVE | SNV |
| PIK3CA E547K | p.E547K | COSM29315 | NEGATIVE | SNV |
| PIK3CA Y1021N | p.Y1021N | COSM6143 | NEGATIVE | SNV |
| PIK3CA T1025P | p.T1025P | COSM6023925 | NEGATIVE | SNV |
| PIK3CA T1025I | p.T1025I | COSM36285 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM29313 | NEGATIVE | SNV |
| PIK3CA N1044Y | p.N1044Y | COSM36288 | NEGATIVE | SNV |
| PIK3CA D1045N | p.D1045N | COSM25086 | NEGATIVE | SNV |
| PIK3CA A1046E | p.A1046E | COSM27156 | NEGATIVE | SNV |
| PIK3CA H1047P | p.H1047P | COSM249874 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM24714 | NEGATIVE | SNV |
| PIK3CA H1048R | p.H1048R | COSM36289 | NEGATIVE | SNV |
| PIK3CA G1049R | p.G1049R | COSM12597 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38H | p.R38H | COSM745 | NEGATIVE | SNV |
| PIK3CA E81K | p.E81K | COSM27502 | NEGATIVE | SNV |
| PIK3CA G106A | p.G106A | COSM166339 | NEGATIVE | SNV |
| PIK3CA R108L | p.R108L | COSM729828 | NEGATIVE | SNV |
| PIK3CA K111N | p.K111N | COSM12580 | NEGATIVE | SNV |
| PIK3CA V344G | p.V344G | COSM22540 | NEGATIVE | SNV |
| PIK3CA N345I | p.N345I | COSM94978 | NEGATIVE | SNV |
| PIK3CA D538Y | p.D538Y | COSM1716811 | NEGATIVE | SNV |
| PIK3CA E542R | p.E542R | COSM1659152 | NEGATIVE | mnp |
| PIK3CA E545K | p.E545K | COSM763 | NEGATIVE | SNV |
| PIK3CA E545D | p.E545D | COSM27374 | NEGATIVE | SNV |
| PIK3CA Q546L | p.Q546L | COSM25041 | NEGATIVE | SNV |
| PIK3CA E547D | p.E547D | COSM729819 | NEGATIVE | SNV |
| PIK3CA Y1021F | p.Y1021F | COSM328025 | NEGATIVE | SNV |
| PIK3CA M1043V | p.M1043V | COSM12591 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM773 | NEGATIVE | SNV |
| PIK3CA N1044K | p.N1044K | COSM27504 | NEGATIVE | SNV |
| PIK3CA A1046V | p.A1046V | COSM6146 | NEGATIVE | SNV |
| PIK3CA H1047L | p.H1047L | COSM776 | NEGATIVE | SNV |
| PIK3CA H1048Y | p.H1048Y | COSM249875 | NEGATIVE | SNV |
| PIK3CA G1049C | p.G1049C | COSM1168056 | NEGATIVE | SNV |
| PIK3CA R38C | p.R38C | COSM744 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA R38L | p.R38L | COSM729833 | NEGATIVE | SNV |
| PIK3CA E39V | p.E39V | COSM1420731 | NEGATIVE | SNV |
| PIK3CA P104T | p.P104T | COSM1666660 | NEGATIVE | SNV |
| PIK3CA P104L | p.P104L | COSM163485 | NEGATIVE | SNV |
| PIK3CA G106V | p.G106V | COSM748 | NEGATIVE | SNV |
| PIK3CA R108P | p.R108P | COSM749 | NEGATIVE | SNV |
| PIK3CA G118D | p.G118D | COSM751 | NEGATIVE | SNV |
| PIK3CA N345D | p.N345D | COSM1420788 | NEGATIVE | SNV |
| PIK3CA D538N | p.D538N | COSM21467 | NEGATIVE | SNV |
| PIK3CA P539R | p.P539R | COSM759 | NEGATIVE | SNV |
| PIK3CA E542Q | p.E542Q | COSM17442 | NEGATIVE | SNV |
| PIK3CA E542V | p.E542V | COSM762 | NEGATIVE | SNV |
| PIK3CA E545Q | p.E545Q | COSM27133 | NEGATIVE | SNV |
| PIK3CA E545V | p.E545V | COSM27155 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM5881064 | NEGATIVE | SNV |
| PIK3CA Q546H | p.Q546H | COSM24712 | NEGATIVE | SNV |
| PIK3CA D1017H | p.D1017H | COSM27377 | NEGATIVE | SNV |
| PIK3CA Y1021H | p.Y1021H | COSM17444 | NEGATIVE | SNV |
| PIK3CA T1025A | p.T1025A | COSM771 | NEGATIVE | SNV |
| PIK3CA M1043L | p.M1043L | COSM5731063 | NEGATIVE | SNV |
| PIK3CA M1043T | p.M1043T | COSM12463 | NEGATIVE | SNV |
| PIK3CA N1044D | p.N1044D | COSM27134 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA N1044K | p.N1044K | COSM12592 | NEGATIVE | SNV |
| PIK3CA D1045G | p.D1045G | COSM1421003 | NEGATIVE | SNV |
| PIK3CA D1045V | p.D1045V | COSM729813 | NEGATIVE | SNV |
| PIK3CA H1047N | p.H1047N | COSM5029128 | NEGATIVE | SNV |
| PIK3CA H1047Q | p.H1047Q | COSM1041525 | NEGATIVE | SNV |
| PIK3CA H1048Q | p.H1048Q | COSM27157 | NEGATIVE | SNV |
| PIK3CA G1049S | p.G1049S | COSM777 | NEGATIVE | SNV |
| PIK3CA G1049A | p.G1049A | COSM27158 | NEGATIVE | SNV |
| PIK3CA E39G | p.E39G | COSM1041451 | NEGATIVE | SNV |
| PIK3CA P104R | p.P104R | COSM747 | NEGATIVE | SNV |
| PIK3CA K111E | p.K111E | COSM13570 | NEGATIVE | SNV |
| PIK3CA V344E | p.V344E | COSM6746438 | NEGATIVE | SNV |
| PIK3CA C420R | p.C420R | COSM757 | NEGATIVE | SNV |
| PIK3CA P539S | p.P539S | COSM249880 | NEGATIVE | SNV |
| PIK3CA E545G | p.E545G | COSM764 | NEGATIVE | SNV |
| PIK3CA Q546K | p.Q546K | COSM766 | NEGATIVE | SNV |
| PIK3CA Q546P | p.Q546P | COSM767 | NEGATIVE | SNV |
| PIK3CA E547G | p.E547G | COSM1420864 | NEGATIVE | SNV |
| PIK3CA T1025S | p.T1025S | COSM12590 | NEGATIVE | SNV |
| PIK3CA A1046T | p.A1046T | COSM27273 | NEGATIVE | SNV |
| PIK3CA M1043I | p.M1043I | COSM94984 | NEGATIVE | SNV |
| PIK3CA H1047Y | p.H1047Y | COSM774 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PIK3CA H1048N | p.H1048N | COSM1421010 | NEGATIVE | SNV |
| PIK3CA N345H | p.N345H | COSM3846786 | NEGATIVE | SNV |
| PIK3CA H1047R | p.H1047R | COSM775 | NEGATIVE | SNV |
| PIK3CA R38S | p.R38S | COSM87310 | NEGATIVE | SNV |
| PIK3CA H1048L | p.H1048L | COSM3734761 | NEGATIVE | SNV |
| PIK3CA N345S | p.N345S | COSM13476 | NEGATIVE | SNV |
| PIK3CA Y1021C | p.Y1021C | COSM12461 | NEGATIVE | SNV |
| PIK3CA E110del | p.E110del | COSM24710 | NEGATIVE | del |
| PIK3CA K111R | p.K111R | COSM14052 | NEGATIVE | SNV |
| PIK3CA H1048D | p.H1048D | COSM249881 | NEGATIVE | SNV |
| PIK3CA N345K | p.N345K | COSM754 | NEGATIVE | SNV |
| PIK3CA G1049D | p.G1049D | COSM308548 | NEGATIVE | SNV |
| PIK3CA V344M | p.V344M | COSM253279 | NEGATIVE | SNV |
| PIK3CA T1025N | p.T1025N | COSM772 | NEGATIVE | SNV |
| PIK3CA N1044S | p.N1044S | COSM22541 | NEGATIVE | SNV |
| PIK3CA E542A | p.E542A | COSM1041494 | NEGATIVE | SNV |
| PIK3CA P539T | p.P539T | COSM41779 | NEGATIVE | SNV |
| PIK3CA Q546E | p.Q546E | COSM6147 | NEGATIVE | SNV |
| PTEN D24Y | p.D24Y | COSM5237 | NEGATIVE | SNV |
| PTEN D24G | p.D24G | COSM5107 | NEGATIVE | SNV |
| PTEN D24E | p.D24E | COSM5198 | NEGATIVE | SNV |
| PTEN A126T | p.A126T | COSM5051 | NEGATIVE | SNV |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN A126G | p.A126G | COSM4391544 | NEGATIVE | SNV |
| PTEN G129E | p.G129E | COSM28917 | NEGATIVE | SNV |
| PTEN R130Q | p.R130Q | COSM5033 | NEGATIVE | SNV |
| PTEN V133I | p.V133I | COSM5044 | NEGATIVE | SNV |
| PTEN Y155C | p.Y155C | COSM5144 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | COSM6006454 | NEGATIVE | SNV |
| PTEN R173H | p.R173H | COSM5039 | NEGATIVE | SNV |
| PTEN R173P | p.R173P | COSM12735 | NEGATIVE | SNV |
| PTEN F341V | p.F341V | COSM5255 | NEGATIVE | SNV |
| PTEN L345Q | p.L345Q | COSM5213 | NEGATIVE | SNV |
| PTEN N48K | p.N48K | COSM4392678 | NEGATIVE | SNV |
| PTEN A126S | p.A126S | COSM28889 | NEGATIVE | SNV |
| PTEN G129R | p.G129R | COSM246853 | NEGATIVE | SNV |
| PTEN R130P | p.R130P | COSM5277 | NEGATIVE | SNV |
| PTEN C136Y | p.C136Y | COSM12734 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH21 | NEGATIVE | SNV |
| PTEN Y174C | p.Y174C | COSM5575601 | NEGATIVE | SNV |
| PTEN D24H | p.D24H | COSM1603823 | NEGATIVE | SNV |
| PTEN A126D | p.A126D | COSM28884 | NEGATIVE | SNV |
| PTEN A126V | p.A126V | COSM5041 | NEGATIVE | SNV |
| PTEN G129V | p.G129V | COSM5276 | NEGATIVE | SNV |
| PTEN R130G | p.R130G | COSM5219 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| PTEN R130L | p.R130L | COSM5216 | NEGATIVE | SNV |
| PTEN T131I | p.T131I | COSM5104 | NEGATIVE | SNV |
| PTEN C136F | p.C136F | COSM5228 | NEGATIVE | SNV |
| PTEN S170R | p.S170R | MCH20 | NEGATIVE | SNV |
| PTEN Y174N | p.Y174N | COSM5221 | NEGATIVE | SNV |
| PTEN L181P | p.L181P | MCH17 | NEGATIVE | SNV |
| PTEN C71Y | p.C71Y | COSM5102 | NEGATIVE | SNV |
| PTEN G127E | p.G127E | COSM5143 | NEGATIVE | SNV |
| PTEN R130* | p.R130* | COSM5152 | NEGATIVE | SNV |
| PTEN D24N | p.D24N | COSM5079 | NEGATIVE | SNV |
| PTEN A126P | p.A126P | COSM5211 | NEGATIVE | SNV |
| PTEN V343E | p.V343E | MCH18 | NEGATIVE | SNV |
| PTEN S170N | p.S170N | COSM5045 | NEGATIVE | SNV |
| PTEN Y16C | p.Y16C | COSM5133 | NEGATIVE | SNV |
| PTEN R173C | p.R173C | COSM5089 | NEGATIVE | SNV |
| PTEN D24V | p.D24V | COSM4016448 | NEGATIVE | SNV |
| PTEN C136R | p.C136R | COSM5096 | NEGATIVE | SNV |
| RAF1 S259P | p.S259P | COSM1418555 | NEGATIVE | SNV |
| RAF1 S257P | p.S257P | ClinVar40600 | NEGATIVE | SNV |
| RAF1 S259T | p.S259T | ClinVar40601 | NEGATIVE | SNV |
| RAF1 S259F | p.S259F | COSM1536534 | NEGATIVE | SNV |
| RAF1 S257L | p.S257L | COSM181063 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RAF1 S259A | p.S259A | COSM20741 | NEGATIVE | SNV |
| RAF1 S259C | p.S259C | COSM5719965 | NEGATIVE | SNV |
| RAF1 S257W | p.S257W | COSM581519 | NEGATIVE | SNV |
| RET C618G | p.C618G | COSM9214400 | NEGATIVE | SNV |
| RET C618W | p.C618W | OM3403 | NEGATIVE | SNV |
| RET C620Y | p.C620Y | COSM7403807 | NEGATIVE | SNV |
| RET c.1880_1899delinsCTCAC | p.? | OMINDEL1170 | NEGATIVE | complex |
| RET C630G | p.C630G | COSM29806 | NEGATIVE | SNV |
| RET C634_R635insHELC | p.C634_R635insHELC | OMINDEL1174 | NEGATIVE | ins |
| RET D631_L633delinsV | p.D631_L633delinsV | OMINDEL1175 | NEGATIVE | del |
| RET E632_C634dup | p.E632_C634dup | OMINDEL1176 | NEGATIVE | ins |
| RET D631_L633delinsE | p.D631_L633delinsE | COSM983 | NEGATIVE | del |
| RET E632_T636delinsSS | p.E632_T636delinsSS | COSM1048 | NEGATIVE | complex |
| RET E632K | p.E632K | COSM1702268 | NEGATIVE | SNV |
| RET E632_L633delinsV | p.E632_L633delinsV | COSM982 | NEGATIVE | del |
| RET [E632D;L633V;C634R] | p.[E632D;L633V;C634R] | OM3407 | NEGATIVE | mnp |
| RET C634_R635dup | p.C634_R635dup | OMINDEL1177 | NEGATIVE | ins |
| RET C634Y | p.C634Y | COSM974 | NEGATIVE | SNV |
| RET C634W | p.C634W | COSM975 | NEGATIVE | SNV |
| RET L760Q | p.L760Q | OM3412 | NEGATIVE | SNV |
| RET E768D | p.E768D | OM3413 | NEGATIVE | SNV |
| RET R770Q | p.R770Q | COSM6006462 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET I788N | p.I788N | OM3284 | NEGATIVE | SNV |
| RET V804M | p.V804M | OM3195 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3285 | NEGATIVE | SNV |
| RET V804L | p.V804L | OM3286 | NEGATIVE | SNV |
| RET Y806C | p.Y806C | OM3287 | NEGATIVE | SNV |
| RET G810C | p.G810C | OM3557 | NEGATIVE | SNV |
| RET [V882=;A883F] | p.[V882=;A883F] | COSM981 | NEGATIVE | mnp |
| RET A883F | p.A883F | COSM977 | NEGATIVE | mnp |
| RET R886W | p.R886W | COSM6942691 | NEGATIVE | SNV |
| RET V899_D903del | p.V899_D903del | COSM5991595 | NEGATIVE | del |
| RET S904F | p.S904F | COSM6438204 | NEGATIVE | SNV |
| RET C618R | p.C618R | COSM29803 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM5946160 | NEGATIVE | SNV |
| RET C620F | p.C620F | OM3405 | NEGATIVE | SNV |
| RET D631_C634del | p.D631_C634del | OMINDEL1172 | NEGATIVE | del |
| RET C630R | p.C630R | COSM964 | NEGATIVE | SNV |
| RET D631del | p.D631del | OMINDEL1173 | NEGATIVE | del |
| RET C630F | p.C630F | OM3406 | NEGATIVE | SNV |
| RET E632_L633del | p.E632_L633del | COSM968 | NEGATIVE | del |
| RET E632_C634delinsG | p.E632_C634delinsG | COSM973 | NEGATIVE | del |
| RET C634R | p.C634R | COSM966 | NEGATIVE | SNV |
| RET C634F | p.C634F | COSM1237919 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET G736A | p.G736A | OM3411 | NEGATIVE | SNV |
| RET V804G | p.V804G | OM3554 | NEGATIVE | SNV |
| RET Y806F | p.Y806F | OM3556 | NEGATIVE | SNV |
| RET G810R | p.G810R | OM3289 | NEGATIVE | SNV |
| RET A883S | p.A883S | COSM133167 | NEGATIVE | SNV |
| RET E884K | p.E884K | COSM960 | NEGATIVE | SNV |
| RET D898_E901del | p.D898_E901del | COSM962 | NEGATIVE | del |
| RET R912P | p.R912P | OM3419 | NEGATIVE | SNV |
| RET C618S | p.C618S | COSM87267 | NEGATIVE | SNV |
| RET C618S | p.C618S | OM3402 | NEGATIVE | SNV |
| RET C618F | p.C618F | OM3401 | NEGATIVE | SNV |
| RET C620R | p.C620R | COSM29804 | NEGATIVE | SNV |
| RET C620S | p.C620S | COSM29805 | NEGATIVE | SNV |
| RET P628_L633del | p.P628_L633del | OMINDEL1171 | NEGATIVE | del |
| RET D631Y | p.D631Y | COSM1738370 | NEGATIVE | SNV |
| RET C634_T636dup | p.C634_T636dup | COSM9213763 | NEGATIVE | ins |
| RET C634G | p.C634G | COSM1738369 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1666664 | NEGATIVE | SNV |
| RET L730Q | p.L730Q | OM3409 | NEGATIVE | SNV |
| RET E768G | p.E768G | COSM1347811 | NEGATIVE | SNV |
| RET E768D | p.E768D | COSM21338 | NEGATIVE | SNV |
| RET G810A | p.G810A | OM3290 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET A883T | p.A883T | COSM100081 | NEGATIVE | SNV |
| RET A883Y | p.A883Y | OM3415 | NEGATIVE | mnp |
| RET S891A | p.S891A | rs75234356 | NEGATIVE | SNV |
| RET S904C | p.S904C | OM3418 | NEGATIVE | SNV |
| RET R912Q | p.R912Q | COSM3437793 | NEGATIVE | SNV |
| RET C618Y | p.C618Y | COSM980 | NEGATIVE | SNV |
| RET C620G | p.C620G | OM3404 | NEGATIVE | SNV |
| RET C630Y | p.C630Y | COSM9233214 | NEGATIVE | SNV |
| RET E768Q | p.E768Q | COSM1716312 | NEGATIVE | SNV |
| RET C634L | p.C634L | OM3408 | NEGATIVE | mnp |
| RET G810S | p.G810S | OM3288 | NEGATIVE | SNV |
| RET L881V | p.L881V | OM3414 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3417 | NEGATIVE | mnp |
| RET A883V | p.A883V | COSM4594153 | NEGATIVE | SNV |
| RET L730R | p.L730R | OM3410 | NEGATIVE | SNV |
| RET C618T | p.C618T | OM3400 | NEGATIVE | mnp |
| RET R897* | p.R897* | COSM6983129 | NEGATIVE | SNV |
| RET C634S | p.C634S | COSM1237918 | NEGATIVE | SNV |
| RET R912L | p.R912L | COSM188545 | NEGATIVE | SNV |
| RET [D903E;S904P] | p.[D903E;S904P] | OM3416 | NEGATIVE | mnp |
| RET G911D | p.G911D | COSM20888 | NEGATIVE | SNV |
| RET L790F | p.L790F | rs75030001 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| RET E884V | p.E884V | COSM1570338 | NEGATIVE | SNV |
| RET C620W | p.C620W | COSM249815 | NEGATIVE | SNV |
| RET G810V | p.G810V | OM3558 | NEGATIVE | SNV |
| ROS1 G2032K | p.G2032K | OM3533 | NEGATIVE | mnp |
| ROS1 S1986F | p.S1986F | COSM6929870 | NEGATIVE | SNV |
| ROS1 D2033N | p.D2033N | COSM6924065 | NEGATIVE | SNV |
| ROS1 S1986Y | p.S1986Y | OM3170 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN11 | NEGATIVE | SNV |
| ROS1 L1951M | p.L1951M | COSM1072523 | NEGATIVE | SNV |
| ROS1 G2032R | p.G2032R | MAN10 | NEGATIVE | SNV |
| ROS1 L1951R | p.L1951R | OM3264 | NEGATIVE | SNV |
| ROS1 L2026M | p.L2026M | BT151 | NEGATIVE | SNV |
| STK11 D53Gfs*110 | p.D53GfsTer110 | COSM391251 | NEGATIVE | ins |
| STK11 c.289_290+2delAAGT | p.? | OMINDEL1000 | NEGATIVE | del |
| STK11 c.290+1_290+2delGT | p.? | OMINDEL1001 | NEGATIVE | del |
| STK11 c.290+1G>A | p.? | COSM6350392 | NEGATIVE | SNV |
| STK11 c.290+2delT | p.? | OMINDEL1002 | NEGATIVE | del |
| STK11 c.290+2T>G | p.? | COSM25857 | NEGATIVE | SNV |
| STK11 c.291-2A>G | p.? | COSM5575722 | NEGATIVE | SNV |
| STK11 c.291-1G>A | p.? | OM3703 | NEGATIVE | SNV |
| STK11 c.291-1G>T | p.? | COSM6083779 | NEGATIVE | SNV |
| STK11 c.375-6_375-2delCCCCA | p.? | OMINDEL1005 | NEGATIVE | del |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.375-2A>G | p.? | OM3707 | NEGATIVE | SNV |
| STK11 c.375-1C>G | p.? | COSM4074282 | NEGATIVE | SNV |
| STK11 c.465-4_472delGC AGGTACTTCT | p.? | OMINDEL1007 | NEGATIVE | del |
| STK11 c.465-2A>C | p.? | COSM6933102 | NEGATIVE | SNV |
| STK11 c.465-2_465-1del insTC | p.? | OM3708 | NEGATIVE | mnp |
| STK11 c.465-1G>C | p.? | OM3709 | NEGATIVE | SNV |
| STK11 c.465-1G>T | p.? | COSM21570 | NEGATIVE | SNV |
| STK11 D194N | p.D194N | COSM25847 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3710 | NEGATIVE | SNV |
| STK11 c.598-1_612delGG CACTGCACCCGTTC | p.? | OMINDEL1014 | NEGATIVE | del |
| STK11 c.598-2A>G | p.? | COSM6923168 | NEGATIVE | SNV |
| STK11 c.598-1G>A | p.? | COSM3724562 | NEGATIVE | SNV |
| STK11 c.598-1_598delin sTT | p.? | OM3713 | NEGATIVE | mnp |
| STK11 c.727_734+9delGT CACCCTGTAAGTGCC | p.? | OMINDEL1015 | NEGATIVE | del |
| STK11 c.734+1G>C | p.? | COSM6970016 | NEGATIVE | SNV |
| STK11 c.734+2T>A | p.? | OM3715 | NEGATIVE | SNV |
| STK11 c.735-2A>G | p.? | OM3716 | NEGATIVE | SNV |
| STK11 c.735-1G>A | p.? | COSM5732040 | NEGATIVE | SNV |
| STK11 c.862+1G>A | p.? | COSM6929076 | NEGATIVE | SNV |
| STK11 c.862+1G>T | p.? | COSM6149637 | NEGATIVE | SNV |
| STK11 c.862+2T>A | p.? | COSM5576087 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.863-2A>C | p.? | COSM6912582 | NEGATIVE | SNV |
| STK11 c.863-1G>A | p.? | COSM6916725 | NEGATIVE | SNV |
| STK11 R304W | p.R304W | COSM29468 | NEGATIVE | SNV |
| STK11 c.918_920+11delC AGGTGAGCGGCCC | p.? | OMINDEL1019 | NEGATIVE | del |
| STK11 c.920+1G>C | p.? | COSM4412472 | NEGATIVE | SNV |
| STK11 c.920+2T>A | p.? | OM3721 | NEGATIVE | SNV |
| STK11 c.1108+1G>A | p.? | OM3722 | NEGATIVE | SNV |
| STK11 L67P | p.L67P | OM3700 | NEGATIVE | SNV |
| STK11 c.375-1C>T | p.? | COSM6977173 | NEGATIVE | SNV |
| STK11 c.465-1delG | p.? | OMINDEL1010 | NEGATIVE | del |
| STK11 D194Y | p.D194Y | COSM20944 | NEGATIVE | SNV |
| STK11 c.598-2_600delAG GCA | p.? | OMINDEL1012 | NEGATIVE | del |
| STK11 c.598-2A>C | p.? | OM3712 | NEGATIVE | SNV |
| STK11 c.734+2_734+10de lTAAGTGCCC | p.? | OMINDEL1017 | NEGATIVE | del |
| STK11 c.850_862+4delGA CCTGCTGAAAGGTGG | p.? | OMINDEL1018 | NEGATIVE | del |
| STK11 c.920+1G>A | p.? | OM3720 | NEGATIVE | SNV |
| STK11 c.375-2A>C | p.? | OM3706 | NEGATIVE | SNV |
| STK11 c.465-4_479delGC AGGTACTTCTGTCAGCT | p.? | OMINDEL1008 | NEGATIVE | del |
| STK11 c.465-2_465-1del AG | p.? | OMINDEL1009 | NEGATIVE | del |
| STK11 D194V | p.D194V | COSM20957 | NEGATIVE | SNV |
| STK11 c.734+1G>T | p.? | COSM51523 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 A205T | p.A205T | COSM20953 | NEGATIVE | SNV |
| STK11 S240W | p.S240W | OM3714 | NEGATIVE | SNV |
| STK11 c.863-1G>T | p.? | COSM6925330 | NEGATIVE | SNV |
| STK11 c.290+1G>C | p.? | OM3701 | NEGATIVE | SNV |
| STK11 c.290+2T>A | p.? | COSM6925006 | NEGATIVE | SNV |
| STK11 c.291-1delG | p.? | OMINDEL1004 | NEGATIVE | del |
| STK11 c.291-1G>C | p.? | OM3704 | NEGATIVE | SNV |
| STK11 c.291-1_291delinsTT | p.? | OM3705 | NEGATIVE | mnp |
| STK11 c.375-2A>T | p.? | COSM6967966 | NEGATIVE | SNV |
| STK11 c.465-2A>G | p.? | COSM21387 | NEGATIVE | SNV |
| STK11 D194E | p.D194E | OM3711 | NEGATIVE | SNV |
| STK11 c.598-2A>T | p.? | COSM25858 | NEGATIVE | SNV |
| STK11 c.598-1G>T | p.? | COSM51521 | NEGATIVE | SNV |
| STK11 c.734+2T>G | p.? | COSM6961577 | NEGATIVE | SNV |
| STK11 c.735-2A>T | p.? | COSM25856 | NEGATIVE | SNV |
| STK11 c.862+3delG | p.? | COSM8554973 | NEGATIVE | del |
| STK11 c.863-2A>T | p.? | COSM401786 | NEGATIVE | SNV |
| STK11 c.920+1G>T | p.? | COSM6923458 | NEGATIVE | SNV |
| STK11 c.598-1delG | p.? | OMINDEL1013 | NEGATIVE | del |
| STK11 c.734+1G>A | p.? | COSM6916684 | NEGATIVE | SNV |
| STK11 c.735-1G>T | p.? | COSM51522 | NEGATIVE | SNV |
| STK11 c.862+2T>G | p.? | OM3718 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| STK11 c.290+1G>T | p.? | OM3702 | NEGATIVE | SNV |
| STK11 c.731_734+9delCCCTGTAAGTGCC | p.? | OMINDEL1016 | NEGATIVE | del |
| STK11 c.465-1G>A | p.? | COSM25855 | NEGATIVE | SNV |
| STK11 L290P | p.L290P | OM3719 | NEGATIVE | SNV |
| STK11 c.465-1_465delinsTT | p.? | COSM6928792 | NEGATIVE | mnp |
| STK11 c.291-2A>T | p.? | COSM49010 | NEGATIVE | SNV |
| STK11 c.862+1G>C | p.? | OM3717 | NEGATIVE | SNV |
| STK11 c.465-2A>T | p.? | COSM1523962 | NEGATIVE | SNV |
| STK11 c.289_290+1delinsGAT | p.? | COSM6922318 | NEGATIVE | mnp |
| STK11 c.375-2_386delACGTATATGGTGAT | p.? | OMINDEL1006 | NEGATIVE | del |
| STK11 D176N | p.D176N | COSM4827690 | NEGATIVE | SNV |
| TP53 E286G | p.E286G | COSM43565 | NEGATIVE | SNV |
| TP53 E285V | p.E285V | COSM44227 | NEGATIVE | SNV |
| TP53 E285K | p.E285K | COSM10722 | NEGATIVE | SNV |
| TP53 R282Q | p.R282Q | COSM44338 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43906 | NEGATIVE | SNV |
| TP53 D281Y | p.D281Y | COSM11516 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44233 | NEGATIVE | SNV |
| TP53 R280G | p.R280G | COSM11123 | NEGATIVE | SNV |
| TP53 P278H | p.P278H | COSM43755 | NEGATIVE | SNV |
| TP53 C277F | p.C277F | COSM10749 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275Y | p.C275Y | COSM10893 | NEGATIVE | SNV |
| TP53 V274L | p.V274L | COSM44443 | NEGATIVE | SNV |
| TP53 R273C | p.R273C | COSM10659 | NEGATIVE | SNV |
| TP53 V272A | p.V272A | COSM44294 | NEGATIVE | SNV |
| TP53 V272M | p.V272M | COSM10891 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM11205 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM46410 | NEGATIVE | SNV |
| TP53 E258Q | p.E258Q | COSM10751 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10785 | NEGATIVE | SNV |
| TP53 R249W | p.R249W | COSM43629 | NEGATIVE | SNV |
| TP53 R248Q | p.R248Q | COSM10662 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM10757 | NEGATIVE | SNV |
| TP53 M246K | p.M246K | COSM44103 | NEGATIVE | SNV |
| TP53 G245A | p.G245A | COSM43965 | NEGATIVE | SNV |
| TP53 G245S | p.G245S | COSM6932 | NEGATIVE | SNV |
| TP53 G244C | p.G244C | COSM11524 | NEGATIVE | SNV |
| TP53 C242F | p.C242F | COSM10810 | NEGATIVE | SNV |
| TP53 C242R | p.C242R | COSM11738 | NEGATIVE | SNV |
| TP53 S241Y | p.S241Y | COSM10935 | NEGATIVE | SNV |
| TP53 S240G | p.S240G | COSM43973 | NEGATIVE | SNV |
| TP53 N239S | p.N239S | COSM44094 | NEGATIVE | SNV |
| TP53 C238W | p.C238W | COSM44676 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C238Y | p.C238Y | COSM11059 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM11063 | NEGATIVE | SNV |
| TP53 M237T | p.M237T | COSM45329 | NEGATIVE | SNV |
| TP53 Y236F | p.Y236F | COSM3958809 | NEGATIVE | SNV |
| TP53 Y236H | p.Y236H | COSM44326 | NEGATIVE | SNV |
| TP53 Y234S | p.Y234S | COSM43865 | NEGATIVE | SNV |
| TP53 Y220F | p.Y220F | COSM1564197 | NEGATIVE | SNV |
| TP53 Y220H | p.Y220H | COSM44637 | NEGATIVE | SNV |
| TP53 V216E | p.V216E | COSM44274 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM46272 | NEGATIVE | SNV |
| TP53 H214Y | p.H214Y | COSM44112 | NEGATIVE | SNV |
| TP53 R213Q | p.R213Q | COSM10735 | NEGATIVE | SNV |
| TP53 R213G | p.R213G | COSM44102 | NEGATIVE | SNV |
| TP53 V197M | p.V197M | COSM43779 | NEGATIVE | SNV |
| TP53 I195T | p.I195T | COSM11089 | NEGATIVE | SNV |
| TP53 L194R | p.L194R | COSM44571 | NEGATIVE | SNV |
| TP53 L194V | p.L194V | COSM46117 | NEGATIVE | SNV |
| TP53 H193P | p.H193P | COSM43833 | NEGATIVE | SNV |
| TP53 P191del | p.P191del | COSM45140 | NEGATIVE | del |
| TP53 H179Q | p.H179Q | COSM11249 | NEGATIVE | SNV |
| TP53 H179P | p.H179P | COSM44218 | NEGATIVE | SNV |
| TP53 C176W | p.C176W | COSM11114 | NEGATIVE | SNV |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C176G | p.C176G | COSM44692 | NEGATIVE | SNV |
| TP53 R175P | p.R175P | COSM45416 | NEGATIVE | SNV |
| TP53 R175S | p.R175S | COSM43931 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM43559 | NEGATIVE | SNV |
| TP53 V157F | p.V157F | COSM10670 | NEGATIVE | SNV |
| TP53 P152L | p.P152L | COSM10790 | NEGATIVE | SNV |
| TP53 P152A | p.P152A | COSM44788 | NEGATIVE | SNV |
| TP53 P151H | p.P151H | COSM11476 | NEGATIVE | SNV |
| TP53 L145P | p.L145P | COSM43899 | NEGATIVE | SNV |
| TP53 C141Y | p.C141Y | COSM43708 | NEGATIVE | SNV |
| TP53 A138V | p.A138V | COSM43818 | NEGATIVE | SNV |
| TP53 C135Y | p.C135Y | COSM10801 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM10991 | NEGATIVE | SNV |
| TP53 K132T | p.K132T | COSM43912 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45492 | NEGATIVE | SNV |
| TP53 E286Q | p.E286Q | COSM44250 | NEGATIVE | SNV |
| TP53 E285Q | p.E285Q | COSM45138 | NEGATIVE | SNV |
| TP53 R282W | p.R282W | COSM10704 | NEGATIVE | SNV |
| TP53 D281G | p.D281G | COSM11232 | NEGATIVE | SNV |
| TP53 R280S | p.R280S | COSM44171 | NEGATIVE | SNV |
| TP53 G279E | p.G279E | COSM43714 | NEGATIVE | SNV |
| TP53 P278A | p.P278A | COSM10814 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C275S | p.C275S | COSM45413 | NEGATIVE | SNV |
| TP53 R273L | p.R273L | COSM10779 | NEGATIVE | SNV |
| TP53 R273S | p.R273S | COSM43909 | NEGATIVE | SNV |
| TP53 V272L | p.V272L | COSM45898 | NEGATIVE | SNV |
| TP53 G266R | p.G266R | COSM10794 | NEGATIVE | SNV |
| TP53 E258G | p.E258G | COSM44168 | NEGATIVE | SNV |
| TP53 R249S | p.R249S | COSM10817 | NEGATIVE | SNV |
| TP53 R249G | p.R249G | COSM10668 | NEGATIVE | SNV |
| TP53 R248G | p.R248G | COSM11564 | NEGATIVE | SNV |
| TP53 M246T | p.M246T | COSM11355 | NEGATIVE | SNV |
| TP53 G245D | p.G245D | COSM43606 | NEGATIVE | SNV |
| TP53 G244A | p.G244A | COSM12013 | NEGATIVE | SNV |
| TP53 C242W | p.C242W | COSM11356 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM44935 | NEGATIVE | SNV |
| TP53 S241P | p.S241P | COSM44578 | NEGATIVE | SNV |
| TP53 N239I | p.N239I | COSM307333 | NEGATIVE | SNV |
| TP53 C238F | p.C238F | COSM43778 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM43700 | NEGATIVE | SNV |
| TP53 M237K | p.M237K | COSM43952 | NEGATIVE | SNV |
| TP53 Y236S | p.Y236S | COSM44693 | NEGATIVE | SNV |
| TP53 Y234C | p.Y234C | COSM10725 | NEGATIVE | SNV |
| TP53 Y220C | p.Y220C | COSM10758 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216G | p.V216G | COSM43681 | NEGATIVE | SNV |
| TP53 H214Q | p.H214Q | COSM44407 | NEGATIVE | SNV |
| TP53 H214D | p.H214D | COSM45115 | NEGATIVE | SNV |
| TP53 R213* | p.R213* | COSM10654 | NEGATIVE | SNV |
| TP53 R196* | p.R196* | COSM10705 | NEGATIVE | SNV |
| TP53 I195S | p.I195S | COSM44539 | NEGATIVE | SNV |
| TP53 L194P | p.L194P | COSM43827 | NEGATIVE | SNV |
| TP53 H193L | p.H193L | COSM11066 | NEGATIVE | SNV |
| TP53 H193N | p.H193N | COSM43935 | NEGATIVE | SNV |
| TP53 H179Q | p.H179Q | COSM44214 | NEGATIVE | SNV |
| TP53 H179D | p.H179D | COSM44776 | NEGATIVE | SNV |
| TP53 C176Y | p.C176Y | COSM10687 | NEGATIVE | SNV |
| TP53 R175H | p.R175H | COSM10648 | NEGATIVE | SNV |
| TP53 V173A | p.V173A | COSM44327 | NEGATIVE | SNV |
| TP53 V172F | p.V172F | COSM44240 | NEGATIVE | SNV |
| TP53 P152R | p.P152R | COSM45505 | NEGATIVE | SNV |
| TP53 P151L | p.P151L | COSM44288 | NEGATIVE | SNV |
| TP53 P151T | p.P151T | COSM43911 | NEGATIVE | SNV |
| TP53 C141G | p.C141G | COSM45794 | NEGATIVE | SNV |
| TP53 C135F | p.C135F | COSM10647 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44910 | NEGATIVE | SNV |
| TP53 K132E | p.K132E | COSM10813 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 E286V | p.E286V | COSM43936 | NEGATIVE | SNV |
| TP53 E286A | p.E286A | COSM45892 | NEGATIVE | SNV |
| TP53 E285G | p.E285G | COSM45649 | NEGATIVE | SNV |
| TP53 E285A | p.E285A | COSM43614 | NEGATIVE | SNV |
| TP53 R282L | p.R282L | COSM44470 | NEGATIVE | SNV |
| TP53 R282P | p.R282P | COSM44306 | NEGATIVE | SNV |
| TP53 D281E | p.D281E | COSM43837 | NEGATIVE | SNV |
| TP53 D281V | p.D281V | COSM45729 | NEGATIVE | SNV |
| TP53 D281H | p.D281H | COSM10943 | NEGATIVE | SNV |
| TP53 R280K | p.R280K | COSM10728 | NEGATIVE | SNV |
| TP53 P278L | p.P278L | COSM10863 | NEGATIVE | SNV |
| TP53 P278S | p.P278S | COSM10939 | NEGATIVE | SNV |
| TP53 P278T | p.P278T | COSM43697 | NEGATIVE | SNV |
| TP53 V274F | p.V274F | COSM10769 | NEGATIVE | SNV |
| TP53 R273P | p.R273P | COSM43896 | NEGATIVE | SNV |
| TP53 R273G | p.R273G | COSM43843 | NEGATIVE | SNV |
| TP53 G266V | p.G266V | COSM10958 | NEGATIVE | SNV |
| TP53 E258D | p.E258D | COSM44962 | NEGATIVE | SNV |
| TP53 E258V | p.E258V | COSM44450 | NEGATIVE | SNV |
| TP53 E258K | p.E258K | COSM10988 | NEGATIVE | SNV |
| TP53 R249K | p.R249K | COSM44091 | NEGATIVE | SNV |
| TP53 R248L | p.R248L | COSM6549 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 R248W | p.R248W | COSM10656 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM46136 | NEGATIVE | SNV |
| TP53 M246R | p.M246R | COSM11376 | NEGATIVE | SNV |
| TP53 M246L | p.M246L | COSM45992 | NEGATIVE | SNV |
| TP53 G245V | p.G245V | COSM11196 | NEGATIVE | SNV |
| TP53 G244D | p.G244D | COSM10883 | NEGATIVE | SNV |
| TP53 G244R | p.G244R | COSM44221 | NEGATIVE | SNV |
| TP53 C242S | p.C242S | COSM11133 | NEGATIVE | SNV |
| TP53 C242Y | p.C242Y | COSM10646 | NEGATIVE | SNV |
| TP53 S241F | p.S241F | COSM10812 | NEGATIVE | SNV |
| TP53 S241C | p.S241C | COSM10709 | NEGATIVE | SNV |
| TP53 S241T | p.S241T | COSM44067 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44510 | NEGATIVE | SNV |
| TP53 N239T | p.N239T | COSM43801 | NEGATIVE | SNV |
| TP53 N239Y | p.N239Y | COSM45870 | NEGATIVE | SNV |
| TP53 C238S | p.C238S | COSM44653 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM44415 | NEGATIVE | SNV |
| TP53 M237V | p.M237V | COSM44525 | NEGATIVE | SNV |
| TP53 Y236C | p.Y236C | COSM10731 | NEGATIVE | SNV |
| TP53 Y236N | p.Y236N | COSM43826 | NEGATIVE | SNV |
| TP53 Y234N | p.Y234N | COSM43956 | NEGATIVE | SNV |
| TP53 Y220N | p.Y220N | COSM44672 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V216A | p.V216A | COSM44567 | NEGATIVE | SNV |
| TP53 V216M | p.V216M | COSM10667 | NEGATIVE | SNV |
| TP53 H214L | p.H214L | COSM5632535 | NEGATIVE | SNV |
| TP53 H214P | p.H214P | COSM1640841 | NEGATIVE | SNV |
| TP53 I195F | p.I195F | COSM44633 | NEGATIVE | SNV |
| TP53 L194I | p.L194I | COSM45330 | NEGATIVE | SNV |
| TP53 H193R | p.H193R | COSM10742 | NEGATIVE | SNV |
| TP53 H193D | p.H193D | COSM44002 | NEGATIVE | SNV |
| TP53 Q192* | p.Q192* | COSM10733 | NEGATIVE | SNV |
| TP53 P177_C182del | p.P177_C182del | COSM43570 | NEGATIVE | del |
| TP53 H179L | p.H179L | COSM43635 | NEGATIVE | SNV |
| TP53 H179Y | p.H179Y | COSM10768 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44645 | NEGATIVE | SNV |
| TP53 R175L | p.R175L | COSM10718 | NEGATIVE | SNV |
| TP53 R175C | p.R175C | COSM43680 | NEGATIVE | SNV |
| TP53 V173G | p.V173G | COSM44383 | NEGATIVE | SNV |
| TP53 V173M | p.V173M | COSM11084 | NEGATIVE | SNV |
| TP53 R156P | p.R156P | COSM10760 | NEGATIVE | SNV |
| TP53 G154V | p.G154V | COSM6815 | NEGATIVE | SNV |
| TP53 P152Q | p.P152Q | COSM44613 | NEGATIVE | SNV |
| TP53 P152T | p.P152T | COSM44561 | NEGATIVE | SNV |
| TP53 C141W | p.C141W | COSM44204 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C141F | p.C141F | COSM44911 | NEGATIVE | SNV |
| TP53 C141R | p.C141R | COSM43901 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM44564 | NEGATIVE | SNV |
| TP53 C135S | p.C135S | COSM44643 | NEGATIVE | SNV |
| TP53 C135G | p.C135G | COSM44829 | NEGATIVE | SNV |
| TP53 K132N | p.K132N | COSM43963 | NEGATIVE | SNV |
| TP53 K132M | p.K132M | COSM43592 | NEGATIVE | SNV |
| TP53 K132Q | p.K132Q | COSM11224 | NEGATIVE | SNV |
| TP53 H179N | p.H179N | COSM44151 | NEGATIVE | SNV |
| TP53 R213L | p.R213L | COSM43650 | NEGATIVE | SNV |
| TP53 E286K | p.E286K | COSM10726 | NEGATIVE | SNV |
| TP53 R282G | p.R282G | COSM10992 | NEGATIVE | SNV |
| TP53 D281A | p.D281A | COSM11665 | NEGATIVE | SNV |
| TP53 R280I | p.R280I | COSM11287 | NEGATIVE | SNV |
| TP53 C275R | p.C275R | COSM43902 | NEGATIVE | SNV |
| TP53 R273H | p.R273H | COSM10660 | NEGATIVE | SNV |
| TP53 V272E | p.V272E | COSM44580 | NEGATIVE | SNV |
| TP53 G266E | p.G266E | COSM10867 | NEGATIVE | SNV |
| TP53 E258A | p.E258A | COSM44719 | NEGATIVE | SNV |
| TP53 R248P | p.R248P | COSM11491 | NEGATIVE | SNV |
| TP53 M246I | p.M246I | COSM44310 | NEGATIVE | SNV |
| TP53 G245C | p.G245C | COSM11081 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 G244V | p.G244V | COSM43652 | NEGATIVE | SNV |
| TP53 G244S | p.G244S | COSM10941 | NEGATIVE | SNV |
| TP53 S241A | p.S241A | COSM44224 | NEGATIVE | SNV |
| TP53 N239K | p.N239K | COSM44647 | NEGATIVE | SNV |
| TP53 C238R | p.C238R | COSM44321 | NEGATIVE | SNV |
| TP53 M237I | p.M237I | COSM10834 | NEGATIVE | SNV |
| TP53 Y234F | p.Y234F | COSM44315 | NEGATIVE | SNV |
| TP53 Y234H | p.Y234H | COSM11152 | NEGATIVE | SNV |
| TP53 Y220D | p.Y220D | COSM11847 | NEGATIVE | SNV |
| TP53 V216L | p.V216L | COSM11210 | NEGATIVE | SNV |
| TP53 H214R | p.H214R | COSM43687 | NEGATIVE | SNV |
| TP53 I195L | p.I195L | COSM45773 | NEGATIVE | SNV |
| TP53 L194F | p.L194F | COSM10995 | NEGATIVE | SNV |
| TP53 P190L | p.P190L | COSM43657 | NEGATIVE | SNV |
| TP53 C176F | p.C176F | COSM10645 | NEGATIVE | SNV |
| TP53 V173L | p.V173L | COSM44057 | NEGATIVE | SNV |
| TP53 P152S | p.P152S | COSM43582 | NEGATIVE | SNV |
| TP53 P151R | p.P151R | COSM44003 | NEGATIVE | SNV |
| TP53 P151A | p.P151A | COSM44944 | NEGATIVE | SNV |
| TP53 K132R | p.K132R | COSM11582 | NEGATIVE | SNV |
| TP53 C141S | p.C141S | COSM293910 | NEGATIVE | SNV |
| TP53 Y234D | p.Y234D | COSM43768 | NEGATIVE | SNV |

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| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 C242G | p.C242G | COSM44135 | NEGATIVE | SNV |
| TP53 I195N | p.I195N | COSM44877 | NEGATIVE | SNV |
| TP53 P278R | p.P278R | COSM10887 | NEGATIVE | SNV |
| TP53 H179R | p.H179R | COSM10889 | NEGATIVE | SNV |
| TP53 D281N | p.D281N | COSM43596 | NEGATIVE | SNV |
| TP53 M237R | p.M237R | COSM45032 | NEGATIVE | SNV |
| TP53 P250L | p.P250L | COSM10771 | NEGATIVE | SNV |
| TP53 G262V | p.G262V | COSM11198 | NEGATIVE | SNV |
| TP53 R175G | p.R175G | COSM10870 | NEGATIVE | SNV |
| TP53 H193Y | p.H193Y | COSM10672 | NEGATIVE | SNV |
| TP53 E286D | p.E286D | COSM45439 | NEGATIVE | SNV |
| TP53 C176R | p.C176R | COSM44948 | NEGATIVE | SNV |
| TP53 R213P | p.R213P | COSM11860 | NEGATIVE | SNV |
| TP53 M246V | p.M246V | COSM43555 | NEGATIVE | SNV |
| TP53 R249M | p.R249M | COSM43871 | NEGATIVE | SNV |
| TP53 C135W | p.C135W | COSM44219 | NEGATIVE | SNV |
| TP53 P151S | p.P151S | COSM10905 | NEGATIVE | SNV |
| TP53 M237L | p.M237L | COSM44965 | NEGATIVE | SNV |
| TP53 V272G | p.V272G | COSM44870 | NEGATIVE | SNV |
| TP53 I195M | p.I195M | COSM562637 | NEGATIVE | SNV |
| TP53 L194H | p.L194H | COSM43623 | NEGATIVE | SNV |
| TP53 V157L | p.V157L | COSM45120 | NEGATIVE | SNV |

| Variant Name | AA Change | Variant ID | Test Result | Variant Type |
|---|---|---|---|---|
| TP53 V272L | p.V272L | COSM10859 | NEGATIVE | SNV |
| TP53 C135R | p.C135R | COSM10684 | NEGATIVE | SNV |
| TP53 G266A | p.G266A | COSM45488 | NEGATIVE | SNV |
| TP53 C176S | p.C176S | COSM44146 | NEGATIVE | SNV |
| TP53 C275G | p.C275G | COSM11501 | NEGATIVE | SNV |
| TP53 N239D | p.N239D | COSM10777 | NEGATIVE | SNV |
| TP53 V173E | p.V173E | COSM45731 | NEGATIVE | SNV |
| TP53 R283P | p.R283P | COSM10743 | NEGATIVE | SNV |
| TP53 R280T | p.R280T | COSM10724 | NEGATIVE | SNV |
| TP53 R249T | p.R249T | COSM43665 | NEGATIVE | SNV |
| TP53 Y220S | p.Y220S | COSM43850 | NEGATIVE | SNV |
| TP53 C238G | p.C238G | COSM46336 | NEGATIVE | SNV |
| TP53 G245R | p.G245R | COSM10957 | NEGATIVE | SNV |
| TP53 C275W | p.C275W | COSM43823 | NEGATIVE | SNV |
| TP53 Y236D | p.Y236D | COSM43602 | NEGATIVE | SNV |

## Fusions

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYH10-ALK fusion | MYH10(38) - ALK(20) | Fusion | NEGATIVE |
| MPRIP-ALK fusion | MPRIP(21) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(30) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |
| CLTC-ALK fusion | CLTC(31) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PRKAR1A-ALK fusion | PRKAR1A(2) - ALK(20) | Fusion | NEGATIVE |
| PRKAR1A-ALK fusion | PRKAR1A(10) - ALK(20) | Fusion | NEGATIVE |
| RNF213-ALK fusion | RNF213(20) - ALK(20) | Fusion | NEGATIVE |
| KANK2-ALK fusion | KANK2(4) - ALK(16) | Fusion | NEGATIVE |
| BEND5-ALK fusion | BEND5(3) - ALK(20) | Fusion | NEGATIVE |
| TPM3-ALK fusion | TPM3(7) - ALK(20) | Fusion | NEGATIVE |
| LMNA-ALK fusion | LMNA(2) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(15) - ALK(20) | Fusion | NEGATIVE |
| TPR-ALK fusion | TPR(4) - ALK(20) | Fusion | NEGATIVE |
| CENPF-ALK fusion | CENPF(11) - ALK(20) | Fusion | NEGATIVE |
| MYT1L-ALK fusion | MYT1L(14) - ALK(20) | Fusion | NEGATIVE |
| ITSN2-ALK fusion | ITSN2(14) - ALK(2) | Fusion | NEGATIVE |
| NCOA1-ALK fusion | NCOA1(21) - ALK(1) | Fusion | NEGATIVE |
| OTOF-ALK fusion | OTOF(2) - ALK(3) | Fusion | NEGATIVE |
| MAPRE3-ALK fusion | MAPRE3(7) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(35) - ALK(20) | Fusion | NEGATIVE |
| CAD-ALK fusion | CAD(36) - ALK(20) | Fusion | NEGATIVE |
| PPM1G-ALK fusion | PPM1G(1) - ALK(18) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(5) - ALK(20) | Fusion | NEGATIVE |
| PPP1CB-ALK fusion | PPP1CB(7) - ALK(20) | Fusion | NEGATIVE |
| SPDYA-ALK fusion | SPDYA(6) - ALK(20) | Fusion | NEGATIVE |
| TRMT61B-ALK fusion | TRMT61B(1) - ALK(9) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP4-ALK fusion | CLIP4(10) - ALK(20) | Fusion | NEGATIVE |
| CLIP4-ALK fusion | CLIP4(14) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| YPEL5-ALK fusion | YPEL5(1) - ALK(18) | Fusion | NEGATIVE |
| GALNT14-ALK fusion | GALNT14(1) - ALK(19) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(2) - ALK(7) | Fusion | NEGATIVE |
| MEMO1-ALK fusion | MEMO1(1) - ALK(2) | Fusion | NEGATIVE |
| BIRC6-ALK fusion | BIRC6(10) - ALK(20) | Fusion | NEGATIVE |
| LTBP1-ALK fusion | LTBP1(19) - ALK(20) | Fusion | NEGATIVE |
| STRN-ALK fusion | STRN(3) - ALK(20) | Fusion | NEGATIVE |
| SLC8A1-ALK fusion | SLC8A1(1) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(2) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(4) - ALK(13) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(16) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(18) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(3) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(19) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(6) - ALK(17) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(7) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(12) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(13) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(14) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(15) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(17) - ALK(20) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-ALK fusion | EML4(18) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(19) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(20) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(20) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(21) - ALK(19) | Fusion | NEGATIVE |
| EML4-ALK fusion | EML4(22) - ALK(20) | Fusion | NEGATIVE |
| LRPPRC-ALK fusion | LRPPRC(23) - ALK(20) | Fusion | NEGATIVE |
| PPM1B-ALK fusion | PPM1B(1) - ALK(20) | Fusion | NEGATIVE |
| CAMKMT-ALK fusion | CAMKMT(3) - ALK(20) | Fusion | NEGATIVE |
| PRKCE-ALK fusion | PRKCE(1) - ALK(20) | Fusion | NEGATIVE |
| MCFD2-ALK fusion | MCFD2(1) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(7) - ALK(20) | Fusion | NEGATIVE |
| PPP1R21-ALK fusion | PPP1R21(17) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(5) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(6) - ALK(20) | Fusion | NEGATIVE |
| SPTBN1-ALK fusion | SPTBN1(7) - ALK(20) | Fusion | NEGATIVE |
| CCDC88A-ALK fusion | CCDC88A(12) - ALK(20) | Fusion | NEGATIVE |
| PPP4R3B-ALK fusion | PPP4R3B(10) - ALK(2) | Fusion | NEGATIVE |
| PNPT1-ALK fusion | PNPT1(5) - ALK(4) | Fusion | NEGATIVE |
| BCL11A-ALK fusion | BCL11A(4) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ACTG2-ALK fusion | ACTG2(1) - ALK(19) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(1) - ALK(17) | Fusion | NEGATIVE |
| ACTG2-ALK fusion | ACTG2(2) - ALK(18) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(29) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(27) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(26) - ALK(20) | Fusion | NEGATIVE |
| DCTN1-ALK fusion | DCTN1(15) - ALK(20) | Fusion | NEGATIVE |
| EIF2AK3-ALK fusion | EIF2AK3(2) - ALK(20) | Fusion | NEGATIVE |
| ZNF2-ALK fusion | ZNF2(1) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(13) - ALK(20) | Fusion | NEGATIVE |
| GCC2-ALK fusion | GCC2(19) - ALK(20) | Fusion | NEGATIVE |
| RANBP2-ALK fusion | RANBP2(18) - ALK(20) | Fusion | NEGATIVE |
| CREB1-ALK fusion | CREB1(1) - ALK(9) | Fusion | NEGATIVE |
| ATIC-ALK fusion | ATIC(7) - ALK(20) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(42) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(41) - ALK(16) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(27) - ALK(18) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(23) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(20) - ALK(19) | Fusion | NEGATIVE |
| FN1-ALK fusion | FN1(15) - ALK(18) | Fusion | NEGATIVE |
| IGFBP5-ALK fusion | IGFBP5(1) - ALK(19) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(32) - ALK(13) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TNS1-ALK fusion | TNS1(28) - ALK(20) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(26) - ALK(11) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(20) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(17) - ALK(12) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(17) | Fusion | NEGATIVE |
| TNS1-ALK fusion | TNS1(16) - ALK(20) | Fusion | NEGATIVE |
| DES-ALK fusion | DES(7) - ALK(19) | Fusion | NEGATIVE |
| FBXO36-ALK fusion | FBXO36(2) - ALK(20) | Fusion | NEGATIVE |
| SP100-ALK fusion | SP100(4) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| ATG16L1-ALK fusion | ATG16L1(2) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| AGAP1-ALK fusion | AGAP1(6) - ALK(18) | Fusion | NEGATIVE |
| MLPH-ALK fusion | MLPH(11) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(6) - ALK(20) | Fusion | NEGATIVE |
| SATB1-ALK fusion | SATB1(5) - ALK(20) | Fusion | NEGATIVE |
| LRRFIP2-ALK fusion | LRRFIP2(26) - ALK(20) | Fusion | NEGATIVE |
| PRKAR2A-ALK fusion | PRKAR2A(2) - ALK(20) | Fusion | NEGATIVE |
| TPM4-ALK fusion | TPM4(7) - ALK(20) | Fusion | NEGATIVE |
| SLMAP-ALK fusion | SLMAP(11) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(4) - ALK(19) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TFG-ALK fusion | TFG(5) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(5) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(6) - ALK(20) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(19) | Fusion | NEGATIVE |
| TFG-ALK fusion | TFG(7) - ALK(20) | Fusion | NEGATIVE |
| STK32B-ALK fusion | STK32B(11) - ALK(20) | Fusion | NEGATIVE |
| CCDC149-ALK fusion | CCDC149(10) - ALK(20) | Fusion | NEGATIVE |
| SLC34A2-ALK fusion | SLC34A2(1) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(22) - ALK(20) | Fusion | NEGATIVE |
| SEC31A-ALK fusion | SEC31A(21) - ALK(20) | Fusion | NEGATIVE |
| MANBA-ALK fusion | MANBA(14) - ALK(20) | Fusion | NEGATIVE |
| COL25A1-ALK fusion | COL25A1(3) - ALK(20) | Fusion | NEGATIVE |
| TERT-ALK fusion | TERT(11) - ALK(5) | Fusion | NEGATIVE |
| MROH2B-ALK fusion | MROH2B(21) - ALK(12) | Fusion | NEGATIVE |
| PDE8B-ALK fusion | PDE8B(11) - ALK(20) | Fusion | NEGATIVE |
| NPM1-ALK fusion | NPM1(4) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(20) | Fusion | NEGATIVE |
| SQSTM1-ALK fusion | SQSTM1(5) - ALK(19) | Fusion | NEGATIVE |
| PHACTR1-ALK fusion | PHACTR1(4) - ALK(20) | Fusion | NEGATIVE |
| KCNQ5-ALK fusion | KCNQ5(1) - ALK(10) | Fusion | NEGATIVE |
| SFT2D1-ALK fusion | SFT2D1(1) - ALK(6) | Fusion | NEGATIVE |
| PRKAR1B-ALK fusion | PRKAR1B(4) - ALK(20) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| GTF2IRD1-ALK fusion | GTF2IRD1(4) - ALK(20) | Fusion | NEGATIVE |
| GTF2IRD1-ALK fusion | GTF2IRD1(7) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(30) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(28) - ALK(20) | Fusion | NEGATIVE |
| HIP1-ALK fusion | HIP1(21) - ALK(20) | Fusion | NEGATIVE |
| PCM1-ALK fusion | PCM1(23) - ALK(20) | Fusion | NEGATIVE |
| INTS9-ALK fusion | INTS9(8) - ALK(20) | Fusion | NEGATIVE |
| PABPC1-ALK fusion | PABPC1(9) - ALK(20) | Fusion | NEGATIVE |
| TRAF1-ALK fusion | TRAF1(6) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(24) - ALK(19) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(17) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| KIF5B-ALK fusion | KIF5B(15) - ALK(20) | Fusion | NEGATIVE |
| VCL-ALK fusion | VCL(16) - ALK(20) | Fusion | NEGATIVE |
| ATRNL1-ALK fusion | ATRNL1(19) - ALK(20) | Fusion | NEGATIVE |
| CARS-ALK fusion | CARS(17) - ALK(20) | Fusion | NEGATIVE |
| PLEKHA7-ALK fusion | PLEKHA7(26) - ALK(19) | Fusion | NEGATIVE |
| CTNND1-ALK fusion | CTNND1(16) - ALK(20) | Fusion | NEGATIVE |
| CPSF7-ALK fusion | CPSF7(5) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(8) - ALK(20) | Fusion | NEGATIVE |
| EEF1G-ALK fusion | EEF1G(6) - ALK(20) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MALAT1-ALK fusion | MALAT1(1) - ALK(16) | Fusion | NEGATIVE |
| NUMA1-ALK fusion | NUMA1(19) - ALK(20) | Fusion | NEGATIVE |
| DDX6-ALK fusion | DDX6(2) - ALK(20) | Fusion | NEGATIVE |
| A2M-ALK fusion | A2M(22) - ALK(19) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(4) - ALK(20) | Fusion | NEGATIVE |
| ETV6-ALK fusion | ETV6(5) - ALK(20) | Fusion | NEGATIVE |
| PDE3A-ALK fusion | PDE3A(10) - ALK(8) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(8) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(9) - ALK(20) | Fusion | NEGATIVE |
| PPFIBP1-ALK fusion | PPFIBP1(12) - ALK(20) | Fusion | NEGATIVE |
| HNRNPA1-ALK fusion | HNRNPA1(2) - ALK(18) | Fusion | NEGATIVE |
| CLIP1-ALK fusion | CLIP1(13) - ALK(20) | Fusion | NEGATIVE |
| LMO7-ALK fusion | LMO7(15) - ALK(20) | Fusion | NEGATIVE |
| KTN1-ALK fusion | KTN1(43) - ALK(19) | Fusion | NEGATIVE |
| KLC1-ALK fusion | KLC1(9) - ALK(20) | Fusion | NEGATIVE |
| THBS1-ALK fusion | THBS1(4) - ALK(19) | Fusion | NEGATIVE |
| MYO5C-ALK fusion | MYO5C(30) - ALK(20) | Fusion | NEGATIVE |
| TPM1-ALK fusion | TPM1(8) - ALK(20) | Fusion | NEGATIVE |
| CCDC61-ALK fusion | CCDC61(13) - ALK(20) | Fusion | NEGATIVE |
| RRBP1-ALK fusion | RRBP1(20) - ALK(20) | Fusion | NEGATIVE |
| SPECC1L-ALK fusion | SPECC1L(9) - ALK(20) | Fusion | NEGATIVE |
| TIMP3-ALK fusion | TIMP3(1) - ALK(19) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TIMP3-ALK fusion | TIMP3(1) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(36) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(34) - ALK(20) | Fusion | NEGATIVE |
| MYH9-ALK fusion | MYH9(9) - ALK(6) | Fusion | NEGATIVE |
| DMD-ALK fusion | DMD(55) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(9) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| MSN-ALK fusion | MSN(11) - ALK(20) | Fusion | NEGATIVE |
| AR-V7 | AR(3) - AR(4) | RNAExonVariant | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(4) - BRAF(7) | Fusion | NEGATIVE |
| HERPUD1-BRAF fusion | HERPUD1(6) - BRAF(7) | Fusion | NEGATIVE |
| TOM1L2-BRAF fusion | TOM1L2(11) - BRAF(9) | Fusion | NEGATIVE |
| ZNF207-BRAF fusion | ZNF207(3) - BRAF(10) | Fusion | NEGATIVE |
| RUNDC1-BRAF fusion | RUNDC1(1) - BRAF(9) | Fusion | NEGATIVE |
| LSM12-BRAF fusion | LSM12(3) - BRAF(9) | Fusion | NEGATIVE |
| CDC27-BRAF fusion | CDC27(16) - BRAF(9) | Fusion | NEGATIVE |
| OSBPL1A-BRAF fusion | OSBPL1A(16) - BRAF(10) | Fusion | NEGATIVE |
| ZSCAN30-BRAF fusion | ZSCAN30(3) - BRAF(10) | Fusion | NEGATIVE |
| MBP-BRAF fusion | MBP(2) - BRAF(9) | Fusion | NEGATIVE |
| NFIC-BRAF fusion | NFIC(6) - BRAF(10) | Fusion | NEGATIVE |
| PLIN3-BRAF fusion | PLIN3(1) - BRAF(9) | Fusion | NEGATIVE |
| AGTRAP-BRAF fusion | AGTRAP(5) - BRAF(8) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLCN6-BRAF fusion | CLCN6(2) - BRAF(11) | Fusion | NEGATIVE |
| ZBTB8A-BRAF fusion | ZBTB8A(4) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(5) - BRAF(9) | Fusion | NEGATIVE |
| MACF1-BRAF fusion | MACF1(60) - BRAF(9) | Fusion | NEGATIVE |
| RNF11-BRAF fusion | RNF11(1) - BRAF(11) | Fusion | NEGATIVE |
| EPS15-BRAF fusion | EPS15(22) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(11) - BRAF(10) | Fusion | NEGATIVE |
| OSBPL9-BRAF fusion | OSBPL9(14) - BRAF(9) | Fusion | NEGATIVE |
| BTF3L4-BRAF fusion | BTF3L4(3) - BRAF(11) | Fusion | NEGATIVE |
| FAM73A-BRAF fusion | FAM73A(7) - BRAF(9) | Fusion | NEGATIVE |
| GIPC2-BRAF fusion | GIPC2(3) - BRAF(10) | Fusion | NEGATIVE |
| TPR-BRAF fusion | TPR(14) - BRAF(7) | Fusion | NEGATIVE |
| DSTYK-BRAF fusion | DSTYK(1) - BRAF(10) | Fusion | NEGATIVE |
| SLC45A3-BRAF fusion | SLC45A3(1) - BRAF(8) | Fusion | NEGATIVE |
| PLXNA2-BRAF fusion | PLXNA2(9) - BRAF(2) | Fusion | NEGATIVE |
| EML4-BRAF fusion | EML4(6) - BRAF(10) | Fusion | NEGATIVE |
| BCL2L11-BRAF fusion | BCL2L11(3) - BRAF(10) | Fusion | NEGATIVE |
| TANK-BRAF fusion | TANK(4) - BRAF(9) | Fusion | NEGATIVE |
| DYNC1I2-BRAF fusion | DYNC1I2(7) - BRAF(10) | Fusion | NEGATIVE |
| RAD18-BRAF fusion | RAD18(7) - BRAF(10) | Fusion | NEGATIVE |
| ATG7-BRAF fusion | ATG7(18) - BRAF(9) | Fusion | NEGATIVE |
| RBMS3-BRAF fusion | RBMS3(11) - BRAF(11) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYRIP-BRAF fusion | MYRIP(16) - BRAF(9) | Fusion | NEGATIVE |
| FXR1-BRAF fusion | FXR1(13) - BRAF(10) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(5) - BRAF(9) | Fusion | NEGATIVE |
| DLG1-BRAF fusion | DLG1(3) - BRAF(10) | Fusion | NEGATIVE |
| PAPSS1-BRAF fusion | PAPSS1(5) - BRAF(9) | Fusion | NEGATIVE |
| SLC12A7-BRAF fusion | SLC12A7(17) - BRAF(11) | Fusion | NEGATIVE |
| GHR-BRAF fusion | GHR(1) - BRAF(10) | Fusion | NEGATIVE |
| AP3B1-BRAF fusion | AP3B1(22) - BRAF(9) | Fusion | NEGATIVE |
| PJA2-BRAF fusion | PJA2(7) - BRAF(11) | Fusion | NEGATIVE |
| FCHSD1-BRAF fusion | FCHSD1(13) - BRAF(9) | Fusion | NEGATIVE |
| FAM114A2-BRAF fusion | FAM114A2(9) - BRAF(11) | Fusion | NEGATIVE |
| RNF130-BRAF fusion | RNF130(3) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(10) - BRAF(9) | Fusion | NEGATIVE |
| PRKAR1B-BRAF fusion | PRKAR1B(9) - BRAF(9) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(17) - BRAF(10) | Fusion | NEGATIVE |
| MAD1L1-BRAF fusion | MAD1L1(16) - BRAF(9) | Fusion | NEGATIVE |
| TMEM106B-BRAF fusion | TMEM106B(4) - BRAF(8) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(4) - BRAF(10) | Fusion | NEGATIVE |
| KLHL7-BRAF fusion | KLHL7(5) - BRAF(9) | Fusion | NEGATIVE |
| TAX1BP1-BRAF fusion | TAX1BP1(8) - BRAF(11) | Fusion | NEGATIVE |
| BBS9-BRAF fusion | BBS9(19) - BRAF(4) | Fusion | NEGATIVE |
| NUDCD3-BRAF fusion | NUDCD3(4) - BRAF(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| COBL-BRAF fusion | COBL(7) - BRAF(10) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(3) - BRAF(8) | Fusion | NEGATIVE |
| KCTD7-BRAF fusion | KCTD7(4) - BRAF(8) | Fusion | NEGATIVE |
| CLIP2-BRAF fusion | CLIP2(6) - BRAF(11) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(4) - BRAF(10) | Fusion | NEGATIVE |
| GTF2I-BRAF fusion | GTF2I(20) - BRAF(10) | Fusion | NEGATIVE |
| POR-BRAF fusion | POR(3) - BRAF(11) | Fusion | NEGATIVE |
| PHTF2-BRAF fusion | PHTF2(17) - BRAF(9) | Fusion | NEGATIVE |
| GNAI1-BRAF fusion | GNAI1(1) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(7) - BRAF(11) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(8) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(21) - BRAF(10) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(22) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(28) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(36) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(45) - BRAF(9) | Fusion | NEGATIVE |
| AKAP9-BRAF fusion | AKAP9(48) - BRAF(8) | Fusion | NEGATIVE |
| BAIAP2L1-BRAF fusion | BAIAP2L1(12) - BRAF(9) | Fusion | NEGATIVE |
| ZKSCAN5-BRAF fusion | ZKSCAN5(2) - BRAF(9) | Fusion | NEGATIVE |
| TRIM4-BRAF fusion | TRIM4(6) - BRAF(10) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(8) - BRAF(8) | Fusion | NEGATIVE |
| CUX1-BRAF fusion | CUX1(10) - BRAF(9) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ARMC10-BRAF fusion | ARMC10(3) - BRAF(9) | Fusion | NEGATIVE |
| ARMC10-BRAF fusion | ARMC10(4) - BRAF(11) | Fusion | NEGATIVE |
| PRKAR2B-BRAF fusion | PRKAR2B(1) - BRAF(10) | Fusion | NEGATIVE |
| BCAP29-BRAF fusion | BCAP29(7) - BRAF(11) | Fusion | NEGATIVE |
| DOCK4-BRAF fusion | DOCK4(41) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(9) - BRAF(2) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(10) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(11) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(14) - BRAF(8) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(16) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(9) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(11) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(17) - BRAF(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SND1-BRAF fusion | SND1(18) - BRAF(10) | Fusion | NEGATIVE |
| SND1-BRAF fusion | SND1(18) - BRAF(8) | Fusion | NEGATIVE |
| NRF1-BRAF fusion | NRF1(5) - BRAF(9) | Fusion | NEGATIVE |
| C7orf73-BRAF fusion | C7orf73(2) - BRAF(9) | Fusion | NEGATIVE |
| DGKI-BRAF fusion | DGKI(7) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(3) - BRAF(10) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(5) - BRAF(8) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(6) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(8) - BRAF(11) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(9) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(10) - BRAF(9) | Fusion | NEGATIVE |
| TRIM24-BRAF fusion | TRIM24(11) - BRAF(2) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(18) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(17) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(15) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(14) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(8) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(13) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(11) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(12) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(11) - BRAF(10) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(9) - BRAF(9) | Fusion | NEGATIVE |
| KIAA1549-BRAF fusion | KIAA1549(5) - BRAF(9) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(7) - BRAF(11) | Fusion | NEGATIVE |
| ZC3HAV1-BRAF fusion | ZC3HAV1(3) - BRAF(10) | Fusion | NEGATIVE |
| UBN2-BRAF fusion | UBN2(3) - BRAF(11) | Fusion | NEGATIVE |
| PARP12-BRAF fusion | PARP12(9) - BRAF(11) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | JHDM1D(12) - BRAF(4) | Fusion | NEGATIVE |
| KDM7A-BRAF fusion | KDM7A(11) - BRAF(11) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(9) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(8) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(10) | Fusion | NEGATIVE |
| MKRN1-BRAF fusion | MKRN1(4) - BRAF(11) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(1) - BRAF(10) | Fusion | NEGATIVE |
| TMEM178B-BRAF fusion | TMEM178B(2) - BRAF(9) | Fusion | NEGATIVE |
| AGK-BRAF fusion | AGK(2) - BRAF(8) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(3) - BRAF(9) | Fusion | NEGATIVE |
| FAM131B-BRAF fusion | FAM131B(2) - BRAF(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FAM131B-BRAF fusion | FAM131B(1) - BRAF(10) | Fusion | NEGATIVE |
| CUL1-BRAF fusion | CUL1(7) - BRAF(9) | Fusion | NEGATIVE |
| ZNF767P-BRAF fusion | ZNF767P(1) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(8) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(9) - BRAF(9) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(11) | Fusion | NEGATIVE |
| AGAP3-BRAF fusion | AGAP3(10) - BRAF(9) | Fusion | NEGATIVE |
| NUB1-BRAF fusion | NUB1(3) - BRAF(9) | Fusion | NEGATIVE |
| RBM33-BRAF fusion | RBM33(3) - BRAF(11) | Fusion | NEGATIVE |
| ESYT2-BRAF fusion | ESYT2(8) - BRAF(9) | Fusion | NEGATIVE |
| PCM1-BRAF fusion | PCM1(37) - BRAF(9) | Fusion | NEGATIVE |
| SCRIB-BRAF fusion | SCRIB(27) - BRAF(11) | Fusion | NEGATIVE |
| KANK1-BRAF fusion | KANK1(7) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(8) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(13) - BRAF(9) | Fusion | NEGATIVE |
| CDK5RAP2-BRAF fusion | CDK5RAP2(12) - BRAF(11) | Fusion | NEGATIVE |
| NUP214-BRAF fusion | NUP214(21) - BRAF(10) | Fusion | NEGATIVE |
| CCNY-BRAF fusion | CCNY(1) - BRAF(10) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(2) - BRAF(8) | Fusion | NEGATIVE |
| CCDC6-BRAF fusion | CCDC6(1) - BRAF(9) | Fusion | NEGATIVE |
| SOX6-BRAF fusion | SOX6(6) - BRAF(9) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SOX6-BRAF fusion | SOX6(5) - BRAF(9) | Fusion | NEGATIVE |
| PICALM-BRAF fusion | PICALM(19) - BRAF(9) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(12) - BRAF(10) | Fusion | NEGATIVE |
| ERC1-BRAF fusion | ERC1(17) - BRAF(8) | Fusion | NEGATIVE |
| CCDC91-BRAF fusion | CCDC91(11) - BRAF(9) | Fusion | NEGATIVE |
| GIT2-BRAF fusion | GIT2(14) - BRAF(9) | Fusion | NEGATIVE |
| MZT1-BRAF fusion | MZT1(2) - BRAF(11) | Fusion | NEGATIVE |
| LMO7-BRAF fusion | LMO7(7) - BRAF(9) | Fusion | NEGATIVE |
| STXBP6-BRAF fusion | STXBP6(4) - BRAF(10) | Fusion | NEGATIVE |
| STRN3-BRAF fusion | STRN3(3) - BRAF(10) | Fusion | NEGATIVE |
| CDC42BPB-BRAF fusion | CDC42BPB(36) - BRAF(9) | Fusion | NEGATIVE |
| GATM-BRAF fusion | GATM(2) - BRAF(11) | Fusion | NEGATIVE |
| EPS15L1-BRAF fusion | EPS15L1(16) - BRAF(9) | Fusion | NEGATIVE |
| CEP89-BRAF fusion | CEP89(16) - BRAF(9) | Fusion | NEGATIVE |
| LSM14A-BRAF fusion | LSM14A(9) - BRAF(9) | Fusion | NEGATIVE |
| TMPRSS2-BRAF fusion | TMPRSS2(3) - BRAF(11) | Fusion | NEGATIVE |
| PACSIN2-BRAF fusion | PACSIN2(7) - BRAF(9) | Fusion | NEGATIVE |
| RP2-BRAF fusion | RP2(3) - BRAF(10) | Fusion | NEGATIVE |
| EGFRvIII | EGFR(1) - EGFR(8) | RNAExonVariant | NEGATIVE |
| PLEKHG1-ESR1 fusion | PLEKHG1(3) - ESR1(2) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(8) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(6) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(7) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(10) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(5) | Fusion | NEGATIVE |
| ESR1-CCDC170 fusion | ESR1(2) - CCDC170(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(3) - ARMT1(4) | Fusion | NEGATIVE |
| ESR1-ATP2B2 fusion | ESR1(4) - ATP2B2(4) | Fusion | NEGATIVE |
| ESR1-COA5 fusion | ESR1(4) - COA5(2) | Fusion | NEGATIVE |
| ESR1-ARMT1 fusion | ESR1(6) - ARMT1(2) | Fusion | NEGATIVE |
| ESR1-NKAIN2 fusion | ESR1(6) - NKAIN2(2) | Fusion | NEGATIVE |
| ESR1-SOX9 fusion | ESR1(6) - SOX9(3) | Fusion | NEGATIVE |
| ESR1-MKL1 fusion | ESR1(6) - MKL1(7) | Fusion | NEGATIVE |
| ESR1-BMPER fusion | ESR1(6) - BMPER(8) | Fusion | NEGATIVE |
| ESR1-TNRC6B fusion | ESR1(6) - TNRC6B(14) | Fusion | NEGATIVE |
| ESR1-YAP1 fusion | ESR1(6) - YAP1(4) | Fusion | NEGATIVE |
| ESR1-GYG2 fusion | ESR1(6) - GYG2(2) | Fusion | NEGATIVE |
| ESR1-TFG fusion | ESR1(6) - TFG(4) | Fusion | NEGATIVE |
| ESR1-POLH fusion | ESR1(6) - POLH(2) | Fusion | NEGATIVE |
| ESR1-PCDH11X fusion | ESR1(6) - PCDH11X(6) | Fusion | NEGATIVE |
| ESR1-DAB2 fusion | ESR1(6) - DAB2(3) | Fusion | NEGATIVE |
| ESR1-RMND1 fusion | ESR1(6) - RMND1(9) | Fusion | NEGATIVE |
| ESR1-PLEKHG1 fusion | ESR1(6) - PLEKHG1(12) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(6) - MTHFD1L(21) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ESR1-SYNE1 fusion | ESR1(6) - SYNE1(59) | Fusion | NEGATIVE |
| ESR1-NOP2 fusion | ESR1(6) - NOP2(16) | Fusion | NEGATIVE |
| ESR1-PDE10A fusion | ESR1(7) - PDE10A(6) | Fusion | NEGATIVE |
| ESR1-CDK13 fusion | ESR1(7) - CDK13(13) | Fusion | NEGATIVE |
| ESR1-MTHFD1L fusion | ESR1(7) - MTHFD1L(27) | Fusion | NEGATIVE |
| ESR1-ARNT2 fusion | ESR1(7) - ARNT2(9) | Fusion | NEGATIVE |
| TNS3-ESR1 fusion | TNS3(16) - ESR1(5) | Fusion | NEGATIVE |
| SNX29-FGFR1 fusion | SNX29(1) - FGFR1(4) | Fusion | NEGATIVE |
| MYO18A-FGFR1 fusion | MYO18A(33) - FGFR1(10) | Fusion | NEGATIVE |
| TPR-FGFR1 fusion | TPR(22) - FGFR1(10) | Fusion | NEGATIVE |
| RANBP2-FGFR1 fusion | RANBP2(20) - FGFR1(10) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(27) - FGFR1(5) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(25) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(4) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(23) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(3) | Fusion | NEGATIVE |
| FN1-FGFR1 fusion | FN1(22) - FGFR1(4) | Fusion | NEGATIVE |
| LRRFIP1-FGFR1 fusion | LRRFIP1(8) - FGFR1(10) | Fusion | NEGATIVE |
| ACPP-FGFR1 fusion | ACPP(10) - FGFR1(4) | Fusion | NEGATIVE |
| SQSTM1-FGFR1 fusion | SQSTM1(6) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(5) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP-FGFR1 fusion | FGFR1OP(6) - FGFR1(10) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR1OP-FGFR1 fusion | FGFR1OP(7) - FGFR1(10) | Fusion | NEGATIVE |
| EGFR-FGFR1 fusion | EGFR(24) - FGFR1(10) | Fusion | NEGATIVE |
| CUX1-FGFR1 fusion | CUX1(11) - FGFR1(10) | Fusion | NEGATIVE |
| TRIM24-FGFR1 fusion | TRIM24(11) - FGFR1(10) | Fusion | NEGATIVE |
| ERLIN2-FGFR1 fusion | ERLIN2(8) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(8) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(1) - FGFR1(2) | Fusion | NEGATIVE |
| BAG4-FGFR1 fusion | BAG4(2) - FGFR1(6) | Fusion | NEGATIVE |
| WHSC1L1-FGFR1 fusion | WHSC1L1(14) - FGFR1(5) | Fusion | NEGATIVE |
| FGFR1-HOOK3 fusion | FGFR1(17) - HOOK3(5) | Fusion | NEGATIVE |
| FGFR1-TACC1 fusion | FGFR1(17) - TACC1(7) | Fusion | NEGATIVE |
| FGFR1-ADAM32 fusion | FGFR1(17) - ADAM32(14) | Fusion | NEGATIVE |
| FGFR1-EVI5 fusion | FGFR1(17) - EVI5(12) | Fusion | NEGATIVE |
| KAT6A-FGFR1 fusion | KAT6A(5) - FGFR1(2) | Fusion | NEGATIVE |
| CNTRL-FGFR1 fusion | CNTRL(40) - FGFR1(10) | Fusion | NEGATIVE |
| FGFR1OP2-FGFR1 fusion | FGFR1OP2(4) - FGFR1(10) | Fusion | NEGATIVE |
| CPSF6-FGFR1 fusion | CPSF6(8) - FGFR1(10) | Fusion | NEGATIVE |
| ZMYM2-FGFR1 fusion | ZMYM2(17) - FGFR1(10) | Fusion | NEGATIVE |
| ERVK3-1-FGFR1 fusion | ERVK3-1(3) - FGFR1(10) | Fusion | NEGATIVE |
| BCR-FGFR1 fusion | BCR(4) - FGFR1(10) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(2) | Fusion | NEGATIVE |
| SLC45A3-FGFR2 fusion | SLC45A3(1) - FGFR2(1) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CTNNB1-FGFR2 fusion | CTNNB1(1) - FGFR2(10) | Fusion | NEGATIVE |
| COL25A1-FGFR2 fusion | COL25A1(3) - FGFR2(3) | Fusion | NEGATIVE |
| PARK2-FGFR2 fusion | PARK2(9) - FGFR2(11) | Fusion | NEGATIVE |
| FGFR2-CBX5 fusion | FGFR2(17) - CBX5(2) | Fusion | NEGATIVE |
| FGFR2-ANKRD24 fusion | FGFR2(17) - ANKRD24(3) | Fusion | NEGATIVE |
| FGFR2-NOL4 fusion | FGFR2(17) - NOL4(7) | Fusion | NEGATIVE |
| FGFR2-PPHLN1 fusion | FGFR2(17) - PPHLN1(3) | Fusion | NEGATIVE |
| FGFR2-PKD2L1 fusion | FGFR2(17) - PKD2L1(7) | Fusion | NEGATIVE |
| FGFR2-UTRN fusion | FGFR2(17) - UTRN(51) | Fusion | NEGATIVE |
| FGFR2-TACC3 fusion | FGFR2(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR2-CREB5 fusion | FGFR2(17) - CREB5(8) | Fusion | NEGATIVE |
| FGFR2-CLIP2 fusion | FGFR2(17) - CLIP2(10) | Fusion | NEGATIVE |
| FGFR2-GAB2 fusion | FGFR2(17) - GAB2(2) | Fusion | NEGATIVE |
| FGFR2-OFD1 fusion | FGFR2(17) - OFD1(3) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(7) | Fusion | NEGATIVE |
| FGFR2-WARS fusion | FGFR2(17) - WARS(3) | Fusion | NEGATIVE |
| FGFR2-SBNO2 fusion | FGFR2(17) - SBNO2(19) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(2) | Fusion | NEGATIVE |
| FGFR2-NRBF2 fusion | FGFR2(17) - NRBF2(4) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(2) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(10) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(16) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-PHC1 fusion | FGFR2(17) - PHC1(2) | Fusion | NEGATIVE |
| FGFR2-TFEC fusion | FGFR2(17) - TFEC(2) | Fusion | NEGATIVE |
| FGFR2-SMN1 fusion | FGFR2(17) - SMN1(2) | Fusion | NEGATIVE |
| FGFR2-OGDH fusion | FGFR2(17) - OGDH(3) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-KRT8 fusion | FGFR2(17) - KRT8(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(6) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(5) | Fusion | NEGATIVE |
| FGFR2-KIAA1217 fusion | FGFR2(17) - KIAA1217(3) | Fusion | NEGATIVE |
| FGFR2-TXLNA fusion | FGFR2(17) - TXLNA(6) | Fusion | NEGATIVE |
| FGFR2-INA fusion | FGFR2(17) - INA(2) | Fusion | NEGATIVE |
| FGFR2-UBQLN1 fusion | FGFR2(17) - UBQLN1(8) | Fusion | NEGATIVE |
| FGFR2-CIT fusion | FGFR2(17) - CIT(23) | Fusion | NEGATIVE |
| FGFR2-TACC2 fusion | FGFR2(17) - TACC2(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(18) | Fusion | NEGATIVE |
| FGFR2-PASD1 fusion | FGFR2(17) - PASD1(10) | Fusion | NEGATIVE |
| FGFR2-TBC1D1 fusion | FGFR2(17) - TBC1D1(9) | Fusion | NEGATIVE |
| FGFR2-FAM76A fusion | FGFR2(17) - FAM76A(2) | Fusion | NEGATIVE |
| FGFR2-AHCYL1 fusion | FGFR2(17) - AHCYL1(2) | Fusion | NEGATIVE |
| FGFR2-MCU fusion | FGFR2(17) - MCU(4) | Fusion | NEGATIVE |
| FGFR2-CELF2 fusion | FGFR2(17) - CELF2(4) | Fusion | NEGATIVE |
| FGFR2-COL14A1 fusion | FGFR2(17) - COL14A1(34) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-AFF3 fusion | FGFR2(17) - AFF3(7) | Fusion | NEGATIVE |
| FGFR2-CCDC6 fusion | FGFR2(17) - CCDC6(1) | Fusion | NEGATIVE |
| FGFR2-WAC fusion | FGFR2(17) - WAC(5) | Fusion | NEGATIVE |
| FGFR2-MGEA5 fusion | FGFR2(17) - MGEA5(12) | Fusion | NEGATIVE |
| FGFR2-DDX21 fusion | FGFR2(17) - DDX21(11) | Fusion | NEGATIVE |
| FGFR2-BICC1 fusion | FGFR2(17) - BICC1(3) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(7) | Fusion | NEGATIVE |
| FGFR2-VCL fusion | FGFR2(17) - VCL(2) | Fusion | NEGATIVE |
| FGFR2-ZMYM4 fusion | FGFR2(17) - ZMYM4(10) | Fusion | NEGATIVE |
| FGFR2-PCM1 fusion | FGFR2(17) - PCM1(7) | Fusion | NEGATIVE |
| FGFR2-CCAR2 fusion | FGFR2(17) - CCAR2(4) | Fusion | NEGATIVE |
| FGFR2-EIF4A2 fusion | FGFR2(17) - EIF4A2(8) | Fusion | NEGATIVE |
| FGFR2-RABGAP1L fusion | FGFR2(17) - RABGAP1L(20) | Fusion | NEGATIVE |
| FGFR2-RASAL2 fusion | FGFR2(17) - RASAL2(3) | Fusion | NEGATIVE |
| FGFR2-LAMC1 fusion | FGFR2(17) - LAMC1(27) | Fusion | NEGATIVE |
| FGFR2-STK26 fusion | FGFR2(17) - STK26(3) | Fusion | NEGATIVE |
| FGFR2-CASP7 fusion | FGFR2(17) - CASP7(2) | Fusion | NEGATIVE |
| FGFR2-TJP2 fusion | FGFR2(17) - TJP2(5) | Fusion | NEGATIVE |
| FGFR2-SHTN1 fusion | FGFR2(17) - SHTN1(5) | Fusion | NEGATIVE |
| FGFR2-KCTD1 fusion | FGFR2(17) - KCTD1(2) | Fusion | NEGATIVE |
| FGFR2-OPTN fusion | FGFR2(17) - OPTN(5) | Fusion | NEGATIVE |
| FGFR2-ROCK1 fusion | FGFR2(17) - ROCK1(2) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR2-NRAP fusion | FGFR2(17) - NRAP(24) | Fusion | NEGATIVE |
| FGFR2-ACSL5 fusion | FGFR2(17) - ACSL5(15) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(6) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(10) | Fusion | NEGATIVE |
| APIP-FGFR2 fusion | APIP(1) - FGFR2(6) | Fusion | NEGATIVE |
| PDHX-FGFR2 fusion | PDHX(1) - FGFR2(7) | Fusion | NEGATIVE |
| CD44-FGFR2 fusion | CD44(1) - FGFR2(3) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(8) - FGFR2(7) | Fusion | NEGATIVE |
| SNX19-FGFR2 fusion | SNX19(7) - FGFR2(7) | Fusion | NEGATIVE |
| KLK2-FGFR2 fusion | KLK2(1) - FGFR2(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(13) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-ELAVL3 fusion | FGFR3(17) - ELAVL3(2) | Fusion | NEGATIVE |
| FGFR3-RBBP4 fusion | FGFR3(17) - RBBP4(12) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-AMBRA1 fusion | FGFR3(17) - AMBRA1(14) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-BAIAP2L1 fusion | FGFR3(17) - BAIAP2L1(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(14) | Fusion | NEGATIVE |
| FGFR3-NBR1 fusion | FGFR3(17) - NBR1(7) | Fusion | NEGATIVE |
| FGFR3-ADD1 fusion | FGFR3(17) - ADD1(3) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-FBXO28 fusion | FGFR3(17) - FBXO28(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(6) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-BRAP fusion | FGFR3(17) - BRAP(5) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-AES fusion | FGFR3(17) - AES(2) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(8) | Fusion | NEGATIVE |
| FGFR3-JAKMIP1 fusion | FGFR3(17) - JAKMIP1(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(16) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(4) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(7) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(4) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(9) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(1) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(17) - TACC3(11) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(10) | Fusion | NEGATIVE |
| FGFR3-CAMK2A fusion | FGFR3(18) - CAMK2A(14) | Fusion | NEGATIVE |
| FGFR3-TACC3 fusion | FGFR3(18) - TACC3(11) | Fusion | NEGATIVE |
| ETV6-FGFR3 fusion | ETV6(5) - FGFR3(9) | Fusion | NEGATIVE |
| EPS15-MET fusion | EPS15(21) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(36) - MET(15) | Fusion | NEGATIVE |
| TPR-MET fusion | TPR(4) - MET(15) | Fusion | NEGATIVE |
| DCTN1-MET fusion | DCTN1(26) - MET(15) | Fusion | NEGATIVE |
| LRRFIP1-MET fusion | LRRFIP1(19) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(5) - MET(15) | Fusion | NEGATIVE |
| TFG-MET fusion | TFG(7) - MET(15) | Fusion | NEGATIVE |
| SLC34A2-MET fusion | SLC34A2(4) - MET(15) | Fusion | NEGATIVE |
| HLA-DRB1-MET fusion | HLA-DRB1(5) - MET(15) | Fusion | NEGATIVE |
| STARD3NL-MET fusion | STARD3NL(6) - MET(15) | Fusion | NEGATIVE |
| CLIP2-MET fusion | CLIP2(11) - MET(15) | Fusion | NEGATIVE |
| GTF2I-MET fusion | GTF2I(6) - MET(15) | Fusion | NEGATIVE |
| BAIAP2L1-MET fusion | BAIAP2L1(9) - MET(15) | Fusion | NEGATIVE |
| TRIM4-MET fusion | TRIM4(6) - MET(15) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZKSCAN1-MET fusion | ZKSCAN1(3) - MET(15) | Fusion | NEGATIVE |
| MET exon 14 skipping (RNA) | MET(13) - MET(15) | RNAExonVariant | NEGATIVE |
| MET KDD | MET(21) - MET(15) | RNAExonVariant | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(1) - MET(6) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(2) - MET(15) | Fusion | NEGATIVE |
| CAPZA2-MET fusion | CAPZA2(4) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(11) | Fusion | NEGATIVE |
| ST7-MET fusion | ST7(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(1) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(2) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(3) - MET(2) | Fusion | NEGATIVE |
| PTPRZ1-MET fusion | PTPRZ1(8) - MET(2) | Fusion | NEGATIVE |
| C8orf34-MET fusion | C8orf34(2) - MET(15) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(9) - MET(13) | Fusion | NEGATIVE |
| OXR1-MET fusion | OXR1(1) - MET(13) | Fusion | NEGATIVE |
| KIF5B-MET fusion | KIF5B(24) - MET(15) | Fusion | NEGATIVE |
| ETV6-MET fusion | ETV6(4) - MET(15) | Fusion | NEGATIVE |
| PPFIBP1-MET fusion | PPFIBP1(9) - MET(15) | Fusion | NEGATIVE |
| SCAF11-MET fusion | SCAF11(1) - MET(2) | Fusion | NEGATIVE |
| CDH1-NRG1 fusion | CDH1(2) - NRG1(2) | Fusion | NEGATIVE |
| VAMP2-NRG1 fusion | VAMP2(4) - NRG1(4) | Fusion | NEGATIVE |
| COX10-AS1-NRG1 fusion | COX10-AS1(1) - NRG1(2) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ROCK1-NRG1 fusion | ROCK1(2) - NRG1(2) | Fusion | NEGATIVE |
| SMAD4-NRG1 fusion | SMAD4(1) - NRG1(6) | Fusion | NEGATIVE |
| VTCN1-NRG1 fusion | VTCN1(2) - NRG1(2) | Fusion | NEGATIVE |
| NOTCH2-NRG1 fusion | NOTCH2(4) - NRG1(6) | Fusion | NEGATIVE |
| ATP1B1-NRG1 fusion | ATP1B1(2) - NRG1(2) | Fusion | NEGATIVE |
| TBL1XR1-NRG1 fusion | TBL1XR1(1) - NRG1(2) | Fusion | NEGATIVE |
| PARP8-NRG1 fusion | PARP8(1) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(2) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(7) - NRG1(6) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(3) | Fusion | NEGATIVE |
| CD74-NRG1 fusion | CD74(6) - NRG1(6) | Fusion | NEGATIVE |
| CLU-NRG1 fusion | CLU(2) - NRG1(6) | Fusion | NEGATIVE |
| HMBOX1-NRG1 fusion | HMBOX1(1) - NRG1(6) | Fusion | NEGATIVE |
| WRN-NRG1 fusion | WRN(30) - NRG1(2) | Fusion | NEGATIVE |
| WHSC1L1-NRG1 fusion | WHSC1L1(1) - NRG1(2) | Fusion | NEGATIVE |
| PDE7A-NRG1 fusion | PDE7A(3) - NRG1(6) | Fusion | NEGATIVE |
| TNC-NRG1 fusion | TNC(10) - NRG1(6) | Fusion | NEGATIVE |
| RAB3IL1-NRG1 fusion | RAB3IL1(9) - NRG1(6) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(2) - NRG1(2) | Fusion | NEGATIVE |
| SLC3A2-NRG1 fusion | SLC3A2(5) - NRG1(6) | Fusion | NEGATIVE |
| TENM4-NRG1 fusion | TENM4(12) - NRG1(2) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| ZMYM2-NRG1 fusion | ZMYM2(2) - NRG1(2) | Fusion | NEGATIVE |
| RALGAPA1-NRG1 fusion | RALGAPA1(20) - NRG1(6) | Fusion | NEGATIVE |
| THBS1-NRG1 fusion | THBS1(6) - NRG1(6) | Fusion | NEGATIVE |
| AKAP13-NRG1 fusion | AKAP13(5) - NRG1(2) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(4) - NRG1(6) | Fusion | NEGATIVE |
| SDC4-NRG1 fusion | SDC4(2) - NRG1(6) | Fusion | NEGATIVE |
| TSHZ2-NRG1 fusion | TSHZ2(1) - NRG1(6) | Fusion | NEGATIVE |
| SETD4-NRG1 fusion | SETD4(2) - NRG1(2) | Fusion | NEGATIVE |
| THAP7-NRG1 fusion | THAP7(3) - NRG1(2) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(11) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(10) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(22) - NTRK1(13) | Fusion | NEGATIVE |
| PPL-NTRK1 fusion | PPL(12) - NTRK1(13) | Fusion | NEGATIVE |
| BANP-NTRK1 fusion | BANP(4) - NTRK1(10) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(11) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(10) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(9) - NTRK1(9) | Fusion | NEGATIVE |
| TP53-NTRK1 fusion | TP53(8) - NTRK1(9) | Fusion | NEGATIVE |
| MPRIP-NTRK1 fusion | MPRIP(21) - NTRK1(12) | Fusion | NEGATIVE |
| RNF213-NTRK1 fusion | RNF213(15) - NTRK1(12) | Fusion | NEGATIVE |
| CTRC-NTRK1 fusion | CTRC(1) - NTRK1(9) | Fusion | NEGATIVE |
| EPHB2-NTRK1 fusion | EPHB2(3) - NTRK1(9) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TRIM63-NTRK1 fusion | TRIM63(8) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| EPS15-NTRK1 fusion | EPS15(21) - NTRK1(9) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(10) | Fusion | NEGATIVE |
| CHTOP-NTRK1 fusion | CHTOP(5) - NTRK1(11) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(8) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(12) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(13) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(9) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(7) - NTRK1(10) | Fusion | NEGATIVE |
| TPM3-NTRK1 fusion | TPM3(6) - NTRK1(11) | Fusion | NEGATIVE |
| GON4L-NTRK1 fusion | GON4L(21) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF2-NTRK1 fusion | ARHGEF2(21) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(2) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(3) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(4) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(10) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(5) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(6) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(13) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(12) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(10) - NTRK1(11) | Fusion | NEGATIVE |
| LMNA-NTRK1 fusion | LMNA(11) - NTRK1(11) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(12) - NTRK1(10) | Fusion | NEGATIVE |
| BCAN-NTRK1 fusion | BCAN(13) - NTRK1(11) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| MRPL24-NTRK1 fusion | MRPL24(1) - NTRK1(9) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(16) - DYNC2H1(85) | Fusion | NEGATIVE |
| NTRK1-DYNC2H1 fusion | NTRK1(17) - DYNC2H1(85) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(2) - NTRK1(9) | Fusion | NEGATIVE |
| PEAR1-NTRK1 fusion | PEAR1(15) - NTRK1(10) | Fusion | NEGATIVE |
| LRRC71-NTRK1 fusion | LRRC71(1) - NTRK1(9) | Fusion | NEGATIVE |
| VANGL2-NTRK1 fusion | VANGL2(1) - NTRK1(12) | Fusion | NEGATIVE |
| SCYL3-NTRK1 fusion | SCYL3(11) - NTRK1(12) | Fusion | NEGATIVE |
| RABGAP1L-NTRK1 fusion | RABGAP1L(14) - NTRK1(16) | Fusion | NEGATIVE |
| RFWD2-NTRK1 fusion | RFWD2(2) - NTRK1(3) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(12) | Fusion | NEGATIVE |

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Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TPR-NTRK1 fusion | TPR(21) - NTRK1(13) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(21) - NTRK1(9) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(16) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(4) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(10) - NTRK1(10) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(8) - NTRK1(11) | Fusion | NEGATIVE |
| TPR-NTRK1 fusion | TPR(6) - NTRK1(12) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(21) - NTRK1(9) | Fusion | NEGATIVE |
| PLEKHA6-NTRK1 fusion | PLEKHA6(16) - NTRK1(10) | Fusion | NEGATIVE |
| MDM4-NTRK1 fusion | MDM4(3) - NTRK1(5) | Fusion | NEGATIVE |
| NFASC-NTRK1 fusion | NFASC(20) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(12) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(11) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(9) | Fusion | NEGATIVE |
| IRF2BP2-NTRK1 fusion | IRF2BP2(1) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(4) - NTRK1(9) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(5) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(6) - NTRK1(10) | Fusion | NEGATIVE |
| TFG-NTRK1 fusion | TFG(7) - NTRK1(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| AFAP1-NTRK1 fusion | AFAP1(4) - NTRK1(10) | Fusion | NEGATIVE |
| SSBP2-NTRK1 fusion | SSBP2(12) - NTRK1(12) | Fusion | NEGATIVE |
| CD74-NTRK1 fusion | CD74(7) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(2) - NTRK1(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(12) | Fusion | NEGATIVE |
| SQSTM1-NTRK1 fusion | SQSTM1(5) - NTRK1(10) | Fusion | NEGATIVE |
| GSN-NTRK1 fusion | GSN(7) - NTRK1(10) | Fusion | NEGATIVE |
| CEL-NTRK1 fusion | CEL(7) - NTRK1(9) | Fusion | NEGATIVE |
| ARHGEF25-NTRK1 fusion | ARHGEF25(11) - NTRK1(8) | Fusion | NEGATIVE |
| GRIPAP1-NTRK1 fusion | GRIPAP1(22) - NTRK1(11) | Fusion | NEGATIVE |
| AGBL4-NTRK2 fusion | AGBL4(6) - NTRK2(16) | Fusion | NEGATIVE |
| NOS1AP-NTRK2 fusion | NOS1AP(9) - NTRK2(11) | Fusion | NEGATIVE |
| NAV1-NTRK2 fusion | NAV1(15) - NTRK2(11) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(14) - NTRK2(5) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(15) | Fusion | NEGATIVE |
| STRN-NTRK2 fusion | STRN(3) - NTRK2(16) | Fusion | NEGATIVE |
| SLMAP-NTRK2 fusion | SLMAP(14) - NTRK2(16) | Fusion | NEGATIVE |
| TP63-NTRK2 fusion | TP63(8) - NTRK2(12) | Fusion | NEGATIVE |
| AFAP1-NTRK2 fusion | AFAP1(14) - NTRK2(12) | Fusion | NEGATIVE |
| KCTD8-NTRK2 fusion | KCTD8(1) - NTRK2(13) | Fusion | NEGATIVE |
| VCAN-NTRK2 fusion | VCAN(6) - NTRK2(10) | Fusion | NEGATIVE |
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(17) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SQSTM1-NTRK2 fusion | SQSTM1(5) - NTRK2(13) | Fusion | NEGATIVE |
| QKI-NTRK2 fusion | QKI(6) - NTRK2(16) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(15) | Fusion | NEGATIVE |
| TRIM24-NTRK2 fusion | TRIM24(12) - NTRK2(16) | Fusion | NEGATIVE |
| GNAQ-NTRK2 fusion | GNAQ(2) - NTRK2(16) | Fusion | NEGATIVE |
| TLE4-NTRK2 fusion | TLE4(7) - NTRK2(15) | Fusion | NEGATIVE |
| GKAP1-NTRK2 fusion | GKAP1(9) - NTRK2(13) | Fusion | NEGATIVE |
| WNK2-NTRK2 fusion | WNK2(24) - NTRK2(16) | Fusion | NEGATIVE |
| TBC1D2-NTRK2 fusion | TBC1D2(6) - NTRK2(12) | Fusion | NEGATIVE |
| DAB2IP-NTRK2 fusion | DAB2IP(1) - NTRK2(17) | Fusion | NEGATIVE |
| NACC2-NTRK2 fusion | NACC2(4) - NTRK2(13) | Fusion | NEGATIVE |
| TRAF2-NTRK2 fusion | TRAF2(9) - NTRK2(15) | Fusion | NEGATIVE |
| VCL-NTRK2 fusion | VCL(16) - NTRK2(12) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(4) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(16) | Fusion | NEGATIVE |
| ETV6-NTRK2 fusion | ETV6(5) - NTRK2(15) | Fusion | NEGATIVE |
| PAN3-NTRK2 fusion | PAN3(1) - NTRK2(17) | Fusion | NEGATIVE |
| STRN3-NTRK2 fusion | STRN3(7) - NTRK2(16) | Fusion | NEGATIVE |
| BCR-NTRK2 fusion | BCR(1) - NTRK2(17) | Fusion | NEGATIVE |
| NTRK3-LOXL2 fusion | NTRK3(3) - LOXL2(5) | Fusion | NEGATIVE |
| ZNF710-NTRK3 fusion | ZNF710(1) - NTRK3(14) | Fusion | NEGATIVE |
| FBXL20-NTRK3 fusion | FBXL20(1) - NTRK3(6) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| EML4-NTRK3 fusion | EML4(2) - NTRK3(14) | Fusion | NEGATIVE |
| EML4-NTRK3 fusion | EML4(6) - NTRK3(14) | Fusion | NEGATIVE |
| TPM4-NTRK3 fusion | TPM4(7) - NTRK3(12) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(15) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(14) | Fusion | NEGATIVE |
| TFG-NTRK3 fusion | TFG(6) - NTRK3(10) | Fusion | NEGATIVE |
| FAT1-NTRK3 fusion | FAT1(2) - NTRK3(7) | Fusion | NEGATIVE |
| SQSTM1-NTRK3 fusion | SQSTM1(5) - NTRK3(14) | Fusion | NEGATIVE |
| HNRNPA2B1-NTRK3 fusion | HNRNPA2B1(7) - NTRK3(14) | Fusion | NEGATIVE |
| RBPMS-NTRK3 fusion | RBPMS(5) - NTRK3(14) | Fusion | NEGATIVE |
| LYN-NTRK3 fusion | LYN(8) - NTRK3(14) | Fusion | NEGATIVE |
| KANK1-NTRK3 fusion | KANK1(3) - NTRK3(14) | Fusion | NEGATIVE |
| UBE2R2-NTRK3 fusion | UBE2R2(3) - NTRK3(4) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(15) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(4) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(14) | Fusion | NEGATIVE |
| ETV6-NTRK3 fusion | ETV6(5) - NTRK3(15) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(13) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(32) - NTRK3(12) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(14) | Fusion | NEGATIVE |
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(11) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| MYO5A-NTRK3 fusion | MYO5A(23) - NTRK3(12) | Fusion | NEGATIVE |
| COX5A-NTRK3 fusion | COX5A(1) - NTRK3(15) | Fusion | NEGATIVE |
| BTBD1-NTRK3 fusion | BTBD1(4) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(14) - NTRK3(14) | Fusion | NEGATIVE |
| AKAP13-NTRK3 fusion | AKAP13(16) - NTRK3(15) | Fusion | NEGATIVE |
| NTRK3-HOMER2 fusion | NTRK3(17) - HOMER2(2) | Fusion | NEGATIVE |
| NTRK3-HOMER1 fusion | NTRK3(17) - HOMER1(2) | Fusion | NEGATIVE |
| MYH9-NTRK3 fusion | MYH9(32) - NTRK3(14) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(15) - NUTM1(2) | Fusion | NEGATIVE |
| MXD1-NUTM1 fusion | MXD1(5) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(14) - NUTM1(2) | Fusion | NEGATIVE |
| BRD4-NUTM1 fusion | BRD4(11) - NUTM1(2) | Fusion | NEGATIVE |
| AFF1-NUTM1 fusion | AFF1(17) - NUTM1(3) | Fusion | NEGATIVE |
| WHSC1L1-NUTM1 fusion | WHSC1L1(7) - NUTM1(2) | Fusion | NEGATIVE |
| ZNF618-NUTM1 fusion | ZNF618(12) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(2) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(10) - NUTM1(5) | Fusion | NEGATIVE |
| BRD3-NUTM1 fusion | BRD3(9) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(16) - NUTM1(4) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(17) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(5) | Fusion | NEGATIVE |
| CIC-NUTM1 fusion | CIC(18) - NUTM1(2) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CIC-NUTM1 fusion | CIC(20) - NUTM1(5) | Fusion | NEGATIVE |
| BCORL1-NUTM1 fusion | BCORL1(1) - NUTM1(2) | Fusion | NEGATIVE |
| MYH10-RET fusion | MYH10(33) - RET(12) | Fusion | NEGATIVE |
| MYH13-RET fusion | MYH13(35) - RET(12) | Fusion | NEGATIVE |
| MPRIP-RET fusion | MPRIP(19) - RET(12) | Fusion | NEGATIVE |
| PRKAR1A-RET fusion | PRKAR1A(7) - RET(12) | Fusion | NEGATIVE |
| KIAA1468-RET fusion | KIAA1468(10) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(16) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(14) - RET(12) | Fusion | NEGATIVE |
| TRIM33-RET fusion | TRIM33(11) - RET(12) | Fusion | NEGATIVE |
| RASAL2-RET fusion | RASAL2(15) - RET(12) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(11) | Fusion | NEGATIVE |
| EML4-RET fusion | EML4(19) - RET(12) | Fusion | NEGATIVE |
| SATB1-RET fusion | SATB1(7) - RET(12) | Fusion | NEGATIVE |
| TFG-RET fusion | TFG(4) - RET(11) | Fusion | NEGATIVE |
| ABI3BP-RET fusion | ABI3BP(34) - RET(12) | Fusion | NEGATIVE |
| PDCD10-RET fusion | PDCD10(4) - RET(11) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(12) | Fusion | NEGATIVE |
| TBL1XR1-RET fusion | TBL1XR1(9) - RET(11) | Fusion | NEGATIVE |
| TNIP2-RET fusion | TNIP2(5) - RET(12) | Fusion | NEGATIVE |
| AFAP1-RET fusion | AFAP1(3) - RET(12) | Fusion | NEGATIVE |
| EPHA5-RET fusion | EPHA5(2) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| TNIP1-RET fusion | TNIP1(11) - RET(12) | Fusion | NEGATIVE |
| RUFY1-RET fusion | RUFY1(17) - RET(12) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(5) - RET(11) | Fusion | NEGATIVE |
| SQSTM1-RET fusion | SQSTM1(6) - RET(12) | Fusion | NEGATIVE |
| KIF13A-RET fusion | KIF13A(18) - RET(12) | Fusion | NEGATIVE |
| TRIM27-RET fusion | TRIM27(3) - RET(12) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(11) - RET(11) | Fusion | NEGATIVE |
| FGFR1OP-RET fusion | FGFR1OP(12) - RET(12) | Fusion | NEGATIVE |
| CUX1-RET fusion | CUX1(10) - RET(12) | Fusion | NEGATIVE |
| TRIM24-RET fusion | TRIM24(9) - RET(12) | Fusion | NEGATIVE |
| PCM1-RET fusion | PCM1(29) - RET(12) | Fusion | NEGATIVE |
| HOOK3-RET fusion | HOOK3(11) - RET(12) | Fusion | NEGATIVE |
| FKBP15-RET fusion | FKBP15(25) - RET(12) | Fusion | NEGATIVE |
| FRMD4A-RET fusion | FRMD4A(12) - RET(12) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(10) | Fusion | NEGATIVE |
| KIAA1217-RET fusion | KIAA1217(11) - RET(11) | Fusion | NEGATIVE |
| ACBD5-RET fusion | ACBD5(11) - RET(12) | Fusion | NEGATIVE |
| WAC-RET fusion | WAC(3) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(8) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(24) - RET(11) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(23) - RET(8) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| KIF5B-RET fusion | KIF5B(22) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(18) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(16) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(12) | Fusion | NEGATIVE |
| KIF5B-RET fusion | KIF5B(15) - RET(11) | Fusion | NEGATIVE |
| RASSF4-RET fusion | RASSF4(3) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(6) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(7) - RET(12) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(8) | Fusion | NEGATIVE |
| NCOA4-RET fusion | NCOA4(9) - RET(12) | Fusion | NEGATIVE |
| PRKG1-RET fusion | PRKG1(3) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(8) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(5) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(2) - RET(12) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(9) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(10) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| CCDC6-RET fusion | CCDC6(1) - RET(11) | Fusion | NEGATIVE |
| ANK3-RET fusion | ANK3(1) - RET(12) | Fusion | NEGATIVE |
| RUFY2-RET fusion | RUFY2(9) - RET(12) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| DLG5-RET fusion | DLG5(13) - RET(12) | Fusion | NEGATIVE |
| ADD3-RET fusion | ADD3(1) - RET(12) | Fusion | NEGATIVE |
| CCDC186-RET fusion | CCDC186(7) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(6) - RET(12) | Fusion | NEGATIVE |
| AFAP1L2-RET fusion | AFAP1L2(2) - RET(12) | Fusion | NEGATIVE |
| PPFIBP2-RET fusion | PPFIBP2(9) - RET(12) | Fusion | NEGATIVE |
| PICALM-RET fusion | PICALM(19) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(7) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(11) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(12) - RET(12) | Fusion | NEGATIVE |
| ERC1-RET fusion | ERC1(17) - RET(12) | Fusion | NEGATIVE |
| ETV6-RET fusion | ETV6(6) - RET(12) | Fusion | NEGATIVE |
| OLFM4-RET fusion | OLFM4(4) - RET(10) | Fusion | NEGATIVE |
| KTN1-RET fusion | KTN1(29) - RET(12) | Fusion | NEGATIVE |
| GPHN-RET fusion | GPHN(9) - RET(12) | Fusion | NEGATIVE |
| GOLGA5-RET fusion | GOLGA5(7) - RET(12) | Fusion | NEGATIVE |
| MYO5C-RET fusion | MYO5C(25) - RET(12) | Fusion | NEGATIVE |
| MYO5A-RET fusion | MYO5A(23) - RET(11) | Fusion | NEGATIVE |
| AKAP13-RET fusion | AKAP13(35) - RET(12) | Fusion | NEGATIVE |
| SNRNP70-RET fusion | SNRNP70(7) - RET(12) | Fusion | NEGATIVE |
| RRBP1-RET fusion | RRBP1(9) - RET(12) | Fusion | NEGATIVE |
| BCR-RET fusion | BCR(4) - RET(12) | Fusion | NEGATIVE |

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| SPECC1L-RET fusion | SPECC1L(10) - RET(11) | Fusion | NEGATIVE |
| SPECC1L-RET fusion | SPECC1L(10) - RET(12) | Fusion | NEGATIVE |
| YWHAE-ROS1 fusion | YWHAE(4) - ROS1(36) | Fusion | NEGATIVE |
| CLTC-ROS1 fusion | CLTC(31) - ROS1(35) | Fusion | NEGATIVE |
| CCDC30-ROS1 fusion | CCDC30(10) - ROS1(36) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(7) - ROS1(35) | Fusion | NEGATIVE |
| TPM3-ROS1 fusion | TPM3(3) - ROS1(36) | Fusion | NEGATIVE |
| SLMAP-ROS1 fusion | SLMAP(10) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(4) - ROS1(35) | Fusion | NEGATIVE |
| TFG-ROS1 fusion | TFG(5) - ROS1(35) | Fusion | NEGATIVE |
| GOLGB1-ROS1 fusion | GOLGB1(9) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(4) - ROS1(35) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(32) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(34) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(33) | Fusion | NEGATIVE |
| SLC34A2-ROS1 fusion | SLC34A2(13) - ROS1(36) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(7) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(34) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(35) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(6) - ROS1(32) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

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| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CD74-ROS1 fusion | CD74(6) - ROS1(33) | Fusion | NEGATIVE |
| CD74-ROS1 fusion | CD74(4) - ROS1(33) | Fusion | NEGATIVE |
| PWWP2A-ROS1 fusion | PWWP2A(1) - ROS1(36) | Fusion | NEGATIVE |
| HLA-A-ROS1 fusion | HLA-A(7) - ROS1(34) | Fusion | NEGATIVE |
| EYS-ROS1 fusion | EYS(12) - ROS1(9) | Fusion | NEGATIVE |
| SLC22A16-ROS1 fusion | SLC22A16(7) - ROS1(16) | Fusion | NEGATIVE |
| SLC16A10-ROS1 fusion | SLC16A10(3) - ROS1(3) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(8) - ROS1(35) | Fusion | NEGATIVE |
| GOPC-ROS1 fusion | GOPC(4) - ROS1(36) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(12) - ROS1(35) | Fusion | NEGATIVE |
| CEP85L-ROS1 fusion | CEP85L(8) - ROS1(36) | Fusion | NEGATIVE |
| TPD52L1-ROS1 fusion | TPD52L1(3) - ROS1(33) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(34) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(35) | Fusion | NEGATIVE |
| EZR-ROS1 fusion | EZR(10) - ROS1(33) | Fusion | NEGATIVE |
| TMEM106B-ROS1 fusion | TMEM106B(3) - ROS1(35) | Fusion | NEGATIVE |
| CCDC6-ROS1 fusion | CCDC6(5) - ROS1(35) | Fusion | NEGATIVE |
| NFKB2-ROS1 fusion | NFKB2(14) - ROS1(36) | Fusion | NEGATIVE |
| ERC1-ROS1 fusion | ERC1(11) - ROS1(36) | Fusion | NEGATIVE |
| PPFIBP1-ROS1 fusion | PPFIBP1(9) - ROS1(35) | Fusion | NEGATIVE |
| LIMA1-ROS1 fusion | LIMA1(10) - ROS1(36) | Fusion | NEGATIVE |
| LRIG3-ROS1 fusion | LRIG3(16) - ROS1(35) | Fusion | NEGATIVE |

## Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| CLIP1-ROS1 fusion | CLIP1(20) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(3) - ROS1(36) | Fusion | NEGATIVE |
| ZCCHC8-ROS1 fusion | ZCCHC8(2) - ROS1(36) | Fusion | NEGATIVE |
| NCOR2-ROS1 fusion | NCOR2(7) - ROS1(36) | Fusion | NEGATIVE |
| MYO5C-ROS1 fusion | MYO5C(30) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(32) - ROS1(35) | Fusion | NEGATIVE |
| MYO5A-ROS1 fusion | MYO5A(23) - ROS1(35) | Fusion | NEGATIVE |
| TSPAN3-ROS1 fusion | TSPAN3(5) - ROS1(31) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(5) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(4) - ROS1(34) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(32) | Fusion | NEGATIVE |
| SDC4-ROS1 fusion | SDC4(2) - ROS1(34) | Fusion | NEGATIVE |
| MSN-ROS1 fusion | MSN(9) - ROS1(34) | Fusion | NEGATIVE |
| GRHL2-RSPO2 fusion | GRHL2(8) - RSPO2(2) | Fusion | NEGATIVE |
| EIF3E-RSPO2 fusion | EIF3E(1) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(3) - RSPO2(2) | Fusion | NEGATIVE |
| NRIP1-RSPO2 fusion | NRIP1(2) - RSPO2(2) | Fusion | NEGATIVE |
| ASH1L-RSPO3 fusion | ASH1L(1) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(10) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(7) - RSPO3(2) | Fusion | NEGATIVE |
| PTPRK-RSPO3 fusion | PTPRK(6) - RSPO3(2) | Fusion | NEGATIVE |

# Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.0.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 26 JUN 2025

| Variant Name | Genes (Exons) | Variant Type | Test Result |
|---|---|---|---|
| PTPRK-RSPO3 fusion | PTPRK(1) - RSPO3(2) | Fusion | NEGATIVE |

ⓘ Fusions include gene fusions and splice variants.

* RNAExonTiles are amplicons that target exonic-junctions in RNA libraries for measurement of 3p/5p expression imbalance and inference of fusion in a partner agnostic manner.

## CNVs

| Variant Name | Test Result |
|---|---|
| AR amplification | NEGATIVE |
| ERBB2 amplification | NEGATIVE |
| ERBB3 amplification | NEGATIVE |
| FGFR1 amplification | NEGATIVE |
| FGFR2 amplification | NEGATIVE |
| FGFR3 amplification | NEGATIVE |
| KRAS amplification | NEGATIVE |
| MET amplification | NEGATIVE |
| PIK3CA amplification | NEGATIVE |

ion torrent

# Genexus™ Dx Software 6.8

## USER GUIDE

Publication Number MAN1000074

Revision B

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
#07-06, Singapore 739256

Products manufactured at this site:

- Genexus™ Dx Integrated Sequencer
- Genexus™ Dx Software

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.

Revision history: MAN1000074 B (English)

| Revision | Date | Description |
|---|---|---|
| B | 25 June 2025 | • Updated “Sign in to a new user account” on page 17 and “Reset a user password (Administrator)” on page 155 to correct password requirements in Genexus™ Dx Software 6.8.2.<br>• Added “Sequencer decommissioning and data sanitization” on page 189. |
| A | 12 June 2024 | New user guide for Genexus™ Dx Software 6.8.2 user guide for FDA submission. |

The information in this guide is subject to change without notice.

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# About this guide

## Purpose of this guide

This user guide provides detailed instructions for using Genexus™ Dx Software 6.8 for sample management, data analysis, and system management.

For detailed instructions for operating the Genexus™ Dx Integrated Sequencer, see the latest version of the *Genexus™ Dx Integrated Sequencer User Guide*.

## Prerequisites

| Category | Prerequisites |
|---|---|
| Instrument | • Genexus™ Dx Integrated Sequencer<br>• Reagents and supplies for operating the Genexus™ Dx Integrated Sequencer |
| Software | • Genexus™ Dx Software 6.8 or later |
| Functional knowledge and understanding | • Key steps in a next-generation sequencing (NGS) workflow<br>• Main functions of the Genexus™ Dx Integrated Sequencer<br>• Main features of the Genexus™ Dx Software 6.8 or later |

# 1 Introduction to Genexus™ Dx Software


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## Software overview

Ion Torrent™ Genexus™ Dx Software supports the Ion Torrent™ Genexus™ Dx Integrated Sequencer workflow from sample preparation through library preparation, template preparation, and sequencing. During and after sequencing, the software generates base calls, trims reads, and determines quality values (primary analysis), then aligns reads, calls variants, and generates reports (secondary analysis).

This documentation provides instructions for using the software to perform the following functions:

- Create and manage samples, libraries, and runs in the software, and manage assays.
- Monitor the status of runs on the Genexus™ Dx Integrated Sequencer, which provides all-in-one library preparation, chip loading, template preparation, and sequencing.
- View data, results, and quality control (QC) reports after a run.
- Download data files.
- Export and print audit records.
- Create and manage user accounts.

## General instrument use

The Ion Torrent™ Genexus™ Dx Integrated Sequencer is an in vitro diagnostic (IVD) medical device intended to be used by trained personnel in a professional laboratory environment for automated, qualitative targeted sequencing of human DNA or RNA extracted from formalin-fixed, paraffin-embedded (FFPE) tumor tissue samples. The Genexus™ Dx Integrated Sequencer is not intended for whole genome or de novo sequencing.

# Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software 6.8 or later. Version-specific information is provided in the software release notes for your version of the software. An administrator-level user can view the software version in the **Settings / Software Updates** screen.

Genexus™ Dx Software is supported on Google Chrome™ browser version 90 and later and is best viewed with 1440 × 900 screen resolution. Google Chrome™ browser is recommended for use with the software.

The operating system of the sequencer is Ubuntu™ 18.04.1 LTS.

# Network and password security requirements

## Network configuration and security

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of your facility administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions.

If external or network drives are connected to the sequencer, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

If a LIMS connection is set up, you can use the LIMS API to get the list of files generated for the assay. For more information, see “LIMS introduction” on page 191.

---

Note: If a LIMS system is configured to retrieve files from the sequencer, you have the option to manually set up an FTP directory or drive mapping. This configuration is not provided as part of sequencer installation, and must be set up by your LIMS system integration or IT group.

---

## Password security

Best practice is to maintain unique passwords for all accounts in use in Genexus™ Dx Software. All passwords must be reset the first time a user signs into the software. Change passwords according to your organization's password policy.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords. This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# Installation and verification run

---

IMPORTANT! The Genexus™ Dx Integrated Sequencer and the Genexus™ Dx Software provides the option to perform a performance qualification verification run.

Notifications are sent when the PQ run is expired. Contact your Field Support Engineer (FSE) or Technical Support if you have questions about an expired PQ status.

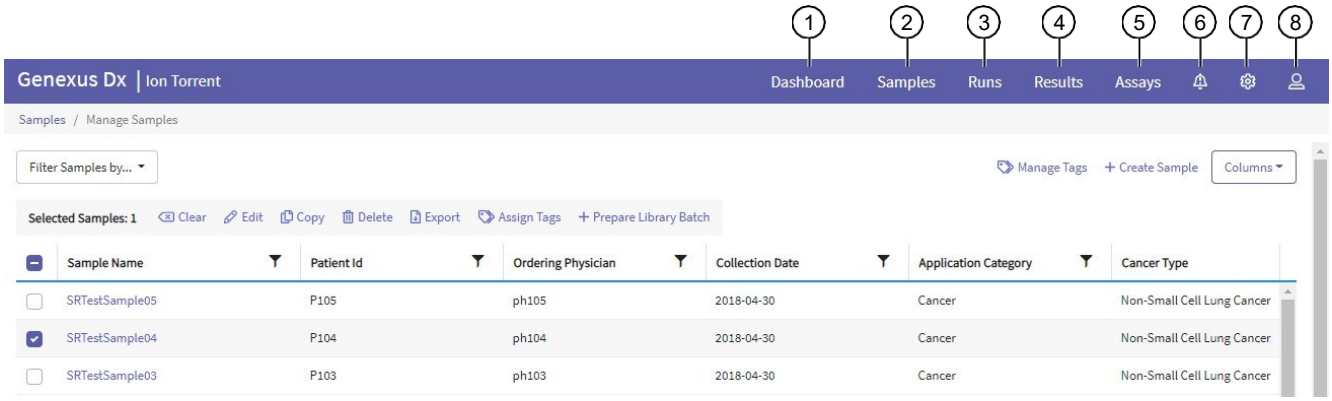
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# 2 Get started with Genexus™ Dx Software


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# About Genexus™ Dx Software

Genexus™ Dx Software provides menus to help you add, select, and manage samples, libraries, runs, and assays. You can also view and manage sequencing results, and manage software settings.




① **Dashboard**—View recent run history, and current sequencing run status.

② **Samples**—Add new samples, import samples, prepare library batches, import library batches and manage attributes.

③ **Runs**—Plan a run starting from a nucleic acid sample or a library. View, edit, and manage runs.

④ **Results**—View sample results, run results, and verification results.

⑤ **Assays**—View and manage assays.

⑥ **Notifications**—Receive alerts and messages for password expiration, system critical service failures, system backup failures, available software updates, and full disk space.

⑦ **Settings**—Access audit records and run logs, configure network settings, manage backup settings, restore runs, link to Connect user accounts, check for software updates, and manage data archiving, disk space, and users.

⑧ **Profile**—Access the Help system, manage and edit user profile settings, configure an SSH key (system administrator only), and sign out.

## Access the End User License Agreement (EULA)

A link to the End User License Agreement (EULA) is located at the bottom of the screen.

1. Scroll to the bottom of the screen.
2. Click the **EULA** link to open a PDF file of the EULA.

## Contact Support

You can access Genexus™ Dx Software technical support from the software.

1. Scroll to the bottom of the screen.
2. Click **Support** to open **https://www.thermofisher.com/contactus**, where you can request help and technical support.

# User-access levels

| Users at this level | Can perform these actions |
|---|---|
| Report | • Access the Sample Results and Run Results screens to view results<br>• Generate, view, and sign variant reports<br>• Send notifications<br>• Download results files<br>• View and edit notes for the sample result<br>• View the audit trail for sample results<br>• View notifications |
| Operator | Report functions, plus:<br>• Add, import, and export sample files<br>• Edit sample files<br>• Prepare library batches<br>• Plan, save, audit, and delete runs<br>• Monitor runs<br>• View results and reports<br>• View the audit trail for samples, library batches, runs, and assays<br>• Reanalyze runs |

*(continued)*

| Users at this level | Can perform these actions |
|---|---|
| Manager | Operator functions, plus:<br>• Create, edit, and obsolete sample attributes<br>• Import assays<br>• Create report templates<br>• Edit and delete sample files<br>• Add or edit Thermo Fisher accounts<br>• Restart a stalled or failed run<br>• Access services information |
| Administrator | Operator and manager functions, plus:<br>• View, export, and print audit records<br>• Configure network settings<br>• View and manage software updates<br>• Configure backup settings and restore runs<br>• Manage sequencer and software log files<br>• Add and manage user accounts |

# System tracking

The system tracks and checks auditable user activities for samples, assays, QC metrics and information related to reagents.

If the software detects an error at any step—for example, a scanned barcode is inconsistent with the information given for the run—the software alerts the user and does not proceed with the run.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# Reagent management

Put in place a reagent management system to inventory and track reagent use.

Proper use of a reagent management system promotes regulatory compliance and enables optimal use of kits, chips, and reagents.

# View notifications

You can view notifications for files that you upload and import in Genexus™ Dx Software.

1. In the menu bar, click **(Notifications)**.
   If you have any unread notifications, the number of unread notifications is shown to the upper right of the notification button.
2. View the list of notifications in the dialog box that appears.
3. To delete a notification, click ✕ **(Remove)** in the section of the notification.

# Sign in to a new user account

Users must request a new user account from administrators before you can sign in to Genexus™ Dx Software. Administrators receive the request and create the user account. After the administrator creates a new account, the software sends an email to the new user with a user name and temporary password. Your user name and password must be unique and must not be shared with other users.

1. Obtain your user name and temporary password that is supplied by your administrator in an email message.
2. Browse to the Genexus™ Dx Software sign in page (for example, *https://<your server address>*), then enter your user name and the temporary password.
3. Press **Enter**, or click **Sign In**.
4. In the **End User License Agreement** that appears at first sign on, read the agreement, then click **Accept**.
5. In the **Change Password** screen, enter your temporary password, type a new password, confirm the password, then click **Change**.

The software opens to the **Dashboard**.

# Sign in

Before you can sign in to Genexus™ Dx Software, you must have a registered account. See “Sign in to a new user account” on page 17.

1. Go to the Genexus™ Dx Software sign in page.
2. Click **Switch Language**, then select the preferred language from the list, if desired.
3. Enter your user name and password, then press **Enter** or click **Sign In**.
   If a message appears that indicates that there is a problem with the security token, refresh the browser, then enter the user name and password again.

The software opens to the **Dashboard** screen.

## Mode switch with a partially used chip installed

In Genexus™ Dx Software you can switch from IVD mode to Assay Development mode, or from Assay Development mode to IVD mode, when you sign in to the software. If you want to switch modes to plan and perform a run in a new mode, and you have a partially used chip installed in the sequencer, you cannot use the partially used chip and sequencing consumables in the new mode. A dialog box appears asking you to confirm the mode switch. You can cancel the mode switch, but if you proceed, you must perform an instrument clean and install a new chip and consumables. You will lose the unused chip lanes and remaining sequencing consumables.

If you want to switch modes to review run data or reanalyze data in the other mode, and do not perform a sequencing run, an instrument clean is not needed. You can proceed with the mode switch and you do not need to replace the installed chip and consumables.

## Switch the software mode

You can switch between two separate software modes in Genexus™ Dx Software:

- Assay Development mode
- IVD mode

1. Go to the Genexus™ Dx Software sign in screen.
2. In **Choose System Mode**, select the mode that you want to use.
3. Click **Switch Language**, then select the preferred language from the dropdown list that appears, if desired.
4. Enter the user name and password, then press **Enter** or click **Sign In**.

---

Note: If a message appears that indicates that there is a problem with the security token, refresh the browser and enter the user name and password again.

---

# Edit a user profile

After your user profile has been established, you can view user profile information, change a password, or change email contact information.

1. In the menu bar, click (Profile)►**My Profile**.
2. In the **Profile** screen, view the profile information.

| To | Do this |
|---|---|
| Change a password | a. Click **Change Password**.<br>b. In the **Change Password** dialog box, enter the current password and a new password. Confirm the password, then click **Change**. |
| Change an email address | a. Click **Change Email**.<br>b. In the **Set Email** dialog box, enter a new email address, and a password, then click **Save**. |
| Edit security questions and answers | a. Click **Edit Security questions and answers**.<br>b. In the **Edit Security questions and answers** dialog box, select a security question from the dropdown list, then enter an answer.<br>c. Enter the password for the account that is associated with the profile.<br>d. Click **Submit**. |

The changes are applied immediately. After the password is changed, you must sign in with the new password.

# Genexus™ Dx Software concepts

The following terms are frequently used in Genexus™ Dx Software.

For more definitions, see “Genexus™ Dx Software definitions” on page 227.

| Term or concept | Description |
|---|---|
| Sample | The information that describes the material from a single source. Each sample is defined by a set of attributes, which are categories of information that describe important aspects of each sample, such as date of sample collection, gender, or a sample type.<br>In the software, a sample is isolated nucleic acid. |
| Sample name | A unique identifier for a particular sample. |
| Assay | A collection of settings and parameters for library preparation, templating, sequencing, and sequencing data analysis. |
| Library | A sample associated with an assay, including barcode information and sample input quantity. Libraries are associated with an assay during library batch creation. |

*(continued)*

| Term or concept | Description |
|---|---|
| Library batch | A collection of libraries that are prepared and sequenced in the same run under the same conditions on the Genexus™ Dx Integrated Sequencer. |
| Library batch ID | A unique identifier for a library batch. |

# Workflows

| Type of workflow | More information |
|---|---|
| Nucleic Acid to Result<br>Library to Result | “Nucleic Acid to Result or Library to Result workflow” on page 21 |

## Nucleic Acid to Result or Library to Result workflow

This is a workflow for a **Nucleic Acid to Result** or **Library to Result** sequencing run using the Genexus™ Dx Integrated Sequencer.

### Nucleic Acid to Result or Library to Result workflow

**View and manage assays (manager/administrator)** (page 139)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Dx Software. You can use the system-installed assays in a run without change.

**Enter samples and libraries** (page 23)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as sample collection date, gender, type, and application category.



**Plan a Nucleic Acid to Result run** (page 51)

**OR**

**Plan a Library to Result run** (page 55)

Runs created in Genexus™ Dx Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

**Dilute samples and load the sample plate (see the instrument or assay user guide)**

Quantify and dilute the nucleic acid samples, then load the sample plate.



**Load the sequencer and start a run (see the instrument or assay user guide)**

Follow the step-by-step instructions in the sequencer touch screen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



## Nucleic Acid to Result or Library to Result workflow

**Monitor the run** (page 64)

Monitor the run in Genexus™ Dx Software in real time.



**Review data and results** (page 69)

Review data and results in Genexus™ Dx Software.

# 3 Samples and library batches


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## About samples and library batches

A sample can be isolated nucleic acid or a specimen that requires nucleic acid isolation. Before you can plan a run on the Genexus™ Integrated Sequencer, you must enter samples into Genexus™ Dx Software. Samples in Genexus™ Dx Software include the data and attributes that characterize the genomic data from a single source. You add this sample information to run plans when Genexus™ Dx Software guides you through the steps to set up a run plan.

A library batch is a group of samples that are sequenced in a library run. You can create library batches from samples you have previously added or uploaded to the software. During library batch preparation, you identify the barcode adapters that were used to prepare the libraries. You add library batches to run plans when Genexus™ Dx Software guides you through the steps to plan a library run.

## Enter and manage samples

There are two ways to add samples in Genexus™ Dx Software.

- Enter information for a single sample or multiple samples into the software.
- Create a file of information for a group of samples and import that file.

If you enter the information for samples into the software, you can select from system-installed sample attributes that are available in the software. For more information, see “Create a new sample” on page 28 and “System-installed sample attributes” on page 218.

System-installed and custom sample attributes are also available in an example file that you can use to import samples. See “Download a template file for sample creation” on page 32 for more information.

---

IMPORTANT! The cancer type attribute is included in the Assay that is added to Genexus™ Dx Software, and is not available as a system attribute. Before you can create a sample in Genexus™ Dx Software, you must upload the assay that includes the cancer type attribute.

---

Samples in Genexus™ Dx Software are listed in the **Manage Samples** screen.

## Customize the samples table

The samples table is organized into columns of sample attributes. You can customize the table with table preferences to determine which columns appear. When you save the table customizations, the changes you make are saved only for your user profile.

1. In the menu bar, click **Samples►Manage Samples**.
2. Click **Columns**.
   - Select the checkbox next to each attribute you would like to display.
   - Deselect the checkbox next to each attribute you would like to remove from the table.
3. Scroll to the bottom of the list.
   - Click **Save** to save your new selections. Your selections persist when you reopen the table.
   - Click **Restore default** to restore the table to the default column settings.

## Find samples

You can sort, search, and filter information to refine and narrow the samples that are listed in the **Samples / Manage Samples** screen in Genexus™ Dx Software. The sort, search, and filter tools help you organize samples and find a samples of interest.

For information to search for a sample by a unique identifier, such as the sample name, see “Search for samples” on page 24.

For information to filter samples by a common trait, such as the date the sample was created, see “Filter the list of samples by the date of sample creation” on page 26.

### Search for samples

Samples are listed in the **Samples / Manage Samples** screen in Genexus™ Dx Software. The most recently created sample is at the top of the list.

You can search for samples with keywords or text that are included in the sample names, tags, or by the application category for the sample.

---

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, in the column heading of interest, click **(Filter)**.

3. In the search field, enter a full or partial name or ID of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.

4. In the filter dialog box, select from the limited list.
   - Select a checkbox next to the name or ID to select one or more items.
   - Click **Select All** to select all of the items listed in the filter dialog box.
   - Click **Clear** to close the filter dialog box.

5. Click **Filter**.
   In the column heading, changes color to indicate that a filter is applied.

The filtered list of samples is shown in the **Manage Samples** screen.

Click **Clear Filters** to remove all filters and view the full list of samples.

## Sort the list of samples

You can sort the list of samples to make it easier to find a sample in Genexus™ Dx Software.

By default, samples are listed in the **Samples / Manage Samples** screen by the date that the sample was created. The most recently created sample is at the top of the list.

You can change the order that the samples are listed in the table.

1. In the menu bar, click **Samples►Manage Samples**.

2. In the **Manage Samples** screen, click a column heading of interest.
   The list of samples is sorted by the sample attribute, in alphabetical or chronological order of the attribute, depending on the heading.

3. Click the column heading again to reverse the order.

## Filter the list of samples by the date of sample creation

You can apply filters to narrow search results, or shorten the list of samples when you view the list of samples in Genexus™ Dx Software.

In addition to the filter tools in the column headers, you can also filter the list of samples with a dropdown list and search by the sample name or other column headers. For more information, see “Refine the list of samples” on page 27 and “Search for samples” on page 24.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select one or more filters.

| Option | Description |
|---|---|
| List only samples created on an exact date | a. In the **Created On** column header, click **(Filter)**.<br>b. In the first dropdown list, select **Is equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| List samples created within a date range | a. In the **Created On** column header, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date that the samples of interest were created.<br>d. In the Boolean operator dropdown list, select **And**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date that the samples of interest were created.<br>g. Click **Filter**.<br>Records are listed for samples created on dates that meet both conditions. |
| List samples according to a date that meets either one condition or a second condition | a. In the **Created On** column header, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date that the samples of interest were created.<br>d. In the Boolean operator dropdown list, select **Or**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date that the samples of interest were created.<br>g. Click **Filter**.<br>Records are listed for samples created on dates that meet at least one of the conditions. |

| Option | Description |
|---|---|
| List only samples that were created before a date that you indicate | a. In the **Created On** column header, click **(Filter)**.<br>b. In the first dropdown list, select **Is before or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| List only samples that were created after a date that you indicate | a. In the **Created On** column header, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column header, changes color to indicate that a filter is applied.

The list of samples is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of samples.

## Refine the list of samples

You can shorten the list of samples that are shown in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, select the type of sample you want to view.

| Option | Description |
|---|---|
| List only the samples that have not been prepared as a library batch. | Select **To Be Prepared**. |
| List all samples. | Select **All**. |

Only the selected type of samples are listed.

## Create a new sample

You can enter information individually for each sample that you add in the software. Complete this procedure to make a sample available for use in a run, or to add the sample to a library batch. If you have many samples to add, see instead “Create multiple samples” on page 28 and “Import samples” on page 29.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Samples/Create Sample** screen, enter a name for the sample, select an **Application Category**, then complete the required fields, and optional fields if needed.
   - In the **Tags** field, enter a full or partial tag name of at least 3 characters, then select one or more tags of interest to assign a tag to the sample.
   - If the tag that you want to assign to the sample does not exist in the software, a manager-or administrator-level user can enter a new tag name in the **Tags** field to create a new tag and assign it to the sample.

   The fields for sample attributes change based on the **Application Category**. Attributes identified with a red asterisk (*) are required.

   ---

   Note: Users are recommended to add custom attributes such as "Specimen Collection Site" and "Specimen Type" (for example, resection, CNB, and FNA). For information about adding custom attributes, see “Add a custom sample attribute (manager/administrator)” on page 32.

   ---
3. Click **Save**.
   The new sample is listed in the **Manage Samples** screen and is available to use in a run plan.

## Create multiple samples

There are two ways you can create multiple new samples in Genexus™ Dx Software.

- You can enter sample information for multiple samples in a table format in the software.
- You can import a file that contains sample data. For more information, see instead “Import samples” on page 29.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.
   A table with columns that are specific for the selected application category appears.
4. *(Optional)* Click **Columns** in the upper right corner of the screen to customize the optional attributes for the samples that you want to create.
   - Select the checkbox for a sample attribute to add a column to the table. Click the row to view fields into which you can enter attribute information.
   - Deselect the checkbox for a sample attribute to remove the attribute from the table.

5. Enter the information for a sample.

   Attributes identified with a red asterisk (*) are required.

6. Click **Add Row**, then enter the information in the new row for each new sample.

7. Repeat step 6 for each new sample.

8. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you will create.

9. Click **Save**.

   The new samples are listed in the **Manage Samples** screen and are available to use in a run plan.

## Import samples

You can enter sample information for multiple samples directly in Genexus™ Dx Software. When you want to create more than a few samples, an easy and fast way to add multiple samples in the software is to create a file of information for a group of samples and import that file.

Sample data files can be used to capture, manage, and edit sample data. You can import sample data files in TSV, XLS, XLSX, or CSV file formats. For a list of the sample attributes that are included in the import file, see “System-installed sample attributes” on page 218. For ease of use, you can download a Microsoft™ Excel™ template file to create an import file.

1. In the menu bar, click **Samples►Create Sample**.

2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.

3. In the **Application Category** dropdown list, select the application category for the samples.

   Tip: Use the search field to search for the application category of interest.

4. Set up a sample file using one of two options.

| Option | Description |
|---|---|
| Download a template file, then edit it to create a new file. | For more information, see “Download a template file for sample creation” on page 32. Upload the edited file using the **Browse** button.<br><br>Note: When you select **Other** as the **Application Category**, you must enter text in the **Application Category** text box, then click anywhere in the screen in order for the template file to be available. |
| Upload the sample data from an existing file. | a. Click **Browse**.<br>b. Navigate to the file, then click **Open**.<br><br>The data contained in the file populates the table in the screen. |

5. In the sample file, edit the sample table and data, if needed.

| Option | Description |
|---|---|
| Remove an attribute column. | Click **Columns**, then deselect the column name. |
| Add an attribute column to the table. | Click **Columns**, then select the column name. The column names that are listed are **Application Category**-specific. |
| Edit sample data. | Click the sample row of interest, then edit the text field or dropdown list for the sample. |
| Add more samples to the table. | Click **Add Row**, then enter the sample data. |
| Remove a sample from the table. | Select the checkbox for the row, then click **Remove Row**. |

6. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample you intend to create.

7. Click **Save**.
   You can place the pointer over ⓘ **(alert)** to view more information if needed.

The new samples are listed in the **Manage Samples** screen and are available to assign to a run plan.

## View sample summary

You can view a summary of the details for the attributes of a sample. The sample summary lists the information for sample attributes for the sample and the date the sample was created in the software. For a list of the information that is shown in the sample summary, see “Key Findings” on page 73.

For more information about sample attributes, see “System-installed sample attributes” on page 218.

To view the sample summary:

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Sample Name** column, click the sample name of interest.
   Details for the sample are shown in the sample summary pane.
3. When you are finished viewing the sample details, click another sample name to view details for another sample, or click ✕ **(Close)** to return to the **Manage Samples** screen.

## Sample summary

You can view details for its attributes when you select a single sample that is listed in the **Samples/Manage Samples** screen in Genexus™ Dx Software. The sample summary lists details that describe sample attributes, such as gender and the date that the sample was created in the software.

Notes for a sample are not shown in the sample summary pane. For more information, see "View notes or add a note to a sample" on page 37.

The sample attributes that are shown in the sample summary pane depend on the application category for the sample. The following table lists the system-installed sample attributes for a sample that has an application category of cancer. Custom sample attributes are also shown in the sample summary pane, but are not listed in the following table.

| Sample attribute | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Patient Id | An identifier for the patient. |
| Date of Birth | The date of birth for the patient. |
| Ordering Physician | The name of the physician who created the order. |
| Collection Date | The date that the sample was collected. |
| Created On | The date the sample was imported or created in the software. |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Sample Type | A term that describes the sample, for example, RNA. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. 'Percent Cellularity' refers to the same measurement as 'Percent Tumor Content'. |
| %Necrosis | The percentage of cellular necrosis in the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected.<br>Stage 0–IV, or Primary, Unknown, or Other. |

## Download a template file for sample creation

You can download a template file, add sample information to it, then use the file to import the sample data for multiple samples.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headings in the **Specimen Format** tab. Use the **Specimen Format** tab to enter samples.

Template files contain both the system-installed and the custom sample attributes as column headings.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.
   Template files are specific for each application category.
4. Click **Download Template** to download the Microsoft™ Excel™ template file.
   The template file contains default sample attributes as columns. If custom sample attributes have been configured in the software, the custom attributes are added to the template file.
5. Save the file to the computer, then open the file, enter sample data in the **Sample Details** tab.
6. When you are finished adding sample information to the file, save the file.

You can now import the file to Genexus™ Dx Software. For more information, see “Import samples” on page 29.

## Add a custom sample attribute (manager/administrator)

Samples attributes in Genexus™ Dx Software are categories of information that describe important aspects of each sample, such as patient ID, date of birth, date of sample collection, gender, or a sample type. The values for each attribute define the data that describes each sample, such as RNA or DNA for the nucleic acid type attribute.

Manager- and administrator-level users can add custom attributes and add or change values of those attributes.

1. To create a custom attribute, navigate to the **Samples►Manage Attributes** screen, then click **+ Add New Attribute**.
2. In the **Add New Attribute** dialog, define the attribute in the **Attribute Name**, **Attribute Category**, and **Data Type** fields, then click **Save**. The user can also determine if the attribute is required or optional.

Custom and predefined sample attributes can be viewed in the Samples table. Each column in the Samples table represents a single sample attribute.

For more information, see “Create a new sample” on page 28, “Create multiple samples” on page 28, “Import samples” on page 29, “Edit a sample” on page 38, and “System-installed sample attributes” on page 218.

# Sample tags

A sample tag is a label that you add to a sample to help you group and track samples in Genexus™ Dx Software. For example, you can assign a tag to each sample in a study or project so that you can easily find that group of samples in the software. A manager- or administrator-level user firsts creates a tag, then any user can assign that tag to a single sample or a group of samples. You can assign multiple tags to each sample.

You can assign a tag when you create a sample or you can add, edit, or remove the tags that are assigned to a sample later. You can apply sample tags to multiple samples at one time. Later, you can filter the table of samples with that tag to list only the samples that have the tag. The tags that you assign to samples are also shown in the list of sample results so sample tags can be used to group and find sample results that are part of a study or project.

If you edit a sample and make a change to the tags associated with the sample, the changes you make are tracked in the software and can be reviewed. For more information, see “Edit a sample” on page 38 and “Review sample edit history” on page 39.

Tags are preserved with the sample when you archive a run that includes a sample with one or more assigned tags.

You can also add tags to sample results. For more information, see “Assign sample tags to a sample result” on page 100 and “Assign sample tags to multiple sample results” on page 101.

## Create a sample tag (manager/administrator)

A manager-or administrator-level user can create sample tags in Genexus™ Dx Software that can be used to label and group samples.

You can create parent tags to nest the labels if desired. For example, if a large project is divided into sections, you can create a parent tag called *Master project* and a child tag called *New variant of concern*. You can create multiple layers of nested tags to help sort projects and sections within projects. When you assign nested tags to a sample, the parent tag and all child tags are listed as tags for the sample. Later, when you search or filter for nested tags, the parent tags are also shown to help you group and track the samples.

1. In the menu bar, click **Samples**►**Manage Samples**.

   Alternatively, you can create a tag when you create a sample. For more information, see “Create a new sample” on page 28 and “Import samples” on page 29.

2. In the **Manage Samples** screen, click **Manage Tags**.
   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.

3. Click ＋ **Create Tag**.

4. In the **Create Tag** dialog box, enter a name for the tag.

   - Select a **Parent Tag** from the list to create a hierarchical tag that is nested below the parent tag, if desired.

5. Click **Save**.

The tag is available to assign to samples.

## Assign sample tags

You can assign tags to samples in Genexus™ Dx Software to label and group samples.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample that you want to tag, then click **Assign Tags**.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
   If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click ＋ **Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the sample, then click **Save**.

The tag is assigned to the sample and is listed in the row for the sample in the samples table.

## Assign a sample tag to multiple samples

You can assign tags to multiple samples at once in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, click the checkbox to the left of each sample of interest, then click **Assign Tags** above the list of samples.
   **Assign Tags** is not shown until you select at least one sample.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.
   If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click ＋ **Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the samples, then click **Save**.

The tag is assigned to the selected samples and is listed in the row for each sample in the samples table.

## Change or remove the tags that are assigned to a sample

You can change or remove the tags that are assigned to a sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Assign Tags**.
3. In the **Assign Tags** dialog box:
   - Deselect the checkbox next to each tag that you want to remove from the sample.
   - Select the checkbox next to each tag that you want to add to the sample.
4. Click **Save**.

## Edit a sample tag (manager/administrator)

A manager-or administrator-level user can rename a tag and change the parent tag associated with a tag in Genexus™ Dx Software to change the hierarchy of the tags. When a tag is edited, the change is applied to all samples assigned with the tag.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, click **Manage Tags**.
   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click **(Edit)**.
4. Change the name of the tag and parent tag assignment if needed, then click **Save**.
5. In the confirmation dialog box that appears, click **Yes** to confirm that you want to edit the sample tag.

All samples that have the tag that you edit are updated with the change. If the name of the tag is changed, the new name is shown for all samples and sample results with the tag. If the hierarchy of the tag is changed, that change is also applied to all samples and sample results with the tag.

## Delete a sample tag (manager/administrator)

A manager-or administrator-level user can delete sample tags in Genexus™ Dx Software. When a tag is deleted, the tag is also removed from any samples assigned with the tag. Additionally, when you delete a parent tag, all tags that are nested under the parent tag are also deleted.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, click **Manage Tags**.
   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click **Delete**.
4. In the **Confirm** dialog box, click **Yes** to confirm the changes.

The tag is no longer listed in the **Manage Tags** list and is removed from all samples and sample results.

# Manage samples

You can find tools for creating, searching, sorting, editing, deleting, and exporting samples, and for viewing the sample history in the **Samples / Manage Samples** screen.

## Copy a sample

You can copy a sample and change the information for the sample, if needed, to create a new sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to copy, then click **Copy**.
   The **Copy Sample** screen opens. The sample information for the source sample is shown. The **Sample Name** for the new sample is the sample name of the sample that was copied, appended with **_Copy**.
3. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample name, if needed.
4. Click **Save**.

The new sample is listed in the **Manage Samples** table and is available to assign to a run plan.

## Copy multiple samples

You can copy multiple samples at once in Genexus™ Dx Software. You can only copy multiple samples that have not been assigned to a run plan. To copy samples that have been assigned to a run plan, you must copy one sample at a time. For more information, see “Copy a sample” on page 36.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
   The actions that you can apply to multiple samples appear.
3. Click **Copy**.
   The **Copy Sample** screen opens. The sample information for each source sample is shown. The **Sample Name** for each new sample is the sample name of the sample that was copied, appended with **_Copy**.
4. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample names, if needed.
5. Click **Save**.

The new samples are listed in the **Manage Samples** table and are available to assign to a run plan.

## Export samples

The **Export** function generates an XLS file that contains details about the selected sample.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to export. To select all samples, select the checkbox in the column heading row.
3. Click **Export**.
   An XLS file is created that contains the details of the selected samples. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the XLS file in an appropriate viewer to review or print.

## View notes or add a note to a sample

You can view existing notes or add new notes to a sample in Genexus™ Dx Software. Use notes to capture time-stamped written comments and observations for a sample. A sample can contain multiple notes from different users.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.

| Action | Description |
|---|---|
| View existing notes for a sample. | In the **Add Notes** dialog box, notes that are associated with the sample are listed by user, date, and time. |
| Add a new note to a sample. | a. In the **Add Notes** dialog box, click **Add Note**.<br>b. Enter the note, then click **Save**.<br>The note is added to the sample and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time. |

## Delete a sample note

You can delete notes that have been added to a sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.
3. In the **Add Notes** dialog box, in the row of the note that you want to delete, click **(Delete)**.

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

## Edit a sample

You can edit the information that describes a sample in Genexus™ Dx Software. When you edit a sample, you can change the application category, update sample data, and remove data that are not required.

You can edit a sample until a run that uses that sample is started. You cannot edit a sample during a run. After a run is complete, you can edit a sample. However, when you edit a sample after the run is complete, the sample status for the result is changed to obsolete and the report and other files associated with the sample are no longer available.

If you edit a sample after the run and the sample result is obsolete, you must reanalyze the sample to generate sample results and a new variant report for the sample. For more information, see “Reanalyze a run” on page 117.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to edit, then click **Edit**.
3. In the **Edit Sample** screen, modify the sample information.
   See “System-installed sample attributes” on page 218 for more information about system-installed sample attributes.
4. Click **Save**.

## Edit multiple samples

You can edit multiple samples at once in Genexus™ Dx Software.

You can edit samples until a run that uses that sample is started. You cannot edit samples during a run. After a run is complete, you can edit samples. However, when you edit samples after the run is complete, the status for the sample results is changed to obsolete and the report and other files associated with the samples are no longer available.

If you edit samples after the run and the sample results are obsolete, you must reanalyze the samples to generate sample results and a new variant report for the samples. For more information, see “Reanalyze a run” on page 117.

Only samples that have the same sample application category can be edited together.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
   The options that you can apply to multiple samples appear.
3. Click **Edit**.
4. In the **Edit Sample** screen, modify the sample information.
   For more information about edit options, see “Create multiple samples” on page 28.
5. Click **Save**.

The changes to the samples are saved and the edited samples are available to assign to run plans.

You can view a history of changes that have been made to each sample. For more information, see “Review sample edit history” on page 39.

## Review sample edit history

The entire history of defining a sample is available to review and export in Genexus™ Dx Software. The history shows the original sample values and any new values or activities. The following actions generate an audit record:

- Create a new sample
- Edit a sample, including changes to sample tags
- Delete a sample

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample of interest, then click **Audit**.
3. In the **Audit Details** screen:

| Option | Description |
|---|---|
| Review the audit trail details. | Each action that was performed for the sample, such as sample creation or edits, is listed. |
| Export the audit record. | Click **Download** to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file. |
| Close the **Audit Details** screen. | Click **Back** to return to the list of samples. |

## Delete samples

You can delete samples that have not been assigned to a run. Samples that are assigned to a run or library batch cannot be deleted.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to delete.
   The options that you can apply to multiple samples appear.
3. Click **Delete**.
   The **Delete Sample** dialog box opens with the message ***Are you sure that you want to delete selected sample(s)?***
4. Click **Yes** to delete the selected samples.

The samples are removed from the software.

# Enter and manage library batches

You can prepare a library batch manually or import library batch information from a file. To change sample or library batch information, you can edit the information in the samples, update library batches, or delete samples or library batches.

## Prepare or import a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Dx Software from samples. You can enter samples in the software or import a sample file. For more information see, “Enter and manage samples” on page 23.

Select the library batch when you plan the run. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 51.

---

Note:

- Each library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

---

### No-template controls

A no-template control (NTC) sample and a positive sample control are included in runs that are planned in Genexus™ Dx Software. The controls are added automatically after you assign samples to a run plan, or when you create a library batch. The configuration of each control is based on the assay that is used. For more information, see the user guide for the panel that you use.

### Prepare a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Dx Software from samples that you have added. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 51.

Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software to prepare the library batch and verify that the barcode scanner is compatible with the computer.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown menu, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

   For more information about how to filter the list of samples, see “Find samples” on page 24.

3. Select samples in the list by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.

   Note: Two control samples are included in each run. The controls are added automatically to a run plan after you assign one or more samples, or to a library batch when you create the library batch.

4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** dropdown menu.

   The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library preparation type, the **Library Prep Type:** automatically fills with the library preparation type for the nucleic acid type that is specified by the assay you selected. For example, **DNA, RNA, or DNA+RNA**.

   The assay determines parameter selections for the run, including NTC and Positive Controls samples for each library batch, and post-run data analysis settings. If you select an assay that includes one library prep type, the library preparation type for the assay listed in the **Library Prep Type:** automatically fills for the nucleic acid type specified by the assay you selected: **DNA, RNA, or DNA+RNA**

5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.

   Note: Library Batch IDs can contain only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).

6. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.

7. Select the barcode ID of the adapter used to prepare each library. If appropriate, swap the default barcodes in the dialog box between DNA, RNA, and Fusions by clicking the **Swap Barcodes** swap image.

   For example, click DNA ⇄ Fusions to swap barcodes.

   Each library in a library batch must have a different barcode ID. When preparing the physical libraries, best practice is to swap barcodes between DNA and RNA libraries in consecutive sequencing runs to prevent carryover contamination. The barcodes that are listed in the **DNA Barcode** or **RNA Barcode** dropdown list belong to the barcode set that was selected when the assay was created.

   IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.

8. *(Optional)* Enter the **Input Quantity** of each DNA, RNA, or DNA+RNA library.

9. Use the connected barcode scanner to scan the barcode from the control kit into the fields in the **DNA Kit Barcodes**, and **RNA Kit Barcodes** columns for the positive controls and the no-template controls, and for each sample, if applicable.

   If needed, you can enter the barcodes manually.

10. Click **Submit** to save and submit your selections.

    The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Import a library batch

You can import library batch information in the form of an XLS or XLSX file. The import file must include all required library and kit information.

1. In the menu bar, click **Samples**►**Manage Library Batches**.
2. In the **Manage Library Batches** screen, click **Import Library Batch**.
3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.

   If the assay includes more than one library preparation type, a library preparation type dropdown list appears.
4. Select the library preparation type, such as **DNA, RNA, or DNA+RNA** for the library batch.
5. Select an assay from the list, then click **Download**.

   The assay name is auto-populated in the Microsoft™ Excel™ template file that downloads to your drive.
6. In the template file, enter or confirm the library batch information.

| Template item | Description |
|---|---|
| Reagents tab | |
| Assay Name | Auto-populated when assay is selected in step 5 (required). |
| Extraction Method Type | Auto-populated when assay and library prep type, if applicable, is selected in step 5 (required). |
| Library Batch ID | Must be alphanumeric (0–9, Aa−Zz), period (.), underscore (_), and hyphen (-) (required). |
| Library Kit Barcode | For example, Genexus™ Dx Library Strips 1 and 2-HD barcode (optional). |
| Panel Kit Barcode | For example, Generic Assay GX v2 barcode (optional). |
| Libraries tab | |
| Sample Name | Must be alphanumeric (0–9, Aa−Zz), period (.), underscore (_), and hyphen (-) (required). |
| Barcode | Barcodes used for each sample and control library preparation (required). |
| Nucleic Acid Type | DNA or RNA (required). |
| Input Quantity | Library input quantity (optional). |
| Control Kit Barcode | The barcode for the Positive Control and NTC kits used for Nucleic Acid to Result runs (required). |
| Extraction Kit Barcode | The barcode for the kit used for nucleic acid extraction of a sample (required). |

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA library must be listed in sequential order per pool for each sample. For example, for a 1-pool DNA and Fusions assay, order should be DNA, RNA for sample 1, DNA, RNA for sample 2. For a 2-pool DNA and Fusions assay, library order should be DNA, RNA (pool 1), DNA, RNA (pool 2) for sample1, then DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 2.

7. Save the file.
8. In the **Import Library Batch** dialog box, click **Select Library batch file**, navigate to the saved file, then select it.
9. Click **Upload**.
   A progress bar followed by an import report displays. If the import process fails, click **View errors** to review the reason for failure. The use of an invalid character is an example of an error. For more troubleshooting support, see “Library batch import fails” on page 223.

## Find library batches

You can sort, search, and filter information to narrow the library batches that are listed in the **Samples / Manage Library Batches** screen in Genexus™ Dx Software. The sort, search, and filter tools help you organize the library batches and find a specific library batch.

For information to search for a library batch by a unique identifier, such as the library batch ID, see “Search for library batches” on page 43.

For information to filter libraries by a common trait, such as the type of nucleic acid in the library batch, see “Filter the list of library batches” on page 45.

### Search for library batches

Library batches are listed in the **Samples / Manage Library Batches** screen in Genexus™ Dx Software. The most recently created library batch is at the top of the list.

You can search for library batches with full or partial keywords of the following items that are associated with the library batches.

- **Library Batch ID**
- **Assay Full Name**
- **Nucleic Acid Type**
- **Sample Name**

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, in the column heading of interest, click **(Filter)**.
3. In the search field, enter a full or partial name or ID of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
4. In the filter dialog box, select from the limited list.
   - Select a checkbox next to the name or ID to select one or more items.
   - Click **Select All** to select all of the items listed in the filter dialog box.
   - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
   In the column heading, changes color to indicate that a filter is applied.

The filtered list of libraries is shown in the **Manage Library Batches** screen.

Click **Clear Filters** to remove all filters and view the full list of library batches.

## Sort the list of library batches

You can sort the list of library batches to make it easier to find a library batch in Genexus™ Dx Software.

By default, library batches are listed in the **Samples / Manage Library Batches** screen by the date the library batch was created. The most recently created library batch is at the top of the list.

You can change the order that the library batches are listed and sort the library batches alphabetically or reverse alphabetically by the **Library Batch ID** or **Assay Full Name**.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, click the column heading of interest.
   The list of library batches is sorted alphabetically.
3. Click the column heading again to reverse the order.

## Filter the list of library batches

You can apply filters to narrow search results, or shorten the list of library batches when you view the list of library batches in Genexus™ Dx Software.

You can also search for library batches with keywords or text. For more information, see “Search for library batches” on page 43.

In addition to the filter tools in the column headings, you can also filter the list of library batches with a dropdown list. For more information, see “Manage library batches” on page 46.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, select one or more filters.

| Option | Description |
|---|---|
| List all library batches that are associated with a sample | In the **Total Samples** column heading, click **(Filter)** then select one or more samples of interest. |
| Filter and search by the **Library Batch ID** or other column headings. | In the **Library Batch ID** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter by the type of nucleic acid in the library. | In the **Nucleic Acid Type** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter by the full assay name in the library batch. | In the **Assay Full Name** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter the list of all samples that are included in library batches. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column heading, changes color to indicate that a filter is applied.

The list of library batches is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of libraries.

## Manage library batches

You can view library batch information, edit, delete, add notes, and review the history of changes to library batches in Genexus™ Dx Software.

You can also search, sort, and filter the list of libraries batches. For more information, see “Find library batches” on page 43.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, perform one or more actions.

| Option | Description |
|---|---|
| View more library batch information, such as the list of samples included in the library batch. | Click a Library Batch ID in the library batches list. Alternatively, in the Total Samples column, click the number of samples in the library batch.<br>In the pane that shows details for the library batch, you can click ••• (More Options) to perform more actions, such as edit, audit, or add notes to the library batch. |
| View all library batches. | In the Filter Library Batches By dropdown menu, select All. |
| View libraries that are not in a run plan. | In the Filter Library Batches By dropdown menu, select To Be Planned. |
| View multiple pages of libraries. | Click the page numbers and arrows at the bottom of the screen. |
| Delete a library batch. | Select the checkbox next to one or more library batch IDs, then click Delete. |
| Edit a library batch. | 1. Place the pointer over the row of a library batch that you want to edit, then click Edit.<br>2. Modify the information in the Edit Library batch screen, then click Submit.<br>Alternatively, in the library batch details pane, click the library batch ID to perform this action.<br>You can edit a library batch only if none of the libraries in the library batch have been assigned to a run. |
| View or export the audit trail of a library batch. | 1. Place the pointer over the row of a library batch that you want to audit, then click Audit.<br>The Audit Records screen opens with time-stamped audit records that include details and changes for each action performed on the library batch.<br>2. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions.<br>3. Click Download in the upper right corner of the screen to download the record as a PDF file. |

*(continued)*

| Option | Description |
|---|---|
| View, add, or delete notes for a library batch. | 1. Place the pointer over the row of a library batch of interest, then click Notes.<br>The Add Notes dialog box opens and notes that are associated with the library batch are listed by user, date, and time.<br>2. In the Add Notes dialog box, click + Add Note.<br>3. Enter the note, then click Save.<br>4. If needed, in the Add Notes dialog box, in the row of the note that you want to delete, click (Delete). |

# 4 Plan and manage runs


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Runs created in Genexus™ Dx Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes. Runs are used to track samples, consumables, and chips throughout the library preparation, templating, sequencing, and data analysis workflow.

There are two types of runs that you can plan in Genexus™ Dx Software.

Types of runs in Genexus™ Dx Software

| Run type | Description |
|---|---|
| Nucleic Acid to Result | A sequencing run that uses nucleic acid samples as input. |
| Library to Result | A sequencing run that uses libraries that you have previously prepared as input. |

Genexus™ Dx Software guides you step-by-step through the process to set up a run. The software prompts you to select required information and consumables, then provides a printed run setup guide to help you load consumables on the Genexus™ Dx Integrated Sequencer.

You can plan sequencing runs that use nucleic acid samples with a **Nucleic Acid to Result** run or that use manually prepared libraries with a **Library to Result** run.

## Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Dx Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter and manage samples” on page 23.
- Ensure that the following conditions are met for a **Library to Result** run.
  - Library batches are prepared and each batch uses a unique library batch ID.
  - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

  For more information, see “Enter and manage library batches” on page 40.

The software returns an error message when any of the conditions are not met.

# Find a run plan

You can sort, search, and filter information to narrow the run plans that are listed in the **Runs / Manage Runs** screen. The sort, search, and filter tools help you organize runs and find a specific run plan in Genexus™ Dx Software. You can also apply the search, sort, and filter tools in the **Assays** step when you plan a run. Use the tools to limit the assays that are listed so that you can easily identify one or more assays to include in the run plan.

For information to search for a run plan by a unique identifier, such as the name of the run plan or the assay applied to the run plan, see “Search for a run plan” on page 49.

For information to filter run plans by a common trait, such as run status, see “Filter the list of run plans” on page 51.

## Search for a run plan

Run plans are listed in the **Runs / Manage Runs** screen in Genexus™ Dx Software. The most recently created run plan is on the top of the list.

You can search for run plans with keywords or text that are included in the name of the run plan or the name of an assay applied to the run plan.

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IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, in the **Run Name** column heading or **Assay Name** column heading, click **(Filter)**.
3. In the search field, enter a full or partial assay or run name of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.

4. In the filter dialog box, select from the limited list.
   - Select a checkbox next to a run or assay name to select one or more items.
   - Click **Select All** to select all items listed in the filter dialog box.
   - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
   In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of run plans is shown in the **Manage Runs** screen.

Click **Clear Filters** to remove all filters and view the full list of run plans.

## Sort the list of run plans

You can sort the list of run plans to make it easier to find run plans in Genexus™ Dx Software.

By default, run plans are listed in the **Runs / Manage Runs** screen by the date the run plan was created. The most recently created run plan is at the top of the list.

You can change the order that the run plans are listed and sort the run plans alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, click the **Run Name** heading.
   The list of runs is sorted alphabetically.
3. Click the column heading again to reverse the order.

## Filter the list of run plans

You can apply filters to narrow search results, or shorten the list of run plans when you view the list of run plans in Genexus™ Dx Software. You can also apply filters in the **Assays** step when you plan a run to help find one or more assays to include in the run plan.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the run name or the assay selected in the run. | For more information, see “Search for a run plan” on page 49. |
| Filter by the number of samples in the run. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run. |
| List runs based on run status. | In the **Run Status** column heading, click **(Filter)** then select a status.<br>• For example, select **Planned** to list all runs that are planned. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The list of run plans is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of run plans.

# Plan a Nucleic Acid to Result run

You can plan a run for sequencing that starts with nucleic acid samples. A run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into steps: **Setup**, **Assays**, **Samples**, **Sample Plate**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Nucleic Acid to Result** screen.

Ensure that the following prerequisites are complete before you plan a **Nucleic Acid to Result** run.

- Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software and verify that the barcode scanner is compatible with the computer.
- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 28, or “Import samples” on page 29.
- Ensure that the assay that you want to use in the run exists in the software. For more information, see “Assays in Genexus™ Dx Software” on page 139.
- Quantify sample concentration, if you are not diluting sample concentrations manually to the target concentration of the assay. For more information, see the user guide for the panel that you use (Pub. No. ).

1. In the menu bar, click **Runs**►**Nucleic Acid to Result**.

   Note: Alternatively, you can click + **Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.

   b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.

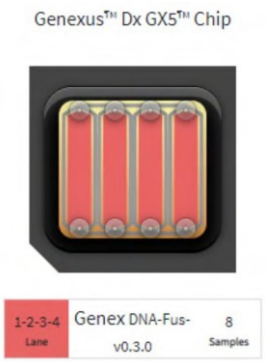
   c. Click **Next**.

3. In the **Samples** step, select the samples that you want to run with each research application of each assay.
   a. Select the checkbox next to each sample that you want to assign to the research application of an assay, then in the **Selected Assays** pane, for the assay and research application that you want to use for the selected samples, click **Assign**.

      The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including a no-template control, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

      If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green.

The run is allowed if the lane capacity is exceeded, but you may not obtain the reads needed per sample to pass QC metrics.




Green lane color denotes lane usage and sample assignment within lane capacity. Red lane color denotes sample assignment that exceeds lane capacity.

b. If needed, edit samples in one of the following ways.
   - Click **View & Remove**, make the selections, then click **Update**.
   - Click **Remove All**, to remove all sample assignments for all assays.

c. Click **Next**.

4. In the **Sample Plate** step, review position assignments in the sample plate. Drag-and-drop samples and no-template controls to edit the location of samples and controls, if applicable.
   a. Use the connected barcode scanner to enter the kit barcode for one or more samples or controls. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples or controls, then click **Assign Kit Barcodes**. In the **Assign Kit Barcodes** dialog box, enter the kit barcode.

      If needed, you can enter the barcodes manually.

   b. Modify the concentration of samples, if needed. For a single sample, in the row of the sample of interest, in the **Conc. (ng/µl)** column, edit the concentration.

      To modify the concentration of multiple samples, select the samples of interest, then click **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration for all selected samples, then click **Submit**. The concentration for each selected sample is updated to the new value.

      If a sample concentration is ≤1,024X the target concentration for the assay, which is displayed as a default value for each sample in the **Sample Plate** screen, the sequencer automatically dilutes the sample to the target concentration during the run. If a sample concentration is

greater than this value, you need to manually dilute the sample to the target concentration, or to a value within range for automated dilution before loading on the sample plate.

Note: The sample volume that is required for library preparation is not adjustable. The volume depends on the number of primer pools in the assay, sample type, and library chemistry.

IMPORTANT! A warning symbol (⚠) with a message that states "Sample concentration out of range" can be shown in error for automated sample dilution in the **Run Plan** screen when you plan a run, or view the **Run Summary** screen, in Genexus™ Dx Software.

- If the sample concentration is below the required minimum concentration and shows a **Dilution Factor** of 1.00 (that is, no dilution), the message is valid.
- If a sample that includes the warning message shows a **Dilution Factor** >1.00 and <1,024.00, the message is *not valid* and can be ignored.

c. Ensure that sample plate information is correct, then click **Next**.

5. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.

Note: To print the run setup guide, you need to allow pop-ups in the browser.

The run plan summary lists the following details.

- the consumables that are required for this run
- how much sample volume to load
- where to load samples and primer pool tubes
- details about the assay.

After saving, the run appears in the **Manage Runs** screen in the run list with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

When the sequencing run is complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the user guide for the panel that you use (Pub. No. ).

# Plan a Library to Result run

You can plan runs for sequencing that start with libraries. A run that starts with prepared libraries is called a **Library to Result** run. Before you can plan a **Library to Result** run, you must first enter sample information and prepare a library batch that associates an assay with the library batch. The library batch must be created with the assay to be used in the run. The assay specifies the barcode set that was used to prepare the sample libraries.

For more information, see “Enter and manage library batches” on page 40.

Genexus™ Dx Software guides you through the four steps of planning a **Library to Result** run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs►Library to Result**.

   Note: You can also click **+ Library to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter a name for the run, then configure the reporting options.
   a. In **Run Name**, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.

   b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.

   c. Click **Next**.

      If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Library Batches** step, select the library batch that you want to use in the run, then click **Next**.

   Note: Only one library batch can be selected per assay. However, you can plan a library run that uses multiple assays if you select multiple compatible assays in the **Assays** step.

   The **Chip View** updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum reads per sample entered at assay setup.




Green lane color denotes lane usage and sample assignment within lane capacity. Red lane color denotes sample assignment that exceeds lane capacity.

If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the **Chip View** updates and shows the lane color as red instead of green. In the example shown at right, eight samples were included in a library batch instead of five. The run is allowed if the lane capacity is exceeded, but you may not obtain the required reads per sample to pass QC metrics.

4. In the **Review** step, review the run plan summary.
   - Click **Save and Print** to print the run setup guide.
   - Click **Save** to save the run without printing.

   Note: To print the run setup guide, you need to allow pop-ups in the browser.

   The run plan summary lists the consumables that are required for the run, where to load the library batch on the sample plate, and how much library volume to load.

   The run appears in the run list on the **Manage Runs** screen with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

# Find the assay that you want to use in a run plan

You can search and filter the list of assays when you plan a run to help you find the assay that you want to use in a run plan in Genexus™ Dx Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for an assay to use in a run plan” on page 56.

For information to filter assays by a common trait, such as the analysis version, see “Filter the list of assays shown when you plan a run” on page 57.

## Search for an assay to use in a run plan

You can search for an assay with keywords or text that are included in the name or full name of an assay to help find the assay that you want to use in a run plan.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

1. In the menu bar, click **Runs**, then click **Nucleic Acid to Result** or **Library to Result**, based on the type of run that you want to plan.
2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.
3. In the **Assays** step, in the **Assay Full Name** column heading, click **(Filter)**.
4. In the search field, enter a full or partial assay name of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
5. In the filter dialog box, select assays from the limited list.
   - Select a checkbox next to an assay name to select one or more assays.
   - Click **Select All** to select all of the assays in the filter dialog box.
   - Click **Clear** to close the filter dialog box.
6. Click **Filter**.
   In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of assays is shown in the **Assays** step. You can select the assay of interest from the limited list and proceed through the remaining steps for the run plan. For more information, “Plan a Nucleic Acid to Result run” on page 51, or “Plan a Library to Result run” on page 55.

Click **Clear Filters** to remove all filters and view the full list of assays.

## Filter the list of assays shown when you plan a run

When you plan a run in Genexus™ Dx Software, you can apply filters to narrow the list of assays that are shown to help find the assay that you want to use in the run plan.

1. In the menu bar, click **Runs**, then click **Nucleic Acid to Result** or **Library to Result**, based on the type of run that you want to plan.
2. Enter and make the selections in the **Setup** step, if applicable, then click **Next**.

3. Select from the filter and search options in the column heading of interest.

| Option | Description |
|---|---|
| List assays according to the analysis version | a. Click ▼ **(Filter)** in the **Analysis Version** column.<br>b. Select one or more of the versions that are shown, then click **Filter**. |
| List assays according to chip type | a. Click ▼ **(Filter)** in the **Chip Type** column.<br>b. Select **Ion Torrent™ GX5™ Chip** then click **Filter**. |
| List assays according to library chemistry | a. Click ▼ **(Filter)** in the **Library Chemistry** column.<br>b. Select the library chemistry:<br>• **AmpliSeq HD** – High sensitivity library preparation chemistry for targeted next-generation sequencing with unique molecular tags to reduce the number of false positives and improve confidence in variant calling.<br>c. Click **Filter**. |

4. *(Optional)* Click **Clear Filters** to return to the entire list of assays.

5. In the shortened list of assays, select one or more checkboxes in the **Application Type** column for each assay that you want to add to the run, then click **Next.**

6. Click the **Filter Assays By** dropdown menu, if applicable, then select **Assigned** to list only the assays that you have selected for the run plan.
   - Click **Filter Assays By►All** dropdown menu, to show all assays again, if desired.

You can proceed through the remaining steps for the run plan. For more information, see “Plan a Nucleic Acid to Result run” on page 51 or “Plan a Library to Result run” on page 55.

# Manage runs

Run plans are listed in the **Runs / Manage Runs** screen. The most recently created run is on the top of the list. Only runs that have not started are listed.

In this screen, you can:

- Plan a run.
- Sort and search the list of run plans.
- Review and print a run plan.
- View the audit trail of a run plan.
- Edit a run plan.
- Delete a run plan.
- View, add, or delete the notes for a run plan.

# View a run plan

You can view detailed run plan information in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, click the run name of interest.
   Run plan details are shown in the run plan summary pane. For more information, see “Run plan details” on page 59.
3. When you are finished viewing the run plan details, click another run name to view details for another run plan, or click ✕ **(Close)** to return to the **Manage Runs** screen.
4. *(Optional)* Click **Print** to print the information about the run.
   To print the run plan, you must allow pop-ups in the browser.

You can click ••• **(More Options)** to perform more actions, such as edit, add notes, or audit the run plan. The actions for each run plan are also available in the **Manage Runs** screen when you place the pointer over the row of a run name. For more information, see “Edit a run” on page 61, “Add a note to a run” on page 62, “Review and download a run history” on page 60.

## Run plan details

Detailed run plan information is shown in the run plan summary pane.

| Run plan detail | Description |
|---|---|
| Generate Report | Indicates if a report is automatically generated for the run. |
| Run Status | The run status. For example, Planned. |
| Run Start Date | This information is available when the run is started. |
| Run Completion Date | This information is available when the run is completed. |
| Sequencing[1] | |
| Consumables Required For This Run | A list of the required consumables, for example, the chip and barcodes. The quantity of each consumable is shown in parentheses before the name of the consumable. |
| Consumables Installed Ready To Use | A list of consumables installed on the sequencer and available for the run. |
| Primer Tube Positions | An image that shows the locations of the primer pool tubes. |
| Sample Locations | Detailed sample information and an image that shows sample locations on the 96-well plate. For more information, see “The run summary” on page 103. |

*(continued)*

| Run plan detail | Description |
|---|---|
| Assays | |
| Assay | Detailed assay information for each assay in the run. For more information, see “The run summary” on page 103. |

[1] The Sequencing section is collapsible and expandable.

## Print a run plan

You can print detailed run plan information in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, click a run name in the list.
3. Click **Print** to print the information about the run plan.
   To print the run plan, you must allow pop-ups in the browser.

Run plan details are shown in the run plan summary pane. You can review the information that was entered or generated when you created the run. For more information, see “Run plan details” on page 59.

## Review and download a run history

The entire history of a run plan is available to review, export, or print in Genexus™ Dx Software. The history shows the original run plan values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Audit**.
   The **Audit Details** screen opens with time-stamped audit records that include run plan details and changes for each action performed on the run plan. Details for the most recent action are shown.
3. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions.

4. Review the audit record.

   Details in the audit record

| Item | Description |
|---|---|
| User name | The name of the person who was signed in when the action was performed. |
| Run Plan | The name of the run plan. |
| Data Object Name | The data object that is affected by the action. |
| Action Performed | Actions performed on the run plan include:<br>• Create<br>• Edit |
| Time Stamp | The time and date of the action. |
| ID | An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier. |
| New Values | A list of all values in the run plan. If a value changed as a result of the action, the new value is shown. |
| Old Values | A list of the previous values in the run plan. |

5. Click **Download** to generate a PDF file, if needed.

   Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.

6. Click **Back** to return to the list of runs.

## Edit a run

You can edit runs that have a run status of **Planned**. Parameters that can be edited include reporting options, assays used in the run, samples included in the run, chip assignment, and sample concentration.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Edit**.
3. In the **Edit Sample Run** workflow, change the desired settings by navigating through the steps clicking **Next**, then click **Save**.

   For more information about the settings, see “Plan a Nucleic Acid to Result run” on page 51 and “Plan a Library to Result run” on page 55.

## Delete a run

You can delete runs from the software.

You must select at least one run in the **Manage Runs** screen before the option to delete a run appears.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, select one or more runs to delete by selecting the checkbox next to the run name.
   Select all of the runs in the screen by selecting the checkbox above the column.
   If needed, click **Clear** to deselect all of the runs.
3. Click **Delete**.
4. In the **Delete Plan** message box, click **Yes** to delete the selected run.

## Add a note to a run

You can add a note or notes to **Nucleic Acid to Result** and **Library to Result** runs that have a run status of **Planned** in Genexus™ Dx Software. A note is any written comment or observation that you want to add to the run.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, click **Add Note**.
4. In the **Add Notes** dialog box, enter the note.
5. Click **Save**.

The note appears in the **Add Notes** pane of a selected run in the list of runs.

### View a run note

You can view the notes that have been added to a run. Run notes are time-stamped written notes to capture comments or observations for a run that is planned or completed. A run can contain multiple notes from different users.

You cannot edit run notes. However, you can add multiple notes to a run.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. Review the note or notes information.

## Delete a run note

You can delete a run note in Genexus™ Dx Software.

Notes record any written comments or observations for a run plan.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, in the row of the note you want to delete, click **(Delete)**.

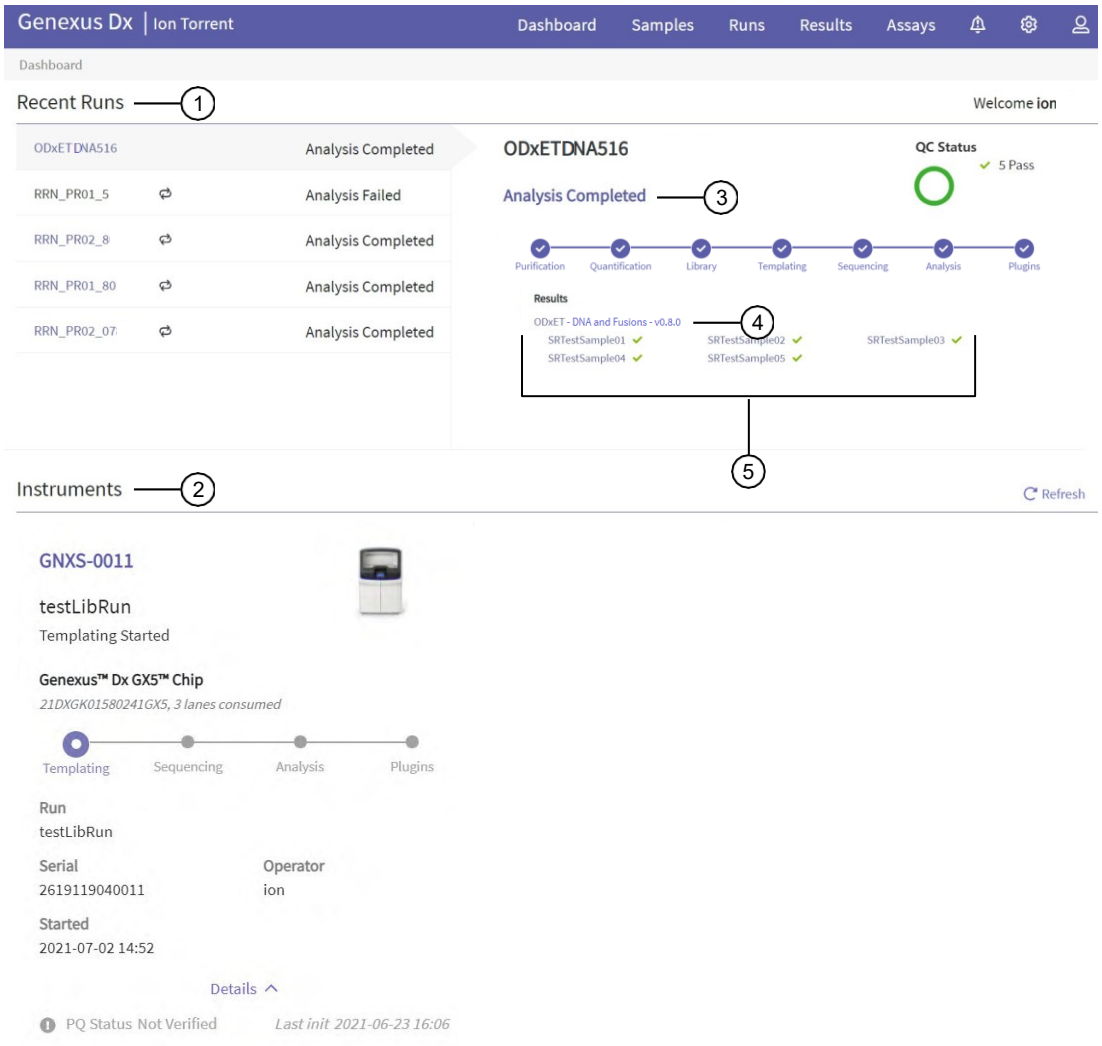
The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

# 5 Genexus™ Dx Software Dashboard


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- Information about connected instruments on the Dashboard ..... 68


In the **Dashboard** screen you can view instrument status and details for recent and current runs.




① The five most recent runs are listed in the left pane.

- You can click the row of a run of interest to view details for the run in the right pane.
- You can click the name of the run to leave the dashboard and view the results for the run.

② Instruments that are integrated with the software are shown.

③ In the details pane on the right, you can place the pointer over the status for the run to view the progress of the run.

④ For the recent run that is selected in the left pane, each assay for the run is shown. You can click the name of an assay to leave the dashboard and open the results for the run and view assay metrics.

⑤ For the recent run that is selected in the left pane, each sample in the run is shown. You can click a sample name to leave the dashboard and open the sample results.

# View run progress on the instrument

You can view details for the five most recent and current runs in the **Dashboard** in Genexus™ Dx Software.

Runs that are reanalyzed are listed with ⟳ **(Reanalysis)** after the run name. For more information, see “Reanalyze a run” on page 117.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

| Option | Description |
|---|---|
| View detailed run result information for completed runs. | In the left side of the screen, click the run name of interest.<br>The **Run Summary** tab opens. For more information, see “The run summary” on page 103.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the quality control information. | In the left side of the screen, click the row of the run of interest.<br>The QC status for the run and for the samples is shown in the details pane in the right side of the screen. For more information, see “QC results” on page 92. |
| View assay-level metrics and results for the run. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, click the assay name of interest.<br>The **Assay Metrics** tab opens. For more information, see “Assay metrics and the run report” on page 106.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the sample results. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, click the sample name of interest.<br>The **Key Findings** tab opens. For more information, see “Key Findings” on page 73.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the progress of the run. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, place the pointer over the status for the run.<br>A progress bar for the run is shown. Steps in the run that are complete are listed with a check mark. Steps that are in progress are shown with a flashing circle. Steps that remain to be completed are listed in grey. If any failures occur, failed steps are also listed in grey. |

Tip: If the recent run includes many samples, limit the samples that are listed. For more information, see “Filter the list of samples shown in the dashboard” on page 67.

## Filter the list of samples shown in the dashboard

You can apply filters to shorten the list of samples shown in the **Dashboard** in Genexus™ Dx Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, click the row of the run of interest. Assays and samples in the run are listed in the details pane in the right side of the screen. In addition, the QC status is shown.
3. In the details pane, filter the list of samples.

| Option | Description |
|---|---|
| Shorten the list of samples. | Click **(Filter)**, select the samples of interest, scroll to the bottom of the filter dialog box, then click **Filter**.<br>**(Filter)** changes color to indicate that a filter is applied. |
| Refine the samples shown by the QC status of the sample result. | In the details pane, in the **QC Status** section, click **Pass** to list only samples that passed QC metrics or click **Fail** to list only samples that failed QC metrics. |

The list of samples is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of samples.

# View the status of an instrument

You can view the status of the Genexus™ Dx Integrated Sequencer with Genexus™ Dx Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Instruments** section, view the status of an instrument.
   The following information is provided for all integrated instruments.
   - instrument name
   - run status of the current or most recent run
   - PQ status
   - **(Alarms)**– instrument alarms, if any
     If the instrument displays one or more alarms, the number of alarms is shown in parentheses. You can place the pointer over **Alarms** to view more information about an alarm.
3. In the **Instruments** section, in an instrument pane, click **Details** to view the current or most recent run name, serial number, the operator, run start and completion times, and instrument alarms, if any.
4. Click **Refresh** to update the information.

# Information about connected instruments on the Dashboard

The following information is available in the **Dashboard** screen. You can expand the **Details** section for each instrument to view more information.

| Item | Description |
|---|---|
| Run | The name of the run that is in progress or the last completed run on the instrument. |
| Serial | The serial number of the instrument. |
| Operator | The name of the operator who started the run on the instrument. |
| Started | The date and time that the run started on the instrument. |
| Last Init | The last initialization completion date and time. |
| PQ Status | Indicates whether the performance qualification (PQ) status of the instrument is Verified or Expired. |
| Next PQ Due | The expiration date of the current PQ verification run. Expiration of the PQ status prevents the instrument from performing a non-PQ run until a valid PQ run has been performed and signed off. |

# 6 Review data and results


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Use the **Results** menu to review results and data analysis, and to perform data management tasks. You can view results sorted by sample or by run.

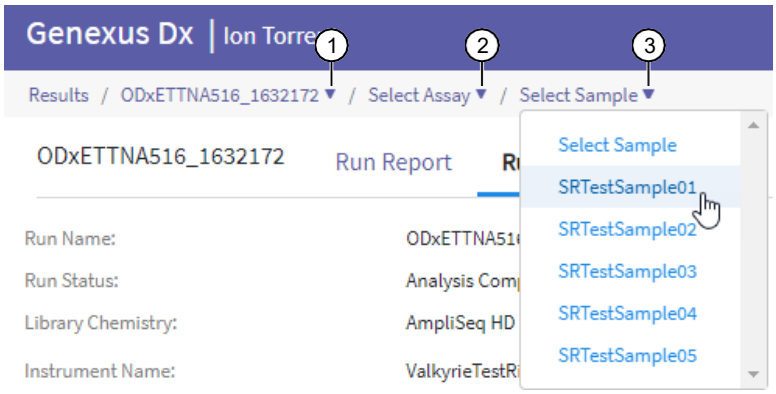
| Selection | Description |
|---|---|
| Click Results►Sample Results | Select this option to review completed sample results and reports. |
| Click Results►Run Results | Select this option to review completed run results and reports by assay. |
| Click Results►Verification Results | Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification. |

## Results navigation bar overview

After you select a run result or a sample result, a results navigation bar appears and allows you to easily toggle between different results screens for each run. You can quickly switch between run-level results, assay-level results, and sample-level results for the run. In addition, you can easily find samples or assays in the run with the search field that appears in each dropdown list.

Assay-level results include assay metrics, such as final read data.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.




| Callout | Dropdown menu | Description |
|---|---|---|
| 1 | Run name | The run name is listed. Multiple runs are listed only if the run has been reanalyzed. For more information, see “Reanalyze a run” on page 117. |
| 2 | Select Assay | You can click the assay name of interest to view assay metrics for the run.<br>You can search for or toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed.<br>You can select Select Assay from the dropdown menu to remove the assay selection. If you remove the assay and sample selections, the Run Summary tab opens with a summary of results for the run selected in the Run Results dropdown menu. Assay metrics are also shown in the Run Report tab. For more information, see “Assay metrics and the run report” on page 106. |
| 3 | Select Sample | You can click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in Select Assay. The search field allows you to find samples in the run.<br>You can select Select Sample from the dropdown menu to remove the sample selection. If you remove the sample selection, the Assay metrics tab for the selected assay opens. If you select Select Assay with Select Sample shown, the Run Summary tab opens. Assay metrics are also shown in the Run Report tab. For more information, see “Assay metrics and the run report” on page 106. |

# Review sample results

You can review completed sample results and reports in Genexus™ Dx Software.

See “Find sample results” on page 95 for information about how to find sample results of interest.

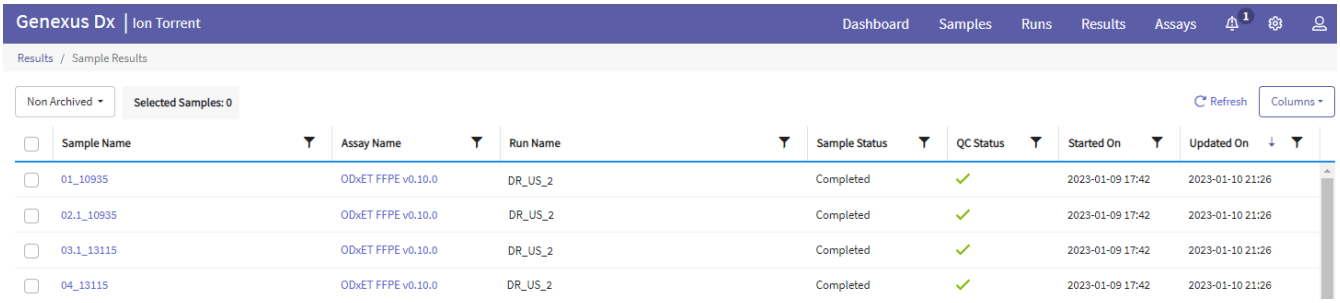
After you open a sample result, you can view different results screens for the run that allow you to toggle between run-level results, assay-level results, and sample-level results. For more information, see “Results navigation bar overview” on page 69.

Assay-level results include assay metrics, such as final read data. Assay metrics are also shown in the **Run Report** tab. For more information, see “Assay metrics and the run report” on page 106.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.

1. Click **Results►Sample Results** to view sequencing results, including variant and fusion calls, for a selected sample.

   In the **Results / Sample Results** screen, samples that have been sequenced are listed by sample name.




   The following information appears in the **Sample Results** screen. You can add columns to the **Sample Results** table to view more information. For example, you can add a column for **Electronic Signatures** to view signature and lock information for variant reports. For more information, see “Customize the samples table” on page 24.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Results screen for the sample. |
| Assay Name | The name of the assay used in the run plan. |
| Run Name | The unique name of the run given when it was created in the software. |
| Sample Status | The status of the run or sample (for example: Completed, Running, Failed, Terminated, Pending, Stalled). |
| QC Status | The QC status of a completed run.<br>• ✔ (Passed) indicates that the sample passed all QC metrics.<br>• ✘ (Failed) indicates that the sample failed a QC metric.<br>• — (Not Calculated) indicates that a sample did not undergo QC analysis. |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |
| Tags | Tags that are assigned to the sample are listed. |

2. In the **Sample Name** column, click a sample name.

   The **Results** screen opens to the **Key Findings** tab, showing the key findings. You can click on other tabs to view QC metrics, variants, and reports. For more information, see “View key findings” on page 72, “View the QC results” on page 91, “View variant results” on page 74, and Chapter 7, “Create, view, and sign reports”.

You can also perform the following actions in the **Sample Results** screen.

| Option | Description |
|---|---|
| Actions | Available action links for a sample are shown when you place the pointer over the row of a sample.<br>• Report—Download the report (available only for samples with a sample status of completed).<br>For more information, see "Variant report" on page 124.<br>• Assign Tags—Add or remove the tags assigned to a sample result.<br>For more information, see "Sample tags" on page 33.<br>• Audit—View the audit trail for the run.<br>For more information, see "Review and download a run history" on page 60.<br>• Notes—View, add, or delete notes for sample result.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>For more information, see "Download a Customer Support Archive" on page 222.<br>• Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step.<br>For more information, see "Reanalyze a sample" on page 118. |
| Actions that you can perform when you select samples | Assign Tags—Assign a tag or tags to selected sample results in one action. The Assign Tags command appears above the Sample Results table after you select one or more samples. |

## View key findings

You can view sample details, biomarker results, and a summary of key metrics for genes that were assayed in a selected run in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.

Review the key findings. For more information, see "Key Findings" on page 73.

## Key Findings

The **Key Findings** table shows details about the sample, a list of biomarkers reported for the sample, and a summary of key metrics for genes assayed in the run.

Note: The **Key Findings** table is by default the first view that is shown for sample results.

The information that is displayed depends on the assay that was used in the run.

Sample Details

| Sample Detail | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Collection Date | The date that the sample was collected. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Sample Type | A term that describes the sample, for example, FFPE, DNA, DNA & RNA |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. "Percent Cellularity" refers to the same measurement as "Percent Tumor Content." |

Biomarker Results

| Result category | Description |
|---|---|
| Variants Detected | Variants that can be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. |

Key Metrics

| Metric | Description |
|---|---|
| Amplicon Summary | |
| Average Base Coverage Depth | The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage. |
| Uniformity Of Base Coverage | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths. |

Key Metrics *(continued)*

| Metric | Description |
|---|---|
| Percent Reads On Target | The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified. |
| Uniformity Of Amplicon Coverage | The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts. |

## View variant results

You can view detailed variant results in the **Variants** table. In the default view, all variants are listed. You can download the information for the variants in XLS or TSV format.

You can also limit the types of variants that are shown and toggle between different variant results: SNVs/Indels, Fusions, and CNVs.

Variant Tables

| Variant type | Description |
|---|---|
| SNVs/Indels | • SNVs: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included.<br>• Indels: insertion and deletion variants.<br>For more information, see “View SNV/INDEL results” on page 76. |
| Fusions | Translocations of genetic material.<br>For more information, see “View fusion results” on page 78. |
| CNVs | Copy number variations (CNVs) are variations of the number of copies of a given gene.<br>For more information, see “View CNV results” on page 83. |

### Search and filter the list of variants

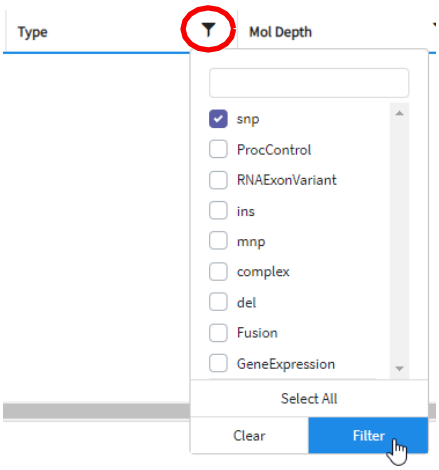
You can search and filter to immediately narrow the list of results that are shown in the variant tables. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name in the **Sample Name** column.
3. In the **Results** screen, click the **Variants** tab.
4. Select the variant class to display the results: **SNVs/Indels**, **Fusions**, or **CNVs**.

5. In the table of variants, in the column heading of interest, click **(Filter)**.
   - In the search field, enter at least 3 characters, then click **Filter**.
   - Select the checkbox in the row of each filter that you want to apply, then click **Filter**.

   The options that are available depend on the column and variant class. For example, you can filter data in the **Type** column to show one specific variant type.

6. Click **Clear Filters** to remove all filters and view the full list of run results.




The column or columns to which you applied a filter change to reflect the filter and selected options.

## Customize the view of the variants table

The **Variants** table includes detailed variant results in Genexus™ Dx Software. You can customize the table to view the information of your choice.

## Variants table column options

When you view the **Variants** table in Genexus™ Dx Software, a default set of columns is included. The table columns are based on various factors, including the samples that are sequenced and the assay used for sequencing.

## Add or remove columns in the variants table

You can add columns to the **Variants** table that you view in Genexus™ Dx Software.

1. In the menu bar, click **Results**▶**Sample Results**.
2. Click a sample name to view results for the sample.
3. Click **Columns**, then select the columns that you want to add to the **Variants** table, or deselect the columns that you want to remove from the table.
   The selected columns are added to the **Variants** table until you close the table.
4. To save the custom table so that the changes to the columns persist the next time the table is viewed, click **Save**.

## Change the width of columns in the variants table

You can change width of columns in the **Variants** table in Genexus™ Dx Software. Not all columns are visible on the screen. You can use the scrollbar under the table to view more columns.

1. In the menu bar, click **Results**▶**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Change the width of each column, as desired.

## Restore default settings for the variants table

You can restore default column headings in the **Variants** table in Genexus™ Dx Software.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click **Columns**►**Restore default**.

The default columns shown in the table are restored.

## View SNV/INDEL results

The **SNVs/Indels** table lists the calls and other information for the SNV and INDEL variants that are analyzed in each sample in a run.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **SNVs/Indels** to view the **SNVs/Indels** table.
   For more information about the data shown in the table, see “SNVs/Indels table” on page 76.
5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the SNV/Indels variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the SNV/Indels variant table, and information for table columns that are available but not currently shown in the SNV/Indels table.

## SNVs/Indels table

The data displayed in the **SNVs/Indels** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 74.

| Column | Description |
|---|---|
| Variant ID | The name of the hotspot as defined in the Browser Extensible Data (BED) file.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 85. |
| Variant Name | The name of the variant. |
| Locus | The chromosome and position of the detected variant. |
| Gene | The gene name. |

*(continued)*

| Column | Description |
|---|---|
| AA Change | Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature. |
| Ref | The reference base or bases at that locus. |
| Alt | The alternate base or bases at that locus. |
| Type | The type of variant that is detected.<br>• snp (single nucleotide polymorphism)<br>• mnp (multi-nucleotide polymorphism)<br>• ins (insertion)<br>• del (deletion)<br>• complex |
| Test Result | Indicates the presence or absence of an SNV/Indel variant.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—Indicates the absence of a variant that differs from the reference allele at a given position. |
| Call Details | The reason why a variant is reported as NO CALL. |
| Phred QUAL Score | The relative probability of either the "reference" hypothesis interval [0,cutoff], or the "variant" hypothesis interval [cutoff,1], Phred-scaled (-10*log10). A higher score means more evidence for the variant call. |
| Raw Read Depth | Total read coverage across amplicon containing SNV/Indel hotspot locations. Count of chip-level reads aligned at this locus that participate in variant calling. |
| Effective Read Depth | The number of reads covering the position. |
| Alt Allele Read Counts | The number of reads containing the alternate allele. |
| Nuc Change | The position and identity of the nucleic acid change. |
| Allele Frequency | The number of variant read counts divided by the total number of read counts for the sample. |
| Allele Frequency (%) | The allele frequency, represented as a percentage. |
| Mut/WT | The ratio of mutant allele to wild-type allele. |
| PPA | Possible Polyploidy Alleles (PPA).<br>A value of Yes indicates variants that are PPA alleles. A value of No indicates variants that are not PPA alleles. |

## View fusion results

You can view calls and other information for the fusions that are analyzed within each sample in a run.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **Fusions** to view the **Fusions** table.
   For more information about the data displayed in the table, see “Fusions table” on page 78.
5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the Fusions variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the Fusions variant table, and information for table columns that are available but not currently shown in the Fusions table.

## Fusions table

The data displayed in the **Fusions** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 74.

| Column | Description |
|---|---|
| Variant ID | The name of the fusion target as defined in the BED file.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 85. |
| Variant Name | The name of the variant.<br>For more information, see “Rules for variant names in Genexus™ Dx Software” on page 215. |
| Locus | The chromosome positions in the reference genome that define the fusion junction. |
| Genes (Exon) | The name of fusion target and representative acceptor and donor exons. |
| Read Counts | The frequency that the fusion was detected in the sample. |
| Type | Assay type, (for example, Fusion, RNA exon variant (exon skipping), RNAExon Tile, Proc Control). |

*(continued)*

| Column | Description |
|---|---|
| Test Result | Indicates the presence or absence of a fusion or RNA exon variant.<br>• POSITIVE—indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—although some evidence for the presence of a fusion exists, the call does not pass one or more filters that are required for a high confidence fusion call.<br>• NEGATIVE—indicates the absence of a fusion due to the variant call not passing read or call thresholds. |
| Call Details | The reason for reporting a fusion as NO CALL or NEGATIVE. |
| Read Counts Per Million | The number of fusion read counts detected per million total reads. |
| Gene Isoform | The name of the fusion target as defined in the BED file.<br>Note: For fusion assays, the value for Gene Isoform is identical to that of Variant ID, |
| Mol Cov. Mutant | The median molecular coverage across a fusion amplicon. |
| Imbalance Score[1] | Each fusion gene exhibits a characteristic Imbalance Score threshold. Scores that exceed the threshold value indicate a high likelihood of the presence of the fusion in the sample. Imbalance score calculation starts with the normalization of reads in the exon-tiling amplicons of the gene, followed by correction with a baseline that represents expression values in normal samples. Reads for observed imbalance scores come from samples. Baseline scores come from normal, fusion-negative samples and are stored in the exon tile assay baseline file for the assay.<br>Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline)<br>where<br>Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples)<br>• 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample.<br>• 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample.<br>Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline)<br>• 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene.<br>• 5' of breakpoint is the total normalized baseline values of the amplicons for the gene. |
| Imbalance P-Value[1] | The statistical significance of measure of imbalance relative to a control gene. |
| Predicted Break-point Range[1] | The exonic range for predicted fusion break point in exon tiling assays. |

*(continued)*

| Column | Description |
|---|---|
| Ratio To Wild Type[1] | The molecular ratio for exon skipping assays relative to wild type control amplicons. |
| Norm Count Within Gene[1] | Exon skipping assay coverage normalized to molecular coverage of wild type (WT) MET control amplicons. |

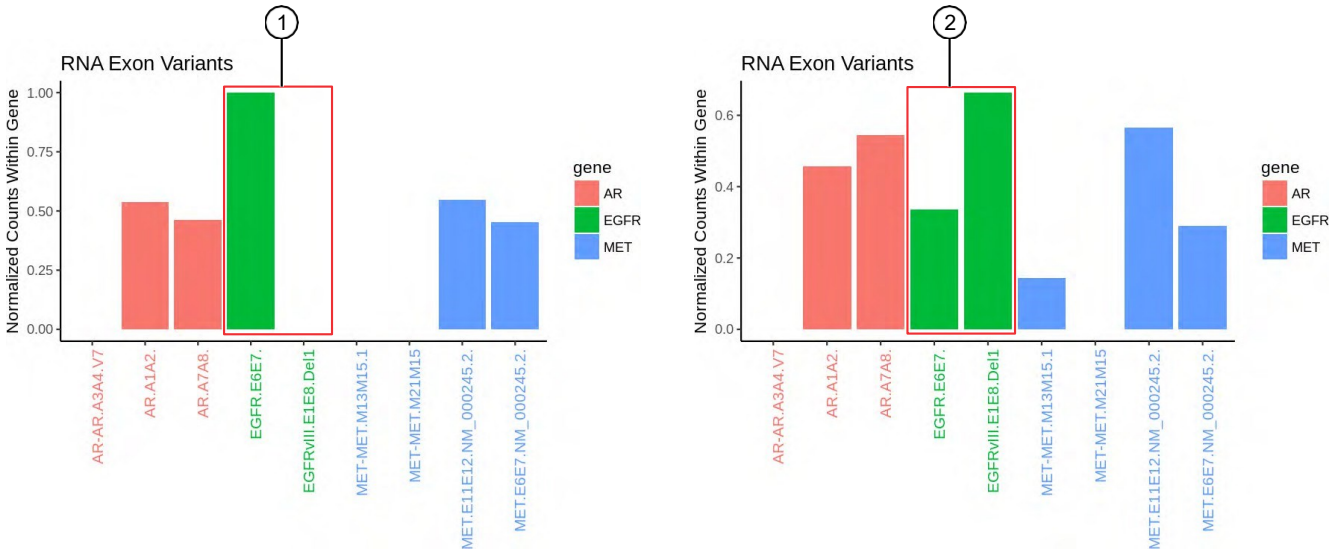
[1] Column appears only in analyses that use the exon tiling fusion detection method.

## View RNA Exon Variants

The **RNA Exon Variant** data view displays a bar graph summary of intragenic exon rearrangements or fusions. The displayed RNA exon variants are defined in the BED file that is associated with an assay. The **RNA Exon Variant** data view is available for all RNA and fusion assays.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 78.
4. In the top right corner of the screen, click **Visualization►RNA Exon Variant**, then review the **RNA Exon Variants** plot.

Representative RNA Exon Variant plots




The X-axis represents specific exon variants, where each variant is labeled with a gene ID followed by a sequence of adjacent exons. The Y-axis measures the read counts for each variant, normalized to the wild type.

① Example result where only the wild type EFGR (EFGR.E6E7) was detected.

② Example result where RNA exon 2–7 deletion occurred in the EFGR gene. The deletion of exons 2–7 resulted in an increase of normalized read counts for the EFGR variant that contains the intragenic fusion of exon 1 and exon 8 (EFGR.E1E8.Del1) and a decrease of normalized read counts for the wild type EFGR (EFGR.E6E7).

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View RNA Exon Tile Fusion Imbalance

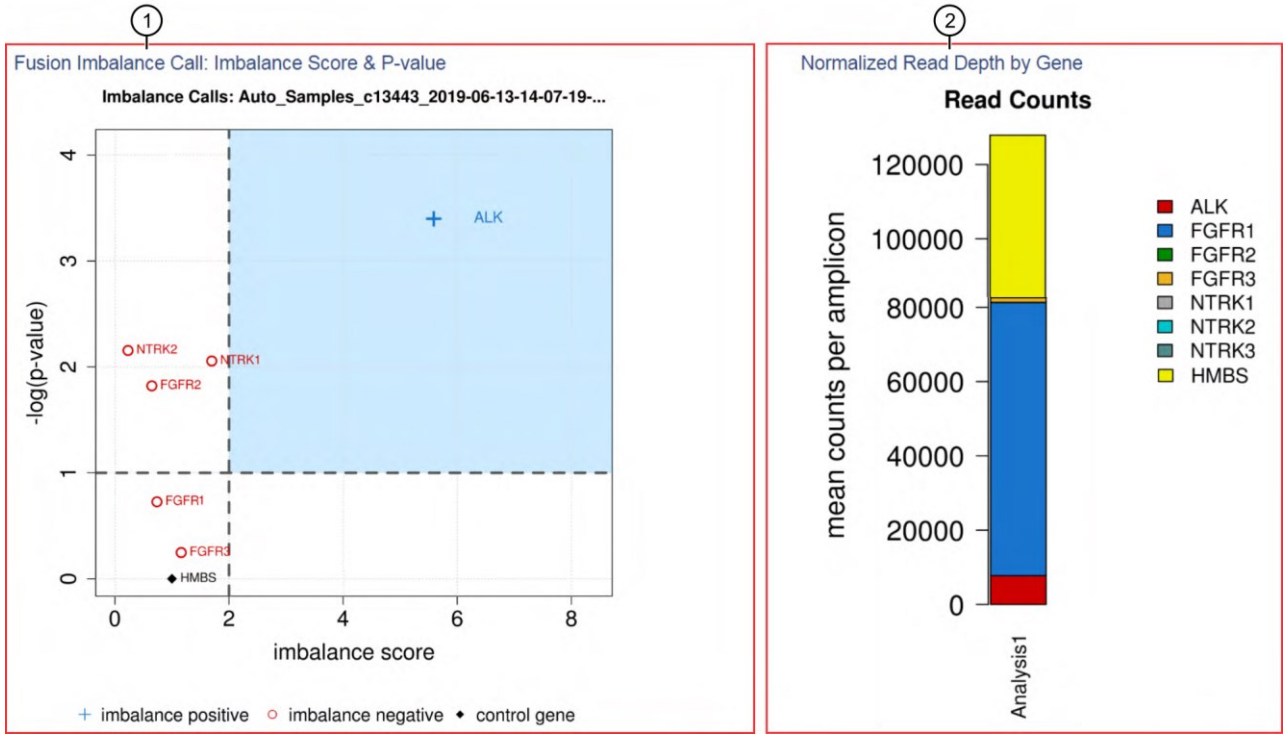
The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

Driver genes, for example ALK, FGFR2, and NTRK1, have amplicons that span exon-exon junctions to probe the expression difference between the 3' and the 5' regions of the gene.

An imbalance score is calculated for each driver gene in a sample to quantify the magnitude of the expression imbalance change between the two parts of the gene. For example, the 3':5' ratio in ALK; and the 5':3' ratio in FGFR2.

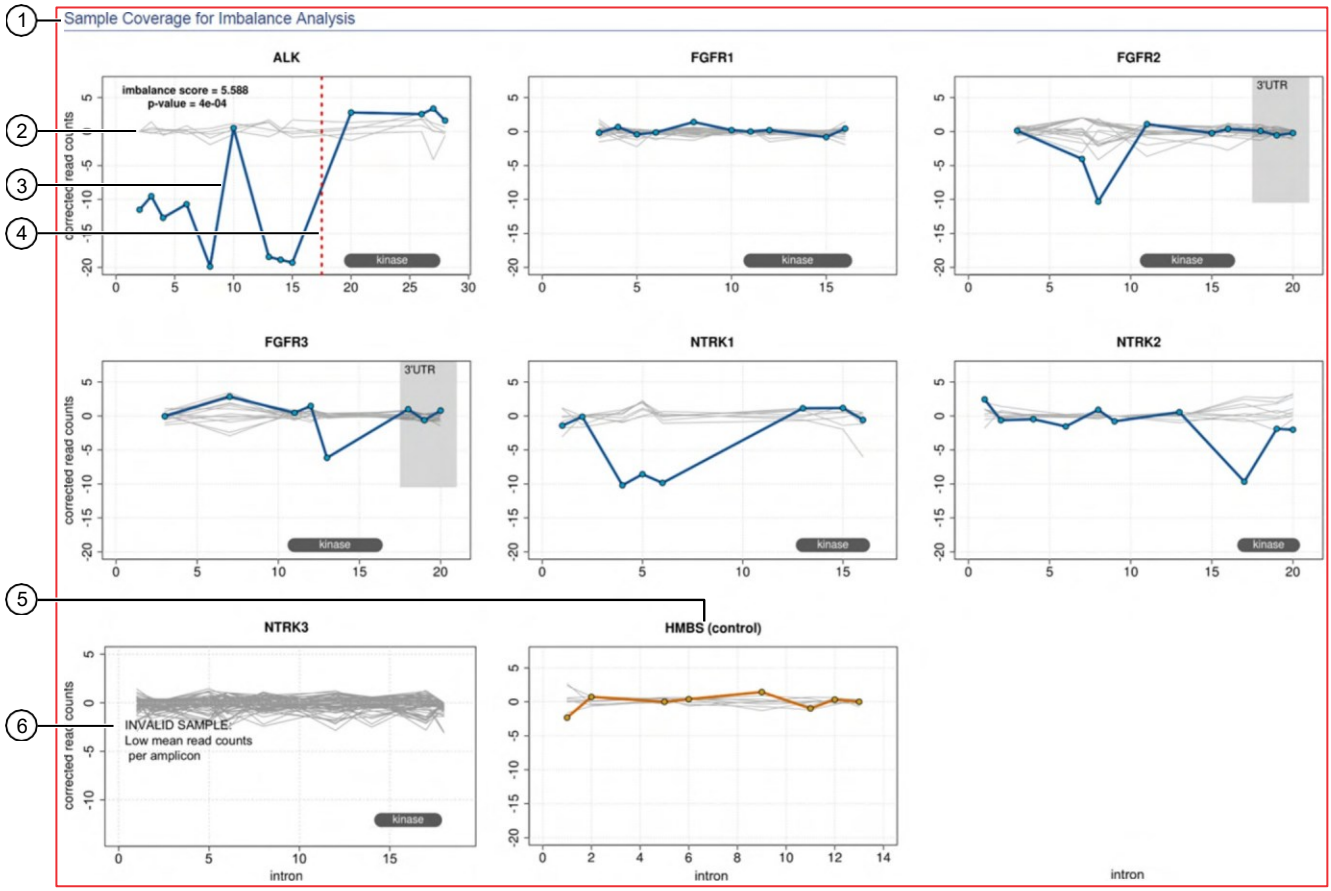
1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see "Fusions table" on page 78.
4. In the top right corner of the screen, click **Visualization**►**RNA Exon Tile Fusion Imbalance**, then review the **RNA Exon Tile Fusion Imbalance** plots.

## Representative RNA Exon Tile Fusion Imbalance plots




① The **Fusion Imbalance Call: Imbalance Score & P-value** plot shows the imbalance scores and p-values for all the genes in the selected sample. The dashed gray lines mark the threshold for an imbalance call, which is applied to all genes across all samples. Points that fall within the blue shaded area of the plot represent fusion-positive genes (✚). All other points that are outside of the blue shaded area represent fusion-negative genes (◯). Control genes are marked with ◆.

② The **Normalized Read Depth by Gene** plot shows the mean read counts of each gene that is captured on the chip for the selected sample. For each gene, the read counts are normalized to the number of amplicons.




1. The **Sample Coverage for Imbalance Analysis** plots show the expression profile for each exon-exon tiling amplicon for each gene. The Y-axis represents the corrected molecular counts. The X-axis represents individual exon-exon junctions, which are listed from 5′ to 3′. The **imbalance score** and **p-value** are listed in the panel of each gene that was called positive for fusion.
2. Baseline (a cluster of gray lines), generated from a fusion-negative sample.
3. Test sample corrected read coverage (blue line), normalized to the baseline. Each point on the line represents a unique exon-exon junction that was covered by the assay and normalized to the baseline.
4. Predicted range for the fusion break point for a fusion-positive gene (dashed red line).
5. Sample coverage profile for the control gene (orange line).
6. If the collected data are insufficient to determine an imbalance score, the **INVALID SAMPLE** message appears in the panel for that gene.

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View CNV results

The **CNVs** table lists the calls and other information for the copy number variants (CNVs) analyzed in each sample in a run.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

4. Click **CNVs** to view the **CNVs** table.

   For more information about the data displayed in the table, see “CNVs table” on page 84.

5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the CNVs variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the CNVs variant table, and information for table columns that are available but not currently shown in the CNVs table.

## CNVs table

The data displayed in the table depends on the assay that was used in the run.

Results in the table can be filtered using the filtering tools. For more information, see “Search and filter the list of variants” on page 74.

| Column | Description |
|---|---|
| Variant ID | The identifier of the CNV variant.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 85. |
| Variant Name | The name of the variant. |
| Locus | The starting position of the first amplicon covering the CNV gene. |
| Gene | The gene name. |
| Copy Number | The copy number of a CNV gene locus per genome. This column is available when a positive call is made. |
| Test Result | Indicates the presence or absence of a CNV.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—The absence of a variant; result is below the detection threshold for a CNV call. |
| P-Value | The statistical significance of the CNV ratio measurement. |
| Call Details | The reason for reporting a CNV as NO CALL. |
| CNV Confidence | The CNV confidence interval associated with the call. The 5% lower confidence bound value is the ploidy estimate, where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate, where it is 95% certain that the true ploidy is below that value. |
| CNV Ratio | The ratio of measured CNV gene locus coverage relative to coverage of non-CNV loci. |
| Valid CNV Amplicons | The number of amplicons spanning the CNV call. |

## View more annotations and annotation sources

In addition to the annotations that are included with Genexus™ Dx Software, you can view annotations and annotation source information from public websites for each variant.

1. In the menu bar, click **Results►Sample Results**.
2. Click the sample name in the **Sample Name** column in the row of a sample of interest.
3. Click the **Variants** tab.
4. In the **Variants** table, click the **Variant ID** of the variant of interest.

   You can click the row even if a **Variant ID** is not listed.

   The **Pileup** view is shown below the variants table. For more information, see “Review the variant pileup view” on page 85.
5. Click the **Annotations** tab.
6. Review annotation information in the **Annotations** tab, and if available, click a link in the row of an annotation to open a website with information about the annotation.

## Review the variant pileup view

You can review variant tracks with the pileup view in Genexus™ Dx Software. The pileup view is a figure that the software generates to show each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup view can be used to identify potential variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

The pileup view is shown below the variants table.

## Change the view of the variant pileup

You can zoom and pan the variant pileup view and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 86.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

| Action | Procedure |
|---|---|
| Track the position as you place the pointer over the variant pileup. | Click **Cursor Guide**. |
| Mark the center of the variant pileup. | Click **Center Line**. |
| Remove the track labels. | Click **Track Labels**. |
| Zoom in to increase the magnification of the image. | Click ⊕ **(Zoom in)** one or more times, or double-click the image. |
| Zoom out to decrease the magnification of the image. | Click ⊖ **(Zoom out)** one or more times. |
| Pan across the image to move the view to another location on the screen with its current magnification. | Click and drag the pointer over the image. |

## Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Dx Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

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Note: For best results, click the **Expand** option to view details about variants and base calls that are shown in each read coverage track

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1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to a track, then select an option to adjust the view of the track.

| Option | Description |
|---|---|
| **Expand** | Select this option to view the tracks with the maximum visible height. |
| **Squish** | Select this option to view the tracks with the minimal height for each track. |
| **Collapse** | Select this option to view the tracks with overlapping transcripts shown along a single line. When **Collapse** is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown. |

## Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Dx Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the reference track.

| Option | Description |
|---|---|
| View the reverse strand | Click **Reverse**.<br>This option is available only when the forward strand is shown. |
| View the forward stand | Click **Forward**.<br>This option is available only when the reverse strand is shown. |
| View the reference sequence translation | Click **Three-Frame Translate**.<br>This option is available only when the amino acid sequence is not shown. |
| Hide the reference sequence translation | Click **Close Translation**.<br>This option is available only when the amino acid sequence is shown. |

## Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Dx Software to dynamically customize the view of the data.

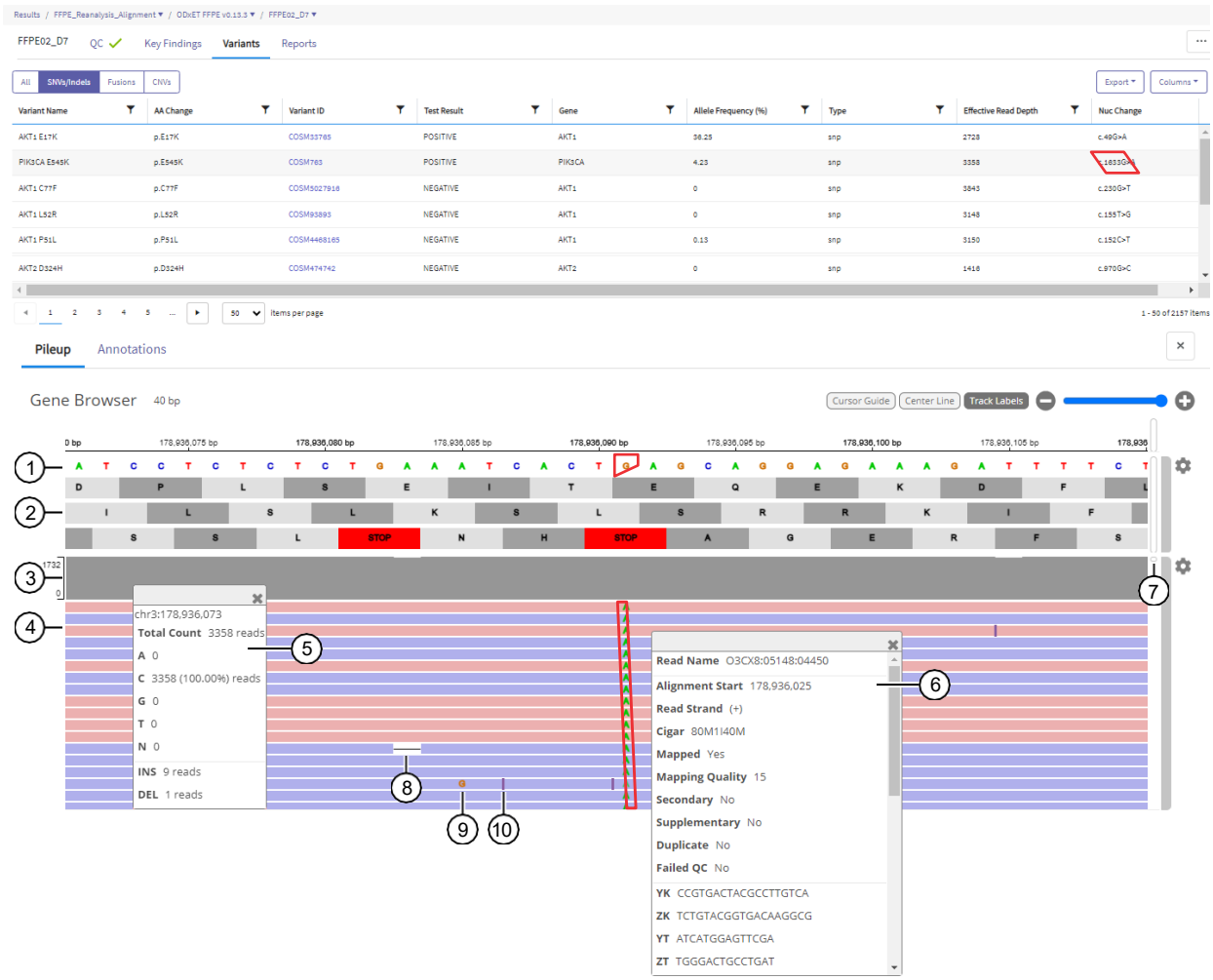
1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the BAM tracks.

| Option | Description |
|---|---|
| View all BAM tracks in monochrome gray | In **Color By**, deselect the checkmark next to **Read Strand**.<br>This option is available only when the BAM tracks are colored by read strand. |
| Distinguish between the direction of the BAM tracks with color | In **Color By**, click **Read Strand**.<br>This option is available only when the BAM tracks are not colored by read strand. |
| Expand, squish or collapse the BAM tracks | For more information, see “Adjust pileup tracks” on page 86. |
| Show all the bases in the BAM tracks | Click **Show all bases**.<br>This option is available only when all bases in the BAM tracks are not shown. |
| Change the minimum threshold for the visibility of BAM tracks | a. Click **Set visibility window**.<br>b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks.<br>When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see “Change the view of the variant pileup” on page 85. You can adjust the visibility window according to the scale shown in the screen. |

## Examples of a variant pileup view

The following figure is an example of a variant pileup view, shown in the **Gene Browser** in the **Variant** tab. The variant pileup view graphically represents the sequencing tracks, the reference track, and the annotation tracks, and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant are indicated in the image, both in the variant table and the in the pileup view. You can customize your pileup view. Reorder the tracks displayed by clicking a gray bar on the right edge of a track, then dragging the track to a different location. You can also click-drag the cursor at any position in the image to move the image left or right in the screen. For more information, see “Adjust pileup tracks” on page 86.




1. The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
2. The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 87.
3. The coverage track.
4. The BAM track.
5. Click the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
6. Click a BAM track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
7. Use the slider to view more tracks.
8. Deletion.
9. Alternate base in the BAM read coverage track.
10. Insertion.

The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.




① The hotspot track.
② The target region track.
③ Click a hotspot track for additional details, such as the start and end positions.
④ Click the target region track for additional details, such as the start and end positions.

## Coverage histogram

When you zoom in on the variant pileup view, you see the coverage histogram and reads from the .bam track. The histogram in gray shows read depth at that location and reads from the .bam track show read direction and location of variants.




① Read depth
② Read location and direction of variants

## View the variants from an assay run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. Review detailed variant data and read coverage tracks.




**Example visualization**

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate a family.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" indicates insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

| Option | Description |
|---|---|
| View the read coverage tracks sorted by family | Click ⚙ **(Actions)** next to the read coverage track, then select **Color►Molecule**. |
| Adjust the BAM tracks | Click ⚙ **(Actions)** next to the read coverage track, then select an option to adjust the view of the track. For more information, see “Adjust pileup tracks” on page 86. |
| Review detailed data about a BAM read | Single-click the read track to obtain information such as the mapping quality, the strand, and the read base. |
| Review distribution of base calls at a selected position | Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions. |

## View the QC results

You can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Dx Software.

Each run includes NTC and positive controls that are added automatically to each run.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.
3. Click the **QC** tab.
   The QC metrics are shown.

Review the QC data. For more information, see “QC results” on page 92.

## QC results

The quality control (QC) metrics for each sample that was sequenced in a run are displayed in Genexus™ Dx Software.

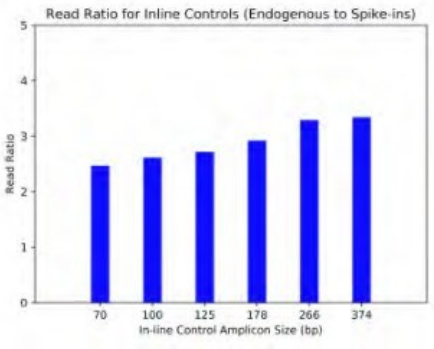
If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test in order to pass. The QC status is divided into the following categories.

- ✔ (Passed) indicates the sample passed all QC metrics.
- ✘ (Failed) indicates the sample failed a QC metric.
- — (Not Calculated) indicates a sample did not undergo QC analysis.

The data shown in the screen depend on the assay that was used in the run. Not all metrics are shown for every assay.

| Metric | Description |
|---|---|
| Run QC | General run quality control information. |
| Key Signal | The average signal after software processing for library ISPs that identically match the library key (TCAG). |
| Key Signal – Resequencing | The average signal after software processing for library ISPs that identically match the library key (TCAG) for the resequencing run. This metric is only shown for assays that include resequencing. |
| Percent Loading | The number of wells with ISPs divided by the number of the total addressable wells in a run. |
| Percent Loading – Resequencing | The number of wells with ISPs divided by the number of the total addressable wells in the resequencing run. This metric is shown only for assays that include resequencing. |
| Raw Read Accuracy | The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as 100 * (1 - (sum(per base error)/sum(per base depth))). |
| Templating QC—CF-1 Control | Sequencing quality metrics of the control fragment. These metrics indicate templating success. |
| Average Reads Per Lane | The number of CF-1 reads divided by the number of chip lanes used in the run. |
| Base Call Accuracy | The probability that a given base is called correctly.<br>1 – (total number of errors for all positions in CF-1) / (total number of CF-1 base reads). |
| Mean AQ20 Read Length (bp) | Average length, in base pairs, at which the accuracy rate is ≥99% for CF-1 reads. |

*(continued)*

| Metric | Description |
|---|---|
| Sample QC—DNA | Sequencing quality metrics of the sample DNA library. |
| MAPD | MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of 0.4 indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls. |
| Mapped Reads | The total valid mapped reads. |
| Mean AQ20 Read Length (bp) | The average length, in base pairs, at which the accuracy rate is ≥99% for all aligned reads of a library. |
| Mean Read Cov | The number of average reads per amplicon. |
| Molecular Uniformity | Uniformity of molecular coverage for all amplicons. |
| Mean Read Length (bp) | The average length, in base pairs, of final library reads for the sample. |
| Mean Mol Cov | Uniformity of molecular coverage for all amplicons. |
| Median Mol Cov | The median number of functional molecule reads per amplicon calculated over all amplicons in the assay. |
| Uniformity of Base Coverage | The percentage of reads with a depth of coverage ≥20% of the mean read coverage at each position. |
| Read Length Histogram | The histogram presents all filtered and trimmed DNA library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of genomic sample reads to control reads for each inline control amplicon.<br>  <br><br>Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for Ion AmpliSeq™ HD chemistries. |

*(continued)*

| Metric | Description |
|---|---|
| Sample QC – RNA | Sequencing quality metrics of the sample RNA library. |
| Mapped Fusion Reads | The total valid mapped reads. That is, the mapped reads that pass the thresholds and filters across all of the RNA targets in the reference file. This number is a subset of the mapped reads shown in the Run Samples table in the Assay Metrics. |
| Mean Read Length (bp) | The average length, in base pairs, of the final library reads for the sample. |
| RNA Expression Ctrls Detected | The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. |
| Mean AQ20 Read Length | The average length, in base pairs, at which the accuracy rate is ≥99% for all aligned reads of a library. |
| Read Length Histogram | The histogram presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of genomic sample reads to control reads for each inline control amplicon. You can use this metric to measure the level of contaminating genomic DNA in RNA libraries when you select the RNA checkbox for Include Inline Controls in assay set up. Using inline controls in RNA assays will reduce the total number of panel reads.<br>Note: The Read Ratio for each inline control amplicon is expected to be approximately 3 with 10 ng DNA input for Ion AmpliSeq™ HD chemistries. |
| NTC QC – DNA | DNA Sequencing quality metrics of the no-template control. |
| Hotspot Calls | The number of positive hotspots detected for the no-template control. |
| Average Base Coverage Depth | The average number of DNA reads of all targeted reference bases. |
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of no-template control sample reads to control reads for each DNA inline control amplicon. |
| NTC QC – RNA | RNA Sequencing quality metrics of the no-template control. |
| Mapped Fusion Reads | The total number of valid mapped reads for the NTC. |
| Total Fusion Calls[1] | The number of fusion variants detected for the no-template control. |
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |

[1] Counts for this metric include fusions, and non-wild type RNAExonVariant and RNAExonTiles variants.

### MAPD copy number QC metric

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on runs that detect CNVs.

MAPD is one of the metrics that we define to assess whether the panel data is useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log2 ratios per tile for a given run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log2 ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log2 ratio belongs to two pairs.

Formally, if `xi` is the log2 ratio at position `i`, with `i` ordered by genomic position:

```
MAPD = median ( | x(i - 1) - x(i) | )
```

MAPD is a per-sequencing run estimate of copy number variability, similar to standard deviation (SD). If one assumes the log2 ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log2 ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

## Find sample results

You can sort, search, and filter information to narrow the sample results that are listed in the **Results / Sample Results** screen. The sort, search, and filter tools help you organize sample results and find a specific sample result in Genexus™ Dx Software.

For information to search for a sample result by a unique identifier, such as the name of a run or the name of a sample, see “Search for sample results” on page 96.

For information to filter sample results by a common trait, such as QC status, see “Filter the list of sample results” on page 97.

## Search for sample results

Sample results are listed in the **Results / Sample Results** screen in Genexus™ Dx Software. The most recent sample result is on the top of the list.

You can search for sample results with keywords or text that are included in the name of the sample, run name, or the name of an assay applied to the run.

---

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. In the menu bar, click **Results**►**Sample Results**.

2. In the **Sample Results** screen, in the **Sample Name**, **Run Name**, or **Assay Name** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay, sample, or run name of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to a sample, run, or assay name to select one or more items.
      - Click **Select All** to select all items listed in the filter dialog box.

      ---

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

      ---

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of sample results is shown in the **Sample Results** screen.

Click **Clear Filters** to remove all filters and view the full list of sample results.

## Sort the list of sample results

You can sort the list of sample results to make it easier to find sample results in Genexus™ Dx Software.

By default, sample results are listed in the **Results / Sample Results** screen by date. The most recent sample result is at the top of the list.

You can change the order that the sample results are listed in the table.

For example, you can sort the **Call** column alphabetically so that the **Absent** calls appear first. This allows you to easily view hotspots that did not pass thresholds for a variant call, such as when the allele fraction is zero.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click a column heading of interest.
   The list of sample results is sorted in alphabetical or chronological order depending on the entries in the column.
3. Click the column heading again to reverse the order.

## Filter the list of sample results

You can apply filters to narrow search results, or shorten the list of sample results when you view the list of sample results in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the name of the sample, the run name or the assay selected in the run. | For more information, see "Search for sample results" on page 96. |
| Show or hide sample results that are archived | In the dropdown list in the upper left corner, select an archive status.<br>• Archived<br>• Non Archived<br>Select All to list all sample results. |
| Filter by the status of the sample result. | 1. In the Sample Status column heading, click ▼ (Filter) then select at least one status.<br>• Completed<br>• Running<br>• Failed<br>• Terminated<br>• Stalled<br>• Aborted<br>• Pending<br>2. Click Filter.<br>3. Records are listed for sample results that meet the sample status criteria. |

*(continued)*

| Option | Description |
|---|---|
| Filter by the QC status. | 1. In the QC Status column heading, click (Filter) then select at least one QC status.<br>• Failed<br>• Not Calculated<br>• Passed<br>2. Click Filter.<br>Records are listed for sample results that meet the QC status criteria. |
| List only sample results from runs that were started on or updated on an exact date. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or updated on the date entered. |
| List only sample results from runs that were started on or updated within a date range. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the earliest date of the range.<br>4. In the Boolean operator dropdown list, select And.<br>5. In the second dropdown list, select Is before or equal to.<br>6. In the calendar that appears, select the latest date of the range.<br>7. Click Filter.<br>Records are listed for sample results from runs started on or updated on dates that meet both conditions. |
| List sample results according to a date that meets either one condition or a second condition | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the earliest date for the sample results of interest.<br>4. In the Boolean operator dropdown list, select Or.<br>5. In the second dropdown list, select Is before or equal to.<br>6. In the calendar that appears, select the latest date for the sample results of interest.<br>7. Click Filter.<br>Records are listed for sample results from runs that were started on or updated on dates that meet at least one of the conditions. |

*(continued)*

| Option | Description |
|---|---|
| List only sample results for runs that were started on or before or updated on or before a date that you indicate. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is before or equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or before the date entered. |
| List only sample results for runs that were started on or after or updated on or after a date that you indicate. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or after, or updated on or after, the date entered. |

In the column heading, **(Filter)** changes color to indicate that a filter is applied.

Click **Clear Filters** to remove all filters and view the full list of sample results.

## Customize the sample results table

By default, the following information is listed in the sample results table in Genexus™ Dx Software.

- Name of the sample, assay, and run.
- Status of the sample result, such as completed.
- QC status.
- Date and time that the run was started and updated.

More information is available to show in the sample results table. You can customize the table to reflect the information that is most relevant.

### Change the columns in the sample results table

The sample results table in Genexus™ Dx Software includes a default set of columns. You can add columns to the table, or remove columns from the table. The columns that are available depend on the assays that are installed in the software. Table columns are based on sample attributes. You can also add or remove a column for information about **Electronic Signatures**.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click **Columns**.

3. Select the columns that you want to add to the sample results table, or deselect the columns that you want to remove from the table.
   The selected columns are added to the table or are removed from the table until you close the table.

4. Click **Save** to save the changes to the table columns.
   The changes that you have made to the table appear each time that you view the sample results table. The changes are saved until you add or remove other columns, then save those changes.

To restore the table to the default view, see “Restore the default columns in the sample results table” on page 100.

### Restore the default columns in the sample results table

You can restore the default view of the columns in the sample results table in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample Results** screen, click **Columns**.

3. At the end of the list of columns that appears, click **Restore default**.

The default columns in the sample results table are restored.

## Sample tags in sample results

You can assign tags to sample results in Genexus™ Dx Software. Tags are used to label and group sample results. A tag can be assigned to a single sample or to multiple samples at once.

### Assign sample tags to a sample result

You can assign tags to sample results in Genexus™ Dx Software to label and group sample results. For more information, see “Sample tags” on page 33.

1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample Results** screen, place the pointer over the row of the sample result that you want to tag, then click **Assign Tags**.

3. In the **Assign Tags** dialog box, search for or scroll to a tag.
   If the tag that you want to assign to a sample does not exist in the software, a manager- or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 33.

4. Select one or more tags that you want to assign to the sample result, then click **Save**.

The tag or tags are assigned to the sample result and are listed in the row for the sample result in the list of sample results.

### Assign sample tags to multiple sample results

You can assign one or more tags to multiple sample results in Genexus™ Dx Software. For more information about sample tags, see “Sample tags” on page 33.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the checkbox to the left of each sample result of interest, then click **Assign Tags** above the list of sample results.

   **Assign Tags** is not shown until you select at least one sample result.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.

   If the tag that you want to assign to a sample result does not exist in the software, a manager- or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 33.
4. Select one or more tags that you want to assign to the sample results, then click **Save**.

The tag or tags are assigned to the selected sample results and are listed in the row for each sample result in the list of sample results.

## Review run results

You can review completed sample results and reports in Genexus™ Dx Software.

See “Find a run result” on page 109 for information about how to find run results of interest.

1. Click **Results►Run Results** to view run results.
   A list of runs that are pending, in progress, or completed are shown in the **Results / Run Results** screen. Runs with a status of failed, aborted, or stalled are also listed.
   The following run information appears in the **Results / Run Results** screen.

| Column | Description |
|---|---|
| Run Name | The unique name of the run given when it was created in the software. Click a run name to open the Run Summary. For more information, see “The run summary” on page 103.<br>Runs that are reanalyzed are listed with (Reanalysis) after the run name. For more information, see “Reanalyze a run” on page 117. |
| Assay Name | The name of the assay selected for the run. You can view the Assay Name and corresponding Assay Full Name for all assays in the Assays►Manage Assays screen. |
| Run Status | The status of the run. For example, Analysis In Progress, Executing Plugins, Analysis Completed, Terminated, Backup: In Progress). |
| Total Samples | The total number of samples in a run. |
| PCR Plate Number | A unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 114. |

*(continued)*

| Column | Description |
|---|---|
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |

2. In the **Run Name** column, click a run name.
   The **Results** screen opens to the **Run Summary** table.

3. View the run results.
   - The **Run Summary** table contains details about the run, including the assays used in the run, samples, reagents and primer tube locations, and whether the run was reanalyzed. For more information, see “View the run summary” on page 103.
   - Click **Run Report** to view detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run. For more information, see “View assay metrics and the run report” on page 106.

You can also perform the following actions in the **Results / Run Results** screen.

| Option | Description |
|---|---|
| Actions[1] | Available action links for a run are shown when you place the pointer over the row of a run.<br>• Delete—Delete the run.<br>• Audit—View the audit trail for the run.<br>For more information, see “Review and download a run history” on page 60.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>For more information, see “Download a Customer Support Archive” on page 222.<br>• Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 114. |

[1] The actions that are available depend on the type of run.

## View the run summary

The run summary provides an overview of the run. The information that is displayed includes a list of the assays used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Name** column, click the run name of interest.
   The **Run Summary** tab opens.
3. Review the run summary.

| Action | Procedure |
|---|---|
| View the assay metrics. | In the Assays section, click the assay name of interest. |
| View the sample locations in an image of a 96-well sample plate. | In the Sample Locations section, click PCR Plate View. |
| Reanalyze a run with a new assay. | In the upper right corner of the screen, click ••• (More Options)►Reanalyze.<br>For more information, see “Reanalyze a run” on page 117. |
| Download customer support archive (CSA) log files for the run to help with troubleshooting. | In the upper right corner of the screen, click ••• (More Options)►CSA.<br>For more information, see “Customer Support Archive (CSA)” on page 222. |
| View the history of variant classifications. | In the upper right corner of the screen, click ••• (More Options)►Variant Audit.<br>The information is available only for results that include variant classifications. |
| View the run report. | Click the Run Report tab. |

### The run summary

The run summary displays run information, assays used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

| Item | Description |
|---|---|
| Run Name | The name of the run. |
| Started On | The date and time when the run was started. |
| Completed On | The date and time when the run was completed. |
| Run Status | The status of the run. |

Run information *(continued)*

| Item | Description |
|---|---|
| Report Template | If the option to generate a report was enabled in the run plan, the report template used is listed. |
| Chip Type | The semiconductor sequencing chip used in the run, such as the Genexus™ Dx GX5™ Chip. |
| Library Chemistry | The type of library chemistry used in the run, such as Ion AmpliSeq™ HD. |

Sequencing instrument information

| Item | Description |
|---|---|
| Instrument Name | The name of the sequencer. |
| Instrument Serial Number | The serial number of the sequencer. |
| Status | The status of the sequencing portion of the run. |
| Operator | The name of the person who was signed in the Genexus™ Dx Integrated Sequencer when the sequencing run was performed. |
| Start Date | The date and time when the run was started on the sequencer. |
| Completion Date | The date and time when the run was completed on the sequencer. |
| Sequencing Flows | The number of flows performed by the sequencer. |

Assays table

| Item | Description |
|---|---|
| Assay Full Name | The name of the assay used in the run. Click the assay name to display the details of the assay. |
| Assay Name | An abbreviated name of the assay or assays used in the run. |
| Analysis Version | The version of the assay used for analysis. |
| Application Type | The application type for the assay, such as DNA and Fusions. |
| Lane | The chip lane or lanes used in the sequencing run for the assay. |
| Total Samples | The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample. |

Sample Locations table

| Item | Description |
|---|---|
| PCR Plate View | Click to view the sample locations in an image of a 96-well sample plate. |
| Well Pos. | The well position indicates the location of the sample on the plate. |
| Library Batch ID | The unique identifier created for the library batch. This information is available only for Library to Result runs. |
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, RNA, or DNA+RNA. |
| Vol. (µl) | The volume of the sample. |
| Conc. (ng/µl) | The concentration of the sample. |
| Dilution Factor | The dilution factor of the sample. |
| Kit Barcodes | The barcode for the kit used for nucleic acid extraction, if applicable. |
| Barcode | The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101. |
| Assay Name | The assay that was used to sequence the sample for the indicated well position. |
| Library Position | The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run. |

Reagents table

| Item | Description |
|---|---|
| Consumable | Consumables used in the run, such as the PanelKit and LibraryKit, are listed. |
| Barcode | The consumable barcode, if applicable. |
| Part # | The consumable part number, if applicable. |
| Expiration Date | The consumable expiration date, if applicable. |
| Lot # | The consumable lot number, if applicable. |

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

# View assay metrics and the run report

The run report provides detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run.

For runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

1. Click **Results►Run Results** to view sequencing results for a particular sample.
2. In the **Run Name** column, click the run name of interest.

| Action | Procedure |
|---|---|
| View the run report | Click the **Run Report** tab. |
| View the assay metrics for a single assay | In the **Select Assay** dropdown list at the top of the screen, select the assay of interest. |

For more information, see “Assay metrics and the run report” on page 106.

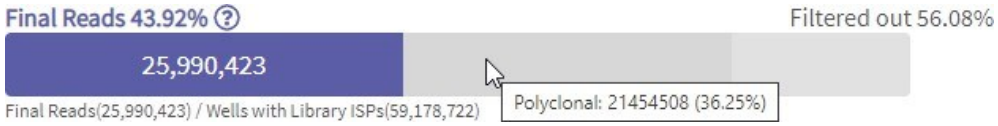
## Assay metrics and the run report

The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.

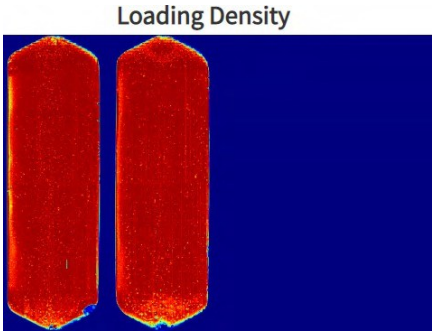
Run Report

| Metric | Description |
|---|---|
| Loading | The number and percentage of total addressable wells on the chip that contain an ISP. |
| Enrichment | The number and percentage of wells ISPs that contain live ISPs. |
| Library | The number and percentage of wells with live ISPs that contain Library ISPs. |
| Final Reads | Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement. |
| Total Bases | The number of filtered and trimmed base pairs that are reported in the output BAM file. |
| Raw Read Accuracy | The raw read accuracy across each individual base position in a read calculated as, (1−[total errors in the sequenced reads]/[total bases sequenced]) × 100. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is *not* based on consensus accuracy across multiple reads for the same base position. |
| Wells with ISPs | The number of wells that contain an ISP. |
| Unenriched | The number and percentage of wells with ISPs that do not contain live ISPs. |

Run Report *(continued)*

| Metric | Description |
|---|---|
| Total Addressable Wells | Wells on the chip that can be physically reached by a library. |
| Empty Wells | The percentage of total addressable wells on the chip that do not contain an ISP. |
| Wells with Live ISPs | Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key. |
| Wells with Library ISPs | Loaded wells with live ISPs with a key signal that is identical to the library key signal. |
| Control ISPs | Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal. |
| Filtered out | The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads. |
| Polyclonal | Wells with a live ISP that carries clones from two or more templates.<br>To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot.<br>  |
| Low Quality | Loaded wells with a low or unrecognizable signal.<br>To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot.<br>  |
| Adapter Dimer | Loaded wells with a library template of an insert size less than 8 bases.<br>Adapter dimer reads represent a small proportion of total reads and can be seen by hovering the pointer over the right end of the Final Reads bar.<br>  |

Run Report *(continued)*

| Metric | Description |
|---|---|
| Loading Density | A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs.<br>  |

## Run Samples

The Run Samples table lists read data for each individual sample in the assay.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered in the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, RNA, or DNA+RNA. |
| Barcode | The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample. |
| Total Reads | The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. |
| Mean Read Length | The average length, in base pairs, of usable library reads for each sample. |
| ≥Q20 Bases | The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample. |
| Mapped Reads | The number of reads that are mapped to the reference file. |
| On Target Reads | The percentage of sequencing reads mapped to any target region of the reference. |
| Mean Depth | The average number of reads of all targeted reference bases. |
| Uniformity | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. |
| Read Length Histogram | A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).<br>  |

### Download information about run samples

Information about each sample in a run, such as the barcode for the sample and the total reads, are shown in the run samples table. You can download the information shown in the run samples table.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Run Samples** table.
4. In the upper right corner above the **Run Samples** table, click **Export to Excel**.

An XLSX file that contains the information shown in the run samples table is downloaded.

Read length histograms are not included in the file.

### View resequencing metrics

For assays that include resequencing, you can view resequencing metrics when a run is complete.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In **Select Assay**, select the assay name of interest.
   The assay metrics for the sequencing run are shown.
4. Click **Resequencing Metrics**.
   The resequencing assay metrics are shown. For more information, see “Assay metrics and the run report” on page 106.

## Find a run result

You can sort, search, and filter information to narrow the run results that are listed in the **Results / Run Results** screen. The sort, search, and filter tools help you organize run results and find a specific run result in Genexus™ Dx Software.

For information to search for a run result by a unique identifier, such as the name of a run, see “Search for run results” on page 110.

For information to filter run results by a common trait, such as run results that have a status of analysis completed, see “Filter the list of run results” on page 112.

## Search for run results

Run results are listed in the **Results / Run Results** screen in Genexus™ Dx Software. The most recent run result is on the top of the list.

You can search for run results with keywords or text that are included in the name of the run name, the name of an assay applied to the run, or by the PCR Plate number. For more information about how to use the PCR Plate number to track libraries, see “Assign PCR Plate” on page 114.

---

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. In the menu bar, click **Results**►**Run Results**.

2. In the **Run Results** screen, in the **Run Name**, **Assay Name**, or **PCR Plate Number** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay or run name or PCR plate number of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to a run or assay name to select one or more items.
      - Click **Select All** to select all items listed in the filter dialog box.

      ---

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

      ---

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

   The filtered list of run results is shown in the **Run Results** screen.

3. Click **Clear Filters** to remove all filters and view the full list of run results.

## Sort the list of run results

You can sort the list of run results to make it easier to find a run result of interest in Genexus™ Dx Software.

By default, run results are listed in the **Runs / Run Results** screen by date. The most recent run result is at the top of the list.

You can change the order that the run results are listed and sort the run results alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs►Run Results**.
2. In the **Run Results** screen, click the **Run Name** heading.
   The list of run results is sorted alphabetically.
3. Click the column heading again to reverse the order.

## Filter the list of run results

You can apply filters to narrow search results, or shorten the list of run results when you view the list of run results in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Run Results**.
2. In the **Run Results** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the run name or the assay selected in the run. | For more information, see “Search for run results” on page 110. |
| Show or hide run results that are archived | a. In the dropdown list in the upper left corner, select an archive status.<br>b. • **Archived** • **Non Archived**<br>c. Select **All** to list all run results. |
| Filter by the status of the run result. | a. In the **Run Status** column heading, click **(Filter)** then select at least one status.<br>• **Analysis Completed**<br>• **Analysis In Progress**<br>• **Analysis Failed**<br>• **Analysis Aborted**<br>• **Analysis Stalled**<br>• **User Aborted**<br>• **Run Aborted**<br>• **Backup In Progress**<br>• **Executing Plugins**<br>• **Library Preparation Aborted**<br>• **Library Preparation Completed**<br>• **Library Preparation Started**<br>• **Selected on Instrument**<br>• **Sequencing Completed**<br>• **Sequencing Started**<br>• **Sequencing Aborted**<br>• **Templating Aborted**<br>• **Templating Completed**<br>• **Templating Started**<br>• **Kit Barcodes Required**<br>b. Click **Filter**.<br>Records are listed for run results that meet the run status criteria. |
| Filter by the number of samples in the run. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run. |
| List only run results from runs that were started on or updated on an exact date. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for run results that were started on or updated on the date entered. |

| Option | Description |
|---|---|
| List only run results from runs that were started on or updated within a date range. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date of the range.<br>d. In the Boolean operator dropdown list, select **And**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date of the range.<br>g. Click **Filter**.<br>Records are listed for sample results started on or updated on dates that meet both conditions. |
| List run results according to a date that meets either one condition or a second condition. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date for the sample results of interest.<br>d. In the Boolean operator dropdown list, select **Or**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date for the sample results of interest.<br>g. Click **Filter**.<br>Records are listed for sample results for runs that were started on or updated on dates that meet at least one of the conditions. |
| List only run results for runs that were started on or before or updated on or before a date that you indicate. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is before or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for run results that were started on or before the date entered. |
| List only run results for runs that were started on or after, or updated on or after, a date that you indicate. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for runs that were started on or after, or updated on or after, the date entered. |

In the column heading, **(Filter)** changes color to indicate that a filter is applied.

Click **Clear Filters** to remove all filters and view the full list of run results.

## Retain run results

You can retain run results in Genexus™ Dx Software. When you retain a run result, run data, including the run result, .wells files, and intermediate files, are retained and cannot be deleted. Users with any level of permissions can retain run results. When a run result is retained, it overrides the automatic deletion settings in the software. For more information, see “Set a schedule to permanently delete files (administrator)” on page 177.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to retain.
3. Click **Retain Data**.

Run results that have been retained are denoted with after the run name in the **Run Results** screen. The run result is retained in the system and cannot be deleted. For information to revert the run to a run that is not retained, see “Release run results” on page 114.

## Release run results

Only run results that have been retained in Genexus™ Dx Software can be released. For more information, see “Retain run results” on page 114.

When you release run results, you can delete the run results. The run results, the associated .wells files, and intermediate files are available to be auto-deleted according to system settings. For more information, see “Set a schedule to permanently delete files (administrator)” on page 177.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, click **Release Data** in the row of the run of interest.
   Run results that have been retained are denoted with (retained) after the run name.
3. Click **Yes** to confirm that you want to proceed with the data release.

The run result is released. The run result can be deleted or auto-deleted.

## Assign PCR Plate

Genexus™ Dx Software lets you track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

---

Note: You should assign a PCR plate to a run if there are leftover libraries that you want to use at a later time to perform a Library to Result run. You can look up the PCR plate number on completed runs to get the library and barcode mapping, then enter the same Barcode information into the Library to Result run.

---

For more information to find a run with a search of the PCR Plate Number, see “Search for run results” on page 110.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.

   The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

# View verification run results

Manager- and administrator-level users can view verification run details, including the run summary, QC summary, and the reagents used in the run. Verification runs are performed during installation to validate the performance of the instrument or instruments. Sequencing verification runs are performed to validate the Genexus™ Dx Integrated Sequencer.

Note: Field Service Engineers use system-installed verification templates to validate the installation of the Genexus™ Dx Integrated Sequencer. Field Service Engineers can view the template information but cannot modify it.

1. In the menu bar, click **Results►Verification Results**.
2. In the **Verification Results** screen, view or download information for the verification run.

| Option | Description |
|---|---|
| View verification results details | In the **Run Name** column, click the verification run name of interest to open the **Verification Results Details** screen.<br>In the **QC** section of the **Verification Results Details** screen, click the **>** symbol to the left of the **Lane** column in the lane of interest to expand the results details pane. |
| Download a verification report that has been signed | Place the pointer over the row of the verification run of interest, then click **Report**. |

3. You can perform the following actions for **Verification Results**. Action links are available when you place the pointer over the row of a verification result.

| Option | Description |
|---|---|
| View Plan | If the run has not started on an instrument, you can view the run plan for the verification run. |
| Audit | Displays the list of users who created and edited the run. You can export and print information from the list from the Audit Trail dialog box. |
| CSA | CSA—Customer Support Archive. Click this link to download the sequencer log files. Log files contained within the CSA can be useful when troubleshooting problems with the sequencer. For more information, see Chapter 10, “Customer Support Archive”. |
| Report | Download a verification report that has been signed. |

## Verification runs

The following information is available in the **Results / Verification Results** screen.

| Column | Description |
|---|---|
| Run Name | The name of the run, created when the run was planned. Click the name to open the Verification Results Details screen. |
| Assay Full Name | The name of the assay used in the run. |
| Field Engineer Name | The name of the support specialist who performed the run. |
| Instrument Name | The name of the sequencer that was verified. |
| Run Status | The current status of the full sequencing run, including analysis. |
| QC Status | Indicates whether a sequencing run passed or failed, based on the sequencing QC metrics selected for the assay. |
| Started On | The date and time that the run was started. |
| Updated On | The date and time that the run was last updated. |

## Sign verification run reports (manager/administrator)

Manager- and administrator-level users can sign results reports for verification runs. However, we recommend that only qualified support specialists sign performance qualification (PQ) reports.

Verification reports can be electronically signed only when all samples pass QC analyses.

1. Click **Results►Verification Results**.
2. In the **Verification Results** screen, click the name of the verification run of interest.
3. In the **Verification Results Details** screen, click **Sign Off**.
4. In the **Sign Off Report** dialog box, enter or make the following selections.
   a. Enter the password.

b. In **Electronic Signature**, select **Approval**.

c. In **Sign off Comments**, enter a comment.

d. In **Laboratory Comments** and **Footer Field**, enter optional information.

5. Click **Sign Off.**

The verification run report is electronically signed.

You can view, print, or download the report in a PDF file. For more information, see “Verification runs” on page 116.

# Reanalyze a run

Runs can be reanalyzed using the same assay in Genexus™ Dx Software. Reanalysis of runs can start from the alignment, basecalling, or signal processing steps. When you reanalyze a run, the reanalysis is applied to all samples in the assay.

| Reanalysis from the | Description |
|---|---|
| Signal processing step | The option to reanalyze a run from the signal processing step is available only when the following conditions are met.<br>• When a run fails during the basecalling step or earlier.<br>• When the run has not already been successfully reanalyzed.<br>Reanalysis at the signal processing step uses DAT files. |
| Basecalling step | The option to reanalyze a run from the basecalling step is available only when a run or reanalysis successfully completes the signal processing step.<br>Reanalysis at the basecalling step uses .wells files. |
| Alignment step | The option to reanalyze a run from the alignment step is available only when a run or reanalysis successfully completes the basecalling step.<br>Reanalysis at the alignment step uses BAM files. |

Note:

- Manager- and administrator-level users can reanalyze a sequencing run only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the run cannot be reanalyzed.
- If a sample fails QC, you can sequence the sample library again. For information to find, recover, and purify the leftover library preparations, see the user guide for the panel that you use. Alternatively, you can prepare a new library for sequencing.

The files needed for the stage of reanalysis that you select must be present in the software. For example, if the .wells files for the run have been removed from the software, you cannot reanalyze from basecalling. Instead, reanalyze the BAM files at alignment. Administrator-level users can manage the settings and schedule to backup and delete files and data. For more information, see “Manage backup settings and restore runs (administrator)” on page 166.

You can reanalyze a run with any compatible assay that exists in the software.

1. In the menu bar, click **Results►Run Results**.
2. In the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In the upper right corner of the screen, click ••• **(More Options)►Reanalyze**.
4. In the **Reanalyze** dialog box, enter or select the following information.
   a. In **Reanalysis Run Name** field, enter a reanalysis run name.
   b. In **Start Reanalysis from** dropdown list, select **Alignment**, **Basecalling** or **Signal Processing**.
   c. In the **Current Assay in Run** column, select the checkbox in the row of the assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for the reanalysis from the dropdown list.
5. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Run Results** screen in the **Run Status** column, and in the **Results / Sample Results** screen in the **Sample Status** column. When reanalysis is complete, the new results can be viewed by clicking the run name corresponding to the reanalysis assay in the **Results / Run Results** screen. Runs that have been reanalyzed are appended with ⟲ after the run name.

# Reanalyze a sample

Samples can be reanalyzed using the same assay in Genexus™ Dx Software. A sample can be reanalyzed only when starting from the alignment step. You can reanalyze all samples in a run from the basecalling or signal processing steps. For more information, see “Reanalyze a run” on page 117.

Reanalysis at the alignment step uses BAM files.

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Note: Manager- and administrator-level users can reanalyze a sample only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the sample cannot be reanalyzed.

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The option to reanalyze a sample from the alignment step is available only when a sample or reanalysis successfully completes the basecalling step.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of a sample of interest, then click **Reanalyze**.

   Alternatively, you can reanalyze a sample when you click a sample name, then click ••• **(More Options)**►**Reanalyze**.
3. In the **Reanalyze** dialog box, enter or select the following information.
   a. In **Reanalysis Run Name** field, enter a reanalysis run name.
   b. In the **Current Assay in Run** column, select the checkbox in the row of the assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for the reanalysis from the dropdown list.
4. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Sample Results** screen in the **Sample Status** column. When reanalysis is complete, the new results can be viewed by clicking the sample name corresponding to the reanalysis assay in the **Results / Sample Results** screen. Samples that have been reanalyzed are listed in the table of sample results with the run name appended with ⟲ .

# Results files

The following files can be downloaded for each sample. The files that are available for download vary depending on the assay. Results files include the sequencing data, results from the analyses, such as variant files, and audit and log files.

For instructions to download results files, see “Download results files for a sample result” on page 121.

| Option | File name | Description |
|---|---|---|
| Variants | | |
| Filtered Variants (.vcf) | `<Sample_name>_<LibPrepID>_filtered.vcf` | Summary of filtered variant results in variant call format (VCF). |
| Variant Summary (.tsv) | `Summary.tsv` | File that lists SNV/INDEL, copy number, and fusion results in tab-separated value format (TSV). |
| Snvindel (.tsv) | `Snvindel.tsv` | File that lists SNV/INDEL variant results in tab-separated value format (TSV). |
| Fusion (.tsv) | `Fusion.tsv` | File that lists fusion results in tab-separated value format (TSV). |
| CNV (.tsv) | `Cnv.tsv` | File that lists copy number variant results in tab-separated value format (TSV). |

*(continued)*

| Option | File name | Description |
|---|---|---|
| Sequencing Results | | |
| DNA Unmapped Bam File (.bam) | `<barcode>_rawlib.basecaller.bam` | Unmapped DNA barcode BAM file; output after mapping reads to reference. |
| DNA Mapped Bam File (.bam) | `merged.bam` | Mapped BAM file of combined barcode reads. |
| DNA Mapped Bam Index File (.bai) | `merged.bam.bai` | Mapped BAM Index file. |
| DNA Basecaller FASTAQ File (.fastq) | `<barcode>_rawlib.basecaller.fastq` | FASTQ file of the DNA barcodes used. |
| DNA Processed Bam File | `merged.bam.ptrim.bam` | Mapped BAM file of combined barcode reads. |
| DNA Processed Bam Index | `merged.bam.ptrim.bam.bai` | Mapped BAM index file. |
| RNA Unmapped Bam File (.bam) | `<barcode>_rawlib.basecaller.bam` | Unmapped RNA BAM file; output of base calling, contains unmapped reads. |
| RNA Mapped Bam File (.bam) | `<barcode>_rawlib.basecaller_alignments.bam` | Mapped BAM file of combined barcode reads. |
| RNA Mapped Bam Index File (.bai) | `<barcode>_rawlib.basecaller_alignments.bam.bai` | Mapped BAM index file. |
| RNA Basecaller FASTAQ File (.fastq) | `<barcode>_rawlib.basecaller.fastq` | FASTQ file generated from the unmapped BAM file of the RNA barcodes used. |
| Test Fragment Basecaller FASTAQ File (.fastq) | `rawtf.basecaller.fastq` | FASTQ file for the test fragment. |
| Audit and Log | | |
| Analysis Log File | `analysis.log` | Analysis log file. |
| Run Audit | `PlannedRun-AuditTrail.pdf` | Contains all audit records pertaining to the run. |
| Reports | | |
| Report | `<language>_<sample name>_Dx_<report template name>_<assay name>_<date>.pdf` | A PDF report that contains sample-specific results. For more information, see “Variant report” on page 124. |

*(continued)*

| Option | File name | Description |
|---|---|---|
| Sample Summary | `Info.csv` | Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. |
| Troubleshooting Files | | |
| Log Files[1] | `analysis.log` | Analysis log file. |
| | `summary.<timestamp>.log` | Start and end time for each time an assay module is executed for the analysis. |
| | Various other files with `.err` `.out` file extensions | Analysis pipeline logs used by field service engineers for troubleshooting. |
| | `Info.csv` | Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. |
| | `PlannedRun-AuditTrail.pdf` | Contains the audit trail of the run plan in PDF format. |
| VCF Files | `analysis.vcf` | Summary of variant results for sample, NTC and positive control in variant call format (VCF). |

[1] Separate folders are generated for each sample. If included in the run, separate folders are also generated for an NTC and positive control.

## Download results files for a sample result

You can download results files for a sample result in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**, or **Results►Run Results**, then do one of the following procedures.

| Option | Selection |
|---|---|
| Download files for a sample | |
| Download results files for a sample from the list of sample results. | In the Results / Sample Results screen, place the pointer over the row of a sample of interest, then click Download Files. |
| Download results files for a selected sample from the Results screen. | **1.** In the Results / Sample Results screen, in the Sample Name column, click the sample name of interest.<br>**2.** Click ••• (More Options)►Download Files. |

*(continued)*

| Option | Selection |
|---|---|
| Download files for a run or assay | |
| Download results files for a run from the list of run results. | In the Results / Run Results screen, place the pointer over the row of a run of interest, then click Download Files. |
| Download results files for a selected run or assay from the Results screen. | 1. In the Results / Run Results screen, in the Run Name column, click the run name of interest.<br>2. Click the run or assay of interest from the results navigation bar.<br>3. Click ••• (More Options)►Download Files. |

2. In the **Download Files** dialog box, select the files to download, then click **Download**.
   For information about the files, see “Results files” on page 119.

The selected results files are downloaded in one ZIP folder.

## Download results files for multiple sample results

You can download results files for multiple sample results in Genexus™ Dx Software. If the run included more than one assay or more than one research application for the assay, you can download the results for each assay or research application at once.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Download**.
   Alternatively, in the **Run Results** screen, click the name of a run, then in the **Run Summary** tab, click ••• **(More Options)►Download Files** to download the results files.
3. Select the assay if the run included more than one assay or more than one research application for the assay.
4. In the **Download Files** dialog box, select the files to download, then click **Download**.
   For information about the files, see “Results files” on page 119.

The selected results files are downloaded in one ZIP folder named `Runname_assayname`. The folder contains subfolders for each sample result except for any NTC and positive control samples.

Control sample information is in the `SmallVariants.vcf`. For more information, see “Results files” on page 119.

# Data files and flow

During a Genexus™ Dx Integrated Sequencer run, raw data (DAT) files from the first flows are available. Genexus™ Dx Software starts processing the data, producing a 1.wells file. Basecalling is performed on the 1.wells file data, producing an unmapped BAM (uBAM) file. Subsequent analysis produces mapped reads (BAM) and variant calls (VCF) files.

The following table shows the flow of data and typical file size generated as the Genexus™ Dx Software processes data.

---

IMPORTANT! Data from approximately 200 sequencing runs can be accommodated on the sequencer before disk space becomes a limiting factor and data archiving is required for runs that use a GX5™ Chip if 1.wells files and intermediate files are marked for deletion, after a cleanup threshold for disk space is reached. You can perform approximately 45 to 50 whole chip runs without deleting any wells files on the system. For more information about data archival of the analysis files, see “Manage backup settings and restore runs (administrator)” on page 166.

---

| Process | File type | File size |
|---|---|---|
| Sequencing[1] | DAT | 2 TB |
| ▼ | | |
| Signal Processing [2] | Wells | 250 GB |
| ▼ | | |
| Base Calls (reads) | uBAM | 15 GB |
| ▼ | | |
| Mapped Reads | BAM | 15–20 GB |
| ▼ | | |
| Variant Calls | VCF | 120 Kb |

[1] The sequencing raw data (DAT) files are deleted from the sequencer after run completion.

[2] A configurable disk space cleanup process is executed nightly to free up space for future runs and to help ensure that sufficient hard disk space is maintained. If the system memory available for storing run analysis files reaches the threshold that is configured, the system auto-deletes 1.wells files for the oldest completed runs. A minimum of 1TB of space is required to start new runs. If the system approaches the 1TB threshold, an alert notifies you when you sign in. New run cannot be started if the 1TB threshold is reached. If desired, you can back up 1.wells files to an external drive.

# 7 Create, view, and sign reports


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- Run report ..... 137


Genexus™ Dx Software provides two types of reports.

- Variant report - A PDF report of the results for a sample in a run. You can customize the layout and contents of a variant report, or you can use a system-installed report template. Multiple users can electronically sign the variant report and one user can be designated to lock the report after sign off.
- Run report - A PDF report of the information that is shown in the **Run Report** tab. The run report includes assay metrics and the record of reagents that were used in a run. For more information, see “Assay metrics and the run report” on page 106.

To save or distribute a variant or run report, you can download the report.

## Variant report

The variant report is a PDF report of the results for each sample in a sequencing run. You can use a system-installed report template to generate a variant report, or you can customize the layout and contents of a variant report.

To make a variant report available for each sample result upon completion of a sequencing run, enable **Generate Report** in the **Setup** step when you plan the run. For more information, see Chapter 4, “Plan and manage runs”). To generate a variant report after a run is complete, see “Generate a variant report” on page 127.

When a variant report has been generated for a sample result, it is available for download in three places:

- In the **Results / Sample Results** screen when you place the pointer over the row for that sample, then click the **Report** link to download the PDF.
- In the variant report pane for the sample results in the **Reports** tab when you click ⋮ **(More)►Download Report**.
- In the **Results** screen for the sample when you click ••• **(More Options)►Download Files**.

Variant reports can be electronically or manually signed by users. Electronic signatures are shown in the Electronically Signed By section of the report, if the section is included in the report template.

For information about the sections that can be included in a variant report, see “Sections of a variant report” on page 128.

## View a variant report

You can view a variant report in Genexus™ Dx Software. A variant report includes details of sample results.

A variant report must be generated before you can view the report. If a report template is set as the default template for the assay associated with the run, one or more variant reports are generated after a run completes successfully. For more information, see “Create a report template (manager/administrator)” on page 132. Alternatively, you can generate one or more variant reports after a run completes successfully. For more information, see “Generate a variant report” on page 127.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.
3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown. The pane for a variant report includes the following information.
   - Report name
   - Report template name
   - Date that the report was generated
   - Username for the user who generated the report
   - Language used for the report
   - If a variant report has been electronically signed at least once, the signature information is listed in the pane for the variant report. If multiple users have signed the report, you can place the pointer over the electronic signature information to view more details. If the electronic signature that is used is associated with a lock-level signature type, the report is locked.
   - If a variant report is locked, **(Lock)** is shown in the upper right corner of the pane.
4. In the pane of the report of interest, click **(More)►Download Report**.

The report opens in a new browser tab. You can download variant reports as PDF files.

## Download a variant report

You can download a variant report for a sample result of interest from the **Results / Sample Results** screen, from the pane that shows the report, or as part of a download of results files.

1. In the menu bar, click **Results**►**Sample Results**.
2. Download the report with one of the following options.

| Option | Procedure |
|---|---|
| Download a ZIP file that contains all variant reports for a sample result. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click ••• **(More Options)**►**Download Files**. |
| Download the variant report in PDF format from the **Reports** tab for the sample results. | Click the **Reports** tab then, in the pane of the report of interest, click ⋮ **(More)**►**Download Report**. |
| Download the variant report as part of the results files for a specific sample in the **Results** screen. | For more information, see “Results files” on page 119. |

   A ZIP file that contains the PDF report is downloaded to the computer if you download reports from the **Sample Results** screen or as part of the results files. A report in PDF format downloads if you use the **Reports** tab for the download.

3. Extract the downloaded files, then open the PDF file in an appropriate viewer.

## Delete a variant report

You can delete a variant report that is not locked for electronic sign-offs if you have account permissions to electronically sign reports.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.
3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown.
4. In the pane of the report of interest, click ⋮ **(More)**►**Download Report**, then click **Delete**.
5. Click **Yes** to confirm that you want to delete the report.
   The pane for the report is removed from the **Reports** tab.

## Generate a variant report

You can generate a new variant report for sample results after a run is complete. A **(Lock)** in the variant report indicates that the electronic signature option for the report is locked. After a variant report is locked, the report cannot be electronically signed by any other user.

You can generate multiple reports for a sample result, if each report is named uniquely, and is generated in a different language, or uses a different report template.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Results / Sample Results** screen, click the sample of interest in the **Sample Name** column.
3. Select the **Reports** tab, then click **+ Generate Variant Report**.
4. In the **Generate Report** dialog box, change the name of the report that is generated by the software, if needed.
   a. Select the report template and language of the report.
   b. If the option to make custom text **Editable on Report Generation** was selected when the report template was created, enter a title in the **Custom Text** section and if needed, a description.

   Report templates that are associated with the assay that was used in the run are included in the **Report Template** list. For more information, see “Create a report template (manager/ administrator)” on page 132.

   If you select the same report template that was used to generate a variant report, and have not locked that report, the new selections you make override the previous variant report.
5. Click **Generate**.
   A draft version of the report is added to the **Reports** tab.

A pane for the new report is added next to the Run Report pane in the **Reports** tab. Reports that have been generated are available for download in the **Reports** tab, in the **Sample Results** screen, and in the ZIP package that contains results files.

## Sections of a variant report

Manager- and administrator-level users can create custom report templates that can include various sections. Each section contains details about a result, so you can include information to meet laboratory requirements.

A variant report can contain the following sections and information, depending on the assay used.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the variant report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other optional fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered into the software. You can customize the format of the Sample Details section when you create a new report template. For more information, see “Create a report template (manager/administrator)” on page 132. |
| Biomarker Results Summary | A summary of results for biomarkers detected in the sample. There can be multiple subsections that indicate levels for the biomarkers, such as Level 1, Level 2, Level 3. The levels for the biomarkers are based on the software assay. |
| QC | • Run QC—A summary of the run quality control metrics<br>• NTC QC – DNA—A summary of the DNA no template control quality control metrics<br>• NTC QC – RNA—A summary of the RNA no template control quality control metrics<br>• Positive Control01 QC – DNA—A summary of the DNA positive control quality control metrics<br>• Positive Control01 QC – RNA—A summary of the RNA positive control quality control metrics<br>• Sample QC – RNA—A summary of RNA sample quality control metrics<br>• Sample QC – DNA—A summary of DNA sample quality control metrics<br>• Templating Control QC – CF-1—A summary of the CF-1 quality control metrics<br>This section is locked and cannot be customized. |
| Sequencing Run Details | Contains the following subsections:<br>• Assay Name—The assay used in the run<br>• Panel—The panel used in the run<br>• Started On—The date and time that the run was started<br>• Completed On—The date and time when the run was completed<br>• Operator—The user who sent the run to the instrument<br>• Instrument—The name of the instrument<br>• Serial Number—The serial number of the instrument<br>This section is locked and cannot be customized. |
| Reagents | Details about run consumables, including part numbers, expiration dates, and lot numbers.<br>This section is locked and cannot be customized. |

*(continued)*

| Section | Description |
|---|---|
| Kit Barcodes | Details about each barcode for the kit that is used in the run, including sample name, assay name, nucleic acid type, extraction kits and control kits. |
| Key Metrics | Includes amplicon information for the results, such as the average base coverage depth, uniformity of base coverage, and percent reads on target. The options that are available to customize in this section depend on the assay that you select for the report template. |
| Electronically Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |
| Footer | The footer is repeated on each page of the report. |
| Signature | A section for a physical signature of the printed report. |
| Custom Text | A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Variant details | Details about variants in the run. You can customize the title of this section and the columns that are included in the report. |

## Sign off variant reports

Manager, administrator, operator, and report-level users can electronically sign a variant report for sample results in Genexus™ Dx Software. A sample result can have multiple variant reports only if each report uses a different report template, and a different language.

IMPORTANT! If you use the same template and language to regenerate an existing report, the existing report will be overwritten.

Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign the report, you will be the last person to sign the report before it is locked.

The signature information appears in the variant report PDF file or a user-created report, if electronic signatures are included in the template that is used to generate the report. For more information, see “Variant report” on page 124.

Multilanguage support for PDF report generation is provided.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

You must have electronic permissions enabled to sign reports. Reports that can be electronically signed must be labeled as **Draft**, and not be locked.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample result of interest in the **Sample Name** column.
3. In the **Reports** tab, in a variant report pane that is labeled as **Draft**, click **Sign Off**.
   The **Sign Off Report** dialog box opens and shows the name of the report, the template used for the report, and the following information, which is based on the profile of the user who signs the report.
   - **Electronic Signature**
   - **User Name**
   - **Language**
4. In **Password**, enter the password.
5. In **Sign Off Comments**, enter a comment.
6. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.
   If you entered footer information in the **Footer Field** when you created a report template, or if text was entered during a previous sign off, the same footer information is shown in the **Sign Off Report** dialog box. You can edit the text in the **Footer Field** to change the information that will be shown in the footer of the PDF report.
7. Click **Sign Off** to confirm the electronic signature.
   The pane for the signed variant report is updated to include the signature information for the variant report. If the electronic signature that is associated with a lock-level signature type is used, a **(Lock)** is shown in the variant report pane and the report is locked.
8. In the menu bar, click **Results►Sample Results** to return to the **Results / Sample Results** screen.
   If the **Electronic Signatures** column is shown in the **Sample Results** screen, the electronic signature types for users who signed the report are listed, and a **(Lock)** is shown for reports that are locked.
9. In the row of the sample of interest, click **Report** to download the report.

The report is signed. The **Electronic Signatures** section of the report pane is updated to include your username and the signature type for your account. You can hover over the signature type to show the list of signature types that are completed for the report. If multiple electronic signatures are added to the report, an electronic signature type for each signer is shown in the **Electronic Signatures** column of the sample results table.

You cannot generate new reports from the variant results if a report from those results is signed off and locked. If you must change the report, reanalyze the sample and generate a report for the reanalyzed sample.

You can send notifications for report sign-offs to other users. For more information, see “Send a notification to another user” on page 131. For information to download the signed report, see “Download a variant report” on page 126.

## Send a notification to another user

You can send notifications to other users in Genexus™ Dx Software. For example, you can notify another user when you have signed a variant report.

To receive email notifications, the recipient must have an email address that is configured in the software. For more information, see “Configure email notifications (manager/administrator)” on page 163.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample result of interest.
3. In the upper right corner of the screen, click ⋯ **(More Options)►Send Notification**.
   The **Send Notification** dialog box opens.
4. In **Send Notification to**, select one or more administrator-level users of interest.
5. In **Comments** enter the message.
6. Click **Send** to send the message.

An email message with the text that you entered is sent to the users that you selected. In addition, when a recipient signs in to the software, an unread notification, (Notifications), is shown. For more information, see “View notifications” on page 17.

# Report templates (manager/administrator)

Manager- and administrator-level users create and manage custom report templates that are used to generate PDF reports after a run. Users select a report template when they plan a run.

## Preview a report template (manager/administrator)

Manager- and administrator-level users can view custom and system-installed report templates in Genexus™ Dx Software.

For more information about system-installed report templates, see “Default report templates” on page 218.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
   Existing report templates are listed in this tab, including system-installed report templates. For more information, see “The report templates table” on page 132.

3. In the **Report Templates** table, place the pointer over the row of the report template that you want to view, then click **Preview**.
   The **Report Template Preview** opens.

4. In the **Report Template Preview** screen, click **Open** to download a PDF file of the report template.
   a. Click **Close** to close the **Report Template Preview** and return to the list of report templates.

You can place the pointer of the row of a report template to perform more actions, such as lock, edit, delete, or add notes to a report template. The options that are available depend on whether the report template is a draft or is locked.

### The report templates table

You can view details about report templates in Genexus™ Dx Software.

Details about report templates

| Item | Description |
|---|---|
| Name | The name of the template. System-installed report templates are included with the software. Custom report templates are named by the person who creates the template. |
| Application Category | The application category that is associated with the report template. |
| Assay Name | The name of assay that is associated with the template. |
| Status | Status of the report template.<br>• Locked<br>• Draft |
| Default | Indicates if the report template is designated as the default report template. |
| Created On | Date on which the report template was created. |

## Create a report template (manager/administrator)

Manager- and administrator-level users create custom report templates that are used to generate PDF reports after a run. As you create the report template, you can reorder sections, and add or remove sections. For more information, see “Sections of a variant report” on page 128.

To create a new report template, you must start from an existing report template, and then make changes to the layout and content of the sections if needed.

IMPORTANT! For a report template to be used in a run, it must first be locked.

1. In the menu bar, click **Assays**►**Manage Presets**.

2. In the **Manage Presets** screen, click the **Report Templates** tab.
   Existing report templates are listed in this tab, including system-installed templates.

3. Click ＋ **Add New**.

4. In the **Report Template** screen, enter a unique report template name, then complete the remaining sections.

   IMPORTANT! When you enter a name for the report template, use only alphanumberic characters (A-Z, a-z, 0-9), and the special characters period (.), hyphen (-), underscore (_), and space.

   If other special characters are used in the report template name, you cannot view or download variant reports that generated with the template.

   Note: The application category determines the sample attributes that are available to add to the report template.

   a. In **Application Category** select the application category that you want to associate with the report.

   b. In **Select Assay**, select the assay that you want to associate with the report.

      Note: After you associate an assay and application category with a report template, that report template becomes available for use with samples for that application category that are processed with the associated assay.

   c. In **Start From Existing Report Template**, select a report template to start from an existing report template if desired.

      Only report templates that are associated with the assay and application category that you select are listed.

   d. In **Languages Setting**, select the language for the report.

   e. Enable or disable the option to set the report template as the default template.

      When a report template is set as default, a report is generated from the report template each time you complete a run with an assay that is associated with the report template. Each assay can be associated with 3 default report templates in various languages. A report pane for each report is visible in the **Reports** tab of the sample results table. If you do not designate a report template as default, you can generate reports from the report template after a run is complete in the **Reports** tab. For more information, see “Generate a variant report” on page 127.

      - To set the new report template as default, keep the **Set as Default** toggle switch enabled.
      - If you do not want the report template to be set as default, disable the **Set as Default** toggle switch.

5. Add information to the **Header** section.

   The **Header** and **Footer** sections are required. All other sections are optional. For more information about the sections that can be included in a report, see “Sections of a variant report” on page 128.

   a. Enter a report title and the organization address.

   b. To include a logo on the report, in the **Logo** field, click **Select File** to browse to the logo file.

      Acceptable formats are PNG, GIF, and JPG.

   c. In **Header Fields**, click the field, then in the list that appears, select a term to add it to the **Header**.

d. Enable or disable the **Report Date** and **Assay Name** checkboxes if needed.

6. In the **Configuration** pane on the left, you can add new sections, then move or delete sections from the template if needed.
   a. In **Add Section**, select the section, then click ➕ **(Add)**.
      The new section is added at the bottom of the list.
   b. Use the following options to move or delete sections from the template.

| Option | Description |
|---|---|
| Add multiple sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, for each section to include, click the section name of interest, then click » **(Add)**.<br>Repeat for each section to include in the report template.<br>c. Click **Apply**. |
| Remove a section. | In the box for the section, click ✕ **(Remove)**. |
| Remove multiple sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click the section name of interest, then click « **(Remove)**.<br>Repeat for each section to remove from the report template.<br>c. Click **Apply**. |
| Add all sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, click **Select All**.<br>c. Click **Apply**. |
| Remove all sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click **Select All**.<br>c. Click **Apply**. |
| Rearrange sections of the report template. | Click-drag the section boxes to rearrange the order. |

You can add the following items and sections to a report template multiple times. Each section, except for a page break, can be uniquely customized.

- **Variant Details**
- **Custom Text**

  You can add text that can be edited when you generate a report. For more information, see “Generate a variant report” on page 127.
- **Page Break**

The following items and sections can appear in the report template in one place.

- **Key Metrics**
- **Manual Signatures**
- **Sample Details**
- **QC**
- **Sequencing Run Details**
- **Reagents**
- **Electronic Signatures**

7. Enter and customize the information for each section of the report template.
   a. For each unlocked section of the template, in the **Configuration** pane on the left, click the section name of interest.
   b. In the **Section** pane on the right, add or remove information for the section.
      You can click any customizable field to populate a list of available options for the field.

   Sections that are locked and cannot be customized are designated with (locked).
8. To preview the report layout, click **Preview Report**.
   You must enter information in the required fields before you can preview a report template.
9. When you are finished, click **Save**.
   The new report template appears in the **Report Templates** list.

## Edit a draft report template (manager/administrator)

You can edit a variant report that is not locked for electronic sign-offs if you have account permissions to electronically sign reports. A locked report template cannot be edited.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to edit, then click **Edit**.
4. Edit the sections of the report template as desired, then click **Save**.

The report template remains a draft until you lock the report template. For more information, see “Lock a draft report template (manager/administrator)” on page 136.

## Delete a draft report template (manager/administrator)

When a report template is in draft status, it can be deleted from the software. Only draft report templates can be deleted.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to delete, then click **Delete**.
4. In the **Delete Report Template** dialog box, click **Yes** to confirm the deletion.

The draft report template is removed from the software.

## Lock a draft report template (manager/administrator)

Manager- and administrator-level users can lock draft report templates in the **Assays / Manage Presets** screen under the **Report Templates** tab to manage report templates.

When a manager or administrator first creates an report template, the status of the report template is **Draft**. After a report template is locked, it cannot be edited or deleted.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to lock, then click **Lock**.

The report template is now locked and cannot be edited or deleted.

## View or add notes to a report template (manager/administrator)

Manager- and administrator-level can view existing notes or add new notes to draft or locked report templates in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template of interest, then click **Notes**.

| Action | Description |
|---|---|
| View existing notes for a report template. | In the **Add Notes** dialog box, notes that are associated with the report template are listed by user, date, and time. |
| Add a new note to a report template. | a. In the **Add Notes** dialog box, click + **Add Note**.<br>b. Enter the note, then click **Save**.<br>The note is added to the report template and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time. |
| Delete a note from a report template | In the **Add Notes** dialog box, in the row of the note that you want to delete, click 🗑 **(Delete)**. |

Click **Close** to return to the list of report templates.

# Run report

The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Assay metrics and the run report” on page 106. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

## View a run report

You can view or download a run report in Genexus™ Dx Software.

For information to download a run report, see “Download a run report” on page 138.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.
3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown. The name of the report is listed at the top of the pane.
4. In the **Run Report** pane, click **View Report**.

The run report opens in a new browser tab.

## Download a run report

You can download a run report summary in PDF format. The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Assay metrics and the run report” on page 106. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
   Multiple panes including a **Run Report** pane, a **Variant Report** pane, and any panes for customized reports that have been generated are shown.
4. In the **Run Report** pane, click **Download Report** to download a run report summary in PDF format.

# 8 View and manage assays (manager/administrator)


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## Assays in Genexus™ Dx Software

Assays contain the settings and parameters for library preparation, templating, controlling the sequencing run, analyzing, and reporting the results. Assays also define the panels, kits, and chips that are used in a run, and specify the reference files and threshold values for quality control and variant detection. The software files that contain the assay settings and parameters are packaged in a ZIP file called an assay definition file (ADF).

Assays can contain system-installed annotation sets, copy number baselines, sequence variant baselines and exon tile fusion baselines.

An assay is a reusable experimental design that contains predefined settings appropriate for use with common types of applications. An assay can be used to plan many runs and plays an important role in enabling rapid throughput across the sequencing instrument. Assays help reduce the chance of errors, because information is stored and then applied to runs instead of entered manually for each run.

## Find an assay

You can sort, search and filter information to narrow the list of assays shown in the **Assays / Manage Assays** screen, then find a specific assay in Genexus™ Dx Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for assays” on page 140.

For information to filter assays by a common trait, such as the name of the creator of the assay, see “Filter and search in the list of assays” on page 141.

## Search for assays

You can search for assays in Genexus™ Dx Software with keywords or text that are included in the name or full name of an assay.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

1. Click **Assays**►**Manage Assays**.
2. In the **Manage Assays** screen, in the **Assay Full Name** column heading or **Assay Name** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay name of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to the name of an assay to select one or more assays.
      - Click **Select All** to select all assays listed in the filter dialog box.

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of assays is shown in the **Manage Assays** screen.

Click **Clear Filters** to remove all filters and view the full list of assays.

## Sort the list of assays

You can sort the list of assays to make it easier to find assays in Genexus™ Dx Software.

1. Click **Assays**►**Manage Assays** to open the **Manage Assays** screen.
2. Click a column heading in the list of assays.
   If the column is sortable, the contents of the column is sorted alphabetically, or chronologically.
3. Click the column heading again to reverse the order.

## Filter and search in the list of assays

You can apply filters to narrow search results, or shorten the list of assays when you view the list of assays in Genexus™ Dx Software.

You can also search for assays in Genexus™ Dx Software with keywords or text that are included in an assay name.

---

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. Click **Assays►Manage Assays** to open the **Manage Assays** screen.
2. Click the **Filter Assays By** dropdown menu, then apply the filters.
   - Select **Status** to expand the list of options for status, then select a status:
     - Obsolete
     - Locked
   - Select **All** to return to the complete list of assays.

3. Select from the filter and search options in the column heading of interest.

| Option | Description |
|---|---|
| Filter and search by name of an object. | Click **(Filter)** in the column heading of interest, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial name for the object. Select one or more matches from the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen. |
| Filter and search by application type. | Click **(Filter)** in the **Application Type** column, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial name of the application type. Select one or more matches from the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen.<br>• Select from the application types shown, then click **Filter**. |
| Filter and search by the creator of the assay. | Click **(Filter)** in the **Created By** column, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial assay name. Select one or more matches listed in the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen.<br>• Select from the user names that are shown, then click **Filter**. |
| Filter by the exact or approximate date on which assays were created. | Click **(Filter)** in the **Created On** column, then use the options to filter by a date or range of dates to filter the list based on the assay creation date. |

4. *(Optional)* Click **Clear Filters** to return to the entire list of assays.

The list of assays is filtered according to the selections.

# View annotation set details

You can view details about an annotation set in Genexus™ Dx Software.

1. Click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, in the **Annotation Sets** tab, click the annotation set name.

The following details for the selected annotation set are shown.

Annotation Set Details

| Item | Description |
|---|---|
| Analysis Pipeline Version | The analysis pipeline version that is used by the annotation set. |
| Last Modified By | For custom annotation sets, the sign-in name of the person who created or modified a custom annotation set. |
| Last Modified On | Date on which a custom annotation set was last modified. |
| Created By | Custom annotation sets show the sign-in name of the person who created the annotation set.<br>Annotation sets that are included with the software are labeled as System Installed. |
| Created On | Date on which a custom annotation set was created. |
| Status | Status of the annotation set, including:<br>• Locked |
| Reference | Genome reference that is available in the annotation set.<br>• hg19<br>• GRCh38 |
| Annotation Versions | A list of the annotations included in the annotation, and the version of each annotation. |

# Copy number baselines (manager/administrator)

A copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). A copy number baseline is associated with a panel file in the software. CNV analysis is based on reads counts for each amplicon.

Known sources of variability include pool imbalance (when an assay has more than one pool of amplicons), total number of reads, per-amplicon attributes of GC proportion, and length of the amplicon insert. In practice, we observe other variability that does not associate with known attributes yet is systematic.

Copy number estimates are made using a proprietary algorithm.

Genexus™ Dx Software includes predefined copy number baselines.

## View copy number baseline details (manager/administrator)

Manager- and administrator-lever users can view details for the system-installed copy number baselines.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
   The **Copy Number Baselines** table lists both system-installed copy number baselines.
3. Click the name of a copy number baseline.
4. Review the details in the **Copy Number Baseline Details** pane.
   The details that are shown depend on the setup of the copy number baseline.

Copy Number Baseline Details

| Item | Description |
|---|---|
| Created By | Sign-in name of the person who created the baseline. System-installed baselines are included with the software. |
| Created On | Date on which the baseline was created. |
| Number of Normal Samples | The number of normal samples that are included in the baseline. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Number of Samples | The total number samples that are included in the baseline. |
| Amplification Type | Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD). |
| Panel Name | Name of the panel that is included in the baseline. |
| Reference | The reference genome for the baseline. |
| Reference File Name | The name of the reference genome file that is used in the baseline. |

# Sequence variant baselines

A sequence variant baseline is used to reduce the number of false positives for panels that detect somatic variants.

Variant calling filters in Genexus™ Dx Software suppress and remove most sequencing errors. However, it is possible that some errors can pass through the filters and be reported as false positives. In somatic panels, sequencing errors that survive filtering are reported as false positives that require investigation.

A sequence variant baseline is a BED file that contains a list of alleles that are known sequencing errors. The baseline file includes detailed properties for each sequencing error. The errors and properties are based on known germline normal samples. When added to an assay in Genexus™ Dx Software, the baseline detects and filters out the errors in sequencing that could lead to false positive variant calls.

Genexus™ Dx Software includes predefined sequence variant baselines.

## View sequence variant baseline details (manager/administrator)

Manager- and administrator-lever users can view details for the system-installed sequence variant baselines.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Sequence Variant Baselines** tab.
   The **Sequence Variant Baselines** table lists the system-installed sequence variant baselines.
3. Click the name of a sequence variant baseline.

Review the details in the **Sequence Variant Baselines** pane. The details that are shown depend on the setup of the sequence variant baseline. Parameters used to create sequence variant baselines are also shown, but are not listed in this table.

Sequence variant baseline details

| Item | Description |
|---|---|
| Created By | Sign-in name of the person who created the baseline. |
| Created On | The date the baseline was created. |
| Number of Samples | The number of samples that are included in the baseline. |
| Amplification Type | Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD). |
| Panel Name | The name of the panel that is included in the baseline. |
| Chip Type | The semiconductor sequencing chip for the panel and baseline. |
| Reference | The reference genome for the panel and baseline. |

# Exon tile assay baselines

An exon tile assay baseline is used to model the amplicon expression variation within a gene. When added to an assay in Genexus™ Dx Software, the exon tile fusion baseline normalizes a test sample, corrects the coverage in exon-tile amplicons, and identifies the presence or absence of expression imbalance in a gene. Exon tile assay baselines that you have created can only be incorporated into some assays. Contact a Field Bioinformatics Specialist (FBS) for guidance.

An exon tile assay baseline is a TXT file that contains the following items.

- The normalized expression values of exon-tile amplicons for genes with imbalance assays.
- The median normalized expression for each amplicon.

Exon tile assay baselines are associated with a panel BED file in the software.

Genexus™ Dx Software includes predefined exon tile assay baselines.

## View exon tile assay baseline details (manager/administrator)

Manager- and administrator-lever users can view details for the system-installed exon tile assay baselines.

1. In the menu bar, click **Assays**►**Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
   The **Exon Tile Assay Baselines** table lists the system-installed exon tile assay baselines.
3. Click the name of an exon tile assay baseline.

Review the details in the **Exon Tile Assay Baselines** pane.

The details that are shown depend on the setup of the exon tile assay baseline.

Exon Tile Assay Baselines

| Item | Description |
|---|---|
| Created By | Sign-in name of the person who created the baseline. System-installed baselines are included with the software. |
| Created On | Date on which the baseline was created. |
| Number of Samples | The total number samples that are included in the baseline. |
| Amplification Type | Library amplification technology that is used in the baseline. |
| Panel Name | Name of the panel that is included in the baseline. |
| Reference | The file name of the fusion reference for the baseline. |

# Manage assays

You can view assay details, and view and download the audit trail of an assay in Genexus™ Dx Software.

Manager- and administrator-level users can also import an assay into the software.

## View assay details

You can view assay details in Genexus™ Dx Software.

1. In the menu bar, click **Assays**►**Manage Assays**.
2. In the **Manage Assays** screen, click the assay name of interest.

The assay details are shown.

If desired, click ••• **(More Options)**►**Audit** to audit the assay. For more information, see “Review and export an assay history” on page 148.

## Assay details

When you view assay details in Genexus™ Dx Software, you can view information about the components of the assay. The information that is shown depends on the type of assay.

You can install and use multiple versions of an assay in the software. When there is a new version of an assay that is available to download, the available assay definition file is listed in the **Settings / Software Updates** screen, in the **Software Updates** table. In some cases, the most recent version (w*x.x.x*) of an assay allows you to perform actions in the software that you cannot perform with previous versions of the assay. For more information about software updates, see “Software updates” on page 163.

Note: For system-installed assays or assays that you install in the software, the end of the assay name, for example w4.1.2 or w5.0.0, is the version number. The assay version number is not the same as the **Version** shown in the assay details. See the following table for more information about **Version**.

| Assay detail[1] | Description |
|---|---|
| Assay creation details | • Application Type–The type of application for the assay, such as DNA and Fusions.<br>• Created By–The sign-in name of the person who created the assay. Assays that are included with the software are labeled as System Installed.<br>• Analysis Pipeline Version–The version of the analysis pipeline used for the assay.<br>• Created On–The date on which the assay was created.<br>• Assay Name–The name and version of the assay.<br>• Version–The revision number of the assay. |
| Panel | In addition to other components, the panel section lists the reference files used in an assay.<br>One item that is listed is the Minimum Read Count Per Sample, which is the minimum number of reads per sample that are needed to achieve the coverage depth required for the assay. The minimum read counts per sample is used to calculate the number of samples that are allowed in a chip lane when you plan a run. |
| Reagents | In addition to other components, the reagents section lists the kits and chips for the assay. |
| • NTC QC – RNA<br>• CF-1 Control<br>• NTC QC – DNA<br>• Run QC<br>• Sample QC | The QC sections list the Quality Control (QC) metric values for the assay. |
| Plugins | The plugins sections lists the plugins that are associated with the assay. |
| Description | A short description of the assay, similar to the assay full name. |

[1] The details that are shown depend on the assay.

## Review and export an assay history

You can review, export, or print the entire history of changes for an assay. The history shows the original values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays►Manage Assays**.
2. In the **Manage Assays** screen, place the cursor in the row for the assay of interest, then click **Audit**.
   The **Audit Details** screen opens with time-stamped audit records that include assay details and changes for each action performed on the assay.
3. *(Optional)* Review the audit record.

   Details in the audit record

| Item | Description |
|---|---|
| User name | The name of the person who was signed in when the action was performed. |
| Assay Full Name | The name of the assay. |
| Data Object Name | The data object that is affected by the action. |
| Action Performed | Actions performed on the assay include:<br>• Create<br>Note: The create action includes imports of assays. |
| Time Stamp | The time and date of the action. |
| ID | An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier. |

4. *(Optional)* Click **Download** to download a print-ready PDF file of the audit record.
   You can open the PDF file in an appropriate viewer, then print the record from within the open document.
5. Click **Back** to return to the **Manage Assays** screen.

## Import an assay (manager/administrator)

A manager- or administrator-level user can import an assay from another Genexus™ Dx Integrated Sequencer if the assay has been first exported to a local drive.

Ensure that the name of the assay, the barcode kit, and the extraction method type for the source system and the target system are identical.

1. In the menu bar, click **Assays►Manage Assays**.
2. In the **Manage Assays** screen, click **Import Assay**.
3. In the **Import Assay** dialog box, click **Select Assay file**, then go to the folder on the computer that contains the exported assay and select the ZIP file for the assay.
4. Click **Upload**.

The assay appears in the list of assays in the **Assays►Manage Assays** screen.

# 9 System administration and management (manager/administrator)


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Manager-level users use tools in the **(Settings)** menu to:

Administrator-level users use tools in the **(Settings)** menu to:

- View and export audit records.
- Configure user email settings, set up laboratory information, and audit these settings.
- Create and manage account settings, and audit these accounts.
- Manage software updates.
- Manage data backup, disk cleanup settings, and disk space usage.
- Manage log files.
- Manage services for servers, services, and active jobs.
- Create and manage user accounts.

# Accounts for use with Thermo Fisher™ Connect Platform in Genexus™ Dx Software (manager/administrator)

Manager- and administrator-level users can configure Genexus™ Dx Software to link to one or more Connect Platform accounts.

The first Connect Platform account that is added in the software is designated as the administrator (Admin) account and cannot be unlinked or removed.

Active Connect Platform accounts can perform the following tasks in the Genexus™ Dx Software:

- Download the latest software updates
- Download assay definition files
- Download configuration packages

Note: Configuration packages contain software updates, such as updates for kits and chips, documents such as an updated End-user License Agreement (EULA), Help documentation, and so on.

## Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)

Manager- and administrator-level users can link their Connect account at Genexus™ Dx Software with a QR code generated in the software.

When a Connect account at **thermofisher.com/connect** is configured and active, administrator-level users can perform the following tasks in the Genexus™ Dx Software:

- Download the latest software updates
- Download assay definition files
- Download software configuration packages

Before you link a Connect account to create a **Thermo Fisher Connect** account, you must have a valid Connect account. You must also have the InstrumentConnect mobile app to link the account with a QR code that is scanned on a mobile device. For more information, see the *Thermo Fisher™ Connect Platform Help System* (Pub. No. MAN0010954).

1. Click **(Settings)►Thermo Fisher Account**.
2. Click **Link Account to Device**.
   The **Link Account to Device** dialog box opens and a QR code image and QR code text are shown.
3. Link the Connect account with the software.

| Option | Description |
|---|---|
| Link a Connect account with the InstrumentConnect mobile app | a. Open the InstrumentConnect app, then sign in.<br>b. In the app, click +.<br>c. Click QR Code, then scan the QR code shown in the **Link Account to Device** dialog box in Genexus™ Dx Software. |
| Link the account through the Connect website | a. Click the link in the QR Code to open the Thermo Fisher Connect website.<br>b. In the Connect website, click **InstrumentConnect** in the left sidebar.<br>c. Click **Link instrument** in the upper right corner of the screen.<br>d. Enter the QR code text shown in the **Link Instrument** screen.<br>e. Click **Link**. |

The linked account is listed in the **Settings / Thermo Fisher Account** screen. A successfully authenticated account has **Active** listed in the **Status** column.

When the **Thermo Fisher Account** is active, software updates are available in Genexus™ Dx Software. Administrator-level users can download and install the updates. For more information, see “Software updates” on page 163.

## Unlink a Thermo Fisher™ Connect Platform account (manager/administrator)

Manager- and administrator-level users can unlink Thermo Fisher™ Connect Platform accounts in Genexus™ Dx Software.

The first Thermo Fisher™ Connect Platform account that is added is designated as the administrator (Admin) account and cannot be unlinked or removed in the software.

1. Click **(Settings)►Thermo Fisher Account**.
2. Place the pointer over the row of the account that you want to unlink, then click **Unlink**.

The Thermo Fisher™ Connect Platform account is no longer linked in Genexus™ Dx Software.

## View the changes made to a connected account (manager/administrator)

Manager- and administrator-level users can view the changes made to connected accounts in Genexus™ Dx Software.

1. Click ⚙ **(Settings)►Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to audit, then click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected account.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made to the selected account.
5. *(Optional)* Click **Download** to export the record of the single change to a PDF file.

# User accounts (administrator)

Administrator-level users can perform user management actions in the **Settings / User Management** screen.

- Add new user accounts
- Assign user privileges (roles)
- Edit user information
- View user account audit trails
- Manage user account security policies

## Add a new user account (administrator)

Administrator-level users add new user accounts to the software, and grant some users permissions to electronically sign reports.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 163.

1. In the menu bar, click ⚙ **(Settings)►User Management**.
2. In the **User Management** screen, click + **Add New**.
3. In the **Create User** dialog box, enter the user account information.
   User names can contain alphanumeric characters and underscores, periods, and hyphens.
4. From the **Role** list, select the appropriate user-access level (**Administrator**, **Manager**, or **Operator**.
   The **Report** role has electronic signature permissions enabled by default.

5. Grant permissions for a manager, administrator, or report user to electronically sign reports.
   a. Enable the **Electronic Signature** toggle switch.
   b. Select an **Electronic Signature Type** to associate with the user account. For example, **Approved**. A **(Lock)** is shown when you select the **Electronic Signature Type** that is configured to lock a report upon sign off.
6. Click **Save**.
   The new account is added to the list in the **User Management** screen. The software sends an email notification containing a temporary password to the email address of the new user.

## Sort the user list (admin)

Administrator-level users can sort the user list by user name, first name, and last name.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, click the column for **User Name**, **First Name**, or **Last Name** to sort the column alphabetically.
3. Click the column name a second time to reverse the order.

## Edit user details (administrator)

Administrator-level users can edit user details in the **Settings / User Management** screen. An administrator can also suspend or disable user accounts. A history of user account changes is maintained by the audit feature.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, place the pointer over the row of the user account, then click **Edit**.
3. In the **Edit User** dialog box, enter changes in the appropriate account information fields.
   - In the **Role** list, select the user-access level (**Administrator**, **Manager**, or **Operator**.
     The **Report** role has electronic signature permissions enabled by default.
   - To grant permissions for administrator, manager or report users to sign reports, enable the **Electronic Signature** toggle switch, and select an **Electronic Signature Type**.
     A **(Lock)** is shown when you select the **Electronic Signature Type** that is configured to lock a report upon sign off.
   - In the **State** list, select the appropriate state (**Active**, **Suspended**, or **Disable**).
4. Click **Save** to make the changes.

Changes that are made to the user account are shown in the **User Management** screen, and shown in the **Audit Trail** screen for the user.

## Reset a user password (Administrator)

An administrator-level user resets passwords for user accounts. A history of user password changes is maintained by Genexus™ Dx Software.

Periodically, the software sends a system-generated email notification to a user that their password is about to expire. The account holder can follow the instructions in the email to change a password. A user can also request that an administrator reset their password.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 163.

Users with administrator accounts can change a password immediately, or send an email to a user who requests to have a password changed.

1. To change a password immediately, do one of the following actions, depending on the account with which you sign in and the type of user account requires a password change.

| Sign-in requirements for password change | Action |
|---|---|
| To change the password for any user account, except for the system adminstrator account, you must be signed in with an administrator account. | • Click ⚙ **(Settings)►User Management**, then place the pointer over the row of the account for which the password will be changed, then click **Reset Password**.<br>• In the **Change Password** dialog box, enter a new password in the **New Password** field and re-enter the new password in the **Confirm Password** field, then click **Change**. |
| To change the password for any user account, including the system administrator account, you must be signed in with the account for which the password will be changed. | • Go to 👤 **(Profile)►My Profile**, then click the **Change Password** link.<br>• In the **Change Password** dialog box, enter the information in the **Current Password**, **New Password**, and **Confirm Password** fields, then click **Change**. |

   The confirmation message "Password changed successfully" is shown to indicate the password reset.

2. To send an email to a user who requests to have a password changed, click ⚙ **(Settings)►User Management**, then place the pointer over the row of the account for which the password will be changed, and click **Reset Password**. Then in the **Change Password** dialog box, click **Send Password in Email** to send a software-generated password in an email message to the user.
   A confirmation message is shown to indicate the email was sent to the user.

## View user account history (administrator)

An administrator-level user can view the user account history in Genexus™ Dx Software. The user account audit trail is a record of when the user account was created and modified.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, place the pointer over the row of the user of interest, then click **Audit**.
3. In the **Audit Details** screen, view the audit trail details for the selected user. For more information, in the **Audit Record Details** column, click the time stamp of interest.
4. *(Optional)* In the **User Management** screen, click **Download** to export the record to a PDF file.

## Set user account security policies (administrator)

An administrator-level user sets security policies for Genexus™ Dx Software user accounts. User account security policies include the permissible number of failed sign-in attempts, password lifetime, and the length of inactivity before users are automatically signed out of the software. These settings apply to all users.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, click **Policies**.
3. In the **Policies** dialog box, select the appropriate values to set the user-account suspension period, password, and session timeout policies.
4. Confirm that the **Enabled** toggle switch is set to enable the session policy settings.
5. Click **Save**.

## Configure an SSH key (system administrator)

System administrator-level users can set up an SSH key in their user profile in the **Profile / My Profile** screen. SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives administrator-level users a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

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IMPORTANT! Ensure that you use only an SSH key that is password protected. The software does not enforce the use of password protected SSH keys, but the use of a password for the SSH keys is strongly recommended.

Do not share the SSH private key or the credentials for the SSH private key. The SSH private key should be used only by the user who creates the SSH private key.

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Configuring an SSH key is optional, but best practice is to configure one or more SSH keys. The ionadmin and ionservice users may need to configure SSH keys so that they can access the system at the operating system level to configure some system-level settings, for example mounting an archive directory on the system. A user can have more than one SSH key.

Before configuring an SSH key, you must first generate keys by using the Keygen command on your local system. When the public and private keys are generated, copy the text string from the public key to use in this procedure.

1. In the menu bar, click **(Profile)►My Profile**.
2. In the **SSH Keys** section, click **Add SSH Key**.
   The **SSH Keys** section is available only to ionadmin and ionservice users.
3. Complete the **Add new SSH Key** dialog box.
   a. In **Label**, type a unique name for this SSH key.
   b. In **SSH Key**, paste or type the SSH key string generated by using the Keygen command on your local system.
   c. Click **Save**.

# Audit records (administrator)

Administrator-level users can use the tools in the **Settings / Audit Records** screen to configure and manage audit records. Information that can be audited includes activities by users on samples, libraries, assays, planned runs, system setup of the sequencer, accounts, and software updates.

The **Audit Records** screen lists actions that are performed on any data object, including the user who performed the action, the action performed, the data object name, and the date and time of the action. Additional details concerning the action are available in an accompanying audit record.

The actions can be searched and sorted, and the audit records can be exported and printed.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

Note: Library batches and runs that are created in a batch with other objects in a single LIMS transaction do not have a "Create" action that is listed in the **Settings / Audit Records** screen. However, you can view complete library batch audit records from the **Samples / Manage Library Batches** and **Runs / Manage Runs** screens. Any subsequent actions that are performed on library batches are listed in the **Audit Records** screen. A library batch or run that is created individually in a LIMS transaction is listed.

## Search audit records (administrator)

An administrator-level user can use the search tools in the **Settings / Audit Records** screen to search for an existing audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.
2. In the **Audit Records** screen, select the search parameters.
   a. To select a date range, click **Calendar** next to **Start Date** and **End Date**.
      If an end date is not selected, the search results include all records from the start date through the current date.
   b. To limit the results list to actions that are performed by a specific user, select the user from the **Select User** list.
   c. To limit the results to specific actions, select from the **Select Action** list.
   d. To list more items in the table, select from the data object list.
      The list is set to **Sample** by default and all items listed in the **Audit Records** screen have the data object of **Sample** in the **Data Object Name** column. You can change the type of objects that are shown in the table. For example, when you select **Planned Runs** in the data object list, all planned runs are listed.
3. Click **Search**.
4. Click **Clear** to return to the default list of records, with the data object of **Sample** in the **Data Object Name** column.

## Sort audit records (administrator)

An administrator-level user can sort the columns in the **Settings / Audit Records** screen to display the list of audit records in a different order. The default order is the newest record on top.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.
2. In the **Audit Records** screen, click the column heading of interest.
   The list of records is reordered based on the heading selected. **User**, **Action Performed**, and **Data Object Name** sort alphabetically.
3. Click the column heading a second time to reverse the order of records displayed.
4. Click **Clear** to return to the sorted column to the default order.
5. Click the **Timestamp** heading to return to the default order, with the most recent record on top.

## Export audit records (administrator)

An administrator-level user uses the **Settings / Audit Records** screen to export a print-ready PDF file of selected audit records.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.
2. In the **Audit Records** screen, select the record or records to be exported by clicking the checkbox next to the record of interest. Select the checkbox above the column to select all of the records.
3. Click **Export**.
   A PDF file is generated. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the PDF file in an appropriate viewer to print.

## Update the audit configuration (administrator)

An administrator-level user configures the audit details by requiring that a reason for a change be included in audit records when a user changes information. When a reason is required, the user must supply a reason for change when editing the information, and the reason is included in the audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings) ►Audit Records**.
2. In the **Audit Records** screen, click **Audit Configuration**.
3. In the **Audit Configuration** dialog box, select the **Require Reason** checkbox next to the **Data Object Name** for which a reason for change is required.
4. Click **Save**.

   To remove a reason for change requirement, open the **Audit Configuration** dialog box, deselect the checkbox, then click **Save**.

# Configure electronic signatures (administrator)

Administrator-level users configure electronic signatures in Genexus™ Dx Software. For more information about electronic signatures, see “Sign off variant reports” on page 129.

## Manage electronic signature types (administrator)

Signature types in Genexus™ Dx Software are associated with electronic signature permissions to designate how a signature is used. Different types of signatures, such as review, approve, and lock, can be required to sign off on a variant report.

Administrator-level users can add new electronic signature types. Each user account can be associated with just one signature type. Signature types can be renamed, made obsolete, restored, or deleted. Changes to electronic signature types are shown in the audit trail for the signature type.

Designate one of the signatures to lock the report during sign-off.

1. Click **(Settings)►Configuration**.
2. In the **Configuration** screen, in the **Electronic Signature** section, manage signatures.

| Option | Description |
|---|---|
| Add new types of electronic signatures. | In the Bulk add list of comma delimited meaning of signature field, enter new types of signatures, separated by commas. For example, enter *Review,Inconclusive* to add two new signature types.<br>Signature types that are added can be associated with users who are granted permission to electrically sign reports. For more information, see “Add a new user account (administrator)” on page 153 and “Edit user details (administrator)” on page 154. |

*(continued)*

| Option | Description |
|---|---|
| Rename a type of signature. | Click Edit, then enter a new name for the signature type. |
| Associate locking a report with a type of signature. A report becomes locked when a user whose account is associated with the selected type of signature electronically signs the report. | In the Select a type of signature to lock report when signing off section, select the radio button next to the type of signature that you want to use to lock a report during report sign-off. |
| Obsolete a type of signature. | Click Obsolete to remove the signature type as an option during variant report sign-off.<br>If an obsolete signature type is associated with a user account, an alert is added to the User Management screen next to Electronic Signature Type. An alert is also shown if no signature type is associated with a user account that includes permissions for electronic signatures. The signature type must be edited by an administrator before the user can electronically sign reports. For more information, see “Edit user details (administrator)” on page 154. |
| Restore a type of signature. | Click Reactivate to reactivate a type of signature that has been made obsolete. |
| Delete a custom type of signature. | Click Delete to delete a type of signature. A custom electronic signature type can be deleted if is not associated with a user, is not set as lock-level, and is not used in electronic sign-offs for any reports. System-installed signature types cannot be deleted. |
| Review the changes made to a type of signature. | 1. Click Audit.<br>2. In the Audit Trail - Electronic Signature screen:<br>3. • Review the audit trail details for the most recent change.<br>• Click a time stamp of interest to view the details for other changes that have been made to the selected signature.<br>• Click Download to export the record of the single change to a PDF file.<br>• Click Back to return to the Configuration screen. |

# Configure email settings (manager/administrator)

Manager- and administrator-level users configure email settings. Administrator-level users can configure support contact information, such as the lab contact and the IT contact for the organization.

# Lab information

The **Lab Information** tab in the **Settings / Configuration** screen provides a reference for support personnel, listing points of contact if a problem with the connected Genexus™ Dx Integrated Sequencer occurs.

- **Lab Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer.
- **IT Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer hardware or the network environment.

IMPORTANT! Administrator and Manager-level users should review the contact information periodically to ensure that it is current and accurate.

## Edit the reference lab information (manager/administrator)

Manager- and administrator-level users can edit the reference lab information in Genexus™ Dx Software. You can view the history of changes to the contact information. For more information, see “View the changes made to the lab information (manager/administrator)” on page 162.

1. Click **(Settings)**►**Configuration**.
2. In the **Lab Information** section, below the **Lab Contact** or **IT Contact** pane, click **Edit**.
   The fields for the **Lab Contact** or **IT Contact** are now available to edit.
3. Enter the updated contact information.
4. Click **Save** to save the new selections or click **Reset** to revert to the original information.

## View the changes made to the lab information (manager/administrator)

Manager- and administrator-level users can view the changes made to the lab information in Genexus™ Dx Software.

1. Click **(Settings)**►**Configuration**.
2. In the **Lab Information** section, in the **Lab Contact** or **IT Contact** pane, click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected contact.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made for the selected contact.
5. *(Optional)* Click **Download** to export the record of the single change to a PDF file.

# Configure email notifications (manager/administrator)

Manager- and administrator-level users configure the Genexus™ Dx Software to send email notifications to a specified administrator.

Administrator-level users receive email notifications when there is a restore system failure. Administrator-level users can also select to receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. For more information, see “Change email notification settings (administrator)” on page 180.

1. Obtain the SMTP server URL and port information from your IT department.
2. Click **(Settings)**►**Configuration**.
3. In the **Configuration** screen, in the **Email Settings** section, enter the server and port information supplied by IT, and the Genexus™ Dx Software user name, password, and email address of the administrator.
   - Keep the **Use TLS** toggle switch enabled to send email notifications with transport layer security.
   - Click the **Use TLS** toggle switch to send email notifications without transport layer security.
4. Click **Update**.
5. Click **Send Test Email** to send a test email to the email address provided in the settings.

# Software updates

An administrator-level user can select **Software Updates** in the **(Settings)** menu to:

- See the currently installed software versions for each software module, assay definition files, plugin, and configuration package installed on the system under the **Installed Software** link.
- Check for new software updates that are available to be downloaded and installed under the **Software Updates** link.

For users who have set up a Thermo Fisher™ Connect Platform account, **(Notifications)** appears in the Genexus™ Dx Software banner on the top of the screen upon login when updated software packages are available for download from the Connect Platform.

Updated software packages are available from the App Store at **apps.thermofisher.com**. Software packages can include assay definition files, for example. Administrator-level users can download and install software directly from the App Store if a Connect Platform account has been configured. Alternatively, a USB drive can be used to install and update the software if the instrument is not connected to the internet.

| Option | For more information, see this section |
|---|---|
| Download and install software directly | "Download and install software from the App Store (administrator)" on page 164 |
| Download and install software with a USB drive | "Format a USB drive for a software update" on page 165<br>"Download and install software with a USB drive (administrator)" on page 165 |

Note: For first-time update of system software from the 5.100.2 version, contact a Field Service Engineer (FSE) to update the system software.

IMPORTANT! Before updating the software, ensure that the instrument is idle, no runs are in progress or are queued to run, and the screen on the instrument is the main menu screen.

## Download and install software from the App Store (administrator)

In order to download and install software updates from the App Store on **thermofisher.com/connect**, you must first:

- Configure a Thermo Fisher Connect account. For instructions, see "Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)" on page 151.
- Ensure that the Genexus™ Dx Integrated Sequencer has the latest software version, Genexus™ Dx Software 6.8.
- Connect the Genexus™ Dx Integrated Sequencer to the internet.

1. In the menu bar, click **(Settings)►Software Updates**, then click **Software Updates** in the upper right corner.
   The **Software Updates** screen opens with **App Store** enabled.
2. *(Optional)* Click the desired package name to view the installation details and version information in the **View Package Details** pane.
3. Click **Download** in the **Actions** column in the row of the software package of interest.
   After the download is complete, the **Download** link changes to an **Install** link.
4. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install.
   If you are performing a system software module installation, a command window opens to show the progress of the installation.

   Note: There may be a delay before the command window opens.
5. When installation of the software package is complete, click **Installed Software** to verify the installation.

## Format a USB drive for a software update

Prerequisites:

- An Ubuntu Linux desktop computer with disk utility software.
- A USB drive with a capacity of 128 GB or more.

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IMPORTANT! Ensure that the USB drive does not contain any viruses or malware before you insert the USB drive into the instrument.

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1. Insert the USB drive into the Ubuntu Linux computer.
2. With Ubuntu running, open the disk utility by navigating to **System ►Administration►Disk Utility**.
3. If the USB drive has partitions, delete the partitions.
   a. Under **Peripheral Devices from Storage Devices**, click the **USB Flash Drive (SMI USB DISK)** to select the USB drive.
   b. Highlight the volumes (New Volume FAT >60 GB).
   c. Click **Unmount Volume**, then click **Delete Partition**.
   d. Click **Delete** to confirm.
4. Select the free USB volume, then click **Create Partition**.
   The **Create partition on SMI USB DISK** window opens.
5. Deselect the **Take ownership of file system** checkbox.
6. Select **ext3** from the **Type** dropdown menu, then select the entire size of the partition. Name the partition *usbinstall*.
   The partition name must be completely lowercase.
7. Click **Mount**.
   The mount point of the USB drive is **/media/usbinstall**.

Proceed to “Download and install software with a USB drive (administrator)”.

## Download and install software with a USB drive (administrator)

In order to download and install software updates, you must first:

- Create or access a Thermo Fisher™ Connect Platform account. For instructions, see “Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)” on page 151.
- Format a USB drive for the software update. For more information, see “Format a USB drive for a software update” on page 165.

1. In the Ubuntu Linux computer where the USB is mounted, sign in to the Thermo Fisher App Store on Connect at **apps.thermofisher.com**.
2. On the left side of the App Store screen, click **(AppConnect)**.
3. On the right side of the screen, under **Resource Libraries**, click **Ion Torrent™ Genexus™**.
4. Click **Software Update**.
5. In the Ion Torrent™ Genexus™ Software table, select the row that contains the software package for the version that you want to install.
6. Click **(Download)** to download the package.
7. Copy the downloaded package (.zip) to the `/media/usbinstall` folder in the USB drive.
8. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the Genexus™ Integrated Sequencer.
9. In the menu bar of Genexus™ Dx Software, click **(Settings)►Software Updates**, then click **Software Updates** in the upper right corner.
10. Select **USB**.
11. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install.

When installation of the software package is complete, click **Installed Software** to verify the installation.

# Manage backup settings and restore runs (administrator)

Administrator-level users can view and manage backup settings and restore runs in Genexus™ Dx Software with the following actions:

- Monitor system storage space usage.
- Manage settings to backup and delete files and data.
- View an audit trail of any changes made.
- View and change email notification settings.
- Restore runs.

## Disk space usage and backup settings (administrator)

An administrator-level user can monitor disk space usage for the Genexus™ Dx Integrated Sequencer and can change data backup settings in Genexus™ Dx Software. System disk space usage information is available in the **Settings / Backup & Restore** menu. You can view the date and time of the last successful backup for data and analysis results and the date and time of the last successful backup for additional files such as sequencing output and intermediate files, based on the backup settings.

A best practice to maintain sufficient disk space is a systematic plan that archives run data, results files, variant information, and signed reports that you want to store to an external file storage system. A system administrator or Field Service Engineer (FSE) sets up a data backup strategy and disk cleanup settings. An administrator-level user then configures the backup schedule. For more information, see “Change the backup settings (administrator)” on page 175.

Before data can be backed up or archived, you must add a mount point. When files are backed up, the files are copied to an external storage device or to a cloud system. The files remain on both the sequencer and the external storage system. After data backup, files are saved in a folder in the external storage that is named `dx`. If multiple sequencers are connected to the software, the software generates a parent folder for each instrument.

When files are cleaned up, the files are deleted from the sequencer, but remain on the external storage system.

Backup and cleanup settings

| Option | Description |
|---|---|
| Back up system data and run results | Backup occurs daily, at approximately midnight, after an administrator-level user adds a mount point.<br>After backup, you can view the results for runs that are backed up in the Run Results and Sample Results screens. |
| Clean up system data and run results | If enabled, run results are deleted from the sequencer after a specified interval of time. For example, 12 months after a run is completed. Runs that are backed up and deleted are marked as Archived.<br>Cleanup settings do not apply for run results that you retain. When you retain run results, the run data, including the run results, 1.wells files, and intermediate files, are retained on the instrument and in the storage system. Runs that have been retained are shown in the software and are not deleted.<br>Archived runs are listed in the Run Results screen with the archived symbol ( ) after the run name. By default, archived runs are not listed. To see the archived runs, select Archived in the Filter runs by list. An administrator-level user can restore the runs to access the data. |

Backup and cleanup settings *(continued)*

| Option | Description |
|---|---|
| Back up intermediate files and 1.wells files | If enabled, intermediate files and 1.wells files are backed up at a specified time. For example, every Friday at 10 pm.<br>Because 1.wells files are 10 times larger than the run results files (BAM files), more time is needed to back up 1.wells files than to back up system data and run results. |
| Clean up intermediate files and 1.wells files | If enabled, intermediate files and 1.wells files are deleted when the system disk space falls below a specified threshold, such as 4 TB. If you have not configured the mount point, or if you have not configured the backup settings for additional files, files are cleaned up when the space threshold is met.<br>Cleanup settings do not apply for run results that you retain. When you retain run results, the run data, including the run results, 1.wells files, and intermediate files, are retained on the instrument and in the storage system. Runs that have been retained are shown in the software and are not deleted.<br>Because 1.wells files are 10 times larger than the run results files (BAM files), more time is needed to back up 1.wells files than to backup system data and run results. |

The software ensures that disk space is available for runs. If the system has less than 1 terabyte (TB) of unused disk space, no new runs can be started on the sequencer. In this case, an alert notifies the user that there is insufficient disk space when a run is set up on the sequencer. The run cannot proceed until data are backed up and deleted.

An administrator-level user can perform the following actions to free up system disk space, if needed.

- In the **Backup & Restore** screen, you can click **Start Now** to start a backup and cleanup at any time.
- In the **Run Results** screen, you can delete one or more run results.
- In the **Run Results** screen, you can archive one or more run results if you have added a mount point. The settings in the **Backup and Restore** screen are not applied when you archive runs in the **Run Results** screen.

Contact a local system administrator to backup and delete data manually. An administrator-level user can also change the schedule to backup and delete data.

Results and reports that have been backed up can be restored in Genexus™ Dx Software. For more information, see “Restore run results for a single run (administrator)” on page 181.

## Types of files for backup

There are three categories of files for backup in Genexus™ Dx Software.

| Category | Types of files | Cumulative size per run | Reanalysis use |
|---|---|---|---|
| Sequencing output | 1.wells | 250–500 GB | Reanalysis from the basecalling step |
| Intermediate files | Troubleshooting | 150–180 GB | Not applicable |
| Analysis results | • BAM<br>• VCF<br>• Reports | 150–180 GB | Reanalysis from the alignment step |

## Specifications and disk space usage

The amount of disk space that the Genexus™ Dx Integrated Sequencer uses for a run depends on the nucleic acid sample type and the assay that is used for sequencing. The system can store 40 to 50 whole chip data runs. If the system has less than 1 terabyte (TB) of available storage space, no new runs can be started. For more information, see “Disk space usage and backup settings (administrator)” on page 167.

The following table lists chip specifications and data usage for a run with the Ion Torrent™ GX5™ chip.

| Assay | Chip specifications and data usage |
|---|---|
| Sample type | FFPE |
| Number of reads per chip (4 lanes) | 48–60 million |
| Number of reads per lane | 12–15 million |
| Signal processing input size per chip[1] | ~2 terabytes |
| Basecalling input size per chip[2] | 200–300 gigabytes |
| Data size of intermediate and results files and logs[3] | 120–180 gigabytes |
| Data size per lane of intermediate and results files and logs | 30–45 gigabytes |
| Data size of backed up files and logs | 30–45 gigabytes |
| Data size of results files per lane[4] | ~8–24 gigabytes |
| Recommended number of samples per lane | 4 samples per lane |
| Data size of the results per sample | ~2–6 gigabytes |

[1] The signal processing input on the sequencer is deleted after analysis is complete and successful.

[2] Administrator- level users can schedule when basecalling input files are deleted. By default, these files are not retained. For more information, see “Set a schedule to permanently delete files (administrator)” on page 177.

[3] Includes QC, plugins, logs, results, and other assay and variant database files.

[4] For more information about results files, see “Results files” on page 119.

## Configure backup settings (administrator)

An administrator-level user views and configures sequencer backup settings in the **Settings / Backup & Restore** screen.

Update backup settings for various reasons, including:

- The backup location has limited free disk space remaining.
- The backup location is not accessible when the system tries to back up a run.

Consult a local Field Service Engineer to set up a backup system. For more information, see Appendix E, “Customer and technical support”.

The following options are available only after a backup directory has been configured:

- View the backup disk space.
- Change the backup settings.

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup Settings** tab, edit the backup settings.

| Option | Description |
|---|---|
| View the system disk space | View the chart that shows the percentage of system disk space that is used.<br>View the total amount of disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes. |
| View the backup disk space | View the chart that shows the percentage of backup disk space that is used.<br>View the total amount of backup disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes. |
| Change the backup directory | Change the backup directory.<br>For more information, see “Add or change the backup directory (administrator)” on page 171. |
| Change the email notification settings | Select when administrator-level users receive email notifications.<br>For more information, see “Change email notification settings (administrator)” on page 180. |
| Change the backup settings | Backup .wells files and intermediate files.<br>For more information, see “Change the backup settings (administrator)” on page 175. |
| Change the disk cleanup settings | Set the schedule to permanently delete analysis results files.<br>Set the conditions to permanently delete other files.<br>For more information, see “Set a schedule to permanently delete files (administrator)” on page 177. |
| Enable backup and cleanup | Click the Backup & Cleanup Enabled toggle switch to the enabled position to start the backup and cleanup process at the scheduled time. |
| Disable backup and cleanup | Click the Backup & Cleanup Enabled toggle switch to the disabled or off position for temporary instances, such as when the backup server is down.<br>This setting overrides all other backup settings. |

*(continued)*

| Option | Description |
|---|---|
| Start backup | Click Start Now to start:<br>• the backup of system data and analysis results to the selected backup directory.<br>• if enabled, the backup of sequencing output and intermediate files to the selected backup directory.<br>• data clean up based on cleanup settings.<br>Backup can take a considerable amount of time when the backup is set up for first time on the system. |

3. Click **Save**.

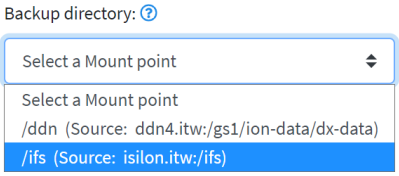
## Add or change the backup directory (administrator)

Administrator-level users can use a Network File System (NFS) to set up an external storage device that is a backup server to store system data. When the current backup directory has limited free disk space remaining, change the backup directory used by the system.

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, select either **Network** or **Cloud** as the connection type.

3. Edit the backup settings.

| Option | Description |
|---|---|
| Add a new mount point | a. In the **Backup Disk Space** section, click **Add Mount**.<br>b. In the **Add Mount** dialog box:<br>• Enter the name of the mount directory. For example, *ddn*.<br>Note: Do not include information from a hostname in the mount directory name. For example, if the host name is *gnx001*, do not use *gnx001* in the mount directory name.<br>You can type a new name to create a new directory.<br>• Enter the storage source. For example, *ddn.itw:/gs1/ion-data/dx-data*<br>c. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is restarted.<br>When the option is unselected, you must mount a directory after a restart. |
| Change the existing backup directory | In the **Backup Disk Space** section, select the directory that you want to use for backups from the **Backup Directory** dropdown menu.<br>You must add a mount point before it can be selected in the dropdown.<br>  |
| Remove a backup directory that is not in use, for example a directory that is full or is no longer available | a. Click **Remove Mount**.<br>b. In the **Remove Mount** dialog box, select the mounted drive you want to remove.<br>c. Click **Remove**.<br>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory. |

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

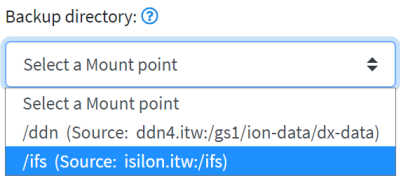
## Add or change the cloud backup directory (administrator)

You can change the cloud backup directory that is used by the system. For example, you might change the directory if the current backup directory has limited storage space. You can also add a new directory, or remove a directory.

1. In the menu bar, click **(Settings)►Backup & Restore**.
   The **Backup & Restore** screen opens.

2. Under **Backup Disk Space** select **Cloud** as the connection type.

3. Edit the backup settings.

| Option | Description |
|---|---|
| Add a new mount point | **1.** In the Backup Disk Space section, click Add Mount.<br>**2.** In the Add Mount dialog box:<br>• Enter the name of the Mount Point directory. For example, *ddn*. You can type a new name to create a new directory.<br>• Enter the Cloud Provider. For example, *AWS-S3*.<br>**3.** Enter the Configuration information.<br>• Nick Name—A name to uniquely identify the directory and mount point on the directory. Do not use spaces or special characters in the name.<br>Note: The following information must be entered correctly to ensure that the mount works correctly. Information should be provided by an administrator for the network or Amazon™ Web Services (AWS).<br>• Bucket Name—The name of the AWS S3 bucket.<br>• Access Key Id—A key identifier that, similar to a username, provides programmatic and CLI access to AWS resource, such as the S3 bucket<br>• Secret Access Key—An access key that, similar to a password, provides programmatic and CLI access to AWS resource, such as the S3 bucket.<br>• Region—The AWS region in which for the S3 bucket resides. For example, *us-east-1*, *us-west-1*.<br>**4.** *(Optional)* Select Automount to allow the system to mount backup directories automatically when the system is rebooted.<br>When the option is unselected, you must mount a directory after a reboot.<br>**5.** Click Save. |
| Change the existing backup directory | In the Backup Disk Space section, select the directory that will be used for backups from the Backup directory drop-down list.<br>You must add a mount point before for the backup directory can be selected.<br>  |
| Remove a backup directory that is not in use, for example a directory that is full or is no longer available | **1.** Click Remove Mount.<br>**2.** In the Remove Mount dialog box, select the mounted drive you want to remove.<br>**3.** Click Remove.<br>**4.** Click Save.<br>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory. |

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

The changes that you made are shown in the **Backup Disk Space** section of the **Backup Settings** screen.

## Set up a backup server (administrator)

Administrator-level users can set up an external storage device to be a backup server to store system data with this procedure. Alternatively, administrator-level users can directly set up a backup server in Genexus™ Dx Software. For more information, see “Add or change the backup directory (administrator)” on page 171.

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IMPORTANT! We recommend Network File System (NFS) version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

---

1. Mount the NFS to the sequencer.

   The following example is for users who have an existing NFS with read and write access. Mount the NFS to the sequencer using the *fstab* file so that the mount point is persistent (NFS mounts automatically after rebooting).

   **Example:**
   For NFS share: `cqtsop4:/md1200`
   Mount point: `nfs1`

2. Create a mount point to mount the external device on nfs share.
   a. Create the directory /nfs1 using the following command:

      ```
      sudo mkdir /nfs1
      ```

   b. Append the mount point entry in /etc/fstab.

      Command example:

      ```
      sudo vi /etc/fstab
      cqtsop4:/md1200 /nfs1 nfs defaults 0 0
      ```

   c. Save, then exit by pressing **Esc** on your keyboard, then type *:wq*

3. Create a permanent or temporary mount on nfs share:
   - Permanent mount command: `sudo mount -a`
   - Temporary mount command (example): `sudo mount cqtsop4:/md1200 /nfs1`

4. Verify that the NFS share is mounted using the df -kh command.

   ```
   ionadmin@eternity1:~$ df -kh
   Filesystem Size Used Avail Use% Mounted on
   /dev/sda9 253G 5.9G 235G 3% /
   none 4.0K 0 4.0K 0% /sys/fs/cgroup
   udev 63G 4.0K 63G 1% /dev
   tmpfs 13G 1.3M 13G 1% /run
   none 5.0M 0 5.0M 0% /run/lock
   none 63G 0 63G 0% /run/shm
   none 100M 0 100M 0% /run/user
   ```

```
/dev/sda5 19G 2.9G 15G 17% /var
/dev/sda1 938M 37M 854M 5% /boot
/dev/sda6 9.1G 1.7G 7.0G 20% /home
/dev/mapper/raid-serverdata 8.4T 4.3T 4.1T 52% /serverdata
/dev/mapper/raid-servershare 1.0T 9.0G 1015G 1% /servershare
/dev/mapper/raid-scratch 1.0T 838M 1023G 1% /scratch
/dev/sda7 92G 61M 87G 1% /tmp
pluto:/md1200 37T 1.1T 36T 3% /nfs2
penguin-ts1:/local 11T 9.3T 978G 91% /nfs
ddn4.itw:/gs1/ion-data/dx-data 1.1P 1001T 115T 90% /ddn
cqtsop4:/md1200 37T 37T 77G 100% /nfs1
```

The mounted directory is listed in the **Backup Directory** dropdown list in the **Backup & Restore** screen.

For more information, see **help.ubuntu.com/lts/serverguide/network-file-system-configuration**.

## External storage devices

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

## Change the backup settings (administrator)

A backup directory must be set up before an administrator-level user can change the backup settings. For more information, see “Disk space usage and backup settings (administrator)” on page 167.

The following types of data are backed up daily at midnight and cannot be changed:

- **System Data**–Includes the system database, reference files, assay definition files, annotation content, and system-installed panel files, hotspot files, fusion reference files, and CNV baseline files.
- **Analysis Results**–The final results generated after sequencing and analysis. Analysis results include run details, all BAM files, filtered results, graphs, run reports, metrics, annotated variant data, and VCF files.

IMPORTANT! The first time you back up the system, it takes a considerable amount of time. The amount of time required for system backup depends on the number of runs that require backup and the amount of disk space used. For more information, see “Specifications and disk space usage” on page 169.

A system restart also takes time. When the system restarts, it resumes the backup where it left off.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, in the **Backup Settings** tab, edit the backup settings.

| Option | Procedure |
|---|---|
| Back up .wells files | Select the Sequencing Output checkbox.<br>Sequencing output backup takes time. |
| Back up intermediate files, such as intermediate BAM files and log files | Select the Intermediate Files checkbox. |
| Change the frequency or timing of the .wells and intermediate files backup | Select Daily or Weekly, then select the time and day, if applicable, for the backup.<br>Intermediate file backup takes time. Consider scheduling the backup of intermediate file on a weekend or at a time when the system is not in use. |

3. Click **Save**.

## View and export the audit trail for backup settings (administrator)

An administrator-level user can view the audit trail for backup settings, disk cleanup settings, and email notification settings in Genexus™ Dx Software.

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, view the audit trail.

| Option | Procedure |
|---|---|
| View the audit trail for the backup disk space | In the Backup Disk Space section, click audit to review changes made to the backup directory. |
| View the audit trail for the backup settings | In the Backup Settings section, click audit to review changes made to the backup settings. |
| View the audit trail for email notification settings | In the Email Notification Settings section, click audit to review changes made to the email notification settings. |
| View the audit trail for disk cleanup settings | In the Disk Cleanup Settings section, click audit to review changes made to the disk cleanup settings. |

3. In the **Audit Trail - Archive Settings** screen:

| Option | Procedure |
|---|---|
| Export the record to a PDF file | Click Download. |
| Return to the Backup Settings tab. | Click Back. |

## Set a schedule to permanently delete files (administrator)

Results files and `.wells` files, and intermediate files that have been backed up can be scheduled to be deleted by an administrator-level user in Genexus™ Dx Software.

Intermediate and .wells files include intermediate BAM files, log files, and Sequencing output files. These files are not backed up by default. When you set a schedule to delete these files, by default, the files are deleted when they are not backed up and the system disk space falls below 4 terabytes (TB). You can change the system disk space threshold to delete these files.

Run results that are retained are never deleted in the software, even when you set a schedule to permanently delete run results in the software. For more information, see “Retain run results” on page 114.

---

IMPORTANT!

- By default, when the **Disk Cleanup Settings** are enabled, the software is set to delete results 12 months after the run was last updated and backed up. You can change this setting.
- Intermediate and .wells files are not backed up unless an administrator-level user sets a schedule to back up these files. By default, these files are auto-deleted when the free disk space threshold is met, even when these files are not backed up. If you select to keep the default auto-delete setting for intermediate and .wells files and the files are deleted, the impact is minimal.

  You cannot perform the following actions when these files are deleted.

  - Reanalyze a run from basecalling.

---

1. In the menu bar, click ⚙ **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, in the **Disk Cleanup Settings** section:

| Option | Procedure |
|---|---|
| Do not auto-delete analysis results at any time. | In Delete Analysis Results, click No. |
| Change the schedule to permanently delete analysis results. | Analysis results are deleted only if the results have been backed up.<br>**1.** In Delete Analysis Results, click Yes.<br>**2.** In months after completion, set the schedule to delete the files:<br>• 3 months<br>• 6 months<br>• 9 months<br>• 12 months<br>• 24 months – default setting<br>• 36 months |
| Do not auto-delete .wells files and intermediate files at any time. | In Delete additional files, click No. |
| Change the schedule to permanently delete .wells files and intermediate files. | In Delete additional files, click Yes, then select the threshold of free disk space on the instrument, from 1 to 10 TB, that triggers the system to delete these files.<br>The system automatically deletes .wells and intermediate files when the disk space falls below the selected threshold.<br>If you have enabled the backup of intermediate files and .wells files, these additional files must be backed up before they can be deleted. |

3. Click **Save**.

## Archive a run result (administrator)

In addition to system cleanup, in which all backed up runs that are older than a specified length of time are archived, an administrator-level user can archive one to five run results at any time. Archived run results are copied to the external storage device or to a cloud system that you have set up. Then the archived run results are removed from the sequencer.

When you archive a run result, the analysis results files are archived. In addition, you can archive the intermediate and 1.wells files, if needed. You can archive run results that are no longer needed to ensure that there is sufficient disk space on the sequencer.

If you archive a run result at the time of a scheduled system archive, the system cleanup is skipped for that instance.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, place the pointer over one or more run result that you want to archive, then click **Archive Data**.
3. In the **Confirm archive data for selected result(s)** dialog box, select **Sequencing Output** and **Intermediate files** to include 1.wells and intermediate files, if needed.
4. Click **Archive**.

Archived runs results are listed in the **Run Results** screen with the archived symbol ( ) after the run name. An administrator-level user can restore the run result to access the data.

## Delete a run result (administrator)

An administrator-level user can delete a run result in Genexus™ Dx Software. When you delete a run result, files for the run result are deleted from the sequencer.

---

IMPORTANT! This action cannot be reversed if you proceed. You can delete a run result even if it is not backed up. Ensure that you do not need the run result before you delete the run result from the sequencer and the software if the run result has not been backed up.

---

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to delete.
3. In the **Confirm delete data for selected result(s)** dialog box, confirm that the data are backed up, if needed, and select the files to delete.
   - **Sequencing Output**
   - **Intermediate Files**
   - **Analysis Results**

   If you delete the analysis results files, the sequencing output and intermediate files are also deleted.
4. Click **Delete**.
5. Click **Yes** to confirm the deletion.

The files for the run result that you select are deleted from the sequencer and are no longer available in the software. If the run results are backed up before the deletion, an administrator-level user can restore the run result to access the data.

You can view the audit trail for a run result that you have deleted in the **Settings / Audit Records** screen. Deleted run results are listed as **Planned Runs**. For more information, see “Search audit records (administrator)” on page 158.

## Data output directory

The data output directory is the primary location on the hard drive where data generated by the Genexus™ Dx Integrated Sequencer is stored.

The directory location is locked and cannot be changed. Users should configure their laboratory information management systems (LIMS) to access this folder location to receive sequencing output files.

After a sequencing run and data analysis are complete, the software creates a separate results folder in the data output directory for each sample, and uses the following naming conventions:

`AssayDev_<Sample Name>_<Assay Name>_<RunPlanShortCode>_<RunPlanShortCode>`

## Email notifications

Administrator-level users receive email notifications when there is a restore run failure. Administrator-level users can also receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. To review or update these settings, see “Change email notification settings (administrator)” on page 180.

Before administrator-level users can receive email notifications, an administrator or manager must first set up notifications for valid Genexus™ Dx Software user accounts. For more information, see “Configure email notifications (manager/administrator)” on page 163.

### Change email notification settings (administrator)

Administrator-level users can select when to receive email notifications, such as when the system has limited disk space remaining. Administrator-level users receive an email message when conditions occur according to the selections in the email notification settings.

1. In the menu bar, click ⚙ **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, in the **Email Notifications Settings** section, select or deselect the checkbox next to one or more of the following options. Administrator-level users receive email notifications for options that are selected. They do not receive email messages for options that are deselected.
   - Backup fails or has insufficient storage space.
   - Backup is successfully completed.
   - Genexus™ Dx Integrated Sequencer has insufficient system disk space. Select the amount of sequencer disk space that must be available in the dropdown list.
   - Backup fails or has insufficient disk space. Select the amount of backup disk space that must be available in the dropdown list.
3. Click **Save**.

Changes to the email notifications take effect immediately.

# Restore run results (administrator)

An administrator-level user can configure Genexus™ Dx Software to automatically back up older run data, results files, and signed reports to external data storage or a cloud storage space. For more information, see “Disk space usage and backup settings (administrator)” on page 167 and “Add or change the backup directory (administrator)” on page 171. In addition, an administrator-level user can set a schedule to automatically delete backed up runs after a specified amount of time. For more information, see “Set a schedule to permanently delete files (administrator)” on page 177. Runs, for which analysis results have been backed up and deleted in the software, are listed in the **Run Results** screen and are denoted with the archived symbol ( ) after the run name.

An administrator-level user can later restore the run results, files, and reports in the software. There are two ways to restore run results in Genexus™ Dx Software.

- For information about how to restore run results for a single run, see “Restore run results for a single run (administrator)” on page 181.
- For information about how to restore or import multiple run results, see “Restore or import multiple archived run results (administrator)” on page 181.

## Restore run results for a single run (administrator)

An administrator-level user can restore run results, files, and reports that have been archived in Genexus™ Dx Software.

Archived runs that can be restored are listed in the **Results / Run Results** screen with the **Restore** link active when you place the pointer over the row of run. Runs, for which analysis results have been backed up and deleted (archived) from the system, are listed in the **Run Results** screen and are denoted with the archived symbol ( ) after the run name.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, click the run name of interest.
3. In ••• **(More Options)**, click **Restore**.
4. Click **OK** in the confirmation dialog box.
5. Click the run name in the **Run Results** screen to view run results.

## Restore or import multiple archived run results (administrator)

An administrator-level user can:

- Restore run results, files, and reports that have been archived.
- Import run results, files and reports from external data storage or another Genexus™ Dx Integrated Sequencer.

---

Note: Do not attempt to import verification runs because they are specific to a single system. After a run is imported, you can access **Run Results** and **Sample Results** in the target system. Genexus™ Integrated Sequencer details are also listed with the instruments in the target system.

---

Results that have been backed up, then deleted in the software, are listed in the **Results / Run Results** screen with (archived) after the run name.

Before you can import a run from external data storage or another Genexus™ Dx Integrated Sequencer:

- The run data must be archived to external data storage, and the same external data storage must be connected to the Genexus™ Dx Integrated Sequencer to which you will import the run.
- The samples for the run or runs that you want to import must have been created in the software with identical names. For more information about how to import a sample from another system, see “Import samples” on page 29.
- The assay for the run or runs that you want to import must exist in the software. The name of the assay, the barcode kit, and the extraction method type, must be identical between the source system and the target system. For information about how to import an assay, see “Import an assay (manager/administrator)” on page 148.

You can search the list of runs that can be restored by the run name or a sample tag name. However, if you restore a run that has an assigned sample tag name that has been deleted in the software, the tag is not shown in the software when you restore the run. In this case, you can reassign the tag if needed. For more information, see “Sample tags” on page 33.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab and select from the following options.

| Option | Description |
|---|---|
| Restore archived run results | **1.** In the type of runs dropdown menu, keep the selection of Archived runs.<br>**2.** In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>**3.** In the Select backup folder to list runs dialog box, navigate to the folder with the name of the system that you use, then select the subfolder that contains the runs you want to restore and click Select.<br>**4.** In the Restore Runs tab, select the checkbox in the row for each run that you want to restore, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>**5.** *(Optional)* After the archived run is restored, click Check Log to view if the process was successful or if there are problems. |

*(continued)*

| Option | Description |
|---|---|
| Import run results from another system | 1. In the type of runs dropdown menu, keep the selection of Archived runs.<br>2. In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>3. In the Select backup folder to list runs dialog box, navigate to the folder for a different system, then click Select.<br>4. In the Restore Runs tab, select the checkbox in the row for each run that you want to import, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>5. *(Optional)* After the archived run is imported, click Check Log to view if the process was successful or if there are any problems, such as if a sample has not been created in the software. |
| Restore runs if there is a system crash | Use this procedure only if there is a system crash and you have worked with a Field Service Engineer (FSE) to restore the data onto a new system. When you select All runs in any other instance, the software overwrites existing data with identical data.<br>1. In the type of runs dropdown menu, select All runs.<br>2. In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>3. In the Select backup folder to list runs dialog box, navigate to the folder that contains the runs that you want to restore, then click Select.<br>4. In the Restore Runs tab, select the checkbox in the row for each run that you want to restore, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>5. *(Optional)* After the archived run is restored, click Check Log to view if the process was successful or if there are problems. |

# Log files (administrator)

Administrator-level users use the **Logs** screen to view log files generated by the software and connected sequencing instruments. Viewing log files is useful for troubleshooting activities.

Administrator-level users can also use the **Logs** screen to manage the storage of log data by setting the retention period for the audit trail function and enabling auto-deletion rules for logs.

## View log files (administrator)

Administrator-level users can view software and instrument log files in Genexus™ Dx Software.

1. Click ⚙ **(Settings)**►**Logs**.
2. In the **Logs** screen, select an option in the software or connected instrument dropdown menu.

| Option | Description |
|---|---|
| View software log files. | Click **Ion Torrent Genexus Software**. |
| View log files for a connected sequencing instrument. | Click **Instrument**. |

3. Click the log file name of interest.

| File name | Description |
|---|---|
| `ApplicationLog_<date>.log` | A log file with information related to some actions on the server that is used for Genexus™ Dx Software, including user sign-ins and sign-outs and failed attempts to sign-in or sign-out. |
| `InstrumentGX5_date.log` | A log of activity on the sequencer instrument. |

The log file opens in a new tab in the browser.

## Manage log settings (administrator)

An administrator-level user manages the length of time that log files are kept and configures the software to allow log files to be automatically deleted.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click ⚙ **(Settings)**►**Logs**.
2. In the **Logs** screen, click **Manage Logs**.
3. In the **Manage Logs** dialog box, in **Retention Period**, select the number of months that logs are to be retained on the sequencer.
4. Select **Enable Auto Deletion** to delete log files automatically after the designated retention period.
5. Click **Save**.

## View the log settings history

Administrator-level users can view the history of changes made to the settings for the retention of log files in Genexus™ Dx Software.

1. Click **(Settings)►Logs**.
2. In the **Logs** screen, click **Audit Trail**.
   The **Audit Trail - Log Settings** screen opens.
3. Click **Back** to return to the list of log files.

# View panel details

You can view details about system-installed and panels that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. View the panel details.

# View hotspot file details

You can view details about system-installed hotspot files and hotspot files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. In the **Hotspots** table, view the details for the hotspot files, including the panel with which the the file is associated, whether the file is **Locked**, if the file is **Active**, who created the file, and the date on which the file was created. You can also view the genomic coordinates and alleles that are included in the hotspot file.

# View fusion reference file details

You can view details for system-installed fusion reference files and fusion reference files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. In the **Fusion References** table, view the details for the fusion reference files.

# View reference genome file details

You can view details for system-installed reference genome files and reference genome files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. In the **References** table, view the details for the reference genome files.

# View barcode set details (manager/administrator)

You can view details for system-installed barcode sets and barcode sets that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.
3. In the **Barcode sets** table, view the list of barcode sets.
4. Click the name of a barcode set in the list to view the barcode IDs, sequences, and other details for the barcode set.
5. Click ✕ **(Remove)** to return to the complete list of barcode sets.

# Control fragments (manager/administrator)

Control fragment sequences are system-installed in Genexus™ Dx Software and are included in reagents. These internal controls are predefined and cannot be modified by users.

Manager- and administrator-level users can view control fragment information in the **Assays / Manage Panels** screen, in the **Control Fragments** tab.

| Column | Description |
|---|---|
| Name | The control fragment name. |
| Sequence | Single-letter nucleotide sequence of the control fragment. |
| Control | The process in which the control fragment is used. |
| Status | Indicates whether the control fragment information is locked and cannot be modified by the user. |

# System services (manager/administrator)

Manager- and administrator-level users can view information about system services using the **Settings / Services** screen. The information can be useful when troubleshooting error messages that are received during an active job or why a run failed.

| Service | Function |
|---|---|
| Jobs Server | Shows a list of server information and active (running) software. |
| Services | Shows the service name, description, and status. The Services list can be used for troubleshooting. If a service has stopped during a run, an option to start the service will be visible for manager- and administrator-level users.<br>For more information, see “Start a stopped service (manager/administrator)” on page 188. |
| Active Jobs | • Shows a list of runs in progress that are active and queued on the sequencer.<br>• The message “No records available” indicates that no jobs are currently active.<br>• Allows manager- and administrator-level users to stop a job.<br>For more information, see “Terminate an active job (manager/administrator)” on page 188. |
| ionCrawler Service Details | Provides information about the data transfer process from the Genexus™ Dx Integrated Sequencer. The information includes whether the service was started or stopped, which user completed the action, and a timestamp for the action. |
| Smart Monitoring Service Details | Allows remote monitoring of the Genexus™ Dx Integrated Sequencer. Allows manager- and administrator-level users to start or stop the service. For more information, see “Start a stopped service (manager/administrator)” on page 188. |

*(continued)*

| Service | Function |
|---|---|
| Download Instrument Diagnostics | Contains the initialization logs of the Genexus™ Dx Integrated Sequencer, and allows you to download the diagnostic archive file in ZIP format. |
| System Check Report | Manager and administrator-level users can click Run System Check to view the system check report. |

## Terminate an active job (manager/administrator)

If an active job is not going to be useful or if a different job is urgent, you can terminate an active job. A manager- or administrator-level user can stop an active job in the **Settings / Services** screen.

1. In the menu bar, click **(Settings)►Services**.
2. In the **Services** screen, in the **Active Jobs** section, find the active job that you want to stop, then click **Terminate**.
   A confirmation message opens, **Are you sure you want to terminate <job name> ?**.
3. Click **Terminate** to terminate the job.

## Start a stopped service (manager/administrator)

A manager- or administrator-level user can use the **Settings / Services** screen to restart a stopped service if a service has stopped running for some reason. The two database services, **ntp** and **deeplaser**, are the only services that can be restarted.

1. In the menu bar, click **(Settings)►Services**.
2. In the **Services** screen, in the **Services** section, find the service that you want to restart, then click **Start**.
   A confirmation message opens, **Are you sure you want to Start <service name>?**.
3. Click **Yes** to restart the service.

## Disable or enable the Smart Monitoring service (manager/administrator)

The Smart Monitoring service allows Thermo Fisher Scientific personnel to monitor the status of the Genexus™ Dx Integrated Sequencer remotely through an internet connection. Smart Monitoring employs multiple layers of security, including a Secure Sockets Layer (SSL) and Lightweight Directory Access Protocol (LDAP) authentication, to provide real-time troubleshooting and problem resolution for the Genexus™ Dx Integrated Sequencer.

The Smart Monitoring service is active by default and can be disabled by manager- or administrator-level users, if required.

- Disable the service.
  a. In the menu bar, click **(Settings)►Services**.

b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Stop**.

c. In the confirmation message, click **Yes** to disable the service.
The **Stop** option changes to **Start**.

- Enable the service.
  a. In the menu bar, click **(Settings)►Services**.

  b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Start**.

  c. In the confirmation message, click **Yes** to enable the service.
  The **Start** option changes to **Stop**.

# Sequencer decommissioning and data sanitization

Contact a Thermo Fisher Scientific Field Service Engineer for any request to decommission a Genexus™ Dx Integrated Sequencer. Decomissioning and data sanitization must be carried out by a Field Service Engineer.

# 10 Customer Support Archive

The Customer Support Archive is a downloadable archive that a technical support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Dx Software. The archive contains log files and other technical data about the software and other files from sequencing runs on a Genexus™ Dx Integrated Sequencer.

For more information, see “Download a Customer Support Archive” on page 222.

# A Genexus™ Dx Software web services API


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Genexus™ Dx Software has a web services API that allows you to automate the retrieval of key information from the system.

This section provides examples to show you how to leverage the API.

## LIMS introduction

Genexus™ Dx Software integrates with laboratory information management systems (LIMS) to help with sample, library batch, and run plan creation. The interaction is through the LIMS calling a set of RESTful APIs provided by the software (see the following table). The LIMS connects to the software by making an HTTP call to the URL of an API, and sending a user name and a password in the HTTP header. The user name and password must be for a registered active user account of the software. The LIMS passes the input information in JSON format within the HTTP body, while the response from the software is returned in JSON format as well. Users must configure their LIMS to create JSON files containing the required attributes for each LIMS request file.

Outlined in the following table are the API names, functions, and URLs of the five RESTful APIs that are provided.

| API | Function | API URL |
|---|---|---|
| Get report templates<br>For more information, see “Get report templates” on page 203. | Returns report templates. | `https://<host>/genexus/api/lims/v2/report-templates` |
| Get a list of assays<br>For more information, see “Get assay list” on page 195. | Returns a list of the assays in the system. PQ assays are excluded. | `https://<host>/genexus/api/lims/v2/assays` |
| Get a list of samples<br>For more information, see “Get samples” on page 196. | Returns a list of the samples in the system. | `https://<host>/genexus/api/lims/v2/samples` |
| Get a list of library batches<br>For more information, see “Get library batches” on page 196. | Returns a list of the library batches in the system. | `https://<host>/genexus/api/lims/v2/library-batches` |
| Get a list of run plans<br>For more information, see “Get run plans” on page 199. | Returns a list of available run plans in the system. | `https://<host>/genexus/api/lims/v2/plans` |
| Get results<br>For more information, see “Get results” on page 204. | Returns results of the run and samples in JSON file format. | `https://<host>/genexus/api/lims/v2/results` |
| Get a list of signed off samples<br>For more information, see “Get signed off samples” on page 209. | Returns a list of the samples that are designated with *(Signed Off)*. | `https://<host>/genexus/api/lims/v2/signedOffSamples` |
| Download files for completed runs<br>For more information, see “Download files for completed runs” on page 210. | Returns a compressed file in ZIP format that contains the requested files for the sample. | `https://<host>/genexus/api/lims/v2/download` |
| Create samples<br>For more information, see “Create samples” on page 210. | Create samples. | `https://<host>/genexus/api/lims/v2/samples` |
| Create a library batch<br>For more information, see “Create a library batch” on page 212. | Create a library batch from existing samples. | `https://<host>/genexus/api/lims/v2/library-batches` |

*(continued)*

| API | Function | API URL |
|---|---|---|
| Create a Nucleic Acid to Result run<br>For more information, see “Create a Nucleic Acid to Result run” on page 213. | Create a Nucleic Acid to Result run from one or more samples and an assay. | `https://<host>/genexus/api/lims/v2/plan-by-samples` |
| Create a library run<br>For more information, see “Create a library run” on page 213. | Create a library run from one or more library batches and an assay. | `https://<host>/genexus/api/lims/v2/plan-by-library` |

## LIMS system FTP setup or drive mapping

To enable the LIMS system to retrieve data analysis files from the sequencer, a manual FTP setup or drive mapping is required. This configuration is not provided as part of the sequencer installation, and must be set up as part of the LIMS system by the system integration or laboratory IT group. See the sections in this appendix for file paths and other setup information.

# API common parameters

If the JSON download process converts `@` symbols to `%40`, do not change the text. Instead, copy the JSON files as is into the browser.

| Parameter | Entry |
|---|---|
| BASE_URL | `https://<host>/genexus/api/lims/v2` |
| Request | `Authorization:Basic`<br>`Accept`:application/json |
| Response | `Content-Type`:application/json |

Response fields

| Code | Status |
|---|---|
| 200 | Successful response |
| 400 | Bad request<br>Error response:<br>`{ "errors":[ { "field":"validation error" } ] }` |
| 401 | Unauthorized |
| 500 | Internal Server Error |

Get requests

| Type of request | Entry |
|---|---|
| Get list of resources | `BASE_URL/resource_url/` |
| Get single object | `BASE_URL/resource_url?id=object_id` |

Common request parameters

| Parameter | Entry | Value |
|---|---|---|
| Starting page number | `pageNumber` | The default value is "1". |
| Number of items to fetch[1] | `pageSize` | The default value is "20". |
| A filed based on which, the result set needs to be ordered | `orderBy` | Only specific values are allowed. If no value is provided, the result set is ordered by ID. |
| Order direction of the result set | `sortOrder` | Valid values are "asc" or "desc".<br>The default value is "asc". |

[1] You can limit the number of items that are fetched when you add parameters to the request. For example, add "status" to the get assay list request.

# Get assay list

| assays | |
|---|---|
| URL | `BASE_URL/assays` |
| Description | Returns a list of the assays in the system. PQ assays are excluded. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/assays` |
| Supported parameters | • `assayName`<br>String. *Optional*.<br>Assay name.<br>• `status`<br>String, *Optional*.<br>Valid values are "draft", "obsolete", or "locked". |
| Example response | {<br>"total": 1,<br>"objects": [<br>{<br>"id": 18,<br>"assayName": "Generic_DNA_and_Fusions_EU_v0.3.0",<br>"assayShortName": "Generic DNA-Fus EU v0.3.0",<br>"assayDisplayedName": "Generic - DNA and Fusions - EU v0.3.0",<br>"application": "DNA+RNA",<br>"createdBy": "System Installed",<br>"createdDate": "2021-07-14T00:05:12.000+0000",<br>"isReanalysisAssay": false,<br>"genomeReference": "hg19",<br>"targetRegions": "Ronin_w2.7.0_v0.3.0_dnafusion_FFPE_DNA_v1.20210604.designed.bed",<br>"hotspots": "Ronin_w2.7.0_dnafusion_FFPE_DNA.v1.20210608.hotspots_0.3.0.bed",<br>"fusionReference": "Ronin_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.fasta",<br>"fusionTargetRegions":<br>"Ronin_w2.7.0_dnafusion_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.bed",<br>"chipType": "Genexus™ Dx GX5™ Chip",<br>"panelKit": "Generic Assay Panel (IVD)",<br>"controlKit": "",<br>"extractionKit": "",<br>"libraryKit": "Genexus™ Dx Library Strips 1 and 2-HD (IVD)",<br>"templatingKit": "Genexus™ Dx Templating Strips 3-GX5™ and 4 (IVD)",<br>"barcodeKit": "Genexus™ Dx Barcodes 1-32 HD (IVD)",<br>"sequencingKit": "Genexus™ Dx Sequencing Kit (IVD)",<br>"flows": 500,<br>"templatingSize": 200,<br>"status": "Locked",<br>"tumorCellularity": "1",<br>"reanalysisAssay": false<br>}<br>]<br>} |

# Get samples

| samples | |
|---|---|
| URL | `BASE_URL/samples` |
| Description | Returns a list of the samples in the system. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/samples` |
| Request query parameters | • `sampleId`<br>String. *Optional*.<br>Sample name. |
| Example response | `{ "total": 1, "objects": [ { "otherAttributes": { "Patient Id": "MRND685", "Date of Birth": "2000-01-01", "Ordering Physician": "Dr Fred" }, "id": 163, "sampleId": "Test_Sample1_224", "collectionDate": "2018-10-02", "gender": "Male", "sampleType": "DNA", "tumorCellularity": "70", "origin": "Non-Small Cell Lung Cancer", "cancerStage": "Stage 0", "diseaseCategory": "Cancer", "tags": [ "LIMS" ] } ] }` |

# Get library batches

| library-batches | |
|---|---|
| URL | `BASE_URL/library-batches` |
| Description | Returns a list of the library batches in the system. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/library-batches` |

*(continued)*

| library-batches | |
|---|---|
| Request query parameters | • `id`<br>String. *Required*.<br>Library batch ID. |

*(continued)*

| library-batches | |
|---|---|
| Example response | {<br>"total": 1,<br>"objects": [<br>{<br>"libraries": [<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_05",<br>"libraryName": "NTC_DNAST_LibBatch_DNA_RNA_3",<br>"inputQuantity": 87.0,<br>"sampleId": "NTC",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165751724060110123456724 0A51339",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_06",<br>"libraryName": "NTC_RNAST_LibBatch_DNA_RNA_3",<br>"inputQuantity": 55.3,<br>"sampleId": "NTC",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165751724060110123456724 0A51339",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_03",<br>"libraryName": "PositiveControl01_DNAST_LibBatch_DNA_RNA_1",<br>"inputQuantity": 87.0,<br>"sampleId": "PositiveControl01",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165441725093010260745524 0A51337",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_04",<br>"libraryName": "PositiveControl01_RNAST_LibBatch_DNA_RNA_1",<br>"inputQuantity": 55.3,<br>"sampleId": "PositiveControl01",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165441725093010260745524 0A51338",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_01",<br>"libraryName": "LIMS_Sample_55_DNAST_LibBatch_DNA_RNA_2",<br>"inputQuantity": 57.0,<br>"sampleId": "LIMS_Sample_55",<br>"tumorCellularity": null,<br>"controlKitBarcode": null,<br>"extractionKitBarcode": "91100090559101234567172110 19"<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_02",<br>"libraryName": "LIMS_Sample_55_RNAST_LibBatch_DNA_RNA_2",<br>"inputQuantity": 51.3,<br>"sampleId": "LIMS_Sample_55",<br>"tumorCellularity": null,<br>"controlKitBarcode": null,<br>"extractionKitBarcode": "91100090560101234567172112 31"<br>}<br>],<br>"id": 13,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",<br>"libraryBatchId": "ST_LibBatch_DNA_RNA",<br>"libraryKitBarcode": "",<br>"panelKitBarcode": "",<br>"libraryPrepType": "DNA+RNA",<br>"ampliSeqHd": false<br>}<br>]<br>} |

# Get run plans

| plans | |
|---|---|
| URL | `BASE_URL/plans` |
| Description | Returns a list of available run plans in the system. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/plans` |
| Request query parameters | • `planName`<br>String. *Optional*.<br>Name of the run plan.<br>• `viewType`<br>String. *Optional*.<br>Valid values are "all" or "toBeProcessed". |
| Example response | {<br>"objects": [<br>{<br>"id": 1,<br>"planName": "EK_Generic_FFPE_EU_Ronin_D5_0.3.0_6.5.26",<br>"planShortId": "EQ55D",<br>"type": "sample",<br>"createdBy": "System Installed",<br>"updatedOn": "2021-07-17T20:33:48.000+0000",<br>"noOfSamples": 8,<br>"noOfLibraries": 16,<br>"samples": [<br>{<br>"id": 8,<br>"sampleId": "NTC",<br>"assayName": "Generic - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_28"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_32"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 5,<br>"sampleId": "SRTestSample64",<br>"assayName": "Generic - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_25"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_29"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}, |

*(continued)*

| plans | |
|---|---|
| Example response, continued | {<br>"id": 6,<br>"sampleId": "SRTestSample65",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_26"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_30"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 2,<br>"sampleId": "SRTestSample61",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_18"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_22"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 7,<br>"sampleId": "SRTestSample66",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_27"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_31"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}, {<br>"id": 3,<br>"sampleId": "SRTestSample62",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_19"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_23"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 4,<br>"sampleId": "SRTestSample63",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_20" |

*(continued)*

| plans | |
|---|---|
| Example response, continued | ]<br>"rnaBarcodes": [<br>"IonDxHDdual_24"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}<br>],<br>"runAssays": [<br>"Generic Assay - DNA and Fusions - EU v0.3.0"<br>]<br>},<br>{<br>"id": 9,<br>"planName": "Re_Ali_EK_ODxET_FFPE_EU_Ronin_D5_0.3.0_6.5.26",<br>"planShortId": "CBLG5",<br>"type": "sample",<br>"createdBy": "System Installed",<br>"updatedOn": "2021-07-15T04:16:31.000+0000",<br>"noOfSamples": 8,<br>"noOfLibraries": 16,<br>"samples": [<br>{<br>"id": 49,<br>"sampleId": "PositiveControl01",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_17"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_21"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 52,<br>"sampleId": "SRTestSample63",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_20"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_24"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}, {<br>"id": 55,<br>"sampleId": "SRTestSample66",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_27"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_31"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>} |

*(continued)*

| plans | |
|---|---|
| Example response, continued | {<br>"id": 50,<br>"sampleId": "SRTestSample61",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_18"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_22"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 53,<br>"sampleId": "SRTestSample64",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_25"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_29"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 54,<br>"sampleId": "SRTestSample65",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_26"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_30"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 51,<br>"sampleId": "SRTestSample62",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_19"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_23"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 56,<br>"sampleId": "NTC",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_28"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_32"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}<br>],<br>"runAssays": [<br>"Generic Assay - DNA and Fusions - EU v0.3.0"<br>]<br>}<br>],<br>"total": 2<br>} |

# Get report templates

| report-templates | |
|---|---|
| URL for a list of resources | `BASE_URL/report-templates` |
| URL for a single object | `BASE_URL/report-templates` |
| Description | Returns report templates. API supports multiple filters. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/report-templates` |
| Request query parameters | `assayName`<br>String. *Optional*.<br>Get the report template for the given assay.<br>Use the Assay Name. Do not use the Assay Full Name. |
| Example response | {<br>"mode": "dx",<br>"reportingTemplate": [<br>{<br>"name": "LAB_ODxET_DNA_QC_6.8_v1.0.0",<br>"id": 22,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - DNA QC v1.0.0",<br>"assayId": 41,<br>"description": "ODxET DNA QC v1.0.0 - Lab Report",<br>"locale": "en"<br>},<br>{<br>"name": "ev_template",<br>"id": 21,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",<br>"assayId": 40,<br>"description": "EV_Template",<br>"locale": "en"<br>}<br>],<br>"total": 2<br>} |

# Get results

| results | |
|---|---|
| URL | `BASE_URL/results` |
| Description | Returns results of the run and samples in JSON file format. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/results` |

*(continued)*

| results | |
|---|---|
| Request query parameters | • `id`<br>String. *Optional*.<br>Results for the selected result id.<br>Example: *123*<br>• `assay_id`<br>String. *Optional*.<br>Results for the assay id.<br>Example: *123*<br>• `plan_id`<br>String. *Optional*.<br>Results for selected run plan id.<br>Example: *123*<br>• `sampleId`<br>String. *Optional*.<br>Results for the selected sample name.<br>Example: *MySample*<br>• `planName`<br>String. *Optional*.<br>Results for selected run plan name.<br>Example: *MyPlan*<br>• `analysisComplete`<br>String. *Optional*.<br>Results for completed runs. Results included for failed and successfully completed runs.<br>Example: *true*<br>• `updatedBefore`<br>String. *Optional*.<br>Returns results updated before the selected date. Use date format: `yyyy-MM-dd HH:MM`.<br>Example: *2019-01-15 10:30*<br>• `updatedAfter`<br>String. *Optional*.<br>Returns results updated after the selected date. Use date format: `yyyy-MM-dd`<br>Example: *2019-01-15*<br>• `orderBy`<br>String. *Optional*.<br>Parameter to sort results by selected field. The default sort is by `updatedBefore` or `updatedAfter`. Results are listed in descending order, with the most recent results listed first. |

| Example response |
|---|
| Output is as follows. |

```
{
    "objects": [
        {
            "id": 328,
            "planName":  "EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-T2.0.0-2",
            "sampleId": "SRTestSample61",
            "assay": {
                "id": 18,
                "value": "Generic Assay - DNA and Fusions - EU v0.3.0",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "planRun": {
                "id": 59,
                "value":  "EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-T2.0.0-2",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "sample": {
                "id": 45,
                "value": "SRTestSample61",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "planShortCode": "YYRU2",
            "resultName":
"Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_328",
            "libraryNames": "SRTestSample61_RNA831, SRTestSample61_DNA831",
            "runStartDate": "2021-07-23T00:00-0400",
            "runCompleteDate": "2021-07-24T00:45-0400",
            "updatedDate": "2021-07-24T00:44-0400",
            "libraryprepId": 428,
            "analysisComplete": true,
            "analysisVersion": "6.5.26",
            "isThumbnail": false,
            "isArchived": false,
            "isReanalysis": false,
            "runStatus": {
                "id": null,
                "name": "Analysis",
                "value": "Completed"
            },
            "qcStatus": "Passed",
            "instrumentName": "ValkyrieTestRig",
            "outputsLink": "/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/outputs",
            "outputsDir": "/serverdata/dxdata/IR/data/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/outputs",
            "loadingMetrics": [
                {
```

```
                "name": "Total Addressable Wells",
                "count": 64847855
            },
            {
                "name": "Wells With ISPs",
                "count": 60145720,
                "percentage": 92.75
            },
            ...
        ],
        "filteringMetrics": [
            {
                "name": "Filtered: Polyclonal",
                "count": 22931697,
                "percentage": 38.27
            },
            {
                "name": "Filtered: Primer-Dimer",
                "count": 9955,
                "percentage": 0.02
            },
            ...
        ],
        "runQCMetrics": [
            {
                "data": [
                    {
                        "metricName": "Key Signal",
                        "value": "75",
                        "referenceRange": "Not Set",
                        "qCStatus": "Not Calculated",
                        "percentValue": false,
                        "iscosmic": false,
                        "isPQControl": false,
                        "uniqueName": "RunQC_KeySignal",
                        "isDisplayMetric": false,
                        "isDisplayQc": false,
                        "libraryType": "NA",
                        "dataType": "INT",
                        "pqcontrol": false,
                        "displayMetric": false,
                        "displayQc": false
                    },
                    ...
                ],
                "sequence": 0,
                "localizationKey": "RunQC",
                "inlineControlQc": false,
                "customApp": "",
                "images": []
            }
        ],
        "purificationQCMetrics": [...],
        "controlQCMetrics": [...],
        "sampleQCMetrics": [...],
        "sampleInlineControlQCMetrics": [],
        "ntcQCMetrics": [],
        "plugins": [
            {
                "results": {
                    "Library Type": "AmpliSeq HD - DNA and Fusions
```

```
(Single Library)",
                        "Min Mapping Quality": "0",
                        "Percent end-to-end reads": "91.62%",
                        "Use Only Non-duplicate Reads": "No",
                        "Sample Tracking": "No",
                        "barcodes": {
                            "IonDxHDdual_18": {
                                "Alignments":
"IonDxHDdual_18_EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-
T2.0.0-2_Auto_R_2021_05_19_14_48_13_ion",
                                "Amplicon base composition bias": "1.141",
                                "Amplicons reading end-to-end": "98.80%",
                                "Amplicons with at least 1 read": "100.00%",
                        ...
                            },
                            ...
                        },
                        "Amplicons reading end-to-end": "86.40%",
                        "Min Aligned Length": "0",
                        "Targeted Regions": "Barcode specific",
                        "Tier 2 Coverage Depth": "350",
                        "Reference Genome": "Barcode specific",
                        "Use Only Uniquely Mapped Reads": "No",
                        "Target Padding": "0",
                        "Launch Mode": "Autostart with default
configuration",
                        "barcoded": "true",
                        "Tier 3 Coverage Depth": "500",
                        "Tier 1 Coverage Depth": "100"
                    },
                    "id": 455,
                    "pluginName": "coverageAnalysis",
                    "pluginVersion": "5.16.0.4",
                    "updatedOn": "2021-07-24T01:58:19.000+0000",
                    "status": "COMPLETED",
                    "path": "/serverdata/dxdata/IR/data/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/plugins/
coverageAnalysis_5.16.0.4/1627091942777",
                    "runType": "SAMPLE"
                },
                ...
            ],
            "biomarkers": [
                {
                    "Variant": "ALK fusion,   AR fusion",
                    "level": "Level 3"
                }
            ]
        }
    ],
    "total": 1
}
```

# Get signed off samples

| signedOffSamples | |
|---|---|
| URL | `BASE_URL/signedOffSamples` |
| Description | Returns a list of the samples that are designated with *(Signed Off)*. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/signedOffSamples` |
| Request query parameters | • `start_date`<br>String. *Optional*.<br>Provide the start date as yyyy-mm-dd.<br>• `end_date`<br>String, *Optional*.<br>Provide the end date as yyyy-mm-dd.<br>• `signedOff`<br>String, *Required*<br>A Boolean data type. Valid values are "true" or a "false".<br>The default value is "true". |
| Example response | `{ "meta": { "pageSize": 20, "next": null, "pageNumber": 0, "previous": null, "total_count": 76 }, "objects": [ { "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample1_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "92", "sample_name": "PQSample1", "planName": "IQ_Lib" } }, { "sample": { "assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0", "base_url": "AssayDev_PQSample2_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS", "checksum_file": "CHECKSUM", "created_date": "2021-06-23", "sample_file": "Info.csv", "sample_id": "93", "sample_name": "PQSample2", "planName": "IQ_Lib" } } ] }` |

# Download files for completed runs

| download | |
|---|---|
| URL | `BASE_URL/download` |
| Description | Returns a compressed file in ZIP format that contains the requested files for the sample. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/download` |
| Request query parameters | • `file_list`<br>String. *Required*.<br>Provide the file name in comma separated format.<br>• `path`<br>String, *Required*.<br>Provide the LIMS folder path of the analyzed sample. You can request the folder path when you get signed off samples. The folder path is listed as the "base_URL". For more information, see “Get signed off samples” on page 209. |
| Example request | 1. Perform a checksum function to query for the list of files for a sample that has been sequenced and analyzed.<br>`https://BASE_URL/download?file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q`<br>2. Parse the checksum to get the file name, then request that file.<br>`https://BASE_URL/download?file_list=Info.csv,BaseCaller.json,analysis.log&path=AssayDev_Sample_C1_qualification_ampliseq_ Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q`<br>3. Ensure that the name of the downloaded file matches the checksum requested in step 1. |

# Create samples

| samples | |
|---|---|
| URL | `BASE_URL/samples` |
| Description | Create 1 to *n* samples. "All" or "None" is followed for all samples in a request. If the creation of any one sample fails, creation of the entire batch fails. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |

*(continued)*

| samples | |
|---|---|
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/samples` |
| Attribute definitions | • `Patient Id`. String. *Required*. A unique identifier for the patient.<br>• `Date of Birth`. String. *Required*. The date of birth for the patient.<br>• `Ordering Physician`. String. *Required*. The name of the physician who created the order for the sample.<br>• `sampleId`. String. *Required*. Sample name.<br>• `sampleType`. String. *Required*. Examples of valid values are "DNA" or "RNA".<br>• `collectionDate`. String. *Required*. The date that the sample was collected.<br>• `gender`. String. *Required*. The biological sex of the sample: Female, Male, or Unknown.<br>• `diseaseCategory`. String. *Required*. The sample application category.<br>• `cancerStage`. String. *Required*. Cancer stage.<br>• `origin`. String. *Required*. Cancer type.<br>• `notes`. String. *Required*. An open-entry field for more sample information.<br>• `necrosis`. String. *Optional*. The percentage of cellular necrosis in the sample.<br>• `tumorCellularity`. String. *Optional*. The percentage of tumor cells over normal cells in the sample.<br>• `condition`. String. *Optional*. The condition of the sample. |
| Example request | {<br>"objects":[<br>{<br>"sampleId":"Sample_NSCLC_99",<br>"diseaseCategory":"Cancer",<br>"sampleType":"DNA & RNA",<br>"origin":"Non-Small Cell Lung Cancer",<br>"cancerStage":"Unknown",<br>"necrosis":100,<br>"tumorCellularity":31,<br>"gender":"Unknown",<br>"collectionDate":"2018-10-02",<br>"condition":"good",<br>"tags":["NSCLC"],<br>"notes":"LIMS API created Cancer sample and existing Tag NSCLC",<br>"otherAttributes":<br>{<br>"Patient Id":"MRND12121",<br>"Ordering Physician":"Dr Fred",<br>"Date of Birth": "2000-01-01"<br>}<br>}<br>]<br>} |

# Create a library batch

| library-batches | |
|---|---|
| URL | `BASE_URL/library-batches` |
| Description | Create a library batch from one or more existing samples. If any requested sample does not exist in the software, the creation of the entire library batch fails. |
| Method | `POST` |
| Example URL | `https://<host>/genexus/api/lims/v2/library-batches` |
| Example request | 
```
{
  "objects":[
    {
    "libraryBatchId": "LibBatch_DNA_RNA_99",
    "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
    "libraryPrepType": "DNA+RNA",
    "libraryKitBarcode": "01ABC00123000000017221123102-321AB234240A51153",
    "panelKitBarcode": "01ABC00123000000017221123102-321AB234240A51345",
    "libraries": [
      {
        "barcodeID": "IonDxHDdual_01",
        "inputQuantity": 57.0,
        "libraryType": "DNA",
        "libraryName": "DNA_Lib1_01",
        "sampleId": "Test_Sample_01",
        "extractionKitBarcode": "A51000"
      },
      {
        "barcodeID": "IonDxHDdual_02",
        "inputQuantity": 51.3,
        "libraryType": "RNA",
        "libraryName": "RNA_Lib1_01",
        "sampleId": "Test_Sample_01",
        "extractionKitBarcode": "A51000"
      },
      {
        "barcodeID": "IonDxHDdual_03",
        "inputQuantity": 87.0,
        "libraryType": "DNA",
        "libraryName": "",
        "sampleId": "PositiveControl01",
        "controlKitBarcode": "A51337"
      },
      {
        "barcodeID": "IonDxHDdual_04",
        "inputQuantity": 55.3,
        "libraryType": "RNA",
        "libraryName": "",
        "sampleId": "PositiveControl01",
        "controlKitBarcode": "A51338"
      },
      {
        "barcodeID": "IonDxHDdual_05",
        "inputQuantity": 87.0,
        "libraryType": "DNA",
        "libraryName": "",
        "sampleId": "NTC",
        "controlKitBarcode": "A51339"
      },
      {
        "barcodeID": "IonDxHDdual_06",
        "inputQuantity": 55.3,
        "libraryType": "RNA",
        "libraryName": "",
        "sampleId": "NTC",
        "controlKitBarcode": "A51339"
      }
    ]
  }
]
}
```
 |

# Create a Nucleic Acid to Result run

| plan-by-samples | |
|---|---|
| URL | `BASE_URL/plan-by-samples` |
| Description | Create a Nucleic Acid to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/plan-by-samples` |
| Example request | <pre>{<br>"reportTemplateName": "",<br>"samples":[<br>{<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"sampleId":"Test_Sample1"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample2"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample3"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample4"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample5"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample6"<br>}<br>],<br>"notes":"Nucleic Acid Run created by LIMS API",<br>"planName":"NucleicAcidRun_DNF_6Samples_Run"<br>}</pre> |

# Create a library run

| plan-by-library | |
|---|---|
| URL | `BASE_URL/plan-by-library` |
| Description | Create a library run from one or more library batches and an assay. The library batch or batches and the assay must exist in the system. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |

*(continued)*

| plan-by-library | |
|---|---|
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/plan-by-library` |
| Example request | `{ "reportTemplateName": "", "samples":[ { "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0", "libraryBatchId":"DNA_RNA_LibBatch_01" } ], "notes":"Library plan run created by LIMS API", "planName":"PlanByLib_DNF_1L_01" }` |

# B Reference information


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## Rules for variant names in Genexus™ Dx Software

Ion Torrent™ Genexus™ Dx Software provides standard naming conventions for genomic variants and other biomarkers. Standard variant names are derived from sample and results data and properties that are available in the software for each variant, such as variant or biomarker type, gene name, and amino acid change. The conventions are used for both de novo variants and variants that are included in a hotspots file. Variant names are shown in the **Variant Name** column of data results tables in Genexus™ Dx Software, and in reports generated by the software.

### Summary of variant types and examples of standardized variant names

Genexus™ Dx Software applies a set of rules to it a variant, then assigns or returns a name for the variant that best describes variant details. Rules that are used to name variants in the software are as follows.

| Variant or biomarker type | Description | Variant name examples |
|---|---|---|
| Variants with protein change | Variants that provide an amino acid change syntax. | BRAF V600E<br>BRAF p.(V600E) c.1799A>C |
| Variants with coding change but no protein change | Variants that provide a coding change, but for which an amino acid is missing or is `p.?`. | MET c.2942-29_2942-28delinsGG |
| Oncomine class-based variants | Any variant with a variant type of `SNV` or `INDEL` at the locus based on Oncomine™ annotations. These changes are typically complex and are frequently referenced in scientific literature. | EGFR exon 19 deletion<br>MET exon 14 skipping |

| | | |
|---|---|---|
| EGFR exon 19 insertion variants | Variant with `Function="nonframeshiftInsertion"` in exon 19 of EGFR gene. | EGFR exon 19 insertion |
| Copy number variants | Copy number gain or loss, signified by a variant type of `CNV`. | ALK amplification<br>PTEN deletion |
| RNA exon variants | Specific RNA exon variants, identified by variant ID. | MET exon 14 skipping<br>EGFRvIII |
| Long deletion Variants | Oncomine Variant Class equals `LongDel` and length attribute is a positive integer. | BRCA1 40 base deletion |
| Exon deletions | A special type of CNV that impacts BRCA genes only. These variants are identified by a type of `CNV`, a subtype of `BigDel`, and call details that specify the exon range affected. | BRCA1 exon 1 deletion |
| Exon duplications | A special type of CNV that impacts BRCA genes only. These variants are identified by a type of `CNV`, a subtype of `BigDup`, and call details that specify the exon range affected. | BRCA1 exon 1 duplication |
| Targeted fusions | Fusions detected by targeting a specific breakpoint. | SLC34A2-ROS1 fusion |
| Non-targeted fusions | Non-targeted fusions | SLC45A3-ETV1 fusion |
| Expression imbalance variants | Variants for which the Oncomine Variant Class is `"ExpressionImbalance";`. This is measured by the partner-agnostic detection of fusions in driver genes using imbalance in exonic expression. | MET fusion |
| All other mutations | Mutation variants that do not match the criteria of other variants in this table. | ADAMTSL4-AS1 mutation |
| Sample MSI | Sample-level microsatellite stability/instability. This biomarker type is a measure of the entire sample rather than an individual variant. | Microsatellite instability-High<br>Microsatellite stable |
| Expression control variants | Variants with `SVTYPE=ExprControl`. | LMNA expression control |
| Gene expression variants | Variants with `SVTYPE=GeneExpression`. | MET expression |
| Sample TMB | Sample-level tumor mutational burden. This biomarker type is a measure of the entire sample rather than an individual variant. | Tumor Mutational Burden |

| | | |
|---|---|---|
| Sample LOH | Sample-level loss of heterozygosity. This biomarker type is a measure of the entire sample rather than an individual variant. | Loss of Heterozygosity |
| Sample TAI | Sample-level telomeric allelic imbalance. This biomarker type is a measure of the entire sample rather than an individual variant. | Telomeric Allelic Imbalance |
| Sample LST | Sample-level large-scale state transition. This biomarker type is a measure of the entire sample rather than an individual variant. | Large-Scale State Transition |
| Gene-level LOH Variants | Oncomine Variant Class equals `"LOH"`. | BARD1 LOH |

# System-installed barcode sets

The Genexus™ Dx Software includes system-installed barcode sets as presets to use in assays. System-installed barcode sets are locked and cannot be changed.

| System-installed barcode set | Description |
|---|---|
| Genexus™ Dx Barcodes 1–32 HD | A unique set of 32 barcode adaptors designed and validated for optimal performance with the Genexus™ Dx Integrated Sequencer. |

# System-installed plugins

Genexus™ Dx Software includes system-installed plugins that expand the analysis capabilities of the software. System-installed plugins are locked and cannot be changed.

You can have results from these plugins automatically included as part of the sample results.

| System-installed plugin | Description |
|---|---|
| coverageAnalysis | This plugin provides statistics and graphs that describe the level of sequence coverage that is produced for targeted genomic regions.<br>This plugin generates QC metrics. For more information, see “QC results” on page 92. |
| molecular Coverage Analysis | This plugin generates QC metrics. For more information, see “QC results” on page 92. |

# System-installed reference genome files

The Genexus™ Dx Software includes system-installed reference genome files as presets to use in assays. System-installed reference genome files are locked and cannot be changed.

| System-installed reference genome file | Description |
|---|---|
| hg19 | Human reference genome. |

# Default report templates

Genexus™ Dx Software supports the default report templates that are imported with assay definition files. The report templates are pre-configured to generate variant reports and can be used as a template to generate new report templates that include additional customizations. The default report templates are locked and cannot be changed.

# System-installed sample attributes

The following table lists and describes system-installed sample attributes. System-installed sample attributes cannot be edited.

| Sample attribute | Description |
|---|---|
| Sample Name[1] | A unique identifier representing the sample.<br>The sample name can contain only alphanumeric characters (0–9, Aa–Zz), periods (.), underscores (_), or hyphens (-), cannot contain spaces, and is limited to a maximum of 20 characters.<br>IMPORTANT! To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample.<br>Note:<br>· Samples that have been used in a run cannot be deleted.<br>· To prevent duplication, the software checks all sample names and returns an error message if a non-unique sample name is detected. |
| Patient Id[1] | A unique identifier for the patient. |
| Date of Birth[1] | The date of birth for the patient. |
| Ordering Physician[1] | The name of the physician who created the order for the sample. |
| Collection Date[1] | The date that the sample was collected.<br>Click Calendar to select the date in the correct format. |

*(continued)*

| Sample attribute | Description |
|---|---|
| Gender[1] | The biological sex of the sample: Female, Male, or Unknown.<br><br>IMPORTANT! Male or Female must be selected for correct measurement of AR CNV. |
| Sample Type[1] | A term that describes the sample, for example, RNA. You can also select Other, then enter a custom sample type. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, or RNA. |
| Application Category[1] | The sample application category, such as Cancer (Germline) or Solid Tumor.<br><br>Note: If you select an oncology application category in this list, the Cancer Stage, Cancer Type, % Cellularity, and % Necrosis attributes listed below become available in the Add New Sample dialog box. |
| Cancer Type[1] | The type of cancer that is represented by the sample.<br><br>Select the type of solid or hematologic cancer. If cancer type is unknown, select Unknown Primary Origin. |
| Cancer Stage[1] | The stage of the cancer from which the sample was collected.<br><br>Select Stage 0–IV, or Primary, Unknown, or Other. |
| % Cellularity | The percentage of tumor cells over normal cells in the sample. This is a whole number between 1 and 100.<br><br>IMPORTANT!<br>▪ If this value is not set, % Cellularity is assumed to be 100% in calculations that use this attribute.<br>▪ % Cellularity is a required attribute for CNV analyses. Do not leave the field blank.<br>▪ If % Cellularity value is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see “CNVs table” on page 84. |
| % Necrosis | The percentage of cellular necrosis in the sample. This is a whole number between 1 and 100. |
| Notes | An open-entry field for more sample information. |

[1] Required attribute

# System-installed verification templates

Field Service Engineers use the system-installed verification templates during Genexus™ Integrated Sequencer installation.

IMPORTANT! The **Assays / Verification Templates** screen lists the verification templates that are used only by service engineers during Genexus™ Dx Integrated Sequencer installation and performance qualification testing. These templates should not be used or modified in any way.

Details about each verification template is available when you click the name of the template in the software.

- **Performance Qualification Assay**

# C Troubleshooting


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## Download Instrument Diagnostics

You can download diagnostics files for troubleshooting purposes. Instrument diagnostics files include:

- Diagnostic archive file (ZIP format)
- Init.log file (TXT format)
- Installation Acceptance Report (PDF format)

You must upload the instrument diagnostics files from the Genexus™ Dx Integrated Sequencer before you can download them in Genexus™ Dx Software.

For more information, see the *user guide for the panel that you use*.

1. In the menu bar, click **(Settings)►Services**.
2. Scroll to the bottom of the screen.
3. In the **Download Instrument Diagnostics** section, in the **Select Instrument** dropdown menu, select the instrument.
4. Click **Download**.

# Customer Support Archive (CSA)

You can download an archive that a Technical Support representative can use to diagnose Genexus™ Dx Software issues. The Customer Support Archive (CSA) contains log files and other technical data about your Genexus™ Dx Software and other files from sequencing runs on a Genexus™ Dx Integrated Sequencer.

You can access customer support archive files from the **Results / Sample Results** and the **Results / Run Results** screens.

## Download a Customer Support Archive

When you download a Customer Support Archive (CSA) in Genexus™ Dx Software, files are included for all samples for a specific assay. If the run includes only one assay, files for all samples are included. If the run includes more than one assay, you can specify the assay of interest when you download the CSA.

1. In the menu bar, click **Results►Sample Results**.
2. In **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.

   Alternatively, you can download CSA files three other ways.

   - Click **Results►Sample Results**. Click the sample name of interest. Then, click ••• **(More Options)►CSA.**
   - Click **Results►Run Results**. Click the run name of interest. Place the pointer over the run of interest, then click **CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.
   - Click **Results►Run Results**. Click the run name of interest. Then, click ••• **(More Options)►CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to the folder that you specified to download files from the browser. This location depends on the browser settings. You can attach the archive to an email to send to Customer Support.

# Troubleshoot Genexus™ Dx Software

| Observation | Possible cause | Recommended action |
|---|---|---|
| Cannot sign in to the Genexus™ Dx Software | You have either forgotten your password or are signed out due to several failed login attempts. | Contact the Genexus™ Dx Software system administrator. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Batch sample import fails | One or more entries in the sample-import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the sample-import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample-import template file, and be careful to copy only those rows that contain actual data. |
| | The sample import spreadsheet contains a nonunique sample name. | Every sample name in the software must be unique. Ensure that the spreadsheet does not contain any duplicate sample names, then repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1. |
| | The headings in the sample import spreadsheet do not match the sample attributes in the software. | The headings must match the sample attributes in the software exactly. Check the headings for spelling or other errors. |
| Library batch import fails | One or more entries in the library batch import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the library batch import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template file, and be careful to copy only those rows that contain actual data. |
| | The library batch import spreadsheet contains a nonunique **Library Batch ID**. | Each **Library Batch ID** in the software must be unique. Ensure that the spreadsheet does not contain any duplicate IDs, then repeat the import. The system check is not case-sensitive. For example, a **Library Batch ID** of ABC1 conflicts with abc1. |
| | A sample name entered in the library batch import spreadsheet does not match a sample name listed in the **Manage Samples** screen. | Ensure that the sample names entered into the spreadsheet are correct and match an existing sample name added to the software. |
| | The Barcode ID name format does not exactly match the format that is used in the **Prepare Library Batch** dialog box. | Use the name format following the Barcode ID name format found in the Barcode Set reference lists (**Settings►References►Barcode Set**), for example: IonDual_0101 through IonDual_0196, or IonHDdual_0101 to IonHDdual_0132. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Library batch import fails *(continued)* | An invalid library, control, or panel kit barcode has been entered in the spreadsheet. | Ensure that you have correctly entered a valid kit barcode in the appropriate cell of the spreadsheet. |
| Cannot download run result files | The run failed. Links to run files are not available for runs that fail QC. | Repeat the run. |

# D Performance characteristics

For performance characteristics of the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

# Customer and technical support


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Visit **thermofisher.com/support** for the latest in services and support, including:

- Worldwide contact telephone numbers
- Product support
- Order and web support
- Safety Data Sheets (SDSs; also known as MSDSs)

Additional product documentation, including user guides and Certificates of Analysis, are available by contacting Customer Support.

## Obtaining Certificates of Analysis

The Certificate of Analysis provides detailed quality control and product qualification information for each product. Certificates of Analysis are printed and shipped with the product.

## Obtaining Certificates of Conformance

The Certificate of Conformance provides information on conformance testing of each instrument provided with the system. Certificates of Conformance are shipped with the instrument, and are also available by contacting Customer Support at **thermofisher.com/support**.

## Serious incidents

Any serious incident that has occurred in relation to the device shall be reported to the manufacturer and the relevant regulatory authority in which the user and/or the patient is established.

# Glossary

## Genexus™ Dx Software definitions

### archive

The process that occurs when runs on the Ion Torrent™ Genexus™ Dx Integrated Sequencer reach the maximum time to be on the sequencer and the run is eligible for deletion from the sequencer.

### adapter dimer (ISPs)

ISPs that carry DNA insert length of less than 8 bp.

### aligned read length

The aligned length of a read at a given accuracy threshold is defined as the greatest position in the read at which the accuracy in the bases up to and including the position meets the accuracy threshold. For example, the AQ17 length of a read is the greatest length at which the read error rate is 2% or less. The "perfect" length is the longest perfectly aligned segment. For all of these calculations, the alignment is constrained to start from position 1 in the read—that is, no 5′ clipping is allowed.

### aligned reads

The number of bases covered by reads aligned to the reference sequence.

### AQ score

An alignment quality (AQ) score defines read accuracy when compared to the reference sequence. The discrepancy can be biological (real variant) or technical (sequencing error). For example, an AQ score of 17 represents 2% aligned read error rate, while an AQ score of 20 represents 1% aligned read error rate when compared to the reference sequence.

### average base coverage depth

The average number of reads of all targeted reference bases.

### average base read depth

The average number of reads of all targeted reference bases that were read at least once.

### backup

The process of data being copied to an external storage device or system from the Ion Torrent™ Genexus™ Dx Integrated Sequencer. Backups of data are done each night, based on backup settings, and data that is backed up is not deleted from the sequencer. When data is backed up, the same data remains on both the Genexus™ Dx Integrated Sequencer and on the external storage system.

### BAM file

A BAM (binary alignment map) file (.bam) is the binary version of a SAM (sequence alignment map) file (.sam). A SAM file is a tab-delimited text file that contains sequence alignment data. A BAM file contains aligned reads sorted by reference location.

### barcode

A barcode is a machine-readable code in the form of numbers and a pattern of parallel lines of varying widths, printed on and identifying a product.

There are several applications for barcodes. Libraries can be molecularly barcoded with unique nucleic acid sequence identifiers. Library barcodes are used during data analysis to sort the sequencing results from sequencing reactions that contain combined libraries. Chips and sample tubes also contain unique numeric barcodes that aid in the setup of the experimental analysis workflow.

### base calling

Base calling is the process by which raw data from the sequencing instrument is converted to nucleotide sequences. This is performed by base calling software that is usually run from the instrument itself.

### basecalling input file

Signal processing input files are converted to a single condensed basecalling input file that represents the processed signal. Basecalling input files are required files for basecalling.

### bead loading

The percentage of chip wells that contain live ISPs. The percentage value considers only potentially addressable wells. Bead loading is calculated as number of loaded ISPs divided by the number of potentially addressable wells.

### bp

Abbreviation for "base pair(s)".

### cellularity (%)

The percentage of tumor cells in a given sample.

### BED file

Browser Extensible Data file—BED file—defines chromosome positions or regions.

### CNV

Copy number variation (CNV) is the variation in copy number of any given gene between two samples. CNV is a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals in the human population.

### contig

A contig (from contiguous) is a set of overlapping DNA segments that together represent a consensus region of DNA.

### CSV file

A comma-separated values (CSV) file is a delimited text file in which each line represents a data record with information fields separated by a comma. A CSV file stores tabular data (numbers and text) in plain text. Each line of the file is a data record.

CSV files are easily opened using spreadsheet software, such as Microsoft™ Excel™ or Apache® OpenOffice™ Calc, where each comma-separated field is listed in a separate column.

### DAT file

A DAT file is a generic data file. It contains data that may be opened or referenced by a specific application. While some DAT files can be opened directly, most contain program data that is referenced the program when it is running. Therefore, most DAT files are not meant to be opened manually.

### empty wells

Wells on a chip that do not contain an ISP.

### enrichment (%)

Predicted number of live ISPs that have a key signal identical to the library key signal. The percent enrichment value reported is the number of loaded ISPs that are library ISPs, after taking out the test fragment ISPs.

### FASTA file

A FASTA file is a text-based format for representing either nucleotide sequences or peptide sequences, in which base pairs or amino acids are represented using single-letter codes. A sequence in FASTA format begins with a single-line description, followed by lines of sequence data.

### FASTQ file

A FASTQ file is a text-based format for storing both a biological sequence (usually nucleotide sequence) and its corresponding quality scores. Both the sequence letter and quality score are each encoded with a single ASCII character for brevity.

### FD (flow disruptiveness)

A data filtering parameter that is used instead of INDEL, SNP, and MNP.

### final library reads

Number (and percentage) of reads, passing all filters that are recorded in the unmapped BAM file. This value may be different from the total number of reads located in the Library Summary Section due to specifications associated with read trimming beyond a minimal requirement resulting in total number of reads being slightly less than Final Library Reads.

### flow

A flow is the event of exposing a chip to one particular dNTP ( deoxyribonucleotide triphosphate), followed by a washing step.

### flow order

The order in which a chip is exposed to each particular dNTP. The default Samba flow order consists of a 32-base sequence, repeated. This flow order resists phase errors by providing opportunities for out-of-phase molecules to catch up and is designed to sample all dimer (nucleotide pair) sequences efficiently. Samba is the default flow order because it improves sequencing accuracy for longer reads by resisting phase errors.

### flow transfer

Progress of the sequencing run expressed as number of total flows completed. For example, a sequencing run set to 500 flows shows 250/500 flows in the status column of the **Runs in Progress** table when the sequencing run is half completed.

### fusions

A target technique used for detection and annotation of gene fusions (or translocation of genetic material) in samples.

### hotspots file

A BED or a VCF file that defines regions in the gene that typically contain variants. Specifying a hotspots file to use in a run enables the Torrent Variant Caller (TVC) module module to identify if a specific variant is present or absent. A hotspots file instructs the Torrent Variant Caller (TVC) module module to include these positions in its output files, including evidence for a variant and the filtering thresholds that disqualified a variant candidate. A hotspots file does not affect other parts of the analysis pipeline.

### IA

Isothermal application (IA) is a template preparation technique that uses nonemulsion isothermal reaction to clonally amplify DNA onto an ISP surface.

### INDEL

INDEL is an abbreviation used to designate an insertion or deletion of bases in the genome of an organism.

### intermediate files

Intermediate files contain information used for debugging runs.

### ISPs

Ion Sphere™ Particles (ISPs) are particles that contain bound copies of a single (ideally) DNA fragment amplified during template preparation.

### key signal

Average 1-mer signal in the library key.

### library ISPs

Live ISPs that have a key signal identical to the library key signal.

### LIMS metadata

Laboratory Information Management System (LIMS) software is used for recording sequencing metadata. Entered text is associated with the Planned Run and can be extracted using APIs for LIMS consumption.

### live ISP

An ISP with a signal of sufficient strength and composition to be associated with the library or test fragment key.

### low quality ISP

An ISP with a low or unrecognizable signal.

### library key

A short known sequence of bases used to distinguish a library fragment from a test fragment (for example, "TCAG").

### MAPD

Acronym for median absolute pairwise difference. Assuming that adjacent amplicons in the genome most likely have the same underlying copy number in a sample, the difference between the log2(read count ratio) values against the reference baseline for all adjacent amplicons contains information for the noise level of the data. The median of the absolute values of all such difference in log2(read count ratio) is the measure for how informative the results fare for copy number estimates.

### mapped reads

Sequencing reads that have been mapped to the reference sequence.

### mean raw accuracy

Average raw accuracy of 1-mers at a specific position in the read.

### missense SNV

A point mutation that changes the amino acid of the respective protein. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

### MNP

Multiple nucleotide polymorphism (MNP) is a genetic mutation in an allele that differs from the reference allele of the same length by >1 nucleotide.

### nonsense SNV

A point mutation that changes one of the 20 amino acids into a stop codon, hence a shorter or unfinished protein product. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

### on target reads

Sequencing reads mapped to any target region of the reference. A read is considered to be on target if at least one aligned base overlaps a target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5′ bases of the read, is not counted.

### primer dimer

A primer dimer (PD) is a potential by-product in PCR, a common biotechnological method. As its name implies, a PD consists of primer molecules that have attached (hybridized) to each other because of strings of complementary bases in the primers

### primer dimer ISP

An ISP that carries an insert length of less than 8 base pairs.

### read length

The length of called reads measured in base pairs.

### read

The sequence of a section of a unique fragment obtained after the end of the sequencing process.

### reference library

A consensus nucleotide sequence that represents the genome of a particular species. The results from a sequencing run are compared to the reference library to identify sequence variants.

### SAM file

A SAM (Sequence Alignment Map) file is a tab-delimited text file that contains sequence alignment data. It stores biological sequences aligned to a reference sequence.

### sample

Genetic material from one source (for example, DNA from one individual).

### signal processing input files

Signal processing input files consist of the raw voltage measurement data collected during the sequencing run. These files are required to reanalyze the run from signal processing.

### SNP

Single nucleotide polymorphism (SNP) is a genetic mutation in an allele that differs from the reference allele of the same length by one nucleotide.

### SSH

SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives users, particularly system administrators, a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

### structural variants

Genetic mutations that cause a change in the organism's chromosome structure, such as insertions, deletions, copy number variations, duplications, inversions, and translocations.

### target base coverage

Summary statistics for targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

### target regions file

A BED file that specifies all of the regions that a panel represents such as the amplified regions that are used with target sequencing. The complete software analysis pipeline is restricted to include only these specified regions instead of the entire reference library.

### test fragment

A test fragment (TF) is a known nucleotide sequence that is used to monitor system characteristics.

### test fragment ISPs

Live ISPs with a key signal that is identical to the test fragment key signal.

### test fragment key

A test fragment key (TF key) is the nucleotide sequence that is used to identify test fragment reads.

### TF key peak counts

Signal strength of the first three bases of the TF key.

### total reads

Total number of filtered and trimmed reads independent of length reported in the output BAM file.

### TSV file

A tab-separated values (TSV) file is a tab-delimited file that is used with spreadsheet software. TSV files are essentially text files, and the raw data can be viewed by text editors, though they are often used when moving raw data between spreadsheets. See also *VCF file*.

### uBAM file

An unmapped BAM (uBAM) file is a variant form of the BAM file format in which the read data does not contain mapping information. This is basically an "off-label" use of the BAM format (which was specifically designed to contain mapping information) that is used for data management reasons. It allows you to attach metadata to the reads from as early on in the analysis process as possible.

### unaligned reads

Nucleotide bases covered by reads that are not aligned to the reference.

### uniformity of base coverage

The percentage of bases in all targeted regions (or whole genome) covered by at least 0.2X the average base coverage depth.

### usable sequence / usable reads

Usable reads consist of library ISPs that pass the polyclonal, low quality, and primer dimer filter.

### VCF file

A variant call format (VCF) file specifies a variant of interest and its location. This file stores the differences between the BAM file and the reference file.

### wells with ISPs

Number of wells that were determined to be positive for the presence of an ISP within the well. Wells containing ISPs have a delayed pH change due to the presence of an ISP slowing the detection of the pH change from the solution.

### XLS file

Files that use the .xls extension. XLS files can be created by Microsoft™ Excel™ and other similar spreadsheet programs.

ion torrent

# Genexus™ Dx Integrated Sequencer

## USER GUIDE

for use with Genexus™ Dx Software 6.8

Catalog Number A53256

Publication Number MAN1000073

Revision B

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
#07-06, Singapore 739256

Products manufactured at this site:

- Genexus™ Dx Integrated Sequencer
- Genexus™ Dx Software

Life Technologies Corporation |
7335 Executive Way |
Frederick, Maryland 21704 USA

Products manufactured at this site:

- Genexus™ Dx Library Strips 1 and 2-HD
- Genexus™ Dx Templating Strips 3-GX5™ and 4
- Genexus™ Dx Sequencing Kit
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Pipette Tips
- Genexus™ Dx Filter
- Genexus™ Dx Conical Bottles

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.
Revision history: MAN1000073 B (English)

| Revision | Date | Description |
|---|---|---|
| B | 25 June 2025 | • Clarified guidance on changing the Genexus™ Dx Filter and Genexus™ Dx Conical Bottles.<br>• Added other minor workflow clarifications and guidance. |
| A | 12 June 2024 | New IVD user guide for the Genexus™ Dx Integrated Sequencer for FDA submission. |

The information in this guide is subject to change without notice.

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# About this guide

CAUTION! **ABBREVIATED SAFETY ALERTS.** Hazard symbols and hazard types specified in procedures may be abbreviated in this document. For the complete safety information, see the "Safety" appendix in this document.

IMPORTANT! Before using this product, read and understand the information in the "Safety" appendix in this document.

## Purpose of this guide

This user guide provides detailed instructions for operating the Genexus™ Dx Integrated Sequencer, as well as product information, troubleshooting, instrument maintenance, and other instrument information. In addition, the guide provides basic instructions for sample entry, run planning, and data review in Genexus™ Dx Software 6.8.

For detailed instructions for using Genexus™ Dx Software for sample management, data analysis, and system management, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system.

This user guide is part of a two-guide set.

- *Genexus™ Dx Integrated Sequencer User Guide* (Pub. No. MAN1000073)
- *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074)

## Prerequisites

| Category | Prerequisites |
|---|---|
| Instrument | • Genexus™ Dx Integrated Sequencer<br>• Reagents and supplies for operating the Genexus™ Dx Integrated Sequencer |
| Software | • Genexus™ Dx Software 6.8 or later |
| Functional knowledge and understanding | • Key steps in a next-generation sequencing (NGS) workflow<br>• Main functions of the Genexus™ Dx Integrated Sequencer<br>• Main features of the Genexus™ Dx Software 6.8 or later |

# 1 Product information


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IMPORTANT! Before using this product, read and understand the information in the “Safety” appendix in this document.

## Genexus™ Dx Integrated Sequencer overview

The Ion Torrent™ Genexus™ Dx Integrated Sequencer is a next-generation sequencing (NGS) system designed to integrate library preparation, template preparation, and sequencing into a single-day, single-instrument automated run. The instrument supports Nucleic Acid to Result and Library to Result sequencing runs that include up to 32 DNA or RNA samples per run. The Genexus™ Dx Integrated Sequencer is used with Ion Torrent™ Genexus™ Dx Software and the reagents and supplies needed for operation.

The Ion Torrent™ Genexus™ Dx Library Strips 1 and 2-HD, Ion Torrent™ Genexus™ Dx Templating Strips 3-GX5™ and 4, Ion Torrent™ Genexus™ Dx Sequencing Kit, Ion Torrent™ Genexus™ Dx GX5™ Chip and Genexus™ Coupler, Ion Torrent™ Genexus™ Dx Pipette Tips, the Ion Torrent™ Genexus™ Dx Filter, and Ion Torrent™ Genexus™ Dx Conical Bottles are a set of reagents and consumables intended for use with the Ion Torrent™ Genexus™ Dx Integrated Sequencer. Nucleic acid reagent kits/tests manufactured and labeled for diagnostic purposes are required for the preparation of libraries targeting specific genomic regions.

# General instrument use

The Ion Torrent™ Genexus™ Dx Integrated Sequencer is an in vitro diagnostic (IVD) medical device intended to be used by trained personnel in a professional laboratory environment for automated, qualitative targeted sequencing of human DNA or RNA extracted from formalin-fixed, paraffin-embedded (FFPE) tumor tissue samples. The Genexus™ Dx Integrated Sequencer is not intended for whole genome or de novo sequencing.

# Special conditions statement

For *in vitro* diagnostic use. For prescription use only.

# Intended user qualifications

The intended user of the Genexus™ Dx Integrated Sequencer is trained in the basic techniques of molecular biology, and has a functional knowledge and understanding of the

- Key steps in the next-generation sequencing (NGS) workflow
- Main functions of the Genexus™ Dx Integrated Sequencer
- Main features of Genexus™ Dx Software 6.8

# Theory of operation

## Overview

Before sequencing, DNA and RNA are isolated from FFPE tissue sections. The amounts of DNA and RNA in a sample are quantified. Nucleic acid samples within an accepted concentration range are added to a sample plate. After addition of positive control DNA and RNA and no-template control, the sample plate is loaded in the Genexus™ Dx Integrated Sequencer. The sequencer is then loaded with consumables needed for the sequencing run, including a customer-derived panel which targets the variants and gene fusions of interest.

During the automated sequencer run, Ion AmpliSeq™ HD libraries are prepared from the samples and controls. The libraries, together with Ion Sphere™ Particles (ISPs), are loaded and templated in the sequencing chip, and the chip is sequenced according to run parameters specified by the assay that is selected in the run plan.

The signal generated by the sequencing reaction is translated into base calls and then reads, which are mapped to a reference sequence. Using analysis parameters in the assay designed for a particular set of targets, Genexus™ Dx Software generates reports that contain a summary of the sample test results.

## Sample preparation

FFPE tissue samples are treated with protease, then loaded into a sample input plate. The Genexus™ Dx Purification Instrument is loaded with the sample plate and consumables included with the Genexus™ Dx FFPE DNA/RNA Purification Kit. During the automated run, the instrument purifies samples using a magnetic bead based-procedure, quantifies sample concentration, then loads the purified samples in an archive plate for storage or use in a Nucleic Acid to Result run.

DNA and RNA can also be manually isolated from FFPE tissue samples using the Ion Torrent Dx FFPE Sample Preparation Kit (Cat. No. A32445), or an equivalent kit.

## Run plan creation

Information about each sample is entered into Genexus™ Dx Software, which directs the progress of the sample from creation of a run plan through automated library preparation, template preparation, sequencing, and analysis. During creation of a run plan, samples are selected and assigned to the run plan. The assay selected in the run plan specifies instrument run and analysis parameters, and directs the reporting output of sample results.

## Library preparation

In the first stage of a Genexus™ Dx Integrated Sequencer run, cDNA is prepared from sample RNA, then sample and control amplicon libraries are prepared from DNA or cDNA. assay panel primers, reagents in Genexus™ Dx Strip 1 and Genexus™ Dx Strip 2-HD, and barcode adapters from the Genexus™ Dx Barcodes 1–32 HD plate are used in library preparation. Each library that is generated in a run has a distinguishing nucleic acid sequence barcode that is incorporated into each amplified molecule.

## Template preparation

In the template preparation stage of the automated Genexus™ Dx Integrated Sequencer run, library molecules are bound onto ISPs and each nucleic acid sequence is amplified over the ISP surface using reagents in the Genexus™ Dx Strip 3-GX5™ and Genexus™ Dx Strip 4. The templated ISPs are enriched and collected using magnetic beads, then the enriched ISPs are magnetically loaded into wells of the sequencing chip. A second amplification step is then carried out with ISPs loaded on the chip, one ISP per well. After amplification, sequencing primer is annealed to the single-stranded template, and sequencing enzyme is added.

## Sequencing

In the sequencing stage of the run, the sequencer flows one of four nucleotides over the Genexus™ Dx GX5™ Chip surface in a programmed series of flows. A base that is incorporated into the newly synthesized strands of an ISP during a flow results in a release of protons and a concomitant pH change in the well. The change in pH is detected by sensors at the base of each well on the chip. Electrical signals that correspond to pH changes generated in each well during sequencing are processed and stored in data files on the system.

Throughout this procedure, as the sample is processed by the instrument, sample and reagent information is recorded and tracked by Genexus™ Dx Software.

## Data analysis

On the internal server of the Genexus™ Dx Integrated Sequencer, the initial signals are processed and each base in a sequence is called (identified). Base calls are assembled into files representing the reads, which are strings of nucleotide bases in the order found in the original library molecules. The reads are then mapped to the reference files provided with the test. Finally, Genexus™ Dx Software assesses the mapped reads at specific nucleotide locations and detects variation in sequence information in comparison to a human reference sequence.

## Results

Using parameters in the specific Assay Definition File (ADF) designed for a particular set of targets, Genexus™ Dx Software generates the following electronic results and reports for each sequenced sample and its associated controls.

Electronic results and reports generated by the software

| Results/report | Description |
|---|---|
| QC, Key Findings, and Variants screens | Contains quality control (QC) and reference information, detailed sequencing analytics, and all variant and gene fusion calls. |
| Run Report | Summary of run results and information, status, reads per barcode, and reagents used. |
| Variant Report | A report that lists the variants associated with the cancer type and detected in the sample that is screened by the assay. |

These reports are subject to approval by a lab manager or administrator via electronic signature.

# Limitations for FFPE samples

The Genexus™ Dx Integrated Sequencer has been validated to detect SNVs, deletions, insertions, and copy number variations in DNA and fusions and splice variants in RNA from FFPE tumor sections. All diagnostic tests developed for use on this instrument require full validation for all aspects of performance.

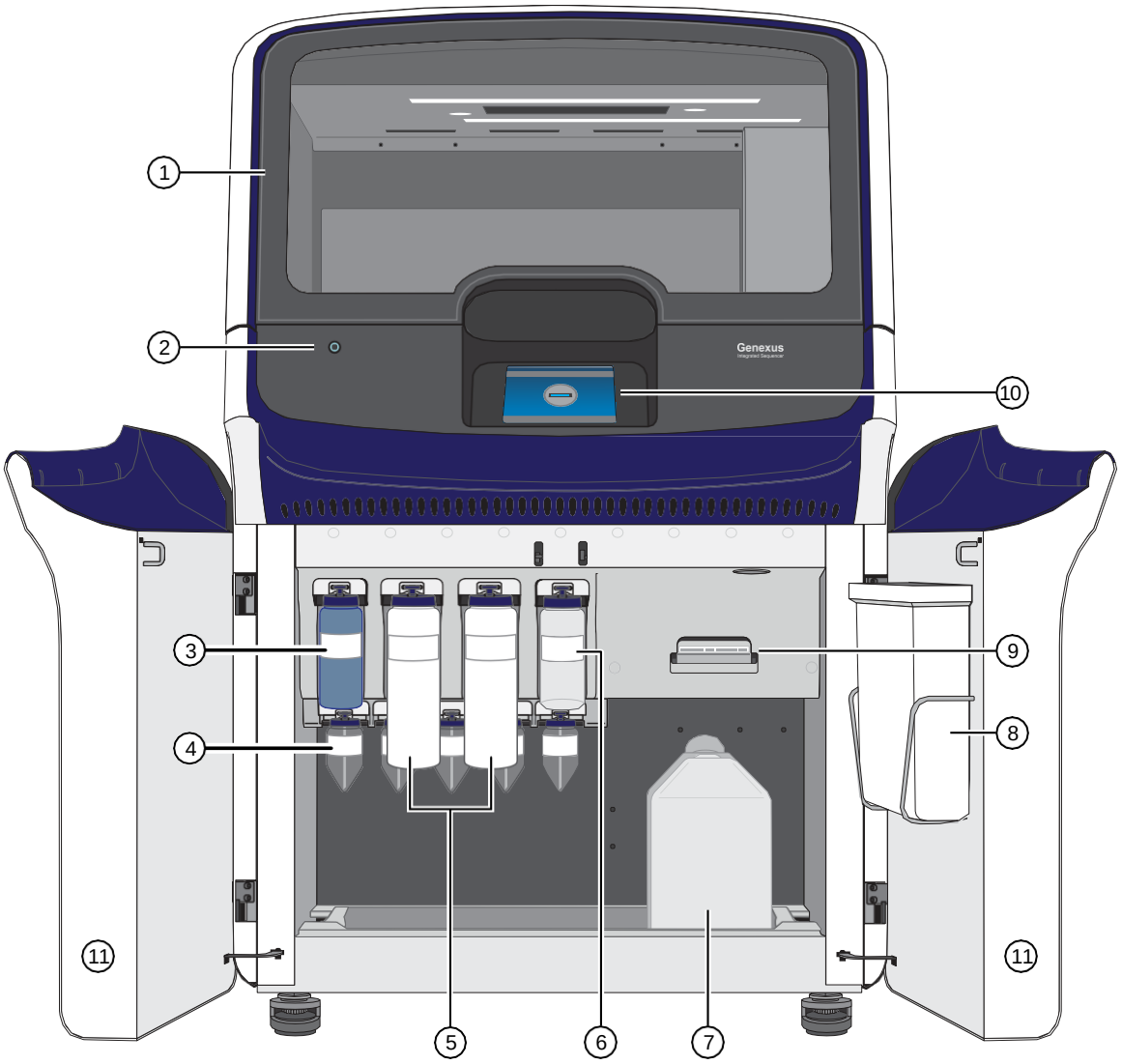
# Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software version 6.8 or later. For a complete description of software compatibility and requirements, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074).

# Genexus™ Dx Integrated Sequencer

| Components | Cat. No. |
|---|---|
| Genexus™ Dx Integrated Sequencer | A53256 |
| Genexus™ Dx Integrated Sequencer Install Kit | A52169 |

# Genexus™ Dx Integrated Sequencer components

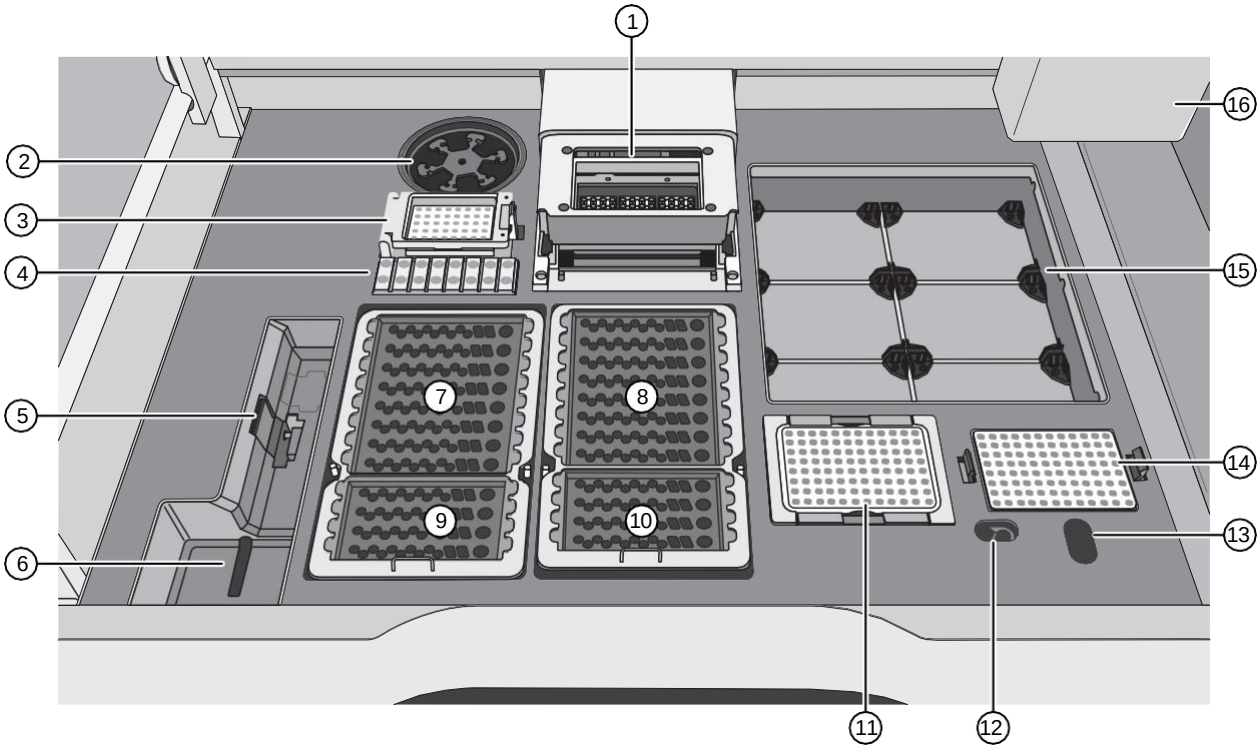



Major features and components of the exterior and sequencing reagents bay of the Genexus™ Dx Integrated Sequencer

1. Door to deck chamber. The door is locked in the closed position during an instrument run.
2. Power button
3. Genexus™ Dx Bottle 1 (Chemical Waste)
4. Genexus™ Dx Conical Bottles (Reusable conical bottles for Genexus™ Dx Cartridge reagent dilution)
5. Genexus™ Dx Bottle 2 (Sequencing Solution)
6. Genexus™ Dx Bottle 3 (Cleaning Solution)
7. Waste carboy
8. Waste pipette tip bin
9. Genexus™ Dx Cartridge
10. Touchscreen
11. Sequencing reagents bay door. Doors are locked in the closed position during an instrument run.

WARNING! This product contains very strong permanent magnets. People wearing a pacemaker or metallic prostheses should not use this product. A pacemaker or prostheses may be affected or damaged if it comes in close contact with a strong magnetic field.

# Genexus™ Dx Integrated Sequencer deck stations

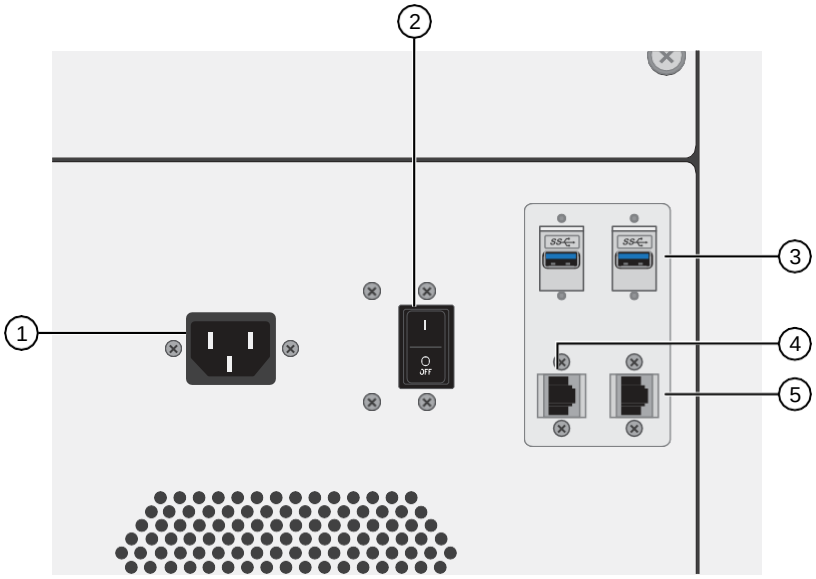



Interior Genexus™ Dx Integrated Sequencer deck components and stations

1. PCR amplification station
2. Microcentrifuge (not used)
3. Genexus™ Dx Barcodes station
4. Genexus™ Dx Primer Pool Tube station
5. Genexus™ Coupler station
6. Chip install station
7. Zone 1 station (Genexus™ Dx Strip 1)
8. Zone 2 station (Genexus™ Dx Strip 2-HD)
9. Zone 3 station (Genexus™ Dx Strip 3-GX5™)
10. Zone 4 station (Genexus™ Dx Strip 4)
11. Enrichment plate station
12. Liquid waste disposal port
13. Waste pipette tip disposal port
14. Sample plate station
15. Genexus™ Dx Pipette Tips station
16. Robotic pipettor

# Genexus™ Dx Integrated Sequencer input and output connections

The connection panel, power port, and an on/off switch are located on the right side of the rear panel of the instrument.

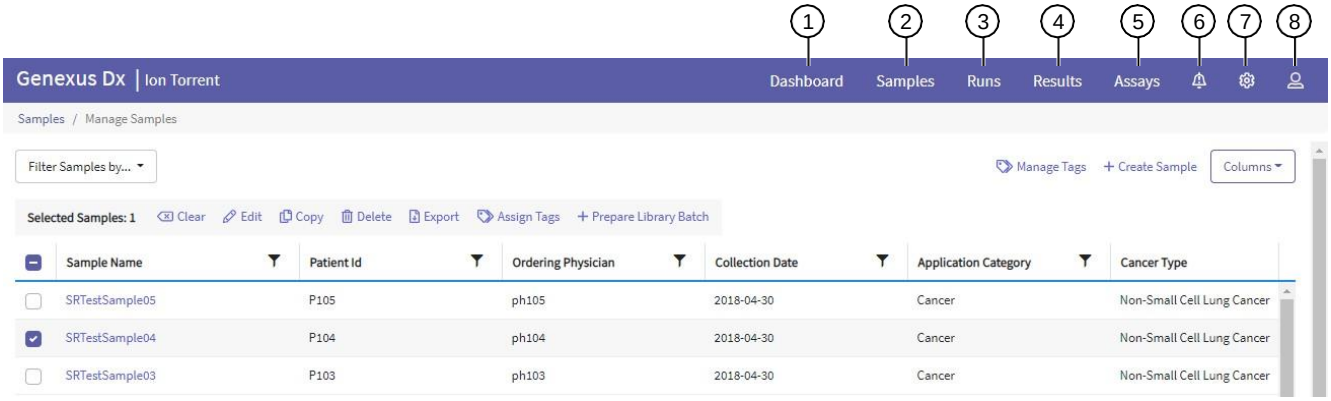



1. Power port—100–240VAC port that provides power to the instrument.
2. On/off switch—Power switch, where the states are on ( | ) or off ( O ).
3. USB ports—Connects a USB device to the instrument.
4. Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and a local area network.
5. Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and an accessory instrument.

# Genexus™ Dx Software overview

Genexus™ Dx Software provides tools to help you add and manage samples, libraries, runs, and assays. You can also view and manage sequencing results, monitor Genexus™ Dx Integrated Sequencer runs in progress, and manage software settings.

Basic software procedures such as sample creation, run planning, and review of sequencing results are described in this user guide. For more information about how to make full use of the software, including sample, run, and system management, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the on-line software help system.




1. **Dashboard**—View recent run history and current sequencing run status.
2. **Samples**—Add new samples, import samples, prepare library batches, import library batches, and manage attributes.
3. **Runs**—Plan a run starting from a nucleic acid sample, or a library. View, edit, and manage runs.
4. **Results**—View sample results, run results, and verification results.
5. **Assays**—Manage and import assays. Manage assay preset parameters and panels.
6. **Notifications**—Receive alerts and messages regarding completion of runs and other processes.
7. **Settings**—Access audit records and run logs, configure network settings, manage backup settings, restore runs, manage gene lists, link to Connect user accounts, check for software updates, and manage data archiving, disk space, and users. Field Service Engineers access verification templates during sequencer installation.
8. **Profile**—Access Help, manage and edit user profile settings and SSH key (system administrator only), sign out.

# Workflow

## Sequencing workflow on the Genexus™ Dx Integrated Sequencer

### Create samples in Genexus™ Dx Software (page 32)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as sample collection date, gender, type, and disease category.



### Plan a run in Genexus™ Dx Software (page 42)

Runs planned in Genexus™ Dx Software contain all of the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

### Dilute the samples and load the sample plate (page 53)

Dilute your nucleic acid samples, if needed, then load the sample plate.



### Load the sequencer and start a run (page 56)

Follow the step-by-step instructions in the sequencer touchscreen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



### Monitor the run (page 72)

Monitor the run in Genexus™ Dx Software in real time.

### Review data and results in Genexus™ Dx Software (page 74)

Review data and results in Genexus™ Dx Software and generate a report for each sample.



# 2 Reagents, supplies, and required materials


- Consumables overview ..... 19
- Required laboratory reagents, supplies, and equipment ..... 21


## Consumables overview

Genexus™ Dx Integrated Sequencer reagents and supplies can be ordered as kits and starter packs, but some consumables can also be ordered individually if needed.

Note: Consumables that have catalog numbers are orderable. Components that have part numbers cannot be ordered individually.

### Genexus™ Dx Library Strips 1 and 2-HD

Ion Torrent™ Genexus™ Dx Library Strips 1 and 2-HD (Cat. No. A50430) are ordered as kits with 8 pairs of strips per kit.

| Component | Carrier color | Part No. | Amount per kit | Storage |
|---|---|---|---|---|
| Genexus™ Dx Strip 1 | Light red | A50862 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 2-HD | Violet | A50861 | 8 strips | −30°C to −10°C |

### Genexus™ Dx Templating Strips 3-GX5™ and 4

Ion Torrent™ Genexus™ Dx Templating Strips 3-GX5™ and 4 (Cat. No. A50431) are ordered as kits with 8 pairs of strips per kit.

| Component | Carrier color | Part No. | Amount per kit | Storage |
|---|---|---|---|---|
| Genexus™ Dx Strip 3-GX5™ | Brown | A50864 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 4 | Yellow | A50863 | 8 strips | −30°C to −10°C |

## Genexus™ Dx Barcodes 1–32 HD

Ion Torrent™ Genexus™ Dx Barcodes 1–32 HD (Cat. No. A50410) are supplied in a plate containing 32 dual barcodes.

| Component | Label color | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx Barcodes 1–32 HD | Violet | 1 plate | 15°C to 30°C |

## Genexus™ Dx GX5™ Chip and Genexus™ Coupler

The Genexus™ Dx GX5™ Chip and Genexus™ Coupler (Cat. No. A50425) are ordered as a set that contains two chips and two couplers, sufficient for up to eight sequencing runs.

| Component | Part No. | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx GX5™ Chip | A50425 | 2 chips | 15°C to 30°C |
| Genexus™ Coupler | 100081252 | 2 couplers | |

## Genexus™ Dx Sequencing Kit

The Ion Torrent™ Genexus™ Dx Sequencing Kit (Cat. No. A50432) provides reagents and solutions sufficient to sequence up to 2 full chips.

| Component | Part No. | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx Cartridge | A50865 | 2 cartridges | −30°C to −10°C |
| Genexus™ Dx Bottle 2 | A50866 | 4 bottles | 15°C to 30°C |
| Genexus™ Dx Bottles 1 and 3 | A50867 | 2 bottles each (4 bottles total) | |

## Genexus™ Dx Pipette Tips

Ion Torrent™ Genexus™ Dx Pipette Tips (Cat. No. A50426) are ordered individually.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Pipette Tips | 12 racks | 15°C to 30°C |

## Genexus™ Dx Conical Bottles

Genexus™ Dx Conical Bottles (Cat. No. A50869) are installed in the sequencing reagents bay and serve as reservoirs for nucleotide reagent dilutions. For information on when and how to change the conical bottles, see “Replace the Genexus™ Dx Conical Bottles” on page 135.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Conical Bottles | 5 bottles | 15°C to 30°C |

## Genexus™ Dx Filter

The Genexus™ Dx Filter (Cat. No. A50868) is installed in the liquid waste disposal port on the instrument deck to prevent liquid waste line blockage. For information about installation, see “Replace the Genexus™ Dx Filter” on page 135.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Filter | 2 filters | 15°C to 30°C |

# Required laboratory reagents, supplies, and equipment

Unless otherwise indicated, all materials are available through **thermofisher.com**. "MLS" indicates that the material is available from **fisherscientific.com** or another major laboratory supplier. Catalog numbers that appear as links open the web pages for those products.

| Item | Source |
|---|---|
| Equipment | |
| Bench top microcentrifuge[1] | **MLS** |
| Sorvall™ ST 8 Small Benchtop Centrifuge (or equivalent), with<br>Thermo Scientific™ M10 Microplate Swinging Bucket Rotor (or equivalent), and<br>Sealed Bucket; Capacity: 4 Standard or 2 Midi-Deepwell plates (Set of 2) (or equivalent) | 75007200<br>75005706<br>75005721 |
| *(Optional)* Uninterruptible Power Provide (UPS)[2] | **MLS** |
| Plates, tubes, and other consumables | |
| MicroAmp™ EnduraPlate™ Optical 96-Well Clear GPLE Reaction Plates with Barcode | 4483348 |
| Thermo Scientific™ Adhesive PCR Plate Foils | AB0626 |
| 2-, 20-, 200-, and 1,000-µL pipettors and appropriate aerosol-resistant filtered pipette tips | **MLS** |
| Eppendorf™ DNA LoBind™ Microcentrifuge Tubes; 1.5-mL | 13-698-791 or **MLS** |
| DNA*Zap*™ PCR DNA Degradation Solutions | AM9890 |
| RNase*Zap*™ RNase Decontamination Solution | AM9780 |
| RNase *AWAY*™ Surface Decontaminant | 7002PK |
| Nuclease-Free Water (not DEPC-Treated) | AM9930 |
| TE Buffer, 1X Solution pH 8.0, Low EDTA | J75793.AP, J75793.AE |
| Vortex mixer with a rubber platform | **MLS** |
| Gloves, powder-free nitrile | **MLS** |

*(continued)*

| Item | Source |
|---|---|
| Ice buckets and ice | — |
| Isopropanol, 70% solution | **MLS** |
| Wipes, disposable lint-free | **MLS** |

[1] Must fit standard 0.2- and 1.5-mL microcentrifuge tubes and generate 15,000 × *g*. To change the RPMs of the centrifuge to × G in units of gravity, see **tools.thermofisher.com/content/sfs/brochures/TR0040-Centrifuge-speed.**.

[2] For laboratories that experience frequent power outages or line voltage fluctuations, recommend that you use an uninterruptible power provide that is compatible with 2500 W output or greater.

# 3 Before you begin


- Tissue input requirements for sample extraction ..... 23
- Sample storage and stability ..... 23
- Procedural guidelines ..... 24
- Power the Genexus™ Dx Integrated Sequencer on or off ..... 29
- Getting started in Genexus™ Dx Software ..... 30
- Network and password security requirements ..... 30


## Tissue input requirements for sample extraction

### FFPE sample purification

The starting material for the purification procedure is an FFPE tissue sample that is unstained and mounted on a slide, or an unstained curl. Confirm the tumor content of each sample based on the area of a hematoxylin and eosin (H&E) stained section.

The recommended number of slide-mounted 5-µm FFPE sections or curls used in extraction varies depending on the sample collection method:

| Sample type | Recommended input |
|---|---|
| FFPE tumor resections | 2 × 5-µm curls/slides or equivalent amount |
| FFPE core needle biopsy (CNB) | 9 × 5-µm curls/slides or equivalent amount |
| FFPE fine needle aspirate (FNA) | 7 × 5-µm curls/slides or equivalent amount |

For further information about how to prepare FFPE samples for purification, see the *Genexus™ Dx Purification System User Guide* (Pub. No. MAN1000137).

## Sample storage and stability

For resection samples, FFPE blocks and slides can be stored at room temperature (15–30°C) for 6 months.

- For fine-needle aspirates (FNAs), FFPE blocks and slides can be stored at room temperature for up to 6 months.
- For core needle biopsies (CNBs), blocks and slides can be stored at room temperature for up to 6 months.
- FFPE curls can be stored at 2–8°C for up to 6 months.

FFPE blocks and slides can be shipped at room temperature, but FFPE curls must be shipped at 2–8°C in low-retention tubes (Eppendorf™ DNA LoBind™ Microcentrifuge Tubes; Cat. No. 13-698-791).

Extracted FFPE DNA can be stored in low-retention tubes at –30°C to –10°C for up to 6 months, including 3 freeze-thaw cycles. Extracted FFPE RNA can be stored in low-retention tubes at –90°C to –70°C for up to 6 months, including 3 freeze-thaw cycles. Store extracted FFPE DNA and RNA in sealed archive plates at –30°C to –10°C for up to 5 months.

# Procedural guidelines

## Definitions

Throughout this guide:

- Room temperature is defined as the temperature range 15−30°C (59–86°F).
- A pulse centrifugation consists of a 3−5 second centrifugation at maximum speed in a mini centrifuge.

## Guidelines for FFPE samples

- The minimum tumor content thresholds are ≥20% for resection and fine needle aspirate (FNA) cell block samples, and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.
- For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.
- For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is <20% and the tumor content in the region of interest is ≥10%. Following tumor enrichment, proceed with the extraction protocol.
- Necrotic samples: Up to 70% necrotic tissue does not appear to interfere with sequencing results.
- Nucleic acid integrity is important for sample performance. Factors such as age of the block, fixation process used, and sample source can impact the quality of the extracted nucleic acid.
- Nucleic acid yield can be impacted by overall tissue area. If an initial extraction leads to insufficient concentrations for DNA and RNA, repeat the extractions with more material whenever possible.
- We recommend that you extract sufficient nucleic acid from samples to enable repeat sequencing runs, if possible.

## Guidelines for RNA

- Wear clean gloves and a clean lab coat.
- Change gloves whenever they may be contaminated.
- Open and close all sample tubes carefully. Avoid splashing or spraying samples.
- Clean lab benches and equipment (including gloves, tube racks, pipettes, centrifuges, and vortexers) with an RNase decontamination solution before and after use.
- Work in a designated RNase-free pre-PCR area.
- Keep RNA on ice or in a –30°C to –10°C chilled benchtop cold box during use.
- Do not vortex RNA. Flick 4 times to mix, then pulse centrifuge to collect.

## Guidelines for panel and reagent use and handling

- Use only the reagents and supplies that have been validated for your assay. For a list of validated reagents and supplies, see Chapter 2, “Reagents, supplies, and required materials”.
- Keep panel tubes capped until immediately before loading in the Genexus™ Dx Integrated Sequencer.
- Thaw positive controls on ice or at 4°C for up to 2 hours. After the positive controls are completely thawed, vortex on a platform vortexer, then centrifuge to collect tube contents. Return to ice or 4°C before loading into the sample plate.

  IMPORTANT! Ensure that contents of the control tubes are completely thawed before adding to the sample plate.

- Equilibrate Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 3-GX5™, and the Genexus™ Dx Cartridge at room temperature for 30 minutes before loading in the sequencer.
- Thaw Genexus™ Dx Strip 4 on ice or at 4°C for at least 30 minutes, or until the tube contents are fully dissolved and ready for loading on the sequencer.

  IMPORTANT! Ensure that strip contents are completely thawed before loading in the sequencer.

- Vortex thawed library and templating strips on a platform vortexer to dissolve precipitate and dislodge air bubbles. After vortexing, centrifuge the strips to collect tube contents using the Genexus™ Strip Centrifuge Adapter to hold strips during centrifugation.

  IMPORTANT! Inspect the bottom of tube 3 of the Genexus™ Dx Strip 2-HD for white precipitate before loading it in the sequencer.

  Note: For information about using the Genexus™ Strip Centrifuge Adapter, see “Centrifuge the library and templating reagent strips” on page 58.

- Verify consistency of reagent volumes across strips and panel tubes by visually inspecting them two at a time after centrifugation. For instructions, see “Centrifuge the library and templating reagent strips” on page 58. Alternatively, use the strip volume guide images provided on page 146 to check appropriate fill volumes of strip tubes.
- If loading is delayed, keep thawed panel tubes, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 4, and the sample plate on ice or at 4°C until you are ready to load the sequencer.
- Do not freeze-thaw the assay panel tubes. Thaw only the number of panel tubes that are needed for an instrument run. Discard unused panel tubes after they are thawed. Store panel tubes at −30°C to −10°C.

  Note: One assay panel tube pair (one tube with forward primers and one tube with reverse primers) is sufficient for preparing up to 4 libraries.

- Do not combine the contents of assay panel tubes. Forward and reverse primers must remain separate, until they are combined by the sequencer as part of the on-instrument library preparation workflow.

## Guidelines for preventing contamination

We recommend following these guidelines to prevent cross-contamination of samples and controls between and within sequencing runs.

Personal protective equipment

- Wear a laboratory coat that is reserved for sequencing work and is laundered frequently. If possible, change to a fresh laboratory coat before setting up a run, or use new sleeve covers.
- Wear fresh gloves to load the instruments, including during the loading of the sequencing chip and coupler. Do not remove gloves to install the sequencing chip.
- Change gloves between dispensing positive controls and no-template control (NTC) in sample plate wells.

Equipment and instrument cleaning

- If you are preparing samples and sample plates in a PCR cabinet or laminar flow hood (recommended), illuminate the hood with UV light for 15 minutes before use.
- Before a run, decontaminate equipment or bench surfaces where samples or sample plates are handled. Use DNA*Zap*™ and either 1) the RNase*Zap*™ RNase Decontamination Solution or 2) the RNase *AWAY*™ Surface Decontaminant reagent. Follow manufacturer instructions for the proper storage and use of each decontamination reagent. Alternatively, a 10% solution of commercial bleach can be used for decontaminating working areas and equipment. Follow with wiping of surfaces with wipes moistened with 70% isopropanol or 70% ethanol.
- Before and after a run, sequentially wipe the deck surface of the Genexus™ Dx Integrated Sequencer with lint-free wipes moistened with the two DNA*Zap*™ decontamination solutions followed by wipes moistened with either 1) the RNase*Zap*™ RNase Decontamination Solution or 2) the RNase *AWAY*™ Surface Decontaminant reagent. Follow with wiping of deck surfaces with wipes moistened with 70% isopropanol or 70% ethanol, or with prepared alcohol wipes. The robotic pipettor arm of the instrument can also be cleaned in this manner.

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  IMPORTANT! Do not spray decontamination solution or alcohol solution directly onto deck surfaces or into deck openings. Instead, use a lint-free wipe moistened with solution to clean surfaces. Do not use bleach to decontaminate instrument surfaces as it can cause corrosion of the metal surfaces.

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- After a sequencing run has completed, ensure that you cover the PCR amplification plate with a foil seal before removing it from the instrument to prevent library contamination.

Workflow tips

- After dispensing a positive control or sample (if applicable) in a sample plate well, do not pass the end of the used tip over wells intended for other samples or NTC. This practice minimizes the chance of depositing microdroplets in adjacent wells.
- If you dilute samples on the sample plate and vortex the plate after sealing, apply the foil seal tightly between wells using an adhesive film applicator before vortexing to prevent contamination between wells. To prevent spillage, use a PCR plate holder while sealing.
- Before disposal, close or cap used sample and positive control tubes to avoid creation of aerosols.
- Avoid touching the foil seals of the reagent strips, barcode plate, and sample plate.

- When loading the sequencer deck for a run, install the sample plate last.
- After a run, immediately seal the PCR amplification plate with a foil seal before removing the plate from the PCR amplification station. Sealing the plate before removal helps prevent contamination of libraries if libraries are recovered for reuse.

## Guidelines for nucleic acid isolation and quantification

- We recommend the Genexus™ Dx FFPE DNA/RNA Purification Kit used with the Genexus™ Dx Purification Instrument. DNA and RNA are automatically quantified and output to an archive plate. For more information, see the *Genexus™ Dx Purification System User Guide* (Pub. No. MAN1000137).
- The Ion Torrent Dx FFPE Sample Preparation Kit (Cat. No. A32445) can also be used for DNA and RNA isolation from FFPE samples. The kit also includes reagents for quantification.
- We recommend the use of fluorometric methods for quantification that are specific for DNA or RNA, if you are not using the Genexus™ Dx Purification Instrument.
- Quantification methods such as densitometry (for example, using a NanoDrop™ spectrophotometer) are not recommended, because they are not specific for DNA or RNA. Use of these methods can lead to gross overestimation of the concentration of sample nucleic acid, under-seeding of the target amplification reaction, and low library yields.
- The Genexus™ Dx Integrated Sequencer has been validated for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.

## Guidelines for Genexus™ Dx Integrated Sequencer operation

- To determine which new consumables must be loaded, and which installed consumables can be reused from a previous run, follow guidance that is provided by Genexus™ Dx Software during run planning. Consumables are listed in the run plan summary, which can be printed or saved for reference.
- To determine how many samples can be processed in a run, follow guidance that is provided by the software during run planning. The number of samples that can be included in a sequencing run depends on the following factors.

| Limiting factor | Description |
|---|---|
| The number of available barcodes in the barcode plate | The maximum number of available barcodes per run is 32.<br>IMPORTANT! When libraries are prepared on the Genexus™ Dx Integrated Sequencer, each target amplification reaction for a library requires a unique barcode. |
| The number of unused lanes on an installed chip | A maximum of 4 lanes are available on a single Genexus™ Dx GX5™ Chip. |
| The minimum read count per sample for an assay | The minimum read count per sample is an assay parameter that helps ensure a sufficient number of read counts per sample is obtained to pass QC metrics and enable high-confidence variant calling. |

- One Genexus™ Dx Strip 1 and one Genexus™ Dx Strip 2-HD is needed for each primer tube position 1–8 that is filled in a run.

- One Genexus™ Dx Strip 3-GX5™ and one Genexus™ Dx Strip 4 is needed for each chip lane that is used in a run.
- If a chip installed in a sequencer has unused lanes, do not remove it unless you are sure that you want to replace it with a new chip. After a partially used chip has been removed from the sequencer, it cannot be reinserted and reused. The sequencer cannot track lane usage after chip removal.
- You can remove a chip in one of the following situations.
  - After all the lanes of a chip are used in a run and a new run is selected, the chip shuttles to the install position and you are asked to remove the used chip.
  - When you select a run plan that requires more lanes than are available on the installed chip, you are asked to remove the partially used chip, and the sequencer performs a post-chip clean. In addition, you must clear consumables from the lower sequencing reagents bay, even if only a single lane of the chip was used. The consumables cannot be reused after a clean has been performed.
- The Genexus™ Dx Integrated Sequencer can track used and unused barcodes in barcode plates, enabling you to exchange plates between runs if needed, and reload a partially used barcode plate for a run if a sufficient number of barcodes is available on the plate.

  IMPORTANT! Do not write or mark on the foil seal of the barcode plate. The instrument vision system will be unable to distinguish used wells from unused wells, which can result in a run failure.
- Before loading pipette tip boxes in the sequencer before a run, ensure that you remove the lids from the tip boxes.
- After initial loading in the sequencer, reusable components (barcode plates, chips, and sequencing reagents bay components) must be used within 14 days for optimal results.
- An assay that is selected in a **Library to Result** run cannot include library batches that share one or more samples and barcodes.

# Precautions

## Avoid nucleic acid contamination

IMPORTANT! A primary source of contamination is spurious nucleic acid fragments from previous sample processing steps. Do not introduce amplified DNA into the work area where the instrument is located.

## Avoid strong electromagnetic radiation



WARNING! Do not use the instrument in close proximity to sources of strong electromagnetic radiation (for example, unshielded intentional RF sources), as these sources can interfere with proper operation.

### Protection by equipment

WARNING! The protection that is provided by the equipment can be impaired if the instrument is operated outside the environment and use specifications, the user provides inadequate maintenance, or the equipment is used in a manner that is not specified by the manufacturer (Thermo Fisher Scientific).

WARNING! This product contains very strong permanent magnets. People wearing a pacemaker or metallic prostheses should not use this product. A pacemaker or prostheses may be affected or damaged if it comes in close contact with a strong magnetic field.

# Power the Genexus™ Dx Integrated Sequencer on or off

## Power on

If the touchscreen is unresponsive, check the power switch on the back of the instrument and ensure that the switch is in the on ( | ) position. If the power switch is in the off ( O ) position, proceed with step 1. If the power switch is already in the on position, proceed to step 2.

1. On the back of the instrument, turn the power switch to the on ( | ) position.
2. On the front of the instrument, press the power button.
   The button illuminates.
3. In the **Sign In** screen, enter the username and password created by the field service engineer when the instrument was set up, or the unique username and password set up for you as an instrument user.
   When the instrument **Home** screen appears, the instrument is ready for use.

## Power off

It is not necessary to power off the instrument overnight or over the weekend, and the instrument does not need to be powered off if it will be idle for an extended period of time. However, if needed, follow the steps below to power off the instrument.

1. In the home screen, tap **Settings►System Tools►Shut down**.
2. Select either **Shutdown** or **Reboot**.
3. If you select **Shutdown**, a confirmation message appears. Select **Yes** to power off the instrument.

   Note: If you power off the sequencer with a partially used chip installed, the chip and consumables status is saved. When you power back on, the saved chip and consumable information enables you to use the chip for up to 14 days after the chip was installed.

IMPORTANT! Do *not* press the power button during a run. Interrupting power to the instrument during a run can result in run failure and loss of sample.

# Getting started in Genexus™ Dx Software

Before you can sign in to and use Genexus™ Dx Software, you must request a new user account from a manager- or administrator-level user. For information on user access levels, creating user accounts, and signing in to the software, and for background information on the software, see "Getting started in Genexus™ Dx Software" in the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or in the software help system.

# Network and password security requirements

## Network configuration, security, and data backup

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of your facility administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configur ation files, utilities, or instructions.

If external or network drives are connected to the sequencer, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

If a Laboratory Information Management System (LIMS) connection is set up, you can use the LIMS API to obtain the list of files generated for the assay. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074).

Note: If a LIMS system is configured to retrieve files from the sequencer, you can manually set up an FTP directory or drive mapping. This configuration is not provided as part of sequencer installation, and must be set up by your LIMS system integration or IT group.

You must have an established setup for regularly backing up and archiving your data. Data backup is solely your responsibility. In the event your system needs repair, Thermo Fisher Scientific is not responsible for data backup or any loss of data.

## Password security

Best practice is to maintain unique passwords for all accounts in use in Genexus™ Dx Software. All passwords must be reset the first time a user signs into the software. Change passwords according to your organization's password policy.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords. This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# 4 Enter samples and prepare library batches in Genexus™ Dx Software


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- Prepare or import a library batch ..... 37
- No-template and positive controls ..... 41


## Enter or import samples

Before planning a Nucleic Acid to Result run in Genexus™ Dx Software, you must first enter sample information in the software to assign sample names and provide other information. From the **Samples** menu, you can create samples in three ways.

- Enter information one sample at a time.
- Enter information for multiple samples in a table format.
- Import information for multiple samples by uploading a file.

A library batch is a group of samples that are sequenced in a Library to Result run. You can create library batches from samples you have previously added or uploaded to the software. During library batch preparation, you identify the barcode adapters that were used to prepare the libraries. You add library batches to run plans when Genexus™ Dx Software guides you through the steps to plan a Library to Result run.

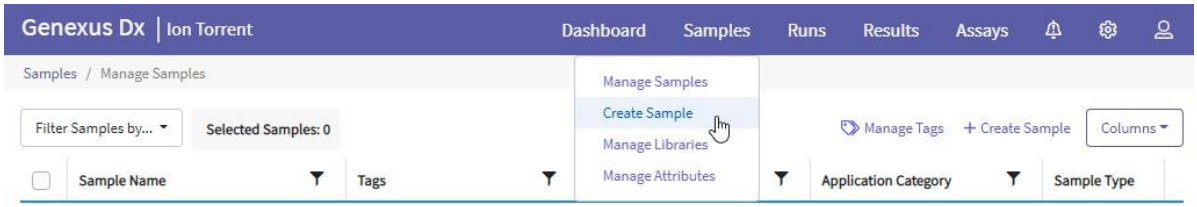
Before entering samples or preparing a library batch, you must sign in to Genexus™ Dx Software in IVD mode.

## Create a new sample

You can enter information individually for each sample that you add in the software. Complete this procedure to make a sample available for use in a run, or to add the sample to a library batch. If you have many samples to add, see “Create multiple samples” on page 33 or “Import samples” on page 35.

The information that you enter for each sample is entered as sample attributes. For more information on sample attributes, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system.

1. In the menu bar, click **Samples►Create Sample**.




Note: You can also click + **Create Sample** in the **Samples / Manage Samples** screen.

2. In the **Samples/Create Sample** screen, enter a name for the sample, select an **Application Category**, then complete the required fields, and optional fields if needed.

   Attributes identified with a red asterisk (*) are required. **Application Category** is set to **Cancer** by default. The fields for sample attributes that are displayed depend on the **Application Category** that is selected.

   - In the optional **Tags** field, enter a full or partial tag name of at least 3 characters if desired, then select one or more tags of interest to assign a tag to the sample.
   - If the tag that you want to assign to the sample does not exist in the software, a manager- or administrator-level user can enter a new tag name in the **Tags** field to create a new tag and assign it to the sample.

3. Click **Save**.

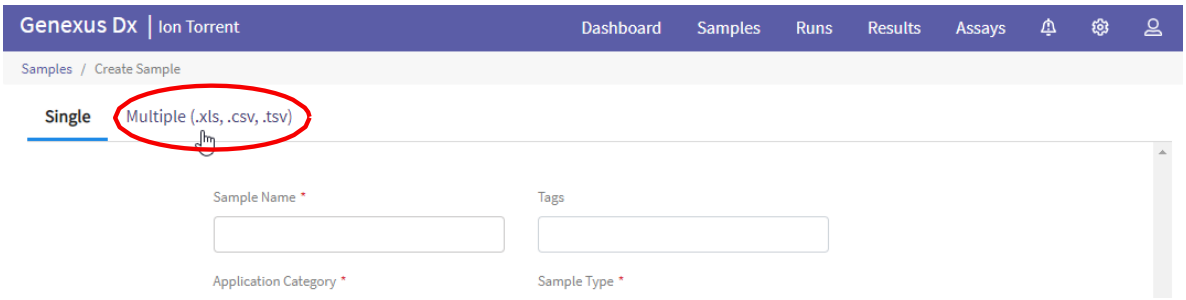
   The new sample is listed in the **Manage Samples** screen and is available to use in a run plan.

## Create multiple samples

There are two ways you can create multiple new samples in Genexus™ Dx Software.

- You can enter sample information for multiple samples in a table format in the software.
- You can import a file that contains sample data. For more information, see instead “Import samples” on page 35.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.




3. In **Application Category**, select the application category for the samples.
   A table with columns that are specific for the selected application category appears.
4. *(Optional)* Click **Columns** in the upper right corner of the screen to customize the optional attributes for the samples that you want to create.
   - Select the checkbox for a sample attribute to add a column to the table. Click the row to view fields into which you can enter attribute information.
   - Deselect the checkbox for a sample attribute to remove the attribute from the table.
5. Enter the information for a sample.
   Attributes identified with a red asterisk (*) are required.
6. Click **Add Row**, then enter the information in the new row for each new sample.
7. Repeat step 6 for each new sample.
8. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you will create.
9. Click **Save**.
   The new samples are listed in the **Manage Samples** screen and are available to use in a run plan.

   Note: Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

## Import samples

You can enter sample information for multiple samples directly in Genexus™ Dx Software. When you want to create more than a few samples, an easy and fast way to add multiple samples in the software is to create a file of information for a group of samples and import that file.

Sample data files can be used to capture, manage, and edit sample data. You can import sample data files in TXT, XLS, XLSX, or CSV file formats. For a list of the sample attributes that are included in the import file, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system. For ease of use, you can download a Microsoft™ Excel™ template file to create an import file.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In the **Application Category** dropdown list, select the application category for the samples.
   Template files are specific for each application category.
4. Select one of two options.

| Option | Description |
|---|---|
| Download a template file, then edit it to create a new file. | a. Click **Download Template**, then edit the file to add sample information. For more information, see “Download a template file for sample creation” on page 36.<br>b. After editing the file, proceed to the second option. |
| Upload the sample data from an existing file. | a. Click **Browse**.<br>b. Navigate to the file, then click **Open**.<br>The data contained in the file populates the table in the screen. |

5. Edit the sample table and data, if needed.

| Option | Description |
|---|---|
| Remove an attribute column | Click **Columns**, then deselect the column name. |
| Add an attribute column to the table | Click **Columns**, then select the column name. The column names that are listed are **Application Category**-specific. |
| Edit sample data | Click the sample row of interest, then edit the text field or dropdown list for the sample. |
| Add more samples to the table | Click **Add Row**, then enter the sample data. |
| Remove a sample from the table | Select the checkbox for the row, then click **Remove Row**. |

6. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample you intend to create.

7. Click **Save**.

   You can place the pointer over the alert (!) to view more information if needed.

The new samples are listed in the **Manage Samples** screen and are available to assign to a run plan.

## Download a template file for sample creation

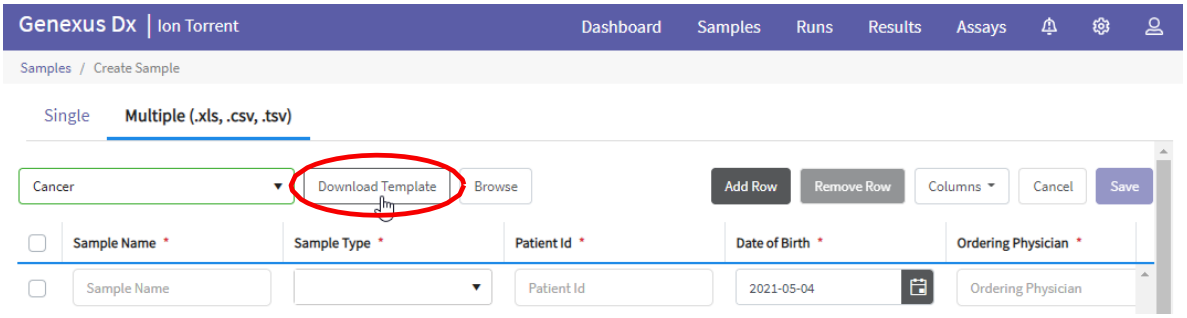
You can download a template file, add sample information to it, then use the file to import the sample data for multiple samples.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headings in the **Specimen Format** tab. Use the **Specimen Format** tab to enter samples.

Template files contain both the system-installed and the custom sample attributes as column headings.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.

   Template files are specific for each application category.
4. Click **Download Template** to download the Microsoft™ Excel™ template file.




   The template file contains default sample attributes as columns. If custom sample attributes have been configured in the software, the custom attributes are added to the template file.

5. Save the file to the computer, then open the file, enter sample data in the **Sample Details** tab.
6. When you are finished adding sample information to the file, save the file.

You can now import the file to Genexus™ Dx Software.

# Prepare or import a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have recovered from a previous sequencing run, you need to first create a library batch in Genexus™ Dx Software from samples. You can enter samples in the software or import a sample file. For more information, see “Enter or import samples” on page 32.

Select the library batch when you plan the run. If you are planning a run starting from nucleic acid samples, omit this step and proceed to “Plan a Nucleic Acid to Result run” on page 43.

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Note:

- Each library in a library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

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## No-template and positive controls

A no-template control (NTC) sample and positive controls are included in runs that are planned in Genexus™ Dx Software. The controls are added automatically after you assign samples to a run plan, or when you create a library batch. The configuration of each control is based on the assay that is used.

## Prepare a library batch

A library batch is a group of prepared libraries that are sequenced in the same library run. If you are planning a run starting from libraries that you have recovered from a previous sequencing run, you need to first create a library batch in Genexus™ Dx Software from samples that you have added. If you are planning a run starting from nucleic acid samples, omit this step and proceed to “Plan a Nucleic Acid to Result run” on page 43.

Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software to prepare the library batch and verify that the barcode scanner is compatible with the computer.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

   ---

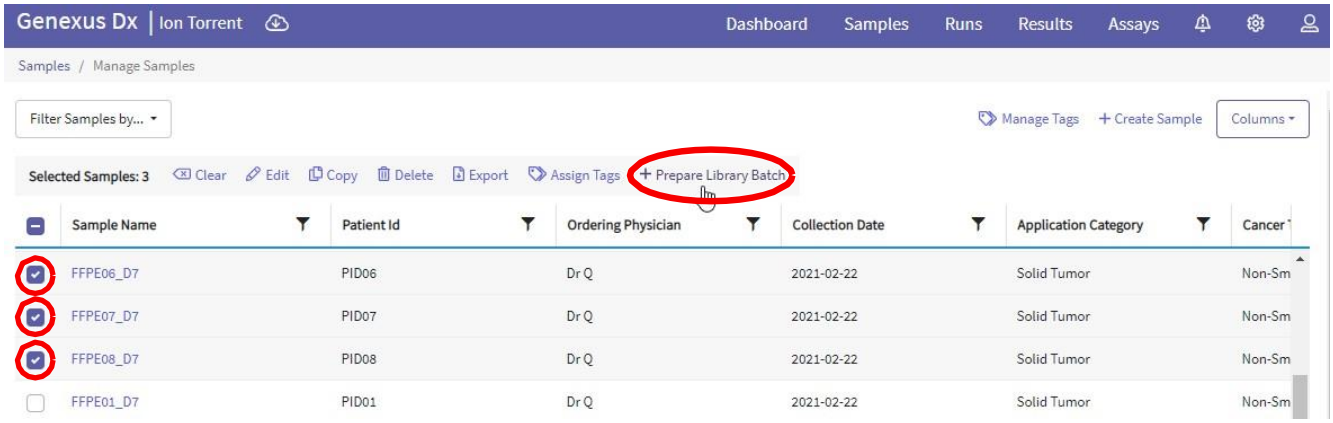
   Note: If the library batch is being created in the same sequencer where the samples were processed initially, in the **Filter Samples by** dropdown list, apply the **All** filter to be able to select the previously processed samples.

   ---

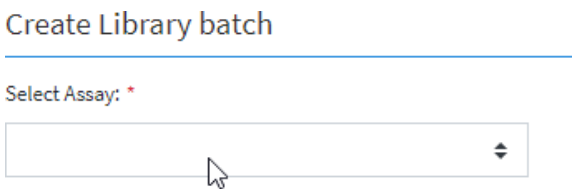
   For more information about how to filter the list of samples, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074).

3. Select samples in the list by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.

Note: Two control samples are included in each run. The controls are added automatically to a run plan after you assign one or more samples, or to a library batch when you create the library batch.



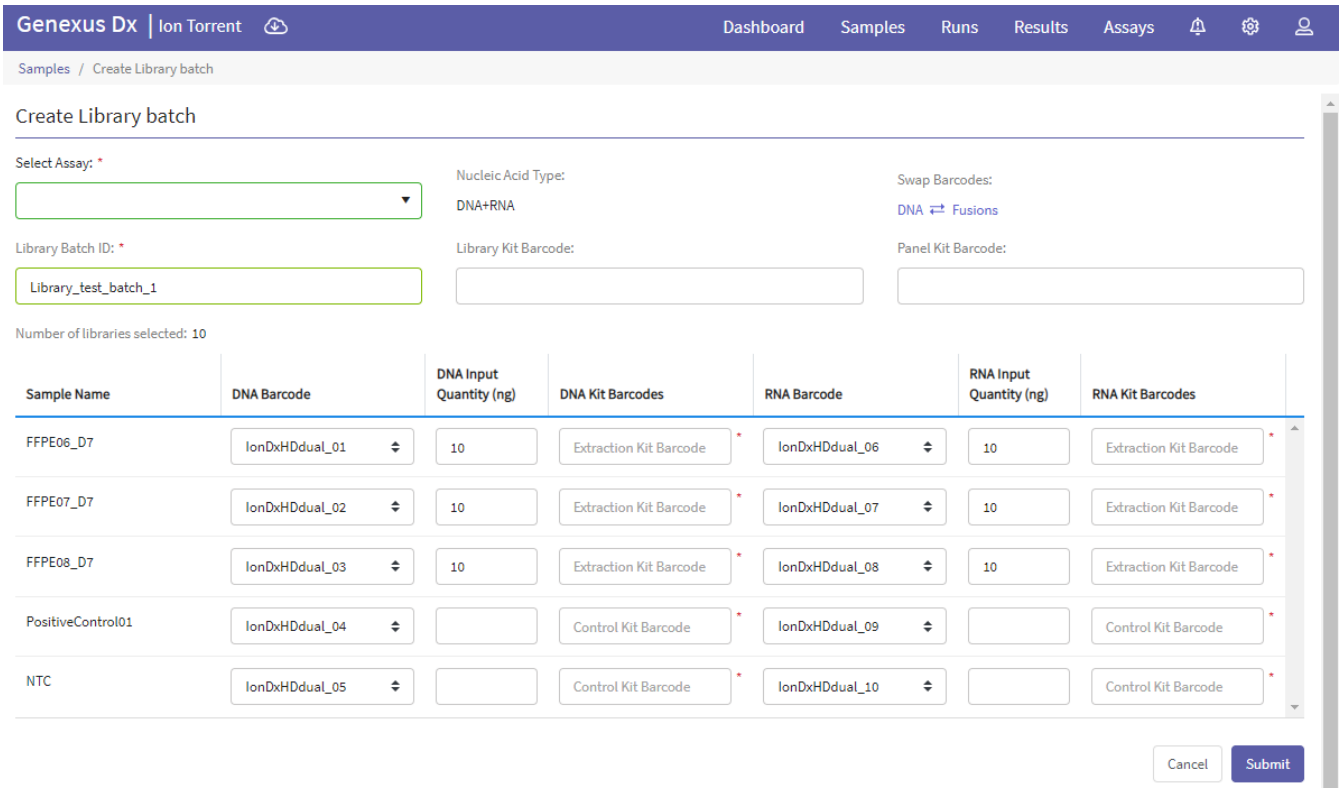

4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** dropdown list.




The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library prep type, the library preparation type automatically fills with the nucleic acid type specified by the assay you selected, for example **DNA**, **RNA**, or **DNA+RNA**.

5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.

   Note: Library Batch IDs can include only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).




6. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.

7. Select the barcode ID used to prepare each library.

   Each library in a library batch must have a different barcode ID. The barcodes that are listed in the **DNA Barcode** or **RNA Barcode** dropdown list belong to the barcode set that was selected when the assay was created.

   IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.

8. Enter the DNA and/or RNA **Input Quantity** for each library.

9. Use the connected barcode scanner to scan the barcode from the control kit into the fields in the **DNA Kit Barcodes** and/or **RNA Kit Barcodes** columns for the positive controls and the no-template controls, and for each sample.

   If needed, you can enter the barcodes manually.

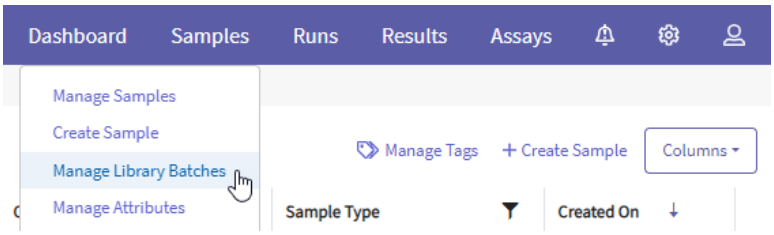
10. Click **Submit** to save and submit your selections and entries.

    The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Import a library batch

You can import library batch information in the form of an .XLS or .XLSX file. The import file must include all required library and kit information.

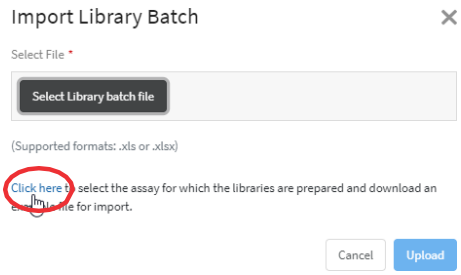
1. In the menu bar, click **Samples►Manage Library Batches**.




2. In the **Manage Library Batches** screen, click **Import Library Batch**.

3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.

   If the assay includes more than one library preparation type, a library preparation type dropdown list appears. Select the library preparation type, such as **DNA, RNA, or DNA+RNA** for the library batch.




4. Select an assay from the list, then click **Download**.
   The assay name is auto-populated in the Microsoft™ Excel™ template file that downloads to your drive.

5. In the template file, enter or confirm the library batch information.

| Template item | Description |
|---|---|
| Reagents tab | |
| Assay Name | Auto-populated when assay is selected in step 4 (required) |
| Extraction Method Type | Auto-populated when assay and library prep type, if applicable, is selected in step 4 (required) |
| Library Batch ID | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required) |
| Library Kit Barcode | For example, barcode (optional) |
| Libraries tab | |
| Sample Name | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required). For the positive control, the sample name should be *PositiveControl01*, and for the no-template control, the sample name should be *NTC*. |
| Barcode | Barcodes used for each sample and control library preparation (required) |
| Nucleic Acid Type | DNA or RNA (required) |

*(continued)*

| Template item | Description |
|---|---|
| Input Quantity | Library input quantity (optional) |
| Control Kit Barcode | The barcode for the Positive Control and NTC kits used for Nucleic Acid to Result runs (required) |
| Extraction Kit Barcode | The barcode for the kit used for nucleic acid extraction of a sample (required) |

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA library must be listed in sequential order per pool for each sample. For example, for a 1-pool DNA and Fusions assay, order should be DNA, RNA for sample 1, DNA, RNA for sample 2. For a 2-pool DNA and Fusions assay, library order should be DNA, RNA (pool 1), DNA, RNA (pool 2) for sample1, then DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 2.

6. Save the file.
7. Click **Select Library batch file**, navigate to the saved file, then select it.
8. Click **Upload**.
   A progress bar followed by an import report displays. If the import process fails, an error message indicates the reason for failure (for example, an invalid character was used). For additional troubleshooting support, see “Library batch import fails” on page 111.

# No-template and positive controls

A no-template control (NTC) sample and positive controls are included in runs that are planned in Genexus™ Dx Software. The controls are added automatically after you assign samples to a run plan, or when you create a library batch. The configuration of each control is based on the assay that is used.

# 5 Plan a run in Genexus™ Dx Software


- Before you plan a run ..... 42
- Mode switch with a partially used chip installed ..... 43
- Plan a Nucleic Acid to Result run ..... 43
- Plan a Library to Result run ..... 49


Before you can sequence samples on the Genexus™ Dx Integrated Sequencer, you need to create a run plan to specify the assay, the samples, and any additional parameters needed for the run. In Genexus™ Dx Software 6.8 and later, the following types of runs can be planned for use on the Genexus™ Dx Integrated Sequencer:

- Nucleic Acid to Result—start from purified nucleic acid samples.
- Library to Result—start from prepared and pooled libraries made previously on the Genexus™ Dx Integrated Sequencer.

In this chapter, Nucleic Acid to Result and Library to Result run planning are described. Library to Result run planning is described as a retest option for libraries that have been previously prepared and pooled on the Genexus™ Dx Integrated Sequencer and have been recovered. For more information about recovering pooled sample libraries and this type of run, see “Recover pooled sample libraries and plan a Library to Result run” on page 141.

## Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Dx Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter or import samples” on page 32.
- Ensure that the following conditions are met for a **Library to Result** run.
  - Library batches are prepared and each batch uses a unique library batch ID.
  - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

  For more information, see “Prepare or import a library batch” on page 37.

The software returns an error message when any of the conditions are not met.

# Mode switch with a partially used chip installed

In Genexus™ Dx Software you can switch from IVD mode to Assay Development mode, or from Assay Development mode to IVD mode, when you sign in to the software. If you want to switch modes to plan and perform a run in a new mode, and you have a partially used chip installed in the sequencer, you cannot use the partially used chip and sequencing consumables in the new mode. A dialog box appears asking you to confirm the mode switch. You can cancel the mode switch, but if you proceed, you must perform an instrument clean and install a new chip and consumables. You will lose the unused chip lanes and remaining sequencing consumables.

If you want to switch modes to review run data or reanalyze data in the other mode, and do not perform a sequencing run, an instrument clean is not needed. You can proceed with the mode switch and you do not need to replace the installed chip and consumables.

# Plan a Nucleic Acid to Result run

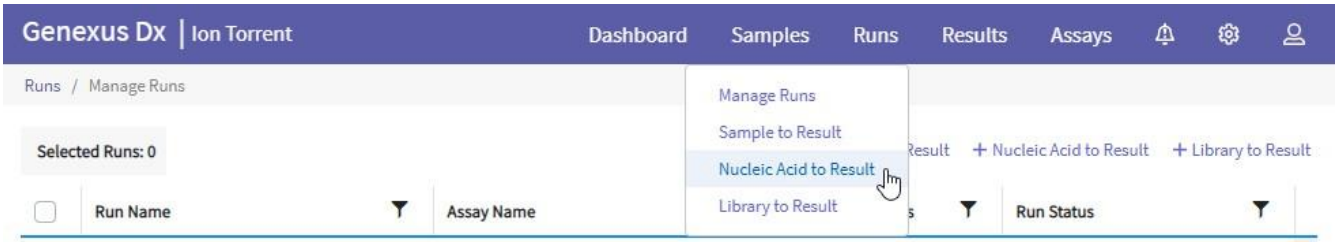
In Genexus™ Dx Software 6.8 and later, a run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Sample Plate**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Nucleic Acid to Result** screen.

Before you plan a run, do the following.

- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 33, or “Import samples” on page 35.
- Quantify sample concentration.
- Connect the barcode scanner to the computer on which you use Genexus™ Dx Software to plan runs and verify that the barcode scanner is compatible with the computer.

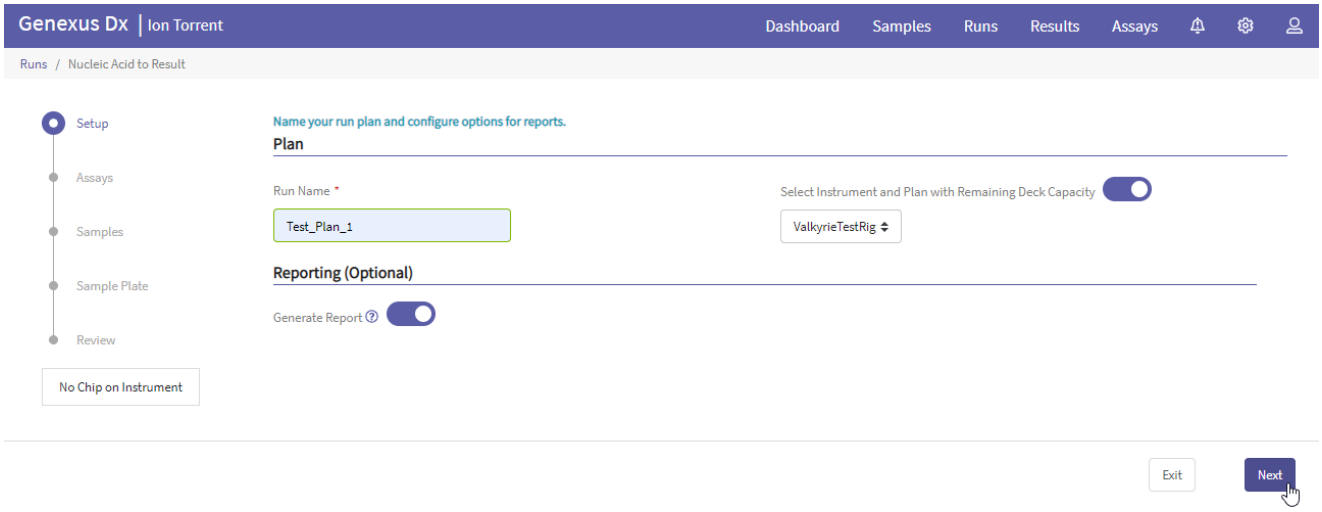
1. In the menu bar, click **Runs►Nucleic Acid to Result**.




Note: Alternatively, you can click + **Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.



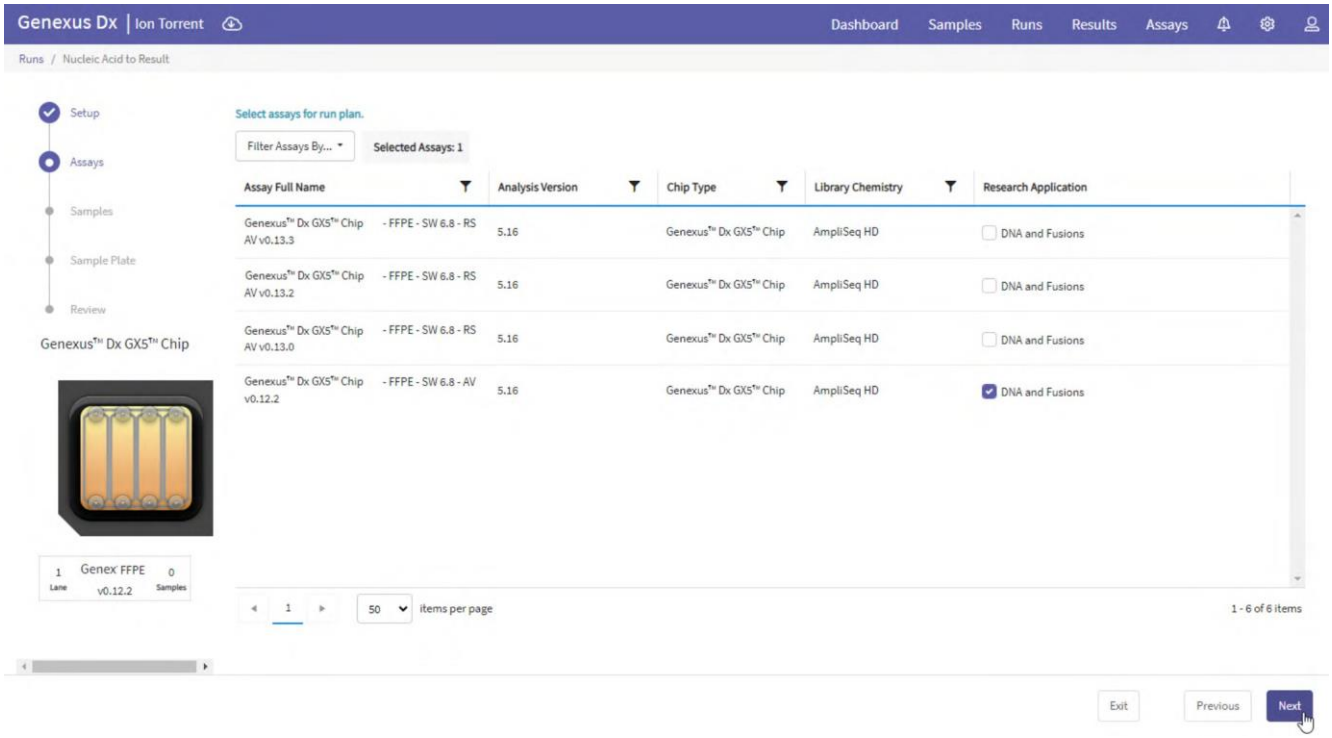

   b. *(Optional)* In the **Reporting (Optional)** section, select **Generate Report**, if needed.

      Note: Leave **Generate Report** enabled to generate a variant report using the default report template. To create a report template, click **Assays►Manage Presets**, then in the **Report Templates** tab, click + **Add New**.

   c. Click **Next**.

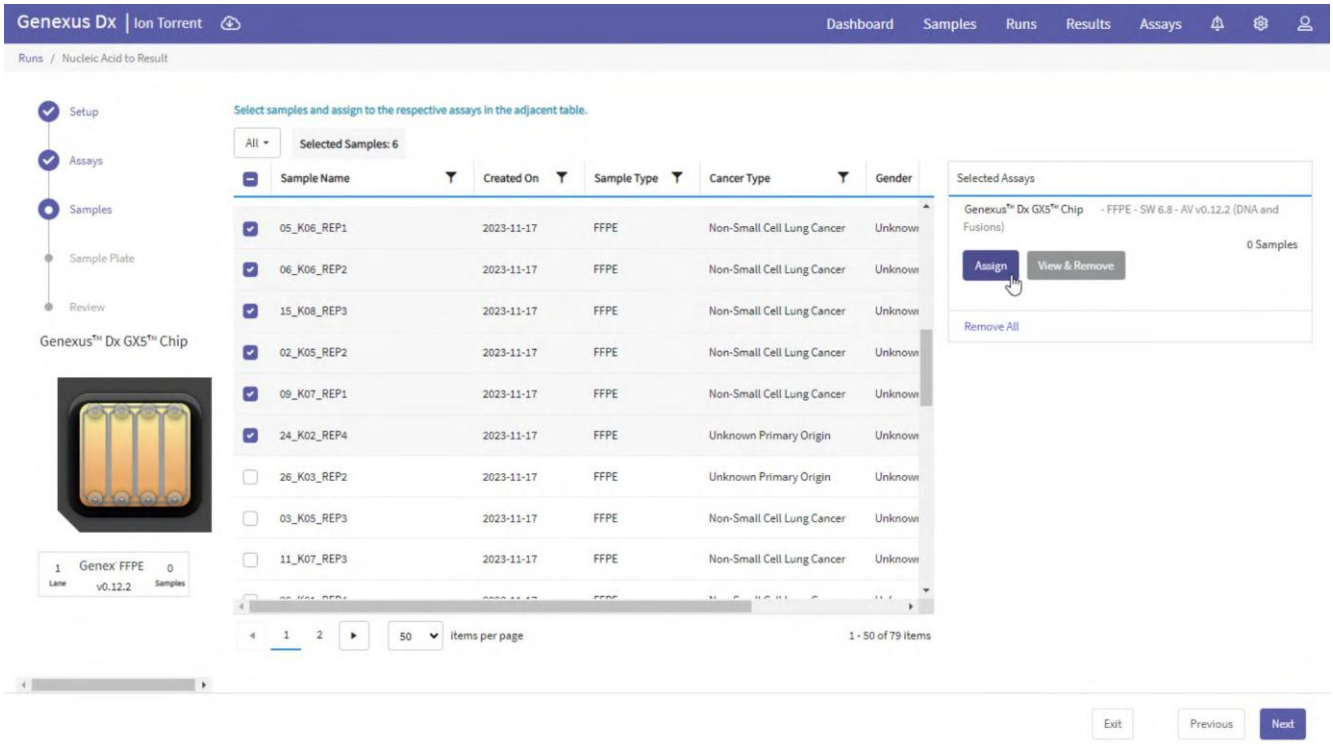
      If a chip is installed in the sequencer, the chip view graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, in the **Research Application** column, select the assay that you want to include in the run, then click **Next**.




After selecting an assay and the research application for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

4. In the **Samples** step, select the samples that you want to run with the assay.
   a. Select the checkbox next to each sample that you want to include in the run, then in the **Selected Assays** pane, for the assay that you want to assign to the selected sample or samples, click **Assign**.




The chip view updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including positive and no-template controls) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity. If the

*minimum read count per sample* × *the number of samples*

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the chip view updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.

Green lane color indicates lane usage and sample assignment within lane capacity. Red lane color indicates sample assignment that exceeds lane capacity.

   b. If you selected more than one assay, repeat substep 4a for each additional assay.

   c. If needed, edit samples in one of the following ways, then click **Next**.
      - Click **View & Remove**, make the selections, then click **Update**.
      - Click **Remove All**, make the selections, then click **Assign**.

   d. Click **Next**.

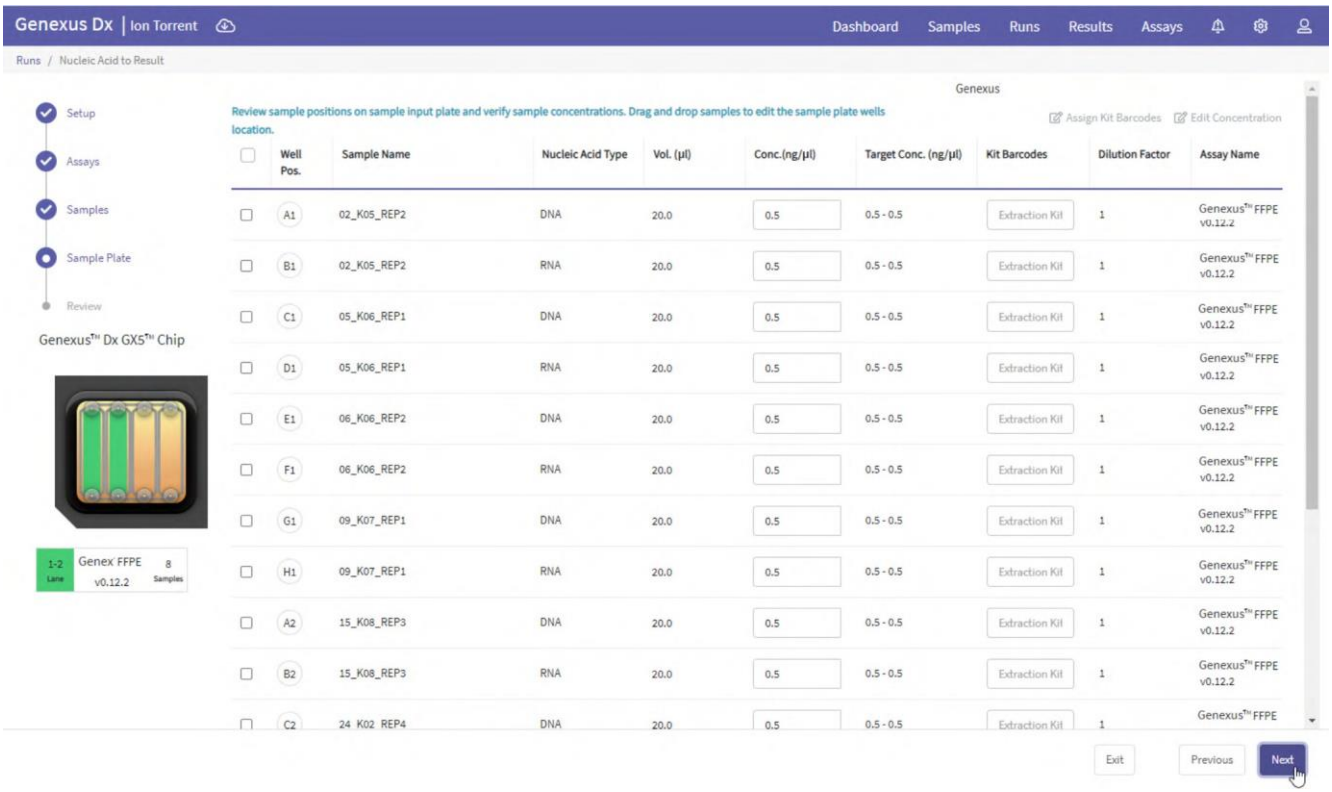
5. In the **Sample Plate** step, review position assignments in the sample plate.

   Note: Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

   a. Use the connected barcode scanner to enter the kit barcode for one or more samples or controls. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples or controls, then click **Assign Kit Barcodes**. In the **Assign Kit Barcodes** dialog box, enter the kit barcode.

      If needed, you can enter the barcodes manually.

   b. Modify the concentration of samples, if needed. For a single sample, in the row of the sample of interest, in the **Conc. (ng/µl)** column, edit the concentration.




   To modify the concentration of multiple samples, select the samples of interest, then click **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration. Click **Submit** to return to the **Sample Plate** screen.

   If a sample concentration is ≤1,024X of the target concentration for the assay, which is displayed as a default value for each sample in the **Sample Plate** screen in the **Target Conc. (ng/µL)** column, the sequencer automatically dilutes the sample to the target concentration during the run. If a sample concentration is greater than this value, you need to manually dilute

the sample to the target concentration, or to a value within range for automated dilution before loading on the sample plate.

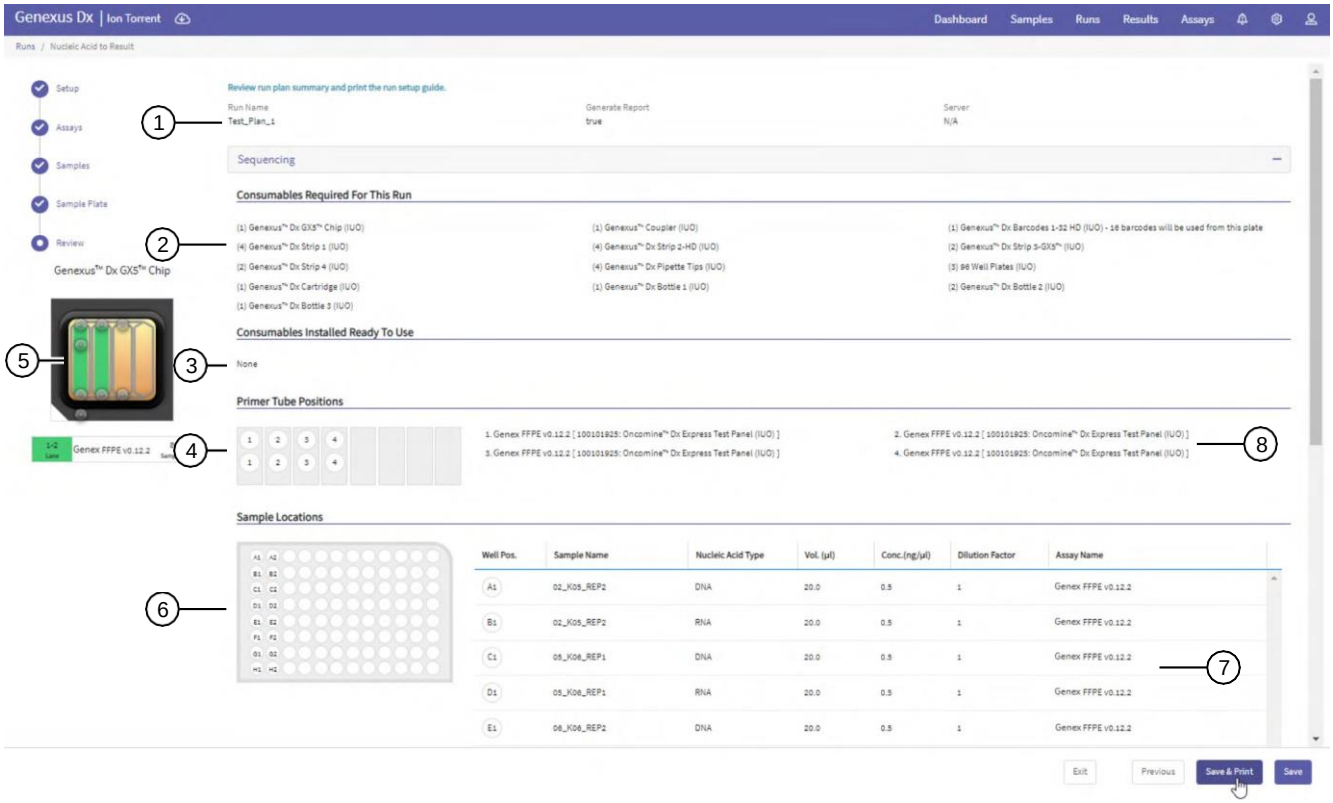
Note: The sample volume that is needed for library preparation is not adjustable. For more information, see “Load the sample plate—Nucleic Acid to Result run” for specific sample volumes to load on the sample plate.

c. Ensure that sample plate information is correct, then click **Next**.

6. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.

   The run plan summary lists the following details:

   - the consumables that are needed for this run
   - how much sample volume to load
   - where to load samples and primer pool tubes
   - details about the assay.




① Run information.
② List of consumables needed for the run.
③ List of consumables that are installed on the sequencer and available for the run.
④ Positions to load primer pool tubes.
⑤ Chip view showing the lanes to be used in the run.
⑥ Positions in the sample plate to load the samples.
⑦ Table listing the sample plate position, sample type, volume to load, concentration, dilution factor, and assay for each sample.
⑧ Description of each primer pool and its position.

The run appears in the run list on the **Manage Runs** screen with the name you specified. Select the run on the sequencer screen during run setup.

# Plan a Library to Result run

You can plan a run that starts with prepared libraries that have been recovered from a previous run. This type of run is called a Library to Result sequencing run. The volume of library recoverable from a previous run depends on the number of samples that were included in the run.

---

IMPORTANT! Before planning a Library to Result run using pooled libraries recovered from a previous run, confirm that the run failed due to a templating or sequencing problem, and that a sufficient volume of pooled sample library is available in well E4 of the PCR amplification plate. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 141.

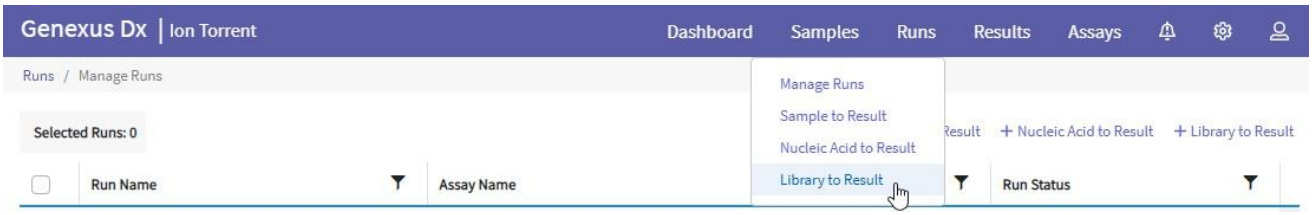
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Before you can plan a Library to Result run, you need to first prepare a library batch that associates an assay to be used in the run with the library batch. For more information, see “Prepare or import a library batch” on page 37.

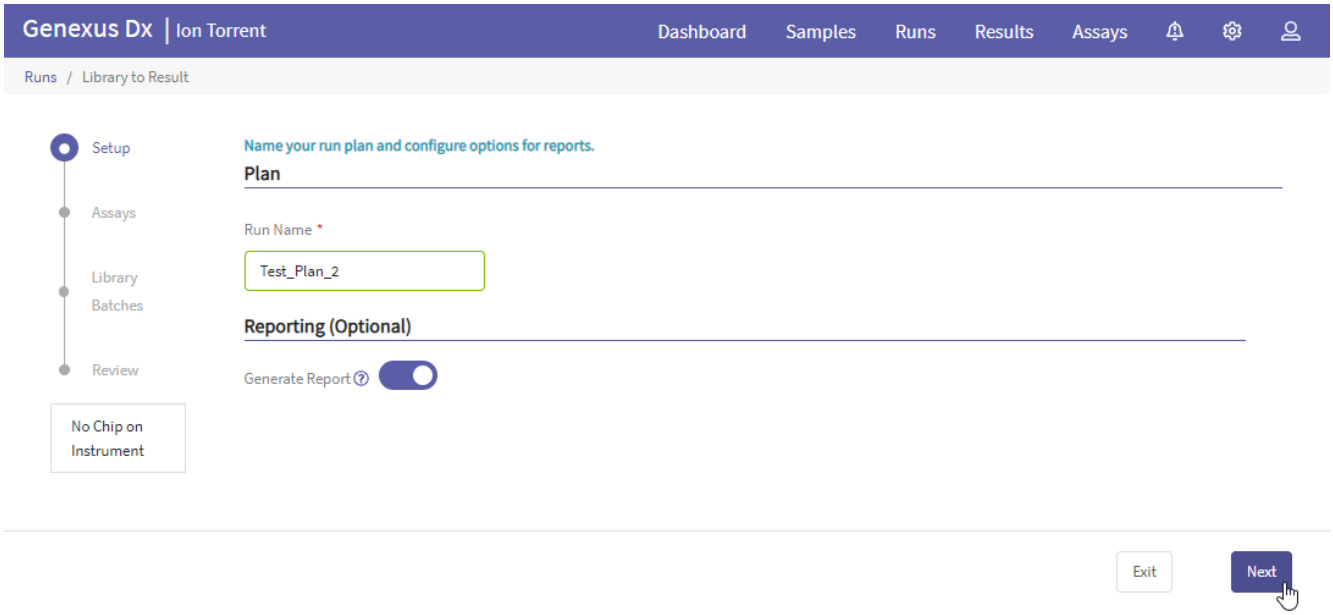
Genexus™ Dx Software guides you through the four steps of planning a Library to Result run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs►Library to Result**.

   Note: You can also click + **Library to Result** in the **Runs / Manage Runs** screen.



2. In the **Setup** step, enter a name for the run, then configure the reporting options.
   a. In **Run Name**, enter a unique name.
      The name is limited to 50 characters and no spaces are allowed.



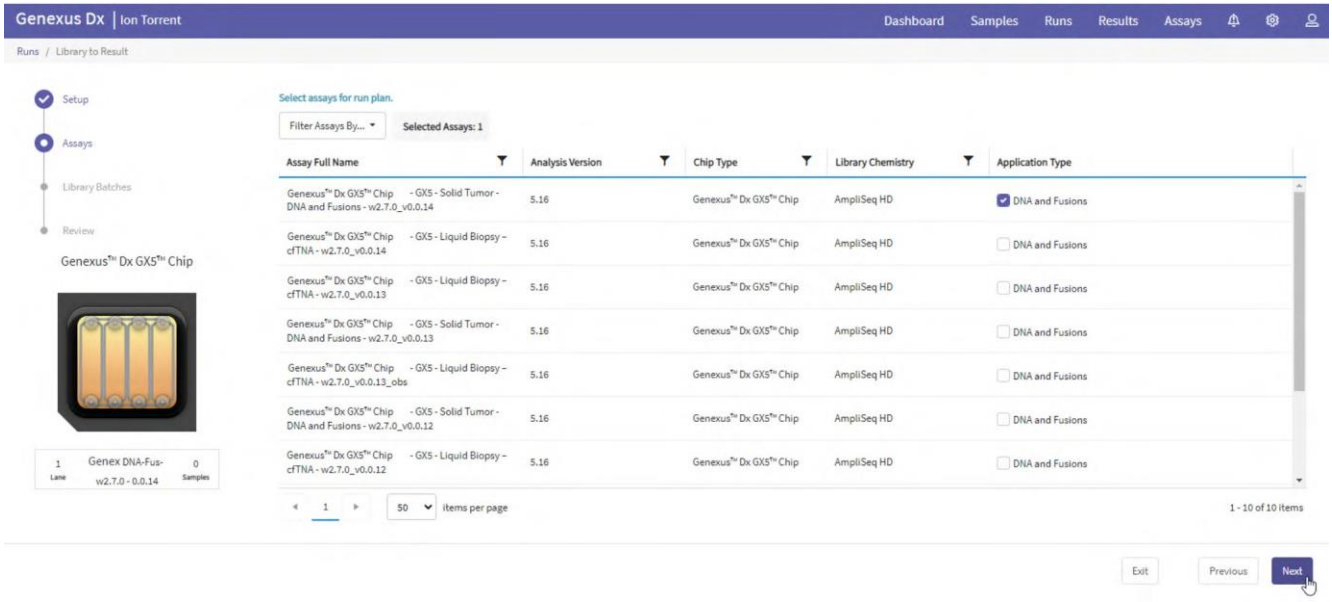

   b. *(Optional)* In the **Reporting (Optional)** section, select **Generate Report**, if needed.

      Note: Leave **Generate Report** enabled to generate a variant report using the default report template. To create a report template, click **Assays**►**Manage Presets**, then in the **Report Templates** tab, click + **Add New**.

   c. Click **Next**.
      If a chip is installed in the sequencer, the chip view graphic in the lower left corner indicates the lanes that are available for sequencing.

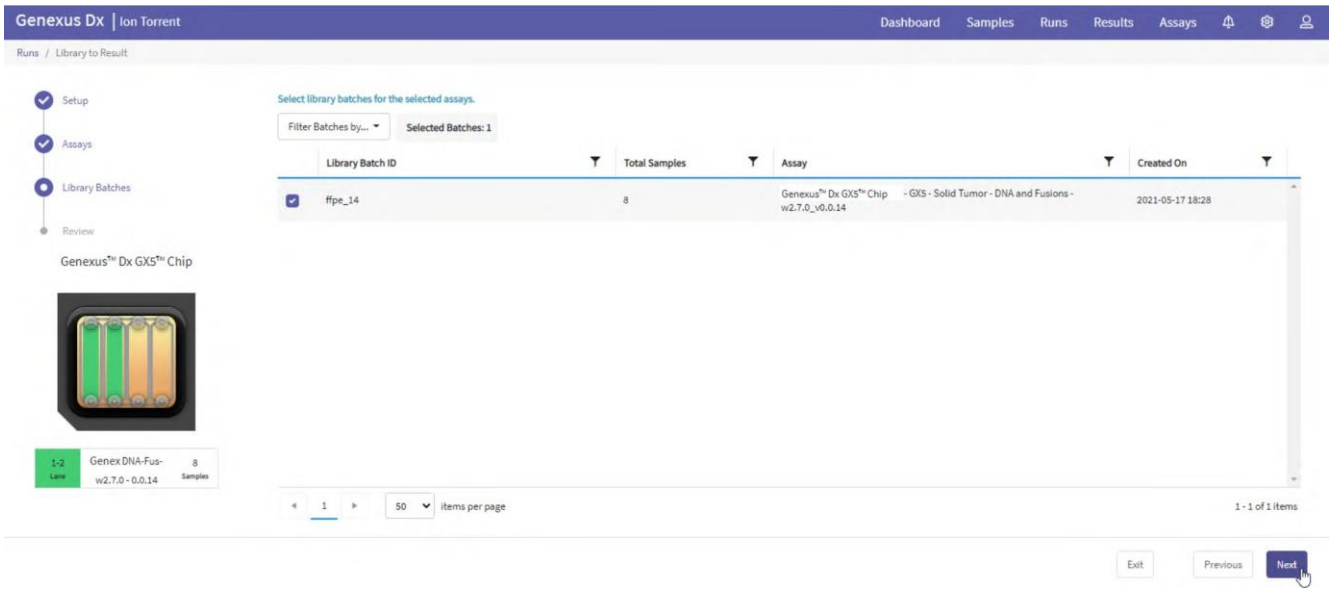
3. In the **Assays** step, select the assay that you want to use in the run, then click **Next**.

   For the assay to be selectable at this step, you need to have prepared a library batch that assigns the assay to the batch.




4. In the **Library Batches** step, select the library batch that you want to use in the run.

   Note: Only one library batch can be selected per assay.




   The chip view updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum read counts per sample for the assay.

   Green lane color indicates lane usage and sample assignment within lane capacity. Red lane color indicates sample assignment that exceeds lane capacity.

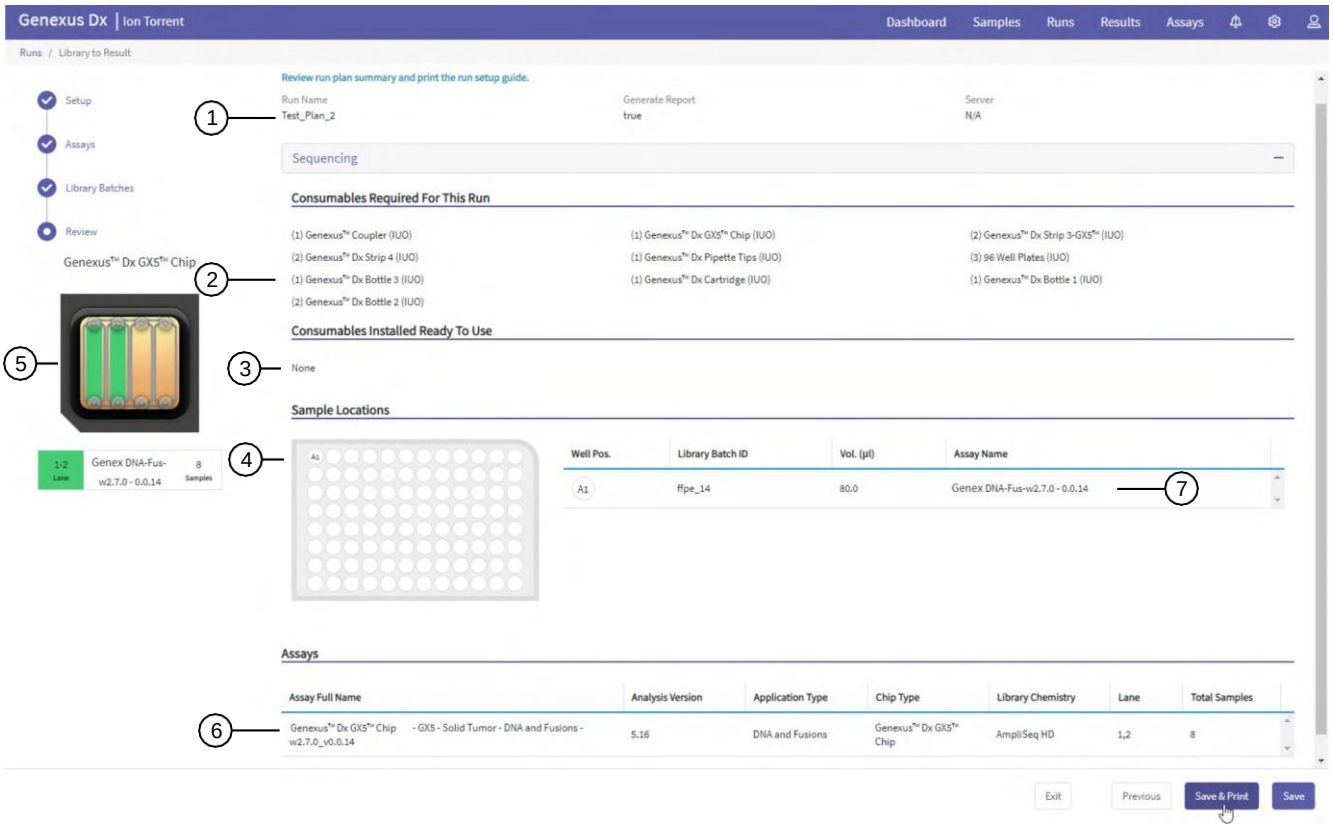
If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the chip view updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not obtain the required reads per sample to pass QC metrics.

5. Click **Next**.

6. In the **Review** step, review the run setup guide.

   - Click **Save and Print** to print the run setup guide
   - Click **Save** to save the run without printing.

   The run plan summary lists the consumables that are needed for the run, where to load the library batch on the sample plate, and how much library volume to load.




① Run information.
② List of consumables needed for the run.
③ List of consumables installed on the sequencer and available for the run.
④ Position or positions in the sample plate to load the library batch.
⑤ Chip view showing the lanes to be used in the run.
⑥ Table listing assay and run information.
⑦ Table listing the well position, library batch ID, volume to load, and assay for each library batch.

The run appears in the run list on the **Manage Runs** screen with the name you specified. Select the run on the sequencer screen during run setup.

# 6 Load the sample plate


- Dilute the samples, if needed—Nucleic Acid to Result run ..... 53
- Load the sample plate—Nucleic Acid to Result run ..... 55


Before starting a Nucleic Acid to Result run on the sequencer, you must quantify the samples, dilute them if needed, then load the sample plate.

## Dilute the samples, if needed—Nucleic Acid to Result run

Nucleic acid samples with concentrations up to 1,024X of the target concentration for an assay (displayed as default values in the **Sample Plate** step screen in run planning) are in range for automated dilution and require no manual dilution. Alternatively, DNA and RNA from FFPE samples can be manually diluted to 0.5 ng/µL. Enter the concentrations during sample run planning at the **Sample Plate** step (see step 5).

For samples with concentrations greater than the upper range for automated dilution, when setting up a Nucleic Acid to Result run, or when processing previously purified nucleic acid, you can manually dilute the sample with nuclease-free water to the target concentration of 0.5 ng/µL DNA and RNA for FFPE samples. For samples that are in range, proceed to "Load the sample plate—Nucleic Acid to Result run". For samples that need manual dilution, proceed to "Procedural guidelines for manual dilution of samples" on page 54.

| Sample type | If the sample concentration is | Action |
|---|---|---|
| FFPE DNA and RNA | ≥0.5 ng/µL, but ≤512 ng/µL | No manual dilution is necessary. The sequencer dilutes the sample to the target concentration during the run. If you are setting up a Nucleic Acid to Result run, or preparing sample for multiple assays, samples can be manually diluted to the target concentration (0.5 ng/µL). |
| | >512 ng/µL | Manually dilute to the target concentration (0.5 ng/µL), or to a concentration in range for automated dilution by the sequencer. |

Note:

- If you enter a concentration <0.05 ng/µL or >10,000 ng/µL, a warning that the concentration is out of range appears, and you are not allowed to proceed to the next step.
- If the concentration is ≥0.05 ng/µL but less than the target concentration of the assay, you can proceed. However, because the sequencer cannot concentrate samples, the sample concentration will be less than the target concentration, and variant information will not be reported.
- If the concentration is ≤10,000 ng/µL, but >1,024X of the target concentration, you can proceed. However, because the sequencer cannot dilute samples more than 1,024-fold, the diluted sample concentration will be greater than the target concentration.

## Procedural guidelines for manual dilution of samples

- Do not pipet volumes less than 2.0 µL. If sample concentration is above 5 ng/µL for FFPE samples, first dilute each sample to an intermediate dilution (for example, 5 ng/µL), using the relationship C1 × V1 = C2 × V2 (C1: initial concentration, V1: Initial volume, C2: target concentration, V2: target volume).
- Unless the test protocol dictates that multiple libraries are prepared from a single sample (for example, replicate libraries), dilute only sufficient sample to prepare a single library (20 µL total).
- If samples were previously frozen, thaw on ice or at 4°C, then vortex and pulse centrifuge to ensure samples are fully resuspended and homogeneous.
- For manual sample quantification, if not using the on-board quantification by the Genexus™ Dx Purification System in standalone configuration, we recommend using the Qubit™ Fluorometer or equivalent fluorometer with a nucleic acid-specific fluorometric kit. Sample concentration determined by UV absorbance is not recommended because UV absorbance is not specific to DNA and RNA.

## Dilute the samples manually, if needed

For samples that need manual dilution, dilute DNA and RNA from FFPE samples to a final concentration of 0.5 ng/µL.

IMPORTANT! Do not perform the following dilution procedure until you are ready to proceed to loading the sample plate. Avoid freeze-thaw cycles of samples that have been diluted to target concentrations.

1. Label a new 1.5-mL nuclease-free, low-retention microcentrifuge tube for each sample being diluted. Place tubes on ice or in a prechilled benchtop cold block until needed.

   Note: When diluting DNA and RNA samples from the same patient, we recommend that you label tubes using different color markers (for example, blue for DNA and red for RNA) to avoid mixing nucleic acid types during manual plate loading.

2. Use the sample concentrations to calculate the volume (X) of each sample needed for 10 ng of FFPE DNA and RNA. Use the following formula: (ng sample input) / (sample concentration, ng/µL) = X µL of sample.

3. Calculate the volume (Y) of nuclease-free water needed to bring FFPE sample volume to 20 µL, using the following formula:

   (10 ng / 0.5 ng/µL) – X µL of sample = Y µL of nuclease-free water

   Note: Dead volume is included in the 20 µL sample volume. However, 10% excess volume can be added if needed by multiplying the X and Y values by 1.1.

4. For each sample, pipet the calculated Y µL of nuclease-free water into the appropriate labeled 1.5-mL microcentrifuge tube.

5. Add the calculated X µL of sample into the appropriate tube, pipet up and down 3 times and vortex to mix, then pulse centrifuge.

6. Keep samples on ice, in a chilled cold block, or in a 2–8°C refrigerator until ready for use.

Proceed to “Load the sample plate—Nucleic Acid to Result run”.

# Load the sample plate—Nucleic Acid to Result run

1. Add samples and controls to the sample plate with the volume and in the well positions that are specified in the run setup guide.

   Note:
   - Do not adjust sample and control volume.
   - We recommend that you load sample plate wells in the following order:
     - Samples
     - Positive control(s)
     - No-template control (NTC)
   - Before adding the NTC, replace your gloves with a new pair of gloves.

   Sample volume

| Sample type | Volume |
|---|---|
| DNA | 20 µL |
| RNA | 20 µL |
| Positive and no-template controls | 20 µL |

2. Seal the plate with an Adhesive PCR Plate Foil (Cat. No. AB0626).

   Note: The use of other plate seals can affect performance.

3. After sealing, centrifuge the plate at 300 × *g* for 15 seconds to collect contents at the bottom of plate wells.

   Note: Ensure that the centrifuge has been wiped down and cleaned before centrifuging the plate.

4. Keep the plate on ice until you are ready to load it in the sequencer.

# 7 Start a sequencing run


- Materials required ..... 56
- Before you begin ..... 57
- Load the sequencer and start a run ..... 59
- Clear the instrument deck and perform a UV Clean ..... 67
- Options for an expired sequencer initialization ..... 70


## Materials required

Genexus™ Dx Integrated Sequencer consumables

- Genexus™ Dx Strip 1
- Genexus™ Dx Strip 2-HD
- Genexus™ Dx Strip 3-GX5™
- Genexus™ Dx Strip 4
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Sequencing Kit
  - Genexus™ Dx Cartridge
  - Genexus™ Dx Bottle 2
  - Genexus™ Dx Bottles 1 and 3
- Genexus™ Dx Pipette Tips
- MicroAmp™ EnduraPlate™ Optical 96-Well Clear Reaction Plates with Barcode (2)

Other items

- Panel, loaded into Genexus™ Primer Pool Tubes
- Genexus™ Strip Centrifuge Adapter

- Sample plate loaded with positive controls, no-template controls, and nucleic acid samples as specified by the run set up guide.

# Before you begin

Before setting up a sequencing run, review general procedural guidelines for handling panels, reagents, and samples to minimize the chance of contamination and help ensure the success of the run. See "Guidelines for panel and reagent use and handling" on page 25 and "Guidelines for preventing contamination" on page 26.

1. Remove panel tubes that are needed for the run from the freezer, then thaw on ice or at 4°C for at least 30 minutes. After thawing, centrifuge at 300 × *g* for 15 seconds using the Genexus™ Strip Centrifuge Adapter to collect contents at the bottom of the tubes. Keep the panel tubes on ice or at 4°C until you load them in the sequencer.

2. If you are installing a new Genexus™ Dx Cartridge, thaw the cartridge at room temperature for 30 minutes before loading in the sequencer.

3. Remove the library and templating strips from the refrigerator or freezer.
   a. Equilibrate Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, and Genexus™ Dx Strip 3-GX5™ at room temperature for 30 minutes before loading in the sequencer.

   b. Thaw Genexus™ Dx Strip 4 by laying the strip on ice or incubating at 4°C for at least 30 minutes, or until the tube contents are fully dissolved and ready for loading in the sequencer.

   IMPORTANT!
   - Ensure that strip contents are completely thawed before proceeding.
   - If loading is delayed, keep thawed Genexus™ Dx Strip 2-HD and Genexus™ Dx Strip 4 on ice or at 4°C until you are ready to load the sequencer.

4. Vortex each strip at maximum speed for 5–10 seconds while rocking the strips from side to side.

   Note: Vortexing helps dislodge magnetic beads that can be trapped in keyholes of Genexus™ Dx Strip 1 and Genexus™ Dx Strip 3-GX5™, and to dissolve precipitate in Genexus™ Dx Strip 2-HD, if present.

   IMPORTANT! Visually check the bottom of tube 3 of Genexus™ Dx Strip 2-HD for white precipitate by doing a pulse-vortex to resuspend and expose precipitate. If precipitate is present, continue to pulse-vortex strips every 10 minutes until precipitate fully dissolves, keeping the strips at room temperature. If precipitate fails to dissolve after 3 rounds of pulse-vortexing, replace the strip with a new Genexus™ Dx Strip 2-HD and repeat the thawing procedure.

Note: Load the Genexus™ Dx Integrated Sequencer within 2 hours of thawing reagents.

Proceed to "Centrifuge the library and templating reagent strips".

## Centrifuge the library and templating reagent strips

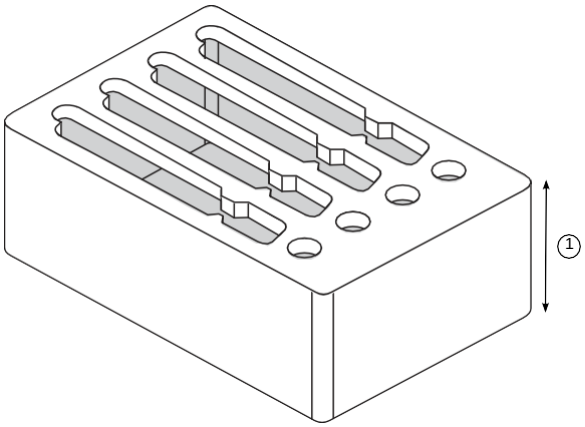
Use the Genexus™ Strip Centrifuge Adapter as a holder for centrifuging library and templating strips to collect contents in the bottom of strip tubes after vortexing. The adapter is provided at sequencer installation.

Use of the strip adapter requires a centrifuge with buckets that support the height of the holder at 44 mm so that buckets swing freely in the centrifuge rotor when loaded with strips. For more information, see “Required laboratory reagents, supplies, and equipment” on page 21. To use the Genexus™ Strip Centrifuge Adapter, follow these steps.

---

Note: The Genexus™ Strip Centrifuge Adapter can also be used for centrifuging primer pool tubes to collect tube contents.

---




Genexus™ Strip Centrifuge Adapter

① 44 mm height

1. After thawing and vortexing, load the strips in the adapters in a balanced orientation. The centrifuge buckets must support the height of the strip adapters loaded with strips.
2. Place each adapter loaded with strips in the centrifuge.
3. Centrifuge the strips at 300 × *g* for 15 seconds.
4. Remove strips from the adapters, then inspect the strips to ensure that contents have been collected and air bubbles are not present. Line up similar strips against each other and ensure that the reagent volume in each well is consistent between the two strips. Repeat this process until all strips are inspected. If volume inconsistencies are observed, do not use the strip.

   ---

   Note: Alternatively, verify well reagent volumes by comparing fill levels with the Genexus™ Dx strip figures provided on page 146.

   ---

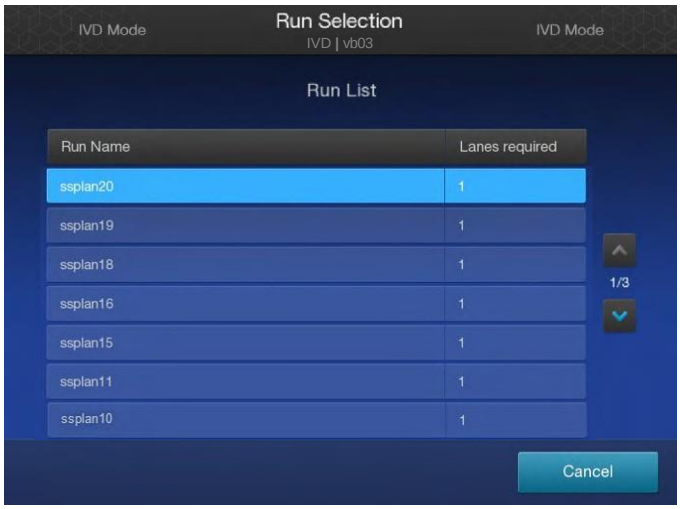
5. Repeat step 1 through step 4 for the remaining library and templating strips to be loaded for a sequencing run, if needed.
6. After centrifugation, return Genexus™ Dx Strip 4 to ice or 4°C until you are ready to load in the sequencer.
7. To verify correct panel volume of primer pool tubes, hold two sets next to each other at an angle above eye-level. If volume inconsistency is suspected, compare against the Genexus™ Dx primer pool tube figures provided on page 146.

# Load the sequencer and start a run

1. Tap **Run** on the sequencer home screen to start the setup and loading procedure.




2. In the **Run Selection** screen, select the run that you want to use from the list.

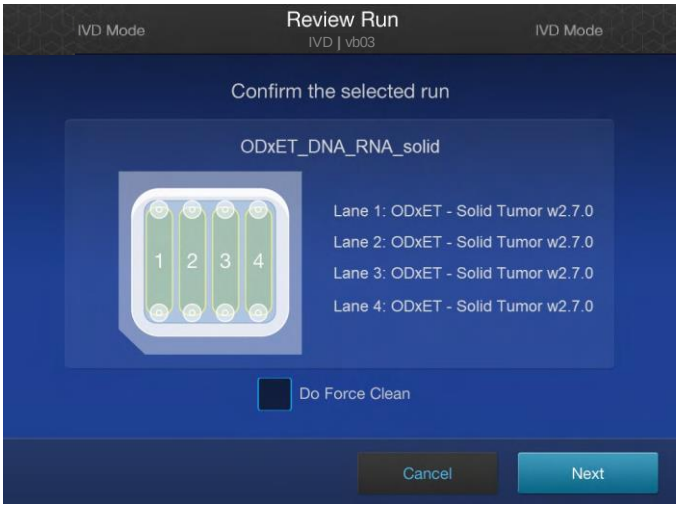



---

Note: If you select a run that requires more lanes than are available on a currently installed chip, a dialog appears giving you the option to install a new chip, or cancel. If you proceed with a new chip, a post-chip clean needs to be performed to remove the installed chip, then the sequencer prompts you to perform the Clear Deck, UV Clean, Load Deck, Clear Sequencing Reagents, and Load Sequencing Reagents steps.

---

3. In the **Review Run** screen, confirm the run and assay selections, then tap **Next**.




The deck door opens automatically.

---

Note:

- If the instrument vision system detects consumables loaded on the deck, the sequencer prompts you to remove the consumables, then starts a UV Clean.
- If all 4 lanes of the installed chip were previously used, the used chip shuttles to the install position. You are prompted to remove it before the UV Clean starts.
- Select the **Do Force Clean** checkbox if there will be 1–3 unused lanes on the installed chip after the run, but you want to start the next run on a new chip after the current run. A force clean automatically cleans the instrument after the run, eliminating the need for an operator to manually perform the **Clean instrument** procedure between the completion of the current run and the next run. Selecting **Do Force Clean** makes all lanes of the chip and installed sequencing reagents unusable after the run.
- If all 4 lanes will be used, the sequencer automatically performs a clean at the end of the run, and you do not need to select the **Do Force Clean** option.

---

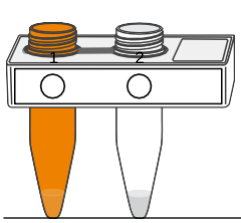
4. In the **Load Deck** screen, the sequencer instructs you step by step to load each required consumable and sample plate in a highlighted position on the deck. The sequencer detects the loading of each consumable in real time and advances to the next component automatically.




IMPORTANT!

- Remove the primer pool tube caps before installing the tube carrier on the deck.
- Load the correct type of barcode plate, library strip, or template strip for the type of run you are setting up. The sequencer displays a warning if you have installed consumables that are incompatible with the run you have selected. For example, a warning appears if an RUO-labelled barcode plate or library strip is loaded in an IVD-mode run.
- Remove the lids from all the boxes of Genexus™ Dx Pipette Tips before you load the boxes in the sequencer.
- After removing a tip box lid, visually inspect the tip box to ensure that all tips are seated level in the tip rack before and after placing on the instrument deck. If a bent or damaged tip is suspected, replace the tip box with a new tip box. Reracking tips from partially used boxes is not supported and can cause some tips to seat at an angle, which can affect run performance.
- Ensure that all tip boxes sit level on the deck by visually inspecting all boxes across the tip station. Reseat the boxes before starting the run, if needed.
- Do not use third-party pipette tips or repackage third-party tips into Genexus™ Dx pipette tip boxes. Use of third-party tips can result in run failure.
- Load instrument plates, including PCR plate, barcode plate, enrichment plate, and sample plate, into position by pressing down firmly and evenly on all sides and corners of each plate.

Note: A primer pool tube carrier can only be installed with the position 1 tube in the back row of the Genexus™ Dx Primer Pool Tube station. Follow the guidance in the run setup guide for loading the primer pool tube carrier or carriers in the correct position and order in the station.



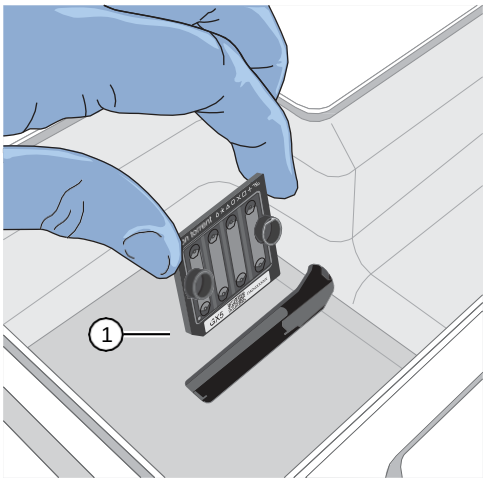

① Position 1
② Position 2

5. Ensure that all the primer pool tube carrier barcodes are visible and are accurately shown as loaded in the instrument software.

IMPORTANT! The instrument cannot detect if the Genexus™ Primer Pool Tube Holder is improperly placed. Ensure that the tube holder is correctly positioned before proceeding.

6. If prompted, insert a new Genexus™ Dx GX5™ Chip and Genexus™ Coupler. Insert the chip into the chip install slot with the chip notch oriented down and toward the front of the instrument.

IMPORTANT! Insert only a new chip and coupler when prompted. Do not insert a partially used chip.






① Notched corner of chip

Note: A chip shuttle under the deck moves the installed chip to loading and sequencing positions during the run.

IMPORTANT! Insert the Genexus™ Coupler so that it is level to help ensure it will properly align with the Genexus™ Dx GX5™ Chip. A coupler that is installed at an angle or is not level will not align properly to the chip and can result in a failed run.

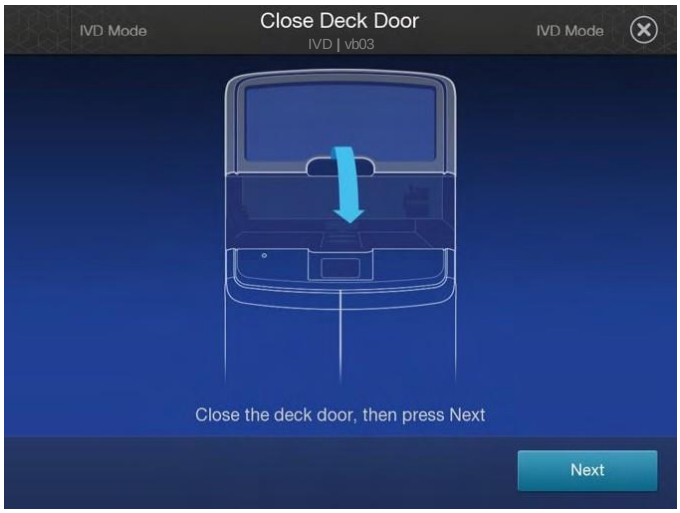
7. When the deck consumables and sample plate have been loaded, lock the library and templating strips in place by sliding the latches toward the rear of the deck.

Note: Load the sample plate last.




If a chip is detected and the strip latches are closed, the **Close Deck Door** screen appears.

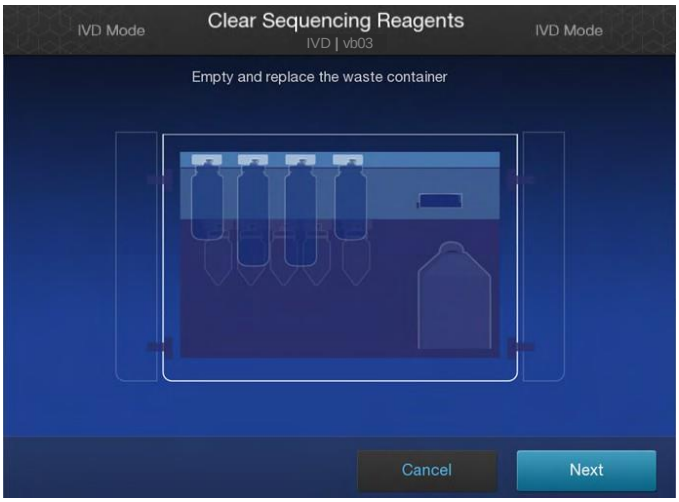
8. Close the deck door, then tap **Next**.




- If you installed a new chip in the sequencer, the sequencer prompts you to open the sequencing reagents bay doors to empty the waste and remove used sequencing reagents bay consumables. Proceed to step 9.
- If you are using a chip that was previously installed and has sufficient lane capacity for the run, the sequencer prompts you to start the run.

IMPORTANT! The cartridge and bottles in the sequencing reagents bay must be replaced every time that a new chip is installed, regardless of how many lanes were used in the previous chip.

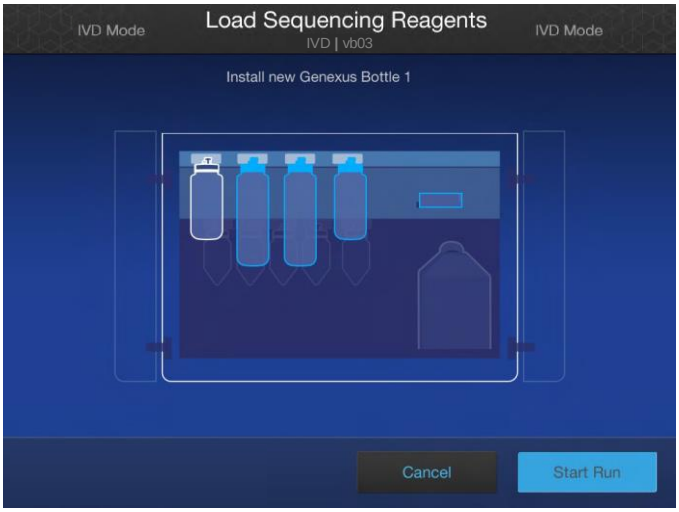
9. Follow on-screen instructions to empty the waste in the Waste carboy, remove waste pipette tips, remove the used Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2, Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge, then tap **Next**.




IMPORTANT!

- Empty and replace the Waste carboy and the waste pipette tip bin.
- After replacing the emptied Waste carboy, ensure that you reinsert the waste tube into the carboy.
- Follow all applicable local, state/provincial, and/or national regulations when recycling or disposing of consumables and liquid waste.

10. Install a new Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2 (two required), Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge.

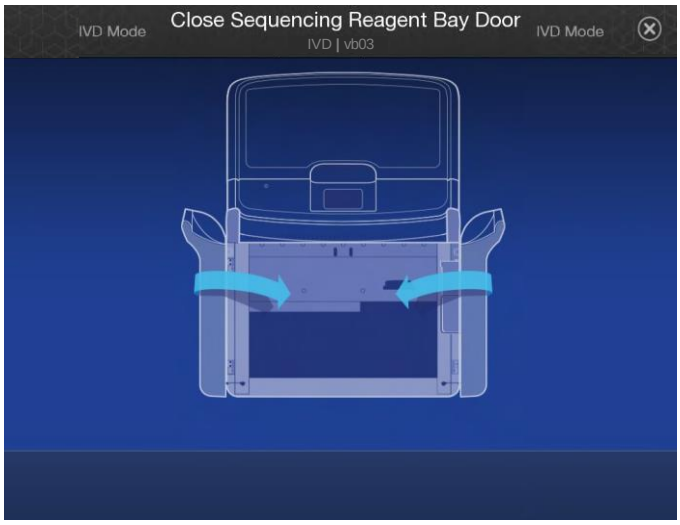


IMPORTANT!

- Before installing, *gently* invert each capped Genexus™ Dx Bottle 2 five times to mix—avoid vigorous mixing. Inspect the plastic nozzles for any pinches or deformations. To avoid pinching or folding of the nozzle, install the bottles straight-on, not at an angle.
- The installed reagents can be used for up to 14 days on the sequencer. The installed reagents on the sequencer should be used within 14 days of installation to avoid run failure, although the sequencer permits use for up to 28 days

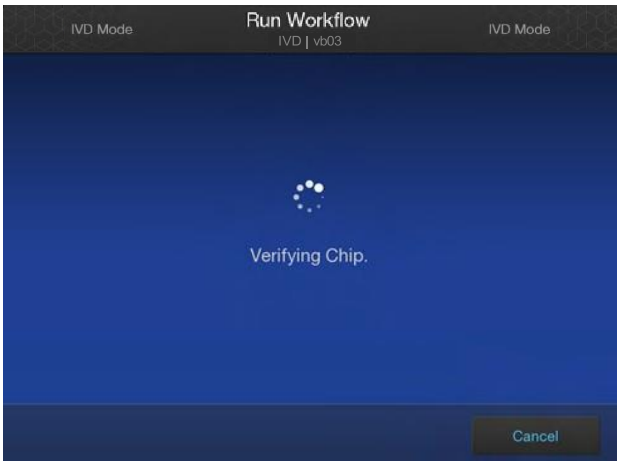
After reagents have been installed, the **Close Sequencing Reagent Bay Door** screen appears.

11. Close the sequencing reagents bay doors.




After the doors are closed, the sequencer automatically starts the run.

At the beginning of the run, the instrument verifies the chip, checks for leaks, then calculates run time.










A sequencing run encompasses the following stages:

1. Starting
2. Initializing
3. Library Prep
4. Templating
5. Presequencing
6. Sequencing
7. Cleaning

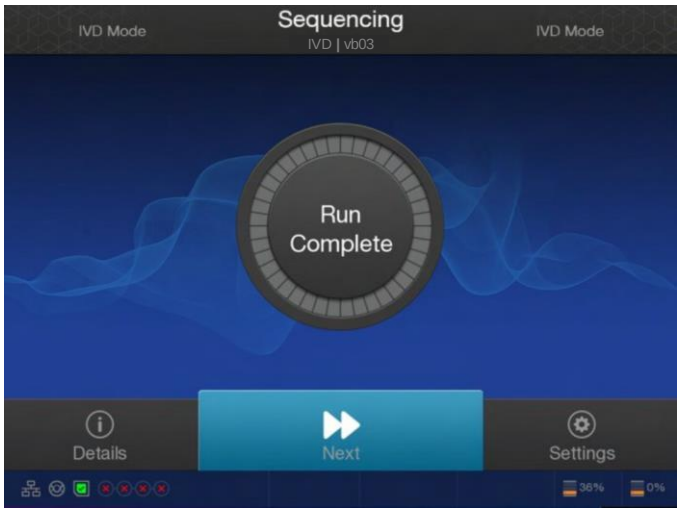
At each stage, the instrument shows the time remaining on the touchscreen.

---

Note:

- The time remaining shown on the screen does not include run analysis time.
- The leak check takes approximatively 22 minutes. Ensure that the leak check has completed and the run has started before leaving the laboratory.

---




When the run finishes, the sequencer displays the **Run Complete** screen.

Note: If all the lanes of a chip are used in a run, the used chip remains engaged in the sequencer until you start a new run. The chip then shuttles to the install position, and you are prompted to remove it before loading the deck with new consumables.

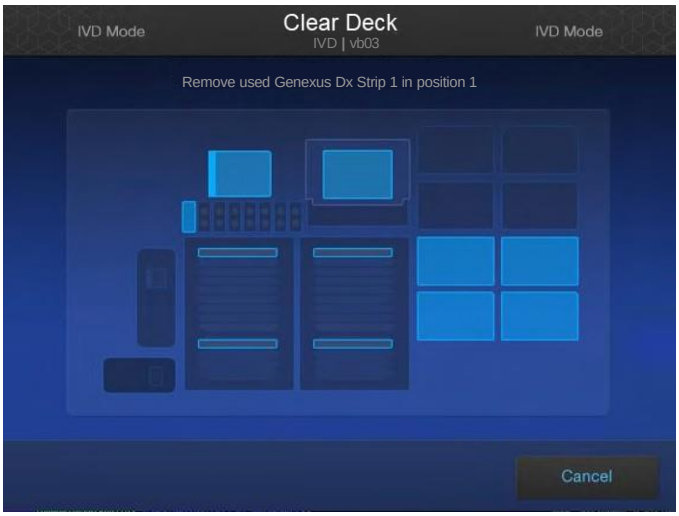
# Clear the instrument deck and perform a UV Clean

After a run completes, remove used consumables from the deck and perform a **UV Clean** to ready the instrument for the next run.

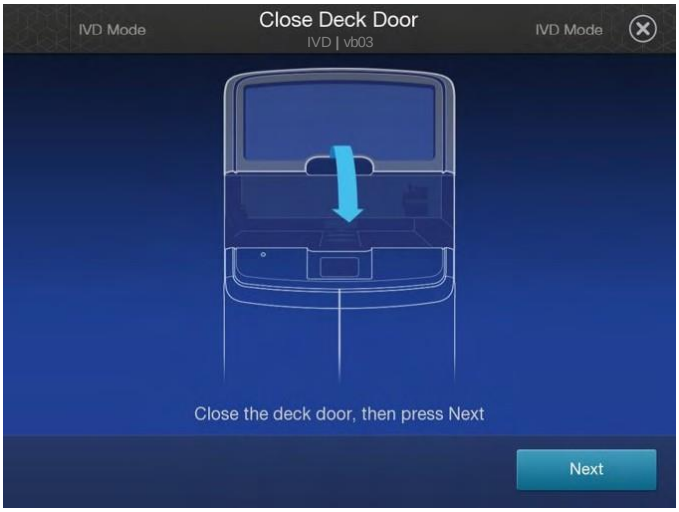
1. In the **Run Complete** screen, tap **Next** to start removal of used consumables.
   The deck door opens.

2. In the **Clear Deck** screen, the sequencer provides step-by-step instructions by highlighting the components to be removed. Unlock the library and templating strips by sliding the latches toward the front of the deck, then remove the used strips. Remove the remaining deck components specified by the sequencer.

   Note: Seal the PCR amplification plate with a foil seal before removing the plate from the PCR amplification station. Sealing the plate before removal helps prevent contamination of libraries if libraries are recovered for reuse. Store the sealed library plate at 4–8°C in a post-amplification area.



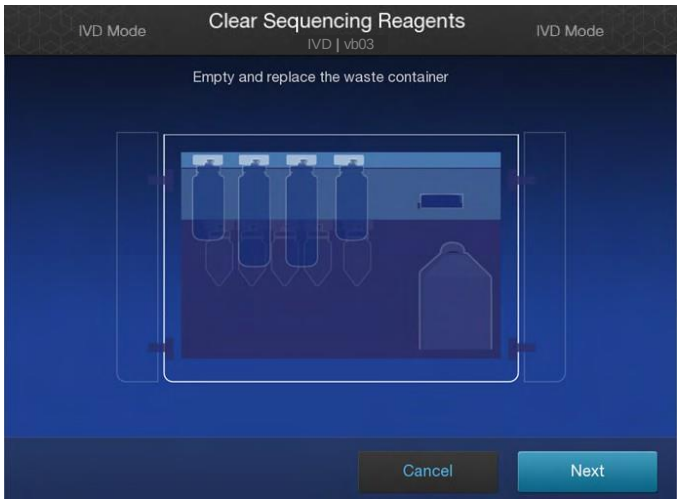

3. Inspect the Genexus™ Dx Filter in the liquid waste disposal port and verify that no standing liquid or debris is present.
   - If no standing liquid is found but debris is present, replace it with a new filter. For more information, see “Replace the Genexus™ Dx Filter” on page 135.
   - If standing liquid is present, contact Technical Support and report a possible filter or deck liquid waste line clog.
4. When finished, close the deck door, then tap **Next**.




A two-minute **UV Clean** starts.




5. After UV cleaning, if all the chip lanes were used, the sequencing reagents bay doors unlock. Open the doors, remove used components from the bay and empty the Waste carboy and waste pipette tip bin, then tap **Next**.



IMPORTANT! Do **not** discard or remove the conical bottles, unless alerted by the sequencer to replace the bottles after a conical bottle flow rate test. A conical bottle change may occur at the end of the run before the cleaning is performed and the instrument is ready for the next run. This process can add approximately 2 hours to when the sequencer can be loaded for the next run. For more information, see “Replace the Genexus™ Dx Conical Bottles” on page 135.

IMPORTANT! Follow all applicable local, state/provincial, and/or national regulations when recycling or disposing of Genexus™ Integrated Sequencer consumables and liquid waste.

CAUTION! The Genexus™ Dx Bottle 1 (small waste bottle) contains small amounts of formamide. Dispose of this waste appropriately.

6. After removal of used components, close the sequencing reagents bay doors, then tap **Next**.
   The sequencer returns to the home screen.

# Options for an expired sequencer initialization

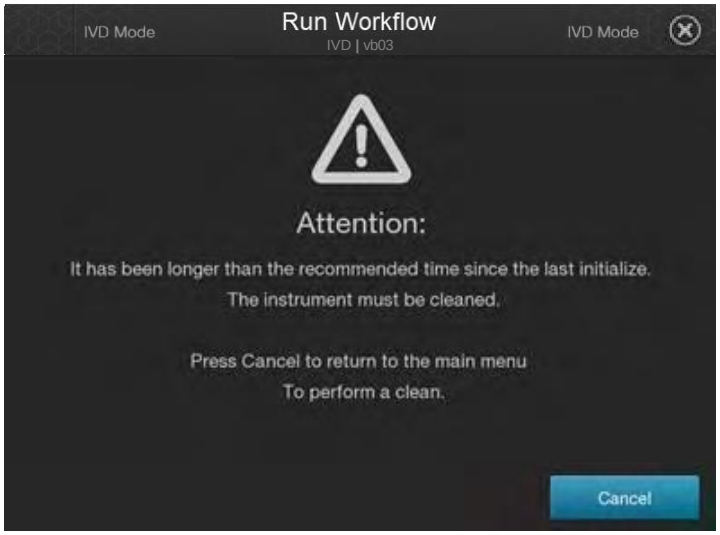
A sequencer initialization is defined by the installation of a new chip, chip coupler, sequencing bottles, and reagent cartridge on the sequencer before a run. Reagents are stable on the sequencer for 14 days, after which you can experience reduced performance. After 14 days, the following on-instrument warning appears:




We recommend that you not bypass the warning by tapping **Ignore**, but instead tap **Cancel** to proceed with an instrument clean. After tapping **Cancel**, select **Settings►Clean instrument** to clean the instrument before starting a run with a new chip. Lane assignment for a new run starts with lane 1.

IMPORTANT! Reagents remain stable on the sequencer for up to 14 days. After this period, performance can decrease, potentially leading to run failure. We recommend that you not bypass the warning by tapping **Ignore**, but instead tap **Cancel** to proceed with an instrument clean.

After 28 days, you can no longer proceed with an expired initialization. The following message appears:



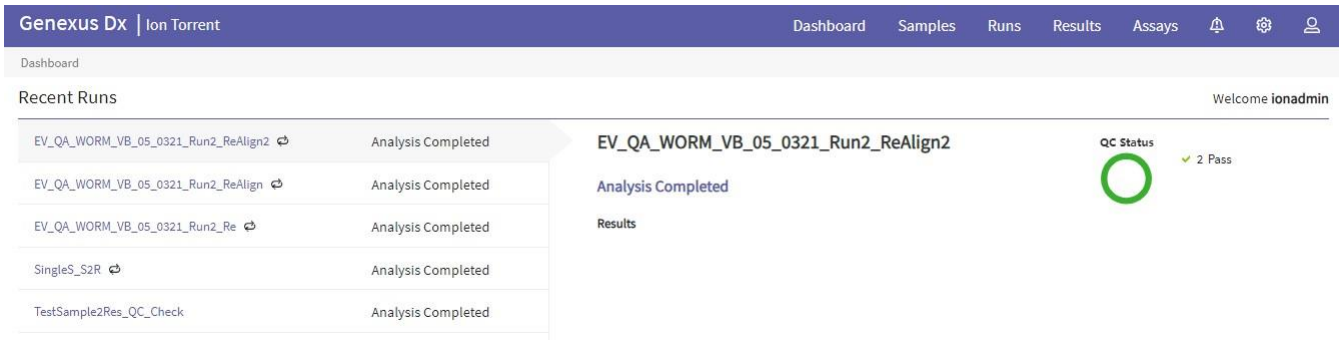

Tap **Cancel** to return to the main menu to perform an instrument clean.

# 8 Monitor the run

## View run progress on the instrument

You can view details for the five most recent and current runs in the **Dashboard** in Genexus™ Dx Software.

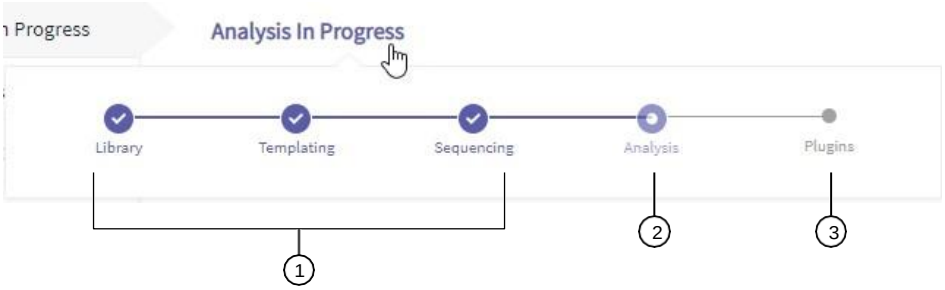
Runs that are reanalyzed are listed with ⟲ **(Reanalysis)** after the run name. For information on reanalyzing runs, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system.




1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

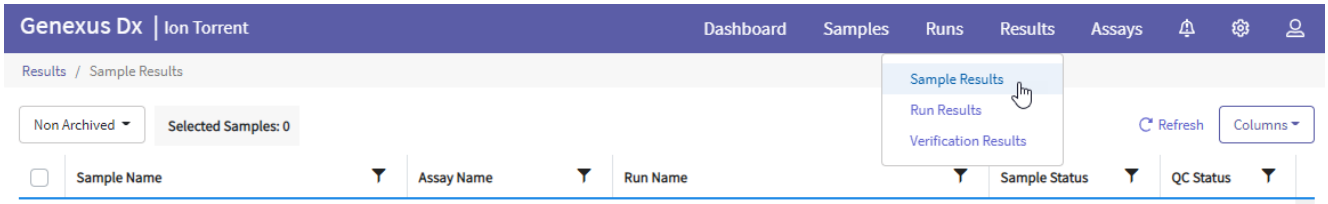
| View option | Description |
|---|---|
| View detailed run result information for completed runs. | In the left side of the screen, click the run name of interest.<br>The Run Summary tab opens. For more information, see “The run summary” on page 88.<br>In the menu bar, click Dashboard to return to the Dashboard. |
| View the quality control information. | In the left side of the screen, click the row of the run of interest.<br>The QC status for the run and for the samples is shown in the details pane in the right side of the screen. For more information, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074) or the software help system. |
| View assay-level metrics and results for the run. | 1. In the left side of the screen, click the row of the run of interest.<br>2. In the run details pane in the right side of the screen, click the assay name of interest.<br>The Assay Metrics tab opens. For more information, see “Assay metrics and the run report” on page 90.<br>In the menu bar, click Dashboard to return to the Dashboard. |

*(continued)*

| View option | Description |
|---|---|
| View the sample results. | **1.** In the left side of the screen, click the row of the run of interest.<br>**2.** In the run details pane in the right side of the screen, click the sample name of interest.<br>The Key Findings tab opens. For more information, see "Key Findings" on page 78.<br>In the menu bar, click Dashboard to return to the Dashboard. |
| View the progress of the run. | **1.** In the left side of the screen, click the row of the run of interest.<br>**2.** In the run details pane in the right side of the screen, place the pointer over the status for the run.<br>A progress bar for the run is shown. Steps in the run that are complete are listed with a check mark. Steps that are in progress are shown with a flashing circle. Steps that remain to be completed are listed in grey. If any failures occur, failed steps are also listed in grey.<br>  <br>① Completed steps are shown with a check mark. In this example, Library preparation, Templating, and Sequencing are complete.<br>② Steps that are in progress are shown with a flashing circle. In this example, Analysis is in progress.<br>③ Steps that remain to be completed are shown in grey. In this example, the Plugins step remains to be started after Analysis is complete. |

# 9 Review data and results


- Results navigation bar overview ..... 74
- Review sample results ..... 76
- Review run results ..... 86
- Variant report ..... 94
- Sign off variant reports ..... 98
- Reanalyze a run or sample ..... 100


Use the **Results** menu to review results and data analysis, and to perform data management tasks. You can view results sorted by sample or by run.

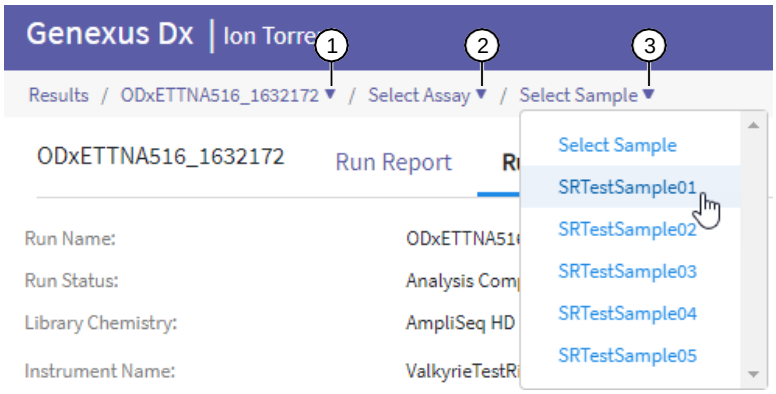
| Selection | Description |
|---|---|
| Click Results▶Sample Results | Select this option to review completed sample results and reports. |
| Click Results▶Run Results | Select this option to review completed run results and reports by assay. |
| Click Results▶Verification Results | Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification. |

## Results navigation bar overview

After you select a run result or a sample result, a results navigation bar appears and allows you to easily toggle between different results screens for each run. You can quickly switch between run-level results, assay-level results, and sample-level results for the run. In addition, you can easily find samples or assays in the run with the search field that appears in each dropdown list.

Assay-level results include assay metrics, such as final read data.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.

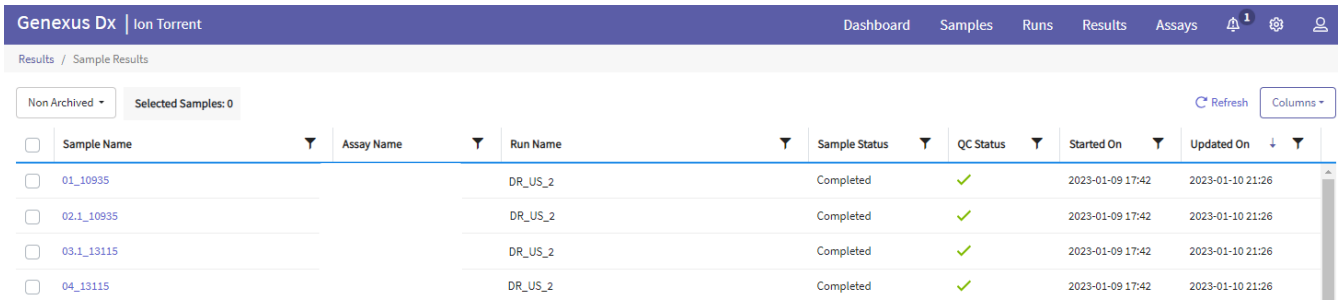



| Callout | Dropdown menu | Description |
|---|---|---|
| 1 | Run name | The run name is listed. Multiple runs are listed only if the run has been reanalyzed. |
| 2 | Select Assay | You can click the assay name of interest to view assay metrics for the run.<br>You can search for or toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed.<br>You can select Select Assay from the dropdown menu to remove the assay selection. If you remove the assay and sample selections, the Run Summary tab opens with a summary of results for the run selected in the Run Results dropdown menu. Assay metrics are also shown in the Run Report tab. For more information, see “View assay metrics and the run report” on page 90. |
| 3 | Select Sample | You can click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in Select Assay. The search field allows you to find samples in the run.<br>You can select Select Sample from the dropdown menu to remove the sample selection. If you remove the sample selection, the Assay metrics tab for the selected assay opens. If you select Select Assay with Select Sample shown, the Run Summary tab opens. Assay metrics are also shown in the Run Report tab. For more information, see “View assay metrics and the run report” on page 90. |

# Review sample results

In the **Results / Sample Results** screen, samples that have been sequenced are listed by sample name.

You can filter the list of results by clicking **(Filter)** in a column of interest, then entering a full or partial sample, assay, or run name.




The following information appears in the **Sample Results** screen. You can add columns to the Sample Results table to view more information. For example, you can add a column for **Electronic Signatures** to view signature and lock information for variant reports. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Results screen for the sample. |
| Assay Name | The name of the assay used in the run plan. |
| Run Name | The unique name of the run given when it was created in the software. |
| Sample Status | The status of the run or sample (for example: Completed, Running, Failed, Terminated, Pending, Stalled). |
| QC Status | The QC status of a completed run.<br>• ✔ (Passed) indicates the sample passed all QC metrics.<br>• ✘ (Failed) indicates the sample failed a QC metric.<br>• — (Not Calculated) indicates a sample did not undergo QC analysis. |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |
| Tags | Tags that are assigned to the sample are listed. |

You can also perform the following actions in the **Sample Results** screen.

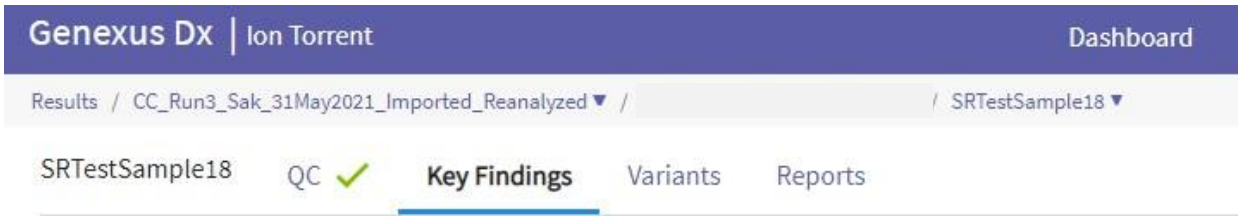
| Option | Description |
|---|---|
| Actions | Available action links for a sample are shown when you place the pointer over the row of a sample.<br>• Report—Download the report (available only for samples with a sample status of completed).<br>• Assign Tags—Add or remove the tags assigned to a sample result.<br>• Audit—View the audit trail for the run.<br>• Notes—View, add, or delete notes for sample result.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>• Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. |
| Actions that you can perform when you select samples | Assign Tags—Assign a tag or tags to selected sample results in one action. The Assign Tags command appears above the Sample Results table after you select one or more samples.<br>Note: For more information on using the Assign Tags feature, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system. |

## View key findings

You can view the **Key Findings** for a sample starting from either sample results or run results.

In the menu bar:

- Click **Results►Sample Results**, then click a sample name.
- Click **Results►Run Results**, then click a run name to open the **Results / Run Results** screen. In the **Run Name** column, click a run name to open the **Results** screen, then select a sample from the **Select Sample** dropdown list.



## Key Findings

The **Key Findings** table shows details about the sample, a list of biomarkers reported for the sample, and a summary of key metrics for genes assayed in the run.

Note: The **Key Findings** table is by default the first view that is shown for sample results.

The information that is displayed depends on the assay that was used in the run.

Sample Details

| Sample Detail | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Collection Date | The date that the sample was collected. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Sample Type | A term that describes the sample, for example, FFPE, DNA, DNA & RNA |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. "Percent Cellularity" refers to the same measurement as "Percent Tumor Content." |

Biomarker Results

| Result category | Description |
|---|---|
| Variants Detected | Variants detected by the assay. |

Key Metrics

| Metric | Description |
|---|---|
| Amplicon Summary | |
| Average Base Coverage Depth | The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage. |
| Uniformity Of Base Coverage | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths. |

Key Metrics *(continued)*

| Metric | Description |
|---|---|
| Percent Reads On Target | The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified. |
| Uniformity Of Amplicon Coverage | The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts. |

## Review the variant pileup view

You can review variant tracks with the pileup view in Genexus™ Dx Software. The pileup view is a figure that the software generates to show each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup view can be used to identify potential variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

The pileup view is shown below the variants table.

### Change the view of the variant pileup

You can zoom and pan the variant pileup view and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 80.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

| Action | Procedure |
|---|---|
| Track the position as you place the pointer over the variant pileup. | Click **Cursor Guide**. |
| Mark the center of the variant pileup. | Click **Center Line**. |
| Remove the track labels. | Click **Track Labels**. |
| Zoom in to increase the magnification of the image. | Click ⊕ **(Zoom in)** one or more times, or double-click the image. |
| Zoom out to decrease the magnification of the image. | Click ⊖ **(Zoom out)** one or more times. |
| Pan across the image to move the view to another location on the screen with its current magnification. | Click and drag the pointer over the image. |

## Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Dx Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

---

Note: For best results, click the **Expand** option to view details about variants and base calls that are shown in each read coverage track

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1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to a track, then select an option to adjust the view of the track.

| Option | Description |
|---|---|
| **Expand** | Select this option to view the tracks with the maximum visible height. |
| **Squish** | Select this option to view the tracks with the minimal height for each track. |
| **Collapse** | Select this option to view the tracks with overlapping transcripts shown along a single line. When **Collapse** is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown. |

## Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Dx Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the reference track.

| Option | Description |
|---|---|
| View the reverse strand | Click **Reverse**.<br>This option is available only when the forward strand is shown. |
| View the forward stand | Click **Forward**.<br>This option is available only when the reverse strand is shown. |
| View the reference sequence translation | Click **Three-Frame Translate**.<br>This option is available only when the amino acid sequence is not shown. |
| Hide the reference sequence translation | Click **Close Translation**.<br>This option is available only when the amino acid sequence is shown. |

## Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Dx Software to dynamically customize the view of the data.

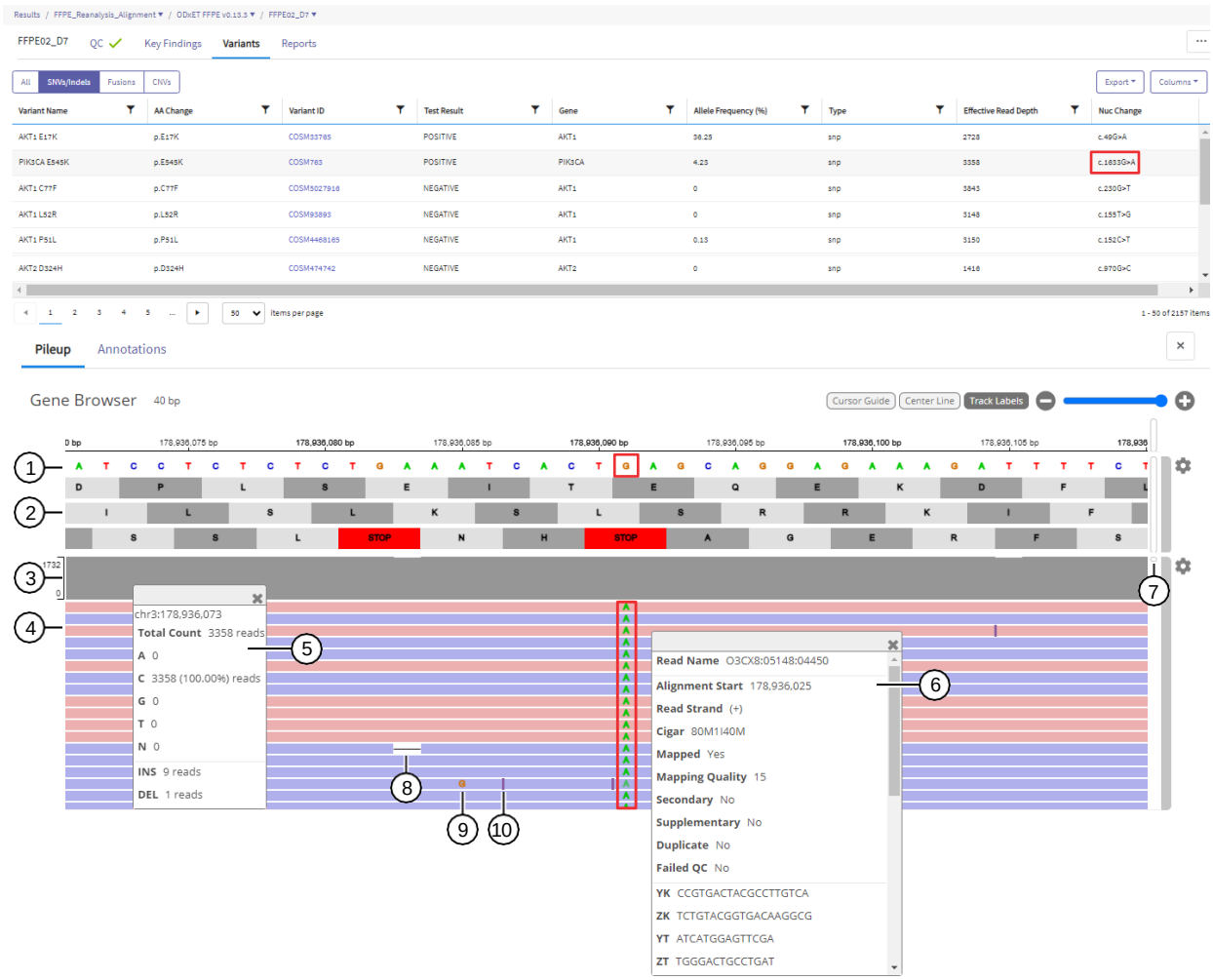
1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the BAM tracks.

| Option | Description |
|---|---|
| View all BAM tracks in monochrome gray | In **Color By**, deselect the checkmark next to **Read Strand**.<br>This option is available only when the BAM tracks are colored by read strand. |
| Distinguish between the direction of the BAM tracks with color | In **Color By**, click **Read Strand**.<br>This option is available only when the BAM tracks are not colored by read strand. |
| Expand, squish or collapse the BAM tracks | For more information, see "Adjust pileup tracks" on page 80. |
| Show all the bases in the BAM tracks | Click **Show all bases**.<br>This option is available only when all bases in the BAM tracks are not shown. |
| Change the minimum threshold for the visibility of BAM tracks | a. Click **Set visibility window**.<br>b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks.<br>When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see "Change the view of the variant pileup" on page 79. You can adjust the visibility window according to the scale shown in the screen. |

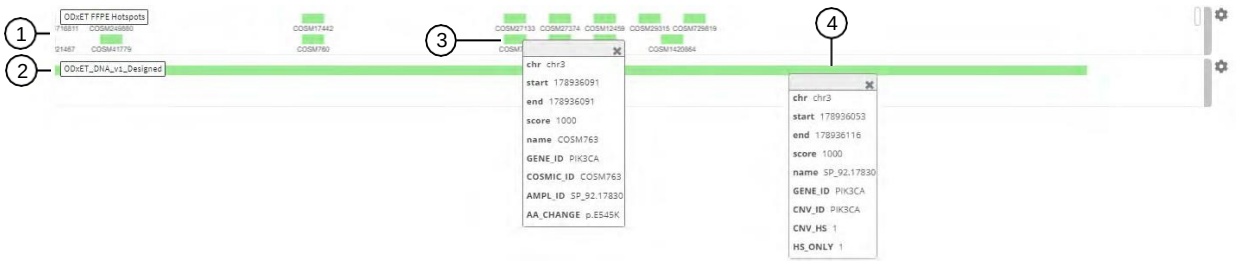
## Examples of a variant pileup view

The following figure is an example of a variant pileup view, shown in the **Gene Browser** in the **Variant** tab. The variant pileup view graphically represents the sequencing tracks, the reference track, and the annotation tracks, and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant are indicated in the image, both in the variant table and the in the pileup view. You can customize your pileup view. Reorder the tracks displayed by clicking a gray bar on the right edge of a track, then dragging the track to a different location. You can also click-drag the cursor at any position in the image to move the image left or right in the screen. For more information, see "Adjust pileup tracks" on page 80.




1. The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
2. The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 81.
3. The coverage track.
4. The BAM track.
5. Click the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
6. Click a BAM track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
7. Use the slider to view more tracks.
8. Deletion.
9. Alternate base in the BAM read coverage track.
10. Insertion.

The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.




① The hotspot track.
② The target region track.
③ Click a hotspot track for additional details, such as the start and end positions.
④ Click the target region track for additional details, such as the start and end positions.

## Coverage histogram

When you zoom in on the variant pileup view, you see the coverage histogram and reads from the .bam track. The histogram in gray shows read depth at that location and reads from the .bam track show read direction and location of variants.




① Read depth
② Read location and direction of variants

## View the variants from an assay run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. Review detailed variant data and read coverage tracks.




**Example visualization**

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate a family.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" indicates insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

| Option | Description |
|---|---|
| View the read coverage tracks sorted by family | Click ⚙ **(Actions)** next to the read coverage track, then select **Color►Molecule**. |
| Adjust the BAM tracks | Click ⚙ **(Actions)** next to the read coverage track, then select an option to adjust the view of the track. For more information, see “Adjust pileup tracks” on page 80. |
| Review detailed data about a BAM read | Single-click the read track to obtain information such as the mapping quality, the strand, and the read base. |
| Review distribution of base calls at a selected position | Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions. |

## View the QC results

Any user can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample Name** column, click the sample name of interest.

3. Click the **QC** tab.
   The QC metrics are shown.

4. Review the QC metrics.
   For more information, see the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074), or the software help system .

# Review run results

In the **Results / Run Results** screen, runs that are pending, in progress, or complete are listed. Runs with a status of failed, aborted, or stalled are also listed.

You can filter the list of run results by clicking **(Filter)** in a column of interest, then entering a full or partial run or assay name, or other applicable filter term.

The following run information appears in the **Results / Run Results** screen.

| Column | Description |
|---|---|
| Run Name | The unique name of the run given when it was created in the software. Click a run name to open the Run Summary.<br>Runs that are reanalyzed are listed with (Reanalysis) after the run name. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074). |
| Assay Name | The name of the assay selected for the run. You can view the Assay Name and corresponding Assay Full Name for all assays in the Assays►Manage Assays screen. |
| Run Status | The status of the run. For example, Analysis In Progress, Executing Plugins, Analysis Completed, Terminated, Archival: In Progress. |
| Total Samples | The total number of samples in a run. |
| PCR Plate Number | A unique identifier for the 96-well plate used for library preparation and templating. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074). |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |

You can also perform the following actions in the **Results / Run Results** screen.

| Option | Description |
|---|---|
| Actions | Available action links for a run are shown when you place the pointer over the row of a run. The actions that are available depend on the type of run.<br>• Delete—Delete the run.<br>• Audit—View the audit trail for the run.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>• Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. |
| Actions | Available action links for a run are shown when you select the checkbox to the left of the Run Name.<br>• Clear—Deselect the selected run or runs.<br>• Retain Data—Click to keep run data regardless of disk cleanup settings. To undo, select Release Data.<br>• Archive Data—Click to archive run data. Select run data options (Sequencing Output, Intermediate Files) in the Archive Confirmation dialog box. Analysis Results are selected by default and are not optional.<br>• Delete—Delete the run data of the selected run or runs. |

## View the run summary

The run summary provides an overview of the run. The information that is displayed includes a list of the assays used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Name** column, click the run name of interest.
   The **Run Summary** tab opens.
3. Review the run summary.

| Action | Procedure |
|---|---|
| View the assay metrics. | In the Assays section, click the assay name of interest. |
| View the sample locations in an image of a 96-well sample plate. | In the Sample Locations section, click PCR Plate View. |
| Reanalyze a run with a new assay. | In the upper right corner of the screen, click ••• (More Options)►Reanalyze.<br>For more information, see the software help system or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074). |
| Download customer support archive (CSA) log files for the run to help with troubleshooting. | In the upper right corner of the screen, click ••• (More Options)►CSA. |

*(continued)*

| Action | Procedure |
|---|---|
| View the history of variant classifications. | In the upper right corner of the screen, click ••• (More Options)►Variant Audit.<br>The information is available only for results that include variant classifications. |
| View the run report. | Click the Run Report tab. |

## The run summary

The run summary displays run information, assays used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

| Item | Description |
|---|---|
| Run Name | The name of the run. |
| Started On | The date and time when the run was started. |
| Completed On | The date and time when the run was completed. |
| Run Status | The status of the run. |
| Report Template | If the option to generate a report was enabled in the run plan, the report template used is listed. |
| Chip Type | The semiconductor sequencing chip used in the run, such as the Genexus™ Dx GX5™ Chip. |
| Library Chemistry | The type of library chemistry used in the run, such as Ion AmpliSeq™ HD. |

Sequencing instrument information

| Item | Description |
|---|---|
| Instrument Name | The name of the sequencer. |
| Instrument Serial Number | The serial number of the sequencer. |
| Status | The status of the sequencing portion of the run. |
| Operator | The name of the person who was signed in the Genexus™ Dx Integrated Sequencer when the sequencing run was performed. |
| Start Date | The date and time when the run was started on the sequencer. |
| Completion Date | The date and time when the run was completed on the sequencer. |
| Sequencing Flows | The number of flows performed by the sequencer. |

Assays table

| Item | Description |
|---|---|
| Assay Full Name | The name of the assay used in the run. Click the assay name to display the details of the assay. |
| Assay Name | An abbreviated name of the assay or assays used in the run. |
| Analysis Version | The version of the assay used for analysis. |
| Application Type | The application type for the assay, such as DNA and Fusions. |
| Lane | The chip lane or lanes used in the sequencing run for the assay. |
| Total Samples | The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample. |

Sample Locations table

| Item | Description |
|---|---|
| PCR Plate View | Click to view the sample locations in an image of a 96-well sample plate. |
| Well Pos. | The well position indicates the location of the sample on the plate. |
| Library Batch ID | The unique identifier created for the library batch. This information is available only for Library to Result runs. |
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, RNA, or DNA+RNA. |
| Vol. (µl) | The volume of the sample. |
| Conc. (ng/µl) | The concentration of the sample. |
| Dilution Factor | The dilution factor of the sample. |
| Kit Barcodes | The barcode for the kit used for nucleic acid extraction, if applicable. |
| Barcode | The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101. |
| Assay Name | The assay that was used to sequence the sample for the indicated well position. |
| Library Position | The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run. |

Reagents table

| Item | Description |
|---|---|
| Consumable | Consumables used in the run, such as the PanelKit and LibraryKit, are listed. |
| Barcode | The consumable barcode, if applicable. |
| Part # | The consumable part number, if applicable. |
| Expiration Date | The consumable expiration date, if applicable. |
| Lot # | The consumable lot number, if applicable. |

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

## View assay metrics and the run report

The run report provides detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run.

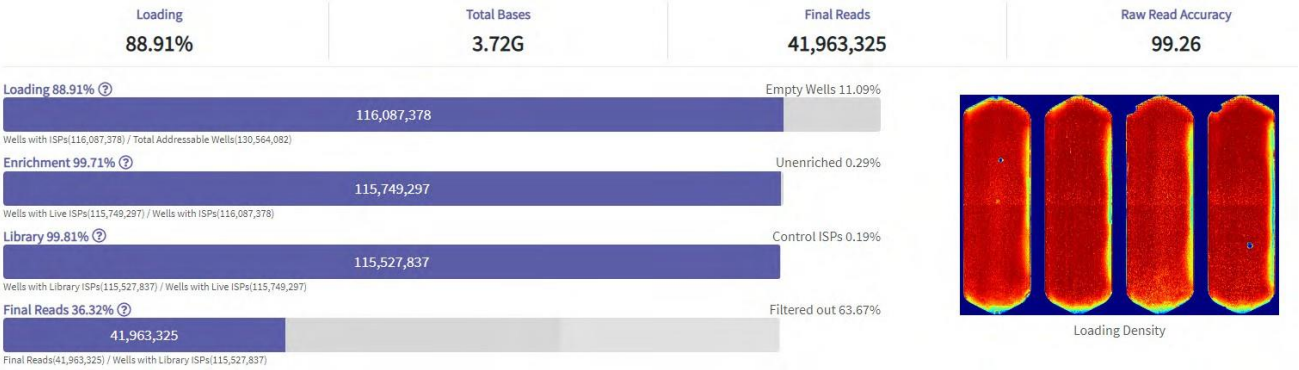
For runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

To view the **Run Report**, in the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest. In the **Run Summary** screen, click the **Run Report** tab.

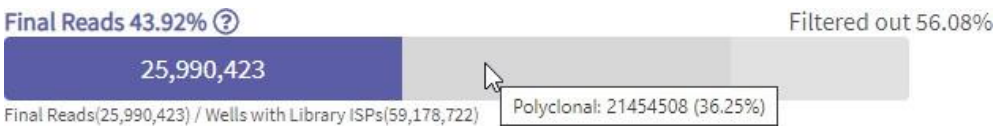
The **Run Report** is assay-specific and cannot be viewed within the sample results screens. To view the **Run Report**, ensure that **Select Sample** is selected in the **Select Sample** dropdown menu.

### Assay metrics and the run report

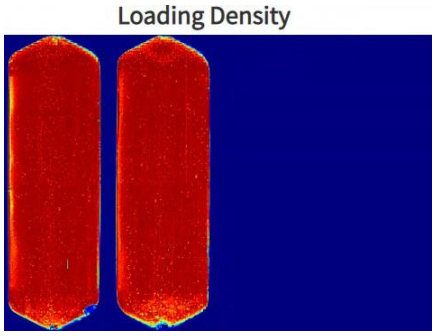
The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.



Run Report

| Metric | Description |
|---|---|
| Loading | The number and percentage of total addressable wells on the chip that contain an ISP. |
| Enrichment | The number and percentage of wells ISPs that contain live ISPs. |
| Library | The number and percentage of wells with live ISPs that contain Library ISPs. |
| Final Reads | Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement. |
| Total Bases | The number of filtered and trimmed base pairs that are reported in the output BAM file. |
| Raw Read Accuracy | The raw read accuracy across each individual base position in a read calculated as, (1−[total errors in the sequenced reads]/[total bases sequenced]) × 100. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is *not* based on consensus accuracy across multiple reads for the same base position. |
| Wells with ISPs | The number of wells that contain an ISP. |
| Unenriched | The number and percentage of wells with ISPs that do not contain live ISPs. |
| Total Addressable Wells | Wells on the chip that can be physically reached by a library. |
| Empty Wells | The percentage of total addressable wells on the chip that do not contain an ISP. |
| Wells with Live ISPs | Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key. |
| Wells with Library ISPs | Loaded wells with live ISPs with a key signal that is identical to the library key signal. |
| Control ISPs | Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal. |
| Filtered out | The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads. |
| Polyclonal | Wells with a live ISP that carries clones from two or more templates.<br>To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot.<br>  |

Run Report *(continued)*

| Metric | Description |
|---|---|
| Low Quality | Loaded wells with a low or unrecognizable signal.<br>To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot.<br>  |
| Adapter Dimer | Loaded wells with a library template of an insert size less than 8 bases.<br>Adapter dimer reads represent a small proportion of total reads and can be seen by hovering the pointer over the right end of the Final Reads bar.<br>  |
| Loading Density | A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs.<br>  |

## Run Samples

The Run Samples table lists read data for each individual sample in the assay.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered in the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, RNA, or DNA+RNA. |
| Barcode | The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample. |
| Total Reads | The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. |
| Mean Read Length | The average length, in base pairs, of usable library reads for each sample. |
| ≥Q20 Bases | The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample. |

Run Samples *(continued)*

| Column | Description |
|---|---|
| Mapped Reads | The number of reads that are mapped to the reference file. |
| On Target Reads | The percentage of sequencing reads mapped to any target region of the reference. |
| Mean Depth | The average number of reads of all targeted reference bases. |
| Uniformity | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. |
| Read Length Histogram | A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).   |

### Download a run report

You can download a run report summary in PDF format. The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “View assay metrics and the run report” on page 90. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
   Multiple panes including a **Run Report** pane, a **Variant Report** pane, and any panes for customized reports that have been generated are shown.
4. In the **Run Report** pane, click **Download Report** to download a run report summary in PDF format.

## Assign PCR Plate

Genexus™ Dx Software lets you track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

Note: You should assign a PCR plate to a run if there are leftover libraries that you want to use at a later time to perform a Library to Result run. You can look up the PCR plate number on completed runs to get the library and barcode mapping, then enter the same Barcode information into the Library to Result run.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.

   The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

# Variant report

The variant report is a PDF report of the results for each sample in a sequencing run. You can use a system-installed report template to generate a variant report, or you can customize the layout and contents of a variant report.

To make a variant report available for each sample result upon completion of a sequencing run, enable **Generate Report** in the **Setup** step when you plan the run. For more information, see page 42). To generate a variant report after a run is complete, see “Generate a variant report” on page 97.

When a variant report has been generated for a sample result, it is available for download in three places:

- In the **Results / Sample Results** screen when you place the pointer over the row for that sample, then click the **Report** link to download the PDF.
- In the variant report pane for the sample results in the **Reports** tab when you click ⋮ **(More)**►**Download Report**.
- In the **Results** screen for the sample when you click ··· **(More Options)**►**Download Files**.

Variant reports can be electronically or manually signed by users. Electronic signatures are shown in the Electronically Signed By section of the report, if the section is included in the report template.

## Sections of a variant report

A variant report can contain the following sections and information, depending on the assay used.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the variant report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other optional fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered into the software. You can customize the format of the Sample Details section when you create a new report template. To create a new report template, click Assays►Manage Presets, then in the Report Templates tab, click + Add New. |
| Biomarker Results Summary | A summary of results for biomarkers detected in the sample. There can be multiple subsections that indicate biomarker information. |
| QC | • Run QC—A summary of the run quality control metrics<br>• NTC QC – DNA—A summary of the DNA no-template control quality control metrics<br>• NTC QC – RNA—A summary of the RNA no-template control quality control metrics<br>• Positive Control01 QC – DNA—A summary of the DNA positive control quality control metrics<br>• Positive Control01 QC – RNA—A summary of the RNA positive control quality control metrics<br>• Sample QC – RNA—A summary of RNA sample quality control metrics<br>• Sample QC – DNA—A summary of DNA sample quality control metrics<br>• Templating Control QC – CF-1—A summary of the CF-1 quality control metrics<br>This section is locked and cannot be customized. |
| Sequencing Run Details | Contains the following subsections:<br>• Assay Name—The assay used in the run<br>• Panel—The panel used in the run<br>• Started On—The date and time that the run was started<br>• Completed On—The date and time when the run was completed<br>• Operator—The user who sent the run to the instrument<br>• Instrument—The name of the instrument<br>• Serial Number—The serial number of the instrument<br>This section is locked and cannot be customized. |
| Reagents | Details about run consumables, including part numbers, expiration dates, and lot numbers.<br>This section is locked and cannot be customized. |
| Kit Barcodes | Details about each barcode for the kit that is used in the run, including sample name, assay name, nucleic acid type, extraction kits and control kits. |
| Key Metrics | Includes amplicon information for the results, such as the average base coverage depth, uniformity of base coverage, and percent reads on target. The options that are available to customize in this section depend on the assay that you select for the report template. |

*(continued)*

| Section | Description |
|---|---|
| Electronically Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |
| Footer | The footer is repeated on each page of the report. |
| Signature | A section for a physical signature of the printed report. |
| Custom Text | A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Variant details | Details about variants in the run. You can customize the title of this section and the columns that are included in the report. |

## Download a variant report

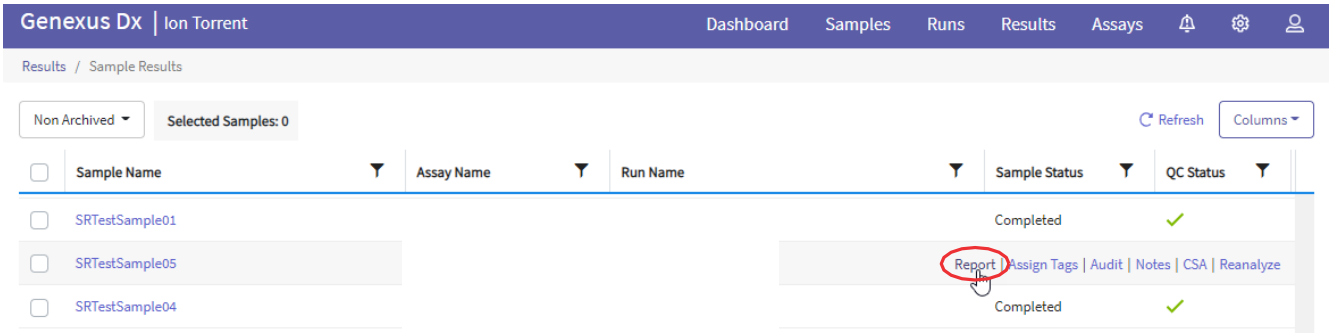
You can download a variant report for a sample result of interest from the **Results / Sample Results** screen, from the pane that shows the report, or as part of a download of results files.

1. In the menu bar, click **Results**►**Sample Results**.
2. Download the report with one of the following options.

| Option | Procedure |
|---|---|
| Download a ZIP file that contains all variant reports for a sample result. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click ••• **(More Options)**►**Download Files**. |
| Download the variant report in PDF format from the **Reports** tab for the sample results. | Click the **Reports** tab then, in the pane of the report of interest, click ⋮ **(More)**►**Download Report**. |
| Download the variant report as part of the results files for a specific sample in the **Results** screen. | For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*. |

A ZIP file that contains the PDF report is downloaded to the computer if you download reports from the **Sample Results** screen or as part of the results files. A report in PDF format downloads if you use the **Reports** tab for the download.

3. In the **Sample Results** screen, place the pointer over the row of the sample of interest, then click **Report**.




A ZIP file that contains the PDF report downloads automatically.

4. Extract the downloaded files, then open the PDF file in an appropriate viewer.

## Generate a variant report

You can generate a new variant report for sample results after a run is complete. A **(Lock)** in the variant report indicates that the electronic signature option for the report is locked. After a variant report is locked, the report cannot be electronically signed by any other user.

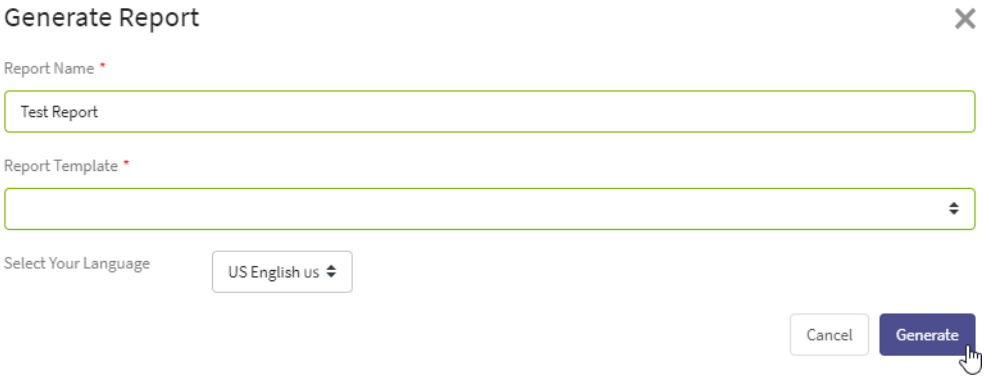
You can generate multiple reports for a sample result, if each report is named uniquely, and is generated in a different language, or uses a different report template.

When generating a customized report, you can update any report template selections. You can use this procedure to generate multiple reports for a sample, if a unique report name is entered for each report. For example, you may want to generate reports for different languages, or reports that use different templates.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Results / Sample Results** screen, click the sample of interest in the **Sample Name** column.
3. Select the **Reports** tab, then click **+ Generate Variant Report**.
4. In the **Generate Report** dialog box, change the name of the report that is generated by the software, if needed.
   a. Select the report template and language of the report.
   b. If the option to make custom text **Editable on Report Generation** was selected when the report template was created, enter a title in the **Custom Text** section and if needed, a description.

The **Report Template** list includes the report templates that are associated with the assay that was used in the run. For information about creating report templates, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*.

If you select the same report template that was used to generate a variant report, and have not locked that report, the new selections you make override the previous variant report.




5. Click **Generate**.
   A draft version of the report is added to the **Reports** tab.

A pane for the new report is added next to the Run Report pane in the **Reports** tab. Reports that have been generated are available for download in the **Reports** tab, in the **Sample Results** screen, and in the ZIP package that contains results files.

# Sign off variant reports

Manager, administrator, operator, and report-level users can electronically sign a variant report for sample results in Genexus™ Dx Software. A sample result can have multiple variant reports only if each report uses a different report template, and a different language.

---

IMPORTANT! If you use the same template and language to regenerate an existing report, the existing report will be overwritten.

---

Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign the report, you will be the last person to sign the report before it is locked.

The signature information appears in the variant report PDF file or a user-created report, if selected. For more information, see “Variant report” on page 94. For information about how to create a report template, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide* (Pub. No. MAN1000074).

Multilanguage support for PDF report generation is provided.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample result of interest in the **Sample Name** column.
3. In the **Reports** tab, in a variant report pane that is labeled as **Draft**, click **Sign Off**.
   The **Sign Off Report** dialog box opens and shows the name of the report, the template used for the report, and the following information, which is based on the profile of the user who signs the report.
   - **Electronic Signature**
   - **User Name**
   - **Language**
4. In **Password**, enter the password.
5. In **Sign Off Comments**, enter a comment.
6. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.
   If you entered footer information in the **Footer Field** when you created a report template, or if text was entered during a previous sign off, the same footer information is shown in the **Sign Off Report** dialog box. You can edit the text in the **Footer Field** to change the information that will be shown in the footer of the PDF report.
7. Click **Sign Off** to confirm the electronic signature.
   The pane for the signed variant report is updated to include the signature information for the variant report. If the electronic signature that is associated with a lock-level signature type is used, a **(Lock)** is shown in the variant report pane and the report is locked.
8. In the menu bar, click **Results►Sample Results** to return to the **Results / Sample Results** screen.
   If the **Electronic Signatures** column is shown in the **Sample Results** screen, the electronic signature types for users who signed the report are listed, and a **(Lock)** is shown for reports that are locked.
9. In the row of the sample of interest, click **Report** to download the report.

The report is signed. The **Electronic Signatures** section of the report pane is updated to include your username and the signature type for your account. You can hover over the signature type to show the list of signature types that are completed for the report. If multiple electronic signatures are added to the report, an electronic signature type for each signer is shown in the **Electronic Signatures** column of the sample results table. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*.

You cannot generate new reports from the variant results if a report from those results is signed off and locked. If you must change the report, reanalyze the sample and generate a report for the reanalyzed sample.

You can send notifications for report sign-offs to other users. For more information, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*.

# Reanalyze a run or sample

If a sequencing analysis fails, you can reanalyze a run or sample using the same assay. For information about how to reanalyze a run or sample, see the software help system, or the *Genexus™ Dx Software 6.8 User Guide*.

# A Troubleshooting


- Customer Support Archive (CSA) ..... 101
- Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls 102
- Genexus™ Dx Integrated Sequencer—general and QC troubleshooting..... 103
- Genexus™ Dx Integrated Sequencer error and warning messages ..... 107
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## Customer Support Archive (CSA)

You can download an archive that a Technical Support representative can use to diagnose Genexus™ Dx Software issues. The Customer Support Archive (CSA) contains log files and other technical data about Genexus™ Dx Software and other files from runs on a Genexus™ Dx Integrated Sequencer.

You can access customer support archive files from the **Results / Sample Results** and the **Results / Run Results** screens.

### Download a Customer Support Archive

When you download a Customer Support Archive (CSA) in Genexus™ Dx Software, files are included for all samples for a specific assay. If the run includes only one assay, files for all samples are included. If the run includes more than one assay, you can specify the assay of interest when you download the CSA.

1. In the menu bar, click **Results►Sample Results**.
2. In **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.

   Alternatively, you can download CSA files three other ways.

   - Click **Results►Sample Results**. Click the sample name of interest. Then, click ••• **(More Options)►CSA.**
   - Click **Results►Run Results**. Click the run name of interest. Place the pointer over the run of interest, then click **CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.
   - Click **Results►Run Results**. Click the run name of interest. Then, click ••• **(More Options)►CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to the folder that you specified to download files from the browser. This location depends on the browser settings. You can attach the archive to an email to send to Customer Support.

# Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls

You can use quality control results to troubleshoot Genexus™ Dx Integrated Sequencer runs to help identify the cause of performance problems. Inclusion of a set of six control amplicons (covering a range of amplicon length) and spike-in nucleic acid into sample library preparation reactions helps determine whether poor performance is due to insufficient sample input and/or poor sample quality, or is unrelated to sample input and quality. With 10 ng sample input, the read ratio of endogenous sample reads to spike-in control reads is expected to be ~3. Using more than 10 ng sample input results in a proportionally higher read ratio. For example, if you load 20 ng of sample, the read ratio should be ~6.

The CF-1 templating control serves as a check on templating and sequencing performance that is independent of library preparation.

Use the following table as a guide to help identify the source of performance problems. For recommended actions, see the troubleshooting topics under "Genexus™ Dx Integrated Sequencer—general and QC troubleshooting" on page 103.

| QC category | Run diagnostic | | | |
|---|---|---|---|---|
| | Successful run | Sample input and/or quality problem | Library preparation problem unrelated to sample | Templating or Sequencing problem |
| Sample QC (endogenous sample reads) | Passed | Failed | Failed | Failed |
| Read ratio for inline controls (endogenous to spike-in reads) | Normal<br>Read ratio ~3 | Low<br>Read ratio <<3 | Normal or variable | — |
| Templating Control QC - CF-1 | Passed | Passed | Passed | Failed |

# Genexus™ Dx Integrated Sequencer—general and QC troubleshooting

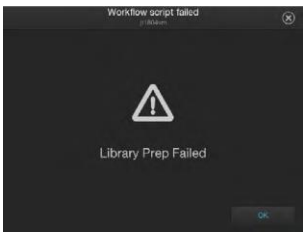
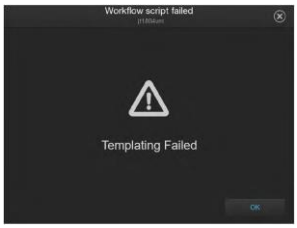
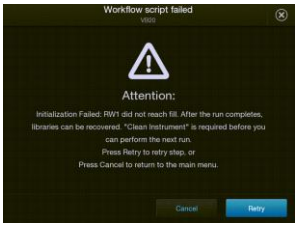
| Observation | Possible cause | Recommended action |
|---|---|---|
| A consumable is not recognized by the sequencer after loading on the deck | The consumable (for example, a strip, pipette tip box) is correctly placed but is not completely inserted into its position, causing it to be misaligned with its expected position. | Ensure that the consumable is pressed completely into place. Apply firm pressure on the item so that it fits snugly into its deck position. |
| | Consumable version does not match the Genexus™ Dx Software version. For example, a consumable compatible with RUO mode is installed in a sequencer running in IVD mode. | Ensure that you are using consumables compatible with the software mode running on the sequencer. |
| | The expiration date of the consumable has passed. | Ensure that you load consumables that have not exceeded their expiration dates. |
| | The instrument camera is not functioning properly. | If no other cause can be found for the issue, there may be a problem with the camera. Contact Technical Support. |
| Run Status = Failed<br>Details: In the Genexus™ Software **Run Result** screen, the **Run Status** for a completed run is listed as "**Failed**". In the **Sample Results** screen, the **Sample Status** is listed as "**BaseCallingActor FAILED**". | Chip calibration failed due to a chip problem, or an instrument problem. | Repeat the run with a new chip. If the problem persists, contact Technical Support. |
| A lane that has been used is not crossed out in the sequencer screen<br>Details: After completion of a run, the lane used in the run was not crossed out, so that the next run could reuse the lane. | A chip problem caused a datacollect failure to read efuse. | In the sequencer screen, tap **Settings►Clean instrument** to perform a clean instrument. For details, see "Perform a Clean instrument procedure" on page 118. After cleaning, start a new run.<br>Note: The **Clean instrument** procedure renders remaining sequencing reagents and unused lanes on the installed chip unusable in a sequencing run after the cleaning. |

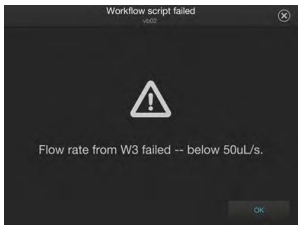
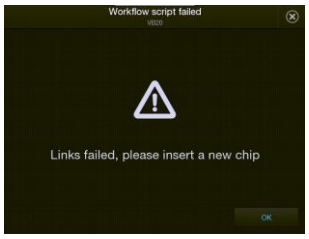
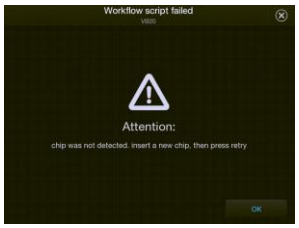
| Observation | Possible cause | Recommended action |
|---|---|---|
| The number of sample reads is low, CF-1 metrics pass QC, but read ratio of inline controls is low<br><br>Details: If CF-1 reads per lane, accuracy, and mean AQ20 read length are good, and read ratio of inline controls (endogenous vs. spike-in) is low (<< 1–3), a problem with sample input is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 102. | Nucleic acid input may have been insufficient, and/or the nucleic acid was degraded. | For a sample run, requantify nucleic acid samples and/or perform sample QC to ensure that the expected nucleic acid input and size was loaded. |
| | | If needed, reisolate and purify nucleic acid samples. |
| The number of sample reads is low, but CF-1 metrics pass QC, and read ratio of inline controls is normal<br><br>Details: If CF-1 metrics passed QC, and read ratio of inline controls is normal (~1–3), a problem in library preparation unrelated to sample input or quality may be indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 102. | One or more of the Genexus™ Dx Strip 1 strips used in the run had magnetic beads trapped in the tube 5 keyhole. | Repeat the run with strips that you have verified have no trapped beads. For more information, see "Before you begin" on page 57. |
| | An incorrect assay was selected for the run, or library amplification parameters were not optimal. | Ensure that you have selected the correct assay and reviewed assay parameters. |
| | Library strips were inadequately equilibrated to room temperature (Genexus™ Dx Strip 1), or incompletely thawed (Genexus™ Dx Strip 2-HD) before loading in the sequencer. | Ensure that Genexus™ Dx Strip 1 strips are fully equilibrated to room temperature, and Genexus™ Dx Strip 2-HD strips are completely thawed before loading in the sequencer. |
| The number of sample reads is low, and CF-1 metrics fail QC<br><br>Details: If CF-1 metrics failed QC, a problem in templating or sequencing is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 102. | One or more of the Genexus™ Dx Strip 3-GX5™ strips used in the run may have had an excessive amount of magnetic beads trapped in the tube 6 or 7 keyhole. | Repeat the run with strips that you have verified have no trapped beads. For more information, see "Before you begin" on page 57. |

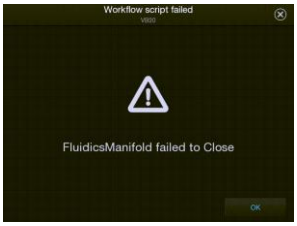
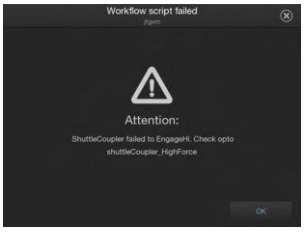
| Observation | Possible cause | Recommended action |
|---|---|---|
| The number of sample reads is low, and CF-1 metrics fail QC<br><br>Details: If CF-1 metrics failed QC, a problem in templating or sequencing is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 102.<br>*(continued)* | Template strips were inadequately equilibrated to room temperature (Genexus™ Dx Strip 3-GX5™), or incompletely thawed (Genexus™ Dx Strip 4) before loading in the sequencer. | Ensure that Genexus™ Dx Strip 3-GX5™ strips are fully equilibrated to room temperature, and Genexus™ Dx Strip 4 strips are completely thawed before loading in the sequencer. |
| | The sequencing chip or coupler was defective or leaky. | Repeat the run with new chip and coupler. If low performance continues, contact Technical Support. |
| | The run was started >14 days after the last initialization was performed, or on an expired initialization. | Perform a Clean instrument procedure (**Settings►Clean instrument**). For more information, see "Perform a Clean instrument procedure" on page 118. After the Clean instrument procedure, install new a chip, and new sequencing reagent bottles and cartridge in the sequencing reagents bay, then repeat the run.<br><br>Note: Reagents are stable on the sequencer for 14 days, after which you may experience decreased performance. For more information, see "Options for an expired sequencer initialization" on page 70. |

<table>
<tr><th>Observation</th><th>Possible cause</th><th>Recommended action</th></tr>
<tr><td>CF-1 AQ20 Mean Read Length QC fails, but positive control/NTC QC passes<br><br>Details: Alternatively, CF-1 AQ20 Mean Read Length QC fails, or is borderline passing, and one or more positive control metrics fail (for example, DNA Control AQ20 Mean Read Length, and DNA or RNA mapped reads).</td><td>Reads were filtered out due to increased low quality ISPs caused by an excess of templated beads generated in the templating reaction.</td><td>Recover the unused pooled sample library from the run, then plan and perform a Library to Result run with diluted library.<br><br>1. Follow the procedure described in “Recover the pooled sample libraries” on page 144 to recover the pooled sample libraries from well E4 of the PCR amplification plate after the run.<br>2. For the number of chip lanes used in the original run, add the volume of recovered pooled libraries and nuclease-free water or Low TE indicated in the following table to well A1 of the sample plate.<br><br><table><tr><td></td><td></td><td></td><td></td></tr><tr><td>1</td><td>17.5 µL</td><td>32.5 µL</td><td>50 µL</td></tr><tr><td>2</td><td>28 µL</td><td>52 µL</td><td>80 µL</td></tr><tr><td>3</td><td>38.5 µL</td><td>71.5 µL</td><td>110 µL</td></tr><tr><td>4</td><td>49 µL</td><td>91 µL</td><td>140 µL</td></tr></table><br>3. Pipet the contents of well A1 up and down 4–5 times to mix.<br>4. Plan and perform a Library to Result run with the diluted library.<br><br>For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 141.</td></tr>
<tr><td>Pipette tips remain in the used tip rack</td><td>Tips were unable to be picked up by the pipettor.</td><td>In Genexus™ Dx Software 6.6 and later, if the pipettor cannot pick up a tip, it leaves the problematic tip in its place and moves to the next available tip. No action is needed.</td></tr>
</table>

# Genexus™ Dx Integrated Sequencer error and warning messages

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error message: Library Prep Failed<br>  | • The sequencer failed xy homing, *or*<br>• The sequencing chip failed to engage at pipette position. | Confirm that no analyses are in progress, then reboot the instrument. Repeat the run. If the failure continues, contact Technical Support. |
| Error message: Templating Failed<br>  | • The sequencer failed xy homing, *or*<br>• The Genexus™ Coupler was misaligned. | 1. Confirm that no analyses are in progress, then reboot the instrument. Repeat the run, and ensure that the coupler is installed with proper alignment. If the failure continues, contact Technical Support.<br>2. If the volume of leftover pooled library in well E4 of the PCR amplification plate is sufficient, perform a Library to Result run using the leftover pooled library. For more information, see "Recover pooled sample libraries and plan a Library to Result run" on page 141.<br>Note: Diluting leftover pooled library more than 2-fold can result in low reads. |
| Error message: Initialization Failed: XXX did not reach fill<br>  | Genexus™ Bottle 2 flow was restricted, or a flow sensor malfunctioned.<br>OR<br>During a post-chip clean, the Genexus™ Bottle 3 was empty, or Genexus™ Bottle 3 flow was restricted.<br>Note: XXX is the name of the conical tube that did not reach fill. | Allow the run to complete, then select **Settings►Clean instrument** to do a clean. After cleaning completes, start a new run. If the error message appears again, contact Technical Support for service.<br>Note: After the run completes, you can recover pooled libraries from the amplification plate, if desired. For more information, see "Recover pooled sample libraries and plan a Library to Result run" on page 141. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error message: Flow rate from W3 failed – below 50 µL/s<br>  | During the post-chip clean the Genexus™ Dx Bottle 3 was empty, or Genexus™ Dx Bottle 3 flow was restricted. | Select **Settings►Clean instrument** to do a clean. Ensure that the Genexus™ Dx Bottle 3 is installed correctly, and the bottle contains sufficient volume. Replace as needed. For details, see “Perform a Clean instrument procedure” on page 118. If the failure persists, contact Technical Support. |
| Error message: PostChipClean failed | The post-chip clean aborted due to a chip error. | In the sequencer screen, tap **Settings►Clean instrument** to re-do a Clean instrument procedure. For details, see “Perform a Clean instrument procedure” on page 118. You may or may not need to replace the chip during deck set up. |
| Error message: Links failed, please insert a new chip<br>  | A chip failed to come up when shuttled to the sequencing position during run setup. | Replace the chip, then try to proceed again. If the error message appears again, contact Technical Support. |
| Error message: Chip was not detected. Insert a new chip, then press retry<br>  | Chip connection was lost and sequencing failed. | 1. Insert a new chip, then press **Retry**.<br>2. If this does not resolve the problem, remove the chip from the instrument. An instrument clean is required before a new run can proceed.<br>3. Force an instrument clean by navigating to **Settings►Clean instrument**.<br>4. After the clean, repeat the run with a new chip and reagents. |
| Error message: Vacuum Clean Failed: Instrument Service might be required | XY motion failed, OR a leak was detected in the vacuum system. | Contact Technical Support for recommended action. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error Message: FluidicsManifold failed to close<br>  | The sequencing clamp hardware did not operate properly. | Instrument service is required to proceed.<br>Note: Pooled libraries can be recovered if the error occurred after library preparation was complete. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 141. |
| Error message: Valve PC4 clog check failed<br>  | A clog was detected during the post-chip clean. | 1. Perform an instrument clean by navigating to **Settings►Clean instrument**.<br>2. After cleaning completes, begin a new run.<br>3. If the error message appears again, contact Technical Support for service. |
| Error message: ShuttleCoupler failed to EngageHi<br>  | Chip shuttle hardware did not operate properly. | Instrument service is required to proceed.<br>Note: Pooled libraries can be recovered if the error occurred after library preparation was complete. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 141. |
| Warning message: Pressure Leak Detected in Vac: vac pressure drop leakrate<br>**Press Ok to open doors and fix the leak.** | Genexus™ Dx Bottle 1 leak was detected. | Remove the Genexus™ Dx Bottle 1 and ensure that the cap is tight, then replace the bottle securely on the instrument. Continue with the run. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Vac: vac time to pressure<br>**Press Ok to open doors and fix the leak.** | A Genexus™ Dx Bottle 1 leak or over-fill was detected. | Ensure that a new Genexus™ Dx Bottle 1 is installed, and the cap is tight and the bottle is secure on the instrument. Continue with the run. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Vac: vac pressure baseline<br>**Press Ok to open doors and fix the leak.** | Genexus™ Dx Bottle 1 leak was detected. | Remove the Genexus™ Dx Bottle 1 and ensure that the cap is tight, then replace the bottle securely on the instrument. Continue with the run. If the pressure leak message appears again, replace Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Warning message: Pressure Leak Detected in Seq: W2P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 2 at the left position. | Remove Genexus™ Dx Bottle 2 installed at the left position, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run. If the pressure leak message is seen again, replace the left Genexus™ Dx Bottle 2 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: W22P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 2 installed at the right position. | Remove the Genexus™ Dx Bottle 2 installed at the right position, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run. If the pressure leak message appears again, replace the right Genexus™ Dx Bottle 2 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: W3P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 3. | Remove the Genexus™ Dx Bottle 3, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run, or repeat the clean if this message appears in a post-chip clean. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 3 with a new bottle. Continue with the run, or repeat the clean if this message appears in a post-chip clean. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: Conicals & Reagent Cartridge **Press Ok to open doors and fix the leak.** | A leak was detected in one or more of the Genexus™ Dx Conical Bottles or the Genexus™ Dx Cartridge. | Tighten all Genexus™ Dx Conical Bottles one bottle at a time to prevent accidental position changes. Inspect the Genexus™ Dx Cartridge for defects. Continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Liquid detected in conicals **Press Ok to open doors and fix the issue.** | The previous post-chip clean did not remove all of the liquid in the Genexus™ Dx Conical Bottles, causing failure of the Conicals empty test and Conical volume test. | Replace the Genexus™ Dx Conical Bottles with new bottles, then tap **Settings►Clean instrument** to clean the instrument. After cleaning, start a new run. |

# Genexus™ Dx Software

| Observation | Possible cause | Recommended action |
|---|---|---|
| Cannot sign in to the Genexus™ Dx Software | You have either forgotten your password or are signed out due to several failed login attempts. | Contact the Genexus™ Dx Software system administrator. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Batch sample import fails | One or more entries in the sample-import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the sample-import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample-import template file, and be careful to copy only those rows that contain actual data. |
| | The sample import spreadsheet contains a nonunique sample name. | Every sample name in the software must be unique. Ensure that the spreadsheet does not contain any duplicate sample names, then repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1. |
| | The headings in the sample import spreadsheet do not match the sample attributes in the software. | The headings must match the sample attributes in the software exactly. Check the headings for spelling or other errors. |
| Library batch import fails | One or more entries in the library batch import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the library batch import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template file, and be careful to copy only those rows that contain actual data. |
| | The library batch import spreadsheet contains a nonunique **Library Batch ID**. | Each **Library Batch ID** in the software must be unique. Ensure that the spreadsheet does not contain any duplicate IDs, then repeat the import. The system check is not case-sensitive. For example, a **Library Batch ID** of ABC1 conflicts with abc1. |
| | A sample name entered in the library batch import spreadsheet does not match a sample name listed in the **Manage Samples** screen. | Ensure that the sample names entered into the spreadsheet are correct and match an existing sample name added to the software. |
| | The Barcode ID name format does not exactly match the format that is used in the **Prepare Library Batch** dialog box. | Use the name format following the Barcode ID name format found in the Barcode Set reference lists (**Settings►References►Barcode Set**), for example: IonDual_0101 through IonDual_0196, or IonHDdual_0101 to IonHDdual_0132. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Library batch import fails *(continued)* | An invalid library, control, or panel kit barcode has been entered in the spreadsheet. | Ensure that you have correctly entered a valid kit barcode in the appropriate cell of the spreadsheet. |
| The results of the run do not appear in the Results / Run Results screen | The instrument disk space is full. | Clear disk space on the sequencer. For more information, see “Manually delete run data” on page 125. |
| Cannot download run result files | The run failed. Links to run files are not available for runs that fail QC. | Repeat the run. |

# B Touchscreen reference

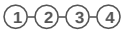

- Sequencer touchscreen icons ..... 113
- Settings ..... 115


## Sequencer touchscreen icons




| Number | Icon | Description |
|---|---|---|
| 1 | | Network connectivity – connected |
| | | Network connectivity – not connected |
| 2 | | Instrument idle |
| | | Sequencing in progress |
| | | Instrument ready |

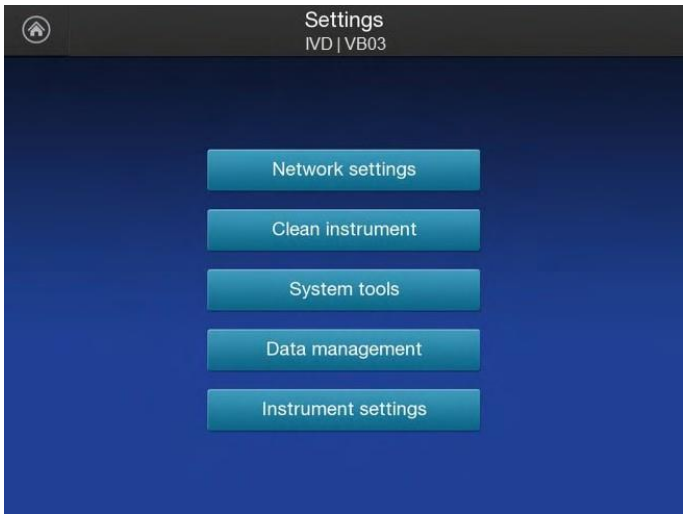
*(continued)*

| Number | Icon | Description |
|---|---|---|
| 2 |  | Error |
| 3 |  | Chip status – Absent[1] |
| |  | Chip status – Standby |
| |  | Chip status – Connecting |
| |  | Chip status – Ready |
| |  | Chip status – Imaging |
| |  | Chip status – Error |
| 4 |  | Chip lane status – 4 lanes available |
| |  | Chip lane status – 1 lane in use or used, 3 lanes available |
| |  | Chip lane status – 2 lanes in use or used, 2 lanes available |
| |  | Chip lane status – 3 lanes in use or used, 1 lane available |
| |  | Chip lane status – 4 lanes in use or used, 0 lanes available |
| 5 |  | Instrument File System Space – the percent of file space used is indicated. The instrument checks for sufficient disk space before each run, and notifies the operator if there is insufficient disk space for a run.<br>The indicator turns yellow when disk space is ≥67% full. The indicator turns red when ≥90% full. |
| 6 |  | Instrument Server File System Space – the percent of file space used is indicated.<br>If the indicator turns red, archive data from the server to free up disk space. See the software help system for information about archiving data. |

[1] If Chip status is Absent, always insert a new chip.

# Settings

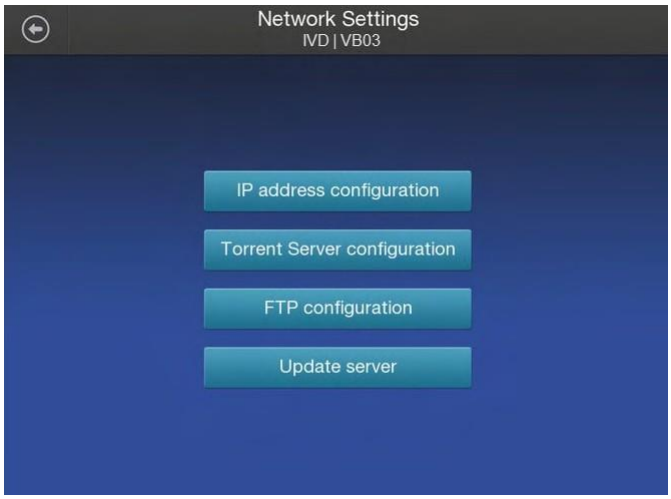
Use the **Settings** menu to view and/or change instrument settings, manage data and network configurations, and clean the instrument.

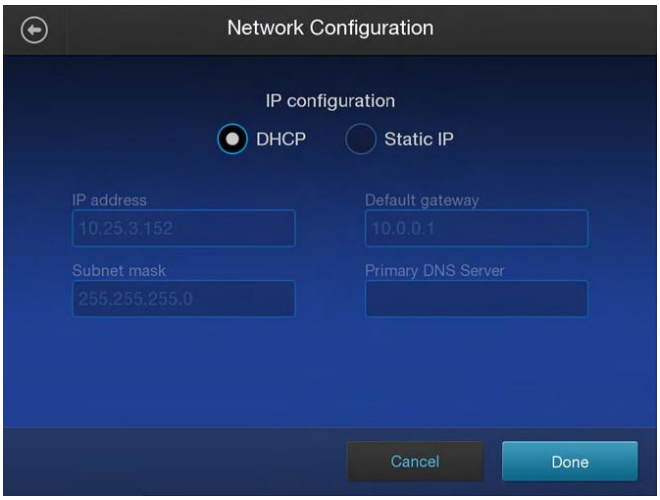
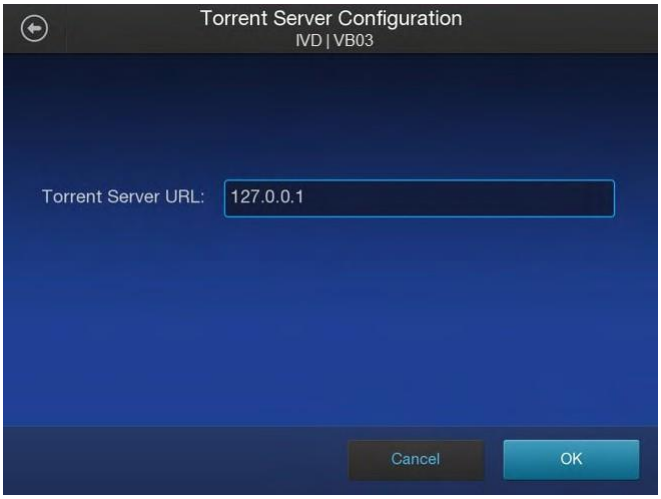



Note: The **System tools** option is for use only by trained service personnel.

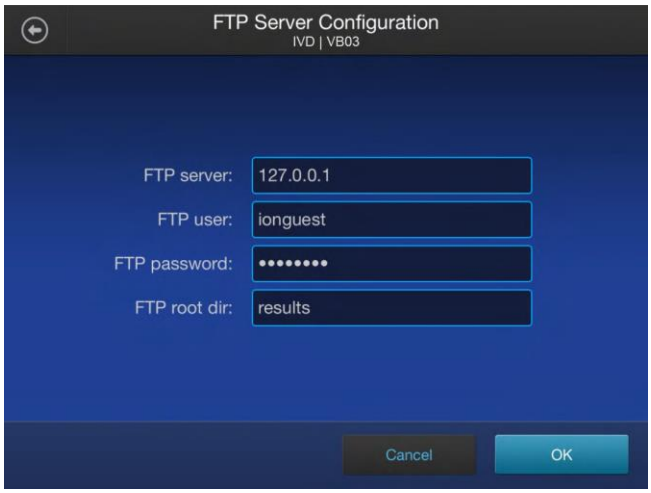
## Network Settings

Use the **Network Settings** menu to configure IP address, Ion Torrent™ Server, and FTP settings.



| Screen | Description | When/How to use |
|---|---|---|
| IP Address Configuration | Set or change the IP configuration (DHCP or Static IP).<br>  | 1. Select either the DHCP or the Static IP option.<br>2. Touch the screen in the field you want to edit to activate the field. A virtual keyboard appears.<br>3. Enter the new information, then press Done. |
| Ion Torrent™ Server Configuration | Change the Ion Torrent™ Server IP address and user information.<br>  <br>Enter the following IP address in the Torrent Server URL field for your instrument: 127.0.0.1 | Use this screen when a change to the Ion Torrent™ Server IP address or user information is required.<br><br>IMPORTANT! Do not change the address unless directed to do so by a Thermo Fisher Scientific field service engineer. |

*(continued)*

| Screen | Description | When/How to use |
|---|---|---|
| FTP Server Configuration | Change the FTP Server IP address and user information.<br>  <br><br>Enter the following IP address in the FTP server field for your instrument: 127.0.0.1 | Use the screen when a change to the FTP Server IP address or user information is required.<br><br>IMPORTANT! Do not change the address unless directed to do so by a Thermo Fisher Scientific field service engineer. |

## Perform a Clean instrument procedure

UV cleaning is normally performed at the end of a sequencing run. Perform a **Clean instrument** procedure from the **Settings** menu if:

- A sequencing run has been aborted, or a power failure occurred during a run.

  IMPORTANT! If the run stoppage or power failure occurred during the first run on a new chip, contact Technical Support to open a service call.

- The normal post-sequencing run cleaning was not complete.
- The sequencer has been idle for ≥2 weeks after a run that used the remaining lanes or all 4 lanes of a chip.
- The sequencer has been powered on after a long-term shutdown as described in the procedure in “Power on after a long-term shutdown” on page 140.

The **Clean instrument** command starts the following cleaning procedures:

- Vacuum clean—Cleaning of the four chip vacuum lines and robotic waste line.
- Post-chip clean—Cleaning of sequencer fluidic lines with a conical flow rate test.
- UV clean—Irradiation of the deck surface with UV light. For more information, see “Clear the instrument deck and perform a UV Clean” on page 67.

IMPORTANT! The **Clean instrument** procedure renders remaining sequencing reagents and unused lanes on the installed chip unusable in a sequencing run after the cleaning.

The following items are needed if the items are not already loaded in the sequencer:

- New Genexus™ Dx Bottle 3
- Used and empty Genexus™ Dx Bottle 2 (2)
- Used Genexus™ Dx Bottle 1
- Used Genexus™ Dx Cartridge
- Used chip
- Used coupler

IMPORTANT! Keep used sequencing reagent bottles and supplies on hand to perform the **Clean instrument** procedure.

1. In the **Settings** menu, tap **Clean instrument**.
2. Follow the on-screen instructions, then tap **Proceed** to go to the following sequence of screens: **Clear Deck** if there are items to be cleared, **Load Deck**, **Clear Sequencing Reagents**, and **Load Sequencing Reagents**. For more information, see “Clear the instrument deck and perform a UV Clean” on page 67.

   Note: If a sequencing run completes with 1–3 unused lanes remaining on a chip, and the operator had selected **Do Force Clean** during run setup, the sequencer proceeds directly to the **Clean instrument** procedure after the run. The sequencing reagents bay doors remain closed and no prompts appear on the sequencer screen.

3. Follow the on-screen instructions in the **Clear Deck** and **Clear Sequencing Reagents** screens. After sequencing reagents are cleared, tap **Next** to return to the home screen.

Note: The **Clean instrument** procedure takes about 2 hours to complete.

## Replace Genexus™ Dx Conical Bottles during a Clean instrument

If the sequencer detects restricted flow from the Genexus™ Dx Conical Bottles during a **Clean instrument** procedure, the following alert is displayed.




If you see this alert, follow these steps to replace the 5 Genexus™ Dx Conical Bottles.

1. Tap **OK**. The screen returns to the run screen.
2. Tap **Settings►Clean instrument** again.
3. Follow the screen instructions to clear the deck, then load the deck.
4. When the sequencing reagents bay doors open, do the following.
   a. Remove, but do not discard, the Genexus™ Dx Bottle 1, the 2 Genexus™ Dx Bottle 2 bottles, and the Genexus™ Dx Bottle 3 to gain easier access to the Genexus™ Dx Conical Bottles.
   b. Remove the 5 used Genexus™ Dx Conical Bottles, then install 5 new conical bottles.
   c. Replace the 4 reagent bottles that you removed in their original positions.
5. Close the sequencing reagents bay doors.
   The **Clean instrument** procedure begins automatically.
6. After the cleaning finishes, follow the screen instructions to clear the deck and remove the sequencing reagents.
   For further information, see “Replace the Genexus™ Dx Conical Bottles” on page 135.

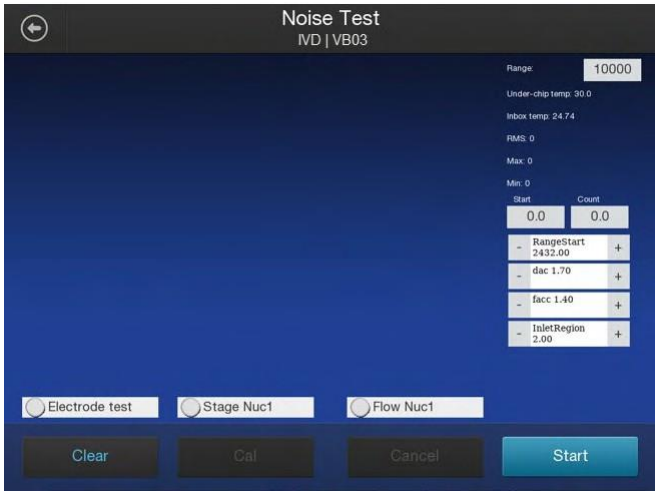
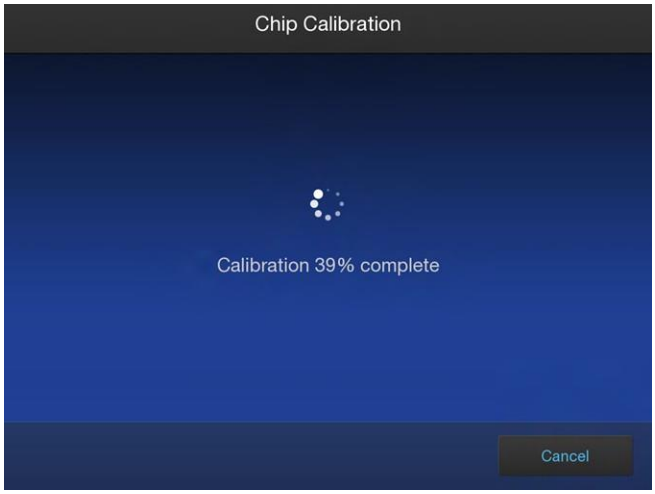
# System Tools

The **System Tools** menu enables you to upload instrument diagnostics, manage data, and shut down or reboot the instrument.

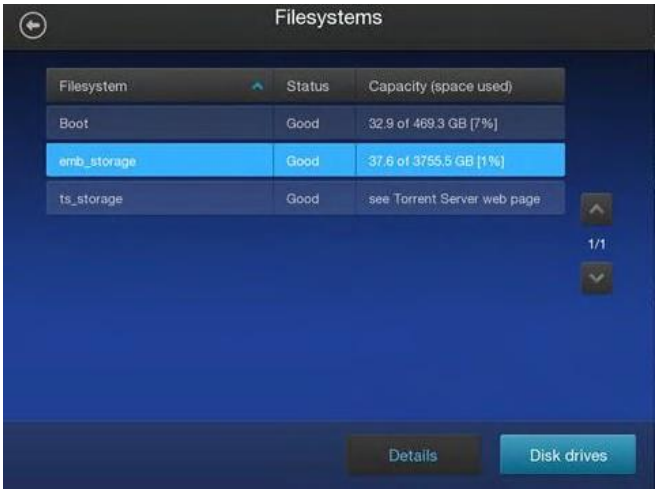
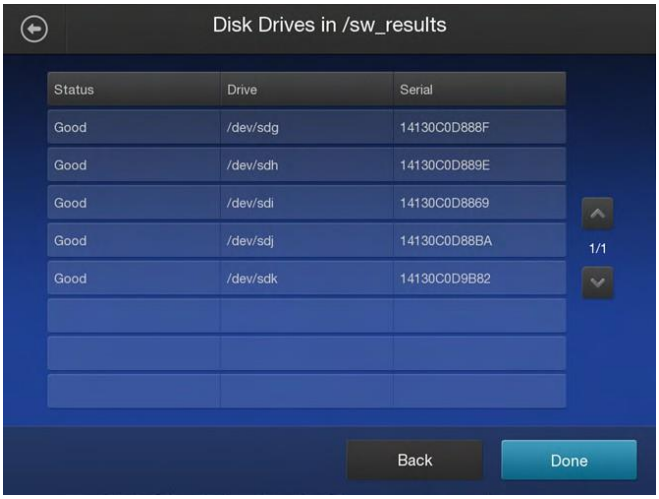
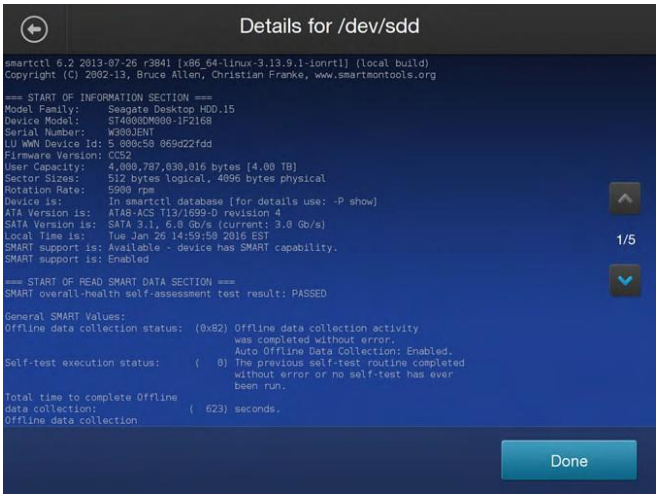



| Tool | Description | When/How to use |
|---|---|---|
| RFID | Lists the product, product expiration date, and remaining uses of the product.<br>  | To determine if reagents that were left on an instrument during a period of instrument shutdown have expired.<br>**1.** Tap the screen within the row of the product to view an RFID Tag Detail page.<br>**2.** Tap Done to return.<br>Note: You can also use the RFID Tags screen to track remaining days of reagent use before a sequencer initialization expires. We recommend that you use reagents within 14 days of initialization. After 28 days, the sequencer displays a warning at run start with options to proceed to a clean and new initialization, ignore to continue without clean, or cancel the run. For further information, see “Options for an expired sequencer initialization” on page 70. |

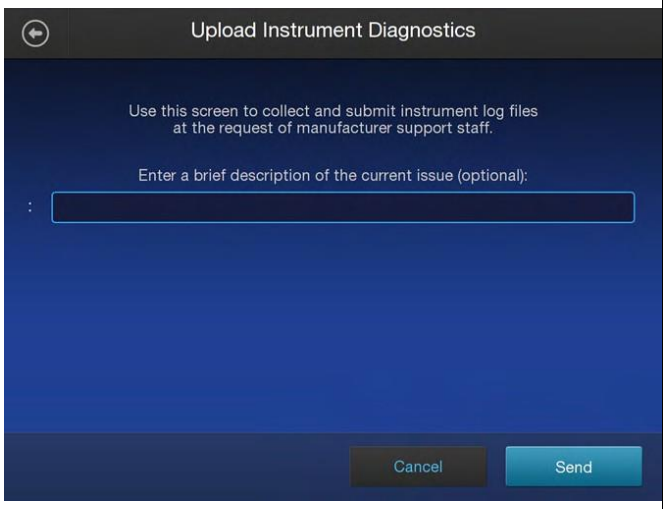
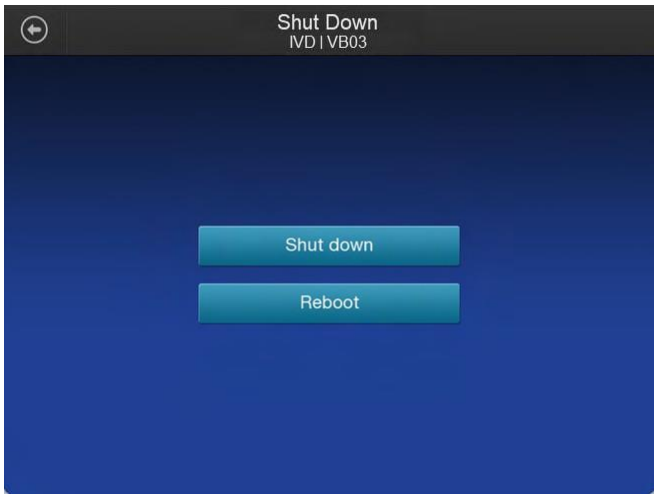
*(continued)*

| Tool | Description | When/How to use |
|---|---|---|
| Noise test | Provides real-time measurement of electrical noise readings on the chip.<br>  | For troubleshooting if directed to do so by Technical Support. |
| Chip calibration | Checks the status of a chip.<br><br>IMPORTANT! Do not perform a chip calibration unless instructed to do so by a field service engineer.<br>  | **1.** Insert a new chip into the chip installation slot.<br>**2.** In the System Tools menu, tap Chip Calibration to start the chip calibration.<br>**3.** In the dropdown lists, select the Image to display and the type of Calibration to perform.<br>**4.** After a chip calibration, perform a Clean instrument before removing the chip.<br><br>Note: Tap Cancel to quit the chip calibration. |

*(continued)*

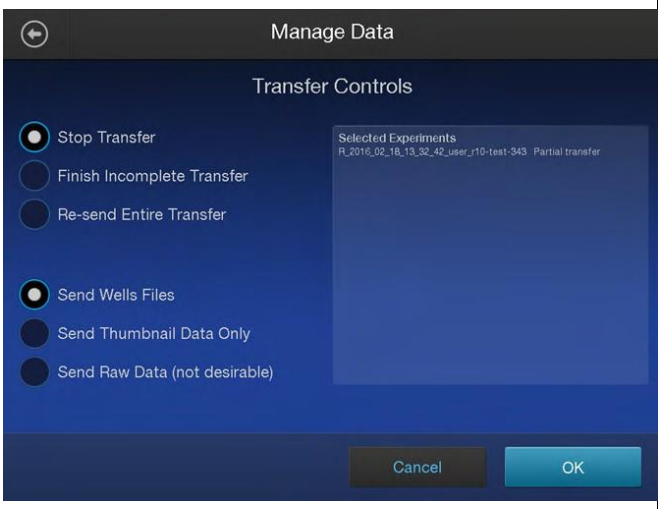
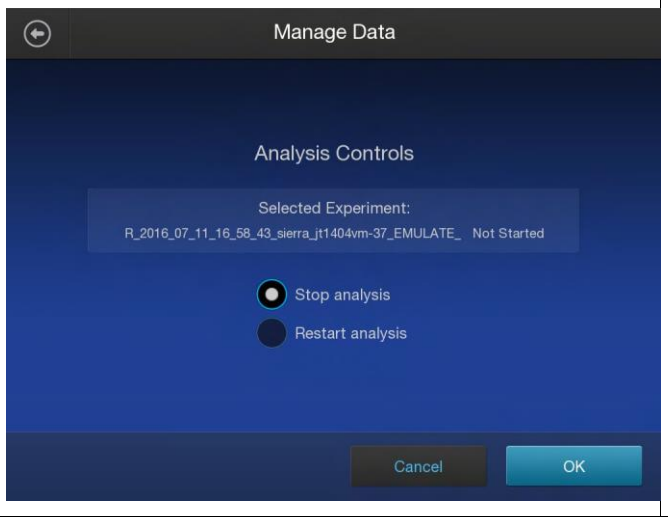
| Tool | Description | When/How to use |
|---|---|---|
| Filesystems and disks | Provides real-time drive activity status and disk space usage.<br>  <br>  <br>  | Use if directed to do so as part of a troubleshooting procedure:<br>**1.** In the System Tools menu, tap Filesystems and disks. The Filesystems screen opens.<br>**2.** Tap the screen within the row of the Filesystem that you want to view, then tap Details. The Disk Drives in <Filesystem> screen opens.<br>**3.** Tap the screen within the row of the Drive to view details of the disk drive. The Details for <Drive> screen opens.<br>**4.** Tap Done to return to the previous screen. |

*(continued)*

| Tool | Description | When/How to use |
|---|---|---|
| Upload diagnostics | Upload instrument diagnostics files for review by support personnel.<br>  | Upload an instrument diagnostics file after an aborted run, or if otherwise directed to do so by Technical Support.<br>Note: The file is pushed to Genexus™ Dx Software. To retrieve the file using Genexus™ Dx Software, navigate to ⚙ (Settings)►About, click Instrument Diagnostics, then follow the on-screen instructions. |
| Shut down | Provides access to "Shut down" and "Reboot" commands.<br>  | Use these commands if directed to do so by Technical Support as part of a troubleshooting procedure, or if the instrument will not be used for more than 3 days.<br>Note: It is not necessary or recommended to power off the instrument overnight or over the weekend. To power on the instrument after a power off, see “Power on” on page 29. |

## Data Management

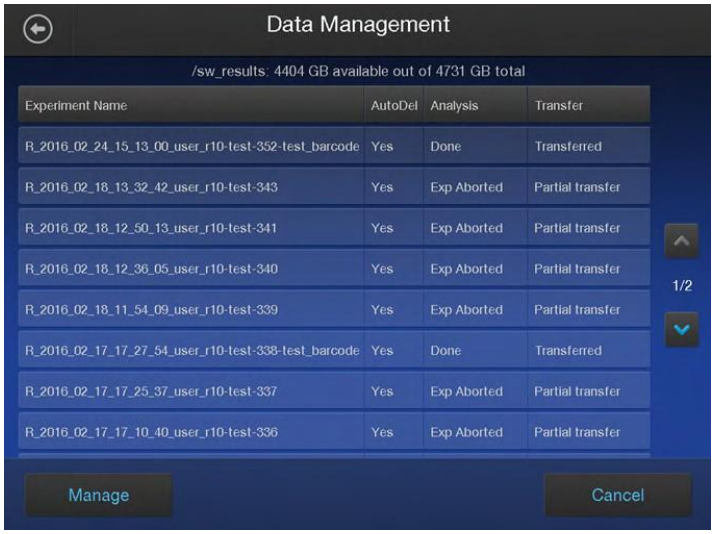
The **Data Management** function allows you to delete run data manually, or transfer data in the event of a failure of automatic transfer. Under normal conditions, run data are automatically transferred to the analysis server, then deleted from the instrument hard drive.

| Item | Description | When/How to use |
|---|---|---|
| Delete data | Manually delete run data from the instrument. | If the instrument hard drive becomes full, see “Manually delete run data” on page 125. |
| Transfer Controls | Transfer run data from the instrument hard drive to the Ion Torrent™ Server.<br>  | 1. Tap the screen in the Experiment Name row of the individual experiment to be transferred.<br>2. Tap Transfer.<br>3. Select the transfer action to be performed.<br>4. Select the file transfer option.<br>5. Tap OK. |
| Analysis Controls | Stop the analysis of the data of individual runs.<br>  | 1. Tap the screen in the Experiment Name row of the individual experiment to be analyzed, then tap Analysis.<br>2. Select Stop Analysis, then tap OK.<br>Note: Restart analysis is currently not supported. |

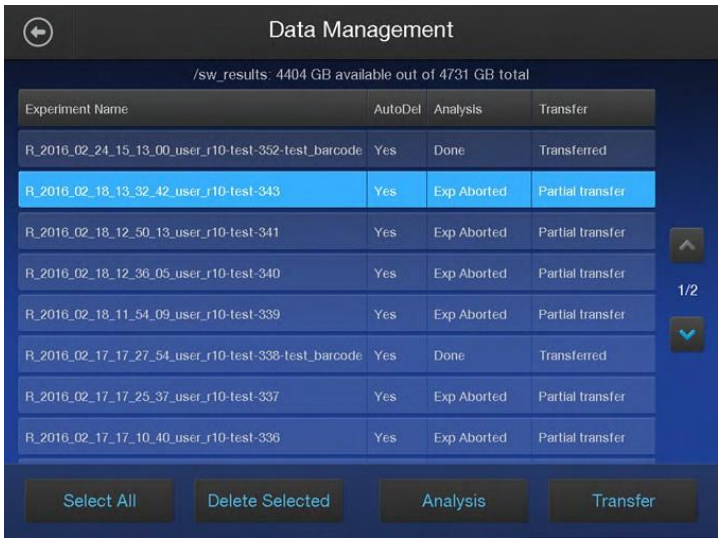
## Manually delete run data

To troubleshoot data management problems, use the **Data Management** function to delete run data manually, or transfer the data to an external server.

1. In the **Settings** menu, tap **Data Management** to access the **Data Management** screen, then tap **Manage**.



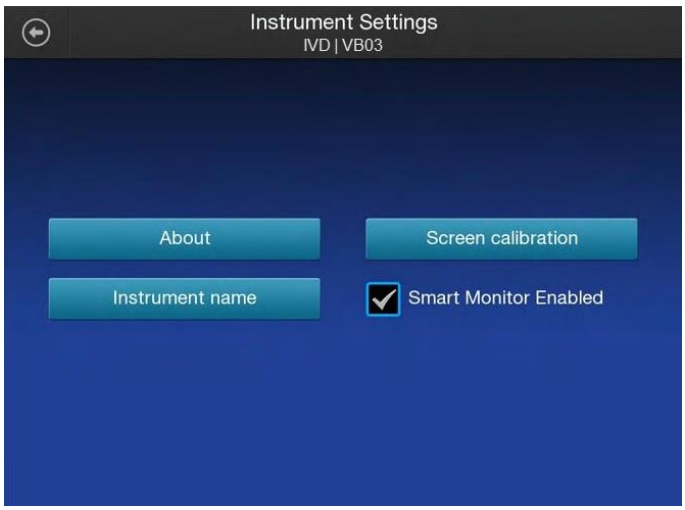

2. Tap **Select All** to select all the available experiments, or tap the screen in the **Experiment Name** row of the individual experiment to be managed.

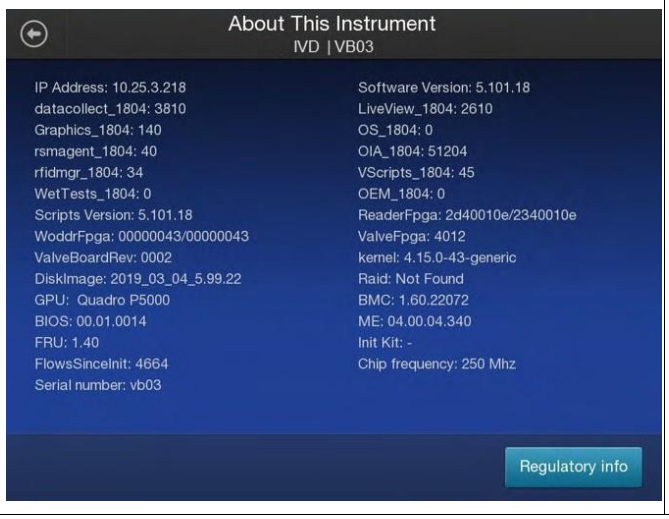



3. Tap **Delete Selected**.

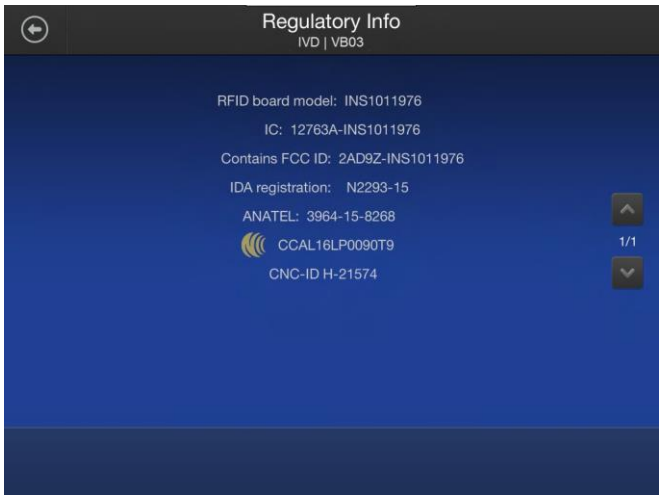
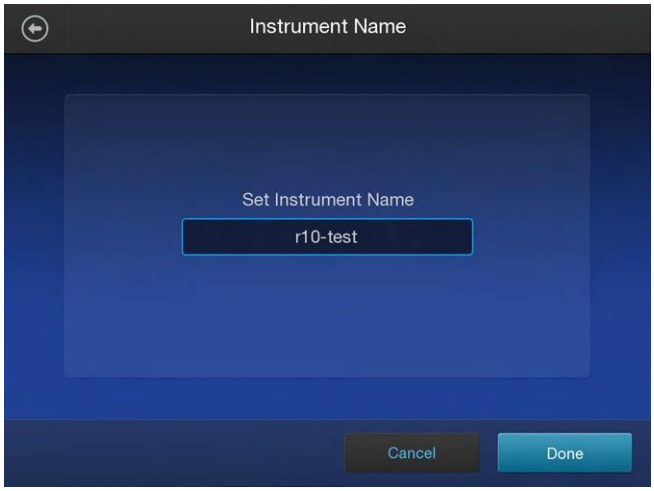
# Instrument settings

The **Instrument Settings** buttons access information about the instrument and allow you to set the instrument name and calibrate the touchscreen.

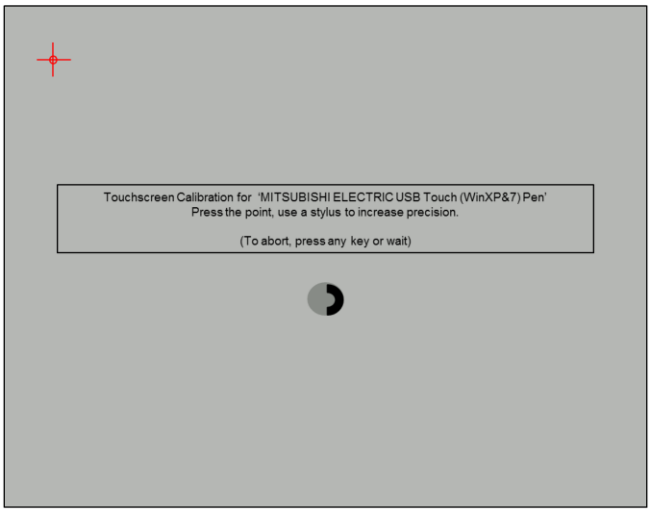



| Button | Description | When/How to use |
|---|---|---|
| About | Provides instrument details.<br>  | Use the screen to view instrument reference information or access regulatory information. Click Regulatory info to view the following screen. |

*(continued)*

| Button | Description | When/How to use |
|---|---|---|
| Regulatory info | Lists instrument-specific regulatory information.<br>  | To view instrument RFID regulatory information. See “Radio compliance standards” on page 159 for compliance information. |
| Instrument name |  | To change the instrument name. |

*(continued)*

| Button | Description | When/How to use |
|---|---|---|
| Screen calibration |  | For troubleshooting if directed to do so by Technical Support.<br>Touch the red cross with your finger or a stylus each time it appears. In total, you touch the screen 4 times, one time in each corner. |
| Smart Monitor Enabled | — | Select the checkbox to enable remote monitoring of instrument runs by Thermo Fisher Scientific for troubleshooting. |

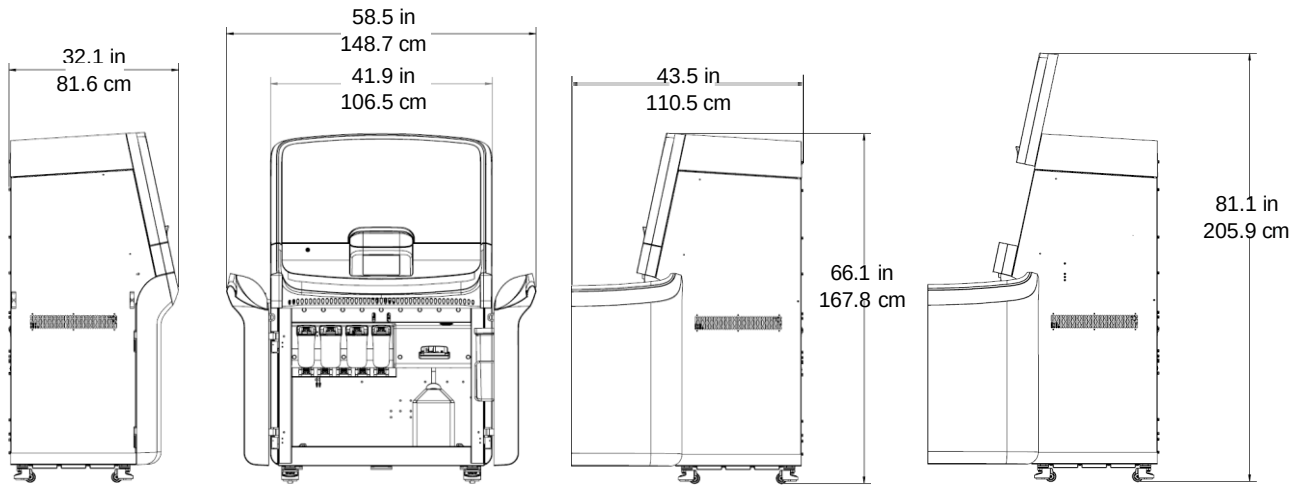
# C Instrument specifications


- Instrument dimensions, weight, and clearances ..... 129
- Environmental requirements ..... 130
- Thermal specifications for the instrument ..... 131
- Ventilation requirements ..... 131
- Electrical requirements ..... 131


## Instrument dimensions, weight, and clearances

### Instrument dimensions and weight

| Component | Height | | Length (depth) | | Width | | Weight |
|---|---|---|---|---|---|---|---|
| | Open | Closed | Open | Closed | Open | Closed | |
| Genexus™ Dx Integrated Sequencer | 81.1 in (205.9 cm) | 66.1 in (167.8 cm) | 43.5 in (110.5 cm) | 32.1 in (81.6 cm) | 58.5 in (148.7 cm) | 41.9 in (106.5 cm) | 450 lb (204.1 kg) |



## Instrument clearances

During instrument setup and maintenance, it is necessary to access the back and sides of the instrument. If the back of an instrument component faces a wall, it is necessary to have enough space to roll the instrument out from the wall to enable access to the back of the instrument.

IMPORTANT! For safety, the power outlet that is used for powering the instrument must be accessible at all times.

| Component | Top | Front | Left/Right | Back |
|---|---|---|---|---|
| Genexus™ Dx Integrated Sequencer | 20 in (50.8 cm) | 18 in (45.7 cm)[1] | 6 in/12 in[2] (15.2 cm/30.5 cm) | 12 in (30.5 cm) |

[1] The instrument requires 36.0 in (90.0 cm) aisle in front of bench for operator access.

[2] To allow sufficient side clearance for the instrument doors to open.

# Environmental requirements

Ensure that the installation room is maintained under correct environmental conditions. Avoid placing the sequencer next to heaters, cooling ducts, or in direct sunlight. Place the sequencer at least 1 meter away from major sources of electronic noise such as refrigerators or microwaves. Fluctuations between day and night temperatures can cause system instability.

| Component | Acceptable range |
|---|---|
| Altitude | Located between sea level and 2,500 m (8,200 ft) above sea level |
| Humidity | 20%–70%, non-condensing |
| Operating temperature | 15°C to 30°C (59°F to 86°F)<br><br>IMPORTANT!<br>▪ The recommended operating temperature for the Genexus™ Dx Integrated Sequencer during install qualification and performance qualification runs is 23°C (73°F).<br>▪ If the sequencer is operated at an elevation of 1,800 m (5,900 ft) or greater above sea level, the maximum operating temperature is 23°C (73°F).<br>▪ The room temperature must not fluctuate more than 2°C (3.6°F) over a 2-hour period. |
| Vibration | Do not install the instrument(s) near equipment that causes vibration (freezers, pumps, and similar equipment). Significant vibration during sequencing can add noise and reduce the quality of the sequencing measurements. |
| Electromagnetic interference | The electromagnetic environment should be evaluated before placement and operation of the instrument. Do not use the instrument in close proximity to sources of strong electromagnetic radiation (for example, unshielded intentional RF sources), as these sources can interfere with proper operation. |

*(continued)*

| Component | Acceptable range |
|---|---|
| Pollution | The Genexus™ Dx Integrated Sequencer is intended to be used in Office or Laboratory controlled environments. |
| Other conditions | For indoor use only. Install the sequencer on a level surface. The installation location must be away from any vents that could expel particulate material on the system components. |

# Thermal specifications for the instrument

During operation, the thermal output based on the typical current draw of the instrument is:

| Component | Typical draw[1] | Thermal output |
|---|---|---|
| Genexus™ Dx Integrated Sequencer | 1000W | 3412 BTU/h |

[1] Maximum draw: 1200W

# Ventilation requirements

Allow at least 12 in (30 cm) of clearance around the Genexus™ Dx Integrated Sequencer for ventilation. Do not block air inlets or outlets that allow proper ventilation.

# Electrical requirements

WARNING! For safety, the power outlet used for powering the instrument must be accessible at all times. See “Instrument clearances” on page 130 for information about the space needed between the wall and the instrument. In case of emergency, you must be able to immediately disconnect the main power supply to the instrument. Allow adequate space between the wall and the equipment so that the power cord can be disconnected in case of emergency.

AVERTISSEMENT ! Par souci de sécurité, la prise de courant alimentant l’instrument doit être accessible à tout moment. En cas d’urgence, il doit être possible de débrancher immédiatement l’alimentation principale de l’ensemble des équipements. Laisser suffisamment d’espace entre le mur et les équipements afin de pouvoir débrancher les câbles d’alimentation sans encombre, en cas d’urgence.

- Electric receptacle required: 2-prong with ground pin
- Mains AC line voltage tolerances can be up to ± 10% percent of nominal voltage
- Use only the specified power cable supplied with the instrument to connect the sequencer to a wall receptacle. Route the power cord away from the workspace to avoid accidental disconnection.

| Component | Rated input voltage | Rated current[1] | Rated frequency | Rated power[2] |
|---|---|---|---|---|
| Genexus™ Dx Integrated Sequencer | 100–240 VAC | 12–5A | 50/60 Hz | 1200W |

[1] Based on rated current at minimum input voltage.
[2] Average power is 1000W.

A power cord is provided with the instrument. If the power cord is not suitable for installation in your region, ensure that any power cord you do use is:

- Maximum 10 ft (3 m) in length
- Grounding type
- Compatible with the power supply receptacles used to connect to main power
- Suitable for the rating of the instrument and mains power supply
- Compliant with local safety requirements (for example, UL Listed for North America, JIS approved for Japan, HAR or agency certified for Europe)

# D Supplemental information


- Maintain the Genexus™ Dx Integrated Sequencer ..... 134
- Recover pooled sample libraries and plan a Library to Result run ..... 141
- Genexus™ Dx Integrated Sequencer Install Kit ..... 145
- Genexus™ Dx reagent strip and panel tube fill volume guide ..... 146

# Maintain the Genexus™ Dx Integrated Sequencer

## Materials required

Cleaning

- Lint-free wipes
- Deionized water
- 70% isopropanol

Liquid waste filter replacement

- Genexus™ Dx Filter (ordered separately [Cat. No. A50868])

Conical bottle replacement

- Genexus™ Dx Conical Bottles (from the Genexus™ Dx Integrated Sequencer Install Kit, or ordered separately [Cat. No. A50869])

## Clean or decontaminate the sequencer

If a spill or leak occurs inside the instrument, perform the following steps to clean or decontaminate the sequencer.

Note: Dispose of all waste in appropriate liquid or solid waste containers.

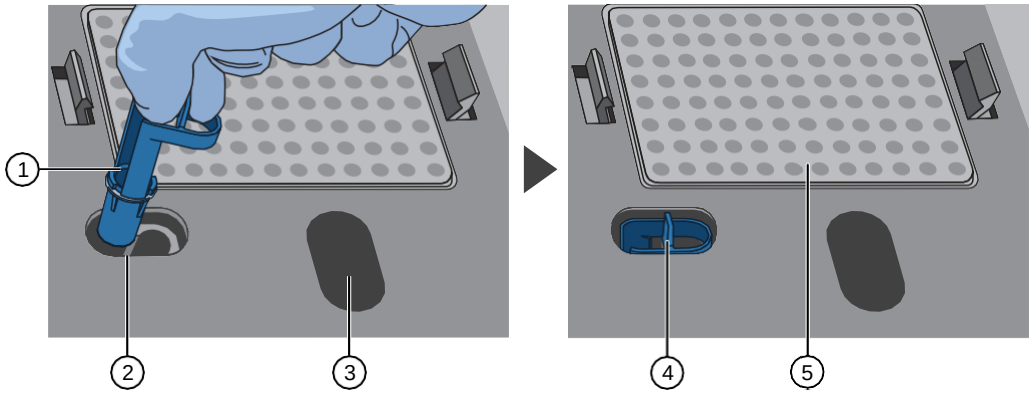
1. Remove each Genexus™ Dx Bottle 2, then remove and empty the Genexus™ Dx Bottle 1 and Waste carboy.
2. Remove the Genexus™ Dx Cartridge.
3. Inspect the floor of the sequencing reagents bay and Genexus™ Dx Cartridge port for liquid.
4. Using lint-free wipes, soak up as much liquid as possible, then clean the affected area with wipes moistened with deionized water.
5. Wipe the affected surfaces with 70% isopropanol, then allow to air-dry.

## Replace the Genexus™ Dx Filter

The Genexus™ Dx Filter captures particulate matter in the liquid waste to prevent blockage of the waste line over time. The filter needs to be replaced with a new filter after one year of regular instrument use as part of annual preventive maintenance, or if a clog is present. For more information, see “Clear the instrument deck and perform a UV Clean” on page 67.

1. Remove the old filter from the liquid waste disposal port on the instrument deck by grasping the filter firmly and pulling up. Discard the filter as regular waste.

2. Insert a new Genexus™ Dx Filter into the liquid waste disposal port, then press firmly to seat the filter O-ring securely.




1. Genexus™ Dx Filter
2. Liquid waste disposal port
3. Used pipette tip disposal port

Installed filter

4. Sample plate station
5.

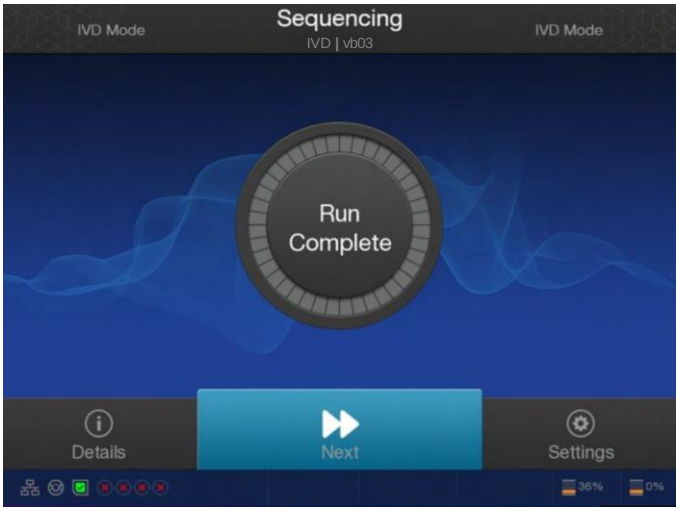
## Replace the Genexus™ Dx Conical Bottles

The Genexus™ Dx Conical Bottles need to be replaced after one year of regular instrument use as part of annual preventive maintenance or when filters in the bottles become partially clogged, causing decreased flow rate. The Genexus™ Dx Integrated Sequencer measures flow rate in a Conical Flow Rate test that is performed during every post-chip clean. If the conical flow rate test fails, the sequencer automatically redirects to the conical bottle replacement procedure. During a normal run, a post-chip clean is performed in three situations:

- At the end of the run if all four lanes have been used.
- At the end of the run if you selected **Do Force Clean** at run setup.
- At run setup when a run is selected but insufficient unused lanes in the installed chip are available for the run. If you continue with the run, a post-chip clean is performed before the run can proceed.

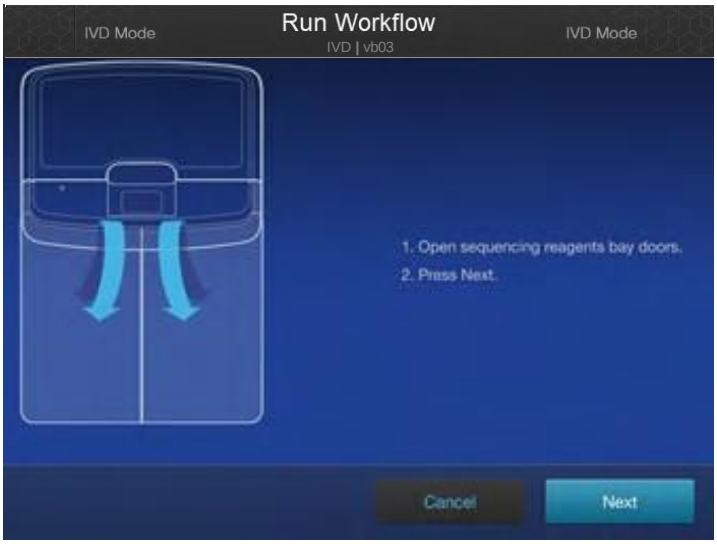
Follow these steps to replace the Genexus™ Dx Conical Bottles.

1. After a run, in the **Run Complete** screen, tap **Next**.




If the Conical Flow Rate test failed, a screen appears directing you to open the sequencing reagents bay doors.

2. Open the sequencing reagents bay doors, then tap **Next**.




The **Remove Consumables** screen appears.

3. Remove, but do not discard, the Genexus™ Dx Bottle 1 (position 1), the two Genexus™ Dx Bottle 2 bottles (positions 2 and 3), and the Genexus™ Dx Bottle 3 (position 4), then tap **Next**.




4. Remove the five used Genexus™ Dx Conical Bottles, install five new conical bottles, then tap **Next**.




Note: Discard the used conical bottles appropriately. Inspect the plastic nozzles on the new bottles for any pinches or deformations. To avoid pinching or folding of the plastic nozzle, install the bottles straight-on, not at an angle.

5. Replace the four reagent bottles that you removed in step 3 in their original positions.




Note: Note: Before installing, *gently* invert each Genexus™ Bottle 2 five times to mix—avoid vigorous mixing. Inspect the plastic nozzles for any pinches or deformations. To avoid pinching or folding of the plastic nozzle, install the bottles straight-on, not at an angle.

6. After replacing the bottles, close the sequencing reagents bay doors, then tap **Next** in the **Insert Consumables** screen.

   After you tap **Next**, the sequencer starts a **Leak Test**.



Note: If you tap **Next** while the sequencing reagents bay doors are open, an alert appears asking you to close the doors before proceeding.

- If the sequencer passes the **Leak Test**, no action is needed. The sequencer proceeds to a cleaning cycle.




- If the sequencer fails the **Leak Test** a notification appears. Tap **OK** to return to step 2 to fix the leak. Open the sequencing reagents bay doors, check the tightness of each conical bottle that you installed, then ensure that the four reagent bottles are installed correctly. Close the sequencing reagents bay doors, then tap **Next** to repeat the **Leak Test**. If the **Leak Test** fails again, contact Technical Support.

After completion of a successful **Leak Test** and cleaning, the sequencer moves to a normal workflow.

- If the sequencer is at the end of the run, the next screen is a **Clear Deck** screen, followed by a **Clear Sequencing Reagents** screen.
- If the sequencer is at the start of the run, the next screen is a **Clear Deck** screen, followed by a **Load Deck** screen.

## Power off and power on the sequencer before and after a long-term shutdown

Follow these procedures to power off the sequencer if it will not be used for more than 28 days, and to power on after a long-term shutdown.

### Power off before a long-term shutdown

- If all four lanes of a chip have been used, and a post-chip clean has completed successfully, follow these steps to power off the sequencer, as described in “Power off” on page 30.
  a. In the home screen, tap **Settings►System Tools►Shut down**.
  b. Select **Shutdown**.
     A confirmation message appears. Select **Yes** to power off the instrument.

c. Turn the power switch located at the back of the instrument to the off ( O ) position.

- If a partially used chip is installed on the instrument, follow these steps to perform a Clean instrument procedure as described in page 118, then power off the instrument.

  a. In the home screen, tap **Settings►Clean Instrument**.

  b. Follow the on-screen instructions to start the cleaning.

  Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

  c. After the cleaning completes, power off the system in the home screen by tapping **Settings►System Tools►Shut down**.

  d. Select **Shutdown**.
  A confirmation message appears. Select **Yes** to power off the instrument.

  e. Turn the power switch located at the back of the instrument to the off ( O ) position.

## Power on after a long-term shutdown

After a long-term shutdown (>28 days), follow these steps to power on the sequencer, as described in “Power on” on page 29.

1. Turn the power switch on the back of the sequencer to the on ( | ) position.
2. Press the power button on the front of the instrument.
   The button illuminates.
3. Sign in with your user name and password.
   After the home screen appears, the sequencer is ready for use. Check for instrument alarms, if any.
4. Perform a Clean instrument procedure as described in “Perform a Clean instrument procedure” on page 118. A Clean instrument procedure must be performed even if the procedure was performed before the long-term shutdown.

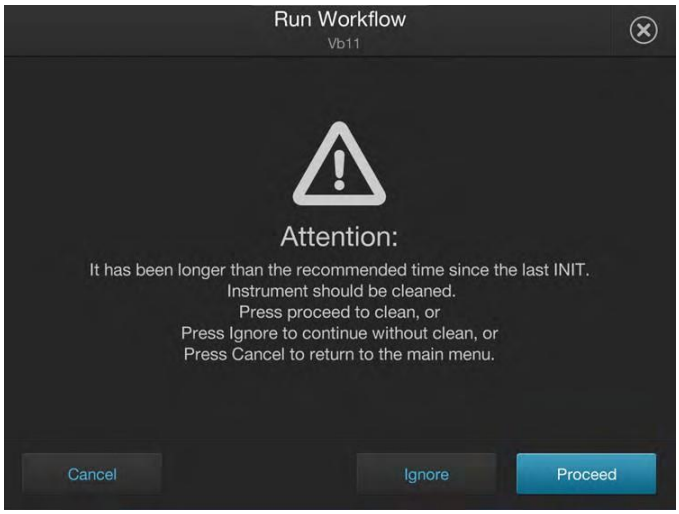
   a. In the home screen, tap **Settings ►Clean Instrument**.

   b. Follow the on-screen instructions to start the cleaning.

   Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

   c. After the cleaning is complete, the instrument is ready to use.

5. If the instrument was in an initialized state before the system was powered off, and shows the following alarm for an expired initialization, perform the following steps to clean the instrument as described in “Options for an expired sequencer initialization” on page 70.




   a. Select **Cancel** to return to the home screen, then tap **Settings ►Clean Instrument**.

   b. Follow the on-screen instructions to start the cleaning.

      Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

   c. After the cleaning is complete, the instrument is ready to use.

## Instrument planned maintenance and performance qualification

The Genexus™ Dx Integrated Sequencer needs annual planned maintenance and displays a notification when planned maintenance is due. To schedule a planned maintenance visit, contact your Thermo Fisher Scientific Field Service Engineer.

Performance qualification is done at the time of sequencer installation by a Thermo Fisher Scientific Field Service Engineer, and needs to be performed annually thereafter. Performance qualification must also be done after a major version upgrade of Genexus™ Dx Software, and after a major repair.

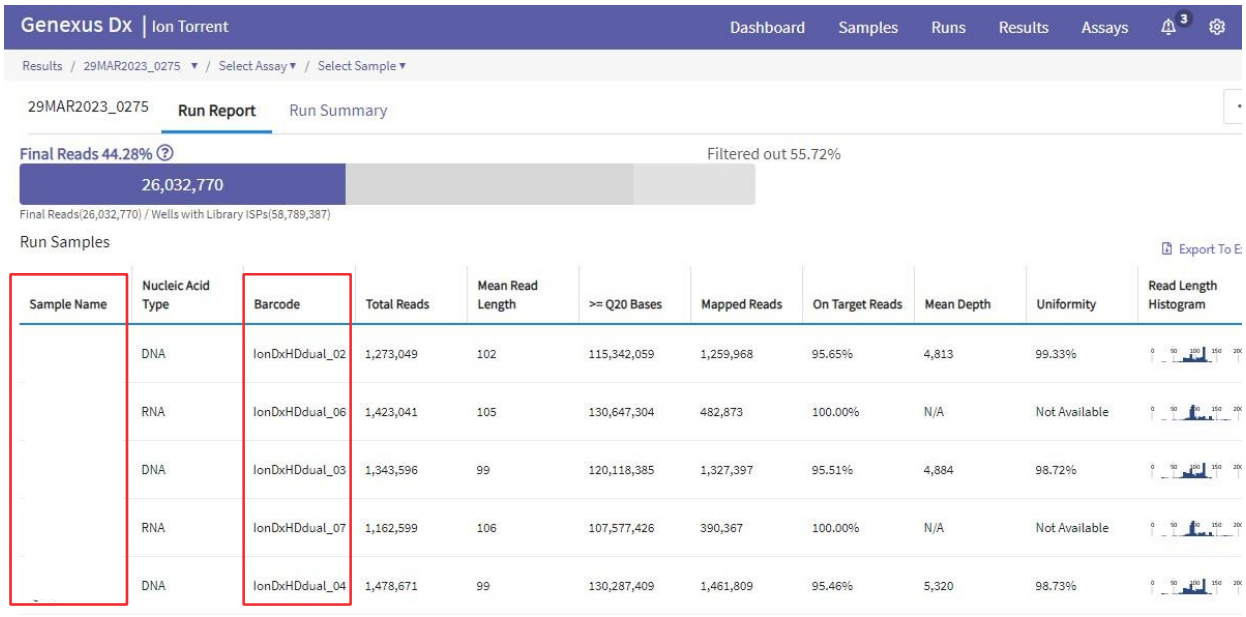
# Recover pooled sample libraries and plan a Library to Result run

If a sequencing run fails due to a templating or sequencing error, you can recover pooled sample libraries from well E4 of the PCR amplification plate from the run and use them in a subsequent Library to Result run. Follow these steps to plan a run and recover the pooled sample libraries.

## Prepare a library batch

If you are planning a run starting from pooled libraries that you have recovered from a previous sequencing run, you need to first prepare a library batch in Genexus™ Dx Software from samples that you have entered. For more detailed instructions, see “Prepare a library batch” on page 37.

To prepare a library batch, have at hand the sample names, HD barcodes, and assay used in the run that you want to repeat. This information can be found in the run report.




1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select all the samples included in the failed run, or contained in the E4 pool, by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.
3. In the **Create Library batch** screen, select the assay used in the previous run from the **Select Assay** dropdown list.
4. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.
5. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.
6. Select the DNA and RNA barcodes used for each sample as specified in the run report.
7. For the FFPE DNA and RNA Positive Controls and NTC, enter or scan with a barcode scanner the barcode from the control kit in the DNA and RNA **Control Kit Barcode** fields. For the NTC, enter or scan the barcode from the NTC kit in the DNA and RNA **Control Kit Barcode** fields.
8. Click **Submit** to create the library batch.
   The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Plan the Library to Result run

After creating the library batch, plan a new Library to Result run using the library batch with its associated assay. For more detailed instructions, see “Plan a Library to Result run” on page 49.

For a 4-lane FFPE DNA/RNA run (12 samples + 1 positive control + 1 NTC), a Library to Result run requires 140 µL of pooled library. If the volume recovered from this run is <140 µL, use nuclease-free water or low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required 140 µL.

For a 2-lane FFPE DNA/RNA run (6 samples + 1 positive control + 1 NTC), a Library to Result run requires 80 µL of pooled library. If the volume recovered from this run is <80 µL, use nuclease-free water or low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required 80 µL.

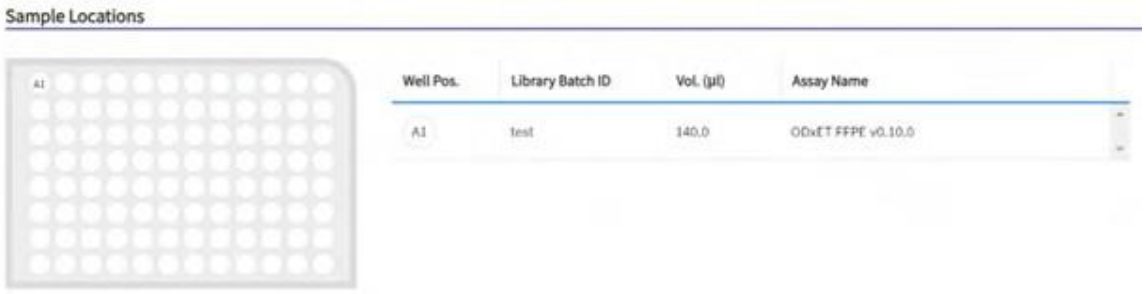
---

IMPORTANT! Run failure due to low loading can occur if insufficient pooled library is recovered and is too diluted. We recommend that you dilute recovered libraries no more than 2-fold when adjusting to the required volume.

---

1. In the menu bar, click **Runs►Library to Result**.
2. In the **Setup** step, enter a name for the run, configure the reporting options, then click **Next**.
3. In the **Assays** step, select the assay that you selected for the library batch, then click **Next**.
4. In the **Library Batches** step, select the library batch that you created, then click **Next**.
5. In the **Review** step, review the run setup guide generated for the sample library pool.

   IMPORTANT! Run failure due to low loading can occur if insufficient library pool is recovered from well E4 and is too diluted.




Proceed to “Recover the pooled sample libraries” to recover the sample library pool from well E4 of the PCR amplification plate.

## Recover the pooled sample libraries

After a run, pooled libraries are in well E4 of the PCR amplification plate for all workflows. The pooled sample libraries in well E4 are covered with a layer of oil. All the oil must be removed to avoid interference with downstream reactions.

IMPORTANT! The following procedure should be performed in a dedicated post-PCR laboratory space and/or laminar flow hood to decrease the possibility of spreading amplicon contamination.

1. After the run is complete, unload the plate from the sequencer deck. Seal the plate with an adhesive PCR plate foil (Cat. No. AB0626), then centrifuge the plate at 1,000 × *g* for 15–30 seconds.
2. Remove the foil, then collect the entire contents of well E4 with a pipette. Transfer the volume to a clear 0.5-ml or 1.5-ml microcentrifuge tube (low nucleic acid-binding).

   Note: A narrow tube is recommended to see the top layer of oil more easily.
3. Centrifuge the tube at 1,000 × *g* for 15–30 seconds, then carefully remove the top oil layer with a pipette. Repeat the centrifugation if needed until all the oil has been removed.
4. Determine the volume of the sample library pool with a pipette. If needed, add nuclease-free water or Low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required volume shown in the run setup guide, then add to well A1 of a new sample plate.

The sample plate is ready to load in the sequencer for a Library to Result run. Proceed to Chapter 7, “Start a sequencing run”.

# Genexus™ Dx Integrated Sequencer Install Kit

The Genexus™ Dx Integrated Sequencer Install Kit (Part No. A52169) is available to first-time owners of a Genexus™ Dx Integrated Sequencer and is shipped with the instrument. The kit contains the following reagents, supplies, and controls that are used during the installation, training, and operation of the instrument.

Genexus™ Dx Integrated Sequencer Install Kit

| Contents | Part No. | Quantity | Storage |
|---|---|---|---|
| Genexus™ Dx Integrated Sequencer PQ Kit<br>• Genexus™ Dx PQ Panel-HD<br>• Genexus™ Dx DNA PQ Control<br>• Genexus™ Dx PQ Library-HD | — | 1 kit | −30°C to −10°C |
| Genexus™ Dx Barcodes 1–32 HD | A50410 | 1 plate | 15°C to 30°C |
| Genexus™ Dx Strip 1 | A50862 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 2-HD | A50861 | 8 strips | −30°C to −10°C |
| Genexus™ Dx Strip 3-GX5™ | A50864 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 4 | A50863 | 8 strips | −30°C to −10°C |
| Genexus™ Dx Cartridge | A50865 | 2 cartridges | −30°C to −10°C |
| Genexus™ Dx GX5™ Chip and Genexus™ Coupler | A50425 | 2 each | 15°C to 30°C |
| Genexus™ Dx Bottles 1 and 3 | A50867 | 2 bottles each | |
| Genexus™ Dx Bottle 2 | A50866 | 4 bottles | |
| Genexus™ Dx Pipette Tips | A50426 | 12 racks | |
| Genexus™ Dx Filter | A50868 | 1 filter | |
| Genexus™ Dx Conical Bottles | A50869 | 2 sets of 5 bottles | |
| Adhesive PCR Plate Foils | AB0626 | 1 box (100 foils) | |

# Genexus™ Dx reagent strip and panel tube fill volume guide

You can use the figures below to check for appropriate fill volume of library and template strip tubes, and assay primer pool tubes. Print this page at 100% and keep as a reference. If a fill volume appears inaccurate, contact Technical Support to report the problem. "E" indicates an empty tube.




1. Genexus™ Dx Strip 1
2. Genexus™ Dx Strip 2-HD
3. Genexus™ Dx Strip 3-GX5™
4. Genexus™ Dx Strip 4
5. Assay primer pool tubes

E

# Safety


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WARNING! **GENERAL SAFETY.** Using this product in a manner not specified in the user documentation may result in personal injury or damage to the instrument or device. Ensure that anyone using this product has received instructions in general safety practices for laboratories and the safety information provided in this document.

- Before using an instrument or device, read and understand the safety information provided in the user documentation provided by the manufacturer of the instrument or device.
- Before handling chemicals, read and understand all applicable Safety Data Sheets (SDSs) and use appropriate personal protective equipment (gloves, gowns, eye protection, and so on). To obtain SDSs, visit **thermofisher.com/support**.

## Symbols on this instrument

Symbols may be found on the instrument to warn against potential hazards or convey important safety information. In this document, the hazard symbol is used along with one of the following user attention words.

- **CAUTION!**—Indicates a potentially hazardous situation that, if not avoided, may result in minor or moderate injury. It may also be used to alert against unsafe practices.
- **WARNING!**—Indicates a potentially hazardous situation that, if not avoided, could result in death or serious injury.
- **DANGER!**—Indicates an imminently hazardous situation that, if not avoided, will result in death or serious injury.

### Standard safety symbols

| Symbol and description |
|---|
| CAUTION! Risk of danger. Consult the manual for further safety information. |

*(continued)*

| Symbol and description |
|---|
|  CAUTION! Risk of electrical shock. |
|  CAUTION! Hot surface. |
|  CAUTION! Potential biohazard. |
|  CAUTION! Ultraviolet light. |

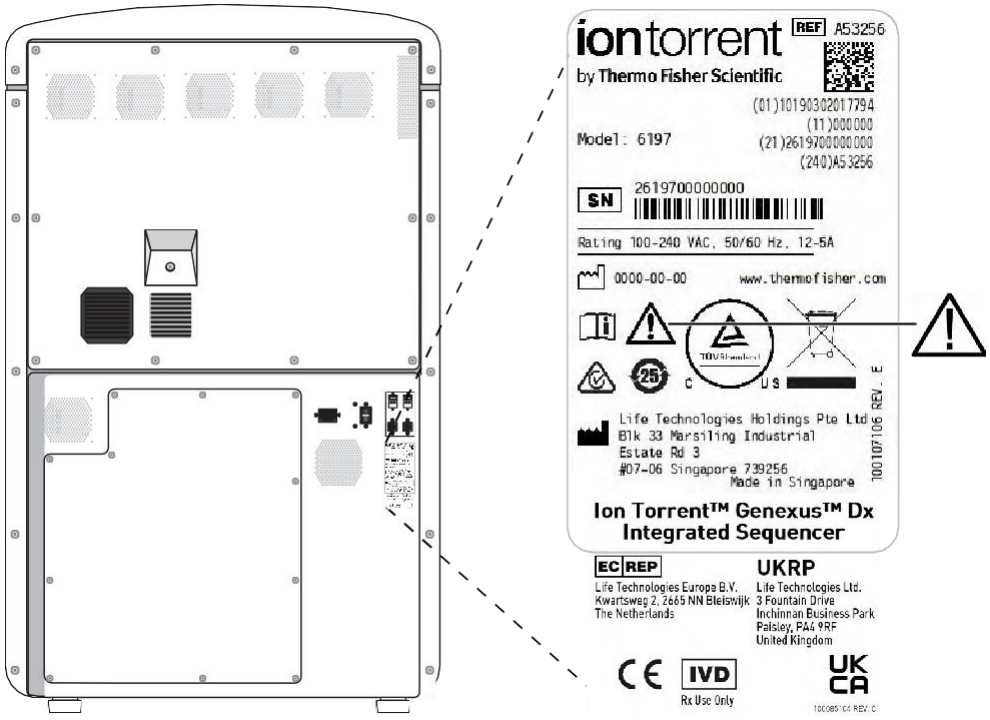
| Symbole et description |
|---|
|  MISE EN GARDE ! Risque de danger. Consulter le manuel pour d'autres renseignements de sécurité. |
|  MISE EN GARDE ! Risque de choc électrique. |
|  MISE EN GARDE ! Surface chaude. |
|  MISE EN GARDE ! Danger biologique potentiel. |
|  MISE EN GARDE ! Rayonnement ultraviolet. |

## Additional safety symbols

| Symbol and description |
|---|
| CAUTION! Moving parts. |
| CAUTION! Piercing hazard. |

| Symbole et description |
|---|
| MISE EN GARDE ! Parties mobiles. |
| MISE EN GARDE ! Danger de perforation. |

## Location of safety labels

| Label and location |
|---|
|  <br>Sequencer rear panel |

## Control and connection symbols

| Symbol | Description |
|---|---|
|  | On (Power) |
|  | Off (Power) |
|  | Protective conductor terminal (main ground) |
|  | Alternating current |

## Conformity symbols

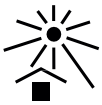
| Conformity mark | Description |
|---|---|
|  | Indicates conformity with safety requirements for Canada and U.S.A. |
|  | Indicates conformity with China RoHS requirements. |
|  | Indicates conformity with European Union requirements. |
|  | Indicates conformity with Australian standards for electromagnetic compatibility. |
|  | Indicates conformity with the WEEE Directive 2012/19/EU.<br><br>CAUTION! To minimize negative environmental impact from disposal of electronic waste, do not dispose of electronic waste in unsorted municipal waste. Follow local municipal waste ordinances for proper disposal provision and contact customer service for information about responsible disposal options. |

# Definition of symbols on product labels or product documents

The following table describes symbols that are present on product labels or product documents. Symbols conform to applicable international regulatory standards.

| Symbol | Description |
|---|---|
|  | MANUFACTURER |
|  | DATE OF MANUFACTURE (YYYY-MM-DD) |
|  | CONSULT ELECTRONIC INSTRUCTIONS FOR USE |
|  | CATALOGUE NUMBER |
|  | UNIQUE DEVICE IDENTIFIER |
|  | BATCH CODE |
|  | SERIAL NUMBER |
|  | CONTAINS SUFFICIENT FOR <n> TESTS |
|  | DO NOT REUSE |

*(continued)*

| Symbol | Description |
|---|---|
|  | USE-BY DATE |
|  | TEMPERATURE LIMIT |
|  | UPPER LIMIT OF TEMPERATURE |
|  | LOWER LIMIT OF TEMPERATURE |
|  | UPPER AND LOWER LIMITS OF HUMIDITY |
|  | KEEP AWAY FROM SUNLIGHT, KEEP AWAY FROM HEAT |
|  | IN VITRO DIAGNOSTIC MEDICAL DEVICE |
|  | PRESCRIPTION USE ONLY |
|  | READ SAFETY DATA SHEET (SDS) |
|  | BIOLOGICAL RISKS |
|  | OBSERVE PRECAUTIONS FOR HANDLING ELECTROSTATIC SENSITIVE DEVICES |

*(continued)*

| Symbol | Description |
|---|---|
|  | CAUTION, CONSULT ACCOMPANYING DOCUMENTS |
|  | FRAGILE, HANDLE WITH CARE |

# Instrument safety

## General



CAUTION! **Do not remove instrument protective covers.** If you remove the protective instrument panels or disable interlock devices, you may be exposed to serious hazards including, but not limited to, severe electrical shock, laser exposure, crushing, or chemical exposure.



MISE EN GARDE ! **Ne retirez pas les couvercles de protection de l'instrument.** Si vous retirez les panneaux de protection des instruments ou si vous désactivez les dispositifs de verrouillage, vous risquez de courir de graves dangers, comme, par exemple, un choc électrique, une exposition au laser, un écrasement ou une exposition à des produits chimiques.

## Physical injury



CAUTION! **Moving and Lifting Injury.** Improper lifting can cause painful and permanent back injury.

Things to consider before lifting or moving the instrument or accessories:

- Depending on the weight, moving or lifting may require two or more persons.
- If you decide to lift or move the instrument after it has been installed, do not attempt to do so without the assistance of others, the use of appropriate moving equipment, and proper lifting techniques.
- Ensure you have a secure, comfortable grip on the instrument or accessory.
- Make sure that the path from where the object is to where it is being moved is clear of obstructions.
- Do not lift an object and twist your torso at the same time. Keep your spine in a good neutral position while lifting with your legs.
- Participants should coordinate lift and move intentions with each other before lifting and carrying.
- For smaller packages, rather than lifting the object from the packing box, carefully tilt the box on its side and hold it stationary while someone else slides the contents out of the box.



MISE EN GARDE ! **Blessures causées par le déplacement et le soulèvement.** Soulever de manière inappropriée peut provoquer des lésions dorsales douloureuses et permanentes.

Éléments à prendre en compte avant de soulever ou de déplacer l'instrument ou ses accessoires:

- Selon le poids, deux personnes ou plus peuvent être nécessaires pour déplacer ou soulever l'instrument.
- Si vous décidez de soulever ou de déplacer l'instrument après son installation, n'essayez pas de le faire seul, sans un équipement approprié et sans avoir recours à des techniques appropriées.
- Assurez-vous d'avoir une prise sûre et confortable sur l'instrument ou l'accessoire.
- Assurez-vous que le chemin entre l'endroit où se trouve l'objet et l'endroit où il est déplacé est libre de tout obstacle.
- Ne soulevez pas un objet et ne pivotez pas votre torse en même temps. Tenez votre colonne vertébrale dans une position bien droite en vous relevant.
- Les participants doivent coordonner leurs mouvements avant de soulever et de porter.
- Pour les petits colis, au lieu de soulever l'objet de son emballage, inclinez soigneusement le carton sur le côté et maintenez-le immobile pendant que quelqu'un d'autre fait glisser le contenu hors du carton.



CAUTION! **Moving Parts.** Moving parts can crush, pinch and cut. Keep hands clear of moving parts while operating the instrument. Disconnect power before servicing.



MISE EN GARDE ! **Pièces mobiles.** Les pièces mobiles peuvent écraser, pincer et couper. Tenez les mains à l'écart des pièces mobiles lors de l'utilisation de l'instrument. Débranchez le câble d'alimentation avant d'entretenir l'unité.

## Electrical safety

WARNING! **Ensure appropriate electrical supply.** For safe operation of the instrument:

- Plug the system into a properly grounded receptacle with adequate current capacity.
- Ensure the electrical supply is of suitable voltage.
- Never operate the instrument with the ground disconnected. Grounding continuity is required for safe operation of the instrument.

AVERTISSEMENT ! **Veiller à utiliser une alimentation électrique appropriée.** Pour garantir le fonctionnement de l'instrument en toute sécurité :

- Brancher le système sur une prise électrique correctement mise à la terre et de puissance adéquate.
- S'assurer que la tension électrique est convenable.
- Ne jamais utiliser l'instrument alors que le dispositif de mise à la terre est déconnecté. La continuité de la mise à la terre est impérative pour le fonctionnement de l'instrument en toute sécurité.

WARNING! **Power Supply Line Cords.** Use properly configured and approved line cords for the power supply in your facility.

AVERTISSEMENT ! **Cordons d'alimentation électrique.** Utiliser des cordons d'alimentation adaptés et approuvés pour raccorder l'instrument au circuit électrique du site.

WARNING! **Disconnecting Power.** To fully disconnect power either detach or unplug the power cord, positioning the instrument such that the power cord is accessible.

AVERTISSEMENT ! **Déconnecter l'alimentation.** Pour déconnecter entièrement l'alimentation, détacher ou débrancher le cordon d'alimentation. Placer l'instrument de manière à ce que le cordon d'alimentation soit accessible.

## Cleaning and decontamination

CAUTION! **Cleaning and Decontamination.** Use only the cleaning and decontamination methods that are specified in the manufacturer user documentation. It is the responsibility of the operator (or other responsible person) to ensure that the following requirements are met:

- No decontamination or cleaning agents are used that can react with parts of the equipment or with material that is contained in the equipment. Use of such agents could cause a HAZARD condition.
- The instrument is properly decontaminated a) if hazardous material is spilled onto or into the equipment, and/or b) before the instrument is serviced at your facility or is sent for repair, maintenance, trade-in, disposal, or termination of a loan. Request decontamination forms from customer service.
- Before using any cleaning or decontamination methods (except methods that are recommended by the manufacturer), confirm with the manufacturer that the proposed method will not damage the equipment.

MISE EN GARDE ! **Nettoyage et décontamination.** Utiliser uniquement les méthodes de nettoyage et de décontamination indiquées dans la documentation du fabricant destinée aux utilisateurs. L'opérateur (ou toute autre personne responsable) est tenu d'assurer le respect des exigences suivantes:

- Ne pas utiliser d'agents de nettoyage ou de décontamination susceptibles de réagir avec certaines parties de l'appareil ou avec les matières qu'il contient et de constituer, de ce fait, un DANGER.
- L'instrument doit être correctement décontaminé a) si des substances dangereuses sont renversées sur ou à l'intérieur de l'équipement, et/ou b) avant de le faire réviser sur site ou de l'envoyer à des fins de réparation, de maintenance, de revente, d'élimination ou à l'expiration d'une période de prêt (des informations sur les formes de décontamination peuvent être demandées auprès du Service clientèle).
- Avant d'utiliser une méthode de nettoyage ou de décontamination (autre que celles recommandées par le fabricant), les utilisateurs doivent vérifier auprès de celui-ci qu'elle ne risque pas d'endommager l'appareil.

## Instrument component and accessory disposal

To minimize negative environmental impact from disposal of electronic waste, do not dispose of electronic waste in unsorted municipal waste. Follow local municipal waste ordinances for proper disposal provision and contact customer service for information about responsible disposal options.

# Safety and electromagnetic compatibility (EMC) standards

The instrument design and manufacture complies with the following standards and requirements for safety and electromagnetic compatibility.

## Safety standards

| Reference | Description |
|---|---|
| EU Directive 2014/35/EU | European Union low voltage directive |
| IEC 61010-1<br>EN 61010-1<br>UL 61010-1<br>CAN/CSA C22.2 No. 61010-1<br>GB 4793.1 (China) | *Safety requirements for electrical equipment for measurement, control, and laboratory use—Part 1: General requirements* |
| IEC 61010-2-010<br>EN 61010-2-010<br>GB 4793.6 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-010: Particular requirements for laboratory equipment for the heating of materials* |
| IEC 61010-2-020<br>EN 61010-2-020<br>GB 4793.7 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-020: Particular requirements for laboratory centrifuges* |
| IEC 61010-2-081<br>EN 61010-2-081<br>GB 4793.9 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-081: Particular requirements for automatic and semi-automatic laboratory equipment for analysis and other purposes* |
| IEC 61010-2-101<br>EN 61010-2-101<br>YY 0648 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-101: Particular requirements for in vitro diagnostic (IVD) medical equipment* |

## EMC standards

| Reference | Description |
|---|---|
| EU Directive 2014/30/EU | European Union EMC directive. |
| EN 61326-1<br>IEC 61326-1<br>GB/T 18268.1 (China) | *Electrical Equipment for Measurement, Control and Laboratory Use. EMC Requirements—Part 1: General Requirements* |
| EN 61326-2-6<br>IEC 61326-2-6 | *Electrical equipment for measurement, control and laboratory use—EMC requirements—Part 2-6: Particular requirements—In vitro diagnostic (IVD) medical equipment* |
| GB/T 18268.26 (China) | *Electrical equipment for measurement, control and laboratory use. EMC requirements. Part 26. Particular requirements. In vitro diagnostic (IVD) medical equipment* |
| FCC Part 18 (47 CFR) | *U.S. Standard for Industrial, Scientific, and Medical Equipment* |
| AS CISPR 11 | Industrial, scientific and medical equipment—Radio frequence disturbance characteristics—Limits and methods of measurement |
| ICES-001, Issue 4 | *Industrial, Scientific and Medical (ISM) Radio Frequency Generators* |
| FCC Part 15 Subpart B (47 CFR) | *U.S. Standard for Radio Frequency Devices*<br>This equipment has been designed and tested to CISPR 11 Class A. In a domestic environment it may cause radio interference, in which case, you may need to take measures to mitigate the interference.<br>Do not use this device in close proximity to sources of strong electromagnetic radiation (for example, unshielded intentional RF sources), because these can interfere with proper operation. |

## Environmental design standards

| Reference | Description |
|---|---|
| Directive 2012/19/EU | European Union WEEE Directive on waste electrical and electronic equipment. |
| Directive 2011/65/EU | European Union RoHS Directive on restriction of hazardous substances in electrical and electronic equipment. |
| SJ/T 11364-2014 | China RoHS Standard—Marking for the restriction of the use of hazardous substances in electronic and electrical products<br>For instrument specific certificates, visit our customer resource page at **www.thermofisher.com/us/en/home/technical-resources/rohs-certificates.html**. |

## Radio compliance standards

| Reference | Description |
|---|---|
| Directive 2014/53/EU | European Union: *RE Directive*—Radio equipment |
| RFID | FCC Notice (for U.S. Customers):<br>This device complies with Part 15 of the FCC Rules.<br>Operation is subject to the following conditions:<br>1. This device may not cause harmful interference, and<br>2. This device must accept any interference received, Including interference that may cause undesired operation.<br>Changes and modifications not expressly approved byThermo Fisher Scientific can void your authority to operate this equipment under Federal Communications Commissions rules. |
| RFID | Canada:<br>This device complies with Industry Canada license-exempt RSS standard(s). Operation is subject to the following two conditions:<br>(1) this device may not cause interference, and (2) this device must accept any interference, including interference that may cause undesired operation of the device. |
| RFID | Canada (Français québécois):<br>Le présent appareil est conforme aux CNR d'Industrie Canada applicables aux appareils radio exempts de licence. L'exploitation est autorisée aux deux conditions suivantes :<br>(1) l'appareil ne doit pas produire de brouillage, et (2) l'utilisateur de l'appareil doit accepter tout brouillage adioélectrique subi, même si le brouillage est susceptible d'en compromettre le fonctionnement. |
| Notice 423 [2005]<br>Industrial and Information Department Radio, BRR[1] | China:<br>Notice on issuing the technical requirements for micropower (short range) radio equipment. |
| GB/T 12572-2008 | China:<br>*Universal requirements and measurement methods of parameters for radio transmitting equipment* |
| ETSI EN 300 330-1 (2015-03) | China:<br>*Electromagnetic compatibility and Radio spectrum Matters (ERM); Short Range Devices (SRD); Radio equipment in the frequency range 9 kHz to 25 MHz and inductive loop systems in the frequency range 9 kHz to 30 MHz; Part 1 Technical characteristics and test methods* |

*(continued)*

| Reference | Description |
|---|---|
| No 1 [2014]1 of Industrial and Information Department Radio, BRR[1] | China:<br>*Methods for the administration of type approval of radio transmitting modules used in independent operation* |
| China: The Ministry of Industry and Information Technology (MIIT) issued a notice on micro-power short-range radio transmitting equipment. | |

[1] Bureau of Radio Regulation of the Ministry of Industry and Information Technology, State Radio Office

# Chemical safety

WARNING! **GENERAL CHEMICAL HANDLING.** To minimize hazards, ensure laboratory personnel read and practice the general safety guidelines for chemical usage, storage, and waste provided below. Consult the relevant SDS for specific precautions and instructions:

- Read and understand the Safety Data Sheets (SDSs) provided by the chemical manufacturer before you store, handle, or work with any chemicals or hazardous materials. To obtain SDSs, see the "Documentation and Support" section in this document.
- Minimize contact with chemicals. Wear appropriate personal protective equipment when handling chemicals (for example, safety glasses, gloves, or protective clothing).
- Minimize the inhalation of chemicals. Do not leave chemical containers open. Use only with sufficient ventilation (for example, fume hood).
- Check regularly for chemical leaks or spills. If a leak or spill occurs, follow the manufacturer cleanup procedures as recommended in the SDS.
- Handle chemical wastes in a fume hood.
- Ensure use of primary and secondary waste containers. (A primary waste container holds the immediate waste. A secondary container contains spills or leaks from the primary container. Both containers must be compatible with the waste material and meet federal, state, and local requirements for container storage.)
- After emptying a waste container, seal it with the cap provided.
- Characterize (by analysis if needed) the waste generated by the particular applications, reagents, and substrates used in your laboratory.
- Ensure that the waste is stored, transferred, transported, and disposed of according to all local, state/provincial, and/or national regulations.
- **IMPORTANT!** Radioactive or biohazardous materials may require special handling, and disposal limitations may apply.

AVERTISSEMENT ! **PRÉCAUTIONS GÉNÉRALES EN CAS DE MANIPULATION DE PRODUITS CHIMIQUES.** Pour minimiser les risques, veiller à ce que le personnel du laboratoire lise attentivement et mette en œuvre les consignes de sécurité générales relatives à l'utilisation et au stockage des produits chimiques et à la gestion des déchets qui en découlent, décrites ci-dessous. Consulter également la FDS appropriée pour connaître les précautions et instructions particulières à respecter :

- Lire et comprendre les fiches de données de sécurité (FDS) fournies par le fabricant avant de stocker, de manipuler ou d'utiliser les matériaux dangereux ou les produits chimiques. Pour obtenir les FDS, se reporter à la section « Documentation et support » du présent document.
- Limiter les contacts avec les produits chimiques. Porter des équipements de protection appropriés lors de la manipulation des produits chimiques (par exemple : lunettes de sûreté, gants ou vêtements de protection).
- Limiter l'inhalation des produits chimiques. Ne pas laisser les récipients de produits chimiques ouverts. Ils ne doivent être utilisés qu'avec une ventilation adéquate (par exemple, sorbonne).
- Vérifier régulièrement l'absence de fuite ou d'écoulement des produits chimiques. En cas de fuite ou d'écoulement d'un produit, respecter les directives de nettoyage du fabricant recommandées dans la FDS.
- Manipuler les déchets chimiques dans une sorbonne.

- Veiller à utiliser des récipients à déchets primaire et secondaire. (Le récipient primaire contient les déchets immédiats, le récipient secondaire contient les fuites et les écoulements du récipient primaire. Les deux récipients doivent être compatibles avec les matériaux mis au rebut et conformes aux exigences locales, nationales et communautaires en matière de confinement des récipients.)
- Une fois le récipient à déchets vidé, il doit être refermé hermétiquement avec le couvercle fourni.
- Caractériser (par une analyse si nécessaire) les déchets générés par les applications, les réactifs et les substrats particuliers utilisés dans le laboratoire.
- Vérifier que les déchets sont convenablement stockés, transférés, transportés et éliminés en respectant toutes les réglementations locales, nationales et/ou communautaires en vigueur.
- **IMPORTANT !** Les matériaux représentant un danger biologique ou radioactif exigent parfois une manipulation spéciale, et des limitations peuvent s'appliquer à leur élimination.



WARNING! **HAZARDOUS WASTE (from instruments).** Waste produced by the instrument is potentially hazardous. Follow the guidelines noted in the preceding General Chemical Handling warning.



WARNING! **4L Reagent and Waste Bottle Safety.** Four-liter reagent and waste bottles can crack and leak. Each 4-liter bottle should be secured in a low-density polyethylene safety container with the cover fastened and the handles locked in the upright position.

# Biological hazard safety



WARNING! **Potential Biohazard.** Depending on the samples used on this instrument, the surface may be considered a biohazard. Use appropriate decontamination methods when working with biohazards.



WARNING! **BIOHAZARD.** Biological samples such as tissues, body fluids, infectious agents, and blood of humans and other animals have the potential to transmit infectious diseases. Conduct all work in properly equipped facilities with the appropriate safety equipment (for example, physical containment devices). Safety equipment can also include items for personal protection, such as gloves, coats, gowns, shoe covers, boots, respirators, face shields, safety glasses, or goggles. Individuals should be trained according to applicable regulatory and company/ institution requirements before working with potentially biohazardous materials. Follow all applicable local, state/provincial, and/or national regulations. The following references provide general guidelines when handling biological samples in laboratory environment.

- U.S. Department of Health and Human Services, *Biosafety in Microbiological and Biomedical Laboratories (BMBL)*, 6th Edition, HHS Publication No. (CDC) 300859, Revised June 2020
  **www.cdc.gov/labs/pdf/CDC-BiosafetyMicrobiologicalBiomedicalLaboratories-2020-P.pdf**
- Laboratory biosafety manual, fourth edition. Geneva: World Health Organization; 2020 (Laboratory biosafety manual, fourth edition and associated monographs)
  **www.who.int/publications/i/item/9789240011311**

# Instrument warranty

For new Ion Torrent™ instruments, Life Technologies warrants to and only to buyer for twelve (12) months from the date of shipping, that the Ion Torrent™ software and Ion Torrent™ instruments are free from defects in material and workmanship and conform to Life Technologies' published specifications in all material respects. Where a valid and timely claim in respect of breach of Ion Torrent™ warranty is submitted to Life Technologies, Life Technologies may, at its discretion, replace, repair or modify the Ion Torrent™ instrument. Any agreed replacement shall be at 1:1, like-kind basis, at no cost to the buyer. For Ion Torrent™ chips or reagents reasonably determined by Life Technologies to be defective, independent of user error, shall be replaced by Life Technologies on a 1:1, like-kind basis at no cost to buyer, provided that such defective Ion Torrent™ chips or reagents were used by buyer prior to their expiration date, or if there is no expiration date, the Ion Torrent™ chips or reagents were used within six (6) months of receipt, and the defect was promptly reported with appropriate detail to Life Technologies' technical support.

NO OTHER WARRANTIES SHALL BE APPLICABLE TO ION TORRENT PRODUCTS (WHETHER OR NOT ANY FURTHER WARRANTY DOCUMENTATION MAY BE INCLUDED IN THE SHIPMENT), WITH THE EXCEPTION OF THIRD PARTY WARRANTIES WITH RESPECT TO THIRD PARTY PRODUCT. ANY THIRD PARTY PRODUCTS ARE NOT COVERED BY THIS SECTION AND ANY WARRANTIES FOR THIRD PARTY PRODUCTS ARE PROVIDED BY THE ORIGINAL MANUFACTURER OF THE THIRD PARTY PRODUCT. Warranties are made only to buyer purchasing the Ion Torrent™ Product directly from Life Technologies, are not transferable and do not extend to the benefit of any other person or entity, unless otherwise expressly stated in writing by Life Technologies. ANY PRODUCT NOT COVERED BY AN EXPRESS WRITTEN WARRANTY IS SOLD AND PROVIDED "AS IS," WITHOUT WARRANTY OF ANY KIND, STATUTORY, EXPRESS OR IMPLIED. Any description of Ion Torrent™ Product recited in Life Technologies' quotation is for the sole purpose of identifying Ion Torrent™ Product, and any such description is not part of any contract between Life Technologies and buyer and does not constitute a warranty that Ion Torrent™ Product shall conform to that description. Any sample or model used in connection with Life Technologies' quotation is for illustrative purposes only, and is not part of any contract between Life Technologies and buyer and does not constitute a warranty that Ion Torrent™ Product will conform to the sample or model. No affirmation of fact or promise made by Life Technologies, whether or not in Life Technologies' quotation, shall constitute a warranty that Ion Torrent™ Product will conform to the affirmation or promise. Unless otherwise specified in writing in documentation shipped with Ion Torrent™ Product or otherwise agreed by Life Technologies in writing. Life Technologies does not provide service or support for custom products or other products made to buyer's specifications. THE WARRANTIES IDENTIFIED IN THIS CLAUSE ARE LIFE TECHNOLOGIES' SOLE AND EXCLUSIVE WARRANTIES WITH RESPECT TO Ion Torrent™ PRODUCT AND ARE IN LIEU OF ALL OTHER WARRANTIES, STATUTORY, EXPRESS OR IMPLIED, ALL OF WHICH OTHER WARRANTIES ARE EXPRESSLY DISCLAIMED, INCLUDING WITHOUT LIMITATION ANY IMPLIED WARRANTY OF MERCHANTABILITY, FITNESS FOR A PARTICULAR PURPOSE, NON-INFRINGEMENT, OR REGARDING RESULTS OBTAINED THROUGH THE USE OF ANY PRODUCT (INCLUDING, WITHOUT LIMITATION, ANY CLAIM OF INACCURATE, INVALID OR INCOMPLETE RESULTS), WHETHER ARISING FROM A STATUTE OR OTHERWISE IN LAW OR FROM A COURSE OF PERFORMANCE, DEALING OR USAGE OF TRADE.

# Customer and technical support

Visit **thermofisher.com/support** for the latest in services and support, including:

- Worldwide contact telephone numbers
- Product support
- Order and web support
- Safety Data Sheets (SDSs; also known as MSDSs)

Additional product documentation, including user guides and Certificates of Analysis, are available by contacting Customer Support.

## Obtaining Certificates of Analysis

The Certificate of Analysis provides detailed quality control and product qualification information for each product. Certificates of Analysis are printed and shipped with the product.

## Obtaining Certificates of Conformance

The Certificate of Conformance provides information on conformance testing of each instrument provided with the system. Certificates of Conformance are shipped with the instrument, and are also available by contacting Customer Support at **thermofisher.com/support**.

## Serious incidents

Any serious incident that has occurred in relation to the device shall be reported to the manufacturer and the relevant regulatory authority in which the user and/or the patient is established.

ion torrent

# Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics

## USER GUIDE

for use with:

Genexus™ Dx Purification System

Genexus™ Dx Integrated Sequencer

Genexus™ Dx Software 6.8

**Publication Number** MAN0024908

**Revision** A

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
#07-06, Singapore 739256

Products manufactured at this site:

- Genexus™ Dx Purification System
- Genexus™ Dx Integrated Sequencer
- Genexus™ Dx Software

Life Technologies Corporation |
7335 Executive Way |
Frederick, Maryland 21704 USA

Products manufactured at this site:

- Oncomine™ Dx Express Test
- Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit
- Genexus™ Dx FFPE DNA/RNA Purification Combo Kit
- Genexus™ Dx Library Strips 1 and 2-HD
- Genexus™ Dx Templating Strips 3-GX5™ and 4
- Genexus™ Dx Sequencing Kit
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Pipette Tips
- Genexus™ Dx Filter
- Genexus™ Dx Conical Bottles

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.

**Revision history: MAN0024908 A (English)**

| Revision | Date | Description |
|---|---|---|
| A | 30 June 2025 | New IVD-labeled part 1 user guide for the Oncomine™ Dx Express Test. |

The information in this guide is subject to change without notice.

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# About this guide

## Purpose of this guide

This user guide describes the intended use, theory of operation, and performance characteristics of the Oncomine™ Dx Express Test when using the Genexus™ Dx Purification System or other IVD-labeled extraction methods, and the Genexus™ Dx Integrated Sequencer. This guide also includes the list of variants detected by the test.

## Oncomine™ Dx Express Test user guides

This user guide is part of a three-guide set.

- *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide*
- *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*
- *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*

All three guides are required to complete the entire Oncomine™ Dx Express Test workflow.

# 1 Product information

## Oncomine™ Dx Express Test

The Ion Torrent™ Oncomine™ Dx Express Test (Cat. No. A54103) is an in vitro diagnostic next-generation sequencing (NGS) test to detect somatic alterations in human DNA and RNA isolated from tumor specimens in formalin-fixed, paraffin-embedded (FFPE) tissue samples. Detection of these variants is performed using the Genexus™ Dx Integrated Sequencer.

For more information about the variants reported by the Oncomine™ Dx Express Test, see Appendix B, "Variants targeted by the Oncomine™ Dx Express Test".

## Intended use

The Oncomine™ Dx Express Test is a qualitative in vitro diagnostic test that uses targeted next-generation sequencing (NGS) technology to detect substitutions, insertions, and deletions in 42 genes from DNA, copy number variants (CNVs) in 10 genes from DNA, and fusions or splice variants in 18 genes from RNA isolated from formalin-fixed, paraffin-embedded (FFPE) tumor samples using the Genexus™ Dx Integrated Sequencer.

The test is intended to be used as companion diagnostic to identify patients who may benefit from treatment with the targeted therapy listed in Table 1 in accordance with the approved therapeutic product labeling.

**Table 1 Companion diagnostic indications**

| Tissue type | Gene | Variant | Targeted therapy |
|---|---|---|---|
| Non-small cell lung cancer (NSCLC) | EGFR | EGFR exon 20 insertions | ZEGFROVY™ (sunvozertinib) |

Additionally, the test is intended to provide tumor mutation profiling information to be used by qualified health care professionals in accordance with professional guidelines in oncology for cancer patients with solid malignant neoplasms. Genomic findings other than those listed in Table 1 are not prescriptive or conclusive for labeled use of any specific therapeutic product.

# Theory of operation

## Overview

Protease-treated FFPE tissue sections are loaded into the Genexus™ Dx Purification System, along with consumables needed for automated purification and quantification of DNA and RNA. The instrument loads the purified and quantified samples into an output plate. After addition of positive control DNA and RNA, and no-template control, the output sample plate is loaded in the Genexus™ Dx Integrated Sequencer. The sequencer is then loaded with consumables needed for the sequencing run, including the Oncomine™ Dx Express Test panel, which targets the variants and gene fusions of interest in the test.

During the automated sequencer run, Ion AmpliSeq™ HD libraries are prepared from the samples and controls. The libraries, together with Ion Sphere™ Particles (ISPs), are loaded and templated in the sequencing chip, and the chip is sequenced according to run parameters specified by the assay that is selected in the run plan.

The signal generated by the sequencing reaction is translated into base calls and then reads, which are mapped to a reference sequence. Using analysis parameters in the assay designed for a particular set of targets, Genexus™ Dx Software generates reports that contain a summary of the sample test results.

The Ion Torrent™ Oncomine™ Dx Express Test reports targeted variants from the following genes:

**Table 2 Alterations reported by the Oncomine™ Dx Express Test**

| Alteration type | Genes |
|---|---|
| Deletions, insertions, and substitutions from DNA | AKT1, AKT2, AKT3, ALK, AR, ARAF, BRAF, CDK4, CHEK2, CTNNB1, EGFR, ERBB2, ERBB3, ERBB4, ESR1, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, GNAS, HRAS, IDH1, IDH2, KEAP1, KIT, KRAS, MAP2K1, MAP2K2, MET, NRAS, NTRK1, NTRK2, NTRK3, PDGFRA, PIK3CA, PTEN, RAF1, RET, ROS1, STK11, TP53 |
| Copy number variants from DNA | AR, EGFR, ERBB2, ERBB3, FGFR1, FGFR2, FGFR3, KRAS, MET, PIK3CA |
| Fusions from RNA | ALK, AR, BRAF, EGFR, ESR1, FGFR1, FGFR2, FGFR3, MET, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, RET, ROS1, RSPO2, RSPO3 |

## Run plan creation

Information about each sample is entered into Genexus™ Dx Software, which directs the progress of the sample from creation of a run plan through automated sample preparation, library preparation, template preparation, sequencing, and analysis. During creation of a run plan, samples are selected and assigned to the run plan. The assay selected in the run plan specifies instrument run and analysis parameters, and directs the reporting output of sample results.

## Sample preparation

Specimens are treated with protease, then loaded into a sample input plate. The Genexus™ Dx Purification System is loaded with the sample plate and consumables specific for the sample and run type. During the automated run, the instrument purifies samples using a magnetic bead-based procedure, quantifies sample concentration, then loads the purified samples in a sequencer-ready output plate. Other IVD-labeled sample extraction methods are acceptable.

The recommended number of slide-mounted 5-µm FFPE sections or curls used in extraction varies depending on the sample collection method:

**Table 3 Sample input recommendations**

| Sample type | Recommended input |
|---|---|
| FFPE tumor resections | 2 × 5-µm curls/slides or equivalent amount |
| FFPE core needle biopsy (CNB) | 9 × 5-µm curls/slides or equivalent amount |
| FFPE fine needle aspirate (FNA) | 7 × 5-µm curls/slides or equivalent amount |

**Table 4 Sample concentration requirements**

| Sample type | Validated nucleic acid concentration | Instrument supported concentration range |
|---|---|---|
| FFPE DNA | 0.5 ng/µL | 0.5–512 ng/µL |
| FFPE RNA | | |

## Library preparation

In the first stage of a Genexus™ Dx Integrated Sequencer run, cDNA is prepared from sample RNA, then sample and control amplicon libraries are prepared from DNA or cDNA. Oncomine™ Dx Express Test panel primers, reagents in Genexus™ Dx Strip 1 and Genexus™ Dx Strip 2-HD, and barcode primers from the Genexus™ Dx Barcodes 1–32 HD plate are used in library preparation. Each library that is generated in a run has a distinguishing nucleic acid sequence barcode that is incorporated into each amplified molecule.

## Template preparation

In the template preparation stage of the automated Genexus™ Dx Integrated Sequencer run, library molecules are bound onto ISPs and each nucleic acid sequence is amplified over the ISP surface using reagents in the Genexus™ Dx Strip 3-GX5™ and Genexus™ Dx Strip 4. The templated ISPs are enriched and collected using magnetic beads, then the enriched ISPs are magnetically loaded into wells of the sequencing chip. A second amplification step is then carried out with ISPs loaded on the chip, one ISP per well. After amplification, sequencing primer is annealed to the single-stranded template, and sequencing enzyme is added.

## Sequencing

In the sequencing stage of the run, the sequencer flows one of four nucleotides over the Genexus™ Dx GX5™ Chip surface in a programmed series of flows. A base that is incorporated into the newly synthesized strands of an ISP during a flow results in a release of protons and a concomitant pH change in the well. The change in pH is detected by sensors at the base of each well on the chip. Electrical signals that correspond to pH changes generated in each well during sequencing are processed and stored in data files on the system.

Throughout this procedure, as the sample is processed by the instrument, sample and reagent information is recorded and tracked by Genexus™ Dx Software.

## Data analysis

On the internal server of the Genexus™ Dx Integrated Sequencer, the initial signals are processed and each base in a sequence is called (identified). Base calls are assembled into files representing the reads, which are strings of nucleotide bases in the order found in the original library molecules. The reads are then mapped to the reference files provided with the test. Finally, Genexus™ Dx Software assesses the mapped reads at specific nucleotide locations and detects variation in sequence information in comparison to a human reference sequence.

## Results

Using parameters specified in the Oncomine™ Dx Express Test Assay Definition File (ADF), Genexus™ Dx Software generates the following electronic results and reports for each sequenced sample and its associated controls.

**Table 5 Electronic results and reports generated by the software**

| Results/report | Description |
|---|---|
| QC, Key Findings, and Variants screens | Display quality control (QC) and reference information, detailed sequencing analytics, and all DNA variant, gene fusion, and copy number variant (CNV) calls. |
| Run Report | A summary of run results and information, status, reads per barcode, and reagents used. |
| Clinical Test Report | A report that lists the CDx biomarkers for any associated therapies included in Table 1 of the “Intended use”, as well as the tumor profiling variants, including DNA variants (SNVs, insertions, deletions, and CNVs), and RNA variants (gene fusions and splice variants), detected in the FFPE sample screened by the Oncomine™ Dx Express Test. No calls and negative calls are also listed. Positive calls are classified into 3 biomarker levels:<br>• Level 1: Companion Diagnostics (CDx) Biomarkers noted in Table 1 of the “Intended use”.<br>• Level 2: Cancer Mutations with Evidence of Clinical Significance<br>• Level 3: Cancer Mutations with Potential Clinical Significance |
| Clinical Laboratory Report | Contains all the information in the Clinical Test Report. In addition, QC metrics for the run, sequencing run details, reagents used, and kit barcodes are also listed. |

These reports are subject to approval by a laboratory manager or administrator via electronic signature.

# Contraindications

There are no known contraindications.

# Assay warnings and limitations

1. Use of this product must be limited to personnel trained in the techniques of PCR, NGS, and the use of the Oncomine™ Dx Express Test, the Genexus™ Dx Purification System, when applicable, and the Genexus™ Dx Integrated Sequencer.
2. Users are cautioned that DNA variant-positive calls in a small number of genomic regions have been observed to generate multiple variant calls with overlapping alleles, even when only one variant is present. For details on these observed cases, see Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”.
3. Due to the nature of hotspot variant calling, when several variants are located in the same genomic region in close proximity to each other, the presence of a positive variant can result in a "No Call" for the neighboring variants instead of a negative call.
4. Genomic findings listed in the Oncomine™ Dx Express Test results report under Cancer Mutations with Evidence of Clinical significance (Level 2) and Cancer Mutations with Potential Clinical Significance (Level 3) are not prescriptive or conclusive for labeled use of any specific therapeutic product, and clinical validation has not been performed. Confirmation of tumor mutation status using an FDA-approved CDx test is needed for therapeutic use.
5. Decisions on patient care and treatment must be based on the independent medical judgment of the treating physician, taking into consideration all applicable information concerning the patient’s condition, such as patient and family history, physical examinations, information from other diagnostic tests, and patient preferences, in accordance with the standard of care in a given community.
6. The Oncomine™ Dx Express Test is for use with the Genexus™ Dx Integrated Sequencer and the Genexus™ Dx Purification System (for integrated workflows), or other IVD-labeled extraction chemistries.
7. The Oncomine™ Dx Express Test is for detecting the following somatic mutations: single-nucleotide variants (SNVs), insertions, deletions, and copy number variants (CNVs) in DNA, and fusions and splice variants in RNA.
8. The Oncomine™ Dx Express Test has been validated to detect and report only the variants listed in Appendix B, “Variants targeted by the Oncomine™ Dx Express Test”, and has not been validated for novel variant detection.
9. The Oncomine™ Dx Express Test is for use with 10 ng each of DNA and RNA per sample for FFPE samples. Input levels less than or greater than this amount are not recommended.
10. Although the Genexus™ Dx Integrated Sequencer can perform on-board dilution of samples up to 1,024-fold, the use of samples at the upper limit of supported concentrations (512 ng/µL for FFPE DNA and RNA) has not been validated.

11. Results for samples with DNA or RNA concentration failure are reported for Nucleic Acid to Result runs. A warning message appears in the run planning screen and the run summary screen to indicate to the user that a sample does not meet the minimum concentration requirement of the assay.
12. The Oncomine™ Dx Express Test is for use with FFPE tumor specimens.
13. Core needle biopsy (CNB) and fine needle aspirate (FNA) cell block specimen types have not been analytically validated for the detection of tumor profiling variants targeted by Oncomine™ Dx Express Test.
14. The effects of potential variations in FFPE specimen fixation have not been evaluated.
15. A potential source of contamination in the procedure is nucleic acid from previous sample extraction steps. Follow good laboratory practices and all precautions and guidelines in these user guides to avoid cross-contamination between samples.
16. The Oncomine™ Dx Express Test is a qualitative test. The test is not for quantitative measurements of percent mutation.
17. The test is not indicated to be used for standalone diagnostic purposes, screening, monitoring, risk assessment, or prognosis.
18. For prescription use only. The test must be ordered by a qualified medical professional in accordance with clinical laboratory regulations.
19. The performance of the Oncomine™ Dx Express Test in samples obtained from patients that have had organ or tissue transplantation has not been evaluated.
20. A negative result does not rule out the presence of a mutation below the Limits of Detection (LoD) of the assay in the patient's tumor and should be followed by additional confirmatory testing.
21. Alterations at allele frequencies below the LoD of the assay may not be detected consistently.
22. The minimum tumor content thresholds for Oncomine™ Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.
23. For optimal CNV results with FFPE samples that have a tumor content of <40%, macrodissect and enrich the samples for tumor content. For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is <20% and the tumor content in the region of interest is ≥10%. For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.
24. The ERBB2 copy number alteration call may not be accurate when tumor content is between 20–39%. Additional confirmatory testing with a clinically validated test is strongly recommended.
25. The presence of >70% necrotic tissue may yield unreliable sequencing results for the detection of EGFR exon 20 insertion mutations.
26. For fusion variants detected by expression imbalance alone (RNA exon tile fusion imbalance), confirmatory testing using a clinically validated assay is recommended.
27. The accuracy of Oncomine™ Dx Express Test for detecting gene fusions in FGFR1, NTRK2, NUTM1, RSPO2, and RSPO3 genes has not been evaluated.
28. When panel reproducibility was evaluated using extracted RNA samples, the ALK fusion imbalance in NSCLC demonstrated a low negative percent agreement of 28.57% (144/504).
29. When panel reproducibility was evaluated using extracted DNA samples, the BRAF V600E variant in colorectal cancer demonstrated a low positive percent agreement of 86.11% (62/72).

# Genexus™ Dx Integrated Sequencer hardware configuration

The Oncomine™ Dx Express Test is approved for use with the following components of the Genexus™ Dx Integrated Sequencer: A51151 FRU and A40001474.

# Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software version 6.8.2 or later. To view the currently installed software version, sign in to the software as an Administrator, click the **(Settings)** menu, then select **Software Updates**. Version-specific information is provided in the software release notes for the version of the software you are using.

Genexus™ Dx Software is supported on Google Chrome™ browser version 90 and later, and is best viewed with 1440 × 900 screen resolution. It has not been tested with other browsers. For a complete description of software compatibility and requirements, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910).

# Oncomine™ Dx Express Test panel, controls, purification and sequencing reagents, and supplies

The following reagents and supplies are available for order as needed. For detailed contents and storage information, see the *Oncomine™ Dx Express Test Part II: Sequencing and Results Reports User Guide* (Pub. No. MAN0024909).

**Table 6 Oncomine™ Dx Express Test components**

| ✓ | Component | Catalog or Part No. |
|---|---|---|
| **Panel and controls** | | |
| | Oncomine™ Dx Express Test Panel | A54103 |
| | Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit | A52167 |
| **Genexus™ Dx FFPE DNA/RNA Purification Combo Kit** | | |
| | Genexus™ Dx FFPE DNA and RNA Purification | A51076 |
| | Genexus™ Dx Nucleic Acid Quantitation | A51078 |
| | Genexus™ Dx Purification Supplies 2 | A51081 |
| **Genexus™ Dx sequencing reagents and supplies** | | |
| | Genexus™ Dx Library Strips 1 and 2-HD | A50430 |
| | Genexus™ Dx Templating Strips 3-GX5™ and 4 | A50431 |
| | Genexus™ Dx Sequencing Kit | A50432 |
| | Genexus™ Dx Barcodes 1–32 HD | A54104 |

**Table 6 Oncomine Dx Express Test components** *(continued)*

| ✓ | Component | Catalog or Part No. |
|---|---|---|
| | Genexus™ Dx GX5™ Chip and Genexus™ Coupler | A54106 |
| | Genexus™ Dx Pipette Tips | A54105 |
| | Genexus™ Dx Filter | A50868 |
| | Genexus™ Dx Conical Bottles | A50869 |

# 2 Pass/fail criteria and repeat strategy

## Quality control pass/fail criteria—FFPE samples (DNA and RNA)

**Table 7 FFPE—Quality Control (QC) specifications for Templating Control, Sample, NTC (No-Template Control), and Purification**

| Metric | Criteria |
|---|---|
| **Templating Control QC - CF-1** | |
| Average Reads per Lane | ≥2383 |
| Mean AQ20 Read Length (bp) | ≥82 |
| **Sample QC - DNA** | |
| MAPD | ≤0.5 |
| Mapped Reads | ≥100,000 |
| Mean AQ20 Read Length (bp) | ≥64 |
| **Sample QC - RNA** | |
| Mapped Fusion Reads | ≥6,000 |
| RNA Expression Controls Detected | ≥5 |
| **NTC QC - DNA** | |
| Hotspot Calls | 0 |
| Average Base Coverage Depth | ≤15 |
| **NTC QC - RNA** | |
| Mapped Reads | ≤2848 |
| Total Fusion Calls | 0 |
| **Purification QC** | |
| Sample Concentration DNA | 0.5 ng/µL minimum – 512 ng/µL maximum |
| Sample Concentration RNA | 0.5 ng/µL minimum – 512 ng/µL maximum |

**Table 8 FFPE—Positive Control QC specifications**

| Metric | Criteria |
|---|---|
| **Positive Control QC - DNA** | |
| COSM1072523_AF | Variant called and AF ≥0.0511 |
| COSM12376_AF | Variant called and AF ≥0.0200 |
| COSM12428_AF | Variant called and AF ≥0.0529 |
| COSM12558_AF | Variant called and AF ≥0.0200 |
| COSM133167_AF | Variant called and AF ≥0.0250 |
| COSM144252_AF | Variant called and AF ≥0.0200 |
| COSM256046_AF | Variant called and AF ≥0.0489 |
| COSM28055_AF | Variant called and AF ≥0.0423 |
| COSM28747_AF | Variant called and AF ≥0.0448 |
| COSM33765_AF | Variant called and AF ≥0.0250 |
| COSM41590_AF | Variant called and AF ≥0.0320 |
| COSM476_AF | Variant called and AF ≥0.0394 |
| COSM48358_AF | Variant called and AF ≥0.0539 |
| COSM516_AF | Variant called and AF ≥0.0250 |
| COSM5219_AF | Variant called and AF ≥0.0555 |
| COSM6223_AF | Variant called and AF ≥0.0348 |
| COSM6224_AF | Variant called and AF ≥0.0471 |
| COSM6240_AF | Variant called and AF ≥0.0250 |
| COSM683_AF | Variant called and AF ≥0.0297 |
| COSM775_AF | Variant called and AF ≥0.0261 |
| COSM962_AF | Variant called and AF ≥0.0309 |
| OMINDEL1177_AF | Variant called and AF ≥0.0334 |
| OMINDEL698_AF | Variant called and AF ≥0.0456 |
| ERBB2 Copy Number | ≥5.0 |
| MET Copy Number | ≥7.0 |
| MAPD | ≤0.50 |
| Mapped Reads | ≥100,000 |

**Table 8 FFPE—Positive Control QC specifications** *(continued)*

| Metric | Criteria |
|---|---|
| Mean AQ20 Read Length | ≥76 |
| **Positive Control QC - RNA** | |
| NTRK2 Targeted Isoform | POSITIVE |
| BRAF Targeted Isoform | POSITIVE |
| AR Targeted Isoform | POSITIVE |
| NUTM1 Targeted Isoform | POSITIVE |
| RET Targeted Isoform | POSITIVE |
| NRG1 Targeted Isoform | POSITIVE |
| EGFR Targeted Isoform | POSITIVE |
| RSPO2 Targeted Isoform | POSITIVE |
| ALK Targeted Isoform | POSITIVE |
| NTRK3 Targeted Isoform | POSITIVE |
| FGFR1 Targeted Isoform | POSITIVE |
| FGFR2 Targeted Isoform | POSITIVE |
| FGFR3 Targeted Isoform | POSITIVE |
| MET Targeted Isoform | POSITIVE |
| RSPO3 Targeted Isoform | POSITIVE |
| ROS1 Targeted Isoform | POSITIVE |
| NTRK1 Targeted Isoform | POSITIVE |
| FGFR2 Expression Imbalance | POSITIVE |
| FGFR3 Expression Imbalance | POSITIVE |
| NTRK1 Expression Imbalance | POSITIVE |
| Mapped Fusion Reads | ≥50,475 |
| RNA Expression Controls Detected | 7 (out of 7) |

# Retest strategy

In general, if you experience a sample failure, repeat the sample preparation at the workflow step in which the failure occurred. The number of libraries that can be made from an extracted sample for repeat testing depends on the quantity of DNA or RNA from the Genexus™ Dx Purification System run, which varies from sample to sample.

Based on quality control results, you can determine whether a sample library requires repeat testing. Refer to the following flowchart and tables to determine the best course of action. See Table 9 for pass/fail and call specifications for FFPE samples. See “Quality control logic and variant reporting” on page 21 for information to guide retest decisions given the specific QC failure that is observed in a run or for a sample. P: Pass; F: Fail; No Call: the call does not pass one or more filters that are needed for a high confidence variant call.

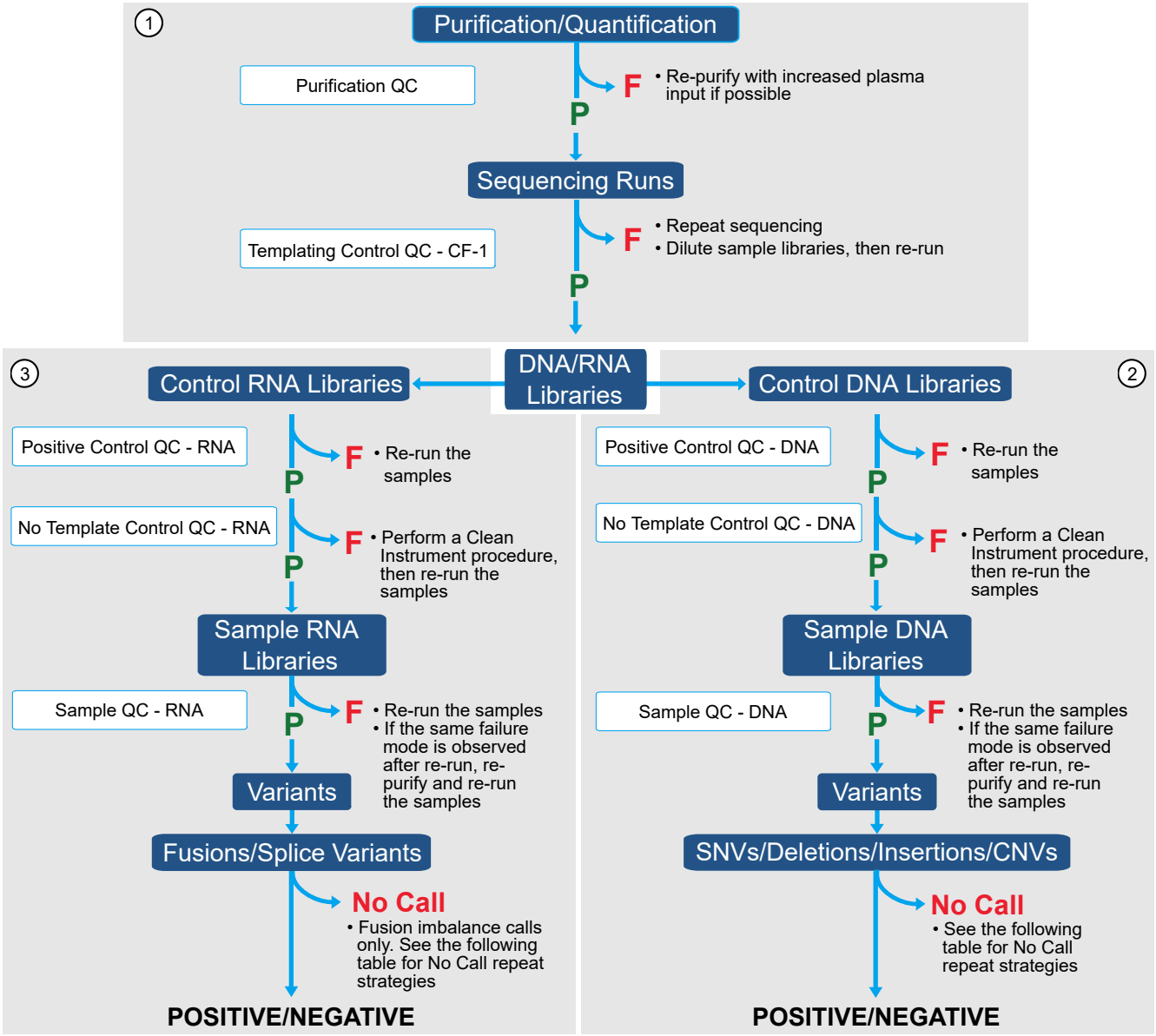



**Figure 1 Repeat strategy for FFPE samples (DNA and RNA)**

**Table 9 Retest strategy for FFPE samples**

| Point of failure | Specification type | Passing criteria | Repeat strategy |
|---|---|---|---|
| 1 | Purification QC | The following minimum concentrations for DNA, RNA, (FFPE) are required:<br>• DNA ≥0.5 ng/µL<br>• RNA ≥0.5 ng/µL | If the minimum concentration requirement is not met for DNA or RNA, the samples must be re-purified using increased tissue input. For FFPE samples, use the set of purified samples where both the DNA and RNA meet the minimum concentration requirement for the test. |
| 1 | Templating Control QC - CF-1 | A run must pass the following specifications to have reportable results for any sample in the run:<br>• CF-1 Mean AQ20 Read Length (bp) must be ≥82<br>• CF-1 Average reads per lane must be ≥2383 | If either CF-1 specification fails, repeat the sequencing run with the same sample set. If libraries have been recovered after the run, the operator can sequence them in a Library to Result run. If either CF-1 specification fails while sample reads are normal or high, dilute the recovered pooled sample libraries before the Library to Result run. For more information, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*. |
| 1 | No Template Control QC (NTC QC) | A run must pass the following NTC specifications to have reportable results for any samples in the run:<br>NTC QC - DNA<br>• Total hotspot calls must = 0<br>• Average base coverage depth must be ≤15<br>NTC QC - RNA<br>• Mapped reads must be ≤2848<br>• Total fusion calls = 0 | If either or both of the NTC QC - DNA and NTC QC - RNA fail, perform a Clean instrument procedure (see "Perform a Clean instrument procedure" in the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*).<br>**Note:** A failed NTC QC - DNA allows reporting of RNA variants for samples in a run, and a failed NTC QC - RNA allows reporting of DNA variants. If either the NTC QC - DNA or the NTC QC - RNA fails in a run, it is not necessary to retest the sample type with a passing NTC QC. For further details, see “Quality control logic and variant reporting” on page 21. |

**Table 9 Retest strategy for FFPE samples** *(continued)*

| Point of failure | Specification type | Passing criteria | Repeat strategy |
|---|---|---|---|
| 2 | Positive Control QC - DNA | The Positive Control QC - DNA must pass the following specifications for DNA samples in the run to have reportable results:<br>• MAPD ≤0.5<br>• Mapped reads ≥100,000<br>• AQ20 Mean Read Length (bp) ≥76<br>• All variants in the Positive Control - DNA must be called "Positive" and pass the allelic frequency threshold for each variant as specified in the assay definition file.<br>• ERBB2 copy number ≥5.0<br>• MET copy number ≥7.0 | If any of these specifications fail, the operator should re-run the DNA samples and verify that 20 µL of the Oncomine™ Dx Express Test FFPE DNA Control is added in the position on the sample plate specified by the run setup guide.<br>**Note:** A failed Positive Control QC - DNA allows reporting of RNA variants. If the Positive Control QC - DNA fails, it is not necessary to retest the RNA for all samples in a run. Similarly, failed CNV QC thresholds allow reporting of SNVs and INDELs, and failed SNV/INDEL QC thresholds allow reporting of CNVs. For further details, see “Quality control logic and variant reporting” on page 21. |
| 3 | Positive Control QC - RNA | The Positive Control QC - RNA must pass the following specification for RNA samples in the run to have reportable results:<br>• Targeted isoforms in the RNA control sample must be called "Positive".<br>• Expression imbalance controls in the RNA control sample must be called "Positive".<br>• RNA expression controls detected =7<br>• Mapped fusion reads ≥50,475 | If any of these specifications fail, the operator should re-run the RNA samples and verify that 20 µL of the Oncomine™ Dx Express Test FFPE RNA Control is added in the position on the sample plate specified by the run setup guide.<br>**Note:** A failed Positive Control QC - RNA allows reporting of DNA variants. If the Positive Control QC - RNA fails, it is not necessary to retest the DNA for all samples in a run. Similarly, failed detection of expression imbalance control genes allows reporting of targeted fusion isoforms, and failed targeted isoform control genes allows reporting fusions through expression imbalance. For further details, see “Quality control logic and variant reporting” on page 21. |
| 2 | Sample QC - DNA | Any individual DNA sample library must meet the following specifications to have reportable results for the DNA sample library.<br>• Mapped reads ≥100,000<br>• MAPD ≤0.5<br>• Mean AQ20 Read Length (bp) ≥64 | If any of these specifications fail, the operator should re-run the sample, or re-purify the sample using increased input material, if possible.<br>**Note:** A failed Sample QC - DNA allows reporting of RNA variants in the sample. If the Sample QC - DNA fails for a sample, it is not necessary to retest the RNA for the sample. For further details, see “Quality control logic and variant reporting” on page 21. |

**Table 9 Retest strategy for FFPE samples** *(continued)*

| Point of failure | Specification type | Passing criteria | Repeat strategy |
|---|---|---|---|
| 3 | Sample QC - RNA | Any individual RNA sample library must meet the following specifications to have reportable results for the RNA sample library.<br>• Mapped fusion reads ≥6,000<br>• RNA Expression Controls detected ≥5 | Re-run the sample alone, or with fewer samples. If the RNA sample library still fails this specification, re-purify using more tissue input, if possible.<br>**Note:** A failed Sample QC - RNA allows reporting of DNA variants in the sample. If the Sample QC - RNA fails for a sample, it is not necessary to retest the DNA for the sample. For further details, see "Quality control logic and variant reporting" on page 21. |
| 2 | SNV, Deletion, Insertion, and CNV specifications | Single Nucleotide Variants (SNVs), Insertions/Deletions (INDELs) and Copy Number Variants (CNVs) must meet allelic frequency or molecular coverage minima and pass all Variant Caller filtering metrics to have a reportable result for the variant.[1]<br>• SNV allelic frequency ≥2.5%<br>• Deletion/Insertion allelic frequency ≥2%<br>• Copy number ≥4 | SNVs, deletions, insertions, and CNVs that do not meet the coverage criteria can result in a "No Call" for the variant. The operator may run the sample alone or with fewer samples to obtain reportable results for the variant. If the repeat run fails to meet the minimum coverage requirement, the operator may re-purify the sample using increased input material to obtain reportable results for the variant. For more information about retest strategies for No Calls, see "Variant call filters and No Call repeat strategies" on page 23. |
| 3 | Targeted fusion isoform specifications | Targeted fusion isoforms must meet the following read or family count minima and pass all Variant Caller filtering metrics to have a reportable result for the variant.[2]<br>• Fusion Read Count ≥21, or<br>• Molecular Family Count ≥3 | For targeted fusion isoforms, fusion calls are Positive or Negative only - No Calls are not reported. |
| 3 | Fusion imbalance specifications | Fusion calls from fusion imbalance exon tile analysis must pass gene level QC, defined in the assay definition file, to have a reportable result for the variant. For a list of genes available for fusion imbalance exon tile analysis, see "Fusions targeted by RNA exon tile fusion imbalance analysis" on page 207. | An imbalance fusion that does not pass gene level QC criteria can result in a "No Call" for the variant. Running the sample alone or with fewer samples sequencing is not likely to change the result. For more information about retest strategies for No Calls, see "Variant call filters and No Call repeat strategies" on page 23. |

[1] Some variants have different allelic frequency thresholds that are defined in the assay definition file.

[2] Several fusions have different Read Count and Molecular Family Count thresholds that are defined in the assay definition file.

# Quality control logic and variant reporting

The following table presents the logic for how a specific failed quality control metric applies to DNA and RNA variant reporting in the same sample and other samples in an Oncomine™ Dx Express Test run.

**Table 10 Quality control metric failure and variant reporting**

| Specification type | Failed metric | Variant reported? | | | | Applies to |
|---|---|---|---|---|---|---|
| | | DNA: SNV/INDEL | DNA: CNV | RNA: Fusion/Splice variant | RNA: Imbalance | |
| Purification QC | Sample Concentration DNA | No | No | Yes | Yes | Failed sample only |
| | Sample Concentration RNA | Yes | Yes | No | No | Failed sample only |
| Templating Control QC | CF-1 Average Reads per Lane | No | No | No | No | All samples in a run |
| | CF-1 Mean AQ20 Read Length | No | No | No | No | All samples in a run |
| NTC QC - DNA | Average Base Coverage Depth | No | No | Yes | Yes | All DNA samples in a run |
| | DNA Hotspot Calls | No | No | Yes | Yes | All DNA samples in a run |
| NTC QC - RNA | Mapped Reads | Yes | Yes | No | No | All RNA samples in a run |
| | Total Fusion Calls | Yes | Yes | No | No | All RNA samples in a run |
| Positive Control QC - DNA | SNV/INDEL Threshold | No | Yes | Yes | Yes | All DNA samples in a run |
| | CNV Threshold | Yes | No | Yes | Yes | All DNA samples in a run |
| | MAPD | Yes | No | Yes | Yes | All DNA samples in a run |

**Table 10 Quality control metric failure and variant reporting** *(continued)*

| Specification type | Failed metric | Variant reported? | | | | Applies to |
|---|---|---|---|---|---|---|
| | | DNA: SNV/INDEL | DNA: CNV | RNA: Fusion/Splice variant | RNA: Imbalance | |
| Positive Control QC - DNA | Mean AQ20 Read Length | No | No | Yes | Yes | All DNA samples in a run |
| | Mapped Reads | No | No | Yes | Yes | All DNA samples in a run |
| Positive Control QC - RNA | Targeted Isoform (Fusion/Splice) | Yes | Yes | No | Yes | All RNA samples in a run |
| | Expression Imbalance | Yes | Yes | Yes | No | All RNA samples in a run |
| | Mapped Fusion Reads | Yes | Yes | No | No | All RNA samples in a run |
| | RNA Expression Controls Detected | Yes | Yes | No | No | All RNA samples in a run |
| Sample QC - DNA | MAPD | Yes | No | Yes | Yes | Failed sample only |
| | Mean AQ20 Read Length | No | No | Yes | Yes | Failed sample only |
| | Mapped Reads | No | No | Yes | Yes | Failed sample only |
| Sample QC - RNA | Mapped Fusion Reads | Yes | Yes | No | No | Failed sample only |
| | RNA Expression Controls Detected | Yes | Yes | No | No | Failed sample only |

# Variant call filters and No Call repeat strategies

The following table lists filters used by the software in variant calling, and potential repeat strategies if a No Call is observed. The cause for a No Call can be determined by examining the value of the FR tag for a given variant, listed in the output VCF file. If coverage or sample quality issues are possible contributing factors to a No Call, repeat strategies are listed.

**Table 11 Variant call filters and No Call repeat strategies**

| Observation (Example FR tag) | Interpretation[1] | Repeat strategy |
|---|---|---|
| **SNV/INDELs** | | |
| *HPLEN* | Homopolymer INDELs of more than the set length are filtered out. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *MINCOV* | A called allele is marked as NOCALL if the coverage at the location of the allele falls below the cut-off value. | Repeat the run with fewer samples per chip to improve coverage. |
| *NOCALLxPredicted SSE* | This filter identifies locations where systematic sequencing errors (SSE) are predicted to occur on both the positive and negative strands. No deletions at these sites are considered reliable due to the consistent error pattern affecting both strands. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *NOCALLxPositive SSE* | This filter identifies locations where only the positive strand shows potential SSE. It helps in filtering out deletion alleles on the positive strand that are not supported by the negative strand. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *NOCALLxNegative SSE* | This filter identifies locations where only the negative strand shows potential SSE. It helps in filtering out deletion alleles on the negative strand that are not supported by the positive strand. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *NODATA* | There are no reads for the sample. | Repeat the failed sample extraction and sequencing from the point of the suspected workflow error. |
| *POSBIAS* and *POSBIASPVAL* | The POSBIAS filter indicates that a variant exhibits significant positional bias, meaning the variant calls might be unevenly distributed across reads from different sequencing positions. POSBIASPVAL is a statistical measure of POSBIAS. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |

**Table 11 Variant call filters and No Call repeat strategies** *(continued)*

| Observation (Example FR tag) | Interpretation[1] | Repeat strategy |
|---|---|---|
| *PosCov* or *NegCov* | This filter indicates that the read coverage on positive or negative strand for the called allele is below the cut-off. | Repeat the run with fewer samples per chip to improve coverage. |
| *PREDICTIONSHIFTx* | This filter highlights variants that can be prone to errors due to base shift discrepancies. It is important for filtering out the false positive calls due to strand-specific sequencing error. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *PREDICTIONVarSHIFTx* or *PREDICTIONRefSHIFTx* | Filters out a deletion or an insertion if the observed clusters deviate from predictions more than the set amount. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *QualityScore* | Filters out low confidence calls. | If the allele frequency of the No Call sample is below or near the cut-off, do not sequence the sample again. Otherwise, repeat the run with greater tissue input to improve the quality score. Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *REJECTION* | Probability that a read comes from none of the models under consideration. Filters out a call with too many outlier reads. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *STDBIAS* and *STDBIASPVAL* | Strand bias and strand bias P value to filter out strand-specific sequencing error. | Repeat with a Nucleic Acid to Result run with greater sample input to improve strand coverage. |
| *STRINGENCY* | A called variant needs to have a mean log-likelihood difference per read greater than this Phred-scaled value. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *VarCov* | Indicates that the variant coverage after flow evaluation of a call is below the cut-off. | Repeat the run with fewer samples per chip to improve coverage. |
| *vc.novel* or a No Call at an adjacent hotspot variant | Contributing factors of this No Call condition are technical issues that cannot be resolved by repeat testing. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| **RNA Imbalance** | | |
| *FAILED_GENE_QC* | An imbalance call cannot be reported for this gene because the gene level QC failed. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |

**Table 11 Variant call filters and No Call repeat strategies** *(continued)*

| Observation (Example FR tag) | Interpretation[1] | Repeat strategy |
|---|---|---|
| **Copy Number Variants** | | |
| *READ_COUNT* | Filters out CNV calls with total mapped reads below the threshold. | Repeat the run with fewer samples per chip to improve coverage. |
| *PERCENT_NON ZERO_AMPLICONS* | Filters out CNV calls when the percentage of amplicons (targeted regions) with non-zero read count is below the threshold. | Re-prepare the sample or repeat the run with fewer samples per chip to improve coverage. |
| *MEDIAN_READS* | Filters out CNV calls when the median number of reads per amplicon is below the threshold. | Repeat the run with fewer samples per chip to improve coverage. |
| *MAPD* | Measures signal noise across the sample. A larger MAPD indicates higher noise level. | Re-prepare the sample with greater tissue input to improve quality. |
| *LOCAL_AVERAGE_ SIGNAL_VARIATION* | This filter assesses the variation in signal intensity between amplicons in a gene. A gene with partial total loss is usually flagged by this filter. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *SEVERE_ GRADIENT* | Indicates a difference in calculated copy number above a certain threshold (range of values >4* MAPD or >0.5) across 8 adjacent amplicons of a CNV target. Here, MAPD is the local MAPD of the CNV target. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *DIFFERENT_MEAN_ SIGNAL* | This filter assesses differences in the mean signal between two groups of co-located amplicons within a CNV region. If the difference exceeds a specified threshold, a flag is raised. | Repeating the sample extraction or rerunning the workflow starting from sequencing is not likely to change the result. |
| *percent_aligned_ reads* | FIlters out samples when the percentage of reads aligned to the panel falls below the cut-off. | Re-prepare the sample with greater tissue input to improve quality. |

[1] Although the presence of multiple reasons for No Call is not required for a No Call assignment, more than one condition can occur simultaneously. In cases where multiple reasons are observed, such that one recommends repeat while the other does not, repeating sample from sample extraction is recommended.

# A Performance characteristics

## Analytical studies

The performance characteristics for the Oncomine™ Dx Express Test were established using DNA and RNA derived from a variety of FFPE tumor types. These FFPE tumor samples were analyzed for CNVs and a broad range of representative substitutions, insertions, and deletions in DNA, as well as fusions and splice variants in RNA, including CDx and tumor profiling variants. Several analytical validation studies were conducted to support the CDx and tumor profiling claims described in the intended use statement. All studies were performed using intended use specimens and sample blends and are summarized in the following section.

### Analytical accuracy studies

#### Analytical accuracy—EGFR exon 20 insertion biomarker

The accuracy of the Oncomine™ Dx Express Test for detecting EGFR exon 20 insertions in patients with NSCLC was evaluated by comparing the test results of the Oncomine™ Dx Express Test with those of a validated NGS-based orthogonal method. A total of 202 tissue samples were obtained from subjects enrolled in the sunvozertinib clinical trial (WU-KONG1B, NCT03974022). Of these, 200 were EGFR exon 20 insertion-positive and 2 were negative, as determined by the clinical trial assays (CTAs) used for patient enrollment. Additionally, 232 samples were procured from commercial vendors and screened using NGS- and polymerase chain reaction (PCR)-based assays representative of the technologies commonly used for patient enrollment in the WU-KONG1B trial to identify EGFR exon 20 insertion-negative cases. Of these biomarker-negative samples, 143 were confirmed as negative, 3 were positive, and 86 were deemed invalid.

A total of 348 samples including 203 EGFR exon 20 insertion-positive samples and 145 EGFR exon 20 insertion–negative samples were initially submitted for testing with the Oncomine™ Dx Express Test. Of the 203 biomarker-positive sample, 150 were tested using the Oncomine™ Dx Express Test. Among these, 144 were confirmed as EGFR exon 20 insertion-positive, 3 were identified as negative, and 3 yielded indeterminate results. All 150 samples tested with the Oncomine™ Dx Express Test were submitted for testing with the orthogonal method; however, 56 samples had insufficient DNA, and only 94 could be evaluated. Of these, 84 were confirmed as positive, 5 were negative, and 5 produced invalid or no call results by the orthogonal method. Among the 145 biomarker-negative samples, 138 were tested with the Oncomine™ Dx Express Test. Of these, 134 were confirmed as EGFR exon 20 insertion-negative, while 4 did not yield valid results. All 138 samples were submitted for testing with the orthogonal method; however, due to insufficient DNA, only 108 were evaluable. Among the evaluable samples, 106 were confirmed as EGFR exon 20 insertion-negative, and 2 produced invalid results.

The analytical concordance between the results from the Oncomine™ Dx Express Test and the orthogonal method for detecting EGFR exon 20 insertions, based on a total of 348 samples (203 EGFR exon 20 insertion-positive samples and 145 EGFR exon 20 insertion-negative samples) initially submitted for testing with the Oncomine™ Dx Express Test, is summarized in Table 12.

**Table 12 Analytical concordance between the Oncomine™ Dx Express Test (ODxET) and orthogonal method for EGFR exon 20 insertions**

| | | Orthogonal method | | | | | Total |
|---|---|---|---|---|---|---|---|
| | | Positive | Negative | Invalid[1] | No Calls | TNP[2] | |
| ODxET | Positive | 84 | 5 | 3 | 2 | 50 | 144 |
| | Negative | 0 | 105 | 1 | 0 | 31 | 137 |
| | Invalid[1] | 0 | 0 | 0 | 0 | 2 | 2 |
| | No Calls | 0 | 1 | 0 | 1 | 3 | 5 |
| | TNP[2] | 0 | 0 | 0 | 0 | 60 | 60 |
| | Total | 84 | 111 | 4 | 3 | 145 | 348 |

[1] Invalid or No calls were considered unknown or unevaluable.

[2] TNP: Test Not Performed due to DNA concentration fail, tumor content fail, insufficient material, or DNA. Samples with TNP are categorized as unknown.

The positive percent agreement (PPA), negative percent agreement (NPA), overall percent agreement (OPA), and their corresponding two-sided 95% Wilson Score confidence intervals (CIs), using the orthogonal method as the reference, are provided in Table 6. When excluding Oncomine™ Dx Express Test unknowns, the point estimates were 100% (84/84) for PPA, 95.5% (105/110) for NPA, and 97.4% (189/194) for OPA. When including Oncomine™ Dx Express Test unknowns, the point estimates were 100% (84/84) for PPA, 94.6% (105/111) for NPA, and 96.9% (189/195) for OPA.

**Table 13 Agreement between the Oncomine™ Dx Express Test (ODxET) and orthogonal method for EGFR exon 20 insertion detection**

| | | Excluding ODxET unknowns | | | Including ODxET unknowns | | |
|---|---|---|---|---|---|---|---|
| Parameter | Agreed (N) | Total (N) | Percent agreement | 95% CI | Total (N) | Percent agreement | 95% CI |
| PPA | 84 | 84 | 100% | (95.6%, 100%) | 84 | 100% | (95.6%, 100%) |
| NPA | 105 | 110 | 95.50% | (89.8%, 98.0%) | 111 | 94.60% | (88.7%, 97.5%) |
| OPA | 189 | 194 | 97.40% | (94.1%, 98.9%) | 195 | 96.90% | (93.5%, 98.6%) |

In the WU-KONG1B trial, the study population was enriched for cases positive for EGFR exon 20 insertions. Consequently, the analytical concordance between Oncomine™ Dx Express Test and the orthogonal method was re-evaluated by calculating prevalence-adjusted values for PPA and NPA, with corresponding two-sided 95% CIs estimated using a percentile bootstrap method. At a prevalence of

1.8%, the adjusted point estimates for PPA and NPA were 100.0% (95.6%, 100.0%) and 99.9% (99.8%, 100.0%), respectively, demonstrating high analytical concordance between the Oncomine™ Dx Express Test and the comparator method.

Additionally, the positive predictive value (PPV) and adjusted negative predictive value (NPV) for the comparison between the Oncomine™ Dx Express Test and the orthogonal method were calculated based on the prevalence of EGFR exon 20 insertions in NSCLC, which is 1.8%. Excluding Oncomine™ Dx Express Test unknowns, the PPV was 95.7% (91.9%, 99.1%), while including unknowns, it was 94.9% (90.9%, 98.3%). The NPV was 100% (96.5%, 100%) regardless of whether ODxET unknowns were excluded or included. The 95% CIs for the PPV were estimated using the percentile bootstrap method, whereas the 95% CIs for the NPV were calculated using the Wilson Score method due to the point estimate being 100%.

## Analytical accuracy for tumor profiling variants

To evaluate the analytical accuracy of the Oncomine™ Dx Express Test for detecting SNVs, insertions and deletions (INDELs), CNVs, fusions, and RNA splice variants in FFPE clinical tumor samples, Oncomine™ Dx Express Test results were compared with those obtained from validated orthogonal methods. The accuracy assessment for tumor profiling variants was conducted across three separate studies. Study I included Level 2 and Level 3 SNVs, insertions, deletions, CNVs, fusions, and RNA splice variants across various tumor types. Study II focused on Level 2 ERBB2 CNVs in breast cancer, ALK fusions in NSCLC, and RET fusions in both NSCLC and thyroid cancer. Study III evaluated Level 2 EGFR SNVs and exon 19 deletions in NSCLC. For all three phases, samples were pre-screened using an orthogonal method similar to Oncomine™ Dx Express Test to confirm the presence or absence of target variants when vendor-provided screening information was insufficient or unclear. In the three analytical accuracy studies conducted for the tumor profiling indication, the PPA and NPA estimates were reported without adjustment for the distribution of samples selected by the pre-screening method. The analytical accuracy results from each study are presented separately below.

## Analytical accuracy study I—Tumor profiling variants

The analytical accuracy study I evaluated the performance of the Oncomine™ Dx Express Test for detecting a range of tumor profiling variants, including SNVs, insertions, deletions, CNVs, fusions, and RNA splice variants, in FFPE clinical tumor samples. The samples were sourced from qualified commercial vendors and were prescreened for target variants using an orthogonal method similar to the Oncomine™ Dx Express Test. The study included 397 SNVs/multi nucleotide variants (MNVs), 5 insertions, 23 deletions, 114 CNVs, and 47 fusion or splice samples. Additionally, 58 unique wild-type (WT) samples and 42 samples pre-identified as negative for entire variant classes were evaluated. Of these, 36 WT samples and 40 variant class-negative samples were included in the concordance analysis. A total of 462 FFPE tumor samples representing 15 tumor types was tested, as shown in Table 14.

**Table 14 Number of samples per cancer type**

| Tissue type | Number of samples |
|---|---|
| Bile duct | 13 |
| Bladder | 56 |
| Brain | 48 |
| Breast | 61 |
| Colon | 70 |
| Endometrium | 26 |
| Gastrointestinal | 1 |
| Head/Neck | 2 |
| Kidney | 2 |
| Lung | 90 |

**Table 14 Number of samples per cancer type** *(continued)*

| Tissue type | Number of samples |
|---|---|
| Pancreas | 9 |
| Prostate | 7 |
| Skin | 48 |
| Soft tissue | 3 |
| Thyroid | 26 |
| **TOTAL** | **462** |

The study compared Oncomine™ Dx Express Test results with those obtained from a validated NGS-based orthogonal method. Concordance analysis was performed at the variant level using results from both the Oncomine™ Dx Express Test and the orthogonal test. PPA and NPA estimates, along with their respective two-sided 95% CIs, were calculated using the orthogonal method as the reference. The accuracy results, excluding unknown results, are summarized by variant type and further stratified according to the FDA's Biomarker Class Level for tumor profiling NGS tests, as shown in Table 15.

**Table 15 Accuracy results for Oncomine™ Dx Express Test tumor profiling variants by variant type and clinical significance—Accuracy Study I**

| Variant type | FDA biomarker category[1] | Total unique variants/genes | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][2] | NPA (n/N) [95% CI][2] |
|---|---|---|---|---|---|---|---|---|
| **All variant types** | **All** | 2944/46 | 586 | 112 | 11 | 800035 | 98.2% (586/597) [96.7%,99.0%] | 100.0% (800035/800147) [100.0%,100.0%] |
| | **Level 2** | 1544/22 | 190 | 19 | 0 | 116800 | 100.0%(190/190) [98.0%,100.0%] | 100.0% (116800/116819) [100.0%,100.0%] |
| | **Level 3** | 1400/43 | 396 | 93 | 11 | 683235 | 97.3% (396/407) [95.3%,98.5%] | 100.0% (683235/683328) [100.0%,100.0%] |
| **SNVs** | **All** | 1324/42 | 397 | 47 | 2 | 387643 | 99.5% (397/399) [98.2%,99.9%] | 100.0%\ (387643/387690) [100.0%,100.0%] |
| | **Level 2** | 265/17 | 137 | 4 | 0 | 10594 | 100.0%(137/137) [97.2%,100.0%] | 100.0%(10594/10598) [99.9%,100.0%] |

**Table 15 Accuracy results for Oncomine Dx Express Test tumor profiling variants by variant type and clinical significance—Accuracy Study I** *(continued)*

| Variant type | FDA biomarker category[1] | Total unique variants/genes | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][2] | NPA (n/N) [95% CI][2] |
|---|---|---|---|---|---|---|---|---|
| **SNVs** | **Level 3** | 1059/38 | 260 | 43 | 2 | 377049 | 99.2% (260/262) [97.3%,99.8%] | 100.0% (377049/377092) [100.0%,100.0%] |
| **MNVs + Complex** | **All** | 287/14 | 0 | 0 | 0 | 6466 | N/E[3] | 100.0%(6466/6466) [99.9%,100.0%] |
| | **Level 2** | 210/12 | 0 | 0 | 0 | 343 | N/E | 100.0%(343/343) [98.9%,100.0%] |
| | **Level 3** | 77/19 | 0 | 0 | 0 | 6123 | N/E | 100.0%(6123/6123) [99.9%,100.0%] |
| **Insertions** | **All** | 75/11 | 5 | 1 | 0 | 61874 | 100.0%(5/5) [56.6%,100.0%] | 100.0%(61874/61875) [100.0%,100.0%] |
| | **Level 2** | 52/3 | 0 | 0 | 0 | 2772 | N/E | 100.0%(2772/2772) [99.9%,100.0%] |
| | **Level 3** | 23/9 | 5 | 1 | 0 | 59102 | 100.0%(5/5) [56.6%,100.0%] | 100.0%(59102/59103) [100.0%,100.0%] |
| **Deletions** | **All** | 258/14 | 23 | 2 | 0 | 78635 | 100.0%(23/23) [85.7%,100.0%] | 100.0%(78635/78637) [100.0%,100.0%] |
| | **Level 2** | 206/5 | 14 | 1 | 0 | 9292 | 100.0%(14/14) [78.5%,100.0%] | 100.0%(9292/9293) [99.9%,100.0%] |
| | **Level 3** | 52/13 | 9 | 1 | 0 | 69343 | 100.0%(9/9) [70.1%,100.0%] | 100.0%(69343/69344) [100.0%,100.0%] |
| **CNVs** | **All** | 10/10 | 114 | 32 | 7 | 4294 | 94.2% (114/121) [88.5%,97.2%] | 99.3%(4294/4326) [99.0%,99.5%] |
| | **Level 2** | 2/2 | 14 | 3 | 0 | 238 | 100.0%(14/14) [78.5%,100.0%] | 98.8% (238/241) [96.4%,99.6%] |
| | **Level 3** | 8/8 | 100 | 29 | 7 | 4056 | 93.5% (100/107) [87.1%,96.8%] | 99.3%(4056/4085) [99.0%,99.5%] |

**Table 15 Accuracy results for Oncomine Dx Express Test tumor profiling variants by variant type and clinical significance—Accuracy Study I** *(continued)*

| Variant type | FDA biomarker category[1] | Total unique variants/genes | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][2] | NPA (n/N) [95% CI][2] |
|---|---|---|---|---|---|---|---|---|
| RNA variants | All | 990/18 | 47 | 30 | 2 | 261123 | 95.9%(47/49) [86.3%,98.9%] | 100.0% (261123/261153) [100.0%,100.0%] |
| | Level 2 | 809/10 | 25 | 11 | 0 | 93561 | 100.0%(25/25) [86.7%,100.0%] | 100.0%(93561/93572) [100.0%,100.0%] |
| | Level 3 | 181/16 | 22 | 19 | 2 | 167562 | 91.7% (22/24) [74.2%, 97.7%] | 100.0% (167562/167581) [100.0%, 100.0%] |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

[3] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

A summary of accuracy results for SNVs in predefined challenging genomic regions (for example, simple sequence repeats [SSRs], homopolymers, GC content), and for insertions and deletions stratified by predefined size bins (for example, 1–5 bp, 6–10 bp, 11–15 bp, >15 bp) and by challenging genomic regions, excluding unknown results, is presented in Table 16.

**Table 16 Accuracy results for SNVs, insertions, and deletions in clinically and analytically meaningful bins—Accuracy Study I**

| Variant type | Bin | Total unique variants | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| SNVs | All | 1324 | 397 | 47 | 2 | 387643 | 99.5% (397/399) [98.2%,99.9%] | 100.0% (387643/387690) [100.0%,100.0%] |
| | Non Homo-polymers | 344 | 90 | 10 | 0 | 106693 | 100.0%(90/90) [95.9%,100.0%] | 100.0% (106693/106703) [100.0%,100.0%] |
| | Homo-polymers | 980 | 307 | 37 | 2 | 280950 | 99.4% (307/309) [97.7%,99.7%] | 100.0% (280950/280987) [100.0%,100.0%] |
| | Non-SSRs | 813 | 230 | 27 | 1 | 241707 | 99.6% (230/231) [97.6%,99.9%] | 100.0% (241707/241734) [100.0%,100.0%] |
| | SSRs | 511 | 167 | 20 | 1 | 145936 | 99.4% (167/168) [96.7%,99.9%] | 100.0% (145936/145956) [100.0%,100.0%] |
| | GC [30%–40%] | 282 | 111 | 3 | 0 | 87285 | 100.0% (111/111) [96.7%,100.0%] | 100.0% (87285/87288) [100.0%,100.0%] |
| | GC (40%–50%] | 237 | 98 | 5 | 0 | 69985 | 100.0% (98/98) [96.2%,100.0%] | 100.0% (69985/69990) [100.0%,100.0%] |
| | GC (50%–60%] | 552 | 143 | 29 | 2 | 163944 | 98.6% (143/145) [95.1%,99.6%] | 100.0% (163944/163973) [100.0%,100.0%] |
| | GC (60%–75%] | 253 | 45 | 10 | 0 | 66429 | 100.0%(45/45) [92.1%,100.0%] | 100.0% (66429/66439) 100.0%,100.0%] |
| MNVs + Complex | All | 287 | 0 | 0 | 0 | 6466 | N/E[2] | 100.0% (6646/6466) [100.0%,100.0%] |
| | Non-homo-polymers | 40 | 0 | 0 | 0 | 0 | N/E | N/E |
| | Homo-polymers | 247 | 0 | 0 | 0 | 6466 | N/E | 100.0% (6646/6466) [100.0%,100.0%] |

**Table 16 Accuracy results for SNVs, insertions, and deletions in clinically and analytically meaningful bins—Accuracy Study I** *(continued)*

| Variant type | Bin | Total unique variants | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| MNVs + Complex | Non-SSRs | 223 | 0 | 0 | 0 | 1484 | N/E | 100.0% (1484/1484) [99.7%,100.0%] |
| | SSRs | 64 | 0 | 0 | 0 | 4982 | N/E | 100.0% (4982/4982) [99.9%,100.0%] |
| | GC [30%–40%] | 63 | 0 | 0 | 0 | 110 | N/E | 100.0% (110/110) [96.6%,100.0%] |
| | GC (40%–50%] | 157 | 0 | 0 | 0 | 8 | N/E | 100.0% (8/8) [67.6%,100.0%] |
| | GC (50%–60%] | 49 | 0 | 0 | 0 | 1366 | N/E | 100.0% (1366/1366) [99.7%,100.0%] |
| | GC (60%–75%] | 18 | 0 | 0 | 0 | 4982 | N/E | 100.0% (4982/4982) [99.9%,100.0%] |
| Insertions | All | 75 | 5 | 1 | 0 | 61874 | 100.0% (5/5) [56.6%,100.0%] | 100.0% (61874/61875) [100.0%,100.0%] |
| | 1–5 bp | 22 | 1 | 1 | 0 | 15958 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (15958/15959) [100.0%,100.0%] |
| | 6–10 bp | 27 | 4 | 0 | 0 | 30858 | 100.0% (4/4) [51.0%,100.0%] | 100.0% (30858/30858) [100.0%,100.0%] |
| | 11–15 bp | 12 | 0 | 0 | 0 | 9839 | N/E | 100.0% (9839/9839) [100.0%,100.0%] |
| | >15 bp | 14 | 0 | 0 | 0 | 5219 | N/E | 100.0% (5219/5219) [99.9%,100.0%] |
| | Non-homo-polymers | 8 | 0 | 0 | 0 | 2169 | N/E | 100.0% (2169/2169) [99.8%,100.0%] |
| | Homo-polymers | 67 | 5 | 1 | 0 | 59705 | 100.0% (5/5) [56.6%,100.0%] | 100.0% (59705/59706) [100.0%,100.0%] |

**Table 16 Accuracy results for SNVs, insertions, and deletions in clinically and analytically meaningful bins—Accuracy Study I** *(continued)*

| Variant type | Bin | Total unique variants | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| Insertions | Non-SSRs | 65 | 0 | 1 | 0 | 19201 | N/E | 100.0% (19201/19202) [100.0%,100.0%] |
| | SSRs | 10 | 5 | 1 | 0 | 42673 | 100.0% (5/5) [56.6%,100.0%] | 100.0% (42673/42673) [100.0%,100.0%] |
| | GC [30%–40%] | 22 | 0 | 0 | 0 | 7387 | N/E | 100.0% (7387/7387) [99.9%,100.0%] |
| | GC (40%–50%] | 7 | 0 | 0 | 0 | 2320 | N/E | 100.0% (2320/2320) [99.8%,100.0%] |
| | GC (50%–60%] | 41 | 0 | 0 | 0 | 11643 | N/E | 100.0% (11643/11643) [100.0%,100.0%] |
| | GC (60%–75%] | 5 | 5 | 1 | 0 | 40524 | 100.0% (5/5) [56.6%,100.0%] | 100.0% (40524/40525) [100.0%,100.0%] |
| Deletions | All | 258 | 23 | 2 | 0 | 78635 | 100.0% (23/23) [85.7%,100.0%] | 100.0% (78635/78637) [100.0%,100.0%] |
| | 1–5 bp | 52 | 8 | 2 | 0 | 15160 | 100.0% (8/8) [67.6%,100.0%] | 100.0% (15160/15162) [100.0%,100.0%] |
| | 6–10 bp | 49 | 0 | 0 | 0 | 14697 | N/E | 100.0% (14697/14697) [100.0%,100.0%) |
| | 11–15 bp | 63 | 14 | 0 | 0 | 19988 | 100.0% (14/14) [78.5%,100.0%] | 100.0% (19988/19988) [100.0%,100.0%] |
| | >15 bp | 94 | 1 | 0 | 0 | 28790 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (28790/28790) [100.0%,100.0%] |
| | Non-homo-polymers | 25 | 0 | 1 | 0 | 7944 | N/E | 100.0% (7944/7945) [99.9%,100.0%] |

**Table 16 Accuracy results for SNVs, insertions, and deletions in clinically and analytically meaningful bins—Accuracy Study I** *(continued)*

| Variant type | Bin | Total unique variants | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| Deletions | Homo-polymers | 233 | 23 | 1 | 0 | 70691 | 100.0% (23/23) [85.7%,100.0%] | 100.0% (70691/70692) [100.0%,100.0%] |
| | Non-SSRs | 210 | 20 | 2 | 0 | 64874 | 100.0% (20/20) [83.9%,100.0%] | 100.0% (64874/64876) [100.0%,100.0%] |
| | SSRs | 48 | 3 | 0 | 0 | 13761 | 100.0% (3/3) [43.9%,100.0%] | 100.0% (13761/13761) [100.0%,100.0%] |
| | GC 30%–40%] | 142 | 7 | 1 | 0 | 43018 | 100.0% (7/7) [64.6%,100.0%] | 100.0% (43018/43019) [100.0%,100.0%] |
| | GC (40%–50%] | 75 | 14 | 0 | 0 | 24834 | 100.0% (14/14) [78.5%,100.0%] | 100.0% (24834/24834) [100.0%,100.0%] |
| | GC (50%–60%] | 15 | 1 | 1 | 0 | 5081 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (5081/5082) [99.9%,100.0%] |
| | GC (60%–75%] | 26 | 1 | 0 | 0 | 5702 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (5702/5702) [99.9%,100.0%] |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

[2] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

A summary of accuracy results for CNVs and RNA variants (including fusions and splice variants), excluding unknown results, is presented at the gene level in Table 17.

**Table 17 Accuracy results for CNV and RNA variants at the gene level—Accuracy Study I**

| Variant type | Gene name | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|
| CNV | All | 114 | 32 | 7 | 4294 | 94.1% (112/119) [88.4%,97.1%] | 99.2% (3861/3893) [98.8%,99.4%] |
| | AR | 3 | 0 | 1 | 441 | 75.0% (3/4) [30.1%,95.4%] | 100.0% (441/441) [99.1%,100.0%] |
| | EGFR | 25 | 8 | 0 | 409 | 100.0% (25/25) [86.7%,100.0%] | 98.1% (409/417) [96.3%,99.0%] |
| | ERBB2 | 22 | 4 | 0 | 419 | 100.0% (22/22) [85.1%,100.0%] | 99.1% (419/423) [97.6%,99.6%] |
| | ERBB3 | 2 | 1 | 0 | 442 | 100.0% (2/2) [34.2%,100.0%] | 99.8% (442/443) [98.7%,100.0%] |
| | FGFR1 | 19 | 1 | 1 | 424 | 95.0% (19/20) [76.4%,99.1%] | 99.8% (424/425) [98.7%,100.0%] |
| | FGFR2 | 9 | 0 | 0 | 436 | 100.0% (9/9) [70.1%,100.0%] | 100.0% (436/436) [99.1%,100.0%] |
| | FGFR3 | 9 | 14 | 1 | 421 | 90.0% (9/10) [59.6%,98.2%] | 96.8% (421/435 [94.7%,98.1%] |
| | KRAS | 10 | 0 | 2 | 433 | 83.3% (10/12) [55.2%,95.3%] | 100.0% (433/433) [99.1%,100.0%] |
| | MET | 9 | 3 | 2 | 431 | 81.8% (9/11) [52.3%,94.9%] | 99.3% (431/434) [98.0%,99.8%] |
| | PIK3CA | 6 | 1 | 0 | 438 | 100.0% (6/6) [61.0%,100.0%] | 99.8% (438/439) [98.7%,100.0%] |
| Fusions | All | 32 | 20 | 1 | 260085 | 97.0% (32/33) [84.7%,99.5%] | 100.0% (260085/260105) [100.0%,100.0%] |
| | ALK | 2 | 2 | 0 | 56654 | 100.0% (2/2) [34.2%,100.0%] | 100.0% (56654/56656) [100.0%,100.0%] |
| | BRAF | 2 | 0 | 0 | 49739 | 100.0% (2/2) [34.2%,100.0%] | 100.0% (49739/49739) [100.0%,100.0%] |
| | ESR1 | 3 | 0 | 0 | 8507 | 100.0% (3/3) [43.9%,100.0%] | 100.0% (8507/8507) [100.0%,100.0%] |
| | FGFR1 | 0 | 0 | 0 | 9310 | N/E[2] | 100.0% (9310/9310) [100.0%,100.0%] |

**Table 17 Accuracy results for CNV and RNA variants at the gene level—Accuracy Study I** *(continued)*

| Variant type | Gene name | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|
| Fusions | FGFR2 | 1 | 1 | 0 | 21012 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (21012/21013) [100.0%,100.0%] |
| | FGFR3 | 8 | 5 | 1 | 11158 | 88.9% (8/9) [56.5%,98.0%] | 100.0% (11158/11163) [99.9%,100.0%] |
| | MET | 1 | 0 | 0 | 8245 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (8245/8245) [100.0%,100.0%] |
| | NRG1 | 1 | 1 | 0 | 9306 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (9306/9307) [99.9%,100.0%] |
| | NTRK1 | 1 | 0 | 0 | 23673 | 100.0% (1/1) [20.7%,100.0%] | 100.0% (23673/23673) [100.0%,100.0%] |
| | NTRK2 | 0 | 1 | 0 | 8245 | N/E | 100.0% (8245/8246) [99.9%,100.0%] |
| | NTRK3 | 4 | 0 | 0 | 8772 | 100.0% (4/4) [51.0%,100.0%] | 100.0% (8772/8772) [100.0%,100.0%] |
| | NUTM1 | 0 | 1 | 0 | 4255 | N/E | 100.0% (4255/4256) [99.9%,100.0%] |
| | RET | 0 | 0 | 0 | 23674 | N/E | 100.0% (23674/23674) [100.0%,100.0%] |
| | ROS1 | 9 | 4 | 0 | 15146 | 100.0% (9/9) [70.1%,100.0%] | 100.0% (15146/15150) [99.9%,100.0%] |
| | RSPO2 | 0 | 0 | 0 | 1064 | N/E | 100.0% (1064/1064) [99.6%,100.0%] |
| | RSPO3 | 0 | 5 | 0 | 1325 | N/E | 99.6% (1325/1330) [99.1%,99.8%] |
| Splice variants/RNA exon variants | All | 15 | 10 | 1 | 1038 | 93.8% (15/16) [71.7%,98.9%] | 99.0% (1038/1048) [98.3%,99.5%] |
| | AR | 6 | 9 | 0 | 251 | 100.0% (6/6) [61.0%,100.0%] | 96.5% (251/260) [93.6%,98.2%] |
| | EGFR | 6 | 0 | 0 | 260 | 100.0% (6/6) [61.0%,100.0%] | 100.0% (260/260) [98.5%,100.0%] |
| | MET | 3 | 1 | 1 | 527 | 75.0% (3/4) [30.1%,95.4%] | 99.8% (527/528) [98.9%,100.0%] |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

[2] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

CNVs in the three genes AR, KRAS, and MET showed relatively low PPA: 75% (3/4) for AR CNV, 83.3% (10/12) for KRAS CNV, and 81.8% (9/11) for MET CNV. All discordant calls were attributed to the presence of target variants in these samples below the limit of detection (LoD) established by the Oncomine™ Dx Express Test.

A summary of accuracy results (PPA and NPA) for all reported variant types, stratified by the LoD level established by Oncomine™ Dx Express Test (for example, at or above 1X LoD and below LoD) and excluding unknown results, is presented in Table 18. For analysis conditional on the Oncomine™ Dx Express Test LoD levels, PPV was calculated for all reported variant types using the same stratification and exclusion criteria. For RNA variants, 'fusion read' refers to a sequencing read that spans a gene fusion breakpoint. For example, a read that provides evidence of a gene fusion event by mapping to two different genomic regions. 'Molecular count' refers to the number of unique fusion molecules present in a sample, as identified by molecular tags.

**Table 18 Accuracy results for all variant types stratified by LoD level—Accuracy Study I**

| Variant type | LoD level | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][1] | NPA (n/N) [95% CI][1] | PPV (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| SNVs | All | 397 | 47 | 2 | 387643 | 99.5% (397/399) [98.2%, 99.9%] | 100.0% (387643/387690) [100.0%, 100.0%] | 89.41% (397/444) [86.21%, 91.95%] |
| | ≥1X LoD | 391 | 12 | 0 | — | 100.0% (391/391) [99.0%, 100.0%] | N/E[2] | 97.02% (391/403) [94.87%, 98.29%] |
| | <1X LoD | 6 | 35 | 2 | — | 75.0% (6/8) [40.9%, 92.9%] | N/E | 14.63% (6/41) [6.88%, 28.44%] |
| Insertions | All | 5 | 1 | 0 | 61874 | 100.0% (5/5) [56.6%, 100.0%] | 100.0% (61874/61875) [100.0%, 100.0%] | 83.3% (5/6) [43.6%, 97.0%] |
| | ≥1X LoD | 5 | 1 | 0 | — | 100.0% (5/5) [56.6%, 100.0%] | N/E | 83.3% (5/6) [43.6%, 97.0%] |
| | <1X LoD | 0 | 0 | 0 | — | N/E | N/E | N/E |
| Deletions | All | 23 | 2 | 0 | 78635 | 100.0% (23/23) [85.7%, 100.0%] | 100.0% (78635/78637) [100.0%, 100.0%] | 92.0% (23/25) [75.03%, 97.78%] |

**Table 18 Accuracy results for all variant types stratified by LoD level—Accuracy Study I** *(continued)*

| Variant type | LoD level | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] | PPV (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| **Deletions** | ≥1X LoD | 23 | 2 | 0 | — | 100.0% (23/23) [85.7%, 100.0%] | N/E | 92.0% (23/25) [75.03%, 97.78%] |
| | <1X LoD | 0 | 0 | 0 | — | N/E | N/E | N/E |
| **CNVs** | All | 114 | 32 | 7 | 4294 | 94.2% (114/121) [88.5%, 97.2%] | 99.3% (4294/4326) [99.0%, 99.5%] | 78.08% (114/146) [70.7%, 84.03%] |
| | ≥1X LoD | 113 | 29 | 0 | — | 100.0% (113/113) [96.7%, 100.0%] | N/E | 79.58% (113/142) [72.21%, 85.39%] |
| | <1X LoD | 1 | 3 | 7 | — | 12.5% (1/8) [2.2%, 47.1%] | N/E | 25% (1/4) [4.56%, 69.94%] |
| **Fusions** | All | 32 | 20 | 1 | 260085 | 97.0% (32/33) [84.7%, 99.5%] | 100.00% (260085/260105) [100.0%, 100.0%] | 61.5% (32/52) [48.0%, 73.5%] |
| | ≥1X LoD (Molecular counts) | 32 | 13 | 0 | — | 100.0% (32/32) [89.3%, 100.0%] | N/E | 71.1% (32/45) [56.6%, 82.3%] |
| | <1X LoD (Molecular counts) | 0 | 7 | 1 | — | 0.0% (0/1) [0.0%, 79.3%] | N/E | 0.0% (0/7) [0.0%, 35.4%] |
| | ≥1X LoD (Fusion reads) | 30 | 15 | 0 | — | 100.0% (30/30) [88.6%, 100.0%] | N/E | 66.7% (30/45) [52.1%, 78.6%] |
| | <1X LoD (Fusion reads) | 2 | 5 | 1 | — | 66.7% (2/3) [20.8%, 93.9%] | N/E | 28.6% (2/7) [8.2%, 64.1%] |

**Table 18 Accuracy results for all variant types stratified by LoD level—Accuracy Study I** *(continued)*

| Variant type | LoD level | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N [95% CI][1] | NPA (n/N) [95% CI][1] | PPV (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| RNA splice variants | All | 15 | 10 | 1 | 1038 | 93.8% (15/16) [71.7%, 98.9%] | 100.0% (1038/1048) [100.0%, 100.0%] | 60.0% (15/25) [40.7%, 76.6%] |
| | ≥1X LoD (Molecular counts) | 14 | 3 | 0 | — | 100.0% (14/14) [78.5%, 100.0%] | N/E | 82.4% (14/17) [59.0%, 93.8%] |
| | <1X LoD (Molecular counts) | 1 | 7 | 1 | — | 50.0% (½) [9.5%, 90.5%] | N/E | 12.5% (1/8) [2.2%, 47.1%] |
| | ≥1X LoD (Fusion reads) | 14 | 4 | 0 | — | 100.0% (14/14) [78.5%, 100.0%] | N/E | 77.8% (14/18) [54.8%, 91.0%] |
| | <1X LoD (Fusion reads) | 1 | 6 | 1 | — | 50.0% (1/2) [9.5%, 90.5%] | N/E | 14.3% (1/7) [2.6%, 51.3%] |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

[2] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

SNVs (75.0%), CNVs (12.5%), and RNA variants (33.3%) showed low PPA when the target variants in these samples were present below the LoD established by the Oncomine™ Dx Express Test.

## Analytical accuracy study II—Tumor profiling variants

The accuracy study II was conducted to evaluate the accuracy of the Oncomine™ Dx Express Test for detecting ERBB2 CNVs in breast cancer, ALK fusions in NSCLC, and RET fusions in NSCLC and thyroid cancer by retrospectively testing FFPE clinical samples. The samples were sourced from qualified commercial vendors and were pre-screened for target variants using an orthogonal method similar to Oncomine™ Dx Express Test. All variants evaluated in this study were Level 2 variants, classified according to FDA's three-tiered approach for reporting biomarkers in tumor profiling NGS tests.

For ERBB2 CNVs, 74 breast cancer samples were tested using both the Oncomine™ Dx Express Test and the fluorescence in situ hybridization (FISH)-based orthogonal method. Of these, 66 samples (29 positive and 37 negative) had concordant results. Four samples were discordant, showing positive results with the Oncomine™ Dx Express Test but negative with the orthogonal method, and four samples were negative with the Oncomine™ Dx Express Test but had unknown results with the orthogonal method. Table 19 shows the 3 × 3 contingency table accounting for all 74 breast ERBB2

CNV samples tested by both the Oncomine™ Dx Express Test (ODxET) and the orthogonal method, along with the PPA and NPA estimates.

**Table 19 Concordance between the Oncomine™ Dx Express Test and orthogonal method for detection of Level 2 ERBB2 CNVs in breast cancer—Accuracy study II**

| | | Orthogonal method | | | |
|---|---|---|---|---|---|
| | | Positive | Negative | Unknown[1] | Total |
| ODxET | Positive | 29 | 4 | 0 | 33 |
| | Negative | 0 | 37 | 4 | 41 |
| | Unknown[1] | 0 | 0 | 0 | 0 |
| | Total | 29 | 41 | 4 | 74 |
| Excluding unknown | PPA (n/N) [95% CI][2] | 100.0% (29/29) [88.3%, 100.0%] | | | |
| | NPA (n/N) [95% CI][2] | 90.2% (37/41) [77.5%, 96.1%] | | | |
| Including unknown | PPA (n/N) [95% CI][2] | 100.0% (29/29) [88.3%, 100.0%] | | | |
| | NPA (n/N) [95% CI][2] | 90.2% (37/41) [77.5%, 96.1%] | | | |

[1] Unknowns are defined as invalid due to insufficient sample, or sample QC sequencing failure resulting in an invalid result or No Call for the variant.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

For ALK fusions, 93 NSCLC samples were tested using the FISH-based orthogonal method. Of these, 67 samples (26 positive and 41 negative) had concordant results. One sample was discordant, showing a positive result with the Oncomine™ Dx Express Test but negative with the orthogonal method. Thirteen samples were positive with the Oncomine™ Dx Express Test but produced unknown results with the orthogonal method. Ten samples were negative with the Oncomine™ Dx Express Test but produced unknown results with the orthogonal method. One sample was positive with the orthogonal method but produced an unknown result with the Oncomine™ Dx Express Test, and one sample produced unknown results with both methods. Table 20 shows the 3 × 3 contingency table accounting for all 93 NSCLC ALK fusion samples tested by both the Oncomine™ Dx Express Test and the orthogonal method, as well as the concordance analysis.

**Table 20 Concordance between Oncomine™ Dx Express Test and orthogonal method for detection**

| | | Orthogonal method | | | |
|---|---|---|---|---|---|
| | | Positive | Negative | Unknown[1] | Total |
| ODxET | Positive | 26 | 1 | 13 | 40 |
| | Negative | 0 | 41 | 10 | 51 |
| | Unknown[1] | 1 | 0 | 1 | 2 |
| | Total | 27 | 42 | 24 | 93 |
| Excluding unknown | PPA (n/N) [95% CI][2] | 100.0% (26/26) [87.1%, 100.0%] | | | |
| | NPA (n/N) [95% CI][2] | 97.6% (41/42) [87.7%, 99.6%] | | | |

**Table 20 Concordance between Oncomine Dx Express Test and orthogonal method for detection of Level 2 ALK fusions in NSCLC—Accuracy study II** *(continued)*

| | | Orthogonal method | | | |
|---|---|---|---|---|---|
| | | Positive | Negative | Unknown[1] | Total |
| Including unknown | PPA (n/N) [95% CI][2] | 96.3% (26/27) [81.7%, 99.4%] | | | |
| | NPA (n/N) [95% CI][2] | 97.6% (41/42) [87.7%, 99.6%] | | | |

[1] Unknowns are defined as invalid due to insufficient sample, or sample QC sequencing failure resulting in an invalid result or No Call for the variant.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

For RET fusions, 54 NSCLC and thyroid samples were tested using the NGS orthogonal method. Of these, 51 samples (17 positive and 34 negative) had concordant results. One sample was discordant, showing a positive result with the Oncomine™ Dx Express Test but negative with the orthogonal method. Two samples were negative with the Oncomine™ Dx Express Test but produced unknown results with the orthogonal method. Table 21 shows the 3 × 3 contingency table accounting for all 54 NSCLC and thyroid RET fusion samples tested by both the Oncomine™ Dx Express Test and the orthogonal method, along with the concordance analysis.

**Table 21 Concordance between Oncomine™ Dx Express Test and orthogonal method for detection of Level 2 RET fusions in NSCLC and thyroid cancer—Accuracy study II**

| | | Orthogonal method | | | |
|---|---|---|---|---|---|
| | | Positive | Negative | Unknown[1] | Total |
| ODxET | Positive | 17 | 1 | 0 | 18 |
| | Negative | 0 | 34 | 2 | 36 |
| | Unknown[1] | 0 | 0 | 0 | 0 |
| | Total | 17 | 35 | 2 | 54 |
| Excluding unknown | PPA (n/N) [95% CI][2] | 100.0% (17/17) [81.6%, 100.0%] | | | |
| | NPA (n/N) [95% CI][2] | 97.1% (34/35) [85.5%, 99.5%] | | | |
| Including unknown | PPA (n/N) [95% CI][2] | 100.0% (17/17) [81.6%, 100.0%] | | | |
| | NPA (n/N) [95% CI][2] | 97.1% (34/35) [85.5%, 99.5%] | | | |

[1] Unknowns are defined as invalid due to insufficient sample, or sample QC sequencing failure resulting in an invalid result or No Call for the variant.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

Concordance analysis results presented in the preceding tables demonstrate robust detection performance of Oncomine™ Dx Express Test for ERBB2 CNVs in breast cancer, ALK fusions in NSCLC, and RET fusions in NSCLC and thyroid cancer, all of which are classified as Level 2 according to the FDA's Biomarker Class Level for tumor profiling NGS tests.

Asummary of accuracy results (PPA and NPA) for ERBB2 CNV, ALK fusion, and RET fusion variants, stratified by the LoD level established by Oncomine™ Dx Express Test (for example, at or above 1× LoD and below LoD), excluding unknown results, is presented in Table 22. For analysis conditional on the Oncomine™ Dx Express Test LoD levels, PPV was calculated for all reported variant types using the same stratification and exclusion criteria.

**Table 22 Accuracy results for Level 2 ERBB2 CNV, ALK fusion, and RET fusion tumor profiling variants stratified by LoD level**

| Gene alterations | LoD level | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA {n/N) [95% CI][1] | NPA (n/N) [95% CI][1] | PPV (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| ERBB2 CNVs | All | 29 | 4 | 0 | 37 | 100.0% (29/29) [88.3%, 100.0%] | 90.2% (37/41) [77.5%, 96.1%] | 87.9% (29/33) [72.7%, 95.2%] |
| | ≥1X LoD | 29 | 4 | 0 | 37 | 100.0% (29/29) [88.3%, 100.0%] | 90.2% (37/41) [77.5%, 96.1%] | 87.9% (29/33) [72.7%, 95.2%] |
| | <1X LoD | 0 | 0 | 0 | 0 | N/E[2] | N/E | N/E |
| ALK fusion | All | 26 | 1 | 0 | 41 | 100.0% (26/26) [87.1%, 100.0%] | 97.6% (41/42) [87.7%, 99.6%] | 96.3% (26/27) [81.7%, 99.3%] |
| | ≥1X LoD (Molecular counts) | 19 | 1 | 0 | 0 | 100.0% (19/19) [83.2%, 100.0%] | N/E | 95.0% (19/20) [76.4%, 99.1%] |
| | <1X LoD (Molecular counts) | 7 | 0 | 0 | 41 | 100.0% (7/7) [64.6%, 100.0%] | N/E | 100.0% (7/7) [64.6%, 100.0% |
| | ≥1X LoD (Fusion reads) | 22 | 1 | 0 | 0 | 100.0% (22/22) [85.1%, 100.0%] | N/E | 95.7% (22/23) [79.0%,99.2%] |
| | <1X LoD (Fusion reads) | 4 | 0 | 0 | 41 | 100.0% (4/4) [51.0%, 100.0%] | N/E | 100.0% (4/4) [51.0%, 100.0%] |
| RET fusion | All | 17 | 1 | 0 | 34 | 97.1% (34/35) [85.5%, 99.5%] | 97.1% (34/35) [85.5%, 99.5%] | 94.4% (17/18) [74.2%, 99.0%] |

**Table 22 Accuracy results for Level 2 ERBB2 CNV, ALK fusion, and RET fusion tumor profiling variants stratified by LoD level** *(continued)*

| Gene alterations | LoD level | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA {n/N) [95% CI][1] | NPA (n/N) [95% CI][1] | PPV (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|---|---|
| RET fusion | ≥1X LoD (Molecular counts) | 17 | 1 | 0 | 34 | 100.0% (17/17) [81.6%, 100.0%] | 97.1% (34/35) [85.5%, 99.5%] | 94.4% (17/18) [74.2%, 99.0%] |
| | <1X LoD (Molecular counts) | 0 | 0 | 0 | 0 | N/E | N/E | N/E |
| | ≥1X LoD (Fusion reads) | 17 | 1 | 0 | 34 | 100.0% (17/17) [81.6%, 100.0%] | 97.1% (34/35) [85.5%, 99.5%] | 94.4% (17/18) [74.2%, 99.0%] |
| | <1X LoD (Fusion reads) | 0 | 0 | 0 | 0 | N/E | N/E | N/E |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

[2] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

All four discordant ERBB2 CNV samples had tumor content levels between 20% and 39%. The CNV algorithm used by Oncomine™ Dx Express Test requires a minimum tumor content threshold of 40%. If a value below 40% is entered, the algorithm defaults to using 40% as the input. Entering a tumor content value higher than the actual level may deflate the copy number estimate, potentially leading to false negatives. Conversely, entering a lower value than the actual tumor content may inflate the estimate, potentially resulting in false positives. Given this threshold, the detection of ERBB2 CNVs in samples with 20%– 39% tumor content may be unreliable, with an increased risk of inaccurate results, including false negatives. According to the Oncomine™ Dx Express Test User Guide, when tumor content is below 20% and tumor content within the region of interest (ROI) is at least 10% in resection samples, macrodissection is performed to enrich tumor content. However, this process does not mitigate the risk of inaccurate CNV results within the 20%–39% tumor content range. To address this issue, limitation language has been included in the device labeling to state that ERBB2 CNV calls may not be accurate when tumor content is between 20% and 39%.

## Analytical accuracy study III—Tumor profiling variants

The analytical accuracy study III was conducted to evaluate the accuracy of the Oncomine™ Dx Express Test for detecting EGFR exon 19 deletions, L858R, and T790M variants in NSCLC FFPE tissue samples by comparing results between the Oncomine™ Dx Express Test and an orthogonal PCR method. All EGFR variants evaluated in this study were Level 2 variants, classified according to FDA's three-tiered approach for reporting biomarkers in tumor profiling NGS tests. Samples were sourced from qualified commercial vendors and were pre-screened for target variants using an orthogonal method similar to the Oncomine™ Dx Express Test.

Of the 70 samples tested, 68 showed concordance between Oncomine™ Dx Express Test and the orthogonal method. Among these, 23 were positive for the variants of interest (11 for EGFR L858R and T790M, and 12 for EGFR exon 19 deletions), and 45 were negative for EGFR variants. One sample was discordant, showing an EGFR variant-positive result with the Oncomine™ Dx Express Test but negative with the orthogonal method. One sample yielded an unknown result with the orthogonal PCR method.

Concordance analysis was performed at the sample level, and PPA and NPA estimates, along with their respective two-sided 95% CIs, were calculated using the orthogonal test results as the reference, excluding unknown results. The accuracy results (PPA and NPA), summarized by gene and further stratified by the FDA's Biomarker Class Level for tumor profiling NGS tests and by predefined challenging genomic regions (for example, homopolymers, simple sequence repeats [SSRs], and GC content), are presented in Table 16. All EGFR variants were tested at levels above 1X LoD.

**Table 23 Accuracy Results for EGFR exon 19 deletions, L858R, and T790M tumor profiling variants—Accuracy study III**

| Variant type | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|
| All EGFR variants | 23 | 1 | 0 | 45 | 100.0% (23/23) [85.7%, 100.0%] | 97.8% (45/46) [88.7%, 99.6%] |
| Level 2 EGFR variants | 23 | 1 | 0 | 45 | 100.0% (23/23) [85.7%, 100.0%] | 97.8% (45/46) [88.7%, 99.6%] |
| Level 3 EGFR variants | 0 | 0 | 0 | 0 | NE[2] | NE |
| EGFR SNVs | 11 | 1 | 0 | 45 | 100.0% (11/11) [74.1%, 100.0%] | 97.8% (45/46) [88.7%, 99.6%] |
| EGFR deletions | 12 | 0 | 0 | 45 | 100.0% (12/12) [75.8%, 100.0%] | 100.0% (45/45) [92.1%, 100.0%] |
| EGFRs_non-homopolymers | 4 | 0 | — | — | 100.0% (4/4) [51.0%, 100.0%] | N/E |
| EGFRs_homopolymers | 19 | 1 | — | — | 100.0% (19/19) [83.2%, 100.0%] | N/E |
| EGFRs_non-SSRs | 19 | 1 | — | — | 100.0% (19/19) [83.2%, 100.0%] | N/E |
| EGFRs_SSRs | 4 | 0 | — | — | 100.0% (4/4) [51.0%, 100.0%] | N/E |
| EGFRs_GC [30%–40%] | 0 | 0 | — | — | N/E | N/E |
| EGFRs_GC [40%–50%] | 11 | 0 | — | — | 100.0% (11/11) [74.1%, 100.0%] | N/E |

**Table 23 Accuracy Results for EGFR exon 19 deletions, L858R, and T790M tumor profiling variants—Accuracy study III** *(continued)*

| Variant type | ODxET + Comparator + | ODxET + Comparator – | ODxET – Comparator + | ODxET – Comparator – | PPA (n/N) [95% CI][1] | NPA (n/N) [95% CI][1] |
|---|---|---|---|---|---|---|
| EGFRs_GC [50%–60%] | 12 | 1 | — | — | 100.0% (12/12) [75.8%, 100.0%] | N/E |
| EGFRs_GC [60%–75%] | 0 | 0 | — | — | N/E | N/E |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

[2] NE: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.

The discordance observed between the Oncomine™ Dx Express Test and the orthogonal assays can be attributed to inherent design differences, including variant calling thresholds and rules between the two assays.

## Limit of Blank (LoB) study

The limit of blank (LoB) study was conducted to estimate the false positive rate (FPR) for each variant class based on replicate testing of FFPE tumor samples known to be WT for all DNA and RNA variants detectable by Oncomine™ Dx Express Test. A total of 24 FFPE clinical samples from 10 different tissue types were used: bile duct (2), bladder (2), brain (4), breast (2), colon (2), endometrium (2), lung (4), pancreas (2), prostate (2), and skin (2). These samples were prescreened using an orthogonal method and served as blank controls for all reportable variants across all genes in the Oncomine™ Dx Express Test panel. The study was conducted using 2 reagent lots. For each tissue sample, 4 replicates were tested per reagent lot, resulting in a total of 192 measurements per DNA and RNA sample (4 replicates × 2 lots × 24 samples).

A total of 19 false positive calls were observed in the SNV variant type across 24 blank samples, resulting in an FPR of 0.006% (19/315, 072) for overall SNVs when calculated using the total number of SNV positions sequenced as the denominator. The per position and per sample FPRs for Level 3 SNVs across 24 blank samples representing all 10 tissue types were 0.0007% (19/2, 750, 976) and 9.9% (19/192), respectively (Table 24). The per position FPR was calculated for each variant type based on the total number of sequenced SNV positions associated with clinically relevant variants across all tested sample replicates, while the per sample FPR was calculated for each variant type using the total number of tested sample replicates as the denominator. All observed false positives were classified as Level 3 SNVs, with allele frequencies ranging from approximately 2.7% to 5.2% (mean: 3.5%). These values are below both the predicted LoD of the orthogonal assay used for prescreening, and the majority fall at or near the lower end of the established LoD of Oncomine™ Dx Express Test for SNVs (3.24%–7.34%), corresponding to approximately 0.56X to 1.06X relative to the Oncomine™ Dx Express Test LoD. These findings suggest that the observed false positives occurred below or near the assay's LoD for SNVs. Additionally, detection of these Level 3 SNVs exhibited tissue-specific patterns. The tissue-specific per position FPR ranged from 0% to 0.06%, detailed as follows: 0% for colon, endometrium, lung, and skin; 0.005% for breast; 0.006% for brain glioma; 0.009% for bile duct, bladder, and pancreas; 0.02% for pancreas; and 0.06% for brain glioblastoma. The tissue-specific per sample FPR ranged from 0% to 75%, with 0% observed in colon, endometrium, lung, and skin; 6.25% (1/16) in breast; 8.33% (2/24) in brain glioma; 12.5% (2/16) in bile duct, bladder, and pancreas; 25% (4/16) in prostate; and 75% (6/8) in brain glioblastoma. To mitigate the risk of reporting false positives

for Level 3 SNVs, Level 3 SNVs with VAFs below 5% were masked from reporting. After masking these 15 Level 3 SNVs from reporting, the overall FPR for SNVs was 0.0003% (1/315, 072) when calculated using the total number of SNV positions sequenced as the denominator. Additionally, after masking these 15 Level 3 SNVs from reporting, the per position and per sample FPRs for Level 3 SNVs across 24 blank samples representing all 10 tissue types remained 0.00004% (1/2, 750, 976) and 0.52% (1/192), respectively (Table 24).

**Table 24 False positive rates per position and per sample before and after masking of Level 3 SNVs with VAFs <5%**

| Category[1] | Before masking certain Level 3 SNVs with VAFs <5% | | After masking certain Level 3 SNVs with VAFs <5% | |
|---|---|---|---|---|
| | Per position false positive rate[2] | Per sample false positive rate[2] | Per position false positive rate[2] | Per sample false positive rate[2] |
| Level 1: EGFR exon 20 insertion (230) | 0% (0/[230 × 192]) | 0% (0/192) | 0% (0/[230 × 192]) | 0% (0/192) |
| Level 2: Panel-wide SNVs (236) | 0% (0/[236 × 192]) | 0% (0/192) | 0% (0/[236 × 192]) | 0% (0/192) |
| Level 2: Panel-wide INDELs (454) | 0% (0/[454 × 192]) | 0% (0/192) | 0% (0/[454 × 192]) | 0% (0/192) |
| Level 2: Panel-wide fusions and splice variants (3286) | 0% (0/[3286 × 192]) | 0% (0/192) | 0% (0/[3286 × 192]) | 0% (0/192) |
| Level 2: Panel-wide CNVs (4) | 0% (0/[4 × 192]) | 0% (0/192) | 0% (0/[4 × 192]) | 0% (0/192) |
| Level 3: Panel-wide SNVs (14328) | 0.0007% (19/[14328 × 192]) | 9.9% (19/192) | 0.00004% (1/[14328 × 192]) | 0.52% (1/192) |
| Level 3: Panel-wide INDELs (8666) | 0% (0/[8666 × 192]) | 0% (0/192) | 0% (0/[8666 × 192]) | 0% (0/192) |
| Level 3: Panel-wide fusions and splice variants (7604) | 0% (0/[7604 × 192]) | 0% (0/192) | 0% (0/[7604 × 192]) | 0% (0/192) |
| Level 3: Panel-wide CNVs (105) | 0% (0/[105 × 192]) | 0% (0/192) | 0% (0/[105 × 192]) | 0% (0/192) |

[1] The number in parentheses in the Category column represents the sum of all variants across all tested cancer type included in the study for each variant category and reporting level.

[2] The false positve rate was calculated from 2 reagent lots (N=192 data points).

A supplemental LoB study was conducted using 12 leftover tissue samples representing 10 different tissue types selected from the original LoB and tested using a different orthogonal assay. The orthogonal assay detected 44 DNA variants confirmed to be true positives, all present at or above the assay's LoD. None of these variants are targeted by Oncomine™ Dx Express Test, as they are not included in the Oncomine™ Dx Express Test panel. Among the 12 samples used in the supplemental study, Oncomine™ Dx Express Test did not detect any false positive reportable variants.

The results and software validation documentation from regression testing on the configuration with restricted reporting of certain Level 3 tumor profiling SNVs with variant allele frequencies (VAFs) below 5%, intended to prevent reporting of false positives, have not yet been provided. These results and documentation will be submitted in a post-market setting.

## Limit of Detection (LoD) study

- Limit of Detection (LoD) for CDx variants

  The LoD of the Oncomine™ Dx Express Test for EGFR exon 20 insertion variants was determined using 4 patient-derived samples containing insertions of 3, 6, 9, and 12 bp. DNA from these samples was blended with DNA extracted from a WT NSCLC sample to create 5 or 6 variant allele frequency (VAF) levels per insertion type. Each blend level was tested using 2 reagent lots, with 10 replicates per lot, resulting in a total of 20 replicates per level. Detection rates across the blend levels were analyzed using probit regression to estimate the LoD, defined as the lowest VAF predicted to achieve a detection rate of at least 95%. The estimated LoD for EGFR exon 20 insertion variants ranged from 3.24% to 4.38% VAF (mean = 3.78%).

- Limit of Detection (LoD) for tumor profiling variants

  The LoD of the Oncomine™ Dx Express Test was determined for SNVs, insertions, deletions, CNVs, fusions, and splice variants. The LoD for SNVs, insertions, and deletions was established using a representative variant approach, while the LoD for CNVs, fusions, and splice variants was assessed at the gene level. The variant set evaluated comprised 19 SNVs, 14 insertion and deletion variants, 15 CNVs, 26 fusions and exon-skipping variants, and 4 RNA imbalance variants. DNA and RNA blends were prepared using clinical specimens representing ten cancer types: bladder, breast, cholangiocarcinoma, colorectal, endometrial, glioma, non-small cell lung, melanoma, pancreatic, and thyroid cancers. For each variant type, at least 120 data points were generated by testing 6 blend levels using 2 reagent lots, with 10 replicates per level per lot (10 replicates per reagent lot × 2 lots × 6 blend levels). LoD values were calculated using probit regression based on the detection rates across blend levels. The LoD was defined as the lowest VAF or copy number predicted to yield a ≥95% detection rate. If the Probit model did not provide a reasonable fit to the data, the empirical hit rate method was used instead, in which the LoD was defined as the lowest level with an observed detection rate of at least 95%.

  The estimated LoDs for SNVs ranged from 3.24% to 7.34% VAF, with a mean of 5.29%. For insertions, LoDs ranged from 2.98% to 5.24% VAF (mean = 4.11%), and for deletions, from 3.08% to 4.67% VAF (mean = 3.88%). CNVs had LoDs ranging from 4.56 to 5.78 copies, with a mean of 4.99 copies.

  The LoDs for the evaluated RNA fusions and splice variants ranged from 5.0 to 15.5 molecular counts (mean = 8.68). Four fusions assessed using imbalance scores based on unequal expression of the 2 alleles of gene demonstrated LoDs ranging from 1.5 to 4.93 (mean = 2.75). The LoD estimates were determined using either the Probit regression method or the empirical hit rate method, depending on the quality of model fit.

## Nucleic acid input study

- DNA and RNA input for CDx variants

  To verify the performance of Oncomine™ Dx Express Test at the product requirement of 10 ng nucleic acid input and to establish the acceptable range of DNA input for accurate detection of EGFR exon 20 insertion variants, 5 DNA-RNA input combinations were evaluated (5 ng, 7.5 ng, 10 ng (standard input), 12.5 ng, and 15 ng). Each combination was tested as a single sample to

assess the impact of varying RNA quantities relative to DNA input. The study used DNA extracted from four FFPE NSCLC patient samples containing EGFR exon 20 insertions of 3, 6, 9, and 12 bp, each blended with WT FFPE NSCLC DNA to achieve VAFs at 1–3X the LoD.

Analysis of variance (ANOVA) was performed to evaluate the relationship between input levels and VAF. No statistically significant differences in VAF were observed across the different input levels. The PPA for all DNA reference locations was 100% across all input levels. The lowest observed NPA was 99.94% excluding no calls, and 97.39% including no calls, across all DNA input levels. These results support the performance of Oncomine™ Dx Express Test at the recommended input of 10 ng and confirm its robustness across a range of DNA and RNA input levels for the accurate detection of EGFR exon 20 insertion variants.

- DNA and RNA input for tumor profiling variants

  The recommended input level for FFPE DNA and RNA samples is 10 ng for detection of tumor profiling variants. To assess the impact of nucleic acid input amounts on Oncomine™ Dx Express Test performance, DNA and RNA extracted from clinical FFPE samples were tested at 3 input levels (5 ng, 10 ng (standard input), and 15 ng) for tumor profiling variants, and at 5 input levels (5 ng, 7.5 ng, 10 ng (standard input), 12.5 ng, and 15 ng) for selected tumor profiling variants. These selected variants included NSCLC ERBB2 exon 20 insertion, NSCLC EGFR exon 19 deletion, EGFR L858R and T790M, BRAF V600E, KRAS G12C, glioma IDH1 and IDH2 SNVs, NSCLC ALK, RET, and ROS1 fusions, and NSCLC MET exon 14 skipping variants.

  The FFPE specimens included variants across 25 genes and represented 11 cancer types, including bladder, breast, cholangiocarcinoma, colorectal, endometrial, glioma, melanoma, non-small cell lung, pancreatic, prostate, and thyroid cancers. The analysis included a range of DNA and RNA variant types, including SNVs, MNVs, deletions, insertions, CNVs, RNA fusions, and splice variants. A total of 37 DNA blends were prepared to achieve VAFs at 1–3X the LoD, and 34 RNA blends were prepared to achieve read counts at 1–3X the LoD for fusions and splice variants. Each blend was tested at each input level using either 6 or 12 replicates per variant to assess the effect of input amount on positive and negative call rates.

  The results demonstrated that the PPA for SNVs, insertions, deletions, and CNVs was 100% across all DNA input levels. The NPA, including no calls at negative variant locations, was ≥99.64% across all DNA input levels, with the exception of one ERBB2 exon 20 insertion variant, which showed an NPA of 97.39% including no calls and 99.95% excluding no calls at the 7.5 ng input level. For all other DNA blends, the NPA excluding no calls was ≥99.84% at all input levels. For RNA fusions and splice variants evaluated, the PPA was 100% across all input levels. The NPA including no calls, was ≥99.26%, and the NPA excluding no calls was ≥99.90% across all input levels. These results confirm the performance of Oncomine™ Dx Express Test at the recommended 10 ng input level for accurate detection of tumor profiling variants.

  The minimum nucleic acid concentration required for the Oncomine™ Dx Express Test is 0.5 ng/µL. To verify Oncomine™ Dx Express Test performance at the minimum concentration for detecting both CDx and tumor profiling variants, variant-positive DNA and RNA samples were tested in 6 replicates under 3 conditions: low concentration (Level 1, 0.25 ng/µL), standard concentration (Level 2, 0.50 ng/µL), and high concentration (Level 3, 0.75 ng/µL). Summary statistics were generated for each concentration level, and equivalence across levels was assessed using Welch's ANOVA test at a significance level of  =0.05. Among 11 CNVs, FGFR1 CNV, and among 10 RNA fusion variants, 6 fusions showed no statistical equivalence in summary statistics across concentration levels. Variant call concordance data (PPA and NPA) were not assessed in this study.

## Tissue input study

This study was performed to verify that FFPE sections cut from resections, core needle biopsies (CNBs), and cell blocks from fine needle aspirate (FNA) samples can yield the minimum nucleic acid concentration required for Oncomine™ Dx Express Test (0.5 ng/µL), and produce the expected variant calls for SNVs, insertions, deletions, CNVs, fusions, and RNA splice variants. The study included samples from a diverse range of tissue types, including bile duct, bladder, brain, breast, colon, endometrium, lung, pancreas, skin, and thyroid. A total of 48 FFPE tissue resection samples, 24 CNB samples, and 12 cell blocks from FNA samples were collected and prescreened using an orthogonal NGS method. In the study, 2 × 5 µm sections from FFPE resection blocks with a tumor cell content of at least 20% were used. For FFPE CNB samples, 9 × 5 µm sections were used, and for FFPE FNA cell blocks, 7 × 5 µm sections were used, each with a tumor cell content of at least 10%. Nucleic acid extractions and quantitation were performed using the Genexus™ Dx Purification System.

When FFPE section requirements as described above for each specimen type were met, the results demonstrated that 98.8% (83/84) of samples met the minimum DNA concentration requirements of 0.5 ng/µL, with one CNB sample yielding a DNA concentration <0.5 ng/µL. All samples (84/84) met theminimum RNA concentration requirements of 0.5 ng/µL. The PPA for the variants of interest between Oncomine™ Dx Express Test and the orthogonal method was 95.52% (64/67) for resection samples, 100% (27/27) for CNB samples, and 100% (13/13) for FNA cell block samples. These data support the performance of Oncomine™ Dx Express Test in detecting both CDx and tumor profiling variants when using the recommended FFPE section input levels: 2 × 5 µm sections from resection samples, 9 × 5 µm sections from CNB, or 7 × 5 µm sections from FNA cell blocks.

## Tumor content study

To evaluate the minimum tumor content needed for detection of EGFR exon 20 insertion variants in NSCLC FFPE tissue using Oncomine™ Dx Express Test, a total of 89 EGFR exon 20 insertion-positive samples and 111 negative reference samples were analyzed. Tumor content was stratified into 5 bins: 0–9%, 10–19%, 20–29%, 30–39%, and ≥40% for each specimen type. No FNA cell block samples were available in the 10–19% tumor content bin.

All samples with valid results from both Oncomine™ Dx Express Test and the comparator method were included in the concordance analysis. Variant calling agreement was analyzed by tumor content bin and specimen type (resection, CNB, and FNA cell block). When excluding unknown results, the PPA was 100% across all tumor content levels for all specimen types. The NPA was 100% for all tumor content levels tested in CNB and FNA cell block samples. For resection samples, the NPA was 100% (52/52) at 10–29% tumor content, 91.9% (34/37) at 30–39%, and 98.4% (60/61) at ≥40%.

A 100% PPA was observed for resection samples with tumor content ≥20% and for CNB samples with tumor content ≥10%. These results support the minimum tumor content requirements of ≥20% for resections and ≥10% for CNBs. However, due to the absence of FNA cell block samples in the 10–19% tumor content bin, and based on available data, the minimum tumor content requirement for FNA cell block samples has been updated from ≥10% to ≥20%.

## Interference study

The impact of potentially interfering substances on the performance of the Oncomine™ Dx Express Test was evaluated using clinical FFPE tissue samples from 10 cancer types: bladder, breast, cholangiocarcinoma, colorectal, endometrial, glioma, NSCLC, melanoma, pancreatic, and thyroid. Each sample contained tumor profiling variants (for example, SNVs, insertions, deletions, CNVs, and fusions) or CDx variants (EGFR exon 20 insertions). Testing was conducted with potentially interfering substances that might be observed in these specific tissue types. Exogenous substances included paraffin, proteinase K, wash solution, and xylene; endogenous substances included hemoglobin, melanin, colloid/thyroglobulin, lipids/triglycerides, and unconjugated bile acids. Table 25 shows the interferent evaluated.

**Table 25 Interferent evaluated**

| Interferent | Condition | Tumor type |
|---|---|---|
| Paraffin | 4X of normally expected levels | All tumor types |
| Proteinase K | >10X of normally expected of the residual Proteinase K post the heat-kill step | All tumor types |
| Wash buffer | Equivalent to approximately 10% wash 2 carried over into eluate | All tumor types |
| Xylene | 6X of normally expected residual levels | All tumor types |
| Hemoglobin | Endogenous protein from blood and tissues (4 mg/mL) | All tumor types |
| Melanin | Endogenous substance from melanoma tissues (0.2 µg/mL) | Melanoma |
| Colloid/Thyroglobulin | Endogenous to thyroid tissues (58.2% Colloid) | Thyroid |
| Colloid/Thyroglobulin | Endogenous to thyroid tissues (54% Colloid) | Thyroid |
| Lipids/Triglycerides | 37 mmol/L | Cholangiocarcinoma and breast |
| Unconjugated bile acids | 30 mol/L | Cholangiocarcinoma |

FFPE samples were processed through the entire Oncomine™ Dx Express Test workflow and spiked with the potentially interfering substances. When needed, variant-positive samples were blended with nucleic acid extracted from WT tissue under the same interferent condition to generate sample blends with VAFs near 1.5X LoD for DNA or near 2X LoD for RNA. Six replicates were tested per condition for each sample or sample blend.

Concordance data (PPA and NPA) comparing the control to each of the 9 tested interferents for variants of interest, stratified by variant type, were analyzed. For all variants of interest, the PPA (excluding no calls) was 100%, and the NPA (excluding no calls) was also 100% across 8 of the 9 tested interferents. In the presence of Proteinase K, the PPA remained at 100%, while the NPA slightly decreased to 99.91% for SNVs and indels; however, the NPA for CNVs and fusions remained at 100%. These results demonstrate that the evaluated interfering substances do not adversely affect Oncomine™ Dx Express Test performance.

## Tumor necrosis study

- Assessment of tumor necrosis impact on CDx variant detection

  The impact of necrotic tissue on Oncomine™ Dx Express Test performance for the detection of CDx variants was evaluated using 2 FFPE NSCLC patient samples harboring EGFR exon 20 insertion variants (6 bp or 9 bp). To simulate high levels of necrosis, DNA from each variant-positive sample was blended with DNA from a highly necrotic (75% necrosis) NSCLC sample lacking the variant of interest. Two unique blends were generated to simulate necrosis levels of 66% and 71%, respectively, while maintaining the EGFR exon 20 insertion variant at a VAF near the LoD. For comparison, two neat (unblended) variant-positive samples were also included. Each sample was tested in 6 sequencing replicates.

  The PPA was assessed by comparing the rate of positive calls between the neat and necrotic conditions. The PPA for EGFR exon 20 insertion variants was 100% in samples with up to 71% necrosis compared to the control condition. These results support that Oncomine™ Dx Express Test can reliably detect EGFR exon 20 insertion variants in FFPE samples with up to approximately 70% necrosis in the region of interest.

- Assessment of tumor necrosis impact on tumor profiling variant detection

  The impact of necrotic tissue on Oncomine™ Dx Express Test performance for the detection of selected tumor profiling variants, including EGFR SNVs (T790M and L858R), EGFR exon 19 deletion, ERBB2 exon 20 insertion, and ERBB2 SNVs, was evaluated using 2 FFPE NSCLC patient samples per variant. To simulate high levels of necrosis, DNA from each variant-positive sample was blended with DNA from a highly necrotic (80% necrosis) NSCLC sample that lacked the variant of interest. The resulting tumor necrosis levels ranged from 56% to 73%. For comparison, 2 neat (unblended) variant-positive samples were also included. Each sample was tested in 6 sequencing replicates.

  The PPA was assessed by comparing the rate of positive calls between the neat and necrotic conditions. The PPA for all tested variants was 100% in samples with the necrosis levels ranging from 56% to 73% for each variant. These results support that Oncomine™ Dx Express Test can reliably detect tumor profiling variants, including SNVs, insertions, and deletions, in FFPE samples with up to approximately 70% necrosis in the region of interest.

## Carryover and cross-contamination study

Cross-contamination throughout the Oncomine™ Dx Express Test workflow, from nucleic acid extraction to sequencing, was assessed. Six FFPE tissue samples containing various types of genetic variants (including SNVs, insertions, CNVs, and fusions) with high VAFs were used to ensure adequate sample quality and to test under challenging conditions. These samples were processed using the entire Oncomine™ Dx Express Test workflow on the Genexus™ Dx System. The evaluated biomarker-positive samples included: EGFR exon 20 insertion (9 bp) in NSCLC, IDH1 R132H in glioma, ERBB2 amplification in bladder cancer, RSPO3 splice variant in colorectal cancer, FGFR3 fusion in bladder cancer, and EGFR vIII splice variant in glioma. Blank samples were included and arranged in a checkerboard pattern alongside variant-positive samples to monitor potential cross-contamination. To evaluate potential carryover contamination, sample positions were alternated across consecutive runs performed on the same instrument, and unique barcodes were applied to each library.

All variant-positive samples were detected by Oncomine™ Dx Express Test with 100% PPA (15/15 per variant). None of the target variants were detected in blank samples, other positive samples with non-target variants, or no template controls (NTCs). The overall FPR was 0% for both DNA variants and RNA variants. However, WT DNA contamination was observed in 6 blank samples across and within

runs, resulting in negative calls. This contamination likely occurred during sample preprocessing steps in the laboratory environment and was not attributed to the automated workflow.

## Primer specificity—*in-silico* analysis study

An *in-silico* cross-reactivity analysis was conducted to assess the specificity of the 1,556 primers used in the Oncomine™ Dx Express Test DNA and RNA panels. Each primer sequence was evaluated for homology to non-target regions in nucleic acids expected to be present in test samples, primarily human total nucleic acid (TNA) consisting of DNA and RNA. The primers underwent a comprehensive specificity assessment against the human genome, human RefSeq Select transcripts, and representative bacterial, fungal, and viral genomes.

*In-silico* analysis identified 883 unintended primer pairings that could potentially generate amplicons from the human genome and 311 unintended pairings from human transcripts. Of these, 38 unintended amplicons (16 from the DNA panel primers and 17 from the RNA panel primers) showed significant sequence similarity to one or more of the 33 intended inserts. However, during Oncomine™ Dx Express Test testing, which included 630 sequencing libraries from 11 FFPE tissue samples and 165 libraries from five cfTNA samples, misprimedor mismapped reads at intended amplicon targets were observed in only 5 cases. None of these 5 instances are predicted to result in false positive or false negative outcomes.

The *in-silico* specificity analysis against representative bacterial, fungal, and viral genomes identified 17 predicted unintended amplicons, each with a mismatch of ≥2 bp relative to the intended amplicons; however, none showed significant similarity to intended targets and are not expected to affect test accuracy. Based on these findings, the DNA and RNA primers used in the Oncomine™ Dx Express Test were deemed specific.

## Precision studies

Two reproducibility studies were conducted to evaluate the precision and reproducibility of the Oncomine™ Dx Express Test: a sample processing reproducibility study and an external panel reproducibility study. The sample processing reproducibility study began with the nucleic acid extraction step to assess the reproducibility and repeatability of the entire sample processing workflow. The panel reproducibility study began with extracted nucleic acid.

### Sample processing reproducibility studies

As the sample processing reproducibility study began at the nucleic acid extraction step, it was conducted using neat (undiluted) samples. Consequently, the variant allele frequency (VAF) levels in these samples were relatively high, and samples with VAFs near the LoD were not assessed in this study. To evaluate reproducibility at or near the assay's LoD, samples with variant levels near the LoD (1–1.5X LoD) were evaluated separately in the panel reproducibility study. For details on the panel reproducibility study, see "Assay panel reproducibility study I—CDx variants" on page 65.

### Sample processing reproducibility study I—CDx variants

The sample processing reproducibility study was conducted at an internal test site to evaluate the reproducibility of the Oncomine™ Dx Express Test workflow for detecting EGFR exon 20 insertion variants in NSCLC FFPE tissue samples. Four NSCLC FFPE samples harboring EGFR exon 20 insertions of 3, 6, 9, and 12 bp in length, along with one WT NSCLC FFPE sample, were used in this study. Testing for each variant was performed by 2 operators using 3 different reagent lots at a

single internal test site. Each operator conducted 3 sequencing runs (2 replicates per sample per run) on separate days, resulting in 12 replicates per sample (2 replicates × 2 operators × 3 lots).

The call rate, no call rate, positive call rate, and negative call rate along with corresponding two-sided 95% CIs using the Wilson Score method, were calculated for each variant of interest. The positive call rate was 100% for all EGFR exon 20 insertion variants, with no instances of No Calls observed, and the negative call rate for the WT sample was also 100% Table 26.

**Table 26 Sample reproducibility for EGFR exon 20 insertion variants**

| Variant | Variant ID | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | Positive call rate (95% CI)[1] | Negative call rate (95% CI)[1] |
|---|---|---|---|---|---|---|---|---|
| EGFR exon 20 insertion (3 bp) | COSM12377 | 2.52X | 12 | 12 | 0 | 0 | 100.0% (75.8%,100.0%) | 0.0% (0.0%,24.2%) |
| EGFR exon 20 insertion (6 bp) | COSM1238028 | 6.94X | 12 | 12 | 0 | 0 | 100.0% (75.8%,100.0%) | 0.0% (0.0%,24.2%) |
| EGFR exon 20 insertion (9 bp) | COSM12376 | 3.76X | 12 | 12 | 0 | 0 | 100.0% (75.8%,100.0%) | 0.0% (0.0%,24.2%) |
| EGFR exon 20 insertion (12 bp) | COSM26720 | 4.48X | 12 | 12 | 0 | 0 | 100.0% (75.8%,100.0%) | 0.0% (0.0%,24.2%) |
| WT | N/A | N/A | 132 | 0 | 132 | 0 | N/A | 100.0% (97.2%,100.0%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

## Sample processing reproducibility study II—Tumor profiling variants

The sample processing reproducibility study was conducted at an internal test site to evaluate the reproducibility of the Oncomine™ Dx Express Test workflow for detecting tumor profiling variants, including SNVs, insertions, deletions, CNVs, fusions, alternate splice variants, and imbalance (unequal expression of the two alleles of gene), in FFPE tissue samples. A total of 57 clinical samples were selected for the study, including 46 biomarker-positive samples, and 11 WT samples across multiple tumor types: bladder, cholangiocarcinoma, melanoma, prostate, NSCLC, thyroid, breast, colorectal, endometrial, and glioma. The biomarker-positive samples included 35 DNA variants (12 SNVs, 1 MNV, 5 insertions, 6 deletions, and 11 CNVs) and 22 RNA variants (18 fusions, 3 splice variants, and 1 imbalance).Testing for each variant was performed at a single internal test site by two operators using 3 different lots of reagents, with each operator performing 3 sequencing runs (2 replicates per sample per run) on separate days, resulting in 12 replicates per sample (2 replicates × 2 operator × 3 lots).

The call rate, no call rate, positive call rate, and negative call rate along with corresponding two-sided 95% CI using the Wilson Score method, were calculated for each variant of interest. As presented in Table 27, the positive call rate was 100% for all SNVs, insertions, and deletions when excluding no calls. The negative call rate in the WT samples was also 100% when excluding no calls

**Table 27 Sample reproducibility for tumor profiling SNVs and INDELs**

| Variant type | Gene | Variant ID | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| SNV | BRAF | COSM476 (V600E) | Colo-rectal | Level 2 | 4.79X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | BRAF | COSM476 (V600E) | NSCLC | Level 2 | 2.52X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | EGFR | COSM6224 (L858R) | NSCLC | Level 2 | 3.1X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | EGFR | COSM6224 (L858R) | NSCLC | Level 2 | 2.84X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | EGFR | COSM6240 (T790M) | NSCLC | Level 2 | 3.98X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | EGFR | COSM6240 (T790M) | NSCLC | Level 2 | 3.86X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | ERBB2 | COSM48358 (S310F) | Bladder | Level 3 | 4.29X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | FGFR3 | COSM715 (S249C) | Bladder | Level 2 | 2.72X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | IDH1 | COSM28747 (R132C) | CCA* | Level 2 | 1.00X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | KRAS | COSM516 (G12C) | NSCLC | Level 2 | 1.96X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |

**Table 27 Sample reproducibility for tumor profiling SNVs and INDELs** *(continued)*

| Variant type | Gene | Variant ID | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| SNV | KRAS2 | COSM518[3] (G12R) | Pan-creatic | Level 3 | 1.67X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | PIK3CA | COSM763 (E545K) | Breast | Level 2 | 1.80X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | PIK3CA | COSM775 (H1047R) | Breast | Level 2 | 1.79X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| SNV | RET3 | COSM964[4] (C630S) | Thyroid | Level 2 | 7.74X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| MNV | BRAF | COSM473 (V600K) | Mela-noma | Level 2 | 5.17X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Insertion | EGFR4 | COSM655155[5] (Exon 20 insertion, 6 bp) | Endo-metrial | Level 3 | 2.35X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Insertion | ERBB2 | COSM12552 (Exon 20 insertion, 3 bp) | NSCLC | Level 2 | 2.17X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Insertion | ERBB2 | COSM303939 (Exon 20 insertion, 6 bp) | NSCLC | Level 2 | 6.11X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Insertion | ERBB2 | COSM20959 (Exon 20 insertion, 12 bp) | NSCLC | Level 2 | 1.92X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Insertion | ERBB2 | COSM20959 (Exon 20 insertion, 12 bp) | Bladder | Level 3 | 4.96X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Deletion | EGFR | COSM13557 (Exon 19 deletion, 9 bp) | NSCLC | Level 2 | 3.67X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Deletion | EGFR | COSM12370 (Exon 19 deletion, 12 bp) | NSCLC | Level 2 | 7.11X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |

**Table 27 Sample reproducibility for tumor profiling SNVs and INDELs** *(continued)*

| Variant type | Gene | Variant ID | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| Deletion | EGFR | COSM6223 (Exon 19 deletion, 15 bp) | NSCLC | Level 2 | 3.84X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Deletion | TP53 | COSM43570 (18 bp deletion) | Breast | Level 3 | 7.64X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Deletion | TP53 | COSM45140 (3 bp deletion) | Pan-creatic | Level 3 | 3.13X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| Deletion | PIK3CA | COSM24710 (3 bp deletion) | Colo-rectal | Level 3 | 3.90X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| WT | WT01 | N/A | Bladder | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT02 | N/A | CCA[6] | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT03 | N/A | Mela-noma | N/A | N/A | 192 | 0 | 191 | 1 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT04 | N/A | Prostate | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT05 | N/A | NSCLC | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT06 | N/A | Thyroid | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT07 | N/A | Breast | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT08 | N/A | Colo-rectal | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |

**Table 27 Sample reproducibility for tumor profiling SNVs and INDELs** *(continued)*

| Variant type | Gene | Variant ID | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| WT | WT09 | N/A | Endo-metrial | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT10 | N/A | Glioma | N/A | N/A | 192 | 0 | 192 | 0 | 0.0% (0.0%, 2.0%) | 100.0% (98.0%, 100.0%) |
| | WT11 | N/A | NSCLC | N/A | N/A | 132 | 0 | 132 | 0 | 0.0% (0.0%, 2.1%) | 100.0% (97.2%, 100.0%) |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.
[2] 95% 2-sided confidence interval calculated via the Wilson Score method.
[3] LoD for COSM518 was estimated using data from COSM516.
[4] LoD for COSM964 was estimated using data from COSM980.
[5] LoD for COSM655155 was estimated using data from COSM1238028.
[6] CCA: cholangiocarcinoma.

Table 28 presents the call rate, No Call rate, positive call rate, and negative call rate for CNVs, along with corresponding two-sided 95% CIs calculated using the Wilson Score method. The positive call rate was 100% for all CNVs when excluding No Calls. The negative call rate in the WT samples was also 100% when excluding No Calls.

**Table 28 Sample reproducibility for tumor profiling CNVs**

| Gene | Tissue type | Reporting level[1] | Variant level (XLoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| FGFR3 | Bladder | Level 3 | 1.95X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| EGFR | Glioma | Level 3 | 1.78X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| MET | Glioma | Level 3 | 1.92X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR1 | Breast | Level 3 | 1.94X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |

**Table 28 Sample reproducibility for tumor profiling CNVs** *(continued)*

| Gene | Tissue type | Reporting level[1] | Variant level (XLoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| PIK3CA | Breast | Level 3 | 1.67X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR2 | Breast | Level 3 | 2.25X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| AR | Breast | Level 3 | 1.11X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ERBB2 | Endometrial | Level 3 | 2.05X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| KRAS | NSCLC | Level 2 | 1.47X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| AR | Prostate | Level 3 | 1.20X | 11 | 11 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ERBB3 | Melanoma | Level 3 | 1.33× | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| WT01 | Bladder | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT02 | CCA[3] | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT03 | Melanoma | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT04 | Prostate | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT05 | NSCLC | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |

**Table 28 Sample reproducibility for tumor profiling CNVs** *(continued)*

| Gene | Tissue type | Reporting level[1] | Variant level (XLoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| WT06 | Thyroid | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT07 | Breast | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT08 | Colorectal | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT09 | Endometrial | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |
| WT10 | Glioma | N/A | N/A | 120 | 0 | 120 | 0 | 0.0% (0.0%, 3.1%) | 100.0% (96.9%, 100.0%) |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

[3] CCA: cholangiocarcinoma.

Table 29 shows the call rate, No Call rate, positive call rate, and negative call rate for fusion and alternate splice variants, along with corresponding two-sided 95% CIs calculated using the Wilson Score method. The positive call rate was 100% for all fusions and alternate splice form variants when excluding no calls. The negative call rate in the WT samples was also 100% when excluding No Calls.

**Table 29 Sample reproducibility for tumor profiling fusions and splice variants**

| Gene | Variant | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| RSPO2 Fusion | EIF3E-RSPO2.E1R2 .COSF1307.1 | Colorectal | Level 3 | 10.75X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| NTRK1 Fusion | TPM3-NTRK1.T7N10 .COSF1329 | Colorectal | Level 2 | 5.05X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| NTRK2 Fusion | BCR-NTRK2 .B1N17 | Glioma | Level 2 | 6.51X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR1 Fusion | FGFR1-TACC1.F17T7 .COSF1362.1 | NSCLC | Level 3 | 76.86X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR3 Fusion | FGFR3-TACC3.F17T7.1 | Bladder | Level 2 | 12.91X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR3 Fusion | FGFR3-TACC3.F17T8 .COSF1353 | Bladder | Level 2 | 131.33X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| MET Fusion | ST7-MET.S1M2 | Glioma | Level 3 | 9.40X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ESR1 Fusion | ESR1-CCDC170 .E2C2 | Breast | Level 3 | 2.27X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| FGFR2 Fusion | FGFR2-CIT .F17C23.1 | CCA[3] | Level 2 | 22.9X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| RSPO3 Fusion | PTPRK-RSPO3.P1R2 .COSF1311.1 | Colorectal | Level 3 | 3.00X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| BRAF Fusion | KIAA1549-BRAF.K15B9 .COSF481.2 | Glioma | Level 2 | 2.85X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |

**Table 29 Sample reproducibility for tumor profiling fusions and splice variants** *(continued)*

| Gene | Variant | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| NRG1 Fusion | CD74-NRG1.C6N6.1 | NSCLC | Level 3 | 8.84X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ROS1 Fusion | CD74-ROS1.C6R33 | NSCLC | Level 2 | 3.42X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ROS1 Fusion | CD74-ROS1.C6R34 .COSF1200.1 | NSCLC | Level 2 | 49.9X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ROS1 Fusion | CD74-ROS1.C6R35.1 | NSCLC | Level 2 | 7.30X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| NTRK3 Fusion | ETV6-NTRK3.E5N14.1 | Thyroid | Level 2 | 4.60X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| RET Fusion | KIF5B-RET.K15R12 .COSF1232.1 | NSCLC | Level 2 | 10.14X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| ALK Fusion | EML4-ALK.E13A20 .COSF408.2 | NSCLC | Level 2 | 5.10X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| EGFR Splice variant | EGFRvIII .E1E8.Del1 | Glioma | Level 3 | 6.57X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| MET Splice variant | MET-MET.M13M15.1 | NSCLC | Level 2 | 3.27X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| AR Splice variant | AR-AR.A3A4.V7 | Prostate | Level 3 | 10.37X | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| WT01 | N/A | Bladder | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT02 | N/A | CCA | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |

**Table 29 Sample reproducibility for tumor profiling fusions and splice variants** *(continued)*

| Gene | Variant | Tissue type | Reporting level[1] | Variant level (X LoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| WT03 | N/A | Melanoma | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT04 | N/A | Prostate | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT05 | N/A | NSCLC | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT06 | N/A | Thyroid | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT07 | N/A | Breast | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT08 | N/A | Colorectal | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT09 | N/A | Endo-metrial | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |
| WT10 | N/A | Glioma | N/A | N/A | 252 | 0 | 252 | 0 | 0.0% (0.0%, 1.5%) | 100.0% (98.5%, 100.0%) |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.
[2] 95% 2-sided confidence interval calculated via the Wilson Score method.
[3] CCA: cholangiocarcinoma.

Table 30 presents the call rate, No Call rate, positive call rate, and negative call rate for imbalance variants, along with corresponding two-sided 95% confidence intervals calculated using the Wilson Score method. The positive call rate was 100% for all imbalance variants when excluding no calls. The negative call rate in the WT samples was also 100% when excluding no calls.

**Table 30 Sample reproducibility for tumor profiling imbalance variants**

| Gene | Tissue type | Reporting level[1] | Variant level (XLoD) | Total calls | Positive calls | Negative calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive call rate (95% CI)[2] | Negative call rate (95% CI)[2] |
| ALK | NSCLC | Level 3 | N/A | 12 | 12 | 0 | 0 | 100.0% (75.8%, 100.0%) | 0.0% (0.0%, 24.2%) |
| WT01 | Bladder | N/A | N/A | 12 | 0 | 0 | 12 | N/E[3] | N/E |
| WT02 | CCA[4] | N/A | N/A | 12 | 0 | 0 | 12 | N/E | N/E |
| WT03 | Melanoma | N/A | N/A | 12 | 0 | 0 | 12 | N/E | N/E |
| WT04 | Prostate | N/A | N/A | 12 | 0 | 0 | 12 | N/E | N/E |
| WT05 | NSCLC | N/A | N/A | 12 | 0 | 0 | 12 | N/E | N/E |
| WT06 | Thyroid | N/A | N/A | 12 | 0 | 0 | 12 | N/E | N/E |
| WT07 | Breast | N/A | N/A | 11 | 0 | 4 | 7 | 0.0% (0.0%, 49.0%) | 100.0% (51.0%, 100.0%) |
| WT08 | Colorectal | N/A | N/A | 12 | 0 | 1 | 11 | 0.0% (0.0%, 79.3%) | 100.0% (20.7%, 100.0%) |
| WT09 | Endometrial | N/A | N/A | 11 | 0 | 0 | 11 | N/E | N/E |
| WT10 | Glioma | N/A | N/A | 12 | 0 | 12 | 0 | 0.0% (0.0%, 24.2%) | 100.0% (75.8%, 100.0%) |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.
[2] 95% 2-sided confidence interval calculated via the Wilson Score method.
[3] N/E: Not evaluable. The statistic cannot be calculated as there were no sample variant results in this category.
[4] CCA: cholangiocarcinoma.

## Assay panel reproducibility studies

For samples evaluated only at the 1–1.5X LoD level in the panel reproducibility study, reproducibility data at higher variant allele frequency (VAF) levels (>2X LoD) were leveraged from the sample processing reproducibility study, which used neat (undiluted) samples with VAFs >2X LoD, and began at the nucleic acid extraction step. For details on the sample processing reproducibility study, see “Sample processing reproducibility study I—CDx variants” on page 54.

## Assay panel reproducibility study I—CDx variants

The panel reproducibility study, starting from DNA, was conducted across 3 test sites to evaluate the repeatability and reproducibility of the Oncomine™ Dx Express Test workflow for detecting EGFR exon 20 insertion variants in NSCLC FFPE tissue samples. The study included 4 NSCLC FFPE

samples harboring EGFR exon 20 insertions of 3, 6, 9, and 12 bp in length, and 2 WT NSCLC FFPE clinical samples. The study was designed to assess reproducibility (variability across 3 testing sites, operators,and instruments) and repeatability (within-run variability) using 3 lots of Oncomine™ Dx Express Test reagents, controls, and Genexus™ Dx Integrated Sequencer reagents and consumables. Each test site included 2 operators and 2 instruments. DNA blends containing EGFR exon 20 insertion variants were prepared to represent two variant levels: 1–1.5X LoD and 2–3X LoD, with the 2–3X LoD level applied only to the 6 bp and 12 bp insertion variants. For each DNA sample blend, a total of 72 replicates were generated per LoD level (2 replicates × 2 operators × 2 instruments per operator × 3 lots × 3 sites), resulting in 144 data points per variant.

The call rate, no call rate, positive call rate, and negative call rate were calculated for each variant of interest. Table 31 presents the mean VAFs, along with the corresponding positive and negative call rates and two-sided 95% CIs (Wilson Score method). The positive call rate was 100% for all EGFR exon 20 insertion variants, with no instances of No Calls observed. The negative call rate, evaluated using 2 WT NSCLC samples (WT01 and WT02) and calculated for relevant hotspots, was also 100%, excluding No Calls.

**Table 31 Panel reproducibility for EGFR exon 20 insertion variants**

| Variant | Variant ID | LoD level | Variant AF mean (%) | Total Calls | Positive Calls | Negative Calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive Call rate (95% CI)[1] | Negative Call rate (95% CI)[1] |
| EGFR exon 20 insertion (3 bp) | COSM12377 | 1.17X | 5.35% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| | | 1.88X | 10.28% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| EGFR exon 20 insertion (6 bp) | COSM12380 | 1.43X | 5.76% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| | | 2.39X | 10.49% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| EGFR exon 20 insertion (9 bp) | COSM13428 | 1.11X | 4.73% | 72 | 72 | 0 | 0 | 100.0% (94.9, -100.0%) | 0.0% (0.0%, 5.1%) |
| | | 1.57X | 6.69% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| EGFR exon 20 insertion (12 bp) | COSM26720 | 1.45X | 4.82% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |

**Table 31 Panel reproducibility for EGFR exon 20 insertion variants** *(continued)*

| Variant | Variant ID | LoD level | Variant AF mean (%) | Total Calls | Positive Calls | Negative Calls | No Calls | No Calls excluded | |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | Positive Call rate (95% CI)[1] | Negative Call rate (95% CI)[1] |
| EGFR exon 20 insertion (12 bp) | COSM26720 | 2.27X | 7.55% | 72 | 72 | 0 | 0 | 100.0% (94.9%, 100.0%) | 0.0% (0.0%, 5.1%) |
| WT01 | N/A | N/A | N/A | 1008 | 0 | 1005 | 3 | 0.0% (0.0%, 0.4%) | 100.0% (99.6%, 100.0%) |
| WT02 | N/A | N/A | N/A | 1008 | 0 | 1008 | 0 | 0.0% (0.0%, 0.4%) | 100.0% (99.6%, 100.0%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

Repeatability for the detection of EGFR exon 20 insertion variants was evaluated across within-run, between system, between-operator, between- site, between-lot, and total variability. When including or excluding No Calls from the reproducibility study data, the within-run repeatability was 100% (Table 32).

**Table 32 Repeatability results—EGFR exon 20 insertions**

| Variant | Variant ID | LoD level | Total Calls | Positive Calls | Negative Calls | No Calls | Total pairs | Concordant pairs | Within-run repeatability | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | No Calls excluded (95% CI)[1] | No Calls included (95% CI)[1] |
| EGFR exon 20 insertion (3 bp) | COSM12377 | 1.17X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.88X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| EGFR exon 20 insertion (6 bp) | COSM12380 | 1.43X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.39X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |

**Table 32 Repeatability results—EGFR exon 20 insertions** *(continued)*

| Variant | Variant ID | LoD level | Total Calls | Positive Calls | Negative Calls | No Calls | Total pairs | Concordant pairs | Within-run repeatability | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | No Calls excluded (95% CI)[1] | No Calls included (95% CI)[1] |
| EGFR exon 20 insertion (9 bp) | COSM13428 | 1.11X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.57X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| EGFR exon 20 insertion (12 bp) | COSM26720 | 1.45X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.27X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| WT01 | WT | N/A | 1008 | 0 | 1005 | 3 | 5004 | 5001 | 100.0% (98.3%, 100.0%) | 100.0% (99.3%, 100.0%) |
| WT02 | WT | N/A | 1008 | 0 | 1008 | 0 | 5004 | 5004 | 100.0% (99.3%, 100.0%) | 100.0% (99.3%, 100.0%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

The PPA and NPA across all three test sites were 100% for all variants at both LoD levels tested.

## Assay panel reproducibility study II—Tumor profiling variants

In the panel reproducibility study, starting from extracted DNA/RNA samples, the repeatability and reproducibility of the Oncomine™ Dx Express Test workflow for detecting tumor profiling variants were evaluated across 3 test sites. The study evaluated a total of 67 across 58 samples (with some samples containing 2 variants), including 41 representative DNA variants (15 SNVs, 6 insertions, 5 deletions, 4 indels, and 11 CNVs), 22 RNA fusions, 3 alternate splice variants, and one imbalance variant. All samples were derived from FFPE tissue samples representing 11 cancer types: bladder, breast, cholangiocarcinoma, colorectal, endometrial, glioma, melanoma, NSCLC, pancreatic, prostate, and thyroid. The study was designed to assess reproducibility (variability across 3 test sites, operators, and instruments) and repeatability (within-run variability) using three lots of Oncomine™ Dx Express Test reagents, controls, and Genexus™ Dx Integrated Sequencer reagents and consumables. Each test site included 2 operators and 2 instruments

DNA/RNA blends containing the variants of interest were prepared at a single target level: 1–1.5X LoD. However, for selected tumor profiling variants in NSCLC, including EGFR L858R, EGFR T790M, BRAF V600E, KRAS G12C, EGFR exon 19 deletion, ERBB2 exon 20 insertion, ALK fusion, ROS1 fusion,

RET fusion, and MET splice variant, DNA/RNA blends were prepared to represent two variant levels: 1–1.5X LoD and 2–3X LoD. For each DNA/RNA sample blend, a total of 72 replicates were generated per LoD level (2 replicates × 2 operators × 2 instruments per operator × 3 lots × 3 sites). As noted above, for samples tested only at the 1–1.5X LoD level, reproducibility data from the sample processing reproducibility study were leveraged to support assay performance at higher LoD levels.

The agreement rates for the evaluated tumor profiling variants are presented in Table 33. The PPA ranged from 87.3% (for BRAF V600E in colorectal cancer) to 100%, with no calls excluded from the analysis.

**Table 33 Panel reproducibility for tumor profiling variants**

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 1 | Level 3 | FGFR2 (Amplifi-cation) | CNV | Breast | 1–1.5X | 6.61 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.47% (153287/155664) (98.41%, 98.53%) |
| | Level 3 | ERBB2 p.Y772_A775dup | Insertion | Breast | 1–1.5X | 7.21 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 2 | Level 2 | BRAF p.V600K | SNV | Melanoma | 1–1.5X | 7.26 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.56% (154330/155016) (99.52%, 99.59%) |
| 3 | Level 2 | PIK3CA p.H1047R | SNV | Breast | 1–1.5X | 5.78 | 91.7% (66/72) (83.0%, 96.1%) | 97.06% (66/68) (89.9%, 99.2%) | 99.09% (153598/155016) (98.81%, 98.91%) |
| 4 | Level 2 | ERBB2 p.G776V | SNV | NSCLC | 1–1.5X | 4.1 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.86% (153177/154944) (98.81%, 98.91%) |
| | Level 2 | MET p.? | SNV | NSCLC | 1–1.5X | 4.91 | 90.3% (65/72) (81.3%, 95.2%) | 94.2% (65/68) (86.0%, 97.7%) | |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Imbalance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 5 | Level 2 | PIK3CA p.E545K | SNV | Breast | 1–1.5X | 7.72 | 97.2% (70/72) (90.4%, 99.2%) | 98.6% (70/71) (92.4%, 99.8%) | 94.37% (146905/155664) (94.26%, 94.49%) |
| | Level 2 | ERBB2 (Amplification) | CNV | Breast | 1–1.5X | 6.45 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 6 | Level 3 | TP53 p.P177_C182del | Deletion | Breast | 1–1.5X | 4.15 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 98.41% (153185/155664) (98.34%, 98.47%) |
| | Level 3 | PIK3CA (Amplification) | CNV | Breast | 1–1.5X | 6.46 | 97.2% (70/72) (90.4%, 99.2%) | 97.2% (70/72) (90.4%, 99.2%) | |
| 7 | Level 2 | IDH1 p.R132C | SNV | Cholangiocarcinoma | 1–1.5X | 6.95 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 97.90% (152402/155664) (97.83%, 97.97%) |
| 8 | Level 3 | EGFR p.S768_D770dup | Insertion | Endometrial | 1–1.5X | 4.74 | 98.6% (71/72) (92.5%, 99.8%) | 100.0% (71/71) (94.9%, 100%) | 99.24% (153839/155016) (99.20%, 99.28%) |
| | Level 2 | ERBB2 (Amplification) | CNV | Endometrial | 1–1.5X | 6.38 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 9 | Level 2 | RET p.C618Y | SNV | Thyroid | 1–1.5X | 9 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 100.0% (576/576) (99.34%, 100%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Imbalance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 10 | Level 3 | AR (Amplification) | CNV | Breast | 1–1.5X | 7.13 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 100.0% (648/648) (99.41%, 100%) |
| | Level 3 | FGFR1 (Amplification) | CNV | Breast | 1–1.5X | 6.28 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 11 | Level 3 | MET (Amplification) | CNV | Glioma | 1–1.5X | 7.04 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 100.0% (648/648) (99.41%, 100%) |
| 12 | Level 3 | EGFR (Amplification) | CNV | Glioma | 1–1.5X | 6.58 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 100.0% (648/648) (99.41%, 100%) |
| 13 | Level 3 | ERBB3 (Amplification) | CNV | Melanoma | 1–1.5X | 6.64 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.09% (154253/155664) (99.05%, 99.14%) |
| 14 | Level 3 | KRAS (Amplification) | CNV | NSCLC | 1–1.5X | 6.61 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 98.42% (152560/155016) (98.35%, 98.48%) |
| | Level 3 | EGFR p.E709_T710delinsD | Deletion | NSCLC | 1–1.5X | 5.94 | 98.6% (71/72) (92.5%, 99.8%) | 100.0% (71/71) (94.9%, 100%) | |
| 15 | Level 2 | KRAS p.G12C | SNV | Pancreatic | 1–1.5X | 6.48 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 97.90% (152402/155664) (97.83%, 97.97%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 16 | Level 2 | RET p.D898_E901del | Deletion | Thyroid | 1–1.5X | 6.44 | 94.4% (68/72) (86.6%, 97.8%) | 100.0% (68/68) (94.7%, 100%) | 99.72% (154580/155016) (99.69%, 99.74%) |
| 17 | Level 3 | ERBB2 p.S310F | SNV | Bladder | 1–1.5X | 4.57 | 93.1% (67/72) (84.8%, 97.0%) | 94.4% (67/71) (86.4%, 97.8%) | 98.60% (153477/155664) (98.54%, 98.65%) |
| | Level 3 | FGFR3 (Amplification) | CNV | Bladder | 1–1.5X | 6.35 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 18 | Level 2 | BRAF p.V600E | SNV | Colorectal | 1–1.5X | 5.32 | 86.1% (62/72) (76.3%, 92.3%) | 87.3% (62/71) (77.6%, 93.2%) | 99.65% (154476/155016) (99.62%, 99.68%) |
| 19 | Level 3 | PIK3CA p.E110del | Deletion | Colorectal | 1–1.5X | 6.58 | 95.8% (69/72) (88.5%, 98.6%) | 97.2% (69/71) (90.3%, 99.2%) | 99.55% (154325/155016) (99.52%, 99.59%) |
| 20 | Level 2 | FGFR3 p.S249C | SNV | Bladder | 1–1.5X | 7.21 | 94.4% (68/72) (86.6%, 97.8%) | 94.4% (68/72) (86.6%, 97.8%) | 91.47% (141734/154944) (91.33%, 91.61%) |
| | Level 3 | ERBB2 p.Y772_A775dup | Insertion | Bladder | 1–1.5X | 5.88 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | |
| 21 | Level 3 | PIK3CA p.E110del | Deletion | Bladder | 1–1.5X | 7.9 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 96.52% (149626/155016) (96.43%, 96.61%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Imbalance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 22 | Level 3 | PTPRZ1-MET.P1M2.1 | Fusion | Glioma | 1–1.5X | 12.13 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.15% (70318/70920) (99.08%, 99.22%) |
| 23 | Level 3 | WHSC1L1-NUTM1.W7N2.1 | Fusion | Thyroid | 1–1.5X | 12.71 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.18% (70342/70920) (99.12%, 99.25%) |
| 24 | Level 3 | EGFRvIII .E1E8.Del1 | Alt Splice | Glioma | 1–1.5X | 17.26 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.13% (70300/70920) (99.05%, 99.19%) |
| 25 | Level 3 | FGFR1-TACC1.F17T7 .COSF1362.1 | Fusion | NSCLC | 1–1.5X | 8.33 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.18% (70336/70920) (99.11%, 99.24%) |
| 26 | Level 2 | TPM3-NTRK1.T7N10 .COSF1329 | Fusion | Colorectal | 1–1.5X | 9.4 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.18% (70337/70920) (99.11%, 99.24%) |
| 27 | Level 2 | FGFR2-CREB5.F17C8.1 | Fusion | Cholangiocarcinoma | 1–1.5X | 7.86 | 97.2% (70/72) (90.4%, 99.2%) | 97.2% (70/72) (90.4%, 99.2%) | 99.18% (70336/70920) (99.11%, 99.24%) |
| 28 | Level 3 | ALK (Fusion Imbalance) | Fusion imbalance | NSCLC | 1–1.5X | 6.09 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 28.57% (144/504) (24.80%, 32.67%) |
| 29 | Level 2 | TRIM24-BRAF.T3B10 | Fusion | Glioma | 1–1.5X | 9.07 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.16% (70327/70920) (99.09%, 99.23%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 30 | Level 2 | CCDC6-RET .C1R12.COS F1271.1 | Fusion | Thyroid | 1–1.5X | 11.47 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.18% (70336/7092 0) (99.11%, 99.24%) |
| 31 | Level 2 | FGFR3-TACC3.F17T 11 .COSF1348.1 | Fusion | Bladder | 1–1.5X | 24.32 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.18% (70336/7092 0) (99.11%, 99.24%) |
| 32 | Level 3 | ESR1-CCDC170 .E2C7.1 | Fusion | Breast | 1–1.5X | 22.88 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.28% (70406/7092 0) (99.21%, 99.34%) |
| 33 | Level 2 | SDC4-ROS1.S2R32 .COSF1265 | Fusion | NSCLC | 1–1.5X | 9.93 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.18% (70342/7092 0) (99.12%, 99.25%) |
| 34 | Level 2 | BCR-NTRK2.B1N1 7 | Fusion | Glioma | 1–1.5X | 7.72 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 99.16% (70326/7092 0) (99.09%, 99.23%) |
| 35 | Level 3 | CD74-NRG1.C6N6. 1 | Fusion | NSCLC | 1–1.5X | 8.85 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 99.19% (70345/7092 0) (99.12%, 99.25%) |
| 36 | Level 3 | AR-AR.A3A4.V7 | Alt Splice | Prostate | 1–1.5X | 18.24 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.19% (70344/7092 0) (99.12%, 99.25%) |
| 37 | Level 2 | SQSTM1-NTRK3.S5N1 4 | Fusion | Thyroid | 1–1.5X | 12.88 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.18% (70342/7092 0) (99.12%, 99.25%) |
| 38 | Level 2 | EML4-ALK.E20A20 .COSF409.2 | Fusion | NSCLC | 1–1.5X | 8.39 | 97.2% (70/72) (90.4%, 99.2%) | 97.2% (70/72) (90.4%, 99.2%) | 99.19% (70344/7092 0) (99.12%, 99.25%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 39 | Level 3 | EIF3E-RSPO2.E1R2.COSF1307.1 | Fusion | Colorectal | 1–1.5X | 9 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 99.19% (70344/70920) (99.12%, 99.25%) |
| 40 | Level 3 | PTPRK-RSPO3.P1R2.COSF1311.1 | Fusion | Colorectal | 1–1.5X | 11.17 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.19% (70344/70920) (99.12%, 99.25%) |
| 41 | Level 2 | EGFR p.L858R | SNV | NSCLC | 1.36X | 7.41 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.43% (154125/155016) (99.39%, 99.46%) |
| | | | | | 2.11X | 11.5 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.53% (154282/155016) (99.49%, 99.56%) |
| | Level 2 | EGFR p.T790M | SNV | NSCLC | 1.21X | 5.76 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.43% (154125/155016) (99.39%, 99.46%) |
| | | | | | 2.40X | 11.39 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.53% (154282/155016) (99.49%, 99.56%) |
| 42 | Level 2 | EGFR p.L747_A750delinsP (9bp) | Deletion | NSCLC | 1.11X | 5.9 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.68% (154594/155088) (99.65%, 99.71%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 42 | Level 2 | EGFR p.L747_A750delinsP (9bp) | Deletion | NSCLC | 1.57X | 10.6 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.70% (154623/155088) (99.67%, 99.73%) |
| | Level 2 | ERBB2 p.Y772_A775dup (12 bp) | Insertion | NSCLC | 1.00X | 5.22 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.68% (154594/155088) (99.65%, 99.71%) |
| | | | | | 1.41X | 7.4 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.70% (154623/155088) (99.67%, 99.73%) |
| 43 | Level 2 | EGFR p.L747_P753delinsS (18 bp) | Deletion | NSCLC | 1.37X | 5.68 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.59% (154450/155088) (99.56%, 99.62%) |
| | | | | | 2.60X | 10.83 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.73% (154674/155088) (99.71%, 99.76%) |
| 44 | Level 2 | EGFR p.L858R | SNV | NSCLC | 1.27X | 6.69 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.47% (154193/155016) (99.43%, 99.50%) |
| | | | | | 1.95X | 10.64 | 98.6% (71/72) (92.5%, 99.8%) | 100.0% (71/71) (94.9%, 100%) | 98.44% (152591/155016) (98.37%, 98.50%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Imbalance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 44 | Level 2 | EGFR p.T790M | SNV | NSCLC | 1.47X | 6.96 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.47% (154193/155016) (99.43%, 99.50%) |
| | | | | | 2.15X | 10.17 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.44% (152591/155016) (98.37%, 98.50%) |
| 45 | Level 2 | KRAS p.G12C | SNV | NSCLC | 1.51X | 5.95 | 98.6% (71/72) (92.5%, 99.8%) | 100.0% (71/71) (94.9%, 100%) | 99.71% (154643/155088) (99.69%, 99.74%) |
| | | | | | 2.76X | 9.93 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.58% (154432/155088) (99.54%, 99.61%) |
| 46 | Level 2 | BRAF p.V600E | SNV | NSCLC | 1.29X | 5.65 | 97.2% (70/72) (90.4%, 99.2%) | 98.6% (70/71) (92.4%, 99.8%) | 99.62% (154423/155016) (99.59%, 99.65%) |
| | | | | | 1.84X | 8.03 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.39% (154065/155016) (99.35%, 99.42%) |
| | Level 2 | EGFR p.E746_A750del (15 bp) | Deletion | NSCLC | 1.50X | 4.63 | 98.6% (71/72) (92.5%, 99.8%) | 98.6% (71/72) (92.5%, 99.8%) | 99.62% (154423/155016) (99.59%, 99.65%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 46 | Level 2 | EGFR p.E746_A750del (15 bp) | Deletion | NSCLC | 2.26X | 6.96 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.39% (154065/155016) (99.35%, 99.42%) |
| 47 | Level 2 | KRAS p.G12C | SNV | NSCLC | 1.47X | 5.61 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.47% (154192/155016) (99.43%, 99.50%) |
| | | | | | 2.70X | 10.28 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.64% (154456/155016) (99.61%, 99.67%) |
| | Level 2 | EGFR p.L747_A750delinsP (9 bp) | Deletion | NSCLC | 1.41X | 5.9 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.47% (154192/155016) (99.43%, 99.50%) |
| | | | | | 2.59X | 10.6 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.64% (154456/155016) (99.61%, 99.67%) |
| 48 | Level 2 | ERBB2 p.G778_P780dup (9 bp) | Insertion | NSCLC | 1.31X | 5.14 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.28% (152417/155088) (98.21%, 98.34%) |
| | | | | | 2.53X | 8.25 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.20% (152302/155088) (98.14%, 98.27%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Imbalance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 49 | Level 2 | BRAF p.V600E | SNV | NSCLC | 1.31X | 5.72 | 98.6% (71/72) (92.5%, 99.8%) | 100.0% (71/71) (94.9%, 100%) | 99.56% (154403/155088) (99.52%, 99.59%) |
| | | | | | 2.59X | 11.35 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.74% (154682/155088) (99.71%, 99.76%) |
| 50 | Level 2 | ERBB2 p.G776 delinsVC (3 bp) | Insertion | NSCLC | 1.38X | 4.12 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.33% (154053/155088) (99.29%, 99.37%) |
| | | | | | 2.66X | 7.92 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 99.26% (153941/155088) (99.22%, 99.30%) |
| 51 | Level 2 | EML4-ALK .E13A20 .COSF408.2 | Fusion | NSCLC | 1.21X | 9.38 | 97.2% (70/72) (90.4%, 99.2%) | 97.2% (70/72) (90.4%, 99.2%) | 98.72% (70459/71370) (98.64%, 98.80%) |
| | | | | | 2.26X | 17.61 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.68% (70430/71370) (98.60%, 98.76%) |
| 52 | Level 2 | KIF5B-RET .K15R12 .COSF1232 | Fusion | NSCLC | 1.22X | 10.44 | 95.8% (69/72) (88.5%, 98.6%) | 95.8% (69/72) (88.5%, 98.6%) | 98.76% (70513/71397) (98.68%, 98.84%) |
| | | | | | 2.09X | 17.93 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.77% (70513/71388) (98.69%, 98.85%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 53 | Level 2 | MET-MET.M13M15.1 | Splice variant | NSCLC | 1.39X | 18.94 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.69% (70474/71406) (98.61%, 98.78%) |
| | | | | | 2.69X | 36.69 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.65% (70444/71406) (98.57%, 98.73%) |
| 54 | Level 2 | CCDC6-RET.C1R12.COSF1271.1 | Fusion | NSCLC | 1.39X | 14.06 | 97.2% (70/72) (90.4%, 99.2%) | 97.2% (70/72) (90.4%, 99.2%) | 98.78% (70496/71370 (98.69%, 98.85%) |
| | | | | | 2.07X | 20.99 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.74% (70469/71370) (98.65%, 98.82%) |
| 55 | Level 2 | SDC4-ROS1.S2R32.COSF1265 | Fusion | NSCLC | 1.80X | 13.94 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.69% (70473/71406) (98.61%, 98.77%) |
| | | | | | 3.09X | 24 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.67% (70459/71406) (98.59%, 98.76%) |
| 56 | Level 2 | EML4-ALK.E20A20.COSF409.2 | Fusion | NSCLC | 1.83X | 9.13 | 94.4% (68/72) (86.6%, 97.8%) | 94.4% (68/72) (86.6%, 97.8%) | 98.73% (70499/71406) (98.65%, 98.81%) |
| | | | | | 2.29X | 11.46 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.72% (70491/71406) (98.63%, 98.80%) |
| 57 | Level 2 | EZR-ROS1.E10R34.COSF1267 | Fusion | NSCLC | 1.61X | 14.39 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.83% (70572/71406) (98.75%, 98.91%) |

**Table 33 Panel reproducibility for tumor profiling variants** *(continued)*

| Variant No. | Reporting level[1] | Variant | Variant type | Tissue type | LoD level | Obs. Mean AF%, CN, Mol. counts, or Im-balance score | PPA (n/N) (95% CI)[2] | | NPA (n/N) (95% CI)[2] No Calls included, per sample |
|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | No Calls included | No Calls excluded | |
| 57 | Level 2 | EZR-ROS1 .E10R34 .COSF1267 | Fusion | NSCLC | 2.40X | 21.44 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.80% (70546/71406) (98.71%, 98.87%) |
| 58 | Level 2 | MET-MET .M13M15.1 | Splice variant | NSCLC | 1.41X | 19.18 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.72% (70498/71415) (98.63%, 98.80%) |
| | | | | | 2.65X | 36.1 | 100.0% (72/72) (94.9%, 100%) | 100.0% (72/72) (94.9%, 100%) | 98.69% (70489/71424) (98.60%, 98.77%) |

[1] Level 2 = Cancer Mutations with Evidence of Clinical Significance, Level 3 = Cancer Mutations with Potential Clinical Significance.
[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

Positive and negative call rates were evaluated for samples containing targeted variants across all variant types detected by Oncomine™ Dx Express Test. Results are summarized by variant type in Table 34 and Table 35, respectively. The positive call rate was >97% for all variant types tested near or above the LoD, both including and excluding no calls. The negative call rate was >97% for all variant types, both including and excluding no calls, except for deletions and fusion imbalance at 1–1.5X LoD. For these exceptions, the negative call rates including no calls were 95.27% for deletions and 49.10% for fusion imbalance.

**Table 34 Positive call rate for tumor profiling variants by variant class**

| Variant type | LoD level | Total calls | Positive calls | Negative calls | No Calls | Positive call rate (95% CI)[1]– No Calls included | Positive call rate (95% CI)[1] – No Calls excluded |
|---|---|---|---|---|---|---|---|
| SNV | 1–1.5X | 1368 | 1329 | 26 | 13 | 97.15% (96.13%, 97.91%) | 98.08% (97.20%, 98.69%) |
| SNV | 2–3X | 576 | 575 | 0 | 1 | 99.83% (99.02%, 99.97%) | 100% (99.34%, 100%) |
| Insertion | 1–1.5X | 504 | 503 | 0 | 1 | 99.80% (98.88%, 99.96%) | 100% (99.24%, 100%) |
| Insertion | 2–3X | 144 | 144 | 0 | 0 | 100% (97.40%, 100%) | 100% (97.40%, 100%) |

**Table 34 Positive call rate for tumor profiling variants by variant class** *(continued)*

| Variant type | LoD level | Total calls | Positive calls | Negative calls | No Calls | Positive call rate (95% CI)[1]– No Calls included | Positive call rate (95% CI)[1] – No Calls excluded |
|---|---|---|---|---|---|---|---|
| Deletion | 1–1.5X | 576 | 561 | 9 | 6 | 97.40% (95.75%, 98.42%) | 98.42% (97.03%, 99.17%) |
| Deletion | 2–3X | 216 | 216 | 0 | 0 | 100% (98.25%, 100%) | 100% (98.25%, 100%) |
| CNV | 1–1.5X | 792 | 789 | 3 | 0 | 99.62% (98.89%, 99.87%) | 99.62% (98.89%, 99.87%) |
| Fusion | 1–1.5X | 1584 | 1555 | 29 | 0 | 98.17% (97.38%, 98.72%) | 98.17% (97.38%, 98.72%) |
| Fusion | 2–3X | 432 | 432 | 0 | 0 | 100% (99.12%, 100%) | 100% (99.12%, 100%) |
| Fusion imbalance | 1–1.5X | 72 | 72 | 0 | 0 | 100% (94.93%, 100%) | 100% (94.93%, 100%) |
| Splice variant | 1–1.5X | 288 | 287 | 1 | 0 | 99.65% (98.06%, 99.94%) | 99.65% (98.06%, 99.94%) |
| Splice variant | 2–3X | 144 | 144 | 0 | 0 | 100% (97.40%, 100%) | 100% (97.40%, 100%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

**Table 35 Negative call reproducibility by variant class**

| Variant type | LoD level | Total calls | Positive calls | Negative calls | No Calls | Negative call rate (95% CI)[1] – No Calls included | Negative call rate (95% CI)[1] – No Calls excluded |
|---|---|---|---|---|---|---|---|
| SNV | 1–1.5X | 2964168 | 1319 | 2945554 | 17295 | 99.37% (99.36%, 99.38%) | 99.96% (99.95%, 99.96%) |
| SNV | 2–3X | 1164456 | 455 | 1160937 | 3064 | 99.70% (99.69%, 99.71%) | 99.96% (99.95%, 99.96%) |
| Insertion | 1–1.5X | 593064 | 1 | 589125 | 3938 | 99.34% (99.31%, 99.36%) | 100.00% (100.0%, 100%) |
| Insertion | 2–3X | 233424 | 0 | 232810 | 614 | 99.74% (99.72%, 99.76%) | 100.00% (100.0%, 100%) |
| Deletion | 1–1.5X | 783648 | 0 | 746619 | 37029 | 95.27% (95.23%, 95.32%) | 100.00% (100.0%, 100%) |
| Deletion | 2–3X | 307872 | 0 | 300911 | 6961 | 97.74% (97.69%, 97.79%) | 100.00% (100.0%, 100%) |

**Table 35 Negative call reproducibility by variant class** *(continued)*

| Variant type | LoD level | Total calls | Positive calls | Negative calls | No Calls | Negative call rate (95% CI)[1] – No Calls included | Negative call rate (95% CI)[1] – No Calls excluded |
|---|---|---|---|---|---|---|---|
| CNV | 1–1.5X | 6408 | 0 | 6408 | 0 | 100% (99.94%,100%) | 100% (99.94%,100%) |
| Fusion | 1–1.5X | 1825056 | 50 | 1821714 | 3292 | 99.82% (99.81%, 99.82%) | 100.00% (100.0%, 100%) |
| Fusion | 2–3X | 564984 | 88 | 561681 | 3215 | 99.42% (99.40%, 99.43%) | 99.98% (99.98%, 99.99%) |
| Fusion imbalance | 1–1.5X | 2880 | 0 | 1414 | 1466 | 49.10% (47.27%, 50.92%) | 100% (99.73%,100%) |
| Splice variant | 1–1.5X | 7200 | 101 | 7099 | 0 | 98.60% (98.30%, 98.84%) | 98.60% (98.30%, 98.84%) |
| Splice variant | 2–3X | 2160 | 0 | 2160 | 0 | 100% (99.82%, 100%) | 100% (99.82%, 100%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

A panel-wide precision analysis based on all detected variants in the tested samples was conducted. PPA was evaluated by variant type and stratified by VAF thresholds (≥0%, ≥5%, ≥8%, ≥10%, ≥15%, and ≥20%). The positive call rate was 100% for all variant types at VAF levels ≥5%, ≥8%, ≥10%, ≥15%, and ≥20%, excluding no calls. At a VAF level ≥0%, the positive call rate was 0.1%.

A variance component analysis was conducted for tumor profiling variants tested near or above the LoD, using the appropriate quantitative metric for each variant type (i.e., VAF for SNVs, insertions, and deletions, copy number for CNVs, and molecular counts for RNA variants). The results showed that the coefficient of variation (CV) was below 10% between operators/instruments for all variants, except for one ALK fusion, one ROS1 fusion, one MET splice variant, which had CVs of 20.95%, 14.59%, and 11.65%, respectively. The CV for between-run variability was below 20% for all variants except for one ALK fusion and one ROS1 fusion, which had CVs of 20.95% and 17.12%, respectively. Percent CVs for reagent lot ranged from 0.0% to 34.15% across all variants. Percent CVs for within-run and within-lab variability varied by variant type, with the highest CVs observed for RNA variants.

Within-run repeatability for detecting selected tumor profiling variants in NSCLC, including EGFR L858R, EGFR T790M, BRAF V600E, KRAS G12C, EGFR exon 19 deletion, ERBB2 exon 20 insertion, ALK fusion, ROS1 fusion, RET fusion, and MET splice variant was evaluated. For DNA variants (Table 36), within-run repeatability was 100% when excluding No Calls, except for one instance of BRAF V600E (COSM476) at 1.29X LoD, which showed a NPA of 97.1% (34/36). For RNA variants (Table 37), within-run repeatability was 100% for both ROS1 fusion and the MET splice variant, regardless of whether no calls were included or excluded. ALK fusion demonstrated within-run repeatability of 94.4% (34/36) at 1.21X LoD and 88.9% (32/36) at 1.83X LoD. RET fusion showed repeatability of 91.7% (33/36) at 1.22X LoD and 94.4% (34/36) at 1.39X LoD.

**Table 36 Within-run repeatability for selected Level 2 tumor profiling DNA variants**

| Variant | Variant ID | LoD level | Total calls | Positive Calls | Negative calls | No Calls | Total pairs | Concordant pairs | Within-run repeatability (95% CI)[1] | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | No Calls included | No Calls excluded |
| EGFRL858R | COSM6224 | 1.27x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.36x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.95x | 72 | 71 | 0 | 1 | 36 | 35 | 97.2% (85.5%, 99.5%) | 100.0% (90.0%, 100.0%) |
| | | 2.11x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| EGFR T790M | COSM6240 | 1.21x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.47x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.15x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.40x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| KRAS G12C | COSM516 | 1.47x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.51x | 72 | 71 | 0 | 1 | 36 | 35 | 97.2% (85.5%, 99.5%) | 100.0% (90.0%, 100.0%) |
| | | 2.70x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.76x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |

**Table 36 Within-run repeatability for selected Level 2 tumor profiling DNA variants** *(continued)*

| Variant | Variant ID | LoD level | Total calls | Positive Calls | Negative calls | No Calls | Total pairs | Concordant pairs | Within-run repeatability (95% CI)[1] | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | No Calls included | No Calls excluded |
| BRAF V600E | COSM476 | 1.29x | 72 | 70 | 1 | 1 | 36 | 34 | 94.4% (81.3%, 99.3%) | 97.1% (85.1%,99.9%) |
| | | 1.31x | 72 | 71 | 0 | 1 | 36 | 35 | 97.2% (85.5%, 99.5%) | 100.0% (90.0%, 100.0%) |
| | | 1.84x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.59x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| ERRB2 exon 20 insertion (3 bp) | COSM12553 | 1.38x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.66x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| ERRB2 exon 20 insertion (9 bp) | COSM12556 | 1.31x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.53x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| ERRB2 exon 20 insertion (12 bp) | COSM20959 | 1.00x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 1.41x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| EGFR exon 19 deletion (9 bp) | COSM12382 | 1.41x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.59x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |

**Table 36 Within-run repeatability for selected Level 2 tumor profiling DNA variants** *(continued)*

| Variant | Variant ID | LoD level | Total calls | Positive Calls | Negative calls | No Calls | Total pairs | Concordant pairs | Within-run repeatability (95% CI)[1] | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | | | | | | | | No Calls included | No Calls excluded |
| EGFR exon 19 deletion (15 bp) | COSM6223 | 1.50x | 72 | 71 | 1 | 0 | 36 | 35 | 97.2% (85.5%, 99.5%) | 100.0% (90.0%, 100.0%) |
| | | 2.26x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| EGFR exon 19 deletion (18 bp) | COSM12370 | 1.37x | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| | | 2.60% | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) | 100.0% (90.3%, 100.0%) |
| WT01 | N/A | N/A | 1008 | 0 | 1005 | 3 | 504 | 501 | 99.40% (97.95%, 99.82%) | 100.0% (99.3%, 100.0%) |
| WT02 | N/A | N/A | 1008 | 0 | 1008 | 0 | 504 | 504 | 100.0% (99.3%, 100.0%) | 100.0% (99.3%, 100.0%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

**Table 37 Within-run repeatability for selected Level 2 tumor profiling RNA variants**

| Variant | Variant ID | LoD level | Total calls | Positive calls | Negative calls | No Calls | Total pairs | Concordant pairs | Within-run Repeatability (95% CI)[1] |
|---|---|---|---|---|---|---|---|---|---|
| ALK fusion | EML4-ALK.E13A20.COSF408.2 | 1.21X | 72 | 70 | 2 | 0 | 36 | 34 | 94.4% (81.3%, 99.3%) |
| | | 2.26X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| ALK fusion | EML4-ALK.E20A20.COSF409.2 | 1.83X | 72 | 68 | 4 | 0 | 36 | 32 | 88.9% (73.9%, 96.9%) |
| | | 2.29X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| RET fusion | KIF5B-RET.K15R12.COSF1232.1 | 1.22X | 72 | 69 | 3 | 0 | 36 | 33 | 91.7% (77.5%, 96.9%) |

**Table 37 Within-run repeatability for selected Level 2 tumor profiling RNA variants** *(continued)*

| Variant | Variant ID | LoD level | Total calls | Positive calls | Negative calls | No Calls | Total pairs | Concordant pairs | Within-run Repeatability (95% CI)[1] |
|---|---|---|---|---|---|---|---|---|---|
| RET fusion | KIF5B-RET.K15R12.COSF1232.1 | 2.09X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0%(90.3%, 100.0%) |
| RET fusion | CCDC6-RET.C1R12.COSF1271.1 | 1.39X | 72 | 70 | 2 | 0 | 36 | 34 | 94.4% (81.3%, 99.3%) |
| | | 2.07X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| ROS1 fusion | SDC4-ROS1.S2R32.COSF1265 | 1.80X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| | | 3.09X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| ROS1 fusion | EZR-ROS1.E10R34.COSF1267 | 1.61X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| | | 2.40X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| MET splice variant | MET-MET.M13M1 5.1 | 1.39X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| | | 2.69X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| MET splice variant | MET-MET.M13M1 5.1 | 1.41X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| | | 2.65X | 72 | 72 | 0 | 0 | 36 | 36 | 100.0% (90.3%, 100.0%) |
| WT01 | NA | NA | 504 | 0 | 504 | 0 | 252 | 252 | 100.0% (98.5%, 100.0%) |
| WT02 | NA | NA | 504 | 0 | 504 | 0 | 252 | 252 | 100.0% (98.5%, 100.0%) |

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

## Stability of assay intermediates study

The stability of intermediate products in the Oncomine™ Dx Express Test workflow was evaluated to determine whether partially completed reactions can be held under defined storage conditions without affecting assay performance. The study incorporated all predefined hold times outlined in the Oncomine™ Dx Express Test User Guide and assessed whether the workflow is robust to pauses at specific steps.

The hold time evaluations were conducted using 12 FFPE clinical samples from 10 tumor types containing representative SNVs, insertion, deletion, CNVs, and fusion variants targeted by the Oncomine™ Dx Express Test panel. Each sample was tested under 3 control conditions (standard operating procedure (SOP)) and 5 test conditions, with each condition performed in duplicate (3 × 2 = 6 extractions and 5 × 2 = 10 extractions per sample, respectively), as outlined in Table 38. With 12 samples included in the study, a total of 192 extractions were performed (16 × 12 = 192). For each replicate, two 5 µm sections were prepared from FFPE tissue blocks. Sections were randomly assigned to sample replicates to minimize potential variability due to tumor content heterogeneity within the blocks. Nucleic acid extraction and quantitation were performed using the Genexus™ Dx Purification System with the Genexus™ Dx FFPE DNA and RNA Purification and the Genexus™ Dx Nucleic Acid Quantitation. All testing was performed on the Genexus™ Dx System using the integrated purification to sequencing configuration, following the standard sample to report workflow.

**Table 38 Designated hold time test conditions**

| Step | Control (SOP) | Test condition |
|---|---|---|
| Lysate stability | Immediate use | • 24 hours at 4°C<br>• 72 hours at 4°C<br>• 168 hours at –20°C |
| Deck set up | Immediate start | 15 minutes delayed start |
| On deck nucleic acid stability | Immediate unloading | 1 hour delayed unloading |

All variants of interest were successfully detected in all samples and replicates across test conditions, resulting in a 100% passing rate when excluding no calls. For each sample, consensus calls from the control runs were used as the comparator. Among samples with SNVs, insertion, deletion, and CNVs variants, 100% concordance was observed under all test conditions compared to the control. Similarly, for samples with RNA variants, 100% concordance was observed for all test conditions, except for lysate stability when stored for 24 hours at 4°C and for 7 days at -20°C, where both negative concordance and overall concordance were 99.99%.

## FFPE tissue block, curl, and slide stability study

The stability of FFPE tissue blocks, tissue sections (curls), and slide-mounted sections was evaluated using clinical samples containing DNA and RNA variants of interest, including CDx variants (EGFR exon 20 insertions) and tumor profiling variants representative SNVs, insertions, deletions, CNVs, and fusion variants, when processed with the Oncomine™ Dx Express Test workflow. For block stability, FFPE tissue blocks were stored at 15°C to 30°C for the duration of the study. At each time point, 5 µm sections were cut and processed immediately. For slide stability, 5 µm sections cut from FFPE blocks were mounted on positively charged slides and stored at 15°C to 30°C for the duration of the study. For curl stability, 5 µm sections were cut and stored in 1.5 mL microcentrifuge tubes at 2°C to 8°C for the

duration of the study. Stability testing was performed at baseline (T0) and 6 months + 2 weeks (T1) time points.

For each FFPE resection sample across the block, slide, and curl formats, duplicate extractions were performed and processed in duplicate runs. Due to limited tissue availability in CNB and FNA samples, at least 1 replicate per sample was extracted and tested. Extractions were performed using 2 × 5 µm sections per resection sample, 9 × 5 µm sections per CNB sample, and 7 × 5 µm sections per FNA cell block sample. All samples were processed using the Oncomine™ Dx Express Test workflow on the Genexus™ Dx System with integrated purification and sequencing configuration. DNA and RNA quantitation was performed to confirm that all samples met the minimum input requirement of 0.5 ng/µL.

All sample replicates achieved a 100% pass rate, with DNA and RNA concentrations meeting the Oncomine™ Dx Express Test minimum input requirement of 0.5 ng/µL at both baseline (T0) and 6-month + 2 weeks (T1) time points. For FFPE resection samples, all DNA and RNA variants of interest demonstrated <30% drift in AF and <40% drift in log fusion read counts relative to baseline measurements at both time points. For CNB and FNA cell block samples, concordance analysis confirmed 100% detection of all DNA and RNA variants of interest at both time points. These results support the stability of FFPE blocks, curls, and slides for up to 6 months under the specified storage conditions.

## Extracted nucleic acid stability study

The storage stability of DNA and RNA extracted from FFPE tissue samples was evaluated using samples containing variants targeted by the Oncomine™ Dx Express Test panel. This included CDx variants (EGFR exon 20 insertions) and representative tumor profiling variants such as SNVs, insertions, deletions, CNVs, fusions, and RNA splice variants. DNA and RNA were extracted from a variety of tissue types, including bile duct, bladder, brain, breast, colon, endometrium, lung, prostate, skin, and thyroid. In total, 12 DNA samples from seven different tissue types and 8 RNA samples from 5 different tissue types were analyzed. Nucleic acids were extracted using the Ion Torrent™ Genexus™ FFPE DNA and RNA Purification Kit on the Genexus™ Dx Purification System. Sample blends were prepared by mixing variant-positive FFPE DNA or RNA with variant-negative material to achieve VAFs of 1–2X limit of detection (LoD) for DNA variants and 1–3X LoD for RNA variants. Extracted nucleic acids were aliquoted into LoBind™ tubes for short- and long-term storage and into archive plates for short-term storage.

DNA samples in tubes were stored at –30°C to –10°C, while RNA samples were stored at –70°C to –90°C. Archive plates containing both DNA and RNA were stored at –30°C to –10°C. Samples stored in tube format were tested at baseline (T0) and at 6 months + 1 week (T1) time points. The extra one-week buffer at each time point was included to accommodate processing time within the Oncomine™ Dx Express Test workflow. Samples stored in archive plates were tested at baseline (T0), 4 weeks (T1), 3 months (T2), and 6 months (T3) time points. To evaluate freeze/thaw stability, aliquots from tubes were subjected to 3 freeze/thaw cycles prior to testing at each time point.

The impact of storage conditions and freeze/thaw cycles on sequencing performance was assessed by measuring drift in VAF for DNA variants and in molecular counts for RNA variants. For samples stored in archive plates, regression analysis showed that drift remained within the allowable limits of 30% for DNA and 40% for RNA. Similarly, for samples stored in tube format, the percent relative change in VAF and molecular counts remained within acceptable limits following storage and freeze/thaw cycles. All variants of interest were successfully detected in all samples stored in both tube and plate formats across all tested time points, demonstrating a 100% detection rate. These results support the stability

of DNA and RNA extracted from FFPE tissue samples for up to 6 months when stored in LoBind™ tubes and for at least 5 months when stored in plate formats, at the recommended temperature.

## Reagent stability study

- **Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents shelf life stability:**

  The real-time stability of the Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents and consumables was evaluated using DNA and RNA samples extracted from FFPE clinical tissue samples, as well as manufactured reagent controls. The DNA and RNA samples included all variant types detected by the Oncomine™ Dx Express Test panel, including CDx variants (EGFR exon 20 insertions) and representative tumor profiling variants such as SNVs, insertions, deletions, CNVs, and fusions. Three Investigational Use Only (IUO) lots of Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents were assessed for stability at 3 time points: baseline (T0), 4 months (T1), and 5 months (T2). The reagents were stored at the temperatures specified on the product labeling throughout the study. At each time point, all DNA and RNA variants of interest were successfully detected with 100% PPA, and all sequencing runs met the predefined QC acceptance criteria. These results demonstrate that the Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents remain stable for a minimum of 4 months when stored as recommended.

- **Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents in-use stability:**

  The in-use stability of the Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents and consumables was evaluated using clinical FFPE samples containing CDx variants (EGFR exon 20 insertions) and representative tumor profiling variants such as SNVs, insertions, deletions, CNVs, fusions, and splice variants. A total of 12 FFPE DNA and 12 FFPE RNA clinical samples or blends, representing 10 different cancer tissue types, were used to assess performance under various test conditions. One IUO lot of Genexus™ Dx Integrated Sequencer reagents and consumables and 1 IUO lot of Oncomine™ Dx Express Test reagents were used to assess stability across 5 test conditions: 24 hours on deck, 14 days on instrument, and 1, 2, and 3 freeze/thaw cycles. Each test condition was compared to baseline (T0). For the 24 hour on deck and freeze/thaw conditions, all DNA and RNA samples across the 10 tissue types were tested.

  At the 24 hour on deck time point, the PPA and NPA were 100%, excluding no calls, for DNA variants including SNVs, indels, and CNVs. For RNA variants, the PPA was 100% and the NPA was 99.99%, excluding no calls. Following 3 freeze/thaw cycles, performance remained consistent with 100% PPA and 100% NPA for DNA variants, and 100% PPA and 99.99% NPA for RNA variants. These data confirm the in-use stability of Oncomine™ Dx Express Test reagents under the tested conditions.

  Overall, the results demonstrate that the Genexus™ Dx GX5™ Chip, Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2, Genexus™ Dx Bottle 3, Genexus™ Dx Cartridge, and Genexus™ Dx Barcodes 1–32 HD remain stable for up to 13 days after initialization of the Genexus™ Dx Integrated Sequencer. The Oncomine™ Dx Express Test panel, Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 3-GX5™, Genexus™ Dx Strip 4, Genexus™ Dx Barcodes 1–32 HD, and Genexus™ Dx GX5™ Chip and Genexus™ Coupler are stable for at least 23 hours at room temperature when placed on the instrument deck. Furthermore, the Oncomine™ Dx Express Test panel, FFPE DNA and RNA controls, Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 3-GX5™, Genexus™ Dx Strip 4, and Genexus™ Dx Cartridge remained stable after up to 2 freeze/thaw cycles.

- **Oncomine™ Dx Express Test and Genexus™ Dx purification and sequencing reagents transport stability:**

  The transport stability of Oncomine™ Dx Express Test and Genexus™ Dx purification and sequencing reagents was evaluated to verify that the shipping configurations developed for the kit components maintain reagent integrity under a range of environmental and physical conditions. The study included 3 lots of Oncomine™ Dx Express Test sequencing and purification reagents and 1 lot of process qualification reagents.

  - **Simulated Transport Conditions**

    Simulated transport testing was performed across 6 shipping conditions to represent typical and extreme scenarios. Test 1 involved a room temperature shipment of the minimum orderable quantity. Test 2 evaluated a room temperature shipment of the maximum orderable quantity. Test 3 simulated a summer shipment using gel ice (–10°C to 11°C) with the maximum orderable quantity, while Test 4 replicated similar conditions for a winter shipment. Test 5 simulated a dry ice shipment (–80°C to –15°C) under summer conditions. Test 6 evaluated dry ice shipment of $CO_2$-sensitive materials under summer conditions, also with the maximum orderable quantity. All test conditions successfully maintained the appropriate temperature ranges, except for Test 6. In this case, a temperature excursion occurred during the simulation of dry ice shipment for $CO_2$-sensitive materials. Specifically, the package containing the Genexus™ Dx Cartridge (A51158) reached 0°C, which is outside the required shipping range of –80°C to –15°C. Despite this deviation, no adverse impact on reagent function was observed.

  - **Package and Label Integrity**

    Across all simulated shipping conditions, the packaging maintained its physical integrity. No significant damage was observed to the pack-outs or primary packaging of Oncomine™ Dx Express Test components. In addition, all primary package labels remained intact, with no signs of smearing or peeling, demonstrating the robustness of the labeling and packaging materials.

  - **Functional Testing Results**

    Functional testing was conducted using the QC release method with Oncomine™ Dx Express Test nucleic acid controls rather than clinical or contrived samples. Following transport simulations, all reagents met the predefined QC acceptance criteria. This includes the Genexus™ Dx Cartridge (A51158) that experienced the temperature excursion during Test 6, as well as all lots of purification and quantification kits. These results confirm that the reagents maintained their functional integrity under all tested shipping scenarios.

    The reagents maintained both physical and functional integrity following simulated environmental and physical challenges, supporting their stability under typical distribution conditions from the manufacturing site to the end user. Additionally, data from the Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer reagents shelf life stability and nucleic acid purification stability studies, which used reagents packaged and shipped under real-world domestic shipping conditions (air and ground) during winter according to established materials handling SOPs and were evaluated through functional testing with clinical samples, were leveraged to support transport stability for Oncomine™ Dx Express Test and Genexus™ Dx purification and sequencing reagents. Overall, the results demonstrate that the shipping configurations developed for Oncomine™ Dx Express Test and Genexus™ Dx purification and sequencing reagents are sufficient to protect product quality during transportation.

## Reagent lot interchangeability—Oncomine™ Dx Express Test and Genexus™ Dx sequencing reagents

Kit lot interchangeability was evaluated through two separate studies: one assessing Oncomine™ Dx Express Test and Genexus™ Dx sequencing reagents, and the other focusing on Genexus™ Dx purification reagents.

Three lots of Oncomine™ Dx Express Test and Genexus™ Dx sequencing reagents and components were tested using a completely randomized split-plot design, resulting in 27 unique kit lot combinations. Each combination was evaluated using 12 FFPE DNA samples/blends and 10 FFPE RNA samples/blends extracted from variant-positive FFPE clinical samples, along with 2 FFPE wild-type RNA samples. The tested samples represented all variant types detected by the Oncomine™ Dx Express Test assay, including CDx variants (EGFR exon 20 insertions) and representative tumor profiling variants such as SNVs, insertions, deletions, CNVs, and fusions. A total of 648 DNA and 648 RNA libraries were planned. Of these, 634 passed QC on the first attempt, yielding a first-time pass rate of 97.8%. Among samples with reportable results, the positive concordance was 100% for SNVs, CNVs, and RNA variants, and 98.15% for insertions and deletions.

Additionally, 3 lots of Genexus™ Dx purification reagents and consumables were evaluated using a completely randomized split-plot design, resulting in 7 kit lot combinations. Each combination was tested using 12 FFPE tissue samples spanning 9 tissue types and covering all variant types detected by Oncomine™ Dx Express Test, including CDx variants (EGFR exon 20 insertions) and representative tumor profiling variants such as SNVs, Indels, CNVs, and fusions. All samples containing target variants met the minimum DNA and RNA input concentrations (≥0.5 ng/µL) required for Oncomine™ Dx Express Test testing, resulting in 168 qualifying extractions. All DNA and RNA variants of interest were consistently detected across every reagent lot combination, with a 100% positive call rate across all variant and tissue types.

These data support the kit lot interchangeability of Oncomine™ Dx Express Test reagents and the Genexus™ Dx purification and sequencing reagents, demonstrating consistent performance across multiple lots.

## Contrived sample functional characterization study

- **Core Needle Biopsy Specimen Type**

  The performance equivalency of the Oncomine™ Dx Express Test was evaluated by comparing results obtained from donor FFPE tissue resection blocks and contrived CNB or stereotactic biopsies (STBs) samples derived from donor FFPE tissue resection blocks. Derivative CNB and STB samples were created either by cutting the resection blocks into smaller rectangular pieces (Method 1) or by using a punch biopsy technique (Method 2). The study included 8 donor FFPE tissue resection blocks with lung tissue, 2 with breast tissue, and one with brain tissue. For each tissue type, 11 derivative samples were generated using both Method 1, and Method 2. A total of 8 DNA variants were evaluated, including 5 SNVs, 2 insertions (one of which was an EGFR exon 20 insertion [9 bp]), and one deletion. Seven variants were tested using donor lung and breast tissue samples, covering tumor content levels ranging from approximately 20% to over 50%. One variant was evaluated in a brain tissue sample with high tumor content (65% and 95%). The resulting derivative samples exhibited a tumor content range from 5% to 95%.

  DNA and RNA yields from each sample were measured, and the percentage of derivative samples meeting the Oncomine™ Dx Express Test minimum input requirement of ≥0.5 ng/µL was calculated. All (100%) derivative samples created using Method 1 met the minimum DNA and RNA input requirement with 9 × 5 µm tissue input. Similarly, all (100%) derivative samples created using

Method 2 met the RNA input requirement of 0.5 ng/µL with 2 × 5 µm tissue input. However, 81.8% of the samples generated using Method 2 did not initially meet the DNA input requirement with 2 × 5 µm tissue input. When these samples were reprocessed using 9 × 5 µm tissue input, the pass rate increased to 100%. These results support the use of 9 × 5 µm tissue input for derivative samples created by both methods to consistently meet the Oncomine™ Dx Express Test input concentration requirements. Variant call results were compared between donor resection and derivative CNB/STB samples to determine sample-level concordance for the variant of interest. All SNVs, insertions, and the deletion variant evaluated, including the EGFR exon 20 insertion, were consistently detected in all derivative samples generated by both methods, resulting in a PPA of 100%.

- **Fine Needle Aspirate Specimen Type**

  The performance equivalency of Oncomine™ Dx Express Test was evaluated by comparing results obtained from donor FFPE tissue resection blocks and contrived FNA cell block samples derived from fresh/frozen tissue adjacent to the FFPE resection block. Three NSCLC patient fresh-frozen tissue samples containing clinically relevant tumor profiling variants, including MET SNV resulting in exon 14 skipping, EGFR exon 19 deletion, and *EGFR* L858R were used to generate contrived FNAs with a 22-gauge needle and saline solution (in place of ethanol) to hydrate, disrupt, and flush cells. FFPE cell blocks were prepared from the contrived FNA specimens, and the performance of these cell blocks was compared with the corresponding FFPE resection samples.

  After the contrived FNA cell block samples were created, a pathologist assessed the cellularity/tumor content using standard Hematoxylin & Eosin (H&E) staining. Samples with adequate tumor content were included in the study. Donor resection FFPE blocks also underwent standard pathological review to assess tumor content and necrosis levels. Both FNA cell blocks and FFPE resection samples were processed using the Oncomine™ Dx Express Test per the instructions in the Oncomine™ Dx Express Test User Guide. Two 5-µm sections of resection FFPE samples and seven 5-µm sections of FNA cell blocks were used as the input. The concordance of variant detection for the variant of interest was assessed between resection FFPE tissue samples and their corresponding derivative FNA cell block samples.

  DNA and RNA yields were measured, and the percentage of samples meeting the Oncomine™ Dx Express Test input requirements (≥0.5 ng/µL) was calculated. All samples in the study (6/6, with two replicates per sample) met the minimum DNA and RNA input concentrations, resulting in a 100% pass rate.

  Variant call results were analyzed, and sample-level concordance for the variants of interest was calculated between donor resection samples and matched FNA cell block samples. All variants of interest were detected in both sample types, resulting a 100% PPA between the resection and FNA cell block samples. In addition to the prespecified variants of interest, several additional variants were detected. Some of these were detected in both the resection FFPE samples and their corresponding FNA cell block samples, while others were observed only in one of the two sample types. Most of these additional variants were present at low allelic frequencies below the Oncomine™ Dx Express Test LoD, and therefore are less likely to be consistently detected across both sample types; however, a few variants were detected at higher frequencies. Although the derivative FNAs and resection FFPE samples were prepared from adjacent areas of the same tissue specimen, tumor heterogeneity may have contributed to the observed differences in variant detection.

  Assay performance to support the use of CNB and FNA specimen types for the tumor profiling indication has not yet been adequately demonstrated for all targeted variant types in the Oncomine™ Dx Express Test panel. A precision study evaluating the detection of tumor profiling variants using CNB and FNA cell block samples, along with a specimen type equivalency study

comparing results from CNB and FNA specimens to those from matched resection (reference) samples, will be conducted in a post-market setting.

## FFPE slide versus curl equivalency study

The performance of Oncomine™ Dx Express Test was evaluated using FFPE slides and curls as sample input types. The study included samples from a broad range of tissue types, including bile duct, bladder, brain, breast, colon, endometrium, lung, pancreas, skin, and thyroid. The evaluated variants included CDx variants (EGFR exon 20 insertions) as well as representative tumor profiling variants such as SNVs, insertions, deletions, CNVs, fusions, and RNA splice variants. A total of 48 FFPE tissue resection samples, 24 CNB samples, and 12 FNA samples were collected and processed into FFPE slides. A subset of these, including 42 samples (24 resections, 12 CNBs, and 6 FNAs), was processed as curls and tested with Oncomine™ Dx Express Test for direct comparison with FFPE slides. All samples contained at least one detectable variant.

All slide and curl samples met the minimum DNA and RNA concentration requirements (≥0.5 ng/µL) using the recommended tissue input, resulting a 100% pass rate (N=42). Variant-level PPA and NPA were calculated for variants of interest between slides and curls. Across all sample types, when excluding no calls, a PPA of 80.46% (70/87) and NPA of 99.98% (130038/130058) were observed. When broken down by sample type, PPA and NPA were 74.58% (44/59) and 99.99% (74511/74522) for resection samples, 90.91% (20/22) and 99.98% (36635/36644) for CNB samples, and 100% for both PPA (6/6) and NPA (18892/18892) in FNA samples. The lower PPA values were primarily attributed to variants detected below the Oncomine™ Dx Express Test LoD, which were only identified in one of the paired input types (either slide or curl). When considering only variants detected above the LoD in slide samples, the PPA increased to 97.10% (66/68) across all input types, with 97.70% (42/43) for resection samples, 95.00% (19/20) for CNBs, and 100% (5/5) for FNAs. These results demonstrate that when variants are present above the LoD, FFPE slides and curls perform equivalently as input types for Oncomine™ Dx Express Test testing.

## Extraction method equivalency study

A nucleic acid extraction method equivalency study was conducted to demonstrate that Oncomine™ Dx Express Test produces comparable variant call data when DNA and RNA samples are extracted from FFPE tissue samples with 3 different extraction methods. The Genexus™ Dx FFPE DNA and RNA Purification (Genexus™) was evaluated alongside 2 alternative extraction approaches: a silica-based column method (Method 1) and a bead-based method (Method 2). DNA and RNA were extracted from 12 clinical FFPE tissue samples representing a broad range of tumor types, including bile duct, bladder, brain, breast, colon, lung, pancreas, prostate, skin, and thyroid. The selected samples contained diverse variant types, including 6 SNVs, 1 insertion (EGFR exon 20 insertions, 9 bp), 1 deletion, 2 CNVs, 1 fusion, and 1 alternative splice variant. For each sample, 2 × 5 µm sections from FFPE tissue blocks were used. To minimize variability due to tumor content, sections were randomly assigned across replicates and extraction methods. Extractions were performed in duplicate by 2 operators over 3 non-consecutive days, resulting in 12 replicates per extraction method (2 operators × 3 days × 2 replicates) and a total of 36 replicates per sample. Sequencing was performed on the Genexus™ Dx Integrated Sequencer in accordance with the Oncomine™ Dx Express Test User Guide.

All extracted samples met the minimum input requirements, with DNA and RNA concentrations of at least 0.5 ng/µL, resulting in a 100% pass rate. Concordance between extraction methods was evaluated. A PPA of 100% was observed between Genexus™ and both Method 1 and Method 2. For the EGFR exon 19 deletion variant, the NPA was 98.86% between Genexus™ and Method 1 and

98.85% between Genexus™ and Method 2. For all other samples, an NPA of 100% was observed between Genexus™ and both comparator methods. Reproducibility of each extraction method was also assessed. All variants of interest were consistently identified in every replicate, yielding a 100% positive call rate. The IDH1 R132C variant showed a negative call rate of 98.47% with the Genexus™ method, while the IDH1 R132H variant and the AR-AR.A3A4.V7 RNA splice variant had negative call rates of 99.23% and 99.14%, respectively, with Method 2. All other variants demonstrated a 100% negative call rate across all methods. These results demonstrate that Oncomine™ Dx Express Test provides consistent and reliable variant detection regardless of the nucleic acid extraction method used.

# Clinical studies

## EGFR exon 20 insertions clinical study—clinical accuracy

To evaluate the ability of the Oncomine™ Dx Express Test to identify the EGFR exon 20 insertion mutation in FFPE non-small cell lung cancer (NSCLC) tumor specimens, 424 samples (192 positive specimens from patients enrolled in clinical trial WU-KONG1B (NCT03974022) and 232 commercially-sourced biomarker-negative samples) were also analyzed by comparing results with the Oncomine™ Dx Express Test to results obtained with the enrolling clinical trial assays (CTAs) used in the trial.

In all, 335 samples (192 CTA+ from clinical trial and 143 CTA– from commercial vendor) were used to evaluate concordance between the Oncomine™ Dx Express Test as an investigational method and the CTAs. Of these 335 samples, a total of 60 samples were invalid or unknown with the Oncomine™ Dx Express Test due to insufficient concentration or sample.

Among the 192 EGFR exon 20 insertion–positive samples, 49 were excluded from testing, resulting in 143 samples tested with the Oncomine™ Dx Express Test. Of these, 138 were confirmed positive, 3 were negative, and 2 did not yield a definitive result.

Of the 232 commercially-sourced samples, 143 were confirmed as EGFR exon 20 insertion-negative by NGS or PCR-based assays, which are representative of the CTAs, 86 were invalid, and 3 were identified as EGFR exon 20 insertion-positive. Following screening, 143 negative samples were selected for Oncomine™ Dx Express Test testing. Of these, 6 were excluded, leaving 137 samples tested with the Oncomine™ Dx Express Test. Among them, 134 were confirmed negative and 3 did not yield a definitive result.

The positive percent agreement (PPA) was defined as the proportion of EGFR exon 20 insertion-positive specimens as called by the CTAs that were also EGFR exon 20 insertion-positive as called by the Oncomine™ Dx Express Test, and the negative percent agreement (NPA) was defined as the proportion of EGFR exon 20 insertion-negative specimens as called by the CTAs that were also EGFR exon 20

insertion-negative as called by the Oncomine™ Dx Express Test. The PPA, NPA, and overall percent agreement (OPA) are shown in Table 39.

**Table 39 EGFR exon 20 insertions—concordance with CTAs**

| Agreement measure | Excluding unknowns[1] | | Including unknowns[1] | |
|---|---|---|---|---|
| | Percent agreement | Wilson Score 95% CI | Percent agreement | Wilson Score 95% CI |
| Positive Percent Agreement (PPA) | 97.9% (138/141) | (93.9%, 99.3%) | 71.9% (138/192) | (65.1%, 77.8%) |
| Negative Percent Agreement (NPA) | 100% (134/134) | (97.2%, 100.0%) | 93.7% (134/143) | (88.5%, 96.7%) |
| Overall Percent Agreement (OPA) | 98.9% (272/275) | (96.8%, 99.6%) | 81.2% (272/335) | (76.7%, 85.0%) |

[1] Unknowns are defined as invalid due to insufficient sample, or sample QC sequencing failure resulting in an invalid result or No Call for the variant.

The study samples were enrolled or screened using CTAs, leading to an enriched CTA-positive population. Because it was not feasible to test all screened subjects on the Oncomine™ Dx Express Test, the positive predictive value (PPV) and negative predictive value (NPV) were calculated and adjusted based on the EGFR exon 20 insertion-positive prevalence of 1.8% to account for potential bias due to enrichment. The adjusted PPV and NPV were both 100%, with 95% CIs of 97.3% to 100% and 99.9% to 100%, respectively.

## EGFR exon 20 insertions clinical study—clinical effectiveness

The clinical effectiveness of ZEGFROVY™ (sunvozertinib) for treating patients with non-small cell lung cancer (NSCLC) that tested positive for an EGFR exon 20 insertion variant was evaluated in the WU-KONG1B (NCT03974022) clinical trial. The full analysis set (FAS) consisted of 192 patients, including the primary efficacy population of 85 patients who received the 200 mg dose of sunvozertinib, and 107 patients who received an unapproved dose. Overall response rates (ORRs) were evaluated by BIRC according to RECIST v1.1 for the primary efficacy population.

In the 200 mg cohort (N=85), the ORR in the clinical trial assay (CTA)-positive population was 46% (39/85), with 6% (5/85) of patients having a complete response (CR) and 40% (34/85) having a partial response (PR). Among these, 58 patients were both CTA-positive and tested positive for EGFR exon 20 insertions using the Oncomine™ Dx Express Test (ODxET+ | CTA+). In this subgroup (N=58), the ORR was 41% (24/58), with 5% (3/58) patients having a CR and 36% (21/58) having a PR. These results, summarized in Table 40, demonstrate that the ORRs for the Oncomine™ Dx Express Test EGFR exon 20 insertion–positive population align with those observed in the CTA EGFR exon 20 insertion-positive population.

In the 200 mg cohort, the median duration of response (DoR) for the CTA-positive population was 11.1 months, with 72% of patients having a DoR ≥6 months and a median follow-up of 15.2 months. For the ODxET+ | CTA+ population, the median DoR was 9.8 months, with 63% of patients having a DoR ≥6 months and a median follow-up of 11.7 months. Additional details are provided in Table 40.

**Table 40 Efficacy results of the WU-KONG1B study**

| Efficacy parameter | ZEGFROVY™ (N=85, CTA +) | ODxET + \| CTA + (N=58) |
|---|---|---|
| **Overall Response Rate (ORR), % (95% CI)**[1] | 46 (35, 57) | 41 (29, 55) |
| Complete Response, % | 6 | 5 |
| Partial Response, % | 40 | 36 |
| **Duration of Response (DoR)** | N=39 | N=24 |
| Median[2] (95% CI) | 11.1 months (8.2, NE) | 9.8 months (5.6, NE) |
| Patients with DoR ≥6 months | 72% | 63% |

[1] 95% CI for ORR was calculated based on Clopper-Pearson exact CI method. CI: confidence interval

[2] Kaplan-Meier estimate using confirmed responses. NE: not evaluable

B

# Variants targeted by the Oncomine™ Dx Express Test

## DNA variants targeted by the Oncomine™ Dx Express Test

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| AKT1 | 3 | p.E17K | c.49G>A | COSM33765 |
| AKT1 | 3 | p.P51L | c.152C>T | COSM4468165 |
| AKT1 | 3 | p.P51S | c.151C>T | COSM5991558 |
| AKT1 | 3 | p.L52R | c.155T>G | COSM93893 |
| AKT1 | 4 | p.P70_C77dup | c.232_233insCCAACACCTTCATCATCCGCTGCC | COSM6568358 |
| AKT1 | 4 | p.R76_C77insWPNTFIIR | c.230_231insGCCCAACACCTTCATCATCCGCTG | COSM5045024 |
| AKT1 | 4 | p.C77F | c.230G>T | COSM5027916 |
| AKT1 | 4 | p.Q79K | c.235C>A | COSM159008 |
| AKT2 | 3 | p.E17K | c.49G>A | COSM159009 |
| AKT2 | 11 | p.D324G | c.971A>G | COSM474741 |
| AKT2 | 11 | p.D324H | c.970G>C | COSM474742 |
| AKT3 | 3 | p.E17K | c.49G>A | COSM224779 |
| AKT3 | 3 | p.L51R | c.152T>G | COSM309035 |
| AKT3 | 4 | p.Q78K | c.232C>A | COSM242892 |
| ALK | 21 | p.G1123S | c.3367G>A | COSM5045108 |
| ALK | 21 | p.G1123D | c.3368G>A | OM3262 |
| ALK | 21 | p.G1128A | c.3383G>C | COSM98475 |
| ALK | 21 | p.E1129V | c.3386A>T | BT117 |
| ALK | 22 | p.T1151M | c.3452C>T | COSM98478 |
| ALK | 22 | p.T1151R | c.3452C>G | OM3260 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ALK | 22 | p.T1151K | c.3452C>A | OM3261 |
| ALK | 22 | p.T1151dup | c.3453_3454insACG | COSM144252 |
| ALK | 22 | p.L1152V | c.3454C>G | OM3513 |
| ALK | 22 | p.L1152P | c.3455T>C | COSM1407659 |
| ALK | 22 | p.L1152R | c.3455T>G | COSM97185 |
| ALK | 22 | p.E1154Q | c.3460G>C | OM3324 |
| ALK | 22 | p.C1156T | c.3466_3467delTGinsAC | OM3350 |
| ALK | 22 | p.C1156F | c.3467G>T | BT144 |
| ALK | 22 | p.C1156Y | c.3467G>A | COSM99136 |
| ALK | 22 | p.S1157F | c.3470C>T | BT145 |
| ALK | 22 | p.I1171H | c.3511_3512delATinsCA | OM3163 |
| ALK | 22 | p.I1171N | c.3512_3513delTCinsAT | COSM1169448 |
| ALK | 22 | p.I1171N | c.3512T>A | COSM28498 |
| ALK | 22 | p.I1171T | c.3512T>C | COSM4381100 |
| ALK | 22 | p.I1171S | c.3512T>G | COSM4440758 |
| ALK | 23 | p.F1174V | c.3520T>G | COSM28054 |
| ALK | 23 | p.F1174L | c.3520T>C | COSM28057 |
| ALK | 23 | p.F1174I | c.3520T>A | COSM28491 |
| ALK | 23 | p.F1174C | c.3521T>G | COSM28059 |
| ALK | 23 | p.F1174S | c.3521T>C | COSM53063 |
| ALK | 23 | p.F1174L | c.3522C>A | COSM28055 |
| ALK | 23 | p.F1174L | c.3522C>G | COSM28061 |
| ALK | 23 | p.V1180L | c.3538G>C | COSM4381101 |
| ALK | 23 | p.R1192P | c.3575G>C | OM3259 |
| ALK | 23 | p.R1192Q | c.3575G>A | OM3372 |
| ALK | 23 | p.L1196M | c.3586C>A | COSM99137 |
| ALK | 23 | p.L1196Q | c.3587T>A | COSM1169447 |
| ALK | 23 | p.L1198F | c.3592C>T | COSM97028 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ALK | 23 | p.L1198V | c.3592C>G | OM3314 |
| ALK | 23 | p.L1198P | c.3593T>C | OM3162 |
| ALK | 23 | p.A1200V | c.3599C>T | COSM317003 |
| ALK | 23 | p.G1202del | c.3603_3605delGGG | OMINDEL698 |
| ALK | 23 | p.G1202R | c.3604G>A | COSM144250 |
| ALK | 23 | p.D1203N | c.3607G>A | COSM6191344 |
| ALK | 23 | p.D1203A | c.3608A>C | OM3313 |
| ALK | 23 | p.S1206C | c.3617C>G | BT147 |
| ALK | 23 | p.S1206Y | c.3617C>A | COSM144251 |
| ALK | 23 | p.S1206F | c.3617C>T | COSM6909200 |
| ALK | 23 | p.R1209Q | c.3626G>A | COSM327056 |
| ALK | 23 | p.E1210K | c.3628G>A | OM3161 |
| ALK | 24 | p.R1231W | c.3691C>T | BT148 |
| ALK | 24 | p.R1231Q | c.3692G>A | COSM5967491 |
| ALK | 24 | p.F1245I | c.3733T>A | COSM28492 |
| ALK | 24 | p.F1245V | c.3733T>G | COSM28499 |
| ALK | 24 | p.F1245C | c.3734T>G | COSM28500 |
| ALK | 24 | p.F1245L | c.3735C>G | COSM28062 |
| ALK | 24 | p.F1245L | c.3735C>A | COSM28493 |
| ALK | 25 | p.I1268V | c.3802A>G | BT149 |
| ALK | 25 | p.G1269A | c.3806G>C | COSM1169707 |
| ALK | 25 | p.R1275Q | c.3824G>A | COSM28056 |
| ALK | 25 | p.R1275L | c.3824G>T | COSM28060 |
| ALK | 25 | p.Y1278S | c.3833A>C | COSM28058 |
| ALK | 26 | p.A1280V | c.3839C>T | COSM6914238 |
| AR | 4 | p.L702H | c.2105T>A | COSM238553 |
| AR | 4 | p.V716M | c.2146G>A | COSM3094417 |
| AR | 5 | p.V731M | c.2191G>A | COSM1468984 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| AR | 5 | p.W742C | c.2226G>T | COSM5572266 |
| AR | 5 | p.W742C | c.2226G>C | COSM5944171 |
| AR | 5 | p.W742L | c.2225G>T | COSM5944172 |
| AR | 6 | p.F814V | c.2440T>G | COSM150793 |
| AR | 8 | p.H875Y | c.2623C>T | COSM238555 |
| AR | 8 | p.H875Q | c.2625T>G | COSM5945766 |
| AR | 8 | p.F877L | c.2629T>C | COSM5945752 |
| AR | 8 | p.F877L | c.2631C>A | COSM5946164 |
| AR | 8 | p.T878A | c.2632A>G | COSM236693 |
| AR | 8 | p.T878S | c.2633C>G | COSM5570418 |
| AR | 8 | p.D880E | c.2640C>A | COSM5945750 |
| AR | 8 | p.L882I | c.2644C>A | COSM5945751 |
| AR | 8 | p.S889G | c.2665A>G | COSM5945753 |
| AR | 8 | p.D891H | c.2671G>C | COSM5945767 |
| AR | 8 | p.E894K | c.2680G>A | COSM5945748 |
| AR | 8 | p.M896V | c.2686A>G | COSM5945749 |
| AR | 8 | p.M896T | c.2687T>C | OM3321 |
| AR | 8 | p.E898G | c.2693A>G | OM3322 |
| ARAF | 7 | p.S214T | c.640T>A | COSM1558277 |
| ARAF | 7 | p.S214A | c.640T>G | COSM1742787 |
| ARAF | 7 | p.S214C | c.641C>G | COSM5044705 |
| ARAF | 7 | p.S214Y | c.641C>A | COSM5416443 |
| ARAF | 7 | p.S214F | c.641C>T | COSM612884 |
| BRAF | 11 | p.R462I | c.1385G>T | COSM447 |
| BRAF | 11 | p.I463S | c.1388T>G | COSM448 |
| BRAF | 11 | p.G464R | c.1390G>C | COSM1111 |
| BRAF | 11 | p.G464R | c.1390G>A | COSM1448615 |
| BRAF | 11 | p.G464E | c.1391G>A | COSM449 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| BRAF | 11 | p.G464V | c.1391G>T | COSM450 |
| BRAF | 11 | p.G466R | c.1396G>C | COSM1112 |
| BRAF | 11 | p.G466R | c.1396G>A | COSM253328 |
| BRAF | 11 | p.G466V | c.1397G>T | COSM451 |
| BRAF | 11 | p.G466A | c.1397G>C | COSM452 |
| BRAF | 11 | p.G466E | c.1397G>A | COSM453 |
| BRAF | 11 | p.G469S | c.1405_1407delGGAinsAGC | COSM1113 |
| BRAF | 11 | p.G469S | c.1405_1407delGGAinsAGT | COSM27912 |
| BRAF | 11 | p.G469R | c.1405G>C | COSM455 |
| BRAF | 11 | p.G469R | c.1405G>A | COSM457 |
| BRAF | 11 | p.G469S | c.1405_1406delGGinsTC | COSM458 |
| BRAF | 11 | p.G469L | c.1405_1406delGGinsTT | COSM6056848 |
| BRAF | 11 | p.G469V | c.1406G>T | COSM459 |
| BRAF | 11 | p.G469A | c.1406G>C | COSM460 |
| BRAF | 11 | p.G469E | c.1406G>A | COSM461 |
| BRAF | 11 | p.Y472C | c.1415A>G | COSM1133046 |
| BRAF | 15 | p.N581T | c.1742A>C | COSM1699452 |
| BRAF | 15 | p.N581I | c.1742A>T | COSM26460 |
| BRAF | 15 | p.N581S | c.1742A>G | COSM462 |
| BRAF | 15 | p.E586K | c.1756G>A | COSM463 |
| BRAF | 15 | p.[G593=;D594N] | c.1779_1780delTGinsGA | COSM211600 |
| BRAF | 15 | p.D594H | c.1780G>C | COSM144576 |
| BRAF | 15 | p.D594N | c.1780G>A | COSM27639 |
| BRAF | 15 | p.D594Y | c.1780G>T | COSM6928839 |
| BRAF | 15 | p.D594A | c.1781A>C | COSM1583010 |
| BRAF | 15 | p.D594V | c.1781A>T | COSM466 |
| BRAF | 15 | p.D594G | c.1781A>G | COSM467 |
| BRAF | 15 | p.D594E | c.1782T>A | COSM1121 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| BRAF | 15 | p.D594E | c.1782T>G | COSM253330 |
| BRAF | 15 | p.F595L | c.1783T>C | COSM21612 |
| BRAF | 15 | p.F595S | c.1784T>C | COSM1123 |
| BRAF | 15 | p.F595L | c.1785T>G | COSM468 |
| BRAF | 15 | p.F595L | c.1785T>A | COSM53198 |
| BRAF | 15 | p.G596R | c.1786G>C | COSM469 |
| BRAF | 15 | p.G596C | c.1786G>T | COSM6936824 |
| BRAF | 15 | p.G596D | c.1787G>A | COSM26506 |
| BRAF | 15 | p.G596V | c.1787G>T | COSM9145962 |
| BRAF | 15 | p.L597S | c.1789_1790delCTinsTC | COSM1126 |
| BRAF | 15 | p.L597V | c.1789C>G | COSM470 |
| BRAF | 15 | p.L597Q | c.1790T>A | COSM1125 |
| BRAF | 15 | p.L597P | c.1790T>C | COSM1448593 |
| BRAF | 15 | p.L597R | c.1790T>G | COSM471 |
| BRAF | 15 | p.A598T | c.1792G>A | COSM28505 |
| BRAF | 15 | p.A598V | c.1793C>T | COSM21549 |
| BRAF | 15 | p.[T599I;V600K] | c.1796_1799delCAGTinsTAAA | COSM1735763 |
| BRAF | 15 | p.T599dup | c.1796_1797insTAC | COSM30730 |
| BRAF | 15 | p.T599R | c.1796C>G | COSM4172020 |
| BRAF | 15 | p.T599I | c.1796C>T | COSM472 |
| BRAF | 15 | p.[T599R;V600G] | c.1796_1799delCAGTinsGAGG | OM3547 |
| BRAF | 15 | p.[T599=;V600R] | c.1797_1799delAGTinsGAG | COSM1127 |
| BRAF | 15 | p.T599dup | c.1797_1798insACA | COSM144982 |
| BRAF | 15 | p.D594_T599dup | c.1798_1799insATTTTGGTCTAGCTACAG | COSM26504 |
| BRAF | 15 | p.T599_V600insEAT | c.1798_1799insAGGCTACAG | COSM4166148 |
| BRAF | 15 | p.T599_V600insETT | c.1798_1799insAGACTACAG | COSM4172018 |
| BRAF | 15 | p.V600M | c.1798G>A | COSM1130 |
| BRAF | 15 | p.V600R | c.1798_1799delGTinsCG | COSM1583011 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| BRAF | 15 | p.V600L | c.1798G>C | COSM219798 |
| BRAF | 15 | p.V600Q | c.1798_1799delGTinsCA | COSM249889 |
| BRAF | 15 | p.V600L | c.1798G>T | COSM33808 |
| BRAF | 15 | p.V600K | c.1798_1799delGTinsAA | COSM473 |
| BRAF | 15 | p.V600R | c.1798_1799delGTinsAG | COSM474 |
| BRAF | 15 | p.V600E | c.1799T>A | COSM476 |
| BRAF | 15 | p.V600_K601delinsE | c.1799_1801delTGA | COSM1133 |
| BRAF | 15 | p.V600A | c.1799T>C | COSM18443 |
| BRAF | 15 | p.V600D | c.1799_1800delTGinsAC | COSM308550 |
| BRAF | 15 | p.V600E | c.1799_1800delTGinsAA | COSM475 |
| BRAF | 15 | p.V600D | c.1799_1800delTGinsAT | COSM477 |
| BRAF | 15 | p.V600G | c.1799T>G | COSM6137 |
| BRAF | 15 | p.K601E | c.1801A>G | COSM478 |
| BRAF | 15 | p.K601R | c.1802A>G | COSM1568586 |
| BRAF | 15 | p.K601I | c.1802A>T | COSM26491 |
| BRAF | 15 | p.K601T | c.1802A>C | COSM3878760 |
| BRAF | 15 | p.K601N | c.1803A>C | COSM1132 |
| BRAF | 15 | p.K601N | c.1803A>T | COSM6265 |
| CDK4 | 2 | p.K22Q | c.64A>C | OM3153 |
| CDK4 | 2 | p.K22R | c.65A>G | COSM232013 |
| CDK4 | 2 | p.K22M | c.65A>T | COSM3463915 |
| CDK4 | 2 | p.R24C | c.70C>T | COSM1677139 |
| CDK4 | 2 | p.R24S | c.70C>A | COSM3463914 |
| CDK4 | 2 | p.R24H | c.71G>A | COSM1989836 |
| CDK4 | 2 | p.R24L | c.71G>T | COSM363684 |
| CHEK2 | 11 | p.[S372=;K373E] | c.1116_1117delCAinsTG | COSM1294063 |
| CHEK2 | 11 | p.K373E | c.1117A>G | COSM42871 |
| CHEK2 | 11 | p.M381V | c.1141A>G | COSM1559601 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| CHEK2 | 15 | p.R523P | c.1568G>C | COSM1559595 |
| CHEK2 | 15 | p.R523H | c.1568G>A | COSM1559596 |
| CTNNB1 | 3 | p.D32Y | c.94G>T | COSM5661 |
| CTNNB1 | 3 | p.D32H | c.94G>C | COSM5668 |
| CTNNB1 | 3 | p.D32N | c.94G>A | COSM5672 |
| CTNNB1 | 3 | p.D32G | c.95A>G | COSM5681 |
| CTNNB1 | 3 | p.D32A | c.95A>C | COSM5690 |
| CTNNB1 | 3 | p.D32V | c.95A>T | COSM5691 |
| CTNNB1 | 3 | p.D32E | c.96C>G | COSM22939 |
| CTNNB1 | 3 | p.D32E | c.96C>A | COSM5766 |
| CTNNB1 | 3 | p.S33T | c.97T>A | COSM27311 |
| CTNNB1 | 3 | p.S33P | c.97T>C | COSM5682 |
| CTNNB1 | 3 | p.S33A | c.97T>G | COSM5683 |
| CTNNB1 | 3 | p.S33L | c.97_98delTCinsCT | COSM6098 |
| CTNNB1 | 3 | p.S33N | c.97_98delTCinsAA | COSM6099 |
| CTNNB1 | 3 | p.S33F | c.98C>T | COSM5669 |
| CTNNB1 | 3 | p.S33Y | c.98C>A | COSM5673 |
| CTNNB1 | 3 | p.S33C | c.98C>G | COSM5677 |
| CTNNB1 | 3 | p.G34I | c.100_101delGGinsAT | COSM1666841 |
| CTNNB1 | 3 | p.G34R | c.100G>C | COSM5684 |
| CTNNB1 | 3 | p.G34R | c.100G>A | COSM5686 |
| CTNNB1 | 3 | p.G34V | c.101G>T | COSM5670 |
| CTNNB1 | 3 | p.G34E | c.101G>A | COSM5671 |
| CTNNB1 | 3 | p.G34A | c.101G>C | COSM5988492 |
| CTNNB1 | 3 | p.I35V | c.103A>G | COSM1566774 |
| CTNNB1 | 3 | p.I35T | c.104T>C | COSM13168 |
| CTNNB1 | 3 | p.I35K | c.104_105delTCinsAA | COSM28727 |
| CTNNB1 | 3 | p.I35S | c.104T>G | COSM5674 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| CTNNB1 | 3 | p.I35N | c.104T>A | COSM5695 |
| CTNNB1 | 3 | p.I35M | c.105C>G | COSM28711 |
| CTNNB1 | 3 | p.H36Y | c.106C>T | COSM5703 |
| CTNNB1 | 3 | p.H36R | c.107A>G | COSM27378 |
| CTNNB1 | 3 | p.H36P | c.107A>C | COSM5678 |
| CTNNB1 | 3 | p.S37A | c.109T>G | COSM5675 |
| CTNNB1 | 3 | p.S37P | c.109T>C | COSM5687 |
| CTNNB1 | 3 | p.S37T | c.109T>A | COSM5729 |
| CTNNB1 | 3 | p.S37F | c.110C>T | COSM5662 |
| CTNNB1 | 3 | p.S37Y | c.110C>A | COSM5666 |
| CTNNB1 | 3 | p.S37C | c.110C>G | COSM5679 |
| CTNNB1 | 3 | p.T41A | c.121A>G | COSM5664 |
| CTNNB1 | 3 | p.T41P | c.121A>C | COSM5688 |
| CTNNB1 | 3 | p.T41S | c.121A>T | COSM5716 |
| CTNNB1 | 3 | p.T41I | c.122C>T | COSM5676 |
| CTNNB1 | 3 | p.T41S | c.122C>G | COSM5701 |
| CTNNB1 | 3 | p.T41N | c.122C>A | COSM5730 |
| CTNNB1 | 3 | p.P44A | c.130C>G | COSM17661 |
| CTNNB1 | 3 | p.P44S | c.130C>T | COSM5704 |
| CTNNB1 | 3 | p.P44L | c.131C>T | COSM5761 |
| CTNNB1 | 3 | p.S45_S47del | c.133_141delTCTCTGAGT | COSM1617582 |
| CTNNB1 | 3 | p.S45P | c.133T>C | COSM5663 |
| CTNNB1 | 3 | p.S45A | c.133T>G | COSM5685 |
| CTNNB1 | 3 | p.S45T | c.133T>A | COSM5719 |
| CTNNB1 | 3 | p.S45_G48del | c.133_144delTCTCTGAGTGGT | COSM5992 |
| CTNNB1 | 3 | p.S45del | c.133_135delTCT | COSM6128 |
| CTNNB1 | 3 | p.[S45E;L46=] | c.133_136delTCTCinsGAGT | COSM86011 |
| CTNNB1 | 3 | p.L46_S47del | c.133_138delTCTCTG | COSM6129 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| CTNNB1 | 3 | p.S45_S47delinsC | c.134_139delCTCTGA | COSM28708 |
| CTNNB1 | 3 | p.S45F | c.134C>T | COSM5667 |
| CTNNB1 | 3 | p.S45C | c.134C>G | COSM5689 |
| CTNNB1 | 3 | p.S45Y | c.134C>A | COSM5692 |
| CTNNB1 | 3 | p.S45del | c.134_136delCTC | COSM6131 |
| CTNNB1 | 3 | p.L46_S47del | c.136_141delCTGAGT | COSM6132 |
| CTNNB1 | 7 | p.K335T | c.1004A>C | COSM1725761 |
| CTNNB1 | 7 | p.K335I | c.1004A>T | COSM17797 |
| CTNNB1 | 7 | p.K335Q | c.1003A>C | COSM4421324 |
| CTNNB1 | 7 | p.K335R | c.1004A>G | COSM5945941 |
| EGFR | 3 | p.R108G | c.322A>G | COSM1451536 |
| EGFR | 3 | p.R108K | c.323G>A | COSM21683 |
| EGFR | 7 | p.A289T | c.865G>A | COSM21686 |
| EGFR | 7 | p.A289D | c.866C>A | COSM21685 |
| EGFR | 7 | p.A289V | c.866C>T | COSM21687 |
| EGFR | 7 | p.V292L | c.874G>T | COSM43067 |
| EGFR | 12 | p.R451C | c.1351C>T | COSM3266583 |
| EGFR | 12 | p.S464L | c.1391C>T | BT126 |
| EGFR | 12 | p.G465R | c.1393G>A | COSM601581 |
| EGFR | 12 | p.G465E | c.1394G>A | BT129 |
| EGFR | 12 | p.K467T | c.1400A>C | BT132 |
| EGFR | 12 | p.I491M | c.1473A>G | BT135 |
| EGFR | 12 | p.S492R | c.1474A>C | COSM236671 |
| EGFR | 12 | p.S492R | c.1476C>A | COSM236670 |
| EGFR | 18 | p.G598A | c.1793G>C | COSM3412196 |
| EGFR | 18 | p.G598V | c.1793G>T | COSM21690 |
| EGFR | 18 | p.E709H | c.2125_2127delGAAinsCAT | COSM12428 |
| EGFR | 18 | p.E709K | c.2125G>A | COSM12988 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 18 | p.E709Q | c.2125G>C | COSM116882 |
| EGFR | 18 | p.E709_T710delinsA | c.2126_2128delAAA | COSM85796 |
| EGFR | 18 | p.E709_T710delinsG | c.2126_2129delAAACinsG | COSM48981 |
| EGFR | 18 | p.E709A | c.2126A>C | COSM13427 |
| EGFR | 18 | p.E709G | c.2126A>G | COSM13009 |
| EGFR | 18 | p.E709V | c.2126A>T | COSM12371 |
| EGFR | 18 | p.E709_T710delinsD | c.2127_2129delAAC | COSM51525 |
| EGFR | 18 | p.L718M | c.2152C>A | COSM1716255 |
| EGFR | 18 | p.L718V | c.2152C>G | OM3316 |
| EGFR | 18 | p.L718Q | c.2153T>A | COSM6503269 |
| EGFR | 18 | p.[L718=;G719C] | c.2154_2155delGGinsTT | COSM18441 |
| EGFR | 18 | p.G719S | c.2155G>A | COSM6252 |
| EGFR | 18 | p.G719C | c.2155G>T | COSM6253 |
| EGFR | 18 | p.G719D | c.2156G>A | COSM18425 |
| EGFR | 18 | p.G719A | c.2156G>C | COSM6239 |
| EGFR | 18 | p.S720I | c.2158_2159delTCinsAT | OM3317 |
| EGFR | 18 | p.S720P | c.2158T>C | COSM88729 |
| EGFR | 18 | p.G721S | c.2161G>A | COSM22992 |
| EGFR | 18 | p.G721R | c.2161G>C | OM3325 |
| EGFR | 18 | p.G721D | c.2162G>A | COSM22993 |
| EGFR | 18 | p.G724S | c.2170G>A | COSM13979 |
| EGFR | 18 | p.I744_E749delinsLKR | c.2230_2246delATCAAGGAATTAAGAGAinsCTTAAGAG | COSM133186 |
| EGFR | 18 | p.I744_A750delinsVK | c.2230_2249delATCAAGGAATTAAGAGAAGCinsGTCAA | COSM85798 |
| EGFR | 18 | p.I740_K745dup | c.2231_2232insTAAAATTCCCGTCGCTAT | COSM12423 |
| EGFR | 19 | p.I744T | c.2231T>C | COSM87245 |
| EGFR | 19 | p.K745_A750del | c.2232_2249delCAAGGAATTAAGAGAAGC | COSM221565 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.E746_A750del | c.2232_2249delCAAGGAATTAAGAGAAGCinsAAA | COSM3734668 |
| EGFR | 19 | p.K745_E749del | c.2233_2247delAAGGAATTAAGAGAA | COSM26038 |
| EGFR | 19 | p.K745_E746insTPVAIK | c.2234_2235insAACTCCCGTCGCTATCAA | COSM255152 |
| EGFR | 19 | p.I740_K745dup | c.2234_2235insAATTCCCGTCGCTATCAA | COSM51504 |
| EGFR | 19 | p.K745_A750delinsT | c.2234_2248delAGGAATTAAGAGAAG | COSM1190791 |
| EGFR | 19 | p.E746del | c.2235_2237delGGA | COSM18420 |
| EGFR | 19 | p.[K745=;E746I;L747P] | c.2235_2240delGGAATTinsAATTCC | OM3355 |
| EGFR | 19 | p.E746_E749del | c.2235_2246delGGAATTAAGAGA | COSM28517 |
| EGFR | 19 | p.E746_A750delinsIP | c.2235_2248delGGAATTAAGAGAAGinsAATTC | COSM13550 |
| EGFR | 19 | p.E746_A750del | c.2235_2249delGGAATTAAGAGAAGC | COSM6223 |
| EGFR | 19 | p.E746_T751delinsIP | c.2235_2251delGGAATTAAGAGAAGCAAinsAATTC | COSM13552 |
| EGFR | 19 | p.E746_T751delinsA | c.2235_2251delGGAATTAAGAGAAGCAAinsAG | COSM13549 |
| EGFR | 19 | p.E746_T751delinsFPS | c.2235_2251delGGAATTAAGAGAAGCAAinsATTCCCGT | COSM6506513 |
| EGFR | 19 | p.E746_T751del | c.2235_2252delGGAATTAAGAGAAGCAAC | COSM24869 |
| EGFR | 19 | p.E746_T751delinsI | c.2235_2252delGGAATTAAGAGAAGCAACinsAAT | COSM13551 |
| EGFR | 19 | p.E746_S752delinsI | c.2235_2255delGGAATTAAGAGAAGCAACATCinsAAT | COSM12385 |
| EGFR | 19 | p.K745_E746insVPVAIK | c.2236_2237insTTCCCGTCGCTATCAAGG | COSM26444 |
| EGFR | 19 | p.E746_A750delinsRP | c.2236_2248delGAATTAAGAGAAGinsAGAC | COSM12413 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.E746_A750delinsIP | c.2236_2248delGAATTAAGAGAAGinsATTC | COSM6966471<br>**Note:** The nucleotide change of COSM6966471 overlaps that of COSM6219, so a positive COSM6966471 sample also results in a positive call for COSM6219. |
| EGFR | 19 | p.E746_A750delinsQP | c.2236_2248delGAATTAAGAGAAGinsCAAC | COSM13557 |
| EGFR | 19 | p.E746_A750del | c.2236_2250delGAATTAAGAGAAGCA | COSM6225 |
| EGFR | 19 | p.E746_A750delinsP | c.2236_2250delGAATTAAGAGAAGCAinsCCT | COSM6947327 |
| EGFR | 19 | p.E746_T751delinsS | c.2236_2251delGAATTAAGAGAAGCAAinsT | COSM26513 |
| EGFR | 19 | p.E746_T751delinsI | c.2236_2252delGAATTAAGAGAAGCAACinsAT | COSM26680 |
| EGFR | 19 | p.E746_T751delinsQ | c.2236_2252delGAATTAAGAGAAGCAACinsCA | COSM22999 |
| EGFR | 19 | p.E746_T751delinsL | c.2236_2252delGAATTAAGAGAAGCAACinsCT | COSM51502 |
| EGFR | 19 | p.E746_T751del | c.2236_2253delGAATTAAGAGAAGCAACA | COSM12728 |
| EGFR | 19 | p.E746_T751delinsM | c.2236_2253delGAATTAAGAGAAGCAACAinsATG | OMINDEL716 |
| EGFR | 19 | p.E746_T751delinsIP | c.2236_2253delGAATTAAGAGAAGCAACAinsATTCCT | COSM51526 |
| EGFR | 19 | p.E746_S752delinsI | c.2236_2255delGAATTAAGAGAAGCAACATCinsAT | COSM133188 |
| EGFR | 19 | p.E746_S752del | c.2236_2256delGAATTAAGAGAAGCAACATCT | COSM133189 |
| EGFR | 19 | p.E746_S752delinsI | c.2236_2256delGAATTAAGAGAAGCAACATCTinsATC | COSM133190 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.E746_P753delinsIS | c.2236_2257delGAATTAAGAGAAGCAACATCTC insATCT | COSM133191 |
| EGFR | 19 | p.E746_P753delinsLS | c.2236_2257delGAATTAAGAGAAGCAACATCTC insCTCT | COSM13200 |
| EGFR | 19 | p.E746_P753del | c.2236_2259delGAATTAAGAGAAGCAACATCTCCG | OMINDEL950 |
| EGFR | 19 | p.E746_A750delinsAP | c.2237_2248delAATTAAGAGAAGinsCAC | COSM144207 |
| EGFR | 19 | p.E746_A750delinsVP | c.2237_2250delAATTAAGAGAAGCAinsTCCCT | COSM28623 |
| EGFR | 19 | p.E746_T751delinsA | c.2237_2251delAATTAAGAGAAGCAA | COSM12678 |
| EGFR | 19 | p.E746_T751delinsAPS | c.2237_2251delAATTAAGAGAAGCAA insCACCAT | COSM6924852 |
| EGFR | 19 | p.E746_T751delinsVP | c.2237_2251delAATTAAGAGAAGCAA insTCC | COSM6968288 |
| EGFR | 19 | p.E746_T751delinsVA | c.2237_2251delAATTAAGAGAAGCAA insTGG | COSM53205 |
| EGFR | 19 | p.E746_T751delinsVP | c.2237_2251delAATTAAGAGAAGCAA insTTC | COSM18421 |
| EGFR | 19 | p.E746_T751delinsVS | c.2237_2251delAATTAAGAGAAGCAA insTTT | COSM6980200 |
| EGFR | 19 | p.E746_T751delinsV | c.2237_2252delAATTAAGAGAAGCAAC insT | COSM12386 |
| EGFR | 19 | p.E746_T751delinsV | c.2237_2253delAATTAAGAGAAGCAACA insTC | COSM133193 |
| EGFR | 19 | p.E746_T751delinsVP | c.2237_2253delAATTAAGAGAAGCAACA insTTCCT | COSM52935 |
| EGFR | 19 | p.E746_T751delinsVA | c.2237_2253delAATTAAGAGAAGCAACA insTTGCT | COSM12416 |
| EGFR | 19 | p.E746_S752delinsA | c.2237_2254delAATTAAGAGAAGCAAC AT | COSM12367 |
| EGFR | 19 | p.E746_S752delinsV | c.2237_2255delAATTAAGAGAAGCAAC ATCinsT | COSM12384 |
| EGFR | 19 | p.E746_S752delinsV | c.2237_2256delAATTAAGAGAAGCAAC ATCTinsTC | COSM18426 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.E746_S752delinsV | c.2237_2256delAATTAAGAGAAGCAACATCTinsTG | COSM674057 |
| EGFR | 19 | p.E746_P753delinsVS | c.2237_2257delAATTAAGAGAAGCAACATCTCinsTCT | COSM18427 |
| EGFR | 19 | p.E746_P753delinsVQ | c.2237_2258delAATTAAGAGAAGCAACATCTCCinsTTCA | COSM51524 |
| EGFR | 19 | p.E746G | c.2237A>G | COSM6849765 |
| EGFR | 19 | p.L747_A750delinsP | c.2238_2248delATTAAGAGAAGinsGC | COSM12422 |
| EGFR | 19 | p.E746_A750delinsDP | c.2238_2248delATTAAGAGAAGinsTC | COSM18428 |
| EGFR | 19 | p.E746_A750delinsD | c.2238_2250delATTAAGAGAAGCAinsC | COSM6974307 |
| EGFR | 19 | p.L747_T751delinsP | c.2238_2251delATTAAGAGAAGCAAinsGC | COSM22944 |
| EGFR | 19 | p.L747_T751delinsQ | c.2238_2252delATTAAGAGAAGCAACinsGCA | COSM12419 |
| EGFR | 19 | p.E746_S752delinsD | c.2238_2255delATTAAGAGAAGCAACATC | COSM6220 |
| EGFR | 19 | p.L747_S752delinsQH | c.2238_2255delATTAAGAGAAGCAACATCinsGCAACA | COSM12421 |
| EGFR | 19 | p.L747_S752delinsQ | c.2238_2256delATTAAGAGAAGCAACATCTinsGCAA | COSM26441 |
| EGFR | 19 | p.E746_P753delinsDT | c.2238_2257delATTAAGAGAAGCAACATCTCinsCA | COSM5340693 |
| EGFR | 19 | p.L747_P753del | c.2238_2258delATTAAGAGAAGCAACATCTCC | COSM255211 |
| EGFR | 19 | p.L747P | c.2239_2240delTTinsCC | COSM24267 |
| EGFR | 19 | p.L747_E749del | c.2239_2247delTTAAGAGAA | COSM6218 |
| EGFR | 19 | p.L747_A750delinsP | c.2239_2248delTTAAGAGAAGinsC | COSM12382<br>**Note:** The nucleotide change of COSM12382 overlaps that of COSM6219, so a positive COSM12382 sample also results in a positive call for COSM6219. |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.L747_A750del | c.2239_2250delTTAAGAGAAGCA | COSM9179903 |
| EGFR | 19 | p.L747_A750delinsP | c.2239_2250delTTAAGAGAAGCAinsCCG | COSM4170220 |
| EGFR | 19 | p.L747_T751delinsP | c.2239_2251delTTAAGAGAAGCAAinsC | COSM12383 |
| EGFR | 19 | p.L747_T751delinsQ | c.2239_2252delTTAAGAGAAGCAACinsCA | COSM12420 |
| EGFR | 19 | p.L747_T751delinsN | c.2239_2253delTTAAGAGAAGCAACAinsAAT | COSM51503 |
| EGFR | 19 | p.L747_A750delinsP | c.2239_2253delTTAAGAGAAGCAACAinsCCAACG | COSM1235296 |
| EGFR | 19 | p.L747_T751delinsA | c.2239_2253delTTAAGAGAAGCAACAinsGCT | COSM23572 |
| EGFR | 19 | p.L747_S752delinsQH | c.2239_2255delTTAAGAGAAGCAACATCinsCAACA | COSM133196 |
| EGFR | 19 | p.L747_S752del | c.2239_2256delTTAAGAGAAGCAACATCT | COSM6255 |
| EGFR | 19 | p.L747_S752delinsQ | c.2239_2256delTTAAGAGAAGCAACATCTinsCAA | COSM12403 |
| EGFR | 19 | p.L747_P753delinsQ | c.2239_2258delTTAAGAGAAGCAACATCTCCinsCA | COSM12387 |
| EGFR | 19 | p.L747_P753delinsQ | c.2239_2259delTTAAGAGAAGCAACATCTCCGinsCAA | COSM51501 |
| EGFR | 19 | p.L747_K754delinsANKG | c.2239_2261delTTAAGAGAAGCAACATCTCCGAAinsGCCAACAAGGG | COSM1667023 |
| EGFR | 19 | p.L747_K754del | c.2239_2262delTTAAGAGAAGCAACATCTCCGAAA | COSM24970 |
| EGFR | 19 | p.L747_A755delinsAN | c.2239_2264delTTAAGAGAAGCAACATCTCCGAAAGCinsGCCAA | COSM85891 |
| EGFR | 19 | p.L747V | c.2239T>G | OM3537 |
| EGFR | 19 | p.L747_A750delinsS | c.2240_2248delTAAGAGAAG | COSM4170221 |
| EGFR | 19 | p.L747_T751delinsS | c.2240_2251delTAAGAGAAGCAA | COSM6210 |
| EGFR | 19 | p.L747_T751del | c.2240_2254delTAAGAGAAGCAACAT | COSM12369 |
| EGFR | 19 | p.L747_P753delinsS | c.2240_2257delTAAGAGAAGCAACATCTC | COSM12370 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.L747_K754delinsST | c.2240_2261delTAAGAGAAGCAACATC TCCGAAinsCGAC | COSM20883 |
| EGFR | 19 | p.L747_A755delinsSMS | c.2240_2263delTAAGAGAAGCAACATC TCCGAAAGinsCGATGT | COSM6933365 |
| EGFR | 19 | p.L747_A755delinsSKG | c.2240_2264delTAAGAGAAGCAACATC TCCGAAAGCinsCGAAAGG | COSM1667026 |
| EGFR | 19 | p.L747S | c.2240T>C | COSM26704 |
| EGFR | 19 | p.L747_T751delinsF | c.2241_2253delAAGAGAAGCAACAinsC | OMINDEL951 |
| EGFR | 19 | p.A750_E758delinsP | c.2248_2273delGCAACATCTCCGAAAG CCAACAAGGAinsCC | COSM26440 |
| EGFR | 19 | p.A750_I759delinsPT | c.2248_2276delGCAACATCTCCGAAAG CCAACAAGGAAATinsCCAAC | COSM5023004 |
| EGFR | 19 | p.A750P | c.2248G>C | COSM6219 |
| EGFR | 19 | p.A750_I759delinsGS | c.2249_2277delCAACATCTCCGAAAGC CAACAAGGAAATCinsGAAGT | COSM5023005 |
| EGFR | 19 | p.T751_A755del | c.2250_2264delAACATCTCCGAAAGC | COSM26718 |
| EGFR | 19 | p.T751_I759delinsN | c.2250_2276delAACATCTCCGAAAGCC AACAAGGAAATinsCAA | COSM1667024 |
| EGFR | 19 | p.T751_I759delinsS | c.2251_2276delACATCTCCGAAAGCCA ACAAGGAAATinsTC | COSM133200 |
| EGFR | 19 | p.T751_I759delinsS | c.2251_2277delACATCTCCGAAAGCCA ACAAGGAAATCinsTCT | COSM22945<br>**Note:** The nucleotide change of COSM22945 overlaps that of COSM24269, so a positive COSM22945 sample also results in a positive call for COSM24269. |
| EGFR | 19 | p.T751_E758del | c.2252_2275delCATCTCCGAAAGCCAA CAAGGAAA | COSM133207 |
| EGFR | 19 | p.T751_I759delinsN | c.2252_2276delCATCTCCGAAAGCCAA CAAGGAAATinsA | COSM96856 |
| EGFR | 19 | p.T751_I759delinsS | c.2252_2276delCATCTCCGAAAGCCAA CAAGGAAATinsG | COSM1667027 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 19 | p.T751_I759delinsN | c.2252_2277delCATCTCCGAAAGCCAACAAGGAAATCinsAT | COSM24270 |
| EGFR | 19 | p.T751_I759delinsREA | c.2252_2277delCATCTCCGAAAGCCAACAAGGAAATCinsGAGAAGCG | COSM22956 |
| EGFR | 19 | p.S752_I759del | c.2253_2276delATCTCCGAAAGCCAACAAGGAAAT | COSM13556 |
| EGFR | 19 | p.S752_I759del | c.2254_2277delTCTCCGAAAGCCAACAAGGAAATC | COSM6256 |
| EGFR | 19 | p.P753_I759del | c.2258_2278delCGAAAGCCAACAAGGAAATCC | COSM24269 |
| EGFR | 19 | p.K754_I759del | c.2260_2277delAAAGCCAACAAGGAAATC | COSM5271797 |
| EGFR | 19 | p.K757R | c.2270A>G | COSM133588 |
| EGFR | 20 | p.? | c.2284-6_2284-5insTCCAGGAAGCCT | COSM26720 |
| EGFR | 20 | p.? | c.2284-9_2284-8insCCCTCCAGGAAG | OMINDEL1050 |
| EGFR | 20 | p.Y764_V765insHH | c.2292_2293insCACCAC | OMINDEL1051 |
| EGFR | 20 | p.A767_S768insYVM | c.2301_2302insTACGTGATG | COSM1651740 |
| EGFR | 20 | p.A767_S768insTLA | c.2302_2303insCGCTGGCCA | COSM12425 |
| EGFR | 20 | p.A767_S768insIA | c.2302_2303insTAGCCA | COSM13559 |
| EGFR | 20 | p.S768G | c.2302A>G | COSM1451595 |
| EGFR | 20 | p.S768delinsRRGH | c.2303_2304insGAGGGGCCA | OMINDEL1059 |
| EGFR | 20 | p.V769_D770insANV | c.2303_2304insTGTGGCCAA | COSM1651741 |
| EGFR | 20 | p.A767_V769dup | c.2303_2304insTGTGGCCAG | COSM20884 |
| EGFR | 20 | p.[S768I;V769L] | c.2303_2305delGCGinsTCC | COSM6984779 |
| EGFR | 20 | p.[S768I;V769L] | c.2303_2305delGCGinsTCT | COSM85750 |
| EGFR | 20 | p.[S768I;V769L] | c.2303_2305delGCGinsTTT | OMINDEL1181 |
| EGFR | 20 | p.S768_V769delinsISW | c.2303_2306delGCGTinsTCAGCTG | OMINDEL1058 |
| EGFR | 20 | p.S768N | c.2303G>A | COSM12989 |
| EGFR | 20 | p.S768T | c.2303G>C | COSM291998 |
| EGFR | 20 | p.S768I | c.2303G>T | COSM6241 |
| EGFR | 20 | p.S768_V769insLDS | c.2304_2305insCTGGACAGC | OMINDEL1802 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.V769M | c.2305G>A | COSM13425 |
| EGFR | 20 | p.V769L | c.2305G>T | COSM6242 |
| EGFR | 20 | p.V769_D770insM | c.2306_2307insCAT | OMINDEL1062 |
| EGFR | 20 | p.V769_D770insP | c.2306_2307insCCC | OMINDEL1072 |
| EGFR | 20 | p.A767_V769dup | c.2306_2307insTGCCAGCGT | A767_V769dup |
| EGFR | 20 | p.[V769G;D770I] | c.2306_2309delTGGAinsGGAT | OM3540 |
| EGFR | 20 | p.V769_D770insTSV | c.2307_2308insACCAGCGTG | OMINDEL1055 |
| EGFR | 20 | p.V769_D770insSTL | c.2307_2308insAGCACCCTG | OMINDEL1066 |
| EGFR | 20 | p.V769_D770insM | c.2307_2308insATG | OMINDEL1063 |
| EGFR | 20 | p.V769_D770insMASVD | c.2307_2308insATGGCCAGCGTGGAC | COSM28638 |
| EGFR | 20 | p.V769_D770insP | c.2307_2308insCCC | OMINDEL1076 |
| EGFR | 20 | p.V769_D770insP | c.2307_2308insCCG | OMINDEL1073 |
| EGFR | 20 | p.V769_D770insRSV | c.2307_2308insCGCAGCGTG | OMINDEL952 |
| EGFR | 20 | p.V769_D770insSTL | c.2307_2308insTCCACCCTG | OMINDEL1065 |
| EGFR | 20 | p.V769_D770insSTL | c.2307_2308insTCCACCTTG | OMINDEL1064 |
| EGFR | 20 | p.V769_D770insSSV | c.2307_2308insTCCAGCGTG | OMINDEL1054 |
| EGFR | 20 | p.V769_D770insCV | c.2307_2308insTGCGTG | COSM12379 |
| EGFR | 20 | p.D770delinsNNN | c.2308_2308delGinsAACAACA | OMINDEL1077 |
| EGFR | 20 | p.D770delinsNNPH | c.2308_2308delGinsAACAACCCCC | OMINDEL1078 |
| EGFR | 20 | p.D770delinsASVH | c.2308_2309insCCAGCGTGC | OMINDEL1080 |
| EGFR | 20 | p.A767_V769dup | c.2308_2309insCCAGCGTGG | COSM12376 |
| EGFR | 20 | p.A767_V769dup | c.2308_2309insCCAGCGTTG | MATNV03 |
| EGFR | 20 | p.A767_V769dup | c.2308_2309insCCAGTGTGG | OMINDEL1803 |
| EGFR | 20 | p.D770delinsGTH | c.2308_2309insGCACAC | COSM6983510 |
| EGFR | 20 | p.V769_D770insGTM | c.2308_2309insGCACCATGG | OMINDEL1061 |
| EGFR | 20 | p.V769_D770insGTR | c.2308_2309insGCACCCGCG | OMINDEL1075 |
| EGFR | 20 | p.V769_D770insGTR | c.2308_2309insGCACCCGGG | OMINDEL1071 |
| EGFR | 20 | p.V769_D770insGTV | c.2308_2309insGCACCGTGG | OMINDEL1060 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.V769_D770insGSV | c.2308_2309insGCAGCGTGG | COSM18429 |
| EGFR | 20 | p.V769_D770insGQ | c.2308_2309insGCCAGG | OMINDEL1070 |
| EGFR | 20 | p.V769_D770insGE | c.2308_2309insGCGAGG | OMINDEL1069 |
| EGFR | 20 | p.V769_D770insGGTASV | c.2308_2309insGCGGGACGGCCAGCGTGG | OMINDEL1052 |
| EGFR | 20 | p.V769_D770insGSV | c.2308_2309insGGAGCGTGG | COSM1235344 |
| EGFR | 20 | p.V769_D770insGG | c.2308_2309insGGGGGG | MATNV07 |
| EGFR | 20 | p.V769_D770insGVV | c.2308_2309insGGGTCGTGG | COSM18430 |
| EGFR | 20 | p.V769_D770insGVASV | c.2308_2309insGGGTGGCCAGCGTGG | OMINDEL1800 |
| EGFR | 20 | p.D770delinsGY | c.2308_2309insGTT | COSM12427 |
| EGFR | 20 | p.V769dup | c.2308_2309insTGG | COSM6506514 |
| EGFR | 20 | p.V769_D770insCV | c.2308_2310delGACinsTGCGTGGAT | OMINDEL1808 |
| EGFR | 20 | p.D770_N771delinsHNPH | c.2308_2311delGACAinsCACAACCCCC | OMINDEL1079<br>**Note:** The nucleotide change of OMINDEL1079 overlaps that of COSM12380, so a positive OMINDEL1079 sample also results in a positive call for COSM12380. |
| EGFR | 20 | p.D770delinsAS | c.2309_2309delAinsCCAG | OMINDEL1810 |
| EGFR | 20 | p.A767_V769dup | c.2309_2310delACinsCCAGCGTGGAT | COSM13558 |
| EGFR | 20 | p.D770delinsGV | c.2309_2310delACinsGGGTT | D770delinsGV |
| EGFR | 20 | p.V769_D770insERG | c.2309_2310insGCGTGGAGA | COSM1651742 |
| EGFR | 20 | p.D770delinsEG | c.2309_2310insGGG | OMINDEL1092 |
| EGFR | 20 | p.D770delinsEF | c.2309_2310insGTT | OMINDEL1091 |
| EGFR | 20 | p.D770_N771insH | c.2309_2310insTCA | OMINDEL1807 |
| EGFR | 20 | p.[D770G;N771H] | c.2309_2311delACAinsGCC | MATNV09 |
| EGFR | 20 | p.D770_N771delinsAGG | c.2309_2312delACAAinsCTGGTGG | COSM12737 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.D770_N771delinsGGT | c.2309_2312delACAAinsGCGGCAC | OMINDEL1089 |
| EGFR | 20 | p.D770_N771delinsGGT | c.2309_2312delACAAinsGGGGCAC | OMINDEL1090 |
| EGFR | 20 | p.D770_P772delinsASVDNR | c.2309_2315delACAACCCinsCCAGCGTGGACAACCG | OMINDEL1801 |
| EGFR | 20 | p.D770_N771insH | c.2310_2311insCAC | OMINDEL1081 |
| EGFR | 20 | p.D770_N771insQRG | c.2310_2311insCAGCGTGGC | COSM4970107 |
| EGFR | 20 | p.D770_N771insP | c.2310_2311insCCC | OMINDEL1096 |
| EGFR | 20 | p.D770dup | c.2310_2311insGAC | OMINDEL1806 |
| EGFR | 20 | p.D770_N771insAPW | c.2310_2311insGCACCGTGG | COSM20886 |
| EGFR | 20 | p.D770_N771insARV | c.2310_2311insGCCCGCGTC | OMINDEL1093 |
| EGFR | 20 | p.D770_N771insARV | c.2310_2311insGCCCGGGTC | OMINDEL1094 |
| EGFR | 20 | p.D770_N771insGD | c.2310_2311insGGAGAC | OMINDEL1804 |
| EGFR | 20 | p.D770_N771insG | c.2310_2311insGGC | COSM13004 |
| EGFR | 20 | p.D770_N771insGNPH | c.2310_2311insGGCAACCCGCAC | D770_N771insGNPH |
| EGFR | 20 | p.D770_N771insGT | c.2310_2311insGGCACA | COSM1238029 |
| EGFR | 20 | p.D770_N771insGTD | c.2310_2311insGGCACCGAC | OMINDEL1074 |
| EGFR | 20 | p.D770_N771insGTD | c.2310_2311insGGCACGGAC | OMINDEL1068 |
| EGFR | 20 | p.D770_N771insGD | c.2310_2311insGGCGAC | COSM22955 |
| EGFR | 20 | p.D770_N771insG | c.2310_2311insGGG | MATNV05 |
| EGFR | 20 | p.D770_N771insGL | c.2310_2311insGGGCTT | OMINDEL1182 |
| EGFR | 20 | p.D770_N771insGD | c.2310_2311insGGGGAC | COSM85795 |
| EGFR | 20 | p.D770_N771insGV | c.2310_2311insGGGGTC | OMINDEL1095 |
| EGFR | 20 | p.D770_N771insGL | c.2310_2311insGGGTTA | COSM48921 |
| EGFR | 20 | p.D770_N771insGF | c.2310_2311insGGGTTT | COSM655155 |
| EGFR | 20 | p.D770_N771insG | c.2310_2311insGGT | COSM12378 |
| EGFR | 20 | p.D770_N771insVDSVDNP | c.2310_2311insGTGGACAGCGTGGACAACCCC | OMINDEL953 |
| EGFR | 20 | p.D770_N771insY | c.2310_2311insTAC | COSM1238030 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.Y764_D770dup | c.2310_2311insTACGTGATGGCCAGCG TGGAC | COSM6962256 |
| EGFR | 20 | p.D770_N771insF | c.2310_2311insTTT | OMINDEL1812 |
| EGFR | 20 | p.D770_N771delinsERG | c.2310_2312delCAAinsGAGAGG | MATNV06 |
| EGFR | 20 | p.N771delinsHH | c.2311_2311delAinsCACC | OMINDEL1118 |
| EGFR | 20 | p.N771delinsPH | c.2311_2311delAinsCCCC | MATNV04 |
| EGFR | 20 | p.N771delinsRH | c.2311_2311delAinsCGCC | OMINDEL1121 |
| EGFR | 20 | p.N771delinsRD | c.2311_2311delAinsCGCG | OMINDEL1120 |
| EGFR | 20 | p.N771delinsGD | c.2311_2311delAinsGGCG | OMINDEL1119 |
| EGFR | 20 | p.N771delinsGY | c.2311_2311delAinsGGTT | COSM53189 |
| EGFR | 20 | p.N771delinsVH | c.2311_2311delAinsGTCC | COSM5023007 |
| EGFR | 20 | p.N771delinsCH | c.2311_2311delAinsTGTC | OMINDEL1814 |
| EGFR | 20 | p.N771delinsGF | c.2311_2312delAAinsGGGTT | COSM18431 |
| EGFR | 20 | p.N771delinsTH | c.2311_2312insCAC | COSM22946 |
| EGFR | 20 | p.D770_N771insT | c.2311_2312insCCA | COSM5023008 |
| EGFR | 20 | p.N771delinsRH | c.2311_2312insGAC | OMINDEL1129 |
| EGFR | 20 | p.N771delinsRD | c.2311_2312insGAG | OMINDEL1127 |
| EGFR | 20 | p.N771delinsSTH | c.2311_2312insGCACCC | COSM6920147 |
| EGFR | 20 | p.N771delinsSQRGH | c.2311_2312insGCCAGCGTGGAC | OMINDEL1815 |
| EGFR | 20 | p.N771delinsSPH | c.2311_2312insGCCCCC | OMINDEL1131 |
| EGFR | 20 | p.D770_N771insSVE | c.2311_2312insGCGTCGAAA | COSM1651743 |
| EGFR | 20 | p.S768_D770dup | c.2311_2312insGCGTGGACA | COSM13428 |
| EGFR | 20 | p.D770_N771insSVE | c.2311_2312insGCGTGGAGA | D770_N771insSVE |
| EGFR | 20 | p.N771delinsRH | c.2311_2312insGGC | OMINDEL1130 |
| EGFR | 20 | p.N771delinsRD | c.2311_2312insGGG | OMINDEL1128 |
| EGFR | 20 | p.N771delinsSH | c.2311_2312insGTC | COSM24434 |
| EGFR | 20 | p.N771delinsSGH | c.2311_2312insGTGGCC | COSM1651744 |
| EGFR | 20 | p.D770_N771insMATP | c.2311_2312insTGGCCACCCCCA | COSM26719 |
| EGFR | 20 | p.N771delinsGC | c.2311_2313delAACinsGGTTGT | OMINDEL904 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.N771_P772delinsHHR | c.2311_2315delAACCCinsCACCACCG | OMINDEL1816 |
| EGFR | 20 | p.N771delinsSVDS | c.2312_2312delAinsGCGTGGACAG | OMINDEL1140 |
| EGFR | 20 | p.N771delinsKH | c.2312_2313insACA | OMINDEL1123 |
| EGFR | 20 | p.D770_N771insKH | c.2312_2313insACACAA | D770_N771insKH_1 |
| EGFR | 20 | p.N771delinsKPP | c.2312_2313insACCCCC | OMINDEL955 |
| EGFR | 20 | p.N771delinsKPPH | c.2312_2313insACCCCCCCA | OMINDEL1125 |
| EGFR | 20 | p.N771delinsKL | c.2312_2313insACT | COSM6438147 |
| EGFR | 20 | p.N771delinsKG | c.2312_2313insAGG | OMINDEL1141 |
| EGFR | 20 | p.N771delinsKH | c.2312_2313insGCA | OMINDEL1124 |
| EGFR | 20 | p.D770_N771insKH | c.2312_2313insGCACAA | D770_N771insKH_2 |
| EGFR | 20 | p.N771delinsKPPH | c.2312_2313insGCCCCCCCA | OMINDEL1126 |
| EGFR | 20 | p.D770_N771insKD | c.2312_2313insGGACAA | OMINDEL1067 |
| EGFR | 20 | p.N771delinsKG | c.2312_2313insGGG | OMINDEL1142 |
| EGFR | 20 | p.N771_P772delinsSVDNR | c.2312_2315delACCCinsGCGTGGACAACCG | COSM13554 |
| EGFR | 20 | p.N771T | c.2312A>C | OM3174 |
| EGFR | 20 | p.N771delinsKG | c.2313_2313delCinsGGGG | OMINDEL1147 |
| EGFR | 20 | p.N771dup | c.2313_2314insAAC | COSM13003 |
| EGFR | 20 | p.N771_P772insNN | c.2313_2314insAACAAC | OMINDEL1085 |
| EGFR | 20 | p.N771_P772insT | c.2313_2314insACA | OMINDEL1148 |
| EGFR | 20 | p.N771_P772insT | c.2313_2314insACC | OMINDEL1138 |
| EGFR | 20 | p.N771_P772insTHN | c.2313_2314insACCCACAAC | OMINDEL1086 |
| EGFR | 20 | p.S768_N771dup | c.2313_2314insAGCGTGGACAAC | S768_N771dup |
| EGFR | 20 | p.D770_N771dup | c.2313_2314insGACAAC | OMINDEL1805 |
| EGFR | 20 | p.N771_P772insG | c.2313_2314insGGC | OMINDEL1143 |
| EGFR | 20 | p.N771_P772insV | c.2313_2314insGTC | COSM6922328 |
| EGFR | 20 | p.V769_N771dup | c.2313_2314insGTGGACAAC | COSM20885 |
| EGFR | 20 | p.N771_P772insL | c.2313_2314insTTG | OMINDEL1149 |
| EGFR | 20 | p.N771_P772insHN | c.2314_2315insACAACC | OMINDEL1084 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.N771_P772insH | c.2314_2315insACC | COSM1238031 |
| EGFR | 20 | p.N771_P772insHH | c.2314_2315insACCACC | COSM6931207 |
| EGFR | 20 | p.N771_P772insRH | c.2314_2315insGGCACC | COSM166390 |
| EGFR | 20 | p.N771_P772insL | c.2314_2315insTCC | OMINDEL1139 |
| EGFR | 20 | p.P772delinsHR | c.2315_2315delCinsACCG | OMINDEL1153 |
| EGFR | 20 | p.P772_H773insGDP | c.2315_2316insAGGCGACCC | OMINDEL954 |
| EGFR | 20 | p.P772_H773insTTP | c.2315_2316insGACAACCCC | OMINDEL1183 |
| EGFR | 20 | p.P772_H773insTHP | c.2315_2316insGACACACCC | COSM48923 |
| EGFR | 20 | p.P772_H773insTTP | c.2315_2316insGACCACCCC | OMINDEL1146 |
| EGFR | 20 | p.P772_H773insSNP | c.2315_2316insGAGCAACCC | OMINDEL1105 |
| EGFR | 20 | p.D770_P772dup | c.2315_2316insGGACAACCC | COSM6845099 |
| EGFR | 20 | p.P772_H773insANP | c.2315_2316insGGCCAACCC | OMINDEL1099 |
| EGFR | 20 | p.P772_H773insGNP | c.2315_2316insGGGCAACCC | OMINDEL1100 |
| EGFR | 20 | p.P772_H773insGHP | c.2315_2316insGGGCCACCC | OMINDEL1135 |
| EGFR | 20 | p.P772_H773insGDP | c.2315_2316insGGGCGACCC | OMINDEL1133 |
| EGFR | 20 | p.P772_H773insYNP | c.2315_2316insGTACAACCC | OMINDEL1083 |
| EGFR | 20 | p.P772_H773insSNP | c.2315_2316insGTCCAACCC | OMINDEL1104 |
| EGFR | 20 | p.N771_H773dup | c.2315_2316insTCACAACCC | OMINDEL1809 |
| EGFR | 20 | p.P772_H773insTP | c.2316_2316delCinsAACCCCT | COSM12388 |
| EGFR | 20 | p.N771_H773dup | c.2316_2316delCinsTCACAACCCT | OMINDEL1811 |
| EGFR | 20 | p.H773_V774insPHPH | c.2316_2316delCinsTCACCCTCACCCT | H773_V774insPHPH |
| EGFR | 20 | p.N771_P772dup | c.2316_2317insAACCCC | MAN123 |
| EGFR | 20 | p.P772_H773insNPNP | c.2316_2317insAACCCCAACCCC | OMINDEL1101 |
| EGFR | 20 | p.P772_H773insNPNP | c.2316_2317insAACCCGAACCCC | OMINDEL1113 |
| EGFR | 20 | p.P772_H773insTPNP | c.2316_2317insACACCCAACCCC | COSM6977296 |
| EGFR | 20 | p.P772_H773insT | c.2316_2317insACC | OMINDEL1152 |
| EGFR | 20 | p.P772_H773insTTP | c.2316_2317insACCACCCCC | OMINDEL1145 |
| EGFR | 20 | p.P772_H773insTRNP | c.2316_2317insACCAGGAACCCC | OMINDEL1116 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.P772_H773insTPNP | c.2316_2317insACCCCCAACCCC | OMINDEL1107 |
| EGFR | 20 | p.P772_H773insTPNP | c.2316_2317insACCCCGAACCCC | OMINDEL1114 |
| EGFR | 20 | p.P772_H773insTRNP | c.2316_2317insACCCGCAACCCC | OMINDEL1108 |
| EGFR | 20 | p.P772_H773insTRNP | c.2316_2317insACCCGGAACCCC | OMINDEL1115 |
| EGFR | 20 | p.P772_H773insTPNP | c.2316_2317insACGCCCAACCCC | OMINDEL1106 |
| EGFR | 20 | p.P772_H773insTRNP | c.2316_2317insACGCGCAACCCC | OMINDEL1109 |
| EGFR | 20 | p.P772_H773insSNP | c.2316_2317insAGCAACCCC | OMINDEL1103 |
| EGFR | 20 | p.S768_P772dup | c.2316_2317insAGCGTGGACAACCCC | OMINDEL1053 |
| EGFR | 20 | p.D770_P772dup | c.2316_2317insGACAACCCC | COSM1651745 |
| EGFR | 20 | p.P772_H773insANP | c.2316_2317insGCCAACCCC | OMINDEL1098 |
| EGFR | 20 | p.P772_H773insGNP | c.2316_2317insGGAAACCCC | OMINDEL1111 |
| EGFR | 20 | p.P772_H773insGNP | c.2316_2317insGGCAACCCC | OMINDEL1088 |
| EGFR | 20 | p.P772_H773insGT | c.2316_2317insGGCACC | MATNV02 |
| EGFR | 20 | p.P772_H773insGHP | c.2316_2317insGGCCACCCC | OMINDEL1134 |
| EGFR | 20 | p.P772_H773insGDP | c.2316_2317insGGCGACCCC | OMINDEL1132 |
| EGFR | 20 | p.V769_P772dup | c.2316_2317insGTGGACAACCCC | OMINDEL1057 |
| EGFR | 20 | p.P772_H773insV | c.2316_2317insGTT | COSM255205 |
| EGFR | 20 | p.P772_H773insYNP | c.2316_2317insTACAACCCC | OMINDEL1082 |
| EGFR | 20 | p.P772_H773insYPNP | c.2316_2317insTACCCCAACCCC | OMINDEL1110 |
| EGFR | 20 | p.P772_H773insYPNP | c.2316_2317insTACCCGAACCCC | OMINDEL1117 |
| EGFR | 20 | p.P772_H773insSNP | c.2316_2317insTCCAACCCC | OMINDEL1102 |
| EGFR | 20 | p.H773delinsNPY | c.2317_2317delCinsAACCCCT | OMINDEL1161 |
| EGFR | 20 | p.H773delinsYNPY | c.2317_2317delCinsTACAACCCCT | OMINDEL1160 |
| EGFR | 20 | p.H773delinsYPNPY | c.2317_2317delCinsTACCCCAACCCCT | OMINDEL1162 |
| EGFR | 20 | p.H773delinsYPNPY | c.2317_2317delCinsTACCCGAACCCCT | OMINDEL1163 |
| EGFR | 20 | p.P772_H773insPNP | c.2317_2318insCCAACCCCC | OMINDEL1087 |
| EGFR | 20 | p.H773delinsPNPY | c.2317_2318insCCAACCCCT | OMINDEL1165 |
| EGFR | 20 | p.P772_H773insPHP | c.2317_2318insCCCACCCCC | OMINDEL1136 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.H773delinsPNPY | c.2317_2318insCTAACCCCT | COSM1735761 |
| EGFR | 20 | p.P772_H773insR | c.2317_2318insGCC | OMINDEL1156 |
| EGFR | 20 | p.P772_H773insRHP | c.2317_2318insGCCACCCCC | OMINDEL1137 |
| EGFR | 20 | p.P772_H773insRVDNP | c.2317_2318insGCGTGGACAACCCCC | OMINDEL1056 |
| EGFR | 20 | p.P772_H773insRNP | c.2317_2318insGGAACCCCC | OMINDEL1813 |
| EGFR | 20 | p.H773delinsRY | c.2317_2318insGTT | MATNV08 |
| EGFR | 20 | p.P772_H773insL | c.2317_2318insTCC | OMINDEL1155 |
| EGFR | 20 | p.P772_H773insQ | c.2318_2319insACA | OMINDEL1158 |
| EGFR | 20 | p.H773delinsQY | c.2318_2319insATA | OMINDEL1166 |
| EGFR | 20 | p.P772_H773insQ | c.2318_2319insGCA | OMINDEL1159 |
| EGFR | 20 | p.H773delinsQY | c.2318_2319insGTA | OMINDEL1167 |
| EGFR | 20 | p.[H773L;V774M] | c.2318_2320delACGinsTCA | COSM6978402 |
| EGFR | 20 | p.H773_V774insNH | c.2319_2320insAACCAC | COSM5023006 |
| EGFR | 20 | p.N771_H773dup | c.2319_2320insAACCCCCAC | COSM12381 |
| EGFR | 20 | p.H773_V774insTQPP | c.2319_2320insACACAACCCCCC | COSM3727813 |
| EGFR | 20 | p.H773_V774insTH | c.2319_2320insACCCAC | OMINDEL1151 |
| EGFR | 20 | p.H773dup | c.2319_2320insCAC | COSM12377 |
| EGFR | 20 | p.H773_V774insHPH | c.2319_2320insCACCCCCAC | OMINDEL1122 |
| EGFR | 20 | p.H773_V774insHV | c.2319_2320insCACGTC | OMINDEL1169 |
| EGFR | 20 | p.H773_V774insQ | c.2319_2320insCAG | COSM131552 |
| EGFR | 20 | p.P772_H773dup | c.2319_2320insCCACAC | OMINDEL1184 |
| EGFR | 20 | p.P772_H773dup | c.2319_2320insCCCCAC | COSM12380<br>**Note:** The nucleotide change of COSM12380 overlaps that of MAN123, so a positive COSM12380 sample also results in a positive call for MAN123. |
| EGFR | 20 | p.H773_V774insPHPH | c.2319_2320insCCCCACCCCCAC | p.H773_V774insPHPH_2 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.H773_V774insY | c.2319_2320insTAC | OMINDEL1168 |
| EGFR | 20 | p.H773_V774insAH | c.2320_2321insCCCACG | COSM1238028 |
| EGFR | 20 | p.H773_V774insAPH | c.2320_2321insCCCCCCACG | OMINDEL1144 |
| EGFR | 20 | p.H773_V774insAPH | c.2320_2321insCGCCCCACG | OMINDEL1150 |
| EGFR | 20 | p.H773_V774insGNPH | c.2320_2321insGCAACCCCCACG | COSM51544 |
| EGFR | 20 | p.H773_V774insGH | c.2320_2321insGCCACG | OMINDEL1154 |
| EGFR | 20 | p.H773_V774insGH | c.2320_2321insGGCACG | OMINDEL1157 |
| EGFR | 20 | p.N771_V774dup | c.2321_2322insCAACCCCCACGT | OMINDEL1097 |
| EGFR | 20 | p.H773_V774dup | c.2321_2322insCCACGT | COSM12389 |
| EGFR | 20 | p.N771_V774dup | c.2322_2323insAACCCCCACGTG | OMINDEL1112 |
| EGFR | 20 | p.H773_V774dup | c.2322_2323insCACGTG | COSM22948 |
| EGFR | 20 | p.V774_C775insPR | c.2322_2323insCCACGT | COSM4170223 |
| EGFR | 20 | p.P772_V774dup | c.2322_2323insCCCCACGTG | COSM6845098 |
| EGFR | 20 | p.C775Y | c.2324G>A | COSM22954 |
| EGFR | 20 | p.P772_C775dup | c.2326_2327insCCCACGTGTGCC | OMINDEL956 |
| EGFR | 20 | p.C775_R776insPYVC | c.2326_2327insCCTACGTGTGCC | OMINDEL1164 |
| EGFR | 20 | p.T790A | c.2368A>G | COSM1272071 |
| EGFR | 20 | p.T790M | c.2369C>T | COSM6240 |
| EGFR | 20 | p.L792Y | c.2374_2375delCTinsTA | OM3266 |
| EGFR | 20 | p.L792V | c.2374C>G | OM3265 |
| EGFR | 20 | p.L792F | c.2374C>T | COSM6493933 |
| EGFR | 20 | p.L792H | c.2375T>A | COSM6493934 |
| EGFR | 20 | p.L792P | c.2375T>C | COSM13190 |
| EGFR | 20 | p.L792R | c.2375T>G | OM3267 |
| EGFR | 20 | p.G796S | c.2386G>A | COSM20891 |
| EGFR | 20 | p.G796R | c.2386G>C | COSM6493935 |
| EGFR | 20 | p.G796C | c.2386G>T | OM3318 |
| EGFR | 20 | p.G796A | c.2387G>C | COSM27568 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| EGFR | 20 | p.C797N | c.2389_2390delTGinsAA | OM3543 |
| EGFR | 20 | p.C797S | c.2389T>A | COSM6493937 |
| EGFR | 20 | p.C797G | c.2389T>G | COSM6493936 |
| EGFR | 20 | p.C797S | c.2390G>C | COSM5945664 |
| EGFR | 20 | p.L798I | c.2392C>A | OM3268 |
| EGFR | 21 | p.L833V | c.2497T>G | COSM13424 |
| EGFR | 21 | p.L833W | c.2498T>G | COSM133204 |
| EGFR | 21 | p.L833F | c.2499G>T | COSM219794 |
| EGFR | 21 | p.H835L | c.2504A>T | COSM6227 |
| EGFR | 21 | p.R836C | c.2506C>T | COSM28604 |
| EGFR | 21 | p.P848L | c.2543C>T | COSM22943 |
| EGFR | 21 | p.T854A | c.2560A>G | COSM28537 |
| EGFR | 21 | p.T854S | c.2560A>T | COSM1235378 |
| EGFR | 21 | p.T854I | c.2561C>T | COSM87246 |
| EGFR | 21 | p.L858K | c.2572_2573delCTinsAA | COSM24268 |
| EGFR | 21 | p.L858R | c.2572_2573delCTinsAG | COSM13553 |
| EGFR | 21 | p.L858M | c.2572C>A | COSM12366 |
| EGFR | 21 | p.L858R | c.2573_2574delTGinsGA | COSM133630 |
| EGFR | 21 | p.L858R | c.2573_2574delTGinsGT | COSM12429 |
| EGFR | 21 | p.L858Q | c.2573T>A | COSM29578 |
| EGFR | 21 | p.L858R | c.2573T>G | COSM6224 |
| EGFR | 21 | p.L861V | c.2581C>G | COSM24848 |
| EGFR | 21 | p.L861Q | c.2582T>A | COSM6213 |
| EGFR | 21 | p.L861R | c.2582T>G | COSM12374 |
| ERBB2 | 8 | p.G309E | c.926G>A | COSM94224 |
| ERBB2 | 8 | p.G309A | c.926G>C | COSM436496 |
| ERBB2 | 8 | p.S310P | c.928T>C | COSM1563863 |
| ERBB2 | 8 | p.S310Y | c.929_930delCCinsAT | COSM6906440 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ERBB2 | 8 | p.S310Y | c.929C>A | COSM94225 |
| ERBB2 | 8 | p.S310F | c.929C>T | COSM48358 |
| ERBB2 | 8 | p.C311R | c.931T>C | COSM35763 |
| ERBB2 | 8 | p.E321G | c.962A>G | COSM35764 |
| ERBB2 | 17 | p.V659E | c.1976_1977delTTinsAA | COSM3724566 |
| ERBB2 | 17 | p.V659E | c.1976_1977delTTinsAG | COSM6503262 |
| ERBB2 | 17 | p.G660D | c.1979G>A | COSM4681497 |
| ERBB2 | 17 | p.R678Q | c.2033G>A | COSM436498 |
| ERBB2 | 18 | p.T733I | c.2198C>T | COSM14059 |
| ERBB2 | 19 | p.L755_T759del | c.2262_2276delGTTGAGGGAAAACAC | COSM7345568 |
| ERBB2 | 19 | p.L755P | c.2263_2264delTTinsCC | COSM683 |
| ERBB2 | 19 | p.L755A | c.2263_2264delTTinsGC | COSM6906940 |
| ERBB2 | 19 | p.L755M | c.2263T>A | COSM1205571 |
| ERBB2 | 19 | p.L755_T759del | c.2264_2278delTGAGGGAAAACACAT | COSM5029269 |
| ERBB2 | 19 | p.L755S | c.2264T>C | COSM14060 |
| ERBB2 | 19 | p.L755W | c.2264T>G | COSM436499 |
| ERBB2 | 19 | p.L755F | c.2265G>C | OBENV02 |
| ERBB2 | 19 | p.L755F | c.2265G>T | OBENV01 |
| ERBB2 | 19 | p.I767M | c.2301C>G | COSM51317 |
| ERBB2 | 19 | p.D769N | c.2305G>A | COSM1302747 |
| ERBB2 | 19 | p.D769H | c.2305G>C | COSM13170 |
| ERBB2 | 19 | p.D769Y | c.2305G>T | COSM1251412 |
| ERBB2 | 20 | p.E770_A771insGIRD | c.2311_2312insGCATACGTGATG | COSM7449692 |
| ERBB2 | 20 | p.Y772_V773insMMAY | c.2316_2317insATGATGGCTTAC | OMINDEL820 |
| ERBB2 | 20 | p.M774delinsWLV | c.2320_2322delATGinsTGGCTTGTA | OMINDEL1178 |
| ERBB2 | 20 | p.M774dup | c.2322_2323insATG | OMINDEL823 |
| ERBB2 | 20 | p.Y772_A775dup | c.2324_2325insATACGTGATGGC | COSM20959 |
| ERBB2 | 20 | p.A775_G776insYVMS | c.2324_2325insATACGTGATGTC | OMINDEL824 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ERBB2 | 20 | p.A775_G776insTVMA | c.2325_2326insACCGTGATGGCT | MAN302 |
| ERBB2 | 20 | p.Y772_A775dup | c.2325_2326insTACGTGATGGCT | COSM12558 |
| ERBB2 | 20 | p.A775_G776insSVMA | c.2325_2326insTCCGTGATGGCT | OMINDEL821 |
| ERBB2 | 20 | p.A775_G776insC | c.2325_2326insTGT | OMINDEL825 |
| ERBB2 | 20 | p.G776delinsIC | c.2326_2326delGinsATAT | OMINDEL826 |
| ERBB2 | 20 | p.G776delinsLC | c.2326_2326delGinsCTTT | COSM12554 |
| ERBB2 | 20 | p.G776delinsLC | c.2326_2326delGinsTTAT | COSM20895 |
| ERBB2 | 20 | p.G776delinsLC | c.2326_2326delGinsTTGT | COSM19875 |
| ERBB2 | 20 | p.G776L | c.2326_2327delGGinsCT | COSM144255 |
| ERBB2 | 20 | p.A775_G776insAVMA | c.2326_2327insCCGTGATGGCTG | OMINDEL816 |
| ERBB2 | 20 | p.G776delinsAVGC | c.2326_2327insCTGTGGGCT | COSM6907033 |
| ERBB2 | 20 | p.A775_G776insV | c.2326_2327insTAG | MAN309 |
| ERBB2 | 20 | p.G776delinsVC | c.2326_2327insTAT | COSM5235349 |
| ERBB2 | 20 | p.A775_G776insV | c.2326_2327insTCG | MAN308 |
| ERBB2 | 20 | p.G776delinsVC | c.2326_2327insTCT | COSM85995 |
| ERBB2 | 20 | p.A775_G776insV | c.2326_2327insTGG | MAN300 |
| ERBB2 | 20 | p.A775_G776insVA | c.2326_2327insTGGCTG | COSM6865891 |
| ERBB2 | 20 | p.A775_G776insVAG | c.2326_2327insTGGCTGGTG | OMINDEL817 |
| ERBB2 | 20 | p.A775_G776insVVMA | c.2326_2327insTGGTGATGGCTG | OMINDEL822 |
| ERBB2 | 20 | p.G776delinsVC | c.2326_2327insTGT | COSM12553 |
| ERBB2 | 20 | p.A775_G776insV | c.2326_2327insTTG | MAN307 |
| ERBB2 | 20 | p.G776delinsVC | c.2326_2327insTTT | COSM12552 |
| ERBB2 | 20 | p.G776_V777delinsSVA | c.2326_2330delGGTGTinsAGTGTGGC | G776_V777delinsSVA_3 |
| ERBB2 | 20 | p.G776S | c.2326G>A | COSM685 |
| ERBB2 | 20 | p.G776R | c.2326G>C | OM3421 |
| ERBB2 | 20 | p.G776C | c.2326G>T | COSM303938 |
| ERBB2 | 20 | p.G776delinsVV | c.2327_2327delGinsTTGT | COSM6977433 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ERBB2 | 20 | p.G776_G778delinsVVGD | c.2327_2334delGTGTGGGCinsTTGTTGGGGAT | OMINDEL1189 |
| ERBB2 | 20 | p.G776A | c.2327G>C | OBENV03 |
| ERBB2 | 20 | p.G776V | c.2327G>T | COSM18609 |
| ERBB2 | 20 | p.G776_V777insL | c.2328_2329insCTT | COSM6438151 |
| ERBB2 | 20 | p.V777M | c.2329G>A | COSM14064 |
| ERBB2 | 20 | p.V777L | c.2329G>C | COSM436500 |
| ERBB2 | 20 | p.V777L | c.2329G>T | COSM14062 |
| ERBB2 | 20 | p.G778_S779insCVG | c.2330_2331insAGGTTGTGT | OMINDEL1179 |
| ERBB2 | 20 | p.V777dup | c.2330_2331insTGT | COSM9869865 |
| ERBB2 | 20 | p.V777A | c.2330T>C | COSM35496 |
| ERBB2 | 20 | p.V777_G778insC | c.2331_2332insTGT | OMINDEL827 |
| ERBB2 | 20 | p.V777_G778insCG | c.2331_2332insTGTGGG | COSM303939 |
| ERBB2 | 20 | p.G778_S779insLPS | c.2333_2334insGCTCCCCAG | COSM5802314 |
| ERBB2 | 20 | p.G778_S779insLPG | c.2333_2334insGCTCCCCGG | COSM7449770 |
| ERBB2 | 20 | p.G778dup | c.2333_2334insGGG | COSM26681 |
| ERBB2 | 20 | p.G778_S779insCPG | c.2335_2336insGCCCAGGCT | OMINDEL1180 |
| ERBB2 | 20 | p.V777_S779dup | c.2336_2337insTGTGGGCTC | COSM681 |
| ERBB2 | 20 | p.G778_P780dup | c.2339_2340insCGGCTCCCC | COSM6865893 |
| ERBB2 | 20 | p.G778_P780dup | c.2339_2340insGGGCTCCCC | COSM12555 |
| ERBB2 | 20 | p.G778_P780dup | c.2339_2340insTGGCTCCCC | COSM303948 |
| ERBB2 | 20 | p.G778_P780dup | c.2340_2341insGGCTCCCCA | COSM12556 |
| ERBB2 | 20 | p.T798I | c.2393C>T | OM3210 |
| ERBB2 | 21 | p.V842I | c.2524G>A | COSM14065 |
| ERBB2 | 21 | p.V842E | c.2525T>A | COSM6005745 |
| ERBB2 | 21 | p.R868W | c.2602C>T | COSM289684 |
| ERBB2 | 21 | p.L869R | c.2606T>G | COSM249793 |
| ERBB2 | 22 | p.R896C | c.2686C>T | COSM14066 |
| ERBB2 | 22 | p.R896H | c.2687G>A | COSM119971 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ERBB3 | 2 | p.M60L | c.178A>T | COSM1606366 |
| ERBB3 | 2 | p.M60K | c.179T>A | COSM254678 |
| ERBB3 | 2 | p.M60R | c.179T>G | COSM941484 |
| ERBB3 | 3 | p.M91I | c.273G>A | COSM122890 |
| ERBB3 | 3 | p.M91I | c.273G>C | COSM1299636 |
| ERBB3 | 3 | p.R103G | c.307C>G | COSM6907312 |
| ERBB3 | 3 | p.V104L | c.310G>C | COSM160824 |
| ERBB3 | 3 | p.V104M | c.310G>A | COSM172423 |
| ERBB3 | 3 | p.V104L | c.310G>T | COSM191840 |
| ERBB3 | 7 | p.G284R | c.850G>C | COSM5413554 |
| ERBB3 | 7 | p.G284R | c.850G>A | COSM941488 |
| ERBB3 | 8 | p.D297Y | c.889G>T | COSM160822 |
| ERBB3 | 8 | p.D297H | c.889G>C | COSM1935392 |
| ERBB3 | 8 | p.D297N | c.889G>A | COSM941489 |
| ERBB3 | 8 | p.D297V | c.890A>T | COSM941490 |
| ERBB3 | 9 | p.E332K | c.994G>A | COSM254677 |
| ERBB3 | 9 | p.E332Q | c.994G>C | COSM4838033 |
| ERBB4 | 18 | p.R711C | c.2131C>T | COSM160827 |
| ERBB4 | 18 | p.R711L | c.2132G>T | OM3150 |
| ESR1 | 5 | p.K303R | c.908A>G | COSM4745827 |
| ESR1 | 6 | p.E380Q | c.1138G>C | COSM3829320 |
| ESR1 | 6 | p.V392I | c.1174G>A | COSM1545070 |
| ESR1 | 8 | p.S463P | c.1387T>C | COSM4771561 |
| ESR1 | 9 | p.P535T | c.1603C>A | COSM5666097 |
| ESR1 | 9 | p.P535H | c.1604C>A | BT24 |
| ESR1 | 9 | p.L536Q | c.1607_1608delTCinsAG | COSM4766050 |
| ESR1 | 9 | p.L536R | c.1607T>G | COSM4774826 |
| ESR1 | 9 | p.Y537N | c.1609T>A | COSM1074635 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| ESR1 | 9 | p.Y537C | c.1610A>G | COSM1074637 |
| ESR1 | 9 | p.Y537S | c.1610A>C | COSM1074639 |
| ESR1 | 9 | p.D538G | c.1613A>G | COSM94250 |
| ESR1 | 9 | p.R555C | c.1663C>T | BT32 |
| FGFR1 | 14 | p.V592M | c.1774G>A | COSM6840954 |
| FGFR1 | 15 | p.K687E | c.2059A>G | COSM35673 |
| FGFR1 | 15 | p.K687M | c.2060A>T | COSM4771556 |
| FGFR1 | 15 | p.K687N | c.2061G>C | COSM6578115 |
| FGFR2 | 7 | p.S252W | c.755C>G | COSM36903 |
| FGFR2 | 7 | p.P253S | c.757C>T | COSM1346291 |
| FGFR2 | 7 | p.P253R | c.758C>G | COSM49170 |
| FGFR2 | 7 | p.P253L | c.758C>T | COSM537801 |
| FGFR2 | 7 | p.K310R | c.929A>G | COSM36901 |
| FGFR2 | 8 | p.A314D | c.941C>A | COSM49171 |
| FGFR2 | 8 | p.A315T | c.943G>A | COSM30777 |
| FGFR2 | 9 | p.Y375H | c.1123T>C | COSM1560916 |
| FGFR2 | 9 | p.Y375C | c.1124A>G | COSM36904 |
| FGFR2 | 9 | p.C382R | c.1144T>C | COSM36906 |
| FGFR2 | 9 | p.C382G | c.1144T>G | COSM6197913 |
| FGFR2 | 9 | p.C382Y | c.1145G>A | COSM915493 |
| FGFR2 | 9 | p.M391R | c.1172T>G | COSM36907 |
| FGFR2 | 9 | p.V395D | c.1184_1185delTCinsAT | COSM250081 |
| FGFR2 | 12 | p.H544Q | c.1632C>G | COSM1474415 |
| FGFR2 | 12 | p.H544Q | c.1632C>A | COSM48382 |
| FGFR2 | 12 | p.I547V | c.1639A>G | COSM36913 |
| FGFR2 | 12 | p.N549H | c.1645A>C | COSM250083 |
| FGFR2 | 12 | p.N549S | c.1646A>G | COSM3665553 |
| FGFR2 | 12 | p.N549K | c.1647T>G | COSM36902 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| FGFR2 | 12 | p.N549K | c.1647T>A | COSM36912 |
| FGFR2 | 12 | p.N549D | c.1645A>G | COSM4604460 |
| FGFR2 | 12 | p.N549T | c.1646A>C | COSM6952624 |
| FGFR2 | 13 | p.V564I | c.1690G>A | OM3247 |
| FGFR2 | 13 | p.V564F | c.1690G>T | OM3293 |
| FGFR2 | 13 | p.V564L | c.1690G>C | OM3564 |
| FGFR2 | 13 | p.E565A | c.1694A>C | OM3292 |
| FGFR2 | 13 | p.E565G | c.1694A>G | OM3563 |
| FGFR2 | 13 | p.G613V | c.1838G>T | COSM94318 |
| FGFR2 | 13 | p.L617V | c.1849T>G | OM3291 |
| FGFR2 | 13 | p.L617M | c.1849T>A | OM3562 |
| FGFR2 | 14 | p.K641R | c.1922A>G | COSM5010388 |
| FGFR2 | 14 | p.A648T | c.1942G>A | COSM29836 |
| FGFR2 | 14 | p.K659E | c.1975A>G | COSM36909 |
| FGFR2 | 14 | p.K659N | c.1977G>T | COSM49173 |
| FGFR2 | 14 | p.K659M | c.1976A>T | COSM49175 |
| FGFR2 | 14 | p.K659N | c.1977G>C | COSM683054 |
| FGFR2 | 15 | p.V679I | c.2035G>A | COSM4822956 |
| FGFR3 | 7 | p.R248C | c.742C>T | COSM714 |
| FGFR3 | 7 | p.R248H | c.743G>A | COSM1757310 |
| FGFR3 | 7 | p.S249T | c.745T>A | COSM29431 |
| FGFR3 | 7 | p.S249C | c.746C>G | COSM715 |
| FGFR3 | 9 | p.G370C | c.1108G>T | COSM716 |
| FGFR3 | 9 | p.G370D | c.1109G>A | COSM5966955 |
| FGFR3 | 9 | p.S371C | c.1111A>T | COSM17461 |
| FGFR3 | 9 | p.Y373C | c.1118A>G | COSM718 |
| FGFR3 | 9 | p.G380R | c.1138G>A | COSM24842 |
| FGFR3 | 9 | p.G380E | c.1139G>A | COSM51545 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| FGFR3 | 9 | p.A391T | c.1171G>A | OM3151 |
| FGFR3 | 9 | p.A391V | c.1172C>T | COSM1737909 |
| FGFR3 | 9 | p.A391E | c.1172C>A | COSM721 |
| FGFR3 | 9 | p.R399C | c.1195C>T | COSM296687 |
| FGFR3 | 13 | p.V553M | c.1657G>A | OM3223 |
| FGFR3 | 13 | p.V555M | c.1663G>A | COSM303914 |
| FGFR3 | 14 | p.D641N | c.1921G>A | COSM29438 |
| FGFR3 | 14 | p.D641G | c.1922A>G | COSM1428721 |
| FGFR3 | 14 | p.K650E | c.1948A>G | COSM719 |
| FGFR3 | 14 | p.K650Q | c.1948A>C | COSM726 |
| FGFR3 | 14 | p.K650M | c.1949A>T | COSM720 |
| FGFR3 | 14 | p.K650T | c.1949A>C | COSM731 |
| FGFR3 | 14 | p.K650R | c.1949A>G | OM3224 |
| FGFR3 | 14 | p.K650N | c.1950G>T | COSM1428730 |
| FGFR3 | 14 | p.K650N | c.1950G>C | COSM3993568 |
| FGFR3 | 15 | p.V677I | c.2029G>A | COSM178247 |
| FGFR3 | 16 | p.K715M | c.2144A>T | COSM732992 |
| FGFR4 | 13 | p.V550L | c.1648G>T | COSM1066172 |
| FGFR4 | 13 | p.V550L | c.1648G>C | COSM3738141 |
| FGFR4 | 13 | p.V550M | c.1648G>A | COSM6163 |
| FGFR4 | 13 | p.V550E | c.1649T>A | COSM9215998 |
| FLT3 | 20 | p.D835F | c.2503_2504delGAinsTT | COSM19844 |
| FLT3 | 20 | p.D835_I836del | c.2503_2508delGATATC | COSM5879385 |
| FLT3 | 20 | p.D835Y | c.2503G>T | COSM783 |
| FLT3 | 20 | p.D835H | c.2503G>C | COSM785 |
| FLT3 | 20 | p.D835N | c.2503G>A | COSM789 |
| FLT3 | 20 | p.D835del | c.2503_2505delGAT | COSM854 |
| FLT3 | 20 | p.D835K | c.2503_2505delGATinsAAG | OM3570 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| FLT3 | 20 | p.D835K | c.2503_2505delGATinsAAA | OM3571 |
| FLT3 | 20 | p.D835L | c.2503_2504delGAinsCT | OM3572 |
| FLT3 | 20 | p.D835G | c.2504A>G | COSM1366188 |
| FLT3 | 20 | p.D835A | c.2504A>C | COSM27650 |
| FLT3 | 20 | p.D835_I836delinsV | c.2504_2506delATA | COSM27651 |
| FLT3 | 20 | p.D835V | c.2504A>T | COSM784 |
| FLT3 | 20 | p.D835E | c.2505T>A | COSM787 |
| FLT3 | 20 | p.D835E | c.2505T>G | COSM788 |
| FLT3 | 20 | p.I836L | c.2506A>C | COSM19649 |
| FLT3 | 20 | p.I836D | c.2506_2507delATinsGA | OM3569 |
| FLT3 | 20 | p.I836S | c.2507T>G | COSM19843 |
| FLT3 | 20 | p.I836T | c.2507T>C | COSM5879386 |
| FLT3 | 20 | p.I836del | c.2508_2510delCAT | COSM19836 |
| FLT3 | 20 | p.D839H | c.2515G>C | COSM3399304 |
| FLT3 | 20 | p.D839N | c.2515G>A | COSM6005768 |
| FLT3 | 20 | p.D839G | c.2516A>G | COSM1166729 |
| FLT3 | 20 | p.D839E | c.2517T>A | COSM5945287 |
| GNAS | 8 | p.R201G | c.601C>G | COSM1757313 |
| GNAS | 8 | p.R201C | c.601C>T | COSM27887 |
| GNAS | 8 | p.R201S | c.601C>A | COSM27899 |
| GNAS | 8 | p.R201H | c.602G>A | COSM27895 |
| GNAS | 8 | p.R201L | c.602G>T | COSM99221 |
| GNAS | 9 | p.Q227K | c.679C>A | COSM28618 |
| GNAS | 9 | p.Q227E | c.679C>G | COSM4962873 |
| GNAS | 9 | p.Q227L | c.680A>T | COSM27888 |
| GNAS | 9 | p.Q227R | c.680A>G | COSM27896 |
| GNAS | 9 | p.Q227H | c.681G>T | COSM27900 |
| GNAS | 9 | p.Q227H | c.681G>C | COSM4168960 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| HRAS | 2 | p.G12N | c.34_35delGGinsAA | COSM4517473 |
| HRAS | 2 | p.G12S | c.34G>A | COSM480 |
| HRAS | 2 | p.G12C | c.34G>T | COSM481 |
| HRAS | 2 | p.G12R | c.34G>C | COSM482 |
| HRAS | 2 | p.G12V | c.35G>T | COSM483 |
| HRAS | 2 | p.G12D | c.35G>A | COSM484 |
| HRAS | 2 | p.G12A | c.35G>C | COSM485 |
| HRAS | 2 | p.G13R | c.37G>C | COSM486 |
| HRAS | 2 | p.G13S | c.37G>A | COSM487 |
| HRAS | 2 | p.G13C | c.37G>T | COSM488 |
| HRAS | 2 | p.G13V | c.38G>T | COSM489 |
| HRAS | 2 | p.G13D | c.38G>A | COSM490 |
| HRAS | 3 | p.Q61K | c.181C>A | COSM496 |
| HRAS | 3 | p.Q61E | c.181C>G | COSM497 |
| HRAS | 3 | p.Q61R | c.181_182delCAinsAG | COSM52979 |
| HRAS | 3 | p.Q61R | c.182_183delAGinsGA | COSM33695 |
| HRAS | 3 | p.Q61L | c.182A>T | COSM498 |
| HRAS | 3 | p.Q61R | c.182A>G | COSM499 |
| HRAS | 3 | p.Q61P | c.182A>C | COSM500 |
| HRAS | 3 | p.Q61R | c.182_183delAGinsGT | COSM501 |
| HRAS | 3 | p.Q61L | c.182_183delAGinsTA | COSM52978 |
| HRAS | 3 | p.Q61H | c.183G>T | COSM502 |
| HRAS | 3 | p.Q61H | c.183G>C | COSM503 |
| IDH1 | 4 | p.G70D | c.209G>A | COSM96531 |
| IDH1 | 4 | p.R132S | c.394_395delCGinsTC | COSM1169216 |
| IDH1 | 4 | p.R132V | c.394_395delCGinsGT | COSM28751 |
| IDH1 | 4 | p.R132C | c.394C>T | COSM28747 |
| IDH1 | 4 | p.R132G | c.394C>G | COSM28749 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| IDH1 | 4 | p.R132S | c.394C>A | COSM28748 |
| IDH1 | 4 | p.R132H | c.395_396delGTinsAC | COSM86993 |
| IDH1 | 4 | p.R132H | c.395G>A | COSM28746 |
| IDH1 | 4 | p.R132L | c.395G>T | COSM28750 |
| IDH1 | 4 | p.R132Q | c.395_396delGTinsAG | OM3221 |
| IDH1 | 4 | p.R132Q | c.395_396delGTinsAA | OM3222 |
| IDH1 | 7 | p.S280F | c.839C>T | OM3315 |
| IDH2 | 4 | p.R140G | c.418C>G | COSM1737874 |
| IDH2 | 4 | p.R140W | c.418C>T | COSM41877 |
| IDH2 | 4 | p.R140L | c.419G>T | COSM41875 |
| IDH2 | 4 | p.R140Q | c.419G>A | COSM41590 |
| IDH2 | 4 | p.R172G | c.514A>G | COSM33731 |
| IDH2 | 4 | p.R172W | c.514A>T | COSM34039 |
| IDH2 | 4 | p.R172V | c.514_515delAGinsGT | OM3327 |
| IDH2 | 4 | p.R172T | c.515G>C | COSM1636997 |
| IDH2 | 4 | p.R172N | c.515_516delGGinsAT | COSM256046 |
| IDH2 | 4 | p.R172M | c.515G>T | COSM33732 |
| IDH2 | 4 | p.R172K | c.515G>A | COSM33733 |
| IDH2 | 4 | p.R172S | c.516G>C | COSM133672 |
| IDH2 | 4 | p.R172S | c.516G>T | COSM34090 |
| IDH2 | 7 | p.Q316E | c.946C>G | OM3320 |
| IDH2 | 7 | p.I319M | c.957C>G | OM3319 |
| KEAP1 | 2 | p.V155F | c.463G>T | COSM710187 |
| KEAP1 | 3 | p.V167F | c.499G>T | COSM710188 |
| KEAP1 | 3 | p.? | c.640-1G>A | COSM6972470 |
| KEAP1 | 3 | p.? | c.640-1G>T | OM3725 |
| KEAP1 | 3 | p.? | c.640-1G>C | OM3726 |
| KEAP1 | 3 | p.? | c.640-2A>C | OM3727 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KEAP1 | 3 | p.S243C | c.728C>G | COSM710191 |
| KEAP1 | 3 | p.R320W | c.958C>T | COSM1524062 |
| KEAP1 | 3 | p.R320L | c.959G>T | COSM2812614 |
| KEAP1 | 3 | p.R320P | c.959G>C | COSM346494 |
| KEAP1 | 3 | p.R320Q | c.959G>A | COSM710195 |
| KEAP1 | 3 | p.G333C | c.997G>T | COSM1193323 |
| KEAP1 | 3 | p.M409* | c.1224delC | COSM6916353 |
| KEAP1 | 3 | p.R415G | c.1243C>G | COSM6196677 |
| KEAP1 | 3 | p.D422N | c.1264G>A | COSM710198 |
| KEAP1 | 4 | p.R470S | c.1408C>A | COSM564846 |
| KEAP1 | 4 | p.R470C | c.1408C>T | COSM564847 |
| KEAP1 | 4 | p.R470G | c.1408C>G | OM3724 |
| KEAP1 | 4 | p.R470H | c.1409G>A | COSM1524064 |
| KEAP1 | 4 | p.? | c.1531+1G>T | COSM6956065 |
| KEAP1 | 4 | p.? | c.1531+2T>G | OM3723 |
| KEAP1 | 5 | p.? | c.1532-2A>G | COSM381459 |
| KEAP1 | 5 | p.? | c.1532-1G>T | COSM6969960 |
| KEAP1 | 5 | p.R554Q | c.1661G>A | COSM1524066 |
| KEAP1 | 5 | p.? | c.1708+2T>A | COSM6149754 |
| KEAP1 | 5 | p.? | c.1708_1708+1delinsTT | COSM6927565 |
| KEAP1 | 5 | p.? | c.1708+1G>T | COSM7738757 |
| KEAP1 | 6 | p.? | c.1709-2A>G | COSM7088100 |
| KEAP1 | 6 | p.? | c.1709-1G>T | COSM7724088 |
| KEAP1 | 6 | p.Y584* | c.1751_1752insA | COSM6201646 |
| KIT | 8 | p.T417_D419delinsRG | c.1250_1256delCTTACGAinsGAGG | COSM1149 |
| KIT | 8 | p.T417_D419delinsI | c.1250_1256delCTTACGAinsT | COSM1148 |
| KIT | 8 | p.D419del | c.1255_1257delGAC | COSM29014 |
| KIT | 8 | p.D419del | c.1255_1258delGACAinsC | COSM1600400 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 8 | p.D419_R420del | c.1255_1260delGACAGG | COSM1578132 |
| KIT | 8 | p.D419_L421delinsF | c.1255_1261delGACAGGCinsT | COSM1152 |
| KIT | 8 | p.D419_L421delinsH | c.1255_1262delGACAGGCTinsCA | COSM1578133 |
| KIT | 8 | p.D419_R420insFL | c.1257_1258insTTCCTC | COSM36332 |
| KIT | 8 | p.D419_R420insFF | c.1257_1258insTTTTTC | COSM36334 |
| KIT | 9 | p.S501_A502dup | c.1505_1506insTTCTGC | COSM5702501 |
| KIT | 9 | p.S501_A502dup | c.1507_1508insCTGCCT | COSM219786 |
| KIT | 9 | p.A502_Y503insFA | c.1507_1508insTTGCCT | COSM53306 |
| KIT | 9 | p.A502_Y503dup | c.1509_1510insGCCTAT | COSM1326 |
| KIT | 9 | p.N505I | c.1514A>T | OM3236 |
| KIT | 10 | p.F522C | c.1565T>G | OM3237 |
| KIT | 10 | p.V530I | c.1588G>A | COSM1155 |
| KIT | 11 | p.K550_P551del | c.1648_1653delAAACCC | COSM1160<br>**Note:** The nucleotide change of COSM1160 overlaps that of COSM1171, so a positive COSM1160 sample also results in a positive call for COSM1171. |
| KIT | 11 | p.K550_E554del | c.1648_1662delAAACCCATGTATGAA | COSM1162<br>**Note:** The nucleotide change of COSM1162 overlaps that of COSM6923694, so a positive COSM1162 sample also results in a positive call for COSM6923694. |
| KIT | 11 | p.K550_V555delinsQ | c.1648_1664delAAACCCATGTATGAAGT insCA | COSM6938892 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.K550_V555del | c.1648_1665delAAACCCATGTATGAAGTA | COSM1158<br>**Note:** The nucleotide change of COSM1158 overlaps that of COSM18827, so a positive COSM1158 sample also results in a positive call for COSM18827. |
| KIT | 11 | p.K550_K558del | c.1648-2_1672delAGAAACCCATGTATGAAGTACAGTGGA | COSM1163<br>**Note:** The nucleotide change of COSM1163 overlaps that of COSM1199, so a positive COSM1163 sample also results in a positive call for COSM1199. |
| KIT | 11 | c.1648-3_1671delCAGAAACCCATGTATGAAGTACAGTGG | c.1648-3_1671delCAGAAACCCATGTATGAAGTACAGTGG | OMINDEL901<br>**Note:** The nucleotide change of OMINDEL901 overlaps that of COSM36309 and COSM6008996, so a positive OMINDEL901 sample also results in positive calls for COSM36309 and COSM6008996. |
| KIT | 11 | p.K550_P551delinsI | c.1649_1653delAACCCinsTT | COSM6971267 |
| KIT | 11 | p.K550_E554delinsI | c.1649_1661delAACCCATGTATGAinsT | COSM6953101<br>**Note:** The nucleotide change of COSM6953101 overlaps that of COSM6944981, so a positive COSM6953101 sample also results in a positive call for COSM6944981. |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.K550_V555delinsI | c.1649_1663delAACCCATGTATGAAG | COSM1164<br>**Note:** The nucleotide change of COSM1164 overlaps that of COSM1185, so a positive COSM1164 sample also results in a positive call for COSM1185. |
| KIT | 11 | p.K550_Q556delinsII | c.1649_1668delAACCCATGTATGAAGTA CAGinsTTATT | COSM1161 |
| KIT | 11 | p.K550_W557delinsIR | c.1649_1669delAACCCATGTATGAAGTA CAGTinsTAC | COSM6977972<br>**Note:** The nucleotide change of COSM6977972 overlaps that of COSM1219, COSM133642, and COSM24748, so a positive COSM6977972 sample also results in positive calls for COSM1219, COSM133642, and COSM24748. |
| KIT | 11 | p.K550_W557delinsIL | c.1649_1670delAACCCATGTATGAAGTA CAGTGinsTTCT | COSM6936468 |
| KIT | 11 | p.K550R | c.1649A>G | COSM1166 |
| KIT | 11 | p.K550I | c.1649A>T | COSM1165 |
| KIT | 11 | p.K550N | c.1650A>C | COSM1167 |
| KIT | 11 | p.P551del | c.1651_1653delCCC | COSM1171 |
| KIT | 11 | p.P551_E554del | c.1651_1662delCCCATGTATGAA | COSM1172<br>**Note:** The nucleotide change of COSM1172 overlaps that of COSM36311, so a positive COSM1172 sample also results in a positive call for COSM36311. |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.P551_V555delinsTL | c.1651_1663delCCCATGTATGAAGinsACCC | COSM30550 |
| KIT | 11 | p.P551_Q556del | c.1651_1668delCCCATGTATGAAGTACAG | COSM1170<br>**Note:** The nucleotide change of COSM1170 overlaps that of COSM1169, COSM1171, COSM1187, and COSM1201, so a positive COSM1170 sample also results in positive calls for COSM1169, COSM1171, COSM1187, and COSM1201. |
| KIT | 11 | p.P551_M552delinsL | c.1652_1654delCCA | COSM1176 |
| KIT | 11 | p.P551_Y553delinsH | c.1652_1657delCCATGT | COSM6934157<br>**Note:** The nucleotide change of COSM6934157 overlaps that of COSM133763 and COSM96863, so a positive COSM6934157 sample also results in positive calls for COSM133763 and COSM96863. |
| KIT | 11 | p.P551_V555delinsL | c.1652_1663delCCATGTATGAAG | COSM1174 |
| KIT | 11 | p.P551_V555del | c.1652_1666delCCATGTATGAAGTAC | COSM1169 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.P551_V560delinsL | c.1652_1678delCCATGTATGAAGTACAGTGGAAGGTTG | COSM27901<br>**Note:** The nucleotide change of COSM27901 overlaps that of COSM1174 and COSM1230, so a positive COSM27901 sample also results in positive calls for COSM1174 and COSM1230. |
| KIT | 11 | p.M552_Y553del | c.1653_1658delCATGTA | COSM96881 |
| KIT | 11 | p.M552_V555del | c.1653_1664delCATGTATGAAGT | COSM6957862 |
| KIT | 11 | p.M552_Q556delinsL | c.1653_1667delCATGTATGAAGTACAinsTCT | COSM1178 |
| KIT | 11 | p.M552_W557del | c.1653_1670delCATGTATGAAGTACAGTG | COSM1177<br>**Note:** The nucleotide change of COSM1177 overlaps that of COSM133642, so a positive COSM1177 sample also results in a positive call for COSM133642. |
| KIT | 11 | p.M552_K558del | c.1653_1674delCATGTATGAAGTACAGTGGAAGinsT | OMINDEL902 |
| KIT | 11 | p.M552_Y553del | c.1654_1659delATGTAT | COSM1179 |
| KIT | 11 | p.M552_E554del | c.1654_1662delATGTATGAA | COSM1181 |
| KIT | 11 | p.M552_V555del | c.1654_1665delATGTATGAAGTA | COSM18827 |
| KIT | 11 | p.M552_Q556del | c.1654_1668delATGTATGAAGTACAG | COSM1327<br>**Note:** The nucleotide change of COSM1327 overlaps that of COSM1187, so a positive COSM1327 sample also results in a positive call for COSM1187. |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.M552_K558del | c.1654_1674delATGTATGAAGTACAGTGGAAG | COSM1182 |
| KIT | 11 | p.M552_Y553delinsN | c.1655_1657delTGT | COSM96863<br>**Note:** The nucleotide change of COSM96863 overlaps that of COSM133763, so a positive COSM96863 sample also results in a positive call for COSM133763. |
| KIT | 11 | p.M552_E554delinsK | c.1655_1660delTGTATG | COSM36311 |
| KIT | 11 | p.M552_Q556delinsK | c.1655_1666delTGTATGAAGTAC | COSM96868 |
| KIT | 11 | p.M552_W557delinsR | c.1655_1669delTGTATGAAGTACAGT | COSM133636<br>**Note:** The nucleotide change of COSM133636 overlaps that of COSM1216, COSM1220, and COSM24728, so a positive COSM133636 sample also results in positive calls for COSM1216, COSM1220, and COSM24728. |
| KIT | 11 | p.M552_W557del | c.1655_1672delTGTATGAAGTACAGTGGA | COSM1180<br>**Note:** The nucleotide change of COSM1180 overlaps that of COSM133700 and COSM36309, so a positive COSM1180 sample also results in positive calls for COSM13370 and COSM36309. |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.M552_E561delinsK | c.1655_1681delTGTATGAAGTACAGTGGAAGGTTGTTG | COSM96880<br>**Note:** The nucleotide change of COSM96880 overlaps that of COSM1206, so a positive COSM96880 sample also results in a positive call for COSM1206. |
| KIT | 11 | p.M552_E554delinsI | c.1656_1661delGTATGA | COSM6944981 |
| KIT | 11 | p.M552_V555delinsI | c.1656_1664delGTATGAAGT | COSM1185 |
| KIT | 11 | p.M552_V559delinsI | c.1656_1676delGTATGAAGTACAGTGGAAGGT | COSM3737059<br>**Note:** The nucleotide change of COSM3737059 overlaps that of COSM1226 and COSM1251, so a positive COSM3737059 sample also results in positive calls for COSM1226 and COSM1251. |
| KIT | 11 | p.Y553_E554del | c.1657_1662delTATGAA | COSM6923694 |
| KIT | 11 | p.Y553_W557del | c.1657_1671delTATGAAGTACAGTGG | COSM1187 |
| KIT | 11 | p.Y553_K558del | c.1657_1674delTATGAAGTACAGTGGAAG | COSM27909 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.Y553_V559del | c.1657_1677delTATGAAGTACAGTGGAAGGTT | COSM1191<br>**Note:** The nucleotide change of COSM1191 overlaps that of COSM11218, COSM1230, COSM1235, COSM1241, and COSM27909, so a positive COSM1191 sample also results in positive calls for COSM11218, COSM1230, COSM1235, COSM1241, and COSM27909. |
| KIT | 11 | p.Y553_V559delinsE | c.1657_1677delTATGAAGTACAGTGGAAGGTTinsGAA | COSM96946<br>**Note:** The nucleotide change of COSM96946 overlaps that of COSM1198 and COSM36315, so a positive COSM96946 sample also results in positive calls for COSM1198 and COSM36315. |
| KIT | 11 | p.Y553N | c.1657T>A | COSM133763 |
| KIT | 11 | p.Y553_Q556del | c.1658_1669delATGAAGTACAGT | COSM1189 |
| KIT | 11 | p.Y553C | c.1658A>G | COSM4413463 |
| KIT | 11 | p.E554_E562del | c.1660_1686delGAAGTACAGTGGAAGGTTGTTGAGGAG | COSM36310 |
| KIT | 11 | p.E554_K558del | c.1661_1675delAAGTACAGTGGAAGG | COSM1192 |
| KIT | 11 | p.E554_V559del | c.1661_1678delAAGTACAGTGGAAGGTTG | OMINDEL701 |
| KIT | 11 | p.E554_N564del | c.1661_1693delAAGTACAGTGGAAGGTTGTTGAGGAGATAAATG | COSM96947 |
| KIT | 11 | p.V555_E562del | c.1662_1685delAGTACAGTGGAAGGTTGTTGAGGA | COSM1194 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.V555_Q556del | c.1663_1668delGTACAG | COSM1201 |
| KIT | 11 | p.V555_I563del | c.1663_1689delGTACAGTGGAAGGTTGTTGAGGAGATA | COSM1196<br>**Note:** The nucleotide change of COSM1196 overlaps that of OMINDEL701, so a positive COSM1196 sample also results in a positive call for OMINDEL701. |
| KIT | 11 | p.V555_G565del | c.1663_1695delGTACAGTGGAAGGTTGTTGAGGAGATAAATGGA | COSM37027<br>**Note:** The nucleotide change of COSM37027 overlaps that of COSM6971633 and OMINDEL701, so a positive COSM37027 sample also results in positive calls for COSM6971633 and OMINDEL701. |
| KIT | 11 | p.V555_V560del | c.1664_1681delTACAGTGGAAGGTTGTTG | COSM1202<br>**Note:** The nucleotide change of COSM1202 overlaps that of COSM1198, COSM1204, COSM1223, COSM1234, COSM1248, and COSM1250, so a positive COSM1202 sample also results in positive calls for COSM1198, COSM1204, COSM1223, COSM1234, COSM1248, and COSM1250. |
| KIT | 11 | p.V555_N566delinsD | c.1664_1696delTACAGTGGAAGGTTGTTGAGGAGATAAATGGAA | COSM96870 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.W557_K558del | c.1665_1672delACAGTGGAinsGC | COSM6986076<br>**Note:** The nucleotide change of COSM6986076 overlaps that of COSM1217, so a positive COSM6986076 sample also results in a positive call for COSM1217. |
| KIT | 11 | p.Q556_V559del | c.1665_1676delACAGTGGAAGGT | COSM1199 |
| KIT | 11 | p.Q556_V560del | c.1665_1679delACAGTGGAAGGTTGT | COSM1198<br>**Note:** The nucleotide change of COSM1198 overlaps that of COSM1247 and COSM36315, so a positive COSM1198 sample also results in positive calls for COSM1247 and COSM36315. |
| KIT | 11 | p.Q556_W557del | c.1666_1671delCAGTGG | COSM36309 |
| KIT | 11 | p.Q556_K558del | c.1666_1674delCAGTGGAAG | COSM23418 |
| KIT | 11 | p.Q556_V559del | c.1666_1677delCAGTGGAAGGTT | COSM36315 |
| KIT | 11 | p.Q556_V560delinsF | c.1666_1678delCAGTGGAAGGTTGinsT | COSM1207 |
| KIT | 11 | p.Q556_V560del | c.1666_1680delCAGTGGAAGGTTGTT | COSM1204 |
| KIT | 11 | p.Q556_V560delinsTTF | c.1666_1680delCAGTGGAAGGTTGTT insACAACCTTC | COSM27904 |
| KIT | 11 | p.Q556_E561del | c.1666_1683delCAGTGGAAGGTTGTTG AG | COSM1206 |
| KIT | 11 | p.Q556_W557delinsR | c.1667_1669delAGT | COSM133642 |
| KIT | 11 | p.Q556_K558delinsR | c.1667_1673delAGTGGAAinsG | COSM28993 |
| KIT | 11 | p.Q556_V560delinsRG | c.1667_1680delAGTGGAAGGTTGTT insGTGGA | COSM6954545 |
| KIT | 11 | p.Q556_I563delinsL | c.1667_1687delAGTGGAAGGTTGTTGA GGAGA | COSM1651755 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.Q556_V559delinsH | c.1668_1676delGTGGAAGGT | COSM133641 |
| KIT | 11 | p.Q556_V559delinsHT | c.1668_1676delGTGGAAGGTinsTAC | COSM1214 |
| KIT | 11 | p.Q556_V560delinsH | c.1668_1679delGTGGAAGGTTGT | COSM1213 |
| KIT | 11 | p.W557_E561delinsPS | c.1668_1682delGTGGAAGGTTGTTGA insACCTTC | COSM6922994 |
| KIT | 11 | p.W557_E561delinsQ | c.1668_1683delGTGGAAGGTTGTTGAG insACAA | COSM6954755 |
| KIT | 11 | p.Q556_E561delinsHH | c.1668_1683delGTGGAAGGTTGTTGAG insTCAT | COSM1208<br>**Note:** The nucleotide change of COSM1208 overlaps that of COSM1263, so a positive COSM1208 sample also results in a positive call for COSM1263. |
| KIT | 11 | p.W557del | c.1669_1671delTGG | COSM1220 |
| KIT | 11 | p.W557_K558delinsQ | c.1669_1672delTGGAinsC | COSM6946777 |
| KIT | 11 | p.W557_K558delinsE | c.1669_1672delTGGAinsG | COSM24748 |
| KIT | 11 | p.W557_K558del | c.1669_1674delTGGAAG | COSM1217<br>**Note:** The nucleotide change of COSM1217 overlaps that of COSM23418, so a positive COSM1217 sample also results in a positive call for COSM23418. |
| KIT | 11 | p.W557_V559delinsI | c.1669_1675delTGGAAGGinsA | COSM1222 |
| KIT | 11 | p.W557_V559del | c.1669_1677delTGGAAGGTT | COSM1218 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.W557_V560del | c.1669_1680delTGGAAGGTTGTT | COSM1223<br>**Note:** The nucleotide change of COSM1223 overlaps that of COSM1234, so a positive COSM1223 sample also results in a positive call for COSM1234. |
| KIT | 11 | p.W557_E561del | c.1669_1683delTGGAAGGTTGTTGAG | COSM1332 |
| KIT | 11 | p.W557_E562del | c.1669_1686delTGGAAGGTTGTTGAGGAG | COSM36312 |
| KIT | 11 | p.W557R | c.1669T>A | COSM1216 |
| KIT | 11 | p.W557R | c.1669T>C | COSM1219 |
| KIT | 11 | p.W557G | c.1669T>G | COSM1221 |
| KIT | 11 | p.W557_K558delinsSS | c.1670_1673delGGAAinsCTTC | COSM96948 |
| KIT | 11 | p.W557_K558delinsFP | c.1670_1674delGGAAGinsTTCCT | COSM1224 |
| KIT | 11 | p.W557_V559delinsF | c.1670_1675delGGAAGG | COSM1226 |
| KIT | 11 | p.W557_V560delinsF | c.1670_1678delGGAAGGTTG | COSM1230 |
| KIT | 11 | p.W557S | c.1670G>C | COSM1227 |
| KIT | 11 | p.W557_K558delinsCT | c.1671_1673delGAAinsCAC | COSM28992 |
| KIT | 11 | p.W557_K558delinsCP | c.1671_1673delGAAinsTCC | COSM21978 |
| KIT | 11 | p.W557_K558delinsC | c.1671_1674delGAAGinsC | COSM96887 |
| KIT | 11 | p.W557_V559delinsC | c.1671_1676delGAAGGT | COSM1233 |
| KIT | 11 | p.W557_V560delinsC | c.1671_1679delGAAGGTTGT | COSM1232 |
| KIT | 11 | p.W557C | c.1671G>C | COSM133700 |
| KIT | 11 | p.W557C | c.1671G>T | COSM19108 |
| KIT | 11 | p.K558del | c.1672_1674delAAG | COSM6008996 |
| KIT | 11 | p.K558_V559delinsSS | c.1672_1676delAAGGTinsTCTTC | COSM28637 |
| KIT | 11 | p.K558_V559del | c.1672_1677delAAGGTT | COSM1235 |
| KIT | 11 | p.K558_V560del | c.1672_1680delAAGGTTGTT | COSM1234 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.K558_V560delinsL | c.1672_1680delAAGGTTGTTinsTTG | COSM6940807<br>**Note:** The nucleotide change of COSM6940807 overlaps that of COSM1235, so a positive COSM6940807 sample also results in a positive call for COSM1225. |
| KIT | 11 | p.K558_N564del | c.1672_1692delAAGGTTGTTGAGGAGA TAAAT | COSM1238 |
| KIT | 11 | p.K558delinsNP | c.1673_1674insTCC | COSM21976 |
| KIT | 11 | p.K558_V560delinsI | c.1673_1678delAGGTTG | COSM1241 |
| KIT | 11 | p.K558_V560delinsM | c.1673_1680delAGGTTGTTinsTG | COSM1242 |
| KIT | 11 | p.K558_E562del | c.1673_1687delAGGTTGTTGAGGAGA | COSM18896 |
| KIT | 11 | p.K558_G565delinsR | c.1673_1693delAGGTTGTTGAGGAGAT AAATG | COSM1240 |
| KIT | 11 | p.K558delinsNP | c.1674_1674delGinsTCCT | COSM1245 |
| KIT | 11 | p.K558_V559delinsN | c.1674_1676delGGT | COSM1244 |
| KIT | 11 | p.K558_V560delinsN | c.1674_1679delGGTTGT | COSM27069 |
| KIT | 11 | p.V559_G565del | c.1674_1694delGGTTGTTGAGGAGATA AATGG | COSM6971633 |
| KIT | 11 | p.K558_G565del | c.1674_1697delGGTTGTTGAGGAGATA AATGGAAA | COSM1236 |
| KIT | 11 | p.K558_Y570delinsN | c.1674_1709delGGTTGTTGAGGAGATA AATGGAAACAATTATGTTTA | COSM28991 |
| KIT | 11 | p.K558N | c.1674G>C | COSM19216 |
| KIT | 11 | p.K558N | c.1674G>T | OM3238 |
| KIT | 11 | p.V559P | c.1675_1676delGTinsCC | COSM1272086 |
| KIT | 11 | p.V559C | c.1675_1676delGTinsTG | OM3239 |
| KIT | 11 | p.V559K | c.1675_1677delGTTinsAAG | COSM96869 |
| KIT | 11 | p.V559_G565del | c.1675_1695delGTTGTTGAGGAGATAA ATGGA | COSM1249 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.V559I | c.1675G>A | COSM1251 |
| KIT | 11 | p.V559_V560del | c.1676_1681delTTGTTG | COSM1248 |
| KIT | 11 | p.V559_E561del | c.1676_1684delTTGTTGAGG | COSM1250 |
| KIT | 11 | p.V559_N566delinsD | c.1676_1696delTTGTTGAGGAGATAAATGGAA | COSM6977976<br>**Note:** The nucleotide change of COSM6977976 overlaps that of COSM18897, so a positive COSM6977976 sample also results in a positive call for COSM18897. |
| KIT | 11 | p.V559_I571del | c.1676_1714delTTGTTGAGGAGATAAATGGAAACAATTATGTTTACATAG | COSM36301 |
| KIT | 11 | p.V559D | c.1676T>A | COSM1252 |
| KIT | 11 | p.V559A | c.1676T>C | COSM1255 |
| KIT | 11 | p.V559G | c.1676T>G | COSM1253 |
| KIT | 11 | p.V560_I563del | c.1677_1688delTGTTGAGGAGAT | COSM18897 |
| KIT | 11 | p.V560E | c.1679_1680delTTinsAG | COSM1333 |
| KIT | 11 | p.V560del | c.1679_1681delTTG | COSM1247 |
| KIT | 11 | p.V560D | c.1679T>A | COSM1257 |
| KIT | 11 | p.V560A | c.1679T>C | COSM36302 |
| KIT | 11 | p.V560G | c.1679T>G | COSM1260 |
| KIT | 11 | p.E562del | c.1680_1682delTGA | COSM1262 |
| KIT | 11 | p.E562del | c.1684_1686delGAG | COSM1263 |
| KIT | 11 | p.E562_V569delinsD | c.1686_1706delGATAAATGGAAACAATTATGT | COSM1267 |
| KIT | 11 | p.I563_D572del | c.1687_1716delATAAATGGAAACAATTATGTTTACATAGAC | COSM1268 |
| KIT | 11 | p.N564_T574del | c.1690_1722delAATGGAAACAATTATGTTTACATAGACCCAACA | COSM24839 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.N564_P573delinsT | c.1691_1717delATGGAAACAATTATGTTTACATAGACC | COSM36299 |
| KIT | 11 | p.N564_P573delinsTS | c.1691_1717delATGGAAACAATTATGTTTACATAGACCinsCCT | COSM22376 |
| KIT | 11 | p.N566_P573del | c.1697_1720delACAATTATGTTTACATAGACCCAA | COSM6948216 |
| KIT | 11 | p.N567_P573del | c.1700_1720delATTATGTTTACATAGACCCAA | COSM1276 |
| KIT | 11 | p.Y568_T574del | c.1702_1722delTATGTTTACATAGACCCAACA | COSM1334 |
| KIT | 11 | p.Y568_L576delinsCV | c.1703_1726delATGTTTACATAGACCCAACACAACinsGTG | COSM1279 |
| KIT | 11 | p.V569_D572del | c.1705_1716delGTTTACATAGAC | COSM1282 |
| KIT | 11 | p.V569_Q575del | c.1705_1725delGTTTACATAGACCCAACACAA | COSM1281 |
| KIT | 11 | p.V569_L576del | c.1705_1728delGTTTACATAGACCCAACACAACTT | COSM133635 |
| KIT | 11 | p.V569_L576delinsG | c.1706_1727delTTTACATAGACCCAACACAACTinsG | COSM1283 |
| KIT | 11 | p.Y570_L576del | c.1708_1728delTACATAGACCCAACACAACTT | COSM1285 |
| KIT | 11 | p.Y570H | c.1708T>C | COSM1430152 |
| KIT | 11 | p.I571_Y578del | c.1710_1733delCATAGACCCAACACAACTTCCTTA | OMINDEL1186 |
| KIT | 11 | p.I571_L576del | c.1711_1728delATAGACCCAACACAACTT | COSM133754 |
| KIT | 11 | p.Q575_P577delinsT | c.1723_1729delCAACTTCinsA | COSM96865 |
| KIT | 11 | p.Q575_P577del | c.1723_1731delCAACTTCCT | COSM219110 |
| KIT | 11 | p.Q575_P577delinsT | c.1723_1731delCAACTTCCTinsACA | COSM219110old |
| KIT | 11 | p.Q575del | c.1724_1726delAAC | COSM96875 |
| KIT | 11 | p.Q575_L576del | c.1724_1729delAACTTC | COSM6987753 |
| KIT | 11 | p.L576F | c.1726C>T | COSM33966 |
| KIT | 11 | p.L576del | c.1727_1729delTTC | COSM1289 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 11 | p.L576P | c.1727T>C | COSM1290 |
| KIT | 11 | p.L576R | c.1727T>G | COSM5945006 |
| KIT | 11 | p.Q575_L576dup | c.1729_1730insAACTTC | COSM1292 |
| KIT | 11 | p.P577del | c.1729_1731delCCT | COSM1684822 |
| KIT | 11 | p.P577_Y578del | c.1729_1734delCCTTAT | COSM1291 |
| KIT | 11 | p.P577_D579del | c.1730_1738delCTTATGATC | COSM98379 |
| KIT | 11 | p.Y578C | c.1733A>G | COSM6503446 |
| KIT | 11 | p.D579del | c.1735_1737delGAT | COSM1294 |
| KIT | 11 | p.T574_D579dup | c.1737_1738insACACAACTTCCTTATGAT | OMINDEL702 |
| KIT | 11 | p.I571_D579dup | c.1737_1738insATAGACCCAACACAACTTCCTTATGAT | COSM23417 |
| KIT | 11 | p.D572_D579dup | c.1737_1738insGACCCAACACAACTTCCTTATGAT | COSM308544 |
| KIT | 11 | p.D579_H580insSYD | c.1737_1738insTCTTATGAT | COSM96866 |
| KIT | 11 | p.H580del | c.1737_1739delTCA | COSM1295 |
| KIT | 11 | p.P573_D579dup | c.1738_1739insCAACACAACTTCCTTATGATC | COSM18668 |
| KIT | 11 | p.P577_H580dup | c.1739_1740insTCCTTATGATCA | COSM1296 |
| KIT | 11 | p.P573_H580dup | c.1740_1741insCCAACACAACTTCCTTATGATCAC | COSM34126 |
| KIT | 11 | p.P577_H580dup | c.1740_1741insCCTTATGATCAC | COSM18666 |
| KIT | 11 | p.Y578_H580dup | c.1740_1741insTATGATCAC | COSM28995 |
| KIT | 11 | p.Y578_P585dup | c.1754_1755insTTATGATCACAAATGGGAGTTTCC | COSM41632 |
| KIT | 13 | p.K642Q | c.1924A>C | COSM96871 |
| KIT | 13 | p.K642E | c.1924A>G | COSM1304 |
| KIT | 13 | p.K642N | c.1926A>C | COSM4125155 |
| KIT | 13 | p.I653T | c.1958T>C | COSM19326 |
| KIT | 13 | p.V654L | c.1960G>T | COSM5941904 |
| KIT | 13 | p.V654A | c.1961T>C | COSM12706 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 13 | p.N655T | c.1964A>C | OM3303 |
| KIT | 13 | p.N655S | c.1964A>G | OM3302 |
| KIT | 13 | p.N655K | c.1965T>A | COSM4413464 |
| KIT | 13 | p.N655K | c.1965T>G | COSM235682 |
| KIT | 14 | p.T670E | c.2008_2009delACinsGA | COSM19293 |
| KIT | 14 | p.T670A | c.2008A>G | OM3304 |
| KIT | 14 | p.T670I | c.2009C>T | COSM12708 |
| KIT | 14 | p.N680K | c.2040T>G | COSM5352270 |
| KIT | 14 | p.H697Y | c.2089C>T | COSM36053 |
| KIT | 17 | p.R796P | c.2386_2387delAGinsCC | COSM1272092 |
| KIT | 17 | p.R796A | c.2386_2387delAGinsGC | COSM1272091 |
| KIT | 17 | p.R796G | c.2386A>G | COSM51381 |
| KIT | 17 | p.R796K | c.2387G>A | COSM1600411 |
| KIT | 17 | p.D816I | c.2446_2447delGAinsAT | COSM21979 |
| KIT | 17 | p.D816F | c.2446_2447delGAinsTT | COSM1312 |
| KIT | 17 | p.D816H | c.2446_2448delGACinsCAT | COSM1600404 |
| KIT | 17 | p.D816N | c.2446G>A | COSM1313 |
| KIT | 17 | p.D816H | c.2446G>C | COSM1311 |
| KIT | 17 | p.D816Y | c.2446G>T | COSM1310 |
| KIT | 17 | p.D816A | c.2447A>C | COSM24675 |
| KIT | 17 | p.D816G | c.2447A>G | COSM12711 |
| KIT | 17 | p.D816V | c.2447A>T | COSM1314 |
| KIT | 17 | p.D816E | c.2448C>A | COSM5749965 |
| KIT | 17 | p.D816E | c.2448C>G | COSM19285 |
| KIT | 17 | p.D820N | c.2458G>A | COSM17947 |
| KIT | 17 | p.D820H | c.2458G>C | COSM22379 |
| KIT | 17 | p.D820Y | c.2458G>T | COSM12710 |
| KIT | 17 | p.D820A | c.2459A>C | COSM133670 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KIT | 17 | p.D820G | c.2459A>G | COSM1316 |
| KIT | 17 | p.D820V | c.2459A>T | COSM1317 |
| KIT | 17 | p.D820E | c.2460T>A | COSM12709 |
| KIT | 17 | p.D820E | c.2460T>G | COSM19280 |
| KIT | 17 | p.N822H | c.2464A>C | COSM1318 |
| KIT | 17 | p.N822Y | c.2464A>T | COSM19109 |
| KIT | 17 | p.N822S | c.2465A>G | COSM33975 |
| KIT | 17 | p.N822I | c.2465A>T | COSM3604523 |
| KIT | 17 | p.N822K | c.2466T>A | COSM1321 |
| KIT | 17 | p.N822K | c.2466T>G | COSM1322 |
| KIT | 17 | p.Y823N | c.2467T>A | COSM22378 |
| KIT | 17 | p.Y823H | c.2467T>C | COSM1651649 |
| KIT | 17 | p.Y823D | c.2467T>G | COSM18681 |
| KIT | 17 | p.Y823C | c.2468A>G | COSM18682 |
| KIT | 17 | p.V825I | c.2473G>A | COSM19110 |
| KIT | 17 | p.V825A | c.2474T>C | COSM1323 |
| KIT | 18 | p.A829P | c.2485G>C | COSM13172 |
| KIT | 18 | p.A829V | c.2486C>T | COSM133757 |
| KIT | 18 | p.P838L | c.2513C>T | OM3240 |
| KRAS | 2 | p.G12N | c.34_35delGGinsAA | COSM13643 |
| KRAS | 2 | p.G12Y | c.34_35delGGinsTA | COSM25081 |
| KRAS | 2 | p.G12I | c.34_35delGGinsAT | COSM34144 |
| KRAS | 2 | p.G12W | c.34_36delGGTinsTGG | COSM36281 |
| KRAS | 2 | p.G12F | c.34_35delGGinsTT | COSM512 |
| KRAS | 2 | p.G12C | c.34_36delGGTinsTGC | COSM513 |
| KRAS | 2 | p.G12L | c.34_35delGGinsCT | COSM514 |
| KRAS | 2 | p.G12C | c.34G>T | COSM516 |
| KRAS | 2 | p.G12S | c.34G>A | COSM517 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KRAS | 2 | p.G12R | c.34G>C | COSM518 |
| KRAS | 2 | p.G12D | c.35_36delGTinsAC | COSM14209 |
| KRAS | 2 | p.G12E | c.35_36delGTinsAG | COSM30566 |
| KRAS | 2 | p.G12V | c.35_36delGTinsTC | COSM515 |
| KRAS | 2 | p.G12E | c.35_36delGTinsAA | COSM519 |
| KRAS | 2 | p.G12V | c.35G>T | COSM520 |
| KRAS | 2 | p.G12D | c.35G>A | COSM521 |
| KRAS | 2 | p.G12A | c.35G>C | COSM522 |
| KRAS | 2 | p.[G12=;G13C] | c.36_37delTGinsAT | COSM87281 |
| KRAS | 2 | p.G13I | c.37_38delGGinsAT | COSM525 |
| KRAS | 2 | p.G13R | c.37_39delGGCinsCGT | COSM526 |
| KRAS | 2 | p.G13C | c.37G>T | COSM527 |
| KRAS | 2 | p.G13S | c.37G>A | COSM528 |
| KRAS | 2 | p.G13R | c.37G>C | COSM529 |
| KRAS | 2 | p.G13N | c.37_38delGGinsAA | COSM53283 |
| KRAS | 2 | p.G13V | c.38_39delGCinsTT | COSM12721 |
| KRAS | 2 | p.G13E | c.38_39delGCinsAG | COSM30567 |
| KRAS | 2 | p.[G13D;V14I] | c.38_40delGCGinsACA | COSM30568 |
| KRAS | 2 | p.G13V | c.38_39delGCinsTG | COSM530 |
| KRAS | 2 | p.G13D | c.38_39delGCinsAT | COSM531 |
| KRAS | 2 | p.G13A | c.38G>C | COSM533 |
| KRAS | 2 | p.G13V | c.38G>T | COSM534 |
| KRAS | 2 | p.G13E | c.38_39delGCinsAA | COSM87280 |
| KRAS | 2 | p.G13D | c.38G>A | COSM532 |
| KRAS | 2 | p.G15S | c.43G>A | COSM538 |
| KRAS | 2 | p.G15D | c.44G>A | COSM539 |
| KRAS | 3 | p.A59S | c.175G>T | COSM1235389 |
| KRAS | 3 | p.A59T | c.175G>A | COSM546 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| KRAS | 3 | p.A59G | c.176C>G | COSM28518 |
| KRAS | 3 | p.A59E | c.176C>A | COSM547 |
| KRAS | 3 | p.[G60=;Q61K] | c.180_181delTCinsAA | COSM87298 |
| KRAS | 3 | p.Q61K | c.181C>A | COSM549 |
| KRAS | 3 | p.Q61E | c.181C>G | COSM550 |
| KRAS | 3 | p.Q61R | c.182_183delAAinsGT | COSM1168052 |
| KRAS | 3 | p.Q61P | c.182A>C | COSM551 |
| KRAS | 3 | p.Q61R | c.182A>G | COSM552 |
| KRAS | 3 | p.Q61L | c.182A>T | COSM553 |
| KRAS | 3 | p.Q61H | c.183A>C | COSM554 |
| KRAS | 3 | p.Q61H | c.183A>T | COSM555 |
| KRAS | 4 | p.K117E | c.349A>G | COSM6854421 |
| KRAS | 4 | p.K117R | c.350A>G | COSM4696721 |
| KRAS | 4 | p.K117N | c.351A>C | COSM19940 |
| KRAS | 4 | p.K117N | c.351A>T | COSM28519 |
| KRAS | 4 | p.A146T | c.436G>A | COSM19404 |
| KRAS | 4 | p.A146P | c.436G>C | COSM19905 |
| KRAS | 4 | p.A146S | c.436G>T | COSM5967494 |
| KRAS | 4 | p.A146G | c.437C>G | COSM1360828 |
| KRAS | 4 | p.A146V | c.437C>T | COSM19900 |
| MAP2K1 | 2 | p.F53I | c.157T>A | COSM3503329 |
| MAP2K1 | 2 | p.F53V | c.157T>G | COSM5077832 |
| MAP2K1 | 2 | p.F53L | c.157T>C | COSM555604 |
| MAP2K1 | 2 | p.F53C | c.158T>G | COSM1562837 |
| MAP2K1 | 2 | p.F53Y | c.158T>A | COSM5588244 |
| MAP2K1 | 2 | p.F53S | c.158T>C | ClinVar13350 |
| MAP2K1 | 2 | p.F53L | c.159T>A | COSM1725008 |
| MAP2K1 | 2 | p.F53L | c.159T>G | COSM2257208 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MAP2K1 | 2 | p.Q56P | c.167A>C | COSM1235481 |
| MAP2K1 | 2 | p.K57E | c.169A>G | COSM5369532 |
| MAP2K1 | 2 | p.K57Q | c.169A>C | OM3155 |
| MAP2K1 | 2 | p.K57T | c.170A>C | COSM4756761 |
| MAP2K1 | 2 | p.K57M | c.170A>T | MAN124 |
| MAP2K1 | 2 | p.K57N | c.171G>T | COSM1235478 |
| MAP2K1 | 2 | p.K57N | c.171G>C | COSM5520914 |
| MAP2K1 | 2 | p.V60M | c.178G>A | OM3576 |
| MAP2K1 | 2 | p.V60E | c.179T>A | COSM2257209 |
| MAP2K1 | 3 | p.L115P | c.344T>C | OM3248 |
| MAP2K1 | 3 | p.C121S | c.361T>A | COSM555601 |
| MAP2K1 | 3 | p.C121S | c.362G>C | COSM1315829 |
| MAP2K1 | 3 | p.P124S | c.370C>T | COSM235614 |
| MAP2K1 | 3 | p.P124Q | c.371C>A | COSM1167912 |
| MAP2K1 | 3 | p.P124L | c.371C>T | COSM1315861 |
| MAP2K1 | 3 | p.G128D | c.383G>A | COSM1374186 |
| MAP2K1 | 3 | p.G128V | c.383G>T | COSM1517232 |
| MAP2K1 | 3 | p.F129L | c.385T>C | OM3249 |
| MAP2K1 | 3 | p.F129L | c.387C>A | COSM1235480 |
| MAP2K1 | 3 | p.F129L | c.387C>G | OM3250 |
| MAP2K1 | 6 | p.E203K | c.607G>A | COSM232755 |
| MAP2K1 | 6 | p.E203V | c.608A>T | COSM3386991 |
| MAP2K1 | 6 | p.V211D | c.632T>A | OM3251 |
| MAP2K1 | 6 | p.L215P | c.644T>C | OM3252 |
| MAP2K2 | 2 | p.V35M | c.103G>A | COSM5855836 |
| MAP2K2 | 3 | p.C125S | c.373T>A | COSM5855814 |
| MAP2K2 | 3 | p.N126D | c.376A>G | COSM1731742 |
| MET | 14 | p.? | c.2942-27_2942-6delAAGCTCTTTCTTTCTCTCTGTT | COSM24688 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.2942-48_2942-29delCCCATGATAGCCGTCTTTAA | COSM29635 |
| MET | 14 | p.? | c.2942-18_2942-8delCTTTCTCTCTG | COSM29636 |
| MET | 14 | p.? | c.2942-21_2942delTTTCTTTCTCTCTGTTTTAAGA | COSM49015 |
| MET | 14 | p.? | c.2942-20_2942-1delTTCTTTCTCTCTGTTTTAAG | COSM6056832<br>**Note:** The nucleotide change of COSM6056832 overlaps that of OM3177, so a positive COSM6056832 sample also results in a positive call for OM3177. |
| MET | 14 | p.? | c.2942-19_2942-3delTCTTTCTCTCTGTTTTA | COSM6056834<br>**Note:** The nucleotide change of COSM6056834 overlaps that of OMINDEL680, so a positive COSM6056834 sample also results in a positive call for OMINDEL680. |
| MET | 14 | p.? | c.2942-18_2942-2delCTTTCTCTCTGTTTTAA | COSM6057022 |
| MET | 14 | p.? | c.2942-19_2942delTCTTTCTCTCTGTTTTAAGA | COSM6930439 |
| MET | 14 | p.? | c.2942-16_2942-2delTTCTCTCTGTTTTAA | COSM6947399 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.2942-15_2946delTCTCTCTGTTTTAAGATCTG | COSM6948341<br>**Note:** The nucleotide change of COSM6948341 overlaps that of OMINDEL679, so a positive COSM6948341 sample also results in a positive call for OMINDEL679. |
| MET | 14 | p.? | c.2942-6_2942-1delinsA | COSM6976260 |
| MET | 14 | p.? | c.2942-21_2942-1delTTTCTTTCTCTCTGTTTTAAG | COSM6980941 |
| MET | 14 | p.? | c.2942-21_2942-20delinsGC | COSM8631602 |
| MET | 14 | p.? | c.2942-1G>A | OM3177<br>**Note:** The nucleotide change of OM3177 overlaps that of OMINDEL808, so a positive OM3177 sample also results in a positive call for OMINDEL808. |
| MET | 14 | p.? | c.2942-1G>T | OM3178 |
| MET | 14 | p.? | c.2942-29_2942-28delinsGG | OM3305 |
| MET | 14 | p.? | c.2942-17_2942delTTTCTCTCTGTTTTAAGA | OMINDEL640 |
| MET | 14 | p.? | c.2942-16_2942-3delTTCTCTCTGTTTTA | OMINDEL642 |
| MET | 14 | p.? | c.2942-22_2942-3delCTTTCTTTCTCTCTGTTTTA | OMINDEL674 |
| MET | 14 | p.? | c.2942-20_2942-2delTTCTTTCTCTCTGTTTTAA | OMINDEL675 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.2942-20_2942-3delTTCTTTCTCTCTGTTTTA | OMINDEL676<br>**Note:** The nucleotide change of OMINDEL676 overlaps that of COSM6056834 and OMINDEL680, so a positive OMINDEL676 sample also results in positive calls for COSM6056834 and OMINDEL680. |
| MET | 14 | p.? | c.2942-17_2942-1delinsAA | OMINDEL677<br>**Note:** The nucleotide change of OMINDEL677 overlaps that of OMINDEL808 and OM3177, so a positive OMINDEL677 sample also results in positive calls for OMINDEL808 and OM3177. |
| MET | 14 | p.? | c.2942-16_2944delTTCTCTCTGTTTTAAGATC | OMINDEL679<br>**Note:** The nucleotide change of OMINDEL679 overlaps that of OMINDEL640 and COSM6948341, so a positive OMINDEL679 sample also results in positive calls for OMINDEL640 and COSM6948341. |
| MET | 14 | p.? | c.2942-14_2942-3delCTCTCTGTTTTA | OMINDEL680 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.2942-14_2944delCTCTCTGTTTTAAGATC | OMINDEL681<br>**Note:** The nucleotide change of OMINDEL681 overlaps that of OM3178, so a positive OMINDEL681 sample also results in a positive call for OM3178. |
| MET | 14 | p.? | c.2942-13_2942-2delTCTCTGTTTTAA | OMINDEL682 |
| MET | 14 | p.? | c.2942-6_2942-2delinsG | OMINDEL683 |
| MET | 14 | p.? | c.2942-5_2944delinsATA | OMINDEL684 |
| MET | 14 | p.? | c.2942-2_2942-1insC | OMINDEL685 |
| MET | 14 | p.? | c.2942-17_2942-1delTTTCTCTCTGTTTTAAG | OMINDEL697 |
| MET | 14 | p.? | c.2942-5_2944delinsA | OMINDEL706 |
| MET | 14 | p.? | c.2942-24_2942-13delCTCTTTCTTTCT | OMINDEL708 |
| MET | 14 | p.? | c.2942-21_2942-7delTTTCTTTCTCTCTGT | OMINDEL709 |
| MET | 14 | p.? | c.2942-20_2942-10delTTCTTTCTCTC | OMINDEL710 |
| MET | 14 | p.? | c.2942-17_2942-7delinsC | OMINDEL712 |
| MET | 14 | p.? | c.2942-36_2942-20delGTCTTTAACAAGCTCTT | OMINDEL802 |
| MET | 14 | p.? | c.2942-35_2942-20delTCTTTAACAAGCTCTT | OMINDEL803 |
| MET | 14 | p.? | c.2942-20_2942-19delinsAGAG | OMINDEL804 |
| MET | 14 | p.? | c.2942-1delG | OMINDEL808 |
| MET | 14 | p.Y1021F | c.3062A>T | COSM339515 |
| MET | 14 | p.Y1021S | c.3062A>C | COSM7450124 |
| MET | 14 | p.? | c.3065_3082+5delGAGCTACTTTTCCAGAAGGTATA | OMINDEL809 |
| MET | 14 | p.A1023IfsTer19 | c.3066_3081delAGCTACTTTTCCAGAA | OMINDEL810 |
| MET | 14 | p.? | c.3069_3082+1delTACTTTTCCAGAAGG | COSM6057025 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.3071_3082+4delCTTTTCCAGAAGGTAT | OMINDEL686 |
| MET | 14 | p.? | c.3072_3082+7delTTTTCCAGAAGGTATATT | COSM6937267 |
| MET | 14 | p.? | c.3072_3082+5delTTTTCCAGAAGGTATA | COSM6977526 |
| MET | 14 | p.? | c.3073_3082+11delTTTCCAGAAGGTATATTTCAG | OMINDEL654<br>**Note:** The nucleotide change of OMINDEL654 overlaps that of OMINDEL656 and OM3188, so a positive OMINDEL654 sample also results in positive calls for OMINDEL656 and OM3188. |
| MET | 14 | p.? | c.3073_3082+1delTTTCCAGAAGG | OMINDEL655 |
| MET | 14 | p.? | c.3073_3082+5delTTTCCAGAAGGTATA | OMINDEL811<br>**Note:** The nucleotide change of OMINEL811 overlaps that of COSM6977526 and OM3188, so a positive OMINDEL811 sample also results in positive calls for COSM6977526 and OM3188. |
| MET | 14 | p.? | c.3074_3082+11delTTCCAGAAGGTATATTTCAG | OMINDEL656 |
| MET | 14 | p.? | c.3074_3082+3delTTCCAGAAGGTA | OMINDEL658 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.3075_3082+9delinsGGTATATTTT | OMINDEL687<br>**Note:** The nucleotide change of OMINDEL687 overlaps that of COSM6924587 and OMINDEL656, so a positive OMINDEL687 sample also results in positive calls for COSM6924587 and OMINDEL656. |
| MET | 14 | p.? | c.3075_3082+9delTCCAGAAGGTATATTTC | OMINDEL688 |
| MET | 14 | p.? | c.3076_3082+1delCCAGAAGG | OMINDEL689 |
| MET | 14 | p.? | c.3076_3082+5delinsG | OMINDEL690 |
| MET | 14 | p.? | c.3077_3082+8delCAGAAGGTATATTT | OMINDEL662 |
| MET | 14 | p.? | c.3077_3082+13delinsATA | OMINDEL812 |
| MET | 14 | p.? | c.3078_3082+7delAGAAGGTATATT | COSM6911117 |
| MET | 14 | p.? | c.3078_3082+2delAGAAGGT | COSM6965225 |
| MET | 14 | p.? | c.3078_3082+13delAGAAGGTATATTTCAGTT | OMINDEL691 |
| MET | 14 | p.? | c.3078_3082+12delAGAAGGTATATTTCAGT | OMINDEL813 |
| MET | 14 | p.? | c.3079_3082+7delGAAGGTATATT | OMINDEL692 |
| MET | 14 | p.? | c.3079_3082+11delGAAGGTATATTTCAG | OMINDEL693 |
| MET | 14 | p.? | c.3079_3082+1delGAAGG | OMINDEL814 |
| MET | 14 | p.? | c.3080_3082+2delAAGGT | OMINDEL668 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.3081_3082+4delAGGTAT | OMINDEL670<br>**Note:** The nucleotide change of OMINDEL670 overlaps that of OMINDEL668, so a positive OMINDEL670 sample also results in a positive call for OMINDEL668. |
| MET | 14 | p.? | c.3081_3082+2delAGGT | OMINDEL694 |
| MET | 14 | p.D1028IfsTer19 | c.3082delG | COSM24687 |
| MET | 14 | p.? | c.3082+2T>C | COSM6108460 |
| MET | 14 | p.? | c.3082+1G>A | COSM6108461 |
| MET | 14 | p.D1028H | c.3082G>C | COSM6438141 |
| MET | 14 | p.D1028Y | c.3082G>T | COSM6917314 |
| MET | 14 | p.? | c.3082+1_3082+2insA | COSM6923013 |
| MET | 14 | p.D1028N | c.3082G>A | COSM6924587 |
| MET | 14 | p.? | c.3082+1G>C | COSM6928344 |
| MET | 14 | p.? | c.3082+1_3082+4delinsAG | COSM6940825 |
| MET | 14 | p.? | c.3082+2T>A | COSM6947995 |
| MET | 14 | p.? | c.3082+2delT | COSM6983140 |
| MET | 14 | p.? | c.3082+3A>G | COSM744044 |
| MET | 14 | p.? | c.3082+2T>G | OM3185 |
| MET | 14 | p.? | c.3082+3A>C | OM3186 |
| MET | 14 | p.? | c.3082+3A>T | OM3188 |
| MET | 14 | p.? | c.3082+2_3082+4delinsACC | OM3217 |
| MET | 14 | p.? | c.3082_3082+3delinsCC | OMINDEL695 |
| MET | 14 | p.? | c.3082+1_3082+2delGT | OMINDEL696 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| MET | 14 | p.? | c.3082_3082+20delGGTATATTTCAGTTTATTGTT | OMINDEL715<br>**Note:** The nucleotide change of OMINDEL715 overlaps that of OMINDEL670, so a positive OMINDEL715 sample also results in a positive call for OMINDEL670. |
| MET | 14 | p.? | c.3082+2_3082+5delTATA | OMINDEL815<br>**Note:** The nucleotide change of OMINDEL815 overlaps that of OM3188, so a positive OMINDEL815 sample also results in a positive call for OM3188. |
| MET | 16 | p.H1112Y | c.3334C>T | COSM696 |
| MET | 16 | p.H1112L | c.3335A>T | COSM698 |
| MET | 16 | p.H1112R | c.3335A>G | COSM703 |
| NRAS | 2 | p.G12N | c.34_35delGGinsAA | COSM12723 |
| NRAS | 2 | p.G12P | c.34_35delGGinsCC | COSM559 |
| NRAS | 2 | p.G12Y | c.34_35delGGinsTA | COSM560 |
| NRAS | 2 | p.G12R | c.34G>C | COSM561 |
| NRAS | 2 | p.G12C | c.34G>T | COSM562 |
| NRAS | 2 | p.G12S | c.34G>A | COSM563 |
| NRAS | 2 | p.G12E | c.35_36delGTinsAG | COSM144577 |
| NRAS | 2 | p.G12D | c.35G>A | COSM564 |
| NRAS | 2 | p.G12A | c.35G>C | COSM565 |
| NRAS | 2 | p.G12V | c.35G>T | COSM566 |
| NRAS | 2 | p.G13N | c.37_38delGGinsAA | COSM24668 |
| NRAS | 2 | p.G13Y | c.37_38delGGinsTA | COSM568 |
| NRAS | 2 | p.G13R | c.37G>C | COSM569 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| NRAS | 2 | p.G13C | c.37G>T | COSM570 |
| NRAS | 2 | p.G13S | c.37G>A | COSM571 |
| NRAS | 2 | p.G13V | c.38_39delGTinsTC | COSM572 |
| NRAS | 2 | p.G13D | c.38G>A | COSM573 |
| NRAS | 2 | p.G13V | c.38G>T | COSM574 |
| NRAS | 2 | p.G13A | c.38G>C | COSM575 |
| NRAS | 3 | p.A59T | c.175G>A | COSM578 |
| NRAS | 3 | p.A59D | c.176C>A | COSM253327 |
| NRAS | 3 | p.[G60=;Q61K] | c.180_181delACinsTA | COSM12730 |
| NRAS | 3 | p.Q61L | c.181_182delCAinsTT | COSM12725 |
| NRAS | 3 | p.Q61K | c.181_183delCAAinsAAG | COSM53223 |
| NRAS | 3 | p.Q61R | c.181_182delCAinsAG | COSM579 |
| NRAS | 3 | p.Q61K | c.181C>A | COSM580 |
| NRAS | 3 | p.Q61E | c.181C>G | COSM581 |
| NRAS | 3 | p.Q61L | c.182_183delAAinsTG | COSM30646 |
| NRAS | 3 | p.Q61R | c.182_183delAAinsGG | COSM33693 |
| NRAS | 3 | p.Q61P | c.182A>C | COSM582 |
| NRAS | 3 | p.Q61L | c.182A>T | COSM583 |
| NRAS | 3 | p.Q61R | c.182A>G | COSM584 |
| NRAS | 3 | p.[Q61H;E62K] | c.183_184delAGinsCA | COSM26494 |
| NRAS | 3 | p.Q61H | c.183A>T | COSM585 |
| NRAS | 3 | p.Q61H | c.183A>C | COSM586 |
| NRAS | 4 | p.K117N | c.351G>T | MAN13 |
| NRAS | 4 | p.A146T | c.436G>A | COSM27174 |
| NRAS | 4 | p.A146V | c.437C>T | COSM4170228 |
| NTRK1 | 14 | p.V573M | c.1717G>A | BT104 |
| NTRK1 | 14 | p.F589L | c.1765T>C | BT106 |
| NTRK1 | 14 | p.F589Y | c.1766T>A | BT105 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| NTRK1 | 14 | p.F589C | c.1766T>G | OM3255 |
| NTRK1 | 14 | p.F589L | c.1767T>A | BT107 |
| NTRK1 | 14 | p.F589L | c.1767T>G | BT108 |
| NTRK1 | 14 | p.G595R | c.1783G>A | BT109 |
| NTRK1 | 14 | p.G595R | c.1783G>C | BT110 |
| NTRK1 | 15 | p.G667C | c.1999G>T | BT111 |
| NTRK1 | 15 | p.G667S | c.1999G>A | BT112 |
| NTRK1 | 15 | p.G667A | c.2000G>C | OM3301 |
| NTRK2 | 18 | p.Q612E | c.1834C>G | OM3272 |
| NTRK2 | 18 | p.Q612P | c.1835A>C | OM3273 |
| NTRK2 | 18 | p.V617M | c.1849_1851delGTCinsATG | OM3274 |
| NTRK2 | 18 | p.V617G | c.1850T>G | OM3275 |
| NTRK2 | 18 | p.V617D | c.1850T>A | OM3306 |
| NTRK2 | 18 | p.F633I | c.1897T>A | OM3276 |
| NTRK2 | 18 | p.F633L | c.1897T>C | OM3277 |
| NTRK2 | 18 | p.F633C | c.1898T>G | OM3278 |
| NTRK2 | 18 | p.F633Y | c.1898T>A | OM3307 |
| NTRK2 | 18 | p.F633L | c.1899T>A | OM3308 |
| NTRK2 | 18 | p.F633L | c.1899T>G | OM3309 |
| NTRK2 | 18 | p.G639R | c.1915G>A | OM3279 |
| NTRK2 | 18 | p.G639R | c.1915G>C | OM3280 |
| NTRK2 | 18 | p.R646K | c.1937G>A | OM3281 |
| NTRK2 | 19 | p.G709A | c.2126G>C | OM3310 |
| NTRK3 | 16 | p.V601M | c.1801_1803delGTCinsATG | OM3300 |
| NTRK3 | 16 | p.V601F | c.1801G>T | OM3312 |
| NTRK3 | 16 | p.F617L | c.1849T>C | OM3299 |
| NTRK3 | 16 | p.F617Y | c.1850T>A | OM3311 |
| NTRK3 | 16 | p.F617L | c.1851T>G | OM3297 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| NTRK3 | 16 | p.F617L | c.1851T>A | OM3298 |
| NTRK3 | 16 | p.G623R | c.1867G>A | COSM6951362 |
| NTRK3 | 16 | p.G623R | c.1867G>C | OM3253 |
| NTRK3 | 17 | p.G696C | c.2086_2088delGGGinsTGT | OM3294 |
| NTRK3 | 17 | p.G696C | c.2086_2088delGGGinsTGC | OM3295 |
| NTRK3 | 17 | p.G696E | c.2087G>A | COSM5850004 |
| NTRK3 | 17 | p.G696A | c.2087G>C | OM3296 |
| PDGFRA | 11 | p.V536E | c.1607T>A | COSM43059 |
| PDGFRA | 11 | p.A530_I543dup | c.1630_1631insCAGTCCTGGTGCTGTT GGTGATTGTGATCATCTCACTTATTG | COSM7002381 |
| PDGFRA | 12 | p.Y555C | c.1664A>G | COSM133645 |
| PDGFRA | 12 | p.W559_R560del | c.1675_1680delTGGAGG | OMINDEL700 |
| PDGFRA | 12 | p.V561A | c.1682T>C | COSM1430077 |
| PDGFRA | 12 | p.V561D | c.1682T>A | COSM739 |
| PDGFRA | 12 | p.S566_E571delinsR | c.1698_1712delCCCAGATGGACATGA | COSM12418 |
| PDGFRA | 12 | p.D568N | c.1702G>A | OM3225 |
| PDGFRA | 12 | p.P577S | c.1729C>T | COSM6438084 |
| PDGFRA | 12 | p.Q579R | c.1736A>G | OM3226 |
| PDGFRA | 14 | p.H650Q | c.1950C>A | OM3228 |
| PDGFRA | 14 | p.H650Q | c.1950C>G | OM3229 |
| PDGFRA | 14 | p.N659Y | c.1975A>T | COSM22416 |
| PDGFRA | 14 | p.N659H | c.1975A>C | COSM51516 |
| PDGFRA | 14 | p.N659R | c.1975_1976delAAinsCG | OM3230 |
| PDGFRA | 14 | p.N659S | c.1976A>G | COSM6960333 |
| PDGFRA | 14 | p.N659K | c.1977C>G | COSM22414 |
| PDGFRA | 14 | p.N659K | c.1977C>A | COSM22415 |
| PDGFRA | 18 | p.R841K | c.2522G>A | OM3233 |
| PDGFRA | 18 | p.D842_M844del | c.2523_2531delAGACATCAT | COSM96895 |
| PDGFRA | 18 | p.D842Y | c.2524G>T | COSM12396 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PDGFRA | 18 | p.D842Y | c.2524_2526delGACinsTAT | COSM12397 |
| PDGFRA | 18 | p.D842I | c.2524_2525delGAinsAT | COSM12398 |
| PDGFRA | 18 | p.D842_M844del | c.2524_2532delGACATCATG | COSM12401 |
| PDGFRA | 18 | p.D842del | c.2524_2526delGAC | COSM12406 |
| PDGFRA | 18 | p.D842_D846delinsS | c.2524_2537delGACATCATGCATGAinsAG | COSM12409 |
| PDGFRA | 18 | p.D842_D846delinsN | c.2524_2536delGACATCATGCATGinsA | COSM12411<br>**Note:** The nucleotide change of COSM12411 overlaps that of COSM12412, so a positive COSM12411 sample also results in a positive call for COSM12412. |
| PDGFRA | 18 | p.D842F | c.2524_2525delGAinsTT | COSM29007 |
| PDGFRA | 18 | p.D842H | c.2524G>C | COSM734174 |
| PDGFRA | 18 | p.D842_H845delinsY | c.2524_2533delGACATCATGCinsT | COSM96896<br>**Note:** The nucleotide change of COSM96896 overlaps that of COSM96893, so a positive COSM96896 sample also results in a positive call for COSM96893. |
| PDGFRA | 18 | p.D842_S847delinsRI | c.2524_2541delGACATCATGCATGATTCGinsCGAATC | OMINDEL900 |
| PDGFRA | 18 | p.D842_D846delinsG | c.2525_2538delACATCATGCATGATinsGA | COSM12412 |
| PDGFRA | 18 | p.D842_H845delinsV | c.2525_2534delACATCATGCAinsT | COSM12414 |
| PDGFRA | 18 | p.D842_I843delinsV | c.2525_2527delACA | COSM405285 |
| PDGFRA | 18 | p.D842V | c.2525A>T | COSM736 |
| PDGFRA | 18 | p.D842_D846delinsE | c.2526_2538delCATCATGCATGATinsG | COSM12408 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PDGFRA | 18 | p.D842_S847delinsEA | c.2526_2541delCATCATGCATGATTCG insGGCC | COSM12415 |
| PDGFRA | 18 | p.I843_D846del | c.2526_2537delCATCATGCATGA | COSM737<br>**Note:** The nucleotide change of COSM737 overlaps that of COSM12411, so a positive COSM737 sample also results in a positive call for COSM12411. |
| PDGFRA | 18 | p.I843_D846del | c.2527_2538delATCATGCATGAT | COSM12400 |
| PDGFRA | 18 | p.I843_S847delinsT | c.2528_2539delTCATGCATGATT | COSM12407 |
| PDGFRA | 18 | p.I843del | c.2529_2531delCAT | COSM4969655 |
| PDGFRA | 18 | p.H845Y | c.2533C>T | COSM96893 |
| PDGFRA | 18 | p.D846Y | c.2536G>T | COSM12399 |
| PDGFRA | 18 | p.N848K | c.2544C>A | COSM28052 |
| PDGFRA | 18 | p.Y849C | c.2546A>G | COSM94952 |
| PDGFRA | 18 | p.Y849S | c.2546A>C | OM3234 |
| PDGFRA | 18 | p.G853D | c.2558G>A | OM3235 |
| PIK3CA | 2 | p.R38G | c.112C>G | COSM40945 |
| PIK3CA | 2 | p.R38C | c.112C>T | COSM744 |
| PIK3CA | 2 | p.R38S | c.112C>A | COSM87310 |
| PIK3CA | 2 | p.R38L | c.113G>T | COSM729833 |
| PIK3CA | 2 | p.R38H | c.113G>A | COSM745 |
| PIK3CA | 2 | p.E39K | c.115G>A | COSM30625 |
| PIK3CA | 2 | p.E39G | c.116A>G | COSM1041451 |
| PIK3CA | 2 | p.E39V | c.116A>T | COSM1420731 |
| PIK3CA | 2 | p.E81K | c.241G>A | COSM27502 |
| PIK3CA | 2 | p.E81A | c.242A>C | COSM5347052 |
| PIK3CA | 2 | p.P104T | c.310C>A | COSM1666660 |
| PIK3CA | 2 | p.P104L | c.311C>T | COSM163485 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PIK3CA | 2 | p.P104R | c.311C>G | COSM747 |
| PIK3CA | 2 | p.G106R | c.316G>C | COSM86046 |
| PIK3CA | 2 | p.G106A | c.317G>C | COSM166339 |
| PIK3CA | 2 | p.G106V | c.317G>T | COSM748 |
| PIK3CA | 2 | p.R108C | c.322C>T | COSM6438095 |
| PIK3CA | 2 | p.R108H | c.323G>A | COSM27497 |
| PIK3CA | 2 | p.R108L | c.323G>T | COSM729828 |
| PIK3CA | 2 | p.R108P | c.323G>C | COSM749 |
| PIK3CA | 2 | p.E109_I112delinsD | c.327_335delAGAAAAGAT | COSM445995 |
| PIK3CA | 2 | p.E110del | c.328_330delGAA | COSM24710 |
| PIK3CA | 2 | p.K111E | c.331A>G | COSM13570 |
| PIK3CA | 2 | p.K111R | c.332A>G | COSM14052 |
| PIK3CA | 2 | p.K111N | c.333G>C | COSM12580 |
| PIK3CA | 2 | p.K111N | c.333G>T | COSM27505 |
| PIK3CA | 3 | p.G118D | c.353G>A | COSM751 |
| PIK3CA | 5 | p.V344M | c.1030G>A | COSM253279 |
| PIK3CA | 5 | p.V344G | c.1031T>G | COSM22540 |
| PIK3CA | 5 | p.V344E | c.1031T>A | COSM6746438 |
| PIK3CA | 5 | p.V344A | c.1031T>C | COSM86951 |
| PIK3CA | 5 | p.N345D | c.1033A>G | COSM1420788 |
| PIK3CA | 5 | p.N345H | c.1033A>C | COSM3846786 |
| PIK3CA | 5 | p.N345S | c.1034A>G | COSM13476 |
| PIK3CA | 5 | p.N345T | c.1034A>C | COSM446002 |
| PIK3CA | 5 | p.N345I | c.1034A>T | COSM94978 |
| PIK3CA | 5 | p.N345K | c.1035T>A | COSM754 |
| PIK3CA | 8 | p.E418K | c.1252G>A | COSM12582 |
| PIK3CA | 8 | p.C420R | c.1258T>C | COSM757 |
| PIK3CA | 10 | p.D538Y | c.1612G>T | COSM1716811 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PIK3CA | 10 | p.D538N | c.1612G>A | COSM21467 |
| PIK3CA | 10 | p.D538G | c.1613A>G | COSM29123 |
| PIK3CA | 10 | p.P539S | c.1615C>T | COSM249880 |
| PIK3CA | 10 | p.P539T | c.1615C>A | COSM41779 |
| PIK3CA | 10 | p.P539R | c.1616C>G | COSM759 |
| PIK3CA | 10 | p.E542R | c.1624_1625delGAinsAG | COSM1659152 |
| PIK3CA | 10 | p.E542Q | c.1624G>C | COSM17442 |
| PIK3CA | 10 | p.E542K | c.1624G>A | COSM760 |
| PIK3CA | 10 | p.E542A | c.1625A>C | COSM1041494 |
| PIK3CA | 10 | p.E542G | c.1625A>G | COSM761 |
| PIK3CA | 10 | p.E542V | c.1625A>T | COSM762 |
| PIK3CA | 10 | p.E545Q | c.1633G>C | COSM27133 |
| PIK3CA | 10 | p.E545K | c.1633G>A | COSM763 |
| PIK3CA | 10 | p.E545A | c.1634A>C | COSM12458 |
| PIK3CA | 10 | p.E545V | c.1634A>T | COSM27155 |
| PIK3CA | 10 | p.E545G | c.1634A>G | COSM764 |
| PIK3CA | 10 | p.E545D | c.1635G>C | COSM27374 |
| PIK3CA | 10 | p.E545D | c.1635G>T | COSM765 |
| PIK3CA | 10 | p.Q546E | c.1636C>G | COSM6147 |
| PIK3CA | 10 | p.Q546K | c.1636C>A | COSM766 |
| PIK3CA | 10 | p.Q546R | c.1637A>G | COSM12459 |
| PIK3CA | 10 | p.Q546L | c.1637A>T | COSM25041 |
| PIK3CA | 10 | p.Q546P | c.1637A>C | COSM767 |
| PIK3CA | 10 | p.Q546H | c.1638G>T | COSM24712 |
| PIK3CA | 10 | p.Q546H | c.1638G>C | COSM5881064 |
| PIK3CA | 10 | p.E547K | c.1639G>A | COSM29315 |
| PIK3CA | 10 | p.E547G | c.1640A>G | COSM1420864 |
| PIK3CA | 10 | p.E547D | c.1641G>C | COSM729819 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PIK3CA | 21 | p.D1017H | c.3049G>C | COSM27377 |
| PIK3CA | 21 | p.Y1021H | c.3061T>C | COSM17444 |
| PIK3CA | 21 | p.Y1021N | c.3061T>A | COSM6143 |
| PIK3CA | 21 | p.Y1021C | c.3062A>G | COSM12461 |
| PIK3CA | 21 | p.Y1021F | c.3062A>T | COSM328025 |
| PIK3CA | 21 | p.T1025S | c.3073A>T | COSM12590 |
| PIK3CA | 21 | p.T1025P | c.3073A>C | COSM6023925 |
| PIK3CA | 21 | p.T1025A | c.3073A>G | COSM771 |
| PIK3CA | 21 | p.T1025I | c.3074C>T | COSM36285 |
| PIK3CA | 21 | p.T1025N | c.3074C>A | COSM772 |
| PIK3CA | 21 | p.M1043V | c.3127A>G | COSM12591 |
| PIK3CA | 21 | p.M1043L | c.3127A>T | COSM5731063 |
| PIK3CA | 21 | p.M1043T | c.3128T>C | COSM12463 |
| PIK3CA | 21 | p.M1043I | c.3129G>A | COSM29313 |
| PIK3CA | 21 | p.M1043I | c.3129G>T | COSM773 |
| PIK3CA | 21 | p.M1043I | c.3129G>C | COSM94984 |
| PIK3CA | 21 | p.N1044D | c.3130A>G | COSM27134 |
| PIK3CA | 21 | p.N1044Y | c.3130A>T | COSM36288 |
| PIK3CA | 21 | p.N1044S | c.3131A>G | COSM22541 |
| PIK3CA | 21 | p.N1044K | c.3132T>A | COSM12592 |
| PIK3CA | 21 | p.N1044K | c.3132T>G | COSM27504 |
| PIK3CA | 21 | p.D1045N | c.3133G>A | COSM25086 |
| PIK3CA | 21 | p.D1045G | c.3134A>G | COSM1421003 |
| PIK3CA | 21 | p.D1045V | c.3134A>T | COSM729813 |
| PIK3CA | 21 | p.A1046T | c.3136G>A | COSM27273 |
| PIK3CA | 21 | p.A1046E | c.3137C>A | COSM27156 |
| PIK3CA | 21 | p.A1046V | c.3137C>T | COSM6146 |
| PIK3CA | 21 | p.H1047N | c.3139C>A | COSM5029128 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PIK3CA | 21 | p.H1047Y | c.3139C>T | COSM774 |
| PIK3CA | 21 | p.H1047P | c.3140A>C | COSM249874 |
| PIK3CA | 21 | p.H1047R | c.3140A>G | COSM775 |
| PIK3CA | 21 | p.H1047L | c.3140A>T | COSM776 |
| PIK3CA | 21 | p.H1047Q | c.3141T>A | COSM1041525 |
| PIK3CA | 21 | p.H1047Q | c.3141T>G | COSM24714 |
| PIK3CA | 21 | p.H1048N | c.3142C>A | COSM1421010 |
| PIK3CA | 21 | p.H1048Y | c.3142C>T | COSM249875 |
| PIK3CA | 21 | p.H1048D | c.3142C>G | COSM249881 |
| PIK3CA | 21 | p.H1048R | c.3143A>G | COSM36289 |
| PIK3CA | 21 | p.H1048L | c.3143A>T | COSM3734761 |
| PIK3CA | 21 | p.H1048Q | c.3144T>G | COSM27157 |
| PIK3CA | 21 | p.G1049C | c.3145G>T | COSM1168056 |
| PIK3CA | 21 | p.G1049R | c.3145G>C | COSM12597 |
| PIK3CA | 21 | p.G1049S | c.3145G>A | COSM777 |
| PIK3CA | 21 | p.G1049A | c.3146G>C | COSM27158 |
| PIK3CA | 21 | p.G1049D | c.3146G>A | COSM308548 |
| PTEN | 1 | p.Y16C | c.47A>G | COSM5133 |
| PTEN | 1 | p.D24H | c.70G>C | COSM1603823 |
| PTEN | 1 | p.D24N | c.70G>A | COSM5079 |
| PTEN | 1 | p.D24Y | c.70G>T | COSM5237 |
| PTEN | 1 | p.D24V | c.71A>T | COSM4016448 |
| PTEN | 1 | p.D24G | c.71A>G | COSM5107 |
| PTEN | 1 | p.D24E | c.72C>G | COSM5198 |
| PTEN | 2 | p.N48K | c.144C>A | COSM4392678 |
| PTEN | 4 | p.C71Y | c.212G>A | COSM5102 |
| PTEN | 5 | p.A126S | c.376G>T | COSM28889 |
| PTEN | 5 | p.A126T | c.376G>A | COSM5051 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PTEN | 5 | p.A126P | c.376G>C | COSM5211 |
| PTEN | 5 | p.A126D | c.377C>A | COSM28884 |
| PTEN | 5 | p.A126G | c.377C>G | COSM4391544 |
| PTEN | 5 | p.A126V | c.377C>T | COSM5041 |
| PTEN | 5 | p.G127E | c.380G>A | COSM5143 |
| PTEN | 5 | p.G129R | c.385G>C | COSM246853 |
| PTEN | 5 | p.G129E | c.386G>A | COSM28917 |
| PTEN | 5 | p.G129V | c.386G>T | COSM5276 |
| PTEN | 5 | p.R130* | c.388C>T | COSM5152 |
| PTEN | 5 | p.R130G | c.388C>G | COSM5219 |
| PTEN | 5 | p.R130Q | c.389G>A | COSM5033 |
| PTEN | 5 | p.R130L | c.389G>T | COSM5216 |
| PTEN | 5 | p.R130P | c.389G>C | COSM5277 |
| PTEN | 5 | p.T131I | c.392C>T | COSM5104 |
| PTEN | 5 | p.V133I | c.397G>A | COSM5044 |
| PTEN | 5 | p.C136R | c.406T>C | COSM5096 |
| PTEN | 5 | p.C136Y | c.407G>A | COSM12734 |
| PTEN | 5 | p.C136F | c.407G>T | COSM5228 |
| PTEN | 5 | p.Y155C | c.464A>G | COSM5144 |
| PTEN | 6 | p.S170R | c.508A>C | MCH20 |
| PTEN | 6 | p.S170N | c.509G>A | COSM5045 |
| PTEN | 6 | p.S170R | c.510T>G | COSM6006454 |
| PTEN | 6 | p.S170R | c.510T>A | MCH21 |
| PTEN | 6 | p.R173C | c.517C>T | COSM5089 |
| PTEN | 6 | p.R173P | c.518G>C | COSM12735 |
| PTEN | 6 | p.R173H | c.518G>A | COSM5039 |
| PTEN | 6 | p.Y174N | c.520T>A | COSM5221 |
| PTEN | 6 | p.Y174C | c.521A>G | COSM5575601 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| PTEN | 6 | p.L181P | c.542T>C | MCH17 |
| PTEN | 8 | p.F341V | c.1021T>G | COSM5255 |
| PTEN | 8 | p.V343E | c.1028T>A | MCH18 |
| PTEN | 8 | p.L345Q | c.1034T>A | COSM5213 |
| RAF1 | 7 | p.S257P | c.769T>C | ClinVar40600 |
| RAF1 | 7 | p.S257L | c.770C>T | COSM181063 |
| RAF1 | 7 | p.S257W | c.770C>G | COSM581519 |
| RAF1 | 7 | p.S259P | c.775T>C | COSM1418555 |
| RAF1 | 7 | p.S259A | c.775T>G | COSM20741 |
| RAF1 | 7 | p.S259T | c.775T>A | ClinVar40601 |
| RAF1 | 7 | p.S259F | c.776C>T | COSM1536534 |
| RAF1 | 7 | p.S259C | c.776C>G | COSM5719965 |
| RET | 10 | p.C618T | c.1852_1853delTGinsAC | OM3400 |
| RET | 10 | p.C618S | c.1852T>A | COSM87267 |
| RET | 10 | p.C618R | c.1852T>C | COSM29803 |
| RET | 10 | p.C618G | c.1852T>G | COSM9214400 |
| RET | 10 | p.C618Y | c.1853G>A | COSM980 |
| RET | 10 | p.C618S | c.1853G>C | OM3402 |
| RET | 10 | p.C618F | c.1853G>T | OM3401 |
| RET | 10 | p.C618W | c.1854C>G | OM3403 |
| RET | 10 | p.C620S | c.1858T>A | COSM5946160 |
| RET | 10 | p.C620R | c.1858T>C | COSM29804 |
| RET | 10 | p.C620G | c.1858T>G | OM3404 |
| RET | 10 | p.C620Y | c.1859G>A | COSM7403807 |
| RET | 10 | p.C620S | c.1859G>C | COSM29805 |
| RET | 10 | p.C620F | c.1859G>T | OM3405 |
| RET | 10 | p.C620W | c.1860C>G | COSM249815 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| RET | 11 | p.D627_L633delinsAH | c.1880_1899delinsCTCAC | OMINDEL1170<br>**Note:** The nucleotide change of OMINDEL1170 overlaps that of OMINDEL1172, so a positive OMINDEL1170 sample also results in a positive call for OMINDEL1172. |
| RET | 11 | p.P628_L633del | c.1882_1899delCCACTGTGCGACGAGCTG | OMINDEL1171 |
| RET | 11 | p.C630R | c.1888T>C | COSM964 |
| RET | 11 | p.C630G | c.1888T>G | COSM29806 |
| RET | 11 | p.C630Y | c.1889G>A | COSM9233214 |
| RET | 11 | p.C630F | c.1889G>T | OM3406 |
| RET | 11 | p.D631_C634del | c.1891_1902delGACGAGCTGTGC | OMINDEL1172<br>**Note:** The nucleotide change of OMINDEL1172 overlaps that of OMINDEL1173, so a positive OMINDEL1172 sample also results in a positive call for OMINDEL1173. |
| RET | 11 | p.D631Y | c.1891G>T | COSM1738370 |
| RET | 11 | p.D631_L633delinsV | c.1892_1897delACGAGC | OMINDEL1175 |
| RET | 11 | p.D631del | c.1893_1895delCGA | OMINDEL1173 |
| RET | 11 | p.D631_L633delinsE | c.1893_1898delCGAGCT | COSM983 |
| RET | 11 | p.E632_L633del | c.1894_1899delGAGCTG | COSM968 |
| RET | 11 | p.E632_T636delinsSS | c.1894_1906delGAGCTGTGCCGCAinsAGCT | COSM1048 |
| RET | 11 | p.E632K | c.1894G>A | COSM1702268 |
| RET | 11 | p.E632_L633delinsV | c.1895_1897delAGC | COSM982 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| RET | 11 | p.E632_C634delinsG | c.1895_1900delAGCTGT | COSM973 |
| RET | 11 | p.E632_C634delinsDVR | c.1896_1900delGCTGTinsCGTGC | OM3407 |
| RET | 11 | p.C634S | c.1900T>A | COSM1237918 |
| RET | 11 | p.C634R | c.1900T>C | COSM966 |
| RET | 11 | p.C634G | c.1900T>G | COSM1738369 |
| RET | 11 | p.C634L | c.1901_1902delGCinsTG | OM3408 |
| RET | 11 | p.C634Y | c.1901G>A | COSM974 |
| RET | 11 | p.C634S | c.1901G>C | COSM1666664 |
| RET | 11 | p.C634F | c.1901G>T | COSM1237919 |
| RET | 11 | p.E632_C634dup | c.1902_1903insGAGCTGTGC | OMINDEL1176 |
| RET | 11 | p.C634W | c.1902C>G | COSM975 |
| RET | 11 | p.C634_R635insHELC | c.1903_1904insACGAGCTGTGCC | OMINDEL1174 |
| RET | 11 | p.C634_R635dup | c.1905_1906insTGCCGC | OMINDEL1177 |
| RET | 11 | p.C634_T636dup | c.1908_1909insTGCCGCACG | COSM9213763 |
| RET | 12 | p.L730Q | c.2189T>A | OM3409 |
| RET | 12 | p.L730R | c.2189T>G | OM3410 |
| RET | 12 | p.G736A | c.2207G>C | OM3411 |
| RET | 12 | p.L760Q | c.2279T>A | OM3412 |
| RET | 13 | p.E768Q | c.2302G>C | COSM1716312 |
| RET | 13 | p.E768G | c.2303A>G | COSM1347811 |
| RET | 13 | p.E768D | c.2304G>C | COSM21338 |
| RET | 13 | p.E768D | c.2304G>T | OM3413 |
| RET | 13 | p.R770Q | c.2309G>A | COSM6006462 |
| RET | 13 | p.I788N | c.2363T>A | OM3284 |
| RET | 13 | p.L790F | c.2370G>C | rs75030001 |
| RET | 14 | p.V804M | c.2410G>A | OM3195 |
| RET | 14 | p.V804L | c.2410G>C | OM3285 |
| RET | 14 | p.V804L | c.2410G>T | OM3286 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| RET | 14 | p.V804G | c.2411T>G | OM3554 |
| RET | 14 | p.Y806C | c.2417A>G | OM3287 |
| RET | 14 | p.Y806F | c.2417A>T | OM3556 |
| RET | 14 | p.G810S | c.2428G>A | OM3288 |
| RET | 14 | p.G810R | c.2428G>C | OM3289 |
| RET | 14 | p.G810C | c.2428G>T | OM3557 |
| RET | 14 | p.G810A | c.2429G>C | OM3290 |
| RET | 14 | p.G810V | c.2429G>T | OM3558 |
| RET | 15 | p.L881V | c.2641C>G | OM3414 |
| RET | 15 | p.A883F | c.2646_2648delAGCinsTTT | COSM981 |
| RET | 15 | p.A883Y | c.2647_2648delGCinsTA | OM3415 |
| RET | 15 | p.A883F | c.2647_2648delGCinsTT | COSM977 |
| RET | 15 | p.A883T | c.2647G>A | COSM100081 |
| RET | 15 | p.A883S | c.2647G>T | COSM133167 |
| RET | 15 | p.A883V | c.2648C>T | COSM4594153 |
| RET | 15 | p.E884K | c.2650G>A | COSM960 |
| RET | 15 | p.E884V | c.2651A>T | COSM1570338 |
| RET | 15 | p.R886W | c.2656C>T | COSM6942691 |
| RET | 15 | p.S891A | c.2671T>G | rs75234356 |
| RET | 15 | p.R897* | c.2689C>T | COSM6983129 |
| RET | 15 | p.D898_E901del | c.2694_2705delTGTTTATGAAGA | COSM962 |
| RET | 15 | p.V899_D903del | c.2695_2709delGTTTATGAAGAGGAT | COSM5991595 |
| RET | 15 | p.D903_S904delinsEP | c.2709_2710delTTinsAC | OM3416 |
| RET | 15 | p.D903_S904delinsEP | c.2709_2710delTTinsGC | OM3417 |
| RET | 15 | p.S904C | c.2711C>G | OM3418 |
| RET | 15 | p.S904F | c.2711C>T | COSM6438204 |
| RET | 16 | p.G911D | c.2732G>A | COSM20888 |
| RET | 16 | p.R912W | c.2734C>T | COSM3415038 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| RET | 16 | p.R912Q | c.2735G>A | COSM3437793 |
| RET | 16 | p.R912P | c.2735G>C | OM3419 |
| RET | 16 | p.R912L | c.2735G>T | COSM188545 |
| ROS1 | 36 | p.L1951M | c.5851C>A | COSM1072523 |
| ROS1 | 36 | p.L1951R | c.5852T>G | OM3264 |
| ROS1 | 37 | p.S1986F | c.5957C>T | COSM6929870 |
| ROS1 | 37 | p.S1986Y | c.5957C>A | OM3170 |
| ROS1 | 38 | p.L2026M | c.6076C>A | BT151 |
| ROS1 | 38 | p.G2032R | c.6094G>A | MAN10 |
| ROS1 | 38 | p.G2032R | c.6094G>C | MAN11 |
| ROS1 | 38 | p.G2032K | c.6094_6095delGGinsAA | OM3533 |
| ROS1 | 38 | p.D2033N | c.6097G>A | COSM6924065 |
| STK11 | 1 | p.D53Gfs*110 | c.157_158insG | COSM391251 |
| STK11 | 1 | p.L67P | c.200T>C | OM3700 |
| STK11 | 1 | c.289_290+1delinsGAT | c.289_290+1delinsGAT | COSM6922318 |
| STK11 | 1 | c.289_290+2delAAGT | c.289_290+2delAAGT | OMINDEL1000 |
| STK11 | 1 | c.290+1_290+2delGT | c.290+1_290+2delGT | OMINDEL1001 |
| STK11 | 1 | c.290+1G>A | c.290+1G>A | COSM6350392 |
| STK11 | 1 | c.290+1G>C | c.290+1G>C | OM3701 |
| STK11 | 1 | c.290+1G>T | c.290+1G>T | OM3702 |
| STK11 | 1 | c.290+2delT | c.290+2delT | OMINDEL1002 |
| STK11 | 1 | c.290+2T>A | c.290+2T>A | COSM6925006 |
| STK11 | 1 | c.290+2T>G | c.290+2T>G | COSM25857 |
| STK11 | 2 | c.291-1_291delinsTT | c.291-1_291delinsTT | OM3705 |
| STK11 | 2 | c.291-1delG | c.291-1delG | OMINDEL1004 |
| STK11 | 2 | c.291-1G>A | c.291-1G>A | OM3703 |
| STK11 | 2 | c.291-1G>C | c.291-1G>C | OM3704 |
| STK11 | 2 | c.291-1G>T | c.291-1G>T | COSM6083779 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| STK11 | 2 | c.291-2A>G | c.291-2A>G | COSM5575722 |
| STK11 | 2 | c.291-2A>T | c.291-2A>T | COSM49010 |
| STK11 | 3 | c.375-1C>G | c.375-1C>G | COSM4074282 |
| STK11 | 3 | c.375-1C>T | c.375-1C>T | COSM6977173 |
| STK11 | 3 | c.375-2_386delACGTATATGGTGAT | c.375-2_386delACGTATATGGTGAT | OMINDEL1006 |
| STK11 | 3 | c.375-2A>C | c.375-2A>C | OM3706 |
| STK11 | 3 | c.375-2A>G | c.375-2A>G | OM3707 |
| STK11 | 3 | c.375-2A>T | c.375-2A>T | COSM6967966 |
| STK11 | 3 | c.375-6_375-2delCCCCA | c.375-6_375-2delCCCCA | OMINDEL1005 |
| STK11 | 4 | c.465-1_465delinsTT | c.465-1_465delinsTT | COSM6928792 |
| STK11 | 4 | c.465-1delG | c.465-1delG | OMINDEL1010 |
| STK11 | 4 | c.465-1G>A | c.465-1G>A | COSM25855 |
| STK11 | 4 | c.465-1G>C | c.465-1G>C | OM3709 |
| STK11 | 4 | c.465-1G>T | c.465-1G>T | COSM21570 |
| STK11 | 4 | c.465-2_465-1delAG | c.465-2_465-1delAG | OMINDEL1009 |
| STK11 | 4 | c.465-2_465-1delinsTC | c.465-2_465-1delinsTC | OM3708 |
| STK11 | 4 | c.465-2A>C | c.465-2A>C | COSM6933102 |
| STK11 | 4 | c.465-2A>G | c.465-2A>G | COSM21387 |
| STK11 | 4 | c.465-2A>T | c.465-2A>T | COSM1523962 |
| STK11 | 4 | c.465-4_472delGCAGGTACTTCT | c.465-4_472delGCAGGTACTTCT | OMINDEL1007 |
| STK11 | 4 | c.465-4_479delGCAGGTACTTCTGTCAGCT | c.465-4_479delGCAGGTACTTCTGTCAGCT | OMINDEL1008 |
| STK11 | 4 | p.D176N | c.526G>A | COSM4827690 |
| STK11 | 4 | p.D194N | c.580G>A | COSM25847 |
| STK11 | 4 | p.D194H | c.580G>C | COSM5325765 |
| STK11 | 4 | p.D194Y | c.580G>T | COSM20944 |
| STK11 | 4 | p.D194V | c.581A>T | COSM20957 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| STK11 | 4 | p.D194E | c.582C>A | OM3710 |
| STK11 | 4 | p.D194E | c.582C>G | OM3711 |
| STK11 | 5 | c.598-1_598delinsTT | c.598-1_598delinsTT | OM3713 |
| STK11 | 5 | c.598-1_612delGGCACTGCACCCGTTC | c.598-1_612delGGCACTGCACCCGTTC | OMINDEL1014 |
| STK11 | 5 | c.598-1delG | c.598-1delG | OMINDEL1013 |
| STK11 | 5 | c.598-1G>A | c.598-1G>A | COSM3724562 |
| STK11 | 5 | c.598-1G>T | c.598-1G>T | COSM51521 |
| STK11 | 5 | c.598-2_600delAGGCA | c.598-2_600delAGGCA | OMINDEL1012 |
| STK11 | 5 | c.598-2A>C | c.598-2A>C | OM3712 |
| STK11 | 5 | c.598-2A>G | c.598-2A>G | COSM6923168 |
| STK11 | 5 | c.598-2A>T | c.598-2A>T | COSM25858 |
| STK11 | 5 | p.A205T | c.613G>A | COSM20953 |
| STK11 | 5 | p.S240W | c.719C>G | OM3714 |
| STK11 | 5 | c.727_734+9delGTCACCCTGTAAGTGCC | c.727_734+9delGTCACCCTGTAAGTGCC | OMINDEL1015 |
| STK11 | 5 | c.731_734+9delCCCTGTAAGTGCC | c.731_734+9delCCCTGTAAGTGCC | OMINDEL1016 |
| STK11 | 5 | c.734+1G>A | c.734+1G>A | COSM6916684 |
| STK11 | 5 | c.734+1G>C | c.734+1G>C | COSM6970016 |
| STK11 | 5 | c.734+1G>T | c.734+1G>T | COSM51523 |
| STK11 | 5 | c.734+2_734+10delTAAGTGCCC | c.734+2_734+10delTAAGTGCCC | OMINDEL1017 |
| STK11 | 5 | c.734+2T>A | c.734+2T>A | OM3715 |
| STK11 | 5 | c.734+2T>G | c.734+2T>G | COSM6961577 |
| STK11 | 6 | c.735-1G>A | c.735-1G>A | COSM5732040 |
| STK11 | 6 | c.735-1G>T | c.735-1G>T | COSM51522 |
| STK11 | 6 | c.735-2A>G | c.735-2A>G | OM3716 |
| STK11 | 6 | c.735-2A>T | c.735-2A>T | COSM25856 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| STK11 | 6 | c.850_862+4delGACCTGCTGAAAGGTGG | c.850_862+4delGACCTGCTGAAAGGTGG | OMINDEL1018 |
| STK11 | 6 | c.862+1G>A | c.862+1G>A | COSM6929076 |
| STK11 | 6 | c.862+1G>C | c.862+1G>C | OM3717 |
| STK11 | 6 | c.862+1G>T | c.862+1G>T | COSM6149637 |
| STK11 | 6 | c.862+2T>A | c.862+2T>A | COSM5576087 |
| STK11 | 6 | c.862+2T>G | c.862+2T>G | OM3718 |
| STK11 | 6 | c.862+3delG | c.862+3delG | COSM8554973 |
| STK11 | 7 | c.863-1G>A | c.863-1G>A | COSM6916725 |
| STK11 | 7 | c.863-1G>T | c.863-1G>T | COSM6925330 |
| STK11 | 7 | c.863-2A>C | c.863-2A>C | COSM6912582 |
| STK11 | 7 | c.863-2A>T | c.863-2A>T | COSM401786 |
| STK11 | 7 | p.L290P | c.869T>C | OM3719 |
| STK11 | 7 | p.R304W | c.910C>T | COSM29468 |
| STK11 | 7 | c.918_920+11delCAGGTGAGCGGCCC | c.918_920+11delCAGGTGAGCGGCCC | OMINDEL1019 |
| STK11 | 7 | c.920+1G>A | c.920+1G>A | OM3720 |
| STK11 | 7 | c.920+1G>C | c.920+1G>C | COSM4412472 |
| STK11 | 7 | c.920+1G>T | c.920+1G>T | COSM6923458 |
| STK11 | 7 | c.920+2T>A | c.920+2T>A | OM3721 |
| STK11 | 8 | c.1108+1G>A | c.1108+1G>A | OM3722 |
| TP53 | 5 | p.K132Q | c.394A>C | COSM11224 |
| TP53 | 5 | p.K132E | c.394A>G | COSM10813 |
| TP53 | 5 | p.K132T | c.395A>C | COSM43912 |
| TP53 | 5 | p.K132R | c.395A>G | COSM11582 |
| TP53 | 5 | p.K132M | c.395A>T | COSM43592 |
| TP53 | 5 | p.K132N | c.396G>C | COSM43963 |
| TP53 | 5 | p.K132N | c.396G>T | COSM10991 |
| TP53 | 5 | p.C135S | c.403T>A | COSM44910 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 5 | p.C135R | c.403T>C | COSM10684 |
| TP53 | 5 | p.C135G | c.403T>G | COSM44829 |
| TP53 | 5 | p.C135Y | c.404G>A | COSM10801 |
| TP53 | 5 | p.C135S | c.404G>C | COSM44643 |
| TP53 | 5 | p.C135F | c.404G>T | COSM10647 |
| TP53 | 5 | p.C135W | c.405C>G | COSM44219 |
| TP53 | 5 | p.A138V | c.413C>T | COSM43818 |
| TP53 | 5 | p.C141S | c.421T>A | COSM44564 |
| TP53 | 5 | p.C141R | c.421T>C | COSM43901 |
| TP53 | 5 | p.C141G | c.421T>G | COSM45794 |
| TP53 | 5 | p.C141Y | c.422G>A | COSM43708 |
| TP53 | 5 | p.C141S | c.422G>C | COSM293910 |
| TP53 | 5 | p.C141F | c.422G>T | COSM44911 |
| TP53 | 5 | p.C141W | c.423C>G | COSM44204 |
| TP53 | 5 | p.L145P | c.434T>C | COSM43899 |
| TP53 | 5 | p.P151T | c.451C>A | COSM43911 |
| TP53 | 5 | p.P151A | c.451C>G | COSM44944 |
| TP53 | 5 | p.P151S | c.451C>T | COSM10905 |
| TP53 | 5 | p.P151H | c.452C>A | COSM11476 |
| TP53 | 5 | p.P151R | c.452C>G | COSM44003 |
| TP53 | 5 | p.P151L | c.452C>T | COSM44288 |
| TP53 | 5 | p.P152T | c.454C>A | COSM44561 |
| TP53 | 5 | p.P152A | c.454C>G | COSM44788 |
| TP53 | 5 | p.P152S | c.454C>T | COSM43582 |
| TP53 | 5 | p.P152Q | c.455C>A | COSM44613 |
| TP53 | 5 | p.P152R | c.455C>G | COSM45505 |
| TP53 | 5 | p.P152L | c.455C>T | COSM10790 |
| TP53 | 5 | p.G154V | c.461G>T | COSM6815 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 5 | p.R156P | c.467G>C | COSM10760 |
| TP53 | 5 | p.V157L | c.469G>C | COSM45120 |
| TP53 | 5 | p.V157F | c.469G>T | COSM10670 |
| TP53 | 5 | p.V172F | c.514G>T | COSM44240 |
| TP53 | 5 | p.V173M | c.517G>A | COSM11084 |
| TP53 | 5 | p.V173L | c.517G>C | COSM44057 |
| TP53 | 5 | p.V173L | c.517G>T | COSM43559 |
| TP53 | 5 | p.V173E | c.518T>A | COSM45731 |
| TP53 | 5 | p.V173A | c.518T>C | COSM44327 |
| TP53 | 5 | p.V173G | c.518T>G | COSM44383 |
| TP53 | 5 | p.R175S | c.523C>A | COSM43931 |
| TP53 | 5 | p.R175G | c.523C>G | COSM10870 |
| TP53 | 5 | p.R175C | c.523C>T | COSM43680 |
| TP53 | 5 | p.R175H | c.524G>A | COSM10648 |
| TP53 | 5 | p.R175P | c.524G>C | COSM45416 |
| TP53 | 5 | p.R175L | c.524G>T | COSM10718 |
| TP53 | 5 | p.C176S | c.526T>A | COSM44146 |
| TP53 | 5 | p.C176R | c.526T>C | COSM44948 |
| TP53 | 5 | p.C176G | c.526T>G | COSM44692 |
| TP53 | 5 | p.C176Y | c.527G>A | COSM10687 |
| TP53 | 5 | p.C176S | c.527G>C | COSM44645 |
| TP53 | 5 | p.C176F | c.527G>T | COSM10645 |
| TP53 | 5 | p.C176W | c.528C>G | COSM11114 |
| TP53 | 5 | p.P177_C182del | c.529_546delCCCCACCATGAGCGCTGC | COSM43570 |
| TP53 | 5 | p.H179N | c.535C>A | COSM44151 |
| TP53 | 5 | p.H179D | c.535C>G | COSM44776 |
| TP53 | 5 | p.H179Y | c.535C>T | COSM10768 |
| TP53 | 5 | p.H179P | c.536A>C | COSM44218 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 5 | p.H179R | c.536A>G | COSM10889 |
| TP53 | 5 | p.H179L | c.536A>T | COSM43635 |
| TP53 | 5 | p.H179Q | c.537T>A | COSM44214 |
| TP53 | 5 | p.H179Q | c.537T>G | COSM11249 |
| TP53 | 6 | p.P190L | c.569C>T | COSM43657 |
| TP53 | 6 | p.P191del | c.572_574delCTC | COSM45140 |
| TP53 | 6 | p.Q192* | c.574C>T | COSM10733 |
| TP53 | 6 | p.H193N | c.577C>A | COSM43935 |
| TP53 | 6 | p.H193D | c.577C>G | COSM44002 |
| TP53 | 6 | p.H193Y | c.577C>T | COSM10672 |
| TP53 | 6 | p.H193P | c.578A>C | COSM43833 |
| TP53 | 6 | p.H193R | c.578A>G | COSM10742 |
| TP53 | 6 | p.H193L | c.578A>T | COSM11066 |
| TP53 | 6 | p.L194I | c.580C>A | COSM45330 |
| TP53 | 6 | p.L194V | c.580C>G | COSM46117 |
| TP53 | 6 | p.L194F | c.580C>T | COSM10995 |
| TP53 | 6 | p.L194H | c.581T>A | COSM43623 |
| TP53 | 6 | p.L194P | c.581T>C | COSM43827 |
| TP53 | 6 | p.L194R | c.581T>G | COSM44571 |
| TP53 | 6 | p.I195L | c.583A>C | COSM45773 |
| TP53 | 6 | p.I195F | c.583A>T | COSM44633 |
| TP53 | 6 | p.I195N | c.584T>A | COSM44877 |
| TP53 | 6 | p.I195T | c.584T>C | COSM11089 |
| TP53 | 6 | p.I195S | c.584T>G | COSM44539 |
| TP53 | 6 | p.I195M | c.585C>G | COSM562637 |
| TP53 | 6 | p.R196* | c.586C>T | COSM10705 |
| TP53 | 6 | p.V197M | c.589G>A | COSM43779 |
| TP53 | 6 | p.R213G | c.637C>G | COSM44102 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 6 | p.R213* | c.637C>T | COSM10654 |
| TP53 | 6 | p.R213Q | c.638G>A | COSM10735 |
| TP53 | 6 | p.R213P | c.638G>C | COSM11860 |
| TP53 | 6 | p.R213L | c.638G>T | COSM43650 |
| TP53 | 6 | p.H214D | c.640C>G | COSM45115 |
| TP53 | 6 | p.H214Y | c.640C>T | COSM44112 |
| TP53 | 6 | p.H214P | c.641A>C | COSM1640841 |
| TP53 | 6 | p.H214R | c.641A>G | COSM43687 |
| TP53 | 6 | p.H214L | c.641A>T | COSM5632535 |
| TP53 | 6 | p.H214Q | c.642T>A | COSM46272 |
| TP53 | 6 | p.H214Q | c.642T>G | COSM44407 |
| TP53 | 6 | p.V216M | c.646G>A | COSM10667 |
| TP53 | 6 | p.V216L | c.646G>T | COSM11210 |
| TP53 | 6 | p.V216E | c.647T>A | COSM44274 |
| TP53 | 6 | p.V216A | c.647T>C | COSM44567 |
| TP53 | 6 | p.V216G | c.647T>G | COSM43681 |
| TP53 | 6 | p.Y220N | c.658T>A | COSM44672 |
| TP53 | 6 | p.Y220H | c.658T>C | COSM44637 |
| TP53 | 6 | p.Y220D | c.658T>G | COSM11847 |
| TP53 | 6 | p.Y220S | c.659A>C | COSM43850 |
| TP53 | 6 | p.Y220C | c.659A>G | COSM10758 |
| TP53 | 6 | p.Y220F | c.659A>T | COSM1564197 |
| TP53 | 7 | p.Y234N | c.700T>A | COSM43956 |
| TP53 | 7 | p.Y234H | c.700T>C | COSM11152 |
| TP53 | 7 | p.Y234D | c.700T>G | COSM43768 |
| TP53 | 7 | p.Y234S | c.701A>C | COSM43865 |
| TP53 | 7 | p.Y234C | c.701A>G | COSM10725 |
| TP53 | 7 | p.Y234F | c.701A>T | COSM44315 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 7 | p.Y236N | c.706T>A | COSM43826 |
| TP53 | 7 | p.Y236H | c.706T>C | COSM44326 |
| TP53 | 7 | p.Y236D | c.706T>G | COSM43602 |
| TP53 | 7 | p.Y236S | c.707A>C | COSM44693 |
| TP53 | 7 | p.Y236C | c.707A>G | COSM10731 |
| TP53 | 7 | p.Y236F | c.707A>T | COSM3958809 |
| TP53 | 7 | p.M237V | c.709A>G | COSM44525 |
| TP53 | 7 | p.M237L | c.709A>T | COSM44965 |
| TP53 | 7 | p.M237K | c.710T>A | COSM43952 |
| TP53 | 7 | p.M237T | c.710T>C | COSM45329 |
| TP53 | 7 | p.M237R | c.710T>G | COSM45032 |
| TP53 | 7 | p.M237I | c.711G>A | COSM10834 |
| TP53 | 7 | p.M237I | c.711G>C | COSM44415 |
| TP53 | 7 | p.M237I | c.711G>T | COSM11063 |
| TP53 | 7 | p.C238S | c.712T>A | COSM43700 |
| TP53 | 7 | p.C238R | c.712T>C | COSM44321 |
| TP53 | 7 | p.C238G | c.712T>G | COSM46336 |
| TP53 | 7 | p.C238Y | c.713G>A | COSM11059 |
| TP53 | 7 | p.C238S | c.713G>C | COSM44653 |
| TP53 | 7 | p.C238F | c.713G>T | COSM43778 |
| TP53 | 7 | p.C238W | c.714T>G | COSM44676 |
| TP53 | 7 | p.N239D | c.715A>G | COSM10777 |
| TP53 | 7 | p.N239Y | c.715A>T | COSM45870 |
| TP53 | 7 | p.N239T | c.716A>C | COSM43801 |
| TP53 | 7 | p.N239S | c.716A>G | COSM44094 |
| TP53 | 7 | p.N239I | c.716A>T | COSM307333 |
| TP53 | 7 | p.N239K | c.717C>A | COSM44647 |
| TP53 | 7 | p.N239K | c.717C>G | COSM44510 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 7 | p.S240G | c.718A>G | COSM43973 |
| TP53 | 7 | p.S241T | c.721T>A | COSM44067 |
| TP53 | 7 | p.S241P | c.721T>C | COSM44578 |
| TP53 | 7 | p.S241A | c.721T>G | COSM44224 |
| TP53 | 7 | p.S241Y | c.722C>A | COSM10935 |
| TP53 | 7 | p.S241C | c.722C>G | COSM10709 |
| TP53 | 7 | p.S241F | c.722C>T | COSM10812 |
| TP53 | 7 | p.C242S | c.724T>A | COSM44935 |
| TP53 | 7 | p.C242R | c.724T>C | COSM11738 |
| TP53 | 7 | p.C242G | c.724T>G | COSM44135 |
| TP53 | 7 | p.C242Y | c.725G>A | COSM10646 |
| TP53 | 7 | p.C242S | c.725G>C | COSM11133 |
| TP53 | 7 | p.C242F | c.725G>T | COSM10810 |
| TP53 | 7 | p.C242W | c.726C>G | COSM11356 |
| TP53 | 7 | p.G244S | c.730G>A | COSM10941 |
| TP53 | 7 | p.G244R | c.730G>C | COSM44221 |
| TP53 | 7 | p.G244C | c.730G>T | COSM11524 |
| TP53 | 7 | p.G244D | c.731G>A | COSM10883 |
| TP53 | 7 | p.G244A | c.731G>C | COSM12013 |
| TP53 | 7 | p.G244V | c.731G>T | COSM43652 |
| TP53 | 7 | p.G245S | c.733G>A | COSM6932 |
| TP53 | 7 | p.G245R | c.733G>C | COSM10957 |
| TP53 | 7 | p.G245C | c.733G>T | COSM11081 |
| TP53 | 7 | p.G245D | c.734G>A | COSM43606 |
| TP53 | 7 | p.G245A | c.734G>C | COSM43965 |
| TP53 | 7 | p.G245V | c.734G>T | COSM11196 |
| TP53 | 7 | p.M246V | c.736A>G | COSM43555 |
| TP53 | 7 | p.M246L | c.736A>T | COSM45992 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 7 | p.M246K | c.737T>A | COSM44103 |
| TP53 | 7 | p.M246T | c.737T>C | COSM11355 |
| TP53 | 7 | p.M246R | c.737T>G | COSM11376 |
| TP53 | 7 | p.M246I | c.738G>A | COSM44310 |
| TP53 | 7 | p.M246I | c.738G>C | COSM10757 |
| TP53 | 7 | p.M246I | c.738G>T | COSM46136 |
| TP53 | 7 | p.R248G | c.742C>G | COSM11564 |
| TP53 | 7 | p.R248W | c.742C>T | COSM10656 |
| TP53 | 7 | p.R248Q | c.743G>A | COSM10662 |
| TP53 | 7 | p.R248P | c.743G>C | COSM11491 |
| TP53 | 7 | p.R248L | c.743G>T | COSM6549 |
| TP53 | 7 | p.R249G | c.745A>G | COSM10668 |
| TP53 | 7 | p.R249W | c.745A>T | COSM43629 |
| TP53 | 7 | p.R249K | c.746G>A | COSM44091 |
| TP53 | 7 | p.R249T | c.746G>C | COSM43665 |
| TP53 | 7 | p.R249M | c.746G>T | COSM43871 |
| TP53 | 7 | p.R249S | c.747G>C | COSM10785 |
| TP53 | 7 | p.R249S | c.747G>T | COSM10817 |
| TP53 | 7 | p.P250L | c.749C>T | COSM10771 |
| TP53 | 7 | p.E258K | c.772G>A | COSM10988 |
| TP53 | 7 | p.E258Q | c.772G>C | COSM10751 |
| TP53 | 7 | p.E258A | c.773A>C | COSM44719 |
| TP53 | 7 | p.E258G | c.773A>G | COSM44168 |
| TP53 | 7 | p.E258V | c.773A>T | COSM44450 |
| TP53 | 7 | p.E258D | c.774A>C | COSM46410 |
| TP53 | 7 | p.E258D | c.774A>T | COSM44962 |
| TP53 | 8 | p.G262V | c.785G>T | COSM11198 |
| TP53 | 8 | p.G266R | c.796G>A | COSM10794 |

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| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 8 | p.G266R | c.796G>C | COSM11205 |
| TP53 | 8 | p.G266E | c.797G>A | COSM10867 |
| TP53 | 8 | p.G266A | c.797G>C | COSM45488 |
| TP53 | 8 | p.G266V | c.797G>T | COSM10958 |
| TP53 | 8 | p.V272M | c.814G>A | COSM10891 |
| TP53 | 8 | p.V272L | c.814G>C | COSM45898 |
| TP53 | 8 | p.V272L | c.814G>T | COSM10859 |
| TP53 | 8 | p.V272E | c.815T>A | COSM44580 |
| TP53 | 8 | p.V272A | c.815T>C | COSM44294 |
| TP53 | 8 | p.V272G | c.815T>G | COSM44870 |
| TP53 | 8 | p.R273S | c.817C>A | COSM43909 |
| TP53 | 8 | p.R273G | c.817C>G | COSM43843 |
| TP53 | 8 | p.R273C | c.817C>T | COSM10659 |
| TP53 | 8 | p.R273H | c.818G>A | COSM10660 |
| TP53 | 8 | p.R273P | c.818G>C | COSM43896 |
| TP53 | 8 | p.R273L | c.818G>T | COSM10779 |
| TP53 | 8 | p.V274L | c.820G>C | COSM44443 |
| TP53 | 8 | p.V274F | c.820G>T | COSM10769 |
| TP53 | 8 | p.C275R | c.823T>C | COSM43902 |
| TP53 | 8 | p.C275G | c.823T>G | COSM11501 |
| TP53 | 8 | p.C275Y | c.824G>A | COSM10893 |
| TP53 | 8 | p.C275S | c.824G>C | COSM45413 |
| TP53 | 8 | p.C275F | c.824G>T | COSM10701 |
| TP53 | 8 | p.C275W | c.825T>G | COSM43823 |
| TP53 | 8 | p.C277F | c.830G>T | COSM10749 |
| TP53 | 8 | p.P278T | c.832C>A | COSM43697 |
| TP53 | 8 | p.P278A | c.832C>G | COSM10814 |
| TP53 | 8 | p.P278S | c.832C>T | COSM10939 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 8 | p.P278H | c.833C>A | COSM43755 |
| TP53 | 8 | p.P278R | c.833C>G | COSM10887 |
| TP53 | 8 | p.P278L | c.833C>T | COSM10863 |
| TP53 | 8 | p.G279E | c.836G>A | COSM43714 |
| TP53 | 8 | p.R280G | c.838A>G | COSM11123 |
| TP53 | 8 | p.R280K | c.839G>A | COSM10728 |
| TP53 | 8 | p.R280T | c.839G>C | COSM10724 |
| TP53 | 8 | p.R280I | c.839G>T | COSM11287 |
| TP53 | 8 | p.R280S | c.840A>C | COSM44233 |
| TP53 | 8 | p.R280S | c.840A>T | COSM44171 |
| TP53 | 8 | p.D281N | c.841G>A | COSM43596 |
| TP53 | 8 | p.D281H | c.841G>C | COSM10943 |
| TP53 | 8 | p.D281Y | c.841G>T | COSM11516 |
| TP53 | 8 | p.D281A | c.842A>C | COSM11665 |
| TP53 | 8 | p.D281G | c.842A>G | COSM11232 |
| TP53 | 8 | p.D281V | c.842A>T | COSM45729 |
| TP53 | 8 | p.D281E | c.843C>A | COSM43906 |
| TP53 | 8 | p.D281E | c.843C>G | COSM43837 |
| TP53 | 8 | p.R282G | c.844C>G | COSM10992 |
| TP53 | 8 | p.R282W | c.844C>T | COSM10704 |
| TP53 | 8 | p.R282Q | c.845G>A | COSM44338 |
| TP53 | 8 | p.R282P | c.845G>C | COSM44306 |
| TP53 | 8 | p.R282L | c.845G>T | COSM44470 |
| TP53 | 8 | p.R283P | c.848G>C | COSM10743 |
| TP53 | 8 | p.E285K | c.853G>A | COSM10722 |
| TP53 | 8 | p.E285Q | c.853G>C | COSM45138 |
| TP53 | 8 | p.E285A | c.854A>C | COSM43614 |
| TP53 | 8 | p.E285G | c.854A>G | COSM45649 |

*(continued)*

| Gene | Exon | Amino acid change | Nucleotide change | Variant ID |
|---|---|---|---|---|
| TP53 | 8 | p.E285V | c.854A>T | COSM44227 |
| TP53 | 8 | p.E286K | c.856G>A | COSM10726 |
| TP53 | 8 | p.E286Q | c.856G>C | COSM44250 |
| TP53 | 8 | p.E286A | c.857A>C | COSM45892 |
| TP53 | 8 | p.E286G | c.857A>G | COSM43565 |
| TP53 | 8 | p.E286V | c.857A>T | COSM43936 |
| TP53 | 8 | p.E286D | c.858A>C | COSM45439 |
| TP53 | 8 | p.E286D | c.858A>T | COSM45492 |

# Fusion isoforms targeted by the Oncomine™ Dx Express Test

| ALK fusion isoforms (N=213) | | |
|---|---|---|
| A2M-ALK.A22A19 | EML4-ALK.E3E6A20 | PDE3A-ALK.P10A8 |
| ACTG2-ALK.A1A17 | EML4-ALK.E3p53insA20.1 | PDE8B-ALK.P11A20 |
| ACTG2-ALK.A1A19 | EML4-ALK.E4A13 | PHACTR1-ALK.P4A20 |
| ACTG2-ALK.A2A18 | EML4-ALK.E6A16 | PLEKHA7-ALK.P26A19 |
| AGAP1-ALK.A6A18 | EML4-ALK.E6A17 | PNPT1-ALK.P5A4 |
| ATG16L1-ALK.A2A20 | EML4-ALK.E6A18.1 | PPFIBP1-ALK.P12A20.COSF1461 |
| ATG16L1-ALK.A2ins25ins97A20 | EML4-ALK.E6A19.COSF1296.2 | PPFIBP1-ALK.P8A20ins49.1 |
| ATIC-ALK.A7A20.COSF444 | EML4-ALK.E6aA20.AB374361.1 | PPFIBP1-ALK.P9A20 |
| ATRNL1-ALK.A19A20 | EML4-ALK.E6bA17 | PPM1B-ALK.P1A20 |
| BCL11A-ALK.B4A20 | EML4-ALK.E6bA20.AB374362.1 | PPM1G-ALK.P1A18 |
| BEND5-ALK.B3A20 | EML4-ALK.E6ins18A20.1 | PPP1CB-ALK.P5A20 |
| BIRC6-ALK.B10A20 | EML4-ALK.E7A20.NGS | PPP1CB-ALK.P7A20 |
| CAD-ALK.C35A20 | ETV6-ALK.E4A20 | PPP1R21-ALK.P17A20 |
| CAD-ALK.C36A20 | ETV6-ALK.E5A20 | PPP1R21-ALK.P7A20 |
| CAMKMT-ALK.C3A20 | FBXO36-ALK.F2A20 | PPP4R3B-ALK.P9A2 |
| CARS-ALK.C17A20.COSF437 | FN1-ALK.F15A18 | PRKAR1A-ALK.P10A20 |

*(continued)*

| ALK fusion isoforms (N=213) | | |
|---|---|---|
| CCDC149-ALK.C10A20 | FN1-ALK.F20A19 | PRKAR1A-ALK.P2A20.NGS.2 |
| CCDC61-ALK.C13A20 | FN1-ALK.F23A19.COSF1301 | PRKAR1A-ALK.P5A20 |
| CCDC88A-ALK.C12A20 | FN1-ALK.F27A18 | PRKAR1B-ALK.P4A20 |
| CENPF-ALK.C11A20 | FN1-ALK.F41A16 | PRKAR2A-ALK.P2A20 |
| CEBPZ-ALK.C2A20 | FN1-ALK.F42A16 | PRKCE-ALK.P1A20 |
| CLIP1-ALK.C13A20 | GALNT14-ALK.G1A19 | RANBP2-ALK.R18A20.COSF415 |
| CLIP4-ALK.C10A20 | GCC2-ALK.G13A20 | RNF213-ALK.R20A20 |
| CLIP4-ALK.C14A17 | GCC2-ALK.G19A20 | RRBP1-ALK.R20ins33A20.1 |
| CLTC-ALK.C30A20.COSF470.1 | GTF2IRD1-ALK.G4A20 | SATB1-ALK.S5A20 |
| CLTC-ALK.C31A20.COSF434 | GTF2IRD1-ALK.G7A20 | SATB1-ALK.S6A20 |
| CLTC-ALK.C31ins63A20 | HIP1-ALK.H21A20 | SEC31A-ALK.S21A20.COSF460 |
| COL25A1-ALK.C3A20 | HIP1-ALK.H28A20 | SEC31A-ALK.S22A20.COSF459 |
| CPSF7-ALK.C5A20 | HIP1-ALK.H30A20.COSF1713 | SFT2D1-ALK.S1A6 |
| CREB1-ALK.C1A9 | HNRNPA1-ALK.H2A18 | SLC34A2-ALK.S1A20 |
| CTNND1-ALK.C16A20 | IGFBP5-ALK.I1A19 | SLC8A1-ALK.S1A20 |
| DCTN1-ALK.D15A20 | INTS9-ALK.I8A20 | SLMAP-ALK.S11A20 |
| DCTN1-ALK.D26A20.COSF1626 | ITSN2-ALK.I14A2 | SP100-ALK.S4A20 |
| DCTN1-ALK.D27A20 | KANK2-ALK.K4A16 | SPDYA-ALK.S6A20 |
| DCTN1-ALK.D29A20 | KCNQ5-ALK.K1A10 | SPECC1L-ALK.S9A20 |
| DDX6-ALK.D2A20 | KIF5B-ALK.K15A20.COSF1060.2 | SPTBN1-ALK.S5A20 |
| DES-ALK.D7A19 | KIF5B-ALK.K15A20.COSF1381.1 | SPTBN1-ALK.S6A20 |
| DMD-ALK.D55A20 | KIF5B-ALK.K17A20.COSF1257 | SPTBN1-ALK.S7A20 |
| EEF1G-ALK.E6A20 | KIF5B-ALK.K24A19 | SQSTM1-ALK.S5A19 |
| EEF1G-ALK.E8A20 | KIF5B-ALK.K24A20.COSF1058.1 | SQSTM1-ALK.S5A20.COSF1051 |
| EIF2AK3-ALK.E2A20 | KLC1-ALK.K9A20.COSF1276 | STK32B-ALK.S11A20 |
| EML4-ALK.E12A20 | KTN1-ALK.K43A19 | STRN-ALK.S3A20.COSF1430 |
| EML4-ALK.E13A19 | LMNA-ALK.L2A20 | TERT-ALK.T11A5 |
| EML4-ALK.E13A20 | LMO7-ALK.L15A20 | TFG-ALK.T4A19 |
| EML4-ALK.E13A20.COSF1062.3 | LRPPRC-ALK.L23A20 | TFG-ALK.T4A20.COSF424 |

*(continued)*

| ALK fusion isoforms (N=213) | | |
|---|---|---|
| EML4-ALK.E13A20.COSF408.2 | LRRFIP2-ALK.L26A20 | TFG-ALK.T5A19 |
| EML4-ALK.E13ins90A20.1 | LTBP1-ALK.L19A20 | TFG-ALK.T5A20.COSF426 |
| EML4-ALK.E14A19 | MALAT1-ALK.M1A16.2 | TFG-ALK.T6A20.COSF428.1 |
| EML4-ALK.E14A20.COSF1064.2 | MANBA-ALK.M14A20 | TFG-ALK.T7A19 |
| EML4-ALK.E14A20.COSF477.3 | MAPRE3-ALK.M7A20 | TFG-ALK.T7A20 |
| EML4-ALK.E14del36A20 | MCFD2-ALK.M1A20 | THBS1-ALK.T4A19 |
| EML4-ALK.E14ins124A20.2 | MEMO1-ALK.M1A2 | TIMP3-ALK.T1A19 |
| EML4-ALK.E14ins2del52A20.1 | MEMO1-ALK.M2A7 | TIMP3-ALK.T1A20 |
| EML4-ALK.E15A20.COSF413.2 | MLPH-ALK.M11A20 | TNS1-ALK.T16A17 |
| EML4-ALK.E17A19 | MPRIP-ALK.M21A20 | TNS1-ALK.T16A20 |
| EML4-ALK.E17A20.COSF1366.4 | MROH2B-ALK.M21A12 | TNS1-ALK.T17A12 |
| EML4-ALK.E17A20.COSF1367.3 | MSN-ALK.M11A20.COSF421 | TNS1-ALK.T20A12 |
| EML4-ALK.E17ins59del79A20 | MSN-ALK.M11int12A20 | TNS1-ALK.T26A11 |
| EML4-ALK.E17ins65A20.1 | MSN-ALK.M9A20 | TNS1-ALK.T28A20 |
| EML4-ALK.E18A20.COSF487.1 | MYH10-ALK.M38A20 | TNS1-ALK.T32A13 |
| EML4-ALK.E19A20 | MYH9-ALK.M34A20del23 | TPM1-ALK.T8A20.NGS |
| EML4-ALK.E19A20full | MYH9-ALK.M36A20 | TPM3-ALK.T7A20.COSF439 |
| EML4-ALK.E20A20.COSF409.2 | MYH9-ALK.M9A6ins10.1 | TPM4-ALK.T7A20 |
| EML4-ALK.E20A20.COSF730.2 | MYO5C-ALK.M30A20 | TPM4-ALK.T7A20.COSF441 |
| EML4-ALK.E21A19 | MYT1L-ALK.M14A20 | TPR-ALK.T15A20.1 |
| EML4-ALK.E21A20 | NCOA1-ALK.N21A1.NGS | TPR-ALK.T4A20 |
| EML4-ALK.E22A20 | NPM1-ALK.N4A20.COSF198 | TRAF1-ALK.T6A20 |
| EML4-ALK.E2A19 | NUMA1-ALK.N19A20 | TRMT61B-ALK.T1A9 |
| EML4-ALK.E2A20.COSF478.2 | OTOF-ALK.O2A3 | VCL-ALK.V16A20.COSF1057.1 |
| EML4-ALK.E2A20.COSF479.2 | PABPC1-ALK.P9A20 | YPEL5-ALK.Y1A18 |
| EML4-ALK.E3A20 | PCM1-ALK.P23A20 | ZNF2-ALK.Z1A20 |

| BRAF fusion isoforms (N=187) | | |
|---|---|---|
| AGAP3-BRAF.A10B11 | GIT2-BRAF.G14B9 | PRKAR1B-BRAF.P10B9 |
| AGAP3-BRAF.A10B9 | GNAI1-BRAF.G1B10.COSF1442 | PRKAR1B-BRAF.P9B9 |
| AGAP3-BRAF.A8B11 | GTF2I-BRAF.G20B10 | PRKAR2B-BRAF.P1B10 |
| AGAP3-BRAF.A8B9 | GTF2I-BRAF.G4B10 | RAD18-BRAF.R7B10 |
| AGAP3-BRAF.A9B9 | HERPUD1-BRAF.H4B7.1 | RBM33-BRAF.R3B11 |
| AGK-BRAF.A2B8 | HERPUD1-BRAF.H6B7 | RBMS3-BRAF.R11B11 |
| AGTRAP-BRAF.A5B8.COSF828.2 | KANK1-BRAF.K7B9 | RNF11-BRAF.R1B11 |
| AKAP9-BRAF.A21B10 | KCTD7-BRAF.K3B8 | RNF130-BRAF.R3B9.COSF1483 |
| AKAP9-BRAF.A22B9 | KCTD7-BRAF.K4B8 | RP2-BRAF.R3B10 |
| AKAP9-BRAF.A28B9 | KDM7A-BRAF.K11B11 | RUNDC1-BRAF.R1B9 |
| AKAP9-BRAF.A36B9 | KDM7A-BRAF.K12B4 | SCRIB-BRAF.S27B11 |
| AKAP9-BRAF.A45B9 | KIAA1549-BRAF.K11B10 | SLC12A7-BRAF.S17B11 |
| AKAP9-BRAF.A48B8 | KIAA1549-BRAF.K12B11.1 | SLC45A3-BRAF.S1B8.COSF871 |
| AKAP9-BRAF.A7B11 | KIAA1549-BRAF.K12B9.COSF1474.1 | SND1-BRAF.S10B10 |
| AKAP9-BRAF.A8B9.COSF1013.1 | KIAA1549-BRAF.K13B11 | SND1-BRAF.S10B11 |
| AP3B1-BRAF.A22B9 | KIAA1549-BRAF.K13B8 | SND1-BRAF.S10B8 |
| ARMC10-BRAF.A3B9 | KIAA1549-BRAF.K13B9 | SND1-BRAF.S10B9 |
| ARMC10-BRAF.A4B11 | KIAA1549-BRAF.K14B11.COSF1226 | SND1-BRAF.S11B11 |
| ATG7-BRAF.A18B9 | KIAA1549-BRAF.K14B8 | SND1-BRAF.S14B11 |
| BAIAP2L1-BRAF.B12B9 | KIAA1549-BRAF.K14B9.COSF483 | SND1-BRAF.S14B8 |
| BBS9-BRAF.B19B4 | KIAA1549-BRAF.K15B10.COSF1283.2 | SND1-BRAF.S14B9 |
| BCAP29-BRAF.B7B11 | KIAA1549-BRAF.K15B11.COSF485.2 | SND1-BRAF.S16B9.1 |
| BCL2L11-BRAF.B3B10 | KIAA1549-BRAF.K15B9.COSF481.2 | SND1-BRAF.S17B10 |
| BTF3L4-BRAF.B3B11 | KIAA1549-BRAF.K17B10.COSF509 | SND1-BRAF.S17B11 |
| C7orf73-BRAF.C2B9 | KIAA1549-BRAF.K18B9.COSF511 | SND1-BRAF.S17B9 |
| CCDC6-BRAF.C1B9 | KIAA1549-BRAF.K5B9 | SND1-BRAF.S18B10 |
| CCDC6-BRAF.C2B8 | KIAA1549-BRAF.K9B9 | SND1-BRAF.S18B8 |
| CCDC91-BRAF.C11B9 | KLHL7-BRAF.K4B10 | SND1-BRAF.S9B10 |
| CCNY-BRAF.C1B10 | KLHL7-BRAF.K5B9 | SND1-BRAF.S9B11 |

*(continued)*

| BRAF fusion isoforms (N=187) | | |
|---|---|---|
| CDC27-BRAF.C16B9.1 | LMO7-BRAF.L6B9 | SND1-BRAF.S9B2.1 |
| CDC42BPB-BRAF.C36B9 | LSM12-BRAF.L3B9 | SND1-BRAF.S9B8 |
| CDK5RAP2-BRAF.C12B11 | LSM14A-BRAF.L9B9.COSF1652 | SND1-BRAF.S9B9.1 |
| CDK5RAP2-BRAF.C13B8 | MACF1-BRAF.M5B9 | SOX6-BRAF.S5B9 |
| CDK5RAP2-BRAF.C13B9 | MACF1-BRAF.M60B9 | SOX6-BRAF.S6B9 |
| CEP89-BRAF.C16B9.COSF1650 | MAD1L1-BRAF.M16B9 | STRN3-BRAF.S3B10 |
| CLCN6-BRAF.C2B11.COSF1440 | MAD1L1-BRAF.M17B10 | STXBP6-BRAF.S4B10 |
| CLIP2-BRAF.C6B11 | MBP-BRAF.M2B9 | TANK-BRAF.T4B9 |
| COBL-BRAF.C7B10 | MKRN1-BRAF.M3B10 | TAX1BP1-BRAF.T8B11.1 |
| CUL1-BRAF.C7B9 | MKRN1-BRAF.M4B10 | TMEM106B-BRAF.T4B8 |
| CUX1-BRAF.C10B9 | MKRN1-BRAF.M4B11.COSF1444 | TMEM178B-BRAF.T1B10 |
| CUX1-BRAF.C8B8 | MKRN1-BRAF.M4B8 | TMEM178B-BRAF.T2B9 |
| DGKI-BRAF.D7B10 | MKRN1-BRAF.M4B9 | TMPRSS2-BRAF.T3B11 |
| DLG1-BRAF.D3B10 | MYRIP-BRAF.M16B9 | TOM1L2-BRAF.T11B9 |
| DLG1-BRAF.D5B9 | MZT1-BRAF.M2B11 | TPR-BRAF.T14B7 |
| DOCK4-BRAF.D41B9 | NFIC-BRAF.N6B10 | TRIM24-BRAF.T10B9 |
| DSTYK-BRAF.D1B10 | NRF1-BRAF.N5B9 | TRIM24-BRAF.T11B2.1 |
| DYNC1I2-BRAF.D7B10 | NUB1-BRAF.N3B9 | TRIM24-BRAF.T3B10 |
| EML4-BRAF.E6B10 | NUDCD3-BRAF.N4B9 | TRIM24-BRAF.T3B11 |
| EPS15-BRAF.E22B10 | NUP214-BRAF.N21B10 | TRIM24-BRAF.T5B8 |
| EPS15L1-BRAF.E16B9 | OSBPL1A-BRAF.O16B10 | TRIM24-BRAF.T6B9 |
| ERC1-BRAF.E12B10 | OSBPL9-BRAF.O11B10 | TRIM24-BRAF.T8B11 |
| ERC1-BRAF.E17B8 | OSBPL9-BRAF.O14B9 | TRIM24-BRAF.T9B9.1 |
| ESYT2-BRAF.E8B9 | PACSIN2-BRAF.P7B9 | TRIM4-BRAF.T6B10 |
| FAM114A2-BRAF.F9B11 | PAPSS1-BRAF.P5B9.1 | UBN2-BRAF.U3B11 |
| FAM131B-BRAF.F1B10.COSF1191 | PARP12-BRAF.P9B11 | ZBTB8A-BRAF.Z4B9 |
| FAM131B-BRAF.F2B9.COSF1189.2 | PCM1-BRAF.P37B9 | ZC3HAV1-BRAF.Z3B10 |
| FAM131B-BRAF.F3B9.COSF1193.1 | PHTF2-BRAF.P17B9 | ZC3HAV1-BRAF.Z7B11 |
| FAM73A-BRAF.F7B9 | PICALM-BRAF.P19B9 | ZKSCAN5-BRAF.Z2B9 |

*(continued)*

| BRAF fusion isoforms (N=187) | | |
|---|---|---|
| FCHSD1-BRAF.F13B9.COSF403 | PJA2-BRAF.P7B11 | ZNF207-BRAF.Z3B10 |
| FXR1-BRAF.F13B10 | PLIN3-BRAF.P1B9 | ZNF767P-BRAF.Z1B11 |
| GATM-BRAF.G2B11.COSF1731 | PLXNA2-BRAF.P9B2 | ZSCAN30-BRAF.Z3B10.COSF1729 |
| GHR-BRAF.G1B10 | POR-BRAF.P3B11 | |
| GIPC2-BRAF.G3B10 | PPFIBP2-BRAF.P3B11 | |

| ESR1 fusion isoforms (N=32) | | |
|---|---|---|
| ESR1-ARMT1.E3A4 | ESR1-CDK13.E7C13 | ESR1-PLEKHG1.E6P12 |
| ESR1-ARMT1.E6A2 | ESR1-COA5.E4C2 | ESR1-POLH.E6P2 |
| ESR1-ARNT2.E7A9 | ESR1-DAB2.E6D3 | ESR1-RMND1.E6R9 |
| ESR1-ATP2B2.E4A4 | ESR1-GYG2.E6G2 | ESR1-SOX9.E6S3 |
| ESR1-BMPER.E6B8 | ESR1-MKL1.E6M7 | ESR1-SYNE1.E6S59 |
| ESR1-CCDC170.E2C10.1 | ESR1-MTHFD1L.E6M21 | ESR1-TFG.E6T4 |
| ESR1-CCDC170.E2C2 | ESR1-MTHFD1L.E7M27 | ESR1-TNRC6B.E6T14 |
| ESR1-CCDC170.E2C5 | ESR1-NKAIN2.E6N2 | ESR1-YAP1.E6Y4.1 |
| ESR1-CCDC170.E2C6.1 | ESR1-NOP2.E6N16 | PLEKHG1-ESR1.P3E2 |
| ESR1-CCDC170.E2C7.1 | ESR1-PCDH11X.E6P6 | TNS3-ESR1.T16E5 |
| ESR1-CCDC170.E2C8.1 | ESR1-PDE10A.E7P6.1 | |

| FGFR1 fusion isoforms (N=35) | | |
|---|---|---|
| ACPP-FGFR1.A10F4 | FGFR1-EVI5.F17E12 | FN1-FGFR1.F27F5 |
| BAG4-FGFR1.B1F2 | FGFR1-HOOK3.F17H5 | KAT6A-FGFR1.K5F2 |
| BAG4-FGFR1.B1F8.1 | FGFR1OP2-FGFR1.F4F10.1 | LRRFIP1-FGFR1.L8F10.1 |
| BAG4-FGFR1.B2F6.1 | FGFR1OP-FGFR1.F5F10.1 | MYO18A-FGFR1.M33F10.1 |
| BCR-FGFR1.B4F10.1 | FGFR1OP-FGFR1.F6F10.1 | RANBP2-FGFR1.R20F10int9.1 |
| CNTRL-FGFR1.C40F10.1 | FGFR1OP-FGFR1.F7F10.1 | SNX29-FGFR1.S1F4 |
| CPSF6-FGFR1.C8int8F10.1 | FGFR1-TACC1.F17T7.COSF1362.1 | SQSTM1-FGFR1.S6F10.1 |
| CUX1-FGFR1.C11F10.1 | FN1-FGFR1.F22F3 | TPR-FGFR1.T22F10.1 |
| EGFR-FGFR1.E24F10 | FN1-FGFR1.F22F4 | TRIM24-FGFR1.T11F10.1 |
| ERLIN2-FGFR1.E8F2 | FN1-FGFR1.F23F3.1 | WHSC1L1-FGFR1.W14F5.1 |

*(continued)*

| FGFR1 fusion isoforms (N=35) | | |
|---|---|---|
| ERVK3_1-FGFR1.E3F10.1 | FN1-FGFR1.F23F4.1 | ZMYM2-FGFR1.Z17F10.1 |
| FGFR1-ADAM32.F17A14.1 | FN1-FGFR1.F25F4 | |

| FGFR2 fusion isoforms (N=79) | | |
|---|---|---|
| APIP-FGFR2.A1F10 | FGFR2-EIF4A2.F17E8 | FGFR2-SBNO2.F17S19 |
| APIP-FGFR2.A1F6 | FGFR2-FAM76A.F17F2.1 | FGFR2-SHTN1.F17S5 |
| APIP-FGFR2.A6F10 | FGFR2-GAB2.F17G2 | FGFR2-SHTN1.F17S7.1 |
| CD44-FGFR2.C1F3.1 | FGFR2-INA.F17I2 | FGFR2-SMN1.F17S2 |
| COL25A1-FGFR2.C3F3 | FGFR2-KCTD1.F17K2.1 | FGFR2-STK26.F17S3 |
| CTNNB1-FGFR2.C1F10 | FGFR2-KIAA1217.F17K3 | FGFR2-TACC2.F17T11 |
| FGFR2-ACSL5.F17A15 | FGFR2-KIAA1217.F17K5 | FGFR2-TACC3.F17T11.1 |
| FGFR2-AFF3.F17A8.2 | FGFR2-KIAA1217.F17K6 | FGFR2-TBC1D1.F17T9 |
| FGFR2-AHCYL1.F17A2.1 | FGFR2-KRT8.F17K7 | FGFR2-TFEC.F17T2 |
| FGFR2-ANKRD24.F17A3 | FGFR2-LAMC1.F17L27 | FGFR2-TJP2.F17T5 |
| FGFR2-BICC1.F17B10 | FGFR2-MCU.F17M4 | FGFR2-TXLNA.F17T6.1 |
| FGFR2-BICC1.F17B16 | FGFR2-MCU.F17M4.1 | FGFR2-UBQLN1.F17U8 |
| FGFR2-BICC1.F17B18.1 | FGFR2-MGEA5.F17M12.1 | FGFR2-UTRN.F17U51 |
| FGFR2-BICC1.F17B2.1 | FGFR2-NOL4.F17N7.1 | FGFR2-VCL.F17V2 |
| FGFR2-BICC1.F17B3.2 | FGFR2-NRAP.F17N24 | FGFR2-VCL.F17V7 |
| FGFR2-CCDC186.F17C4 | FGFR2-NRBF2.F17N4 | FGFR2-WAC.F17W5 |
| FGFR2-CASP7.F17C2.1 | FGFR2-OFD1.F17O3.1 | FGFR2-WARS.F17W3 |
| FGFR2-CBX5.F17C2 | FGFR2-OGDH.F17O3 | FGFR2-ZMYM4.F17Z10 |
| FGFR2-CCAR2.F17C4.1 | FGFR2-OPTN.F17O5 | KLK2-FGFR2.K1F5 |
| FGFR2-CCDC6.F17C1.1 | FGFR2-PASD1.F17P10 | PARK2-FGFR2.P9F11.1 |
| FGFR2-CCDC6.F17C2.2 | FGFR2-PCM1.F17P7 | PDHX-FGFR2.P1F7.1 |
| FGFR2-CELF2.F17C4 | FGFR2-PHC1.F17P2 | SLC45A3-FGFR2.S1F1 |
| FGFR2-CIT.F17C23.1 | FGFR2-PKD2L1.F17P7 | SLC45A3-FGFR2.S1F2 |
| FGFR2-CLIP2.F17C10 | FGFR2-PPHLN1.F17P3.1 | SNX19-FGFR2.S7F7.1 |
| FGFR2-COL14A1.F17C34.1 | FGFR2-RABGAP1L.F17R20 | SNX19-FGFR2.S8F7.1 |

*(continued)*

| FGFR2 fusion isoforms (N=79) | |
|---|---|
| FGFR2-CREB5.F17C8.1 | FGFR2-RASAL2.F17R3 |
| FGFR2-DDX21.F17D11 | FGFR2-ROCK1.F17R2 |

| FGFR3 fusion isoforms (N=42) | | |
|---|---|---|
| ETV6-FGFR3.E5F9.1 | FGFR3-TACC3.F17ins7T10 | FGFR3-TACC3.F17T6 |
| FGFR3-ADD1.F17A3 | FGFR3-TACC3.F17Int6ins36T7del4.1 | FGFR3-TACC3.F17T7 |
| FGFR3-AES.F17A2 | FGFR3-TACC3.F17Int9T11.1 | FGFR3-TACC3.F17T7.1 |
| FGFR3-AMBRA1.F17A14 | FGFR3-TACC3.F17intron17T4.1 | FGFR3-TACC3.F17T8.COSF1353 |
| FGFR3-BAIAP2L1.F17B2.COSF1346 | FGFR3-TACC3.F17Intron17T9.1 | FGFR3-TACC3.F17T9.2 |
| FGFR3-BRAP.F17B5 | FGFR3-TACC3.F17T10.COSF1434 | FGFR3-TACC3.F18Int136T11.1 |
| FGFR3-CAMK2A.F18C14 | FGFR3-TACC3.F17T11.1 | FGFR3-TACC3.F18T1.1 |
| FGFR3-ELAVL3.F17E2 | FGFR3-TACC3.F17T11.2 | FGFR3-TACC3.F18T10.1 |
| FGFR3-FBXO28.F17F4 | FGFR3-TACC3.F17T11.COSF1348.1 | FGFR3-TACC3.F18T10.2 |
| FGFR3-JAKMIP1.F17J4.1 | FGFR3-TACC3.F17T13.NGS | FGFR3-TACC3.F18T11del5.1 |
| FGFR3-NBR1.F17N7 | FGFR3-TACC3.F17T14 | FGFR3-TACC3.F18T4and5.1 |
| FGFR3-RBBP4.F17R12 | FGFR3-TACC3.F17T14.1 | FGFR3-TACC3.F18T7.NGS |
| FGFR3-TACC3.F16T4 | FGFR3-TACC3.F17T4 | FGFR3-TACC3.F18T9 |
| FGFR3-TACC3.F17ins1T10 | FGFR3-TACC3.F17T4.1 | FGFR3-TACC3.TruncatedF17T4 |

| MET fusion isoforms (N=31) | | |
|---|---|---|
| BAIAP2L1-MET.B9M15.1 | KIF5B-MET.K24M15 | ST7-MET.S1M11 |
| C8orf34-MET.C2M15 | LRRFIP1-MET.L19M15.1 | ST7-MET.S1M2 |
| CAPZA2-MET.C1M6.1 | OXR1-MET.O1M13 | STARD3NL-MET.S6M15 |
| CAPZA2-MET.C2M15 | OXR1-MET.O9M13 | TFG-MET.T5M15 |
| CAPZA2-MET.C4M11.1 | PPFIBP1-MET.P9M15.1 | TFG-MET.T7M15 |
| CLIP2-MET.C11M15 | PTPRZ1-MET.P1M2.1 | TPR-MET.T36M15 |
| DCTN1-MET.D26M15.1 | PTPRZ1-MET.P2M2 | TPR-MET.T4M15 |
| EPS15-MET.E21M15 | PTPRZ1-MET.P3M2.1 | TRIM4-MET.T6M15 |
| ETV6-MET.E4M15 | PTPRZ1-MET.P8M2.1 | ZKSCAN1-MET.Z3M15 |

*(continued)*

| MET fusion isoforms (N=31) | |
|---|---|
| GTF2I-MET.G6M15 | SCAF11-MET.S1M2 |
| HLA_DRB1-MET.H5M15 | SLC34A2-MET.S4M15 |

| NRG1 fusion isoforms (N=35) | | |
|---|---|---|
| AKAP13-NRG1.A5N2 | NOTCH2-NRG1.N4N6 | TBL1XR1-NRG1.T1N2 |
| ATP1B1-NRG1.A2N2 | PARP8-NRG1.P1N2 | TENM4-NRG1.T12N2.1 |
| CD74-NRG1.C6N3 | PDE7A-NRG1.P3N6.1 | THAP7-NRG1.T3N2 |
| CD74-NRG1.C6N6.1 | RAB3IL1-NRG1.R9N6 | THBS1-NRG1.T6N6 |
| CD74-NRG1.C7N2 | RALGAPA1-NRG1.R20N6 | TNC-NRG1.T10N6 |
| CD74-NRG1.C7N6 | ROCK1-NRG1.R2N2 | TSHZ2-NRG1.T1N6 |
| CD74-NRG1.C7N6.COSF1666 | SDC4-NRG1.S2N6 | VAMP2-NRG1.V4N4.1 |
| CDH1-NRG1.C2N2 | SDC4-NRG1.S4N6 | VTCN1-NRG1.V2N2 |
| CLU-NRG1.C2N6 | SETD4-NRG1.S2N2 | WHSC1L1-NRG1.W1N2 |
| COX10-AS1-NRG1.C1N2 | SLC3A2-NRG1.S2N2 | WRN-NRG1.W30N2 |
| HMBOX1-NRG1.H1N6 | SLC3A2-NRG1.S5N6 | ZMYM2-NRG1.Z2N2 |
| KIF13B-NRG1.K2N2 | SMAD4-NRG1.S1N6 | |

| NTRK1 fusion isoforms (N=89) | | |
|---|---|---|
| AFAP1-NTRK1.A4N10 | LMNA-NTRK1.L3N11 | SQSTM1-NTRK1.S5N12 |
| ARHGEF25-NTRK1.A14N8 | LMNA-NTRK1.L4N12 | SSBP2-NTRK1.S12N12 |
| ARHGEF2-NTRK1.A21N10 | LMNA-NTRK1.L4N12.1 | TFG-NTRK1.T4N9 |
| BANP-NTRK1.B4N10 | LMNA-NTRK1.L5del90N11 | TFG-NTRK1.T5N10.COSF1328 |
| BCAN-NTRK1.B12N10 | LMNA-NTRK1.L5N10 | TFG-NTRK1.T6N10.1 |
| BCAN-NTRK1.B13N11 | LMNA-NTRK1.L5N11 | TFG-NTRK1.T7N12 |
| CD74-NTRK1.C7N10 | LMNA-NTRK1.L6N12 | TP53-NTRK1.T10N9.COSF1658 |
| CEL-NTRK1.C7N9 | LMNA-NTRK1.L6N12.1 | TP53-NTRK1.T11N9.COSF1660 |
| CHTOP-NTRK1.C5N10 | LRRC71-NTRK1.L1N9 | TP53-NTRK1.T8N9.COSF1655 |
| CHTOP-NTRK1.C5N11.1 | MDM4-NTRK1.M3N5 | TP53-NTRK1.T9N9.COSF1657 |
| CTRC-NTRK1.C1N9 | MPRIP-NTRK1.M21N12 | TPM3-NTRK1.T6N11 |
| EPHB2-NTRK1.E3N9 | MRPL24-NTRK1.M1N9 | TPM3-NTRK1.T7N10.COSF1329 |

*(continued)*

| NTRK1 fusion isoforms (N=89) | | |
|---|---|---|
| EPS15-NTRK1.E21N9 | MRPL24-NTRK1.M1N9.1 | TPM3-NTRK1.T7N12 |
| EPS15-NTRK1.E21N9.1 | NFASC-NTRK1.N20N10 | TPM3-NTRK1.T7N13 |
| GON4L-NTRK1.G21N9 | NTRK1-DYNC2H1.N16D85 | TPM3-NTRK1.T7N7.ins.1 |
| GRIPAP1-NTRK1.G22N11 | NTRK1-DYNC2H1.N17D85 | TPM3-NTRK1.T7N9 |
| GSN-NTRK1.G7N10 | PEAR1-NTRK1.P15N10 | TPM3-NTRK1.T7N9b |
| IRF2BP2-NTRK1.I1N10 | PEAR1-NTRK1.P2N9 | TPM3-NTRK1.T8N10 |
| IRF2BP2-NTRK1.I1N10.1 | PLEKHA6-NTRK1.P16N10 | TPR-NTRK1.T10N10 |
| IRF2BP2-NTRK1.I1N11 | PLEKHA6-NTRK1.P21N9 | TPR-NTRK1.T10N4 |
| IRF2BP2-NTRK1.I1N12 | PPL-NTRK1.P12N13.1 | TPR-NTRK1.T16int9N10.1 |
| IRF2BP2-NTRK1.I1N9 | PPL-NTRK1.P22N10.1 | TPR-NTRK1.T21N10.COSF1326 |
| IRF2BP2-NTRK1.I1N9.1 | PPL-NTRK1.P22N11.1 | TPR-NTRK1.T21N12 |
| LMNA-NTRK1.L10N11.1 | PPL-NTRK1.P22N13 | TPR-NTRK1.T21N13 |
| LMNA-NTRK1.L10N12.1 | RABGAP1L-NTRK1.R14N16 | TPR-NTRK1.T21N9 |
| LMNA-NTRK1.L10N13 | RFWD2-NTRK1.R2N3 | TPR-NTRK1.T6N12.COSF1324 |
| LMNA-NTRK1.L11N11.1 | RNF213-NTRK1.R15N12 | TPR-NTRK1.T8N11 |
| LMNA-NTRK1.L2N10 | SCYL3-NTRK1.S11N12 | TRIM63-NTRK1.T8N9 |
| LMNA-NTRK1.L2N11 | SQSTM1-NTRK1.S2N10.1 | VANGL2-NTRK1.V1N12 |
| LMNA-NTRK1.L3N10 | SQSTM1-NTRK1.S5N10.1 | |

| NTRK2 fusion isoforms (N=31) | | |
|---|---|---|
| AFAP1-NTRK2.A14N12 | NAV1-NTRK2.N15N11.1 | TBC1D2-NTRK2.T6N12 |
| AGBL4-NTRK2.A6N16.1 | NOS1AP-NTRK2.N9N11 | TLE4-NTRK2.T7N15 |
| BCR-NTRK2.B1N17 | PAN3-NTRK2.P1N17 | TP63-NTRK2.T8N12.1 |
| DAB2IP-NTRK2.D1N17 | QKI-NTRK2.Q6N16.COSF1446 | TRAF2-NTRK2.T9N15 |
| ETV6-NTRK2.E4N16 | SLMAP-NTRK2.S14N16 | TRIM24-NTRK2.T12N15.1 |
| ETV6-NTRK2.E5N15 | SQSTM1-NTRK2.S5N13 | TRIM24-NTRK2.T12N16 |
| ETV6-NTRK2.E5N16 | SQSTM1-NTRK2.S5N17.1 | VCAN-NTRK2.V6N10 |
| GKAP1-NTRK2.G9N13 | STRN3-NTRK2.S7N16 | VCL-NTRK2.V16N12.1 |
| GNAQ-NTRK2.G2N16 | STRN-NTRK2.S14N5 | WNK2-NTRK2.W24N16 |

*(continued)*

| NTRK2 fusion isoforms (N=31) | | |
|---|---|---|
| KCTD8-NTRK2.K1N13 | STRN-NTRK2.S3N15 | |
| NACC2-NTRK2.N4N13.COSF1448 | STRN-NTRK2.S3N16 | |

| NTRK3 fusion isoforms (N=33) | | |
|---|---|---|
| AKAP13-NTRK3.A14N14.1 | FBXL20-NTRK3.F1N6 | NTRK3-HOMER1.N17H2 |
| AKAP13-NTRK3.A16N15 | HNRNPA2B1-NTRK3.H7N14 | NTRK3-HOMER2.N17H2 |
| BTBD1-NTRK3.B4N14 | KANK1-NTRK3.K3N14 | NTRK3-LOXL2.N3L5 |
| COX5A-NTRK3.C1N15.1 | LYN-NTRK3.L8N14.1 | RBPMS-NTRK3.R5N14 |
| EML4-NTRK3.E2N14.1 | MYH9-NTRK3.M32N14 | SQSTM1-NTRK3.S5N14 |
| EML4-NTRK3.E6N14 | MYO5A-NTRK3.M23N11 | TFG-NTRK3.T6N10 |
| ETV6-NTRK3.E4N14.1 | MYO5A-NTRK3.M23N12 | TFG-NTRK3.T6N14 |
| ETV6-NTRK3.E4N15.COSF823.2 | MYO5A-NTRK3.M23N14 | TFG-NTRK3.T6N15 |
| ETV6-NTRK3.E5N14.1 | MYO5A-NTRK3.M32N12 | TPM4-NTRK3.T7N12 |
| ETV6-NTRK3.E5N15.COSF571.2 | MYO5A-NTRK3.M32N13 | UBE2R2-NTRK3.U3N4 |
| FAT1-NTRK3.F2N7 | MYO5A-NTRK3.M32N14 | ZNF710-NTRK3.Z1N14 |

| NUTM1 fusion isoforms (N=16) | | |
|---|---|---|
| AFF1-NUTM1.A17N3 | BRD4-NUTM1.B14N2del585 | CIC-NUTM1.C20N5 |
| BCORL1-NUTM1.B1N2 | BRD4-NUTM1.B15N2.1 | MXD1-NUTM1.M5N2 |
| BRD3-NUTM1.B10N2.COSF973.1 | CIC-NUTM1.C16N4.1 | WHSC1L1-NUTM1.W7N2.1 |
| BRD3-NUTM1.B10N5 | CIC-NUTM1.C17N5 | ZNF618-NUTM1.Z12N2 |
| BRD3-NUTM1.B9N4 | CIC-NUTM1.C18N2 | |
| BRD4-NUTM1.B11N2.COSF780.1 | CIC-NUTM1.C18N5 | |

| RET fusion isoforms (N=89) | | |
|---|---|---|
| ABI3BP-RET.A34R12 | FGFR1OP-RET.F11R11 | OLFM4-RET.O4R10 |
| ACBD5-RET.A11R12 | FGFR1OP-RET.F12R12 | PCM1-RET.P29R12.COSF1481.1 |
| ADD3-RET.A1R12 | FKBP15-RET.F25R12 | PDCD10-RET.P4R11 |
| AFAP1L2-RET.A2R12 | FRMD4A-RET.F12R12 | PICALM-RET.P19R12 |
| AFAP1L2-RET.A6R12 | GOLGA5-RET.G7R12.COSF1503.1 | PPFIBP2-RET.P9R12 |

*(continued)*

| RET fusion isoforms (N=89) | | |
|---|---|---|
| AFAP1-RET.A3R12 | GPHN-RET.G9R12 | PRKAR1A-RET.P7R12.COSF1511.1 |
| AKAP13-RET.A35R12 | HOOK3-RET.H11R12.COSF1509.1 | PRKG1-RET.P3R12 |
| ANK3-RET.A1R12 | KIAA1217-RET.K11R10 | RASAL2-RET.R15R12 |
| BCR-RET.B4R12 | KIAA1217-RET.K11R11 | RASSF4-RET.R3R12 |
| CCDC186-RET.C7R12 | KIAA1468-RET.K10R12 | RRBP1-RET.R9R12 |
| CCDC6-RET.C1R10 | KIF13A-RET.K18R12 | RUFY1-RET.R17R12 |
| CCDC6-RET.C1R11 | KIF5B-RET.K15R11.COSF1255.2 | RUFY2-RET.R9R12 |
| CCDC6-RET.C1R11.1 | KIF5B-RET.K15R12.COSF1232.1 | SATB1-RET.S7R12 |
| CCDC6-RET.C1R12.COSF1271.1 | KIF5B-RET.K16R12.COSF1230.1 | SNRNP70-RET.S7R12 |
| CCDC6-RET.C1R9 | KIF5B-RET.K18R12 | SPECC1L-RET.S10R11.NGS.1 |
| CCDC6-RET.C2R11 | KIF5B-RET.K22R12.COSF1253.1 | SPECC1L-RET.S10R12 |
| CCDC6-RET.C2R12 | KIF5B-RET.K23R12.COSF1234.1 | SQSTM1-RET.S5R11 |
| CCDC6-RET.C5ins16R11.1 | KIF5B-RET.K23R8 | SQSTM1-RET.S6R12 |
| CCDC6-RET.C8R11 | KIF5B-RET.K24R11.COSF1262.1 | TBL1XR1-RET.T9R11.NGS.1 |
| CCDC6-RET.C8R12 | KIF5B-RET.K24R8.COSF1236.1 | TBL1XR1-RET.T9R12 |
| CUX1-RET.C10R12 | KTN1-RET.K29R12.COSF1513.1 | TFG-RET.T4R11 |
| DLG5-RET.D13R12 | MPRIP-RET.M19R12 | TNIP1-RET.T11R12 |
| EML4-RET.E19R11 | MYH10-RET.M33R12 | TNIP2-RET.T5R12 |
| EML4-RET.E19R12 | MYH13-RET.M35R12 | TRIM24-RET.T9R12.COSF1521.1 |
| EPHA5-RET.E2R12 | MYO5A-RET.M23R11 | TRIM27-RET.T3R12.COSF1519.1 |
| ERC1-RET.E11R12.COSF1507.1 | MYO5C-RET.M25R12 | TRIM33-RET.T11R12 |
| ERC1-RET.E12R12 | NCOA4-RET.N6R12.COSF1340.1 | TRIM33-RET.T14R12 |
| ERC1-RET.E17R12 | NCOA4-RET.N7R12.COSF1491.1 | TRIM33-RET.T16R12.COSF1525.1 |
| ERC1-RET.E7R12.1 | NCOA4-RET.N9R12 | WAC-RET.W3R12 |
| ETV6-RET.E6R12 | NCOA4-RET.N9R8 | |

| ROS1 fusion isoforms (N=57) | | |
|---|---|---|
| CCDC30-ROS1.C10R36 | GOPC-ROS1.G8R35.COSF1139.1 | SLC22A16-ROS1.S7R16 |
| CCDC6-ROS1.C5R35.1 | HLA_A-ROS1.H7R34 | SLC34A2-ROS1.S13R32.COSF1259 |
| CD74-ROS1.C4R33.NGS | LIMA1-ROS1.L10R36 | SLC34A2-ROS1.S13R33 |
| CD74-ROS1.C6R32.COSF1202.1 | LRIG3-ROS1.L16R35.COSF1269.1 | SLC34A2-ROS1.S13R34.COSF1261 |
| CD74-ROS1.C6R33 | MSN-ROS1.M9R34.1 | SLC34A2-ROS1.S13R36 |
| CD74-ROS1.C6R34.COSF1200.1 | MYO5A-ROS1.M23R35.1 | SLC34A2-ROS1.S4R32.COSF1196 |
| CD74-ROS1.C6R35.1 | MYO5A-ROS1.M32R35 | SLC34A2-ROS1.S4R34.COSF1198 |
| CD74-ROS1.C7R34 | MYO5C-ROS1.M30R35 | SLC34A2-ROS1.S4R35 |
| CEP85L-ROS1.C12R35 | NCOR2-ROS1.N7R36.1 | SLMAP-ROS1.S10R35 |
| CEP85L-ROS1.C8R36 | NFKB2-ROS1.N14R36 | TFG-ROS1.T4R35.1 |
| CLIP1-ROS1.C19R36 | PPFIBP1-ROS1.P9R35.1 | TFG-ROS1.T5R35 |
| CLTC-ROS1.C31R35 | PWWP2A-ROS1.P1R36 | TMEM106B-ROS1.T3R35.1 |
| ERC1-ROS1.E11R36 | SDC4-ROS1.S2R32.COSF1265 | TPD52L1-ROS1.T3R33 |
| EYS-ROS1.E12R9 | SDC4-ROS1.S2R34 | TPM3-ROS1.T3R36.1 |
| EZR-ROS1.E10R33 | SDC4-ROS1.S4R32.COSF1278 | TPM3-ROS1.T7R35.COSF1273.1 |
| EZR-ROS1.E10R34.COSF1267 | SDC4-ROS1.S4R34.COSF1280 | TSPAN3-ROS1.T5R31 |
| EZR-ROS1.E10R35.1 | SDC4-ROS1.S5R34 | YWHAE-ROS1.Y4R36 |
| GOLGB1-ROS1.G9R35 | SHTN1-ROS1.S11R36 | ZCCHC8-ROS1.Z2R36 |
| GOPC-ROS1.G4R36.COSF1188 | SLC16A10-ROS1.S3R3 | ZCCHC8-ROS1.Z3R36 |

| RSPO2 fusion isoforms (N=4) | | |
|---|---|---|
| EIF3E-RSPO2.E1R2.COSF1307.1 | NRIP1-RSPO2.N2R2 | NRIP1-RSPO2.N3R2 |
| GRHL2-RSPO2.G8R2 | | |

| RSPO3 fusion isoforms (N=5) | | |
|---|---|---|
| ASH1L-RSPO3.A1R2 | PTPRK-RSPO3.P1R2.COSF1311.1 | PTPRK-RSPO3.P7R2.COSF1313.1 |
| PTPRK-RSPO3.P10R2 | PTPRK-RSPO3.P6R2 | |

# RNA splicing variants targeted by the Oncomine™ Dx Express Test

| Gene | Fusion |
|---|---|
| AR | AR-AR.A3A4.V7 |
| EGFR | EGFRvIII.E1E8.Del1 |
| MET | MET-MET.M13M15.1 |
| | MET-MET.M21M15 |

# Fusions targeted by RNA exon tile fusion imbalance analysis

Non-targeted fusions in the following genes can be detected with Genexus™ Dx Software using RNA exon tile fusion imbalance analysis. The RNA exon tile amplicons used in the analysis for each gene are listed in the table. RNA exon tile fusion imbalance analysis is enabled for FFPE samples only. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.6 Reference User Guide* (MAN0024910).

| Gene | RNA exon tile amplicon set | |
|---|---|---|
| ALK | ALK.I.A2A3 | ALK.I.A13A14 |
| | ALK.I.A3A4 | ALK.I.A14A15 |
| | ALK.I.A4A5 | ALK.I.A15A16 |
| | ALK.I.A6A7 | ALK.I.A20A21 |
| | ALK.I.A8A9 | ALK.I.A26A27 |
| | ALK.I.A10A11 | ALK.I.A28A29 |
| FGFR1 | FGFR1.I.F3F4 | FGFR1.I.F10F11 |
| | FGFR1.I.F4F5 | FGFR1.I.F11F12 |
| | FGFR1.I.F5F6 | FGFR1.I.F12F13 |
| | FGFR1.I.F6F7 | FGFR1.I.F15F16 |
| | FGFR1.I.F8F9 | FGFR1.I.F16F17 |
| FGFR2 | FGFR2.I.F3F4 | FGFR2.I.F16F17 |
| | FGFR2.I.F7F8 | FGFR2-FGFR2.F18F18.1 |
| | FGFR2.I.F8F9 | FGFR2-FGFR2.F18F18.3 |
| | FGFR2.I.F11F12 | FGFR2-FGFR2.F18F18.7 |
| | FGFR2.I.F15F16 | |
| FGFR3 | FGFR3.I.F3F4 | FGFR3-FGFR3.F18F18.1 |
| | FGFR3.I.F11F12 | FGFR3-FGFR3.F18F18.4 |
| | FGFR3.I.F12F13 | FGFR3-FGFR3.F18F18.8 |
| | FGFR3.I.F13F14 | |
| NTRK1 | NTRK1.I.N1N2 | NTRK1.I.N6N7 |
| | NTRK1.I.N2N3 | NTRK1.I.N13N14 |
| | NTRK1.I.N4N5 | NTRK1.I.N15N16 |
| | NTRK1.I.N5N6 | NTRK1.I.N16N17 |

*(continued)*

| Gene | RNA exon tile amplicon set | |
|---|---|---|
| NTRK2 | NTRK2.I.N1N2 | NTRK2.I.N9N10 |
| | NTRK2.I.N2N3 | NTRK2.I.N13N14 |
| | NTRK2.I.N4N5 | NTRK2.I.N17N18 |
| | NTRK2.I.N6N7 | NTRK2.I.N19N20 |
| | NTRK2.I.N8N9 | NTRK2.I.N20N21 |
| NTRK3 | NTRK3.I.N2N3 | NTRK3.I.N12N13 |
| | NTRK3.I.N3N4 | NTRK3.I.N13N14 |
| | NTRK3.I.N5N6 | NTRK3.I.N14N15 |
| | NTRK3.I.N7N8 | NTRK3.I.N15N16 |
| | NTRK3.I.N8N9 | NTRK3.I.N17N18 |
| | NTRK3.I.N9N10 | NTRK3.I.N18N19 |
| | NTRK3.I.N11N12 | |
| HMBS (control) | HMBS.I.H2H3 | HMBS.I.H11H12 |
| | HMBS.I.H5H6 | HMBS.I.H12H13 |
| | HMBS.I.H6H7 | HMBS.I.H13H14 |
| | HMBS.I.H9H10 | |

# Copy number variants targeted by the Oncomine™ Dx Express Test

| Gene / Mutation ID | Display name |
|---|---|
| AR | AR amplification |
| EGFR | EGFR amplification |
| ERBB2 | ERBB2 amplification |
| ERBB3 | ERBB3 amplification |
| FGFR1 | FGFR1 amplification |
| FGFR2 | FGFR2 amplification |
| FGFR3 | FGFR3 amplification |
| KRAS | KRAS amplification |
| MET | MET amplification |
| PIK3CA | PIK3CA amplification |

# Biomarker level 3 SNV variants masked at low allelic frequency

Fifteen unique level 3 SNVs with observed variant allelic frequency below 5% (Table 41) are prospectively masked in reporting.

**Table 41 Variants masked with AF <5%**

| Gene | Variant | Nucleotide change | Variant ID |
|---|---|---|---|
| ALK | ALK T1151M | c.3452C>T | COSM98478 |
| EGFR | EGFR R451C | c.1351C>T | COSM3266583 |
| EGFR | EGFR V769M | c.2305G>A | COSM13425 |
| ESR1 | ESR1 R555C* | c.1663C>T | BT32 |
| FGFR2 | FGFR2 A648T | c.1942G>A | COSM29836 |
| KEAP1 | KEAP1 D422N | c.1264G>A | COSM710198 |
| KEAP1 | KEAP1 R470C | c.1408C>T | COSM564847 |
| KEAP1 | KEAP1 R470H | c.1409G>A | COSM1524064 |
| KEAP1 | KEAP1 p.? | c.640-1G>A | COSM6972470 |
| KIT | KIT L576F | c.1726C>T | COSM33966 |
| KRAS | KRAS G12V | c.35G>T | COSM520 |
| PIK3CA | PIK3CA R38H | c.113G>A | COSM745 |
| PIK3CA | PIK3CA R108H | c.323G>A | COSM27497 |
| TP53 | TP53 R175H | c.524G>A | COSM10648 |
| TP53 | TP53 G266V | c.797G>T | COSM10958 |

# C Customer and technical support

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- Order and web support
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Report any cybersecurity incident that has occurred in relation to the device to Thermo Fisher Scientific. Contact Customer Support at **thermofisher.com/support** or **Contact Us**.

ion torrent

# Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports

## USER GUIDE

for use with:

Genexus™ Dx Integrated Sequencer

Genexus™ Dx Purification System

Genexus™ Dx Software 6.8

Publication Number MAN0024909

Revision A

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Corporation |
7335 Executive Way |
Frederick, Maryland 21704 USA

Products manufactured at this site:

- Oncomine™ Dx Express Test
- Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit
- Genexus™ Dx FFPE DNA/RNA Purification Kit
- Genexus™ Dx Library Strips 1 and 2-HD
- Genexus™ Dx Templating Strips 3-GX5™ and 4
- Genexus™ Dx Sequencing Kit
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Pipette Tips
- Genexus™ Dx Filter
- Genexus™ Dx Conical Bottles

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
#07-06, Singapore 739256

Products manufactured at this site:

- Genexus™ Dx Purification System
- Genexus™ Dx Integrated Sequencer
- Genexus™ Dx Software

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.

Revision history: MAN0024909 A (English)

| Revision | Date | Description |
|---|---|---|
| A | 25 June 2025 | New IVD-labeled part 2 user guide for the Oncomine™ Dx Express Test. |

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# About this guide

CAUTION! **ABBREVIATED SAFETY ALERTS.** Hazard symbols and hazard types specified in procedures may be abbreviated in this document. For the complete safety information, see the "Safety" appendix in this document.

IMPORTANT! Before using this product, read and understand the information in the "Safety" appendix in this document.

## Purpose of this guide

This user guide provides instructions for sample entry and run planning in Genexus™ Dx Software, purifying samples on the Genexus™ Dx Purification System, starting a sequencing run on the Genexus™ Dx Integrated Sequencer, and reviewing results obtained with the Oncomine™ Dx Express Test. This guide also provides troubleshooting, instrument maintenance, and other information.

## Oncomine™ Dx Express Test user guides

This user guide is part of a three-guide set.

- *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide*
- *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*
- *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*

All three guides are required to complete the entire Oncomine™ Dx Express Test workflow.

# 1 Product information

IMPORTANT! Before using this product, read and understand the information in the “Safety” appendix in this document.

## Oncomine™ Dx Express Test

The Ion Torrent™ Oncomine™ Dx Express Test (Cat. No. A54103) is ordered as a set of primer panel tubes sufficient for sequencing up to 28 sample libraries starting from FFPE tissue sections. Accompanying controls, reagents, and supplies for running samples on the Genexus™ Dx Integrated Sequencer are ordered separately, and are described in the following pages.

For a complete description of the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

## Intended use

For the intended use statement for the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

## Theory of operation

For a complete description of the theory of operation, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

## Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software version 6.8.2 or later. For a complete description of software compatibility and requirements, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910).

# Contents and storage

The Oncomine™ Dx Express Test Panel and Oncomine™ Dx Express Test control kits are ordered separately.

Note: Consumables that have catalog numbers are orderable. Components that have part numbers cannot be ordered individually.

## Oncomine™ Dx Express Test

The Oncomine™ Dx Express Test (Cat. No. A54103) is provided in 8 pairs of tubes, where each tube carrier contains one tube with forward (FWD) primers and one tube with reverse (REV) primers. The amount of panel provided is sufficient for up to 32 reactions (samples + controls).

| Contents | Carrier color | Amount | Storage |
|---|---|---|---|
| Oncomine™ Dx Express Test Panel | | | |
| Oncomine™ Dx Express FWD (position 1, amber tube) | Magenta | 8 × 2 tubes, 50 µL/tube | –30°C to –10°C |
| Oncomine™ Dx Express REV (position 2, natural tube) | | | |

## Oncomine™ Dx Express Test controls

Controls available for use with the Oncomine™ Dx Express Test are listed in the following table. The Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit is ordered separately.

| Control | Cap color | Part No. | Amount | Storage |
|---|---|---|---|---|
| Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit (Cat. No. A52167) | | | | |
| Oncomine™ Dx Express Test FFPE DNA Control | Brown | A50834 | 4 × 30 µL | –30°C to –10°C |
| Oncomine™ Dx Express Test FFPE RNA Control | White | A50835 | 4 × 30 µL | –90°C to –60°C |
| Oncomine™ Dx Express Test NTC Control | Purple | A50836 | 4 × 30 µL | 15°C to 30°C |

# Required materials

The following reagents, supplies, and equipment are required to use the Oncomine™ Dx Express Test. The items are ordered separately.

Note: Consumables that have catalog numbers are orderable. Components that have part numbers cannot be ordered individually.

## Genexus™ Dx sample purification kits

The following kits are available for purifying nucleic acids from formalin-fixed, paraffin-embedded (FFPE) tissue specimens on the Genexus™ Dx Purification System.

### Genexus™ Dx FFPE DNA/RNA Purification Kit

The Ion Torrent™ Genexus™ Dx FFPE DNA/RNA Purification Kit (Cat. No. A52170) provides reagents and consumables for 48 DNA and RNA isolations.

IMPORTANT! Store all kit components in the upright orientation.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx FFPE DNA and RNA Purification (Part No. A51076) | | |
| Genexus™ Dx FFPE DNA and RNA Purification Plate 1 | 4 plates | 15°C to 30°C |
| Genexus™ Dx FFPE DNA and RNA Purification Plate 2 | 4 plates | |
| Genexus™ Dx 12-Well Tip Comb | 4 each | |
| Genexus™ Dx DNase | 115 µL | |
| Genexus™ Dx DNase Buffer | 4 × 1.2 mL | |
| Genexus™ Dx Proteinase K | 1.2 mL | |
| Genexus™ Dx FFPE Protease Buffer | 15 mL | |
| Genexus™ Dx Nucleic Acid Quantitation (Part No. A51078) | | |
| Genexus™ Dx Quantitation Plate[1] | 4 plates | 2°C to 8°C |
| Genexus™ Dx Quantitation Tubes[2] | 4 each | 15°C to 30°C |
| Genexus™ Dx Purification Supplies 2 (Part No. A51081) | | |
| Genexus™ Dx 48-Well Nucleic Acid Archive Plate | 4 plates | 15°C to 30°C |
| Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal | 4 each | |
| Genexus™ Dx Purification Tip Cartridge | 8 each | |

*(continued)*

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx AutoLys M Tubes | 2 × 24 each | 15°C to 30°C |
| Genexus™ Dx AutoLys M Caps | 2 × 24 each | |

[1] Store the Genexus™ Dx Quantitation Plate in the dark to prevent photobleaching of the preloaded reagents.
[2] Can be stored at 15°C to 30°C upon receipt.

## Genexus™ Dx Library Strips 1 and 2-HD

Ion Torrent™ Genexus™ Dx Library Strips 1 and 2-HD (Cat. No. A50430) are ordered as kits with 8 pairs of strips per kit.

| Component | Carrier color | Part No. | Amount per kit | Storage |
|---|---|---|---|---|
| Genexus™ Dx Strip 1 | Light red | A50862 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 2-HD | Violet | A50861 | 8 strips | −30°C to −10°C |

## Genexus™ Dx Templating Strips 3-GX5™ and 4

Ion Torrent™ Genexus™ Dx Templating Strips 3-GX5™ and 4 (Cat. No. A50431) are ordered as kits with 8 pairs of strips per kit.

| Component | Carrier color | Part No. | Amount per kit | Storage |
|---|---|---|---|---|
| Genexus™ Dx Strip 3-GX5™ | Brown | A50864 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 4 | Yellow | A50863 | 8 strips | −30°C to −10°C |

## Genexus™ Dx Barcodes 1–32 HD

Ion Torrent™ Genexus™ Dx Barcodes 1–32 HD (Cat. No. A50410) are supplied in a plate containing 32 dual barcodes.

| Component | Label color | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx Barcodes 1–32 HD | Violet | 1 plate | 15°C to 30°C |

## Genexus™ Dx GX5™ Chip and Genexus™ Coupler

The Genexus™ Dx GX5™ Chip and Genexus™ Coupler (Cat. No. A54106) are ordered as a set that contains two chips and two couplers, sufficient for up to eight sequencing runs.

| Component | Part No. | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx GX5™ Chip | 100097356 | 2 chips | 15°C to 30°C |
| Genexus™ Coupler | 100081252 | 2 couplers | |

## Genexus™ Dx Sequencing Kit

The Ion Torrent™ Genexus™ Dx Sequencing Kit (Cat. No. A50432) provides reagents and solutions sufficient to sequence up to 2 full chips.

| Component | Part No. | Amount | Storage |
|---|---|---|---|
| Genexus™ Dx Cartridge | A50865 | 2 cartridges | −30°C to −10°C |
| Genexus™ Dx Bottle 2 | A50866 | 4 bottles | 15°C to 30°C |
| Genexus™ Dx Bottles 1 and 3 | A50867 | 2 bottles each (4 bottles total) | |

## Genexus™ Dx Pipette Tips

Ion Torrent™ Genexus™ Dx Pipette Tips (Cat. No. A50426) are ordered individually.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Pipette Tips | 12 racks | 15°C to 30°C |

## Genexus™ Dx Conical Bottles

Genexus™ Dx Conical Bottles (Cat. No. A50869) are installed in the sequencing reagents bay and serve as reservoirs for nucleotide reagent dilutions. For information on when and how to change the conical bottles, see “Replace the Genexus™ Dx Conical Bottles” on page 211.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Conical Bottles | 5 bottles | 15°C to 30°C |

## Genexus™ Dx Filter

The Genexus™ Dx Filter (Cat. No. A50868) is installed in the liquid waste disposal port on the instrument deck to prevent liquid waste line blockage. For information about installation, see “Replace the Genexus™ Dx Filter” on page 211.

| Component | Amount | Storage |
|---|---|---|
| Genexus™ Dx Filter | 2 filters | 15°C to 30°C |

## Handheld Barcode Scanner

The Handheld Barcode Scanner can be used to scan extraction and control kit package barcodes during run planning.

| Component | Amount | Cat. No. |
|---|---|---|
| Handheld Barcode Scanner (Zebra Technologies Corp. Model No. DS4608) | 1 each | 4488442 |

## Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer

| Components | Cat. No. |
|---|---|
| Genexus™ Dx Purification System instrument | A53257 |
| Genexus™ Dx Integrated Sequencer | A53256 |
| Genexus™ Dx Install Kit | A52172 |

## Required materials and equipment not supplied

Unless otherwise indicated, all materials are available through **thermofisher.com**. "MLS" indicates that the material is available from **fisherscientific.com** or another major laboratory supplier. Catalog numbers that appear as links open the web pages for those products.

| Item | Source |
|---|---|
| Equipment | |
| Microcentrifuge[1] | • Cole-Parmer™ EW-17414-06<br>• Eppendorf™ 022620304 |
| Sorvall™ ST 8 Small Benchtop Centrifuge (or equivalent), with<br>Thermo Scientific™ M10 Microplate Swinging Bucket Rotor (or equivalent), and<br>Sealed Bucket; Capacity: 4 Standard or 2 Midi-Deepwell plates; set of 2) (or equivalent) | 75007200<br>75005706<br>75005721 |
| Platform laboratory mixer (Vortex or equivalent) | **MLS** |
| Economy Standard Incubator (2, 60°C and 90°C), or equivalent | S50441A<br>**fisherscientific.com** |
| Uninterruptible Power Supply (UPS)[2] | **MLS** |
| Equipment and consumables for AutoLys M FFPE sample extraction[3] | |
| PCR cabinet or laminar flow hood (recommended) | **MLS** |
| AutoLys M Tube Rack | A37955 |
| AutoLys M Tube Locking Lid | A37954 |
| AutoLys M TubeLifter or<br>AutoLys M Tube Pliers | A37956<br>A38261 |
| Integra™ Miltex™ Sterile Standard Scalpel; Mfr. No. 4410, rounded blade for tissue macrodissection, if needed | 12-460-451 or **MLS** |
| Integra™ Miltex™ Sterile Standard Scalpel; Mfr. No. 4411, flat blade for scraping FFPE slides | 12-460-452 or **MLS** |

*(continued)*

| Item | Source |
|---|---|
| Fisherbrand™ High Precision Straight Medium Point General Purpose Tweezers/Forceps, or equivalent, for handling FFPE curls | 12-000-121 |
| Plates, tubes, and other consumables | |
| MicroAmp™ EnduraPlate™ Optical 96-Well Clear Reaction Plates with Barcode | 4483354, 4483352 |
| Thermo Scientific™ Adhesive PCR Plate Foils | AB0626 |
| MicroAmp™ Adhesive Film Applicator (recommended) | 4333183 or **MLS** |
| Nuclease-free microcentrifuge tubes, 1.5-mL or 1.7-mL | **MLS** |
| Eppendorf™ DNA LoBind™ Tubes, 1.5 mL (for long term storage of archived samples) | 13-698-791 (Eppendorf™ 022431021) or **MLS** |
| 2-, 20-, 200-, and 1,000-µL pipettors and appropriate aerosol-resistant filtered pipette tips | **MLS** |
| DNA*Zap*™ PCR DNA Degradation Solutions | AM9890 |
| RNase*Zap*™ RNase Decontamination Solution | AM9780 |
| RNase *AWAY*™ Surface Decontaminant | 7002PK |
| TE Buffer, 1X Solution pH 8.0, Low EDTA | J75793.AP or **MLS** |
| Nuclease-Free Water (not DEPC-Treated) | AM9937, AM9930 or **MLS** |
| Gloves, powder-free nitrile | **MLS** |
| Ice buckets and ice | — |
| Isopropanol, 70% solution | **MLS** |
| Wipes, disposable lint-free | **MLS** |

[1] Must fit standard 1.5-mL microcentrifuge tubes and generate 15,000 × *g*. To convert the RPMs of your centrifuge to RCF in units of gravity, see **tools.thermofisher.com/content/sfs/brochures/TR0040-Centrifuge-speed.pdf**.

[2] We require that you use an uninterruptible power supply that is compatible with 2500 W output or greater.

[3] For use with the Genexus™ FFPE DNA and RNA Purification kit.

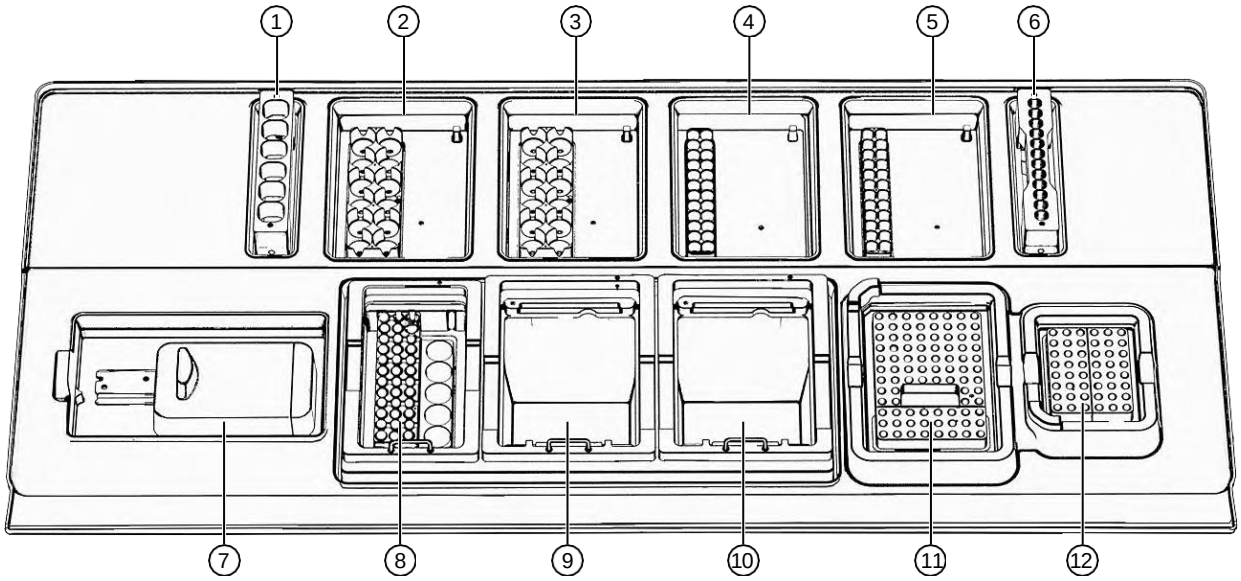
# Genexus™ Dx Purification System features




External features of the Genexus™ Dx Purification System.

① Power button
② USB port
③ Touchscreen

# Genexus™ Dx Purification System deck stations

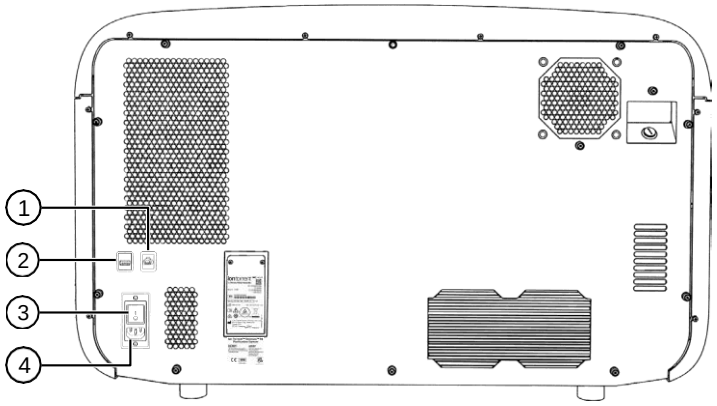



Interior deck stations of the Genexus™ Dx Purification System.

1. 6-well tip comb
2. 24 Deep-well plate 1
3. 24 Deep-well plate 2
4. 96 Deep-well plate 1
5. 96 Deep-well plate 2
6. 12-well tip comb
7. Quantitation module
8. Quantitation tray
9. Tip box 1
10. Tip box 2
11. 96-well output plate
12. 48-well archive plate

# Genexus™ Dx Purification System input and output connections

The connection panel, power port, and an on/off switch are located on the left side of the rear panel of the instrument.



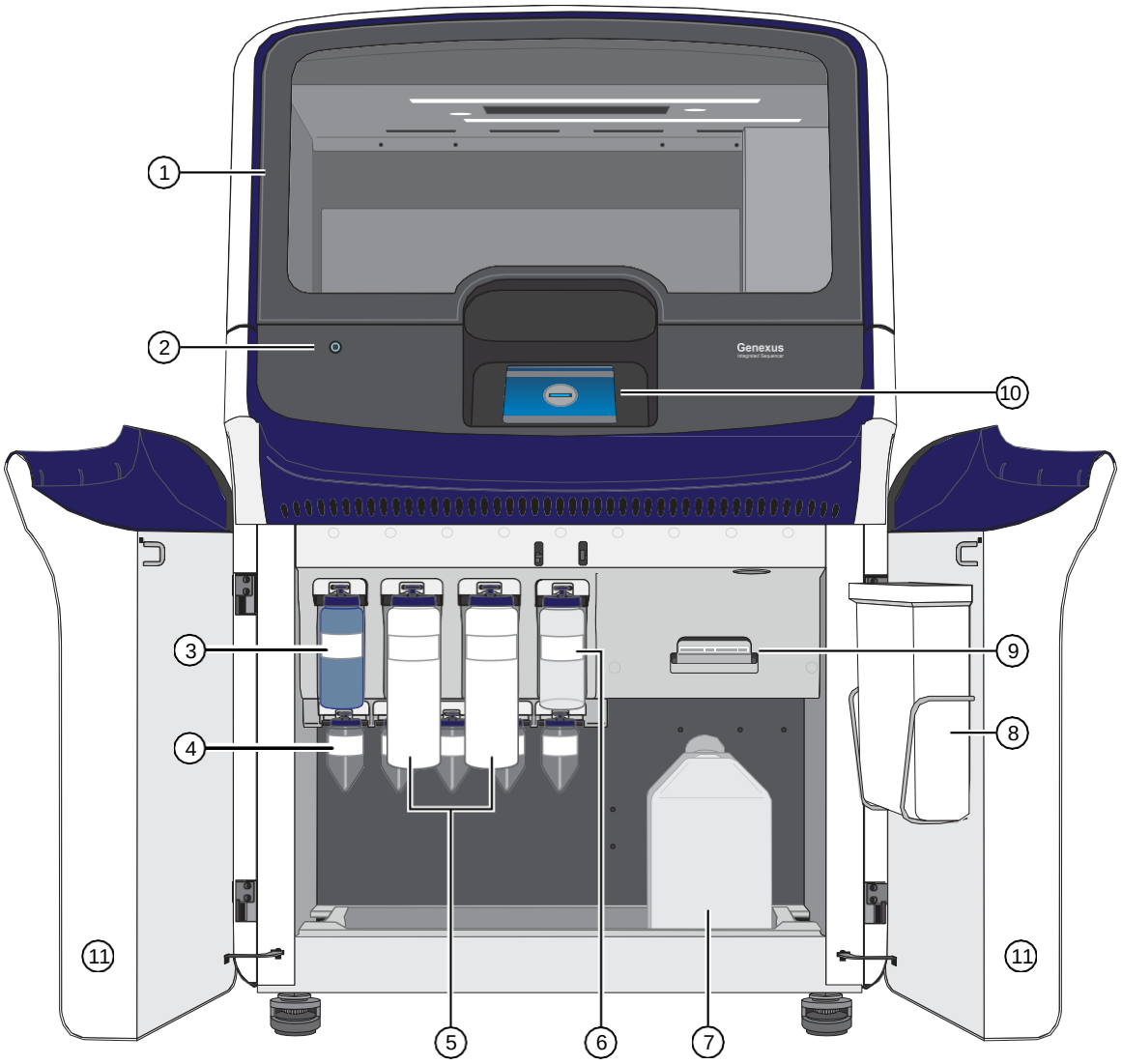

① Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the instrument and a local area network.

② USB port—Connects a USB device to the instrument.

③ On/off switch—Power switch, where the states are on ( | ) or off ( O ).

④ Power port—100–240VAC port that provides power to the instrument.

# Genexus™ Dx Integrated Sequencer components



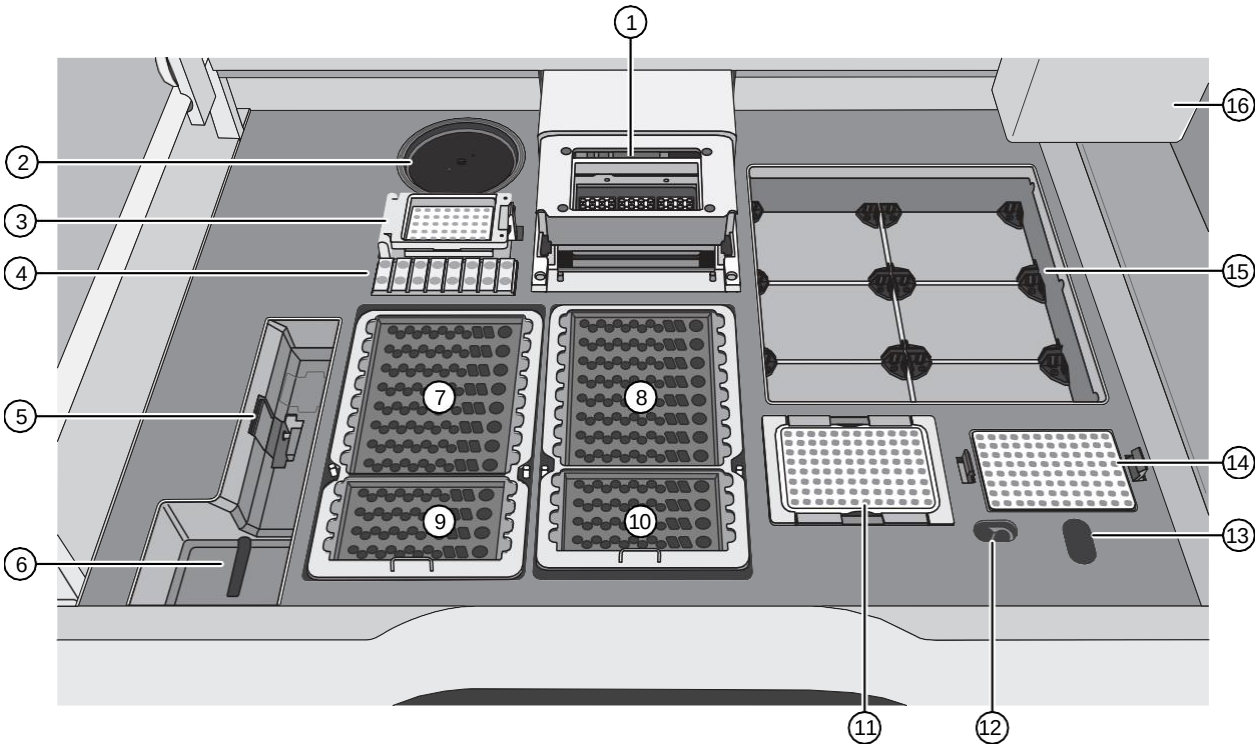

Major features and components of the exterior and sequencing reagents bay of the Genexus™ Dx Integrated Sequencer

1. Door to deck chamber. The door is locked in the closed position during an instrument run.
2. Power button
3. Genexus™ Dx Bottle 1 (Chemical Waste)
4. Genexus™ Dx Conical Bottles (Reusable conical bottles for Genexus™ Dx Cartridge reagent dilution)
5. Genexus™ Dx Bottle 2 (Sequencing Solution)
6. Genexus™ Dx Bottle 3 (Cleaning Solution)
7. Waste carboy
8. Waste pipette tip bin
9. Genexus™ Dx Cartridge
10. Touchscreen
11. Sequencing reagents bay door. Doors are locked in the closed position during an instrument run.



WARNING! This product contains very strong permanent magnets. People wearing a pacemaker or metallic prostheses should not use this product. A pacemaker or prostheses may be affected or damaged if it comes in close contact with a strong magnetic field.

# Genexus™ Dx Integrated Sequencer deck stations

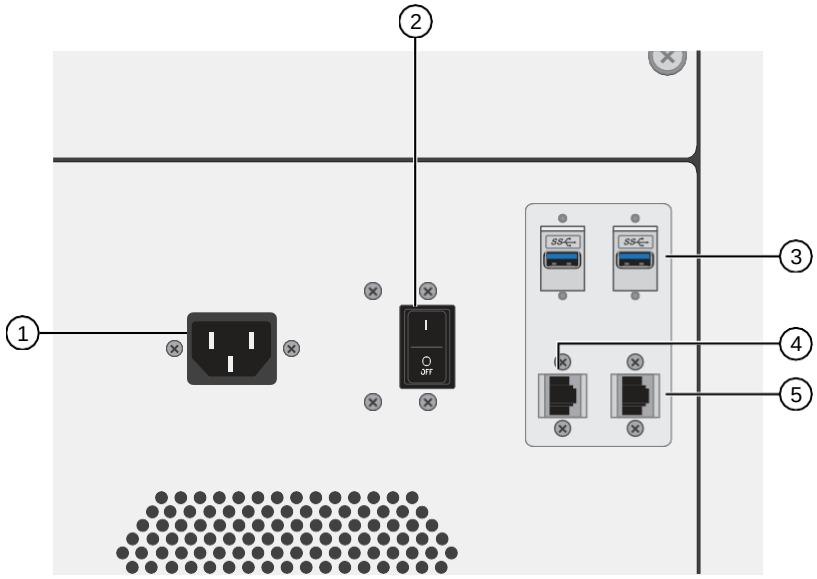



Interior Genexus™ Dx Integrated Sequencer deck components and stations

1. PCR amplification station
2. Microcentrifuge (not used)
3. Genexus™ Dx Barcodes 1–32 HD station
4. Oncomine™ Dx Express Test primer tube station
5. Genexus™ Coupler station
6. Chip install station
7. Zone 1 station (Genexus™ Dx Strip 1)
8. Zone 2 station (Genexus™ Dx Strip 2-HD)
9. Zone 3 station (Genexus™ Dx Strip 3-GX5™)
10. Zone 4 station (Genexus™ Dx Strip 4)
11. Enrichment plate station
12. Liquid waste disposal port
13. Waste pipette tip disposal port
14. Sample plate station
15. Genexus™ Dx Pipette Tips station
16. Robotic pipettor

# Genexus™ Dx Integrated Sequencer input and output connections

The connection panel, power port, and an on/off switch are located on the right side of the rear panel of the instrument.

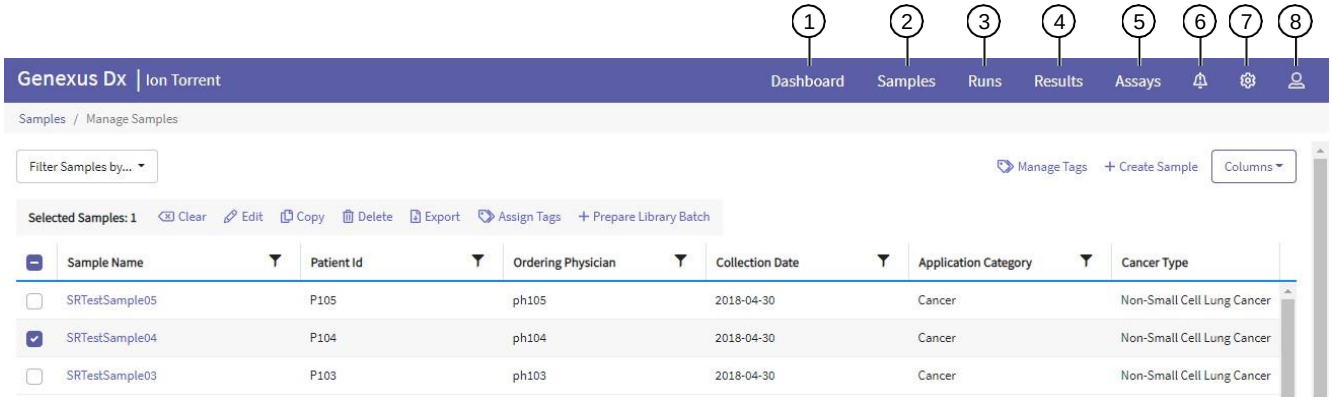



1. Power port—100–240VAC port that provides power to the instrument.
2. On/off switch—Power switch, where the states are on ( | ) or off ( O ).
3. USB ports—Connects a USB device to the instrument.
4. Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and a local area network.
5. Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and an accessory instrument such as the Genexus™ Dx Purification System.

# Genexus™ Dx Software overview

Genexus™ Dx Software provides tools to help you add and manage samples, libraries, runs, and assays. You can also view and manage sequencing results, monitor Genexus™ Dx Integrated Sequencer runs in progress, and manage software settings.

Basic software procedures such as sample creation, run planning, and review of sequencing results are described in this user guide. For more information about how to make full use of the software, including sample, run, and system management, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or the on-line software help system.




① **Dashboard**—View recent run history and current purification or sequencing run status.

② **Samples**—Add new samples, import samples, prepare library batches, import library batches, and manage attributes.

③ **Runs**—Plan a run starting from a sample, a nucleic acid sample, or a library. View, edit, and manage runs. Sample to result runs are for nucleic acid isolation on a Genexus™ Dx Purification System followed by sequencing on a Genexus™ Dx Integrated Sequencer.

④ **Results**—View sample results, run results, and verification results.

⑤ **Assays**—Manage and import assays. Manage assay preset parameters and panels.

⑥ **Notifications**—Receive alerts and messages regarding completion of runs and other processes.

⑦ **Settings**—Access audit records and run logs, configure network settings, manage backup settings, restore runs, manage gene lists, link to Connect user accounts, check for software updates, and manage data archiving, disk space, and users. Field Service Engineers access verification templates during sequencer installation.

⑧ **Profile**—Access Help, manage and edit user profile settings and SSH key (system administrator only), sign out.

# Sample to Result workflow

## Workflow for a Sample to Result purification and sequencing run

### Enter samples in Genexus™ Dx Software (page 36)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as collection date, gender, sample type, and application category.



### Plan a Sample to Result run in Genexus™ Dx Software (page 46)

Runs planned in Genexus™ Dx Software include all the settings that are used in Oncomine™ Dx Express Test sample and library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

### Prepare samples and load the Genexus™ Dx Purification System, then start a purification run for FFPE samples (page 71)

Prepare samples, then follow the step-by-step instructions on the purification instrument touchscreen for automated purification of sequencing-ready DNA and RNA samples from FFPE sections.



### Add controls to the sample plate (page 94)

For a Sample to Result run, add positive controls and no-template controls to the sample plate prepared by the Genexus™ Dx Purification System.



### Load the Genexus™ Dx Integrated Sequencer and start a sequencing run (page 100)

Follow the step-by-step instructions in the sequencer touchscreen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



### Monitor the run (page 116)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



### Review results in Genexus™ Dx Software (page 118)

Review data and results and generate a report for each sample.

# Nucleic Acid to Result workflow

## Workflow for a Nucleic Acid to Result sequencing run

### **Enter samples in Genexus™ Dx Software** (page 36)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as collection date, gender, sample type, and application category.



### **Plan a Nucleic Acid to Result run** (page 56)

Runs planned in Genexus™ Dx Software include all the settings that are used in Oncomine™ Dx Express Test library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

### **Dilute the samples and load the sample plate** (page 96)

Dilute your nucleic acid samples, if needed, then load the sample plate with samples and positive controls and no-template controls.



### **Load the sequencer and start a run** (page 100)

Follow the step-by-step instructions on the sequencer touchscreen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



### **Monitor the run** (page 116)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



### **Review results in Genexus™ Dx Software** (page 118)

Review data and results in Genexus™ Dx Software and generate a report for each sample.

# 2 Before you begin


- Tissue input requirements for sample extraction ..... 27
- Sample storage and stability ..... 28
- Procedural guidelines ..... 28
- Power the Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer on or off ..... 34
- Getting started in Genexus™ Dx Software ..... 34
- Network and password security requirements ..... 35


## Tissue input requirements for sample extraction

### FFPE sample purification

The starting material for the purification procedure is an FFPE tissue sample that is unstained and mounted on a slide, or an unstained curl. Confirm the tumor content of each sample based on the area of a hematoxylin and eosin (H&E) stained section.

The recommended number of slide-mounted 5-µm FFPE sections or curls used in extraction varies depending on the sample collection method:

| Sample type | Recommended input |
|---|---|
| FFPE tumor resections | 2 × 5-µm curls/slides or equivalent amount |
| FFPE core needle biopsy (CNB) | 9 × 5-µm curls/slides or equivalent amount |
| FFPE fine needle aspirate (FNA) | 7 × 5-µm curls/slides or equivalent amount |

For further information about how to prepare FFPE samples for purification, see “Prepare FFPE curls with AutoLys M Tubes” on page 74 or “Prepare FFPE slide sections with AutoLys M Tubes” on page 76.

# Sample storage and stability

For resection samples, FFPE blocks and slides can be stored at room temperature (15–30°C) for 6 months.

- For fine-needle aspirates (FNAs), FFPE blocks and slides can be stored at room temperature for up to 6 months.
- For core needle biopsies (CNBs), blocks and slides can be stored at room temperature for up to 6 months.
- FFPE curls can be stored at 2–8°C for up to 6 months.

FFPE blocks and slides can be shipped at room temperature, but FFPE curls must be shipped at 2–8°C in low-retention tubes (Eppendorf™ DNA LoBind™ Tubes; Cat. No. 13-698-791).

Extracted FFPE DNA can be stored in low-retention tubes at –30°C to –10°C for up to 12 months, including 3 freeze-thaw cycles. Extracted FFPE RNA can be stored in low-retention tubes at –90°C to –65°C for up to 12 months, including 3 freeze-thaw cycles. Store extracted FFPE DNA and RNA in sealed archive plates at –20°C to –10°C for up to 3 months.

# Procedural guidelines

## Definitions

Throughout this guide:

- Room temperature is defined as the temperature range 15−30°C (59–86°F).
- A pulse centrifugation consists of a 3−5 second centrifugation at maximum speed in a mini centrifuge.
- Thawing or storing on ice can be substituted with thawing or storing at 4°C (2–8°C refrigerator or prechilled benchtop cold block).

## Guidelines for FFPE samples

- The minimum tumor content thresholds for the Oncomine™ Dx Express Test are ≥20% for resection and fine needle aspirate (FNA) cell block samples, and ≥10% for core needle biopsy (CNB) samples, regardless of variant type.
- For core needle biopsies and fine needle aspirates, macrodissection is not recommended due to the limiting tissue section surface areas.
- For resection or surgical biopsies, macrodissect and enrich the sample for tumor content if the tumor content is <20% and the tumor content in the region of interest is ≥10%. Following tumor enrichment, proceed with the extraction protocol.
- For optimal CNV results, if the tumor content of an FFPE resection sample is <20%, macrodissect and enrich the samples for tumor content
- Two 5-µm sections from FFPE resection blocks with at least 20% tumor cell content, nine 5-µm sections from FFPE CNB samples, and seven 5-µm sections from FFPE FNA cell blocks are recommended.

- Necrotic samples: Up to 70% necrotic tissue does not appear to interfere with sequencing results for the detection of EGFR exon 20 insertion mutations.
- Nucleic acid integrity is important for sample performance. Factors such as age of the block, fixation process used, and sample source can impact the quality of the extracted nucleic acid.
- Nucleic acid yield can be impacted by overall tissue area. If an initial extraction leads to insufficient concentrations for DNA and RNA, repeat the extractions with more material whenever possible.
- We recommend that you extract sufficient nucleic acid from samples to enable repeat sequencing runs, if possible.

## Guidelines for panel and reagent use and handling

- Use only the reagents and supplies that have been validated for the Oncomine™ Dx Express Test. For a list of validated reagents and supplies, see “Required materials” on page 12 and “Required materials and equipment not supplied” on page 15.
- Keep panel tubes capped until immediately before loading in the Genexus™ Dx Integrated Sequencer.
- Thaw positive controls on ice or at 4°C for up to 2 hours. After the positive controls are completely thawed, vortex on a platform vortexer, then centrifuge to collect tube contents. Return to ice or 4°C before loading into the sample plate.

  IMPORTANT! Ensure that contents of the control tubes are completely thawed before adding to the sample plate.

- Equilibrate Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 3-GX5™, and the Genexus™ Dx Cartridge at room temperature for 30 minutes before loading in the sequencer.
- Thaw Genexus™ Dx Strip 4 on ice or at 4°C for at least 30 minutes, or until the tube contents are fully dissolved and ready for loading on the sequencer.

  IMPORTANT! Ensure that strip contents are completely thawed before loading in the sequencer.

- Vortex thawed library and templating strips on a platform vortexer to dissolve precipitate and dislodge air bubbles. After vortexing, centrifuge the strips to collect tube contents using the Genexus™ Strip Centrifuge Adapter to hold strips during centrifugation.

  IMPORTANT! Inspect the bottom of tube 3 of the Genexus™ Dx Strip 2-HD for white precipitate before loading it in the sequencer.

  Note: For information about using the Genexus™ Strip Centrifuge Adapter, see “Centrifuge the library and templating reagent strips” on page 102.

- Verify consistency of reagent volumes across strips and panel tubes by visually inspecting them two at a time after centrifugation. For instructions, see “Centrifuge the library and templating reagent strips” on page 102. Alternatively, use the strip volume guide images provided on page 222 to check appropriate fill volumes of strip tubes.
- If loading is delayed, keep thawed panel tubes, Genexus™ Dx Strip 2-HD, Genexus™ Dx Strip 4, and the sample plate on ice or at 4°C until you are ready to load the sequencer.

- Do not freeze-thaw the Oncomine™ Dx Express Test panel tubes. Thaw only the number of panel tubes that are needed for an instrument run. Discard unused panel tubes after they are thawed. Store panel tubes at −30°C to −10°C.

  Note: One Oncomine™ Dx Express Test panel tube pair (one tube with forward primers and one tube with reverse primers) is sufficient for preparing up to 4 libraries.

- Do not combine the contents of Oncomine™ Dx Express Test panel tubes. Forward and reverse primers must remain separate, until they are combined by the sequencer as part of the on-instrument library preparation workflow.

## Guidelines for preventing contamination

We recommend following these guidelines to prevent cross-contamination of samples and controls between and within sequencing runs.

Personal protective equipment

- Wear a laboratory coat that is reserved for sequencing work and is laundered frequently. If possible, change to a fresh laboratory coat before setting up a run, or use new sleeve covers.
- Wear fresh gloves to load the instruments, including during the loading of the sequencing chip and coupler. Do not remove gloves to install the sequencing chip.
- Change gloves between dispensing positive controls and no-template control (NTC) in sample plate wells.

Equipment and instrument cleaning

- If you are preparing samples and sample plates in a PCR cabinet or laminar flow hood (recommended), illuminate the hood with UV light for 15 minutes before use.
- Before a run, decontaminate equipment or bench surfaces where samples or sample plates are handled. Use DNA*Zap*™ and either 1) the RNase*Zap*™ RNase Decontamination Solution or 2) the RNase *AWAY*™ Surface Decontaminant reagent. Follow manufacturer instructions for the proper storage and use of each decontamination reagent. Alternatively, a 10% solution of commercial bleach can be used for decontaminating working areas and equipment. Follow with wiping of surfaces with wipes moistened with 70% isopropanol or 70% ethanol.
- Before and after a run, sequentially wipe the deck surface of the Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer with lint-free wipes moistened with the two DNA*Zap*™ decontamination solutions followed by wipes moistened with either 1) the RNase*Zap*™ RNase Decontamination Solution or 2) the RNase *AWAY*™ Surface Decontaminant reagent. Follow with wiping of deck surfaces with wipes moistened with 70% isopropanol or 70% ethanol, or with prepared alcohol wipes. The robotic pipettor arm of each instrument can also be cleaned in this manner.

  IMPORTANT! Do not spray decontamination solution or alcohol solution directly onto deck surfaces or into deck openings. Instead, use a lint-free wipe moistened with solution to clean surfaces. Do not use bleach to decontaminate instrument surfaces as it can cause corrosion of the metal surfaces.

- After a sequencing run has completed, ensure that you cover the PCR amplification plate with a foil seal before removing it from the instrument to prevent library contamination.

Workflow tips

- After dispensing a positive control or sample (if applicable) in a sample plate well, do not pass the end of the used tip over wells intended for other samples or NTC. This practice minimizes the chance of depositing microdroplets in adjacent wells.
- If you dilute samples on the sample plate and vortex the plate after sealing, apply the foil seal tightly between wells using an adhesive film applicator before vortexing to prevent contamination between wells. To prevent spillage, use a PCR plate holder while sealing.
- Before disposal, close or cap used sample and positive control tubes to avoid creation of aerosols.
- Avoid touching the foil seals of the reagent strips, barcode plate, and sample plate.
- When loading the sequencer deck for a run, install the sample plate last.
- After a run, immediately seal the PCR amplification plate with a foil seal before removing the plate from the PCR amplification station. Sealing the plate before removal helps prevent contamination of libraries if libraries are recovered for reuse.

## Guidelines for Genexus™ Dx Purification System operation

- Follow guidance that is provided by Genexus™ Dx Software when you create a run plan to determine which consumables must be loaded for each run.
- Follow guidance that is provided by the software when you create a run plan to determine how many samples can be run with a given purification kit in an instrument run.
- Ensure deep-well plates are placed in the correct stations and in the correct orientation on the instrument deck. Press plates firmly into the appropriate station to seat properly.
- Ensure tip racks are placed in the correct station, in the correct orientation, and locked in place to avoid damage to the automated liquid handler.
- If the instrument detects a problem (for example, stuck pipette tip or obstructed movement of the gantry arm), the run is aborted and you need to restart the run with new consumables.

## Guidelines for Genexus™ Dx Integrated Sequencer operation

- To determine which new consumables must be loaded, and which installed consumables can be reused from a previous run, follow guidance that is provided by Genexus™ Dx Software during run planning. Consumables are listed in the run plan summary, which can be printed or saved for reference.
- To determine how many samples can be processed in a run, follow guidance that is provided by the software during run planning. The number of samples that can be included in a sequencing run depends on the following factors.

| Limiting factor | Description |
|---|---|
| The number of available barcodes in the barcode plate | The maximum number of available barcodes per run is 32.<br>IMPORTANT! When libraries are prepared on the Genexus™ Dx Integrated Sequencer, each target amplification reaction for a library requires a unique barcode. |
| The number of unused lanes on an installed chip | A maximum of 4 lanes are available on a single Genexus™ Dx GX5™ Chip. |
| The minimum read count per sample for an assay | The minimum read count per sample is an assay parameter that helps ensure a sufficient number of read counts per sample is obtained to pass QC metrics and enable high-confidence variant calling. |

- One Genexus™ Dx Strip 1 and one Genexus™ Dx Strip 2-HD is needed for each primer tube position 1–8 that is filled in a run.
- One Genexus™ Dx Strip 3-GX5™ and one Genexus™ Dx Strip 4 is needed for each chip lane that is used in a run.
- If a chip installed in a sequencer has unused lanes, do not remove it unless you are sure that you want to replace it with a new chip. After a partially used chip has been removed from the sequencer, it cannot be reinserted and reused. The sequencer cannot track lane usage after chip removal.
- You can remove a chip in one of the following situations.
  - After all the lanes of a chip are used in a run and a new run is selected, the chip shuttles to the install position and you are asked to remove the used chip.
  - When you select a run plan that requires more lanes than are available on the installed chip, you are asked to remove the partially used chip, and the sequencer performs a post-chip clean. In addition, you must clear consumables from the lower sequencing reagents bay, even if only a single lane of the chip was used. The consumables cannot be reused after a clean has been performed.
- The Genexus™ Dx Integrated Sequencer can track used and unused barcodes in barcode plates, enabling you to exchange plates between runs if needed, and reload a partially used barcode plate for a run if a sufficient number of barcodes is available on the plate.

  IMPORTANT! Do not write or mark on the foil seal of the barcode plate. The instrument vision system will be unable to distinguish used wells from unused wells, which can result in a run failure.

- Before loading pipette tip boxes in the sequencer before a run, ensure that you remove the lids from the tip boxes.

- After initial loading in the sequencer, reusable components (barcode plates, chips, and sequencing reagents bay components) must be used within 14 days for optimal results.
- An assay that is selected in a **Library to Result** run cannot include library batches that share one or more samples and barcodes.

# Precautions

## Avoid nucleic acid contamination

IMPORTANT! A primary source of contamination is spurious nucleic acid fragments from previous sample processing steps. Do not introduce amplified DNA into the work area where the instrument is located.

## Avoid strong electromagnetic radiation



WARNING! Do not use the instrument in close proximity to sources of strong electromagnetic radiation (for example, unshielded intentional RF sources), as these sources can interfere with proper operation.

## Protection by equipment



WARNING! The protection that is provided by the equipment can be impaired if the instrument is operated outside the environment and use specifications, the user provides inadequate maintenance, or the equipment is used in a manner that is not specified by the manufacturer (Thermo Fisher Scientific).



WARNING! This product contains very strong permanent magnets. People wearing a pacemaker or metallic prostheses should not use this product. A pacemaker or prostheses may be affected or damaged if it comes in close contact with a strong magnetic field.

# Power the Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer on or off

## Power on

If the touchscreen is unresponsive, check the power switch on the back of the instrument and ensure that the switch is in the on ( | ) position. If the power switch is in the off ( O ) position, proceed with step 1. If the power switch is already in the on position, proceed to step 2.

1. On the back of the instrument, turn the power switch to the on ( | ) position.
2. On the front of the instrument, press the power button.
   The button illuminates.
3. In the **Sign In** screen, enter the username and password created by the field service engineer when the instrument was set up, or the unique username and password set up for you as an instrument user.
   When the instrument **Home** screen appears, the instrument is ready for use.

## Power off

It is not necessary to power off the instrument overnight or over the weekend, and the instrument does not need to be powered off if it will be idle for an extended period of time. However, if needed, follow the steps below to power off the instrument.

1. In the home screen, tap **Settings►System Tools►Shut down**.
2. Select either **Shutdown** or **Reboot**.
3. If you select **Shutdown**, a confirmation message appears. Select **Yes** to power off the instrument.

Note: If you power off the sequencer with a partially used chip installed, the chip and consumables status is saved. When you power back on, the saved chip and consumable information enables you to use the chip for up to 14 days after the chip was installed.

IMPORTANT! Do *not* press the power button during a run. Interrupting power to the instrument during a run can result in run failure and loss of sample.

# Getting started in Genexus™ Dx Software

Before you can sign in to and use Genexus™ Dx Software, you must request a new user account from a manager- or administrator-level user. For information on user access levels, creating user accounts, and signing in to the software, and for background information on the software, see "Getting started in Genexus™ Dx Software" in the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or in the software help system.

# Network and password security requirements

## Network configuration, security, and data backup

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of your facility administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions.

If external or network drives are connected to the sequencer, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

If a Laboratory Information Management System (LIMS) connection is set up, you can use the LIMS API to obtain the list of files generated for the assay. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910).

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Note: If a LIMS system is configured to retrieve files from the sequencer, you can manually set up an FTP directory or drive mapping. This configuration is not provided as part of sequencer installation, and must be set up by your LIMS system integration or IT group.

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You must have an established setup for regularly backing up and archiving your data. Data backup is solely your responsibility. In the event your system needs repair, Thermo Fisher Scientific is not responsible for data backup or any loss of data.

## Password security

Best practice is to maintain unique passwords for all accounts in use in Genexus™ Dx Software. All passwords must be reset the first time a user signs into the software. Change passwords according to your organization's password policy.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords. This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# 3 Enter samples and prepare library batches in Genexus™ Dx Software


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- Prepare or import a library batch ..... 41
- No-template and positive controls ..... 45


## Enter or import samples

Before planning a Sample to Result or Nucleic Acid to Result run in Genexus™ Dx Software, you must first enter sample information in the software to assign sample names and provide other information. From the **Samples** menu, you can create samples in three ways.

- Enter information one sample at a time.
- Enter information for multiple samples in a table format.
- Import information for multiple samples by uploading a file.

A library batch is a group of samples that are sequenced in a Library to Result run. You can create library batches from samples you have previously added or uploaded to the software. During library batch preparation, you identify the barcode adapters that were used to prepare the libraries. You add library batches to run plans when Genexus™ Dx Software guides you through the steps to plan a Library to Result run.

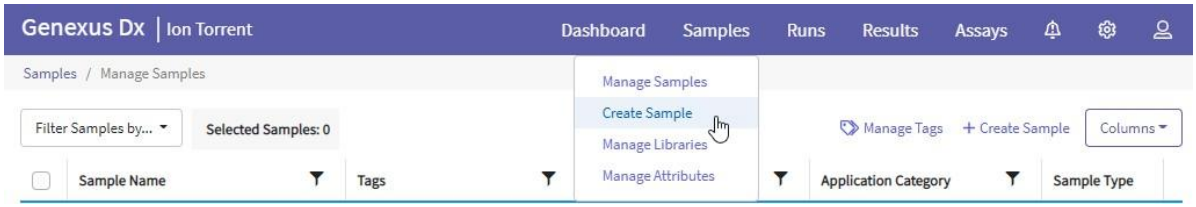
Before entering samples or preparing a library batch, you must sign in to Genexus™ Dx Software in IVD mode.

## Create a new sample

You can enter information individually for each sample that you add in the software. Complete this procedure to make a sample available for use in a run, or to add the sample to a library batch. If you have many samples to add, see “Create multiple samples” on page 37 or “Import samples” on page 39.

The information that you enter for each sample is entered as sample attributes. For more information on sample attributes, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or the software help system.

1. In the menu bar, click **Samples►Create Sample**.




Note: You can also click + **Create Sample** in the **Samples / Manage Samples** screen.

2. In the **Samples/Create Sample** screen, enter a name for the sample, select an **Application Category**, then complete the required fields, and optional fields if needed.

   Attributes identified with a red asterisk (*) are required. **Application Category** is set to **Cancer** by default. The fields for sample attributes that are displayed depend on the **Application Category** that is selected.

   - In the optional **Tags** field, enter a full or partial tag name of at least 3 characters if desired, then select one or more tags of interest to assign a tag to the sample.
   - If the tag that you want to assign to the sample does not exist in the software, a manager- or administrator-level user can enter a new tag name in the **Tags** field to create a new tag and assign it to the sample.

3. Click **Save**.

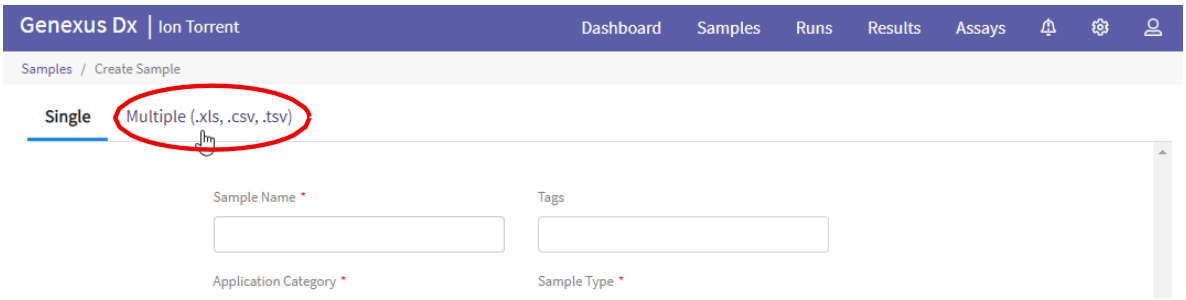
   The new sample is listed in the **Manage Samples** screen and is available to use in a run plan.

## Create multiple samples

There are two ways you can create multiple new samples in Genexus™ Dx Software.

- You can enter sample information for multiple samples in a table format in the software.
- You can import a file that contains sample data. For more information, see instead “Import samples” on page 39.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.




3. In **Application Category**, select the application category for the samples.
   A table with columns that are specific for the selected application category appears.
4. *(Optional)* Click **Columns** in the upper right corner of the screen to customize the optional attributes for the samples that you want to create.
   - Select the checkbox for a sample attribute to add a column to the table. Click the row to view fields into which you can enter attribute information.
   - Deselect the checkbox for a sample attribute to remove the attribute from the table.
5. Enter the information for a sample.
   Attributes identified with a red asterisk (*) are required.
6. Click **Add Row**, then enter the information in the new row for each new sample.
7. Repeat step 6 for each new sample.
8. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you will create.
9. Click **Save**.
   The new samples are listed in the **Manage Samples** screen and are available to use in a run plan.

   Note: Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

## Import samples

You can enter sample information for multiple samples directly in Genexus™ Dx Software. When you want to create more than a few samples, an easy and fast way to add multiple samples in the software is to create a file of information for a group of samples and import that file.

Sample data files can be used to capture, manage, and edit sample data. You can import sample data files in TXT, XLS, XLSX, or CSV file formats. For a list of the sample attributes that are included in the import file, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or the software help system. For ease of use, you can download a Microsoft™ Excel™ template file to create an import file.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In the **Application Category** dropdown list, select the application category for the samples.
   Template files are specific for each application category.
4. Select one of two options.

| Option | Description |
|---|---|
| Download a template file, then edit it to create a new file. | a. Click **Download Template**, then edit the file to add sample information. For more information, see "Download a template file for sample creation" on page 40.<br>b. After editing the file, proceed to the second option. |
| Upload the sample data from an existing file. | a. Click **Browse**.<br>b. Navigate to the file, then click **Open**.<br>The data contained in the file populates the table in the screen. |

5. Edit the sample table and data, if needed.

| Option | Description |
|---|---|
| Remove an attribute column | Click **Columns**, then deselect the column name. |
| Add an attribute column to the table | Click **Columns**, then select the column name. The column names that are listed are **Application Category**-specific. |
| Edit sample data | Click the sample row of interest, then edit the text field or dropdown list for the sample. |
| Add more samples to the table | Click **Add Row**, then enter the sample data. |
| Remove a sample from the table | Select the checkbox for the row, then click **Remove Row**. |

6. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample you intend to create.

7. Click **Save**.

   You can place the pointer over the alert (!) to view more information if needed.

The new samples are listed in the **Manage Samples** screen and are available to assign to a run plan.

## Download a template file for sample creation

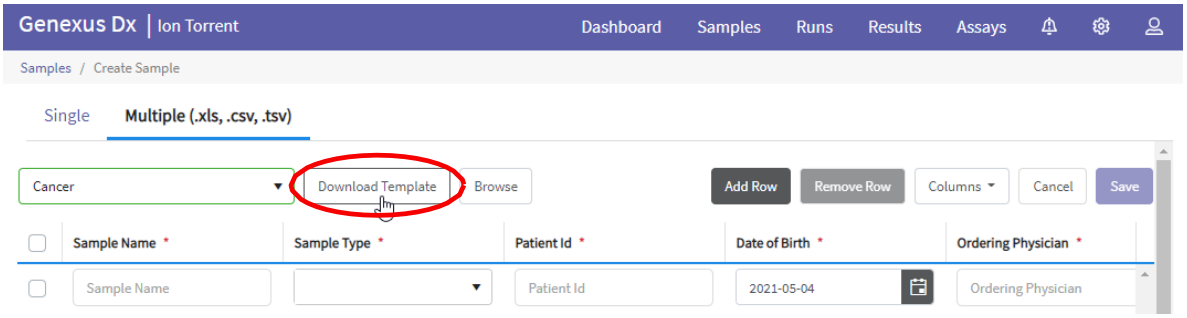
You can download a template file, add sample information to it, then use the file to import the sample data for multiple samples.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headings in the **Specimen Format** tab. Use the **Specimen Format** tab to enter samples.

Template files contain both the system-installed and the custom sample attributes as column headings.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.

   Template files are specific for each application category.
4. Click **Download Template** to download the Microsoft™ Excel™ template file.




   The template file contains default sample attributes as columns. If custom sample attributes have been configured in the software, the custom attributes are added to the template file.

5. Save the file to the computer, then open the file, enter sample data in the **Sample Details** tab.
6. When you are finished adding sample information to the file, save the file.

You can now import the file to Genexus™ Dx Software.

# Prepare or import a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have recovered from a previous sequencing run, you need to first create a library batch in Genexus™ Dx Software from samples. You can enter samples in the software or import a sample file. For more information, see “Enter or import samples” on page 36.

Select the library batch when you plan the run. If you are planning a run starting from FFPE tissue samples, omit this step and proceed to “Plan a Sample to Result run” on page 48.

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Note:

- Each library in a library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

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## Prepare a library batch

A library batch is a group of prepared libraries that are sequenced in the same library run. If you are planning a run starting from libraries that you have recovered from a previous sequencing run, you need to first create a library batch in Genexus™ Dx Software from samples that you have added. If you are planning a run starting from FFPE tissue samples, omit this step and proceed to “Plan a Sample to Result run” on page 48.

Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software to prepare the library batch and verify that the barcode scanner is compatible with the computer.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

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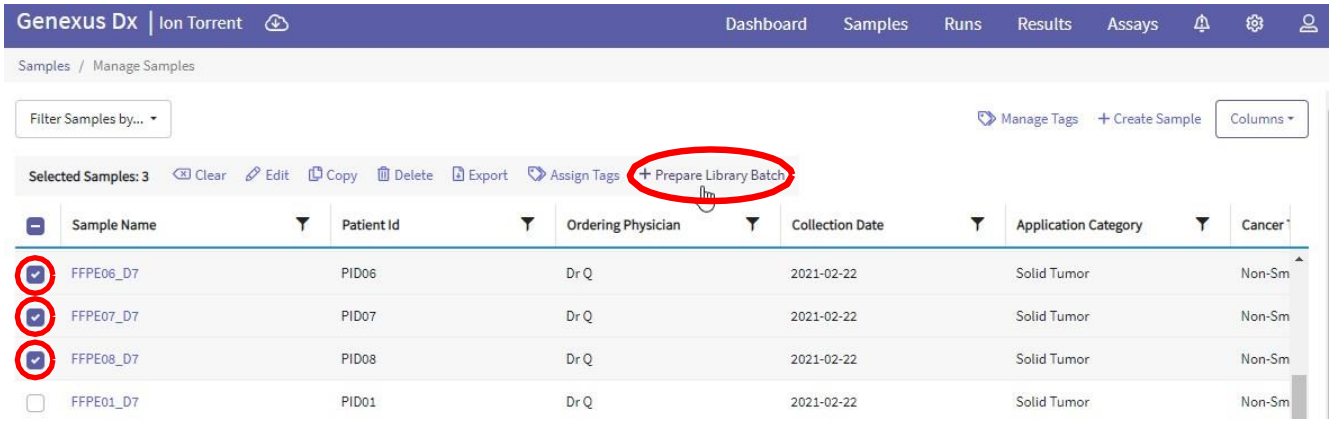
   Note: If the library batch is being created in the same sequencer where the samples were processed initially, in the **Filter Samples by** dropdown list, apply the **All** filter to be able to select the previously processed samples.

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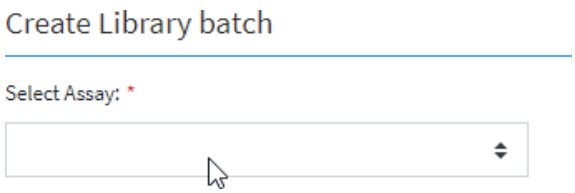
   For more information about how to filter the list of samples, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910).

3. Select samples in the list by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.

Note: Two control samples are included in each run. The controls are added automatically to a run plan after you assign one or more samples, or to a library batch when you create the library batch.



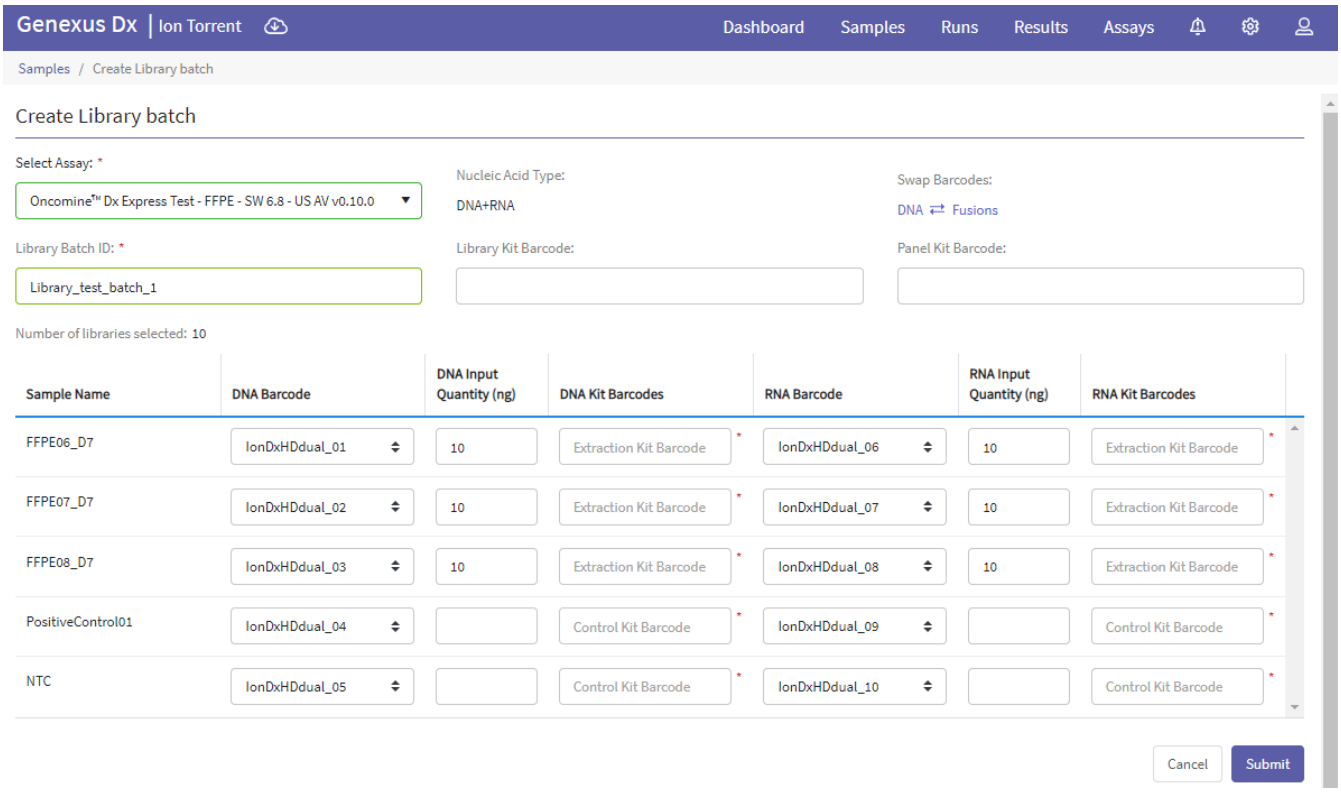

4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** dropdown list.




The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library prep type, the library preparation type automatically fills with the nucleic acid type specified by the assay you selected, for example DNA+RNA.

5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.

   Note: Library Batch IDs can include only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).




6. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.

7. Select the barcode ID used to prepare each library.

   Each library in a library batch must have a different barcode ID. The barcodes that are listed in the **DNA Barcode** or **RNA Barcode** dropdown list belong to the barcode set that was selected when the assay was created.

   IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.

8. Enter the DNA and/or RNA **Input Quantity** for each library.

9. Use the connected barcode scanner to scan the barcode from the control kit into the fields in the **DNA Kit Barcodes** and/or **RNA Kit Barcodes** columns for the positive controls and the no-template controls, and for each sample.

   If needed, you can enter the barcodes manually.

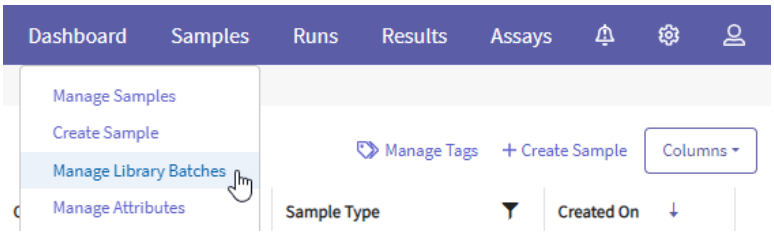
10. Click **Submit** to save and submit your selections and entries.

    The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Import a library batch

You can import library batch information in the form of an .XLS or .XLSX file. The import file must include all required library and kit information.

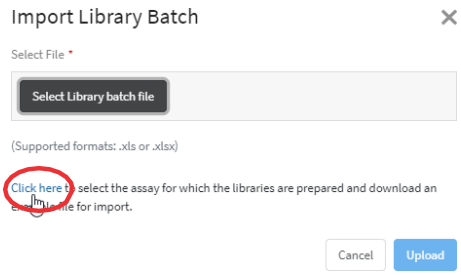
1. In the menu bar, click **Samples►Manage Library Batches**.




2. In the **Manage Library Batches** screen, click **Import Library Batch**.

3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.

   If the assay includes more than one library preparation type, a library preparation type dropdown list appears. Select the library preparation type, such as **DNA+RNA** for the library batch.




4. Select an assay from the list, then click **Download**.
   The assay name is auto-populated in the Microsoft™ Excel™ template file that downloads to your drive.

5. In the template file, enter or confirm the library batch information.

| Template item | Description |
|---|---|
| Reagents tab | |
| Assay Name | Auto-populated when assay is selected in step 4 (required) |
| Extraction Method Type | Auto-populated when assay and library prep type, if applicable, is selected in step 4 (required) |
| Library Batch ID | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required) |
| Library Kit Barcode | For example, Genexus™ Dx Library Strips 1 and 2-HD barcode (optional) |
| Panel Kit Barcode | For example, Oncomine™ Dx Express Test barcode (optional) |
| Libraries tab | |
| Sample Name | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required). For the positive control, the sample name should be *PositiveControl01*, and for the no-template control, the sample name should be *NTC*. |
| Barcode | Barcodes used for each sample and control library preparation (required) |

*(continued)*

| Template item | Description |
|---|---|
| Nucleic Acid Type | DNA or RNA (required) |
| Input Quantity | Library input quantity (optional) |
| Control Kit Barcode | The barcode for the Positive Control and NTC kits used for Sample to Result and Nucleic Acid to Result runs (required) |
| Extraction Kit Barcode | The barcode for the kit used for nucleic acid extraction of a sample (required) |

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA library must be listed in sequential order per pool for each sample.

6. Save the file.
7. Click **Select Library batch file**, navigate to the saved file, then select it.
8. Click **Upload**.
   A progress bar followed by an import report displays. If the import process fails, an error message indicates the reason for failure (for example, an invalid character was used). For additional troubleshooting support, see “Library batch import fails” on page 177.

# No-template and positive controls

A no-template control (NTC) sample and positive controls are included in runs that are planned in Genexus™ Dx Software. The controls are added automatically after you assign samples to a run plan, or when you create a library batch. The configuration of each control is based on the assay that is used.

# 4 Plan a run in Genexus™ Dx Software


- Before you plan a run ..... 47
- Mode switch with a partially used chip installed ..... 47
- Integrated nucleic acid isolation in Sample to Result runs ..... 47
- Plan a Sample to Result run ..... 48
- Plan a Nucleic Acid to Result run ..... 56
- Plan a Library to Result run ..... 63
- Plan a Genexus™ Dx FFPE DNA and RNA Purification run—standalone configuration ..... 67


Before you can sequence samples on the Genexus™ Dx Integrated Sequencer, you need to create a run plan to specify the Oncomine™ Dx Express Test assay, the samples, and any additional parameters needed for the run. In Genexus™ Dx Software 6.8 and later, the following types of runs can be planned for use on the Genexus™ Dx Integrated Sequencer:

- Sample to Result—start from unprocessed samples and isolate nucleic acid on the Genexus™ Dx Purification System in an integrated run configuration, then load the nucleic acid samples in the Genexus™ Dx Integrated Sequencer.
- Nucleic Acid to Result—start from purified nucleic acid samples.
- Library to Result—start from prepared and pooled libraries made previously on the Genexus™ Dx Integrated Sequencer.

In this chapter, Sample to Result, Nucleic Acid to Result, and Library to Result run planning are described. Sample to Result run planning with system-installed assays is enabled only if the Genexus™ Dx Purification System is part of your instrument configuration. Library to Result run planning is described as a retest option for libraries that have been previously prepared and pooled on the Genexus™ Dx Integrated Sequencer and have been recovered. For more information about recovering pooled sample libraries and this type of run, see “Recover pooled sample libraries and plan a Library to Result run” on page 217.

In addition, run planning on the Genexus™ Dx Purification System is described for purification runs using the standalone configuration. In the standalone run configuration, purified nucleic acid is loaded in an archive plate that can be stored and used later in a Nucleic Acid to Result sequencing run.

# Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Dx Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter or import samples” on page 36.
- A Genexus™ Dx Purification System is set up for use in an integrated configuration before you plan a **Sample to Result** run.
- Ensure that the following conditions are met for a **Library to Result** run.
  - Library batches are prepared and each batch uses a unique library batch ID.
  - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

  For more information, see “Prepare or import a library batch” on page 41.

The software returns an error message when any of the conditions are not met.

# Mode switch with a partially used chip installed

In Genexus™ Dx Software you can switch from IVD mode to Assay Development mode, or from Assay Development mode to IVD mode, when you sign in to the software. If you want to switch modes to plan and perform a run in a new mode, and you have a partially used chip installed in the sequencer, you cannot use the partially used chip and sequencing consumables in the new mode. A dialog box appears asking you to confirm the mode switch. You can cancel the mode switch, but if you proceed, you must perform an instrument clean and install a new chip and consumables. You will lose the unused chip lanes and remaining sequencing consumables.

If you want to switch modes to review run data or reanalyze data in the other mode, and do not perform a sequencing run, an instrument clean is not needed. You can proceed with the mode switch and you do not need to replace the installed chip and consumables.

# Integrated nucleic acid isolation in Sample to Result runs

A **Sample to Result** run is an integrated run for sequential and automated nucleic acid purification and sequencing. Administrators, managers, and operators can create **Sample to Result** runs for the formalin-fixed, paraffin-embedded (FFPE) tissue sample type.

Core needle biopsy (CNB) and fine needle aspiration (FNA) samples are considered FFPE sample types in the software and can be grouped with FFPE tissue sections in a nucleic acid isolation batch. The Genexus™ Dx Purification System can run one nucleic acid isolation batch at a time.

# Plan a Sample to Result run

This procedure applies only if a Genexus™ Dx Purification System is used with the Genexus™ Dx Integrated Sequencer.

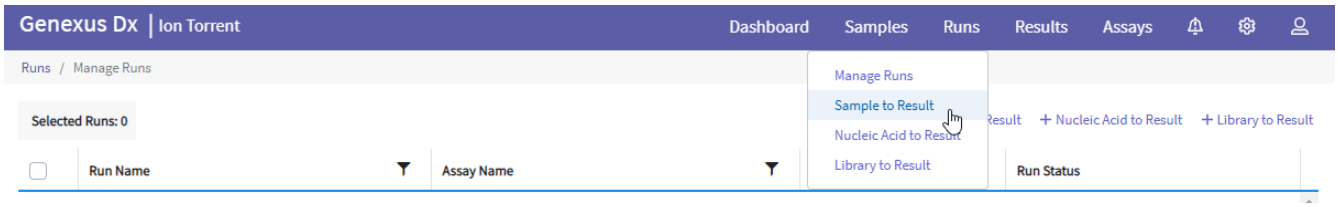
Ensure that the following prerequisites are complete before you plan a **Sample to Result** run.

- Integrate a Genexus™ Dx Purification System with the system.
- Enter sample information into Genexus™ Dx Software. For more information, see "Create a new sample" on page 37, or "Import samples" on page 39.
- Connect the barcode scanner to the computer on which you use Genexus™ Dx Software to plan runs and verify that the barcode scanner is compatible with the computer.

Planning a **Sample to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Purification**, and **Review**. Progress through the steps is tracked in the upper left corner of the **Runs / Sample to Result** screen.

1. In the menu bar, click **Runs►Sample to Result**.

   Note: You can also click + **Sample to Result** in the **Runs / Manage Runs** screen.

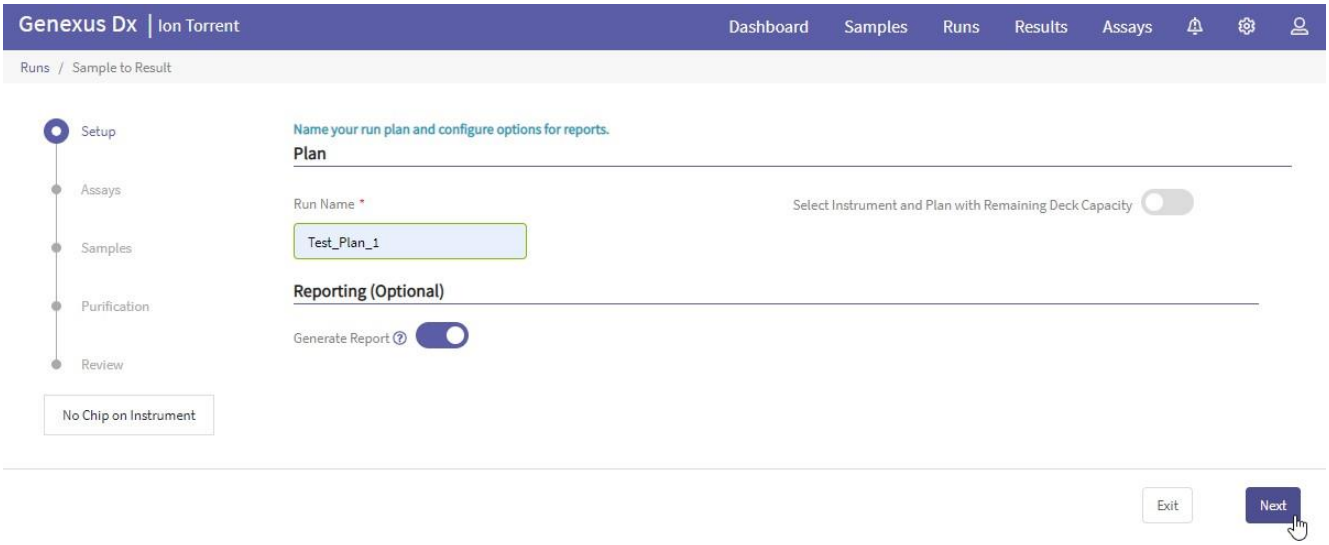



2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.

   b. *(Optional)* In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report that uses the default report template.

c. Click **Next**.



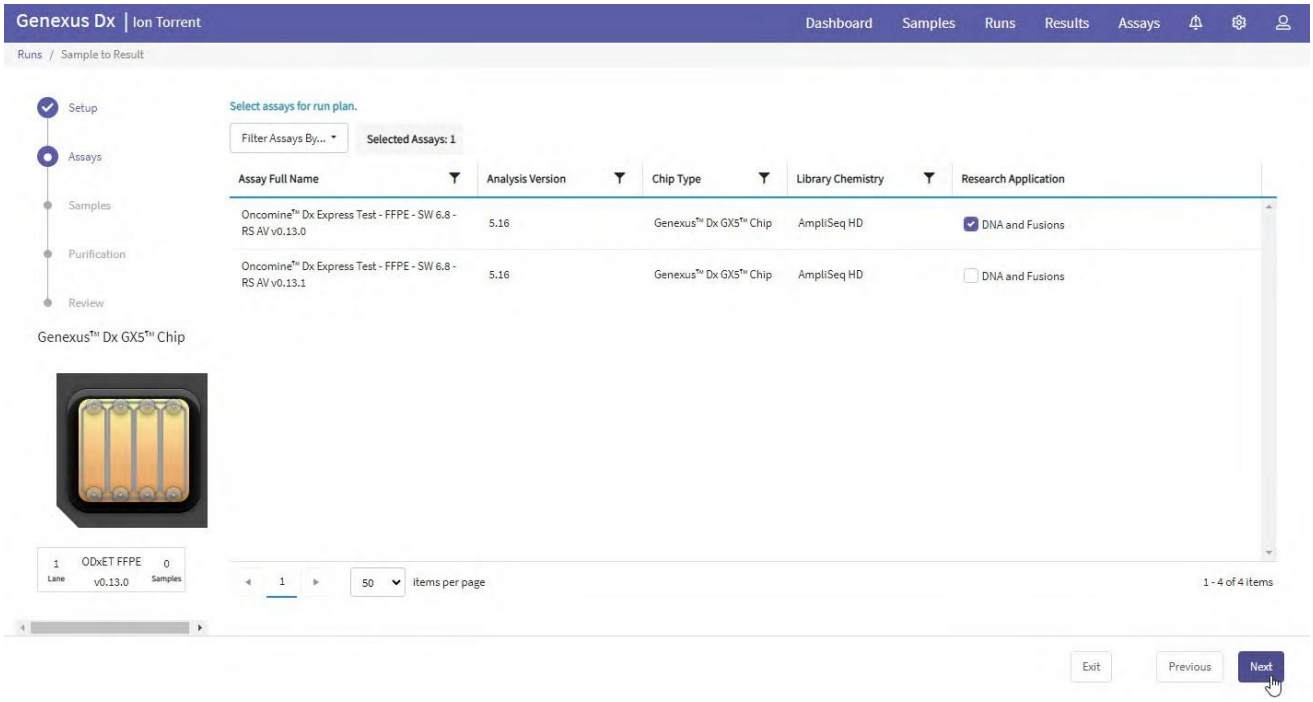

3. In the **Assays** step, select the assay that you want to include in the run.

   Only assays that are configured for **Sample to Result** runs are listed. If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

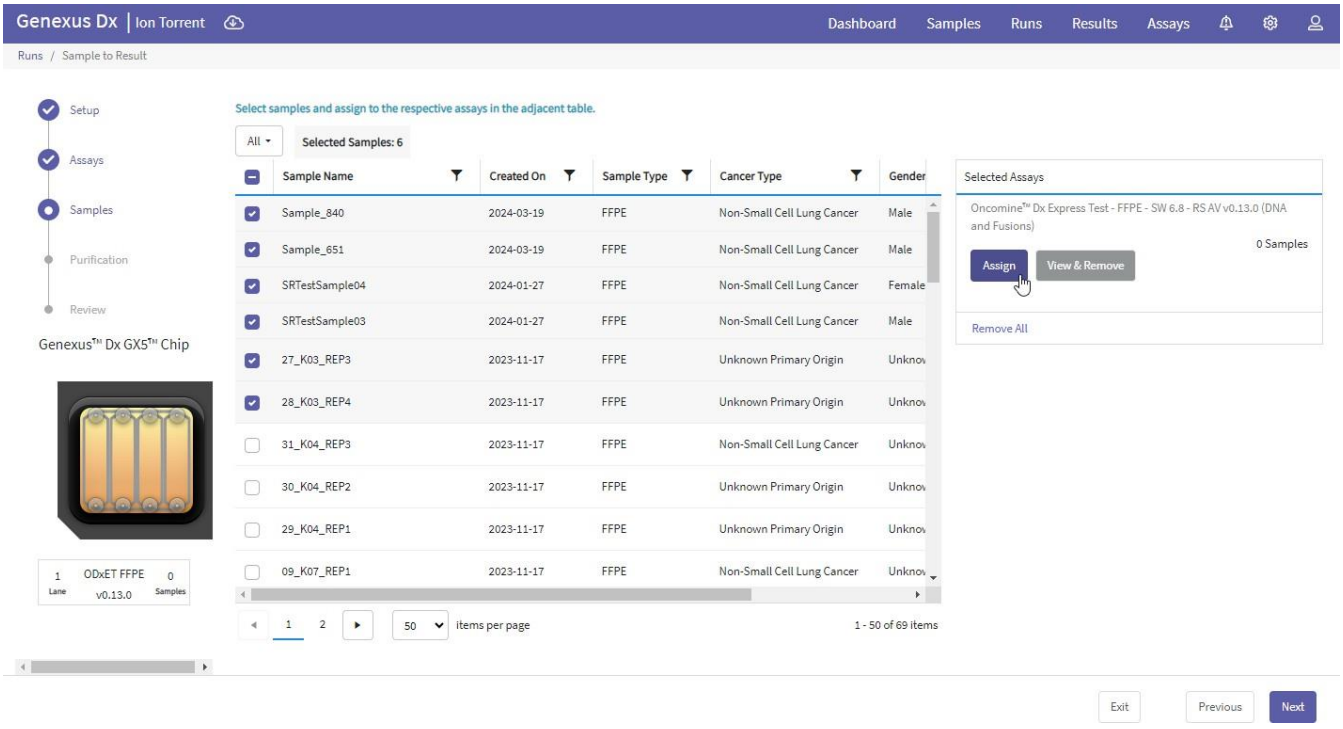
   a. Use the **(Filter)** tools in table column headers to find assays of interest, if desired.

   b. In the **Research Application** column for the assay of interest, select one or more application types, such as **DNA and Fusions**, to include each selected application type for the assay in the run plan.

c. Click **Next**.


Genexus Dx | Ion Torrent
Dashboard
Samples
Runs
Results
Assays
Runs / Sample to Result
Setup
Assays
Samples
Purification
Review
Genexus™ Dx GX5™ Chip
1 Lane
ODxET FFPE v0.13.0
0 Samples
Select assays for run plan.
Filter Assays By...
Selected Assays: 1
Assay Full Name
Analysis Version
Chip Type
Library Chemistry
Research Application
Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0
5.16
Genexus™ Dx GX5™ Chip
AmpliSeq HD
DNA and Fusions
Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.1
5.16
Genexus™ Dx GX5™ Chip
AmpliSeq HD
DNA and Fusions
1
50
items per page
1 - 4 of 4 items
Exit
Previous
Next

4. In the **Samples** step, select the samples that you want to run with each application type of each assay.
   a. Select the checkbox next to each sample that you want to assign to the application type of an assay, then in the **Selected Assays** pane, for the assay and application type that you want to use for the selected samples, click **Assign**.




Note: Two positive control samples and two no-template controls are included for each assay. The controls are added automatically to a run plan after you assign one or more samples.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples and controls, and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

If the

*minimum reads per sample* × *number of samples*

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.



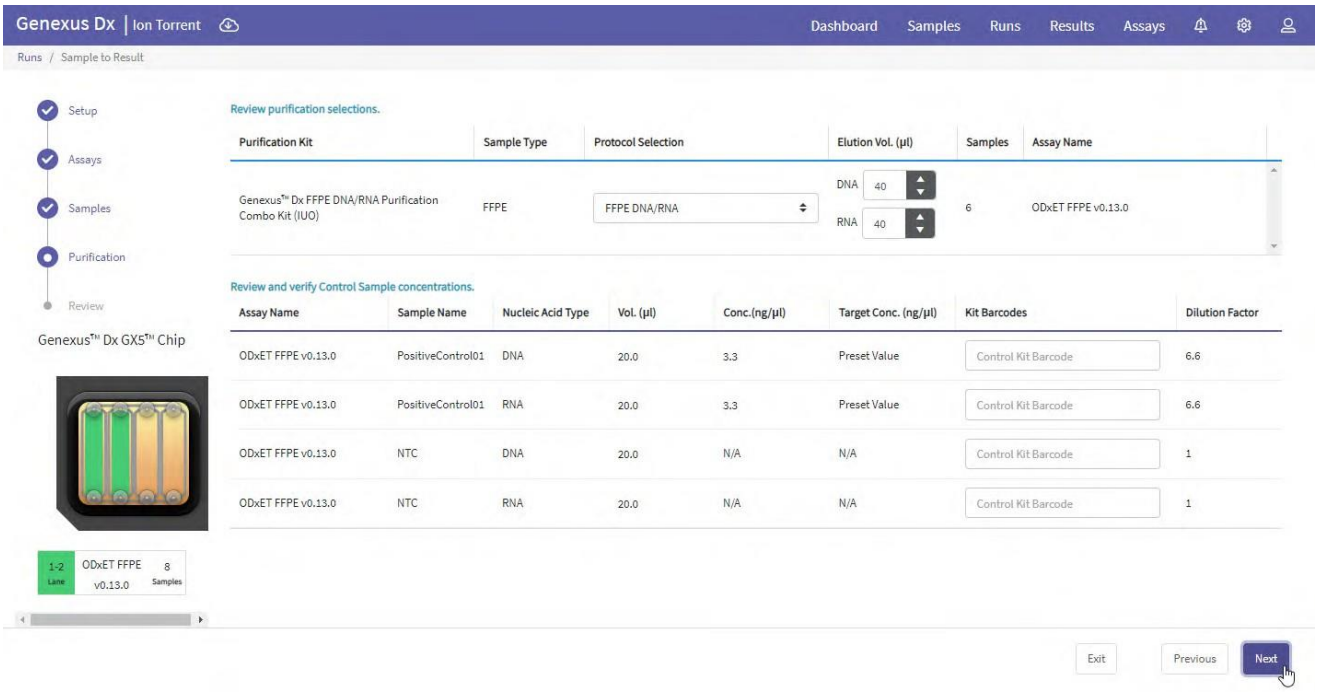

Green lane color denotes lane usage and sample assignment within lane capacity. Red lane color denotes sample assignment that exceeds lane capacity.

b. If needed, edit samples in one of the following ways.
   - Click **View & Remove**, make the selections, then click **Update**.
   - Click **Remove All**, to remove all sample assignments for all assays.

c. Click **Next**.

5. In the **Purification** step, review and edit the purification selections.
   a. Review the purification selections.

| Option | Description |
|---|---|
| Protocol Selection | FFPE DNA/RNA—the protocol auto-populates based on the assay selected and other options are not available. |
| Elution Vol. (µL) | The default 40 µL elution volume is recommended. Modify the elution volume within the allowable range, if needed. |




b. Use the connected barcode scanner to enter the control kit barcodes in the **Kit Barcodes** fields.

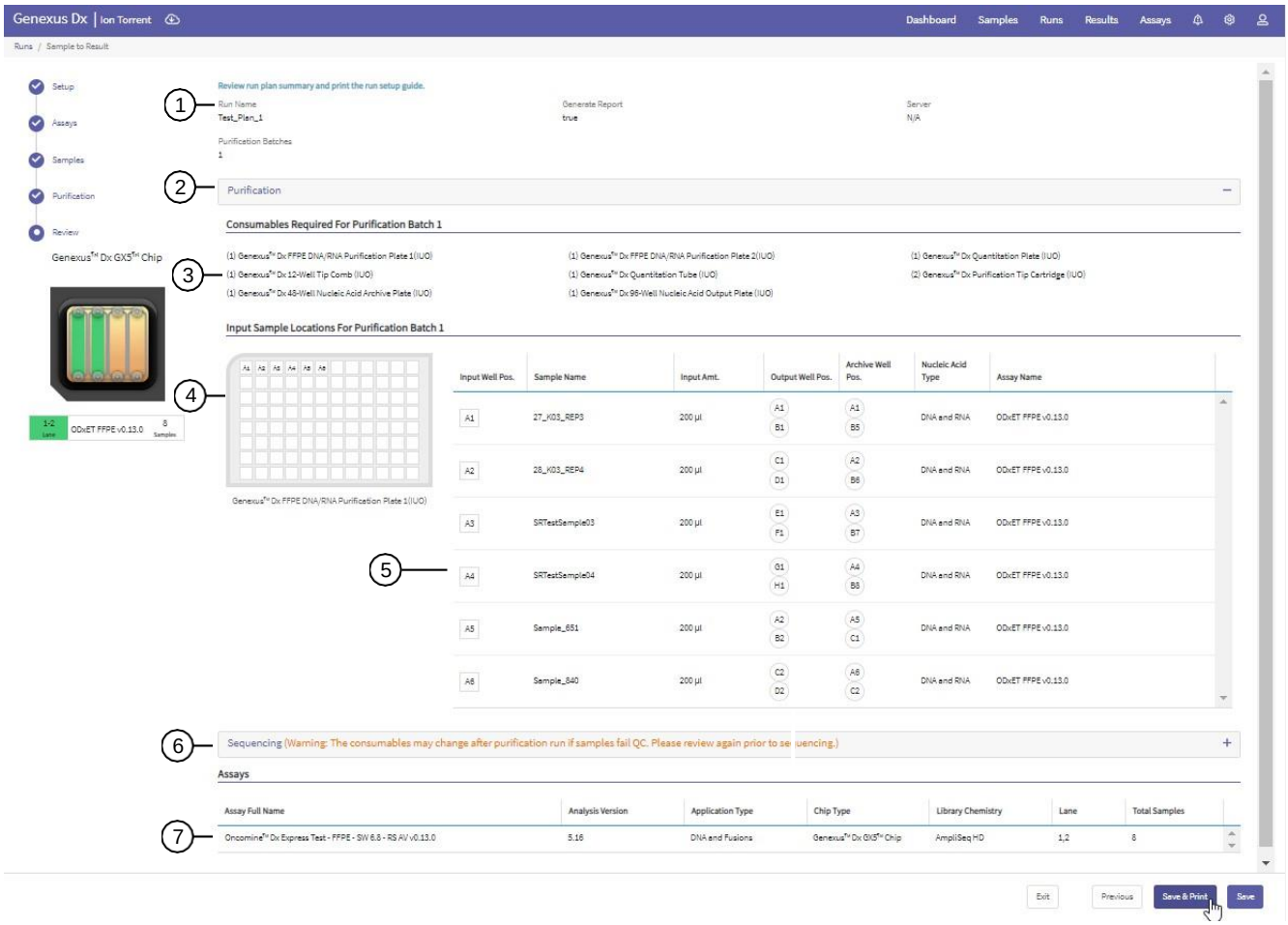
   If needed, you can enter the barcodes manually.

c. Click **Next**.

6. In the **Review** step, review the run setup guide, then click **Save**.

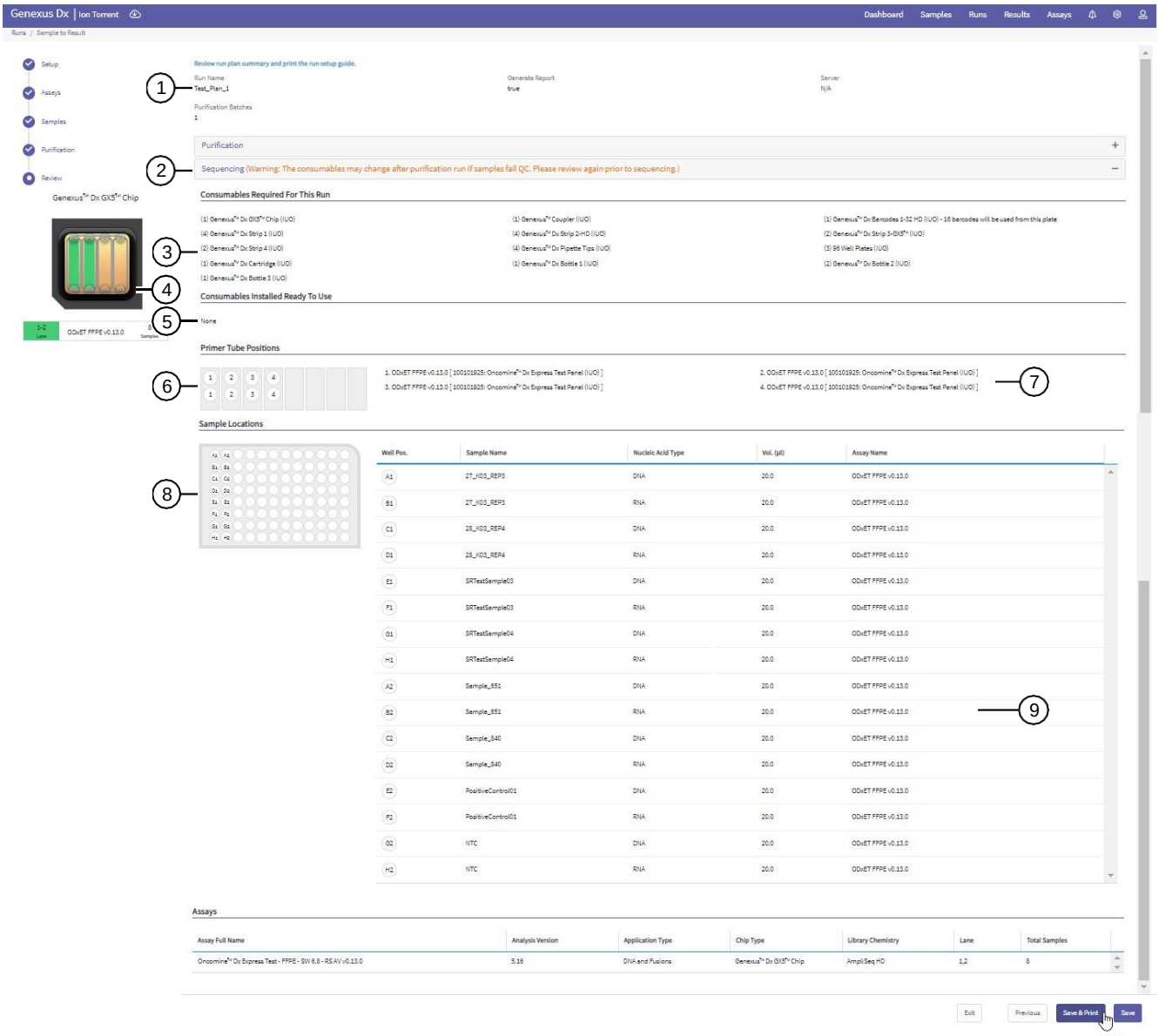
   The run plan summary lists the following details:

   - the consumables that are required for purification
   - how much volume to load
   - where to load samples
   - the consumables that are required for sequencing
   - details about the assay or assays.




① Run information
② Purification section expander
③ List of purification consumables needed for the run
④ Positions in the sample plate to load the samples
⑤ Table listing the input well position, sample name, input amount, output well position, nucleic acid type, and assay for each sample
⑥ Sequencing section expand/collapse bar
⑦ Assay information

Tip: Click **Sequencing** to expand the sequencing section of the run setup guide.




① Run information
② Sequencing section expand/collapse bar
③ List of sequencing consumables needed for the run
④ Chip view showing the lanes to be used in the run
⑤ List of consumables that are installed on the sequencer and available for the run
⑥ Positions to load primer pool tubes
⑦ Description of each primer pool and its position
⑧ Positions in the output plate where purified samples are located
⑨ Table listing the sample plate position, sample name, nucleic acid type, volume, and assay for each sample

IMPORTANT! The consumables required for sequencing can change. After purification, review the run plan in the **Runs / Manage Runs** screen for updated sequencing consumable information.

The run plan appears in the **Manage Runs** screen with the name that you specified.

After you select the run and load the Genexus™ Dx Purification System, the run is started on the purification instrument screen.

# Plan a Nucleic Acid to Result run

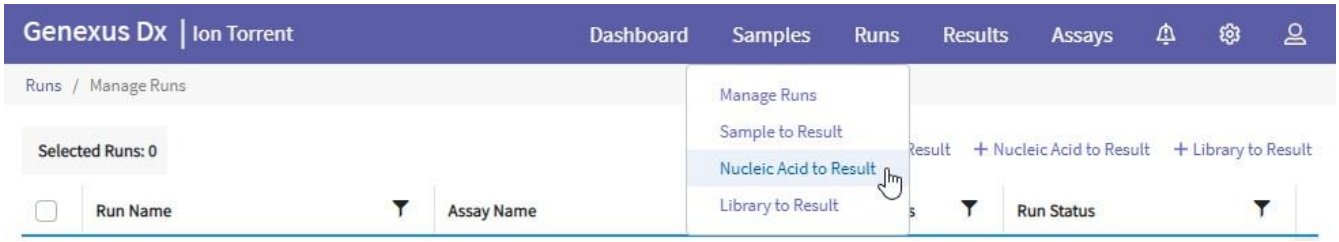
In Genexus™ Dx Software 6.8 and later, a run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Sample Plate**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Nucleic Acid to Result** screen.

Before you plan a run, do the following.

- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 37, or “Import samples” on page 39.
- Quantify sample concentration.
- Connect the barcode scanner to the computer on which you use Genexus™ Dx Software to plan runs and verify that the barcode scanner is compatible with the computer.

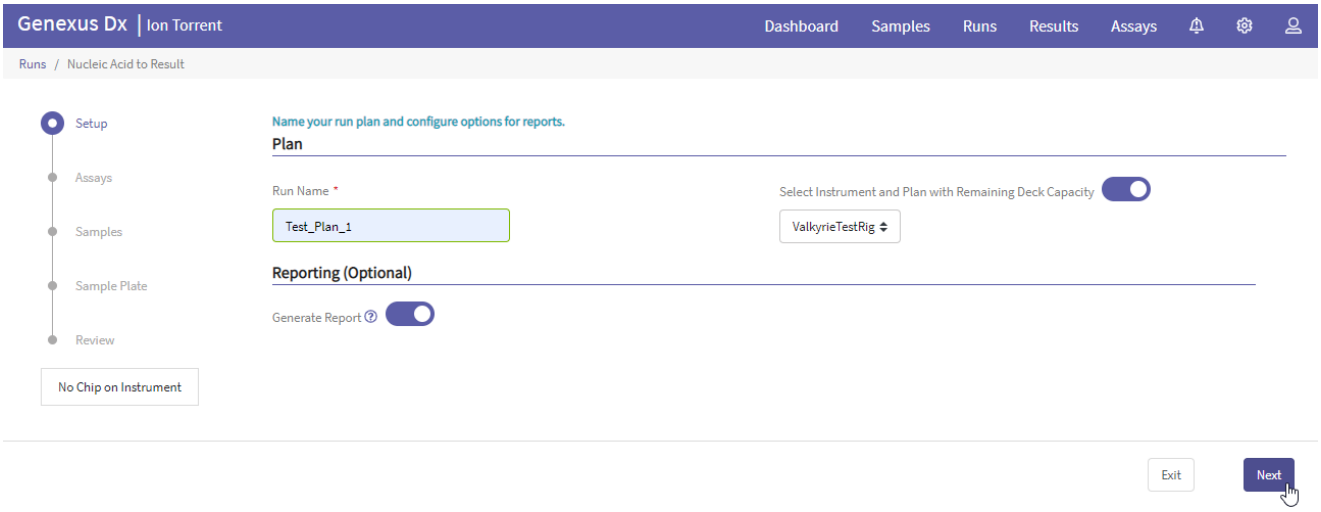
1. In the menu bar, click **Runs►Nucleic Acid to Result**.




Note: Alternatively, you can click + **Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.



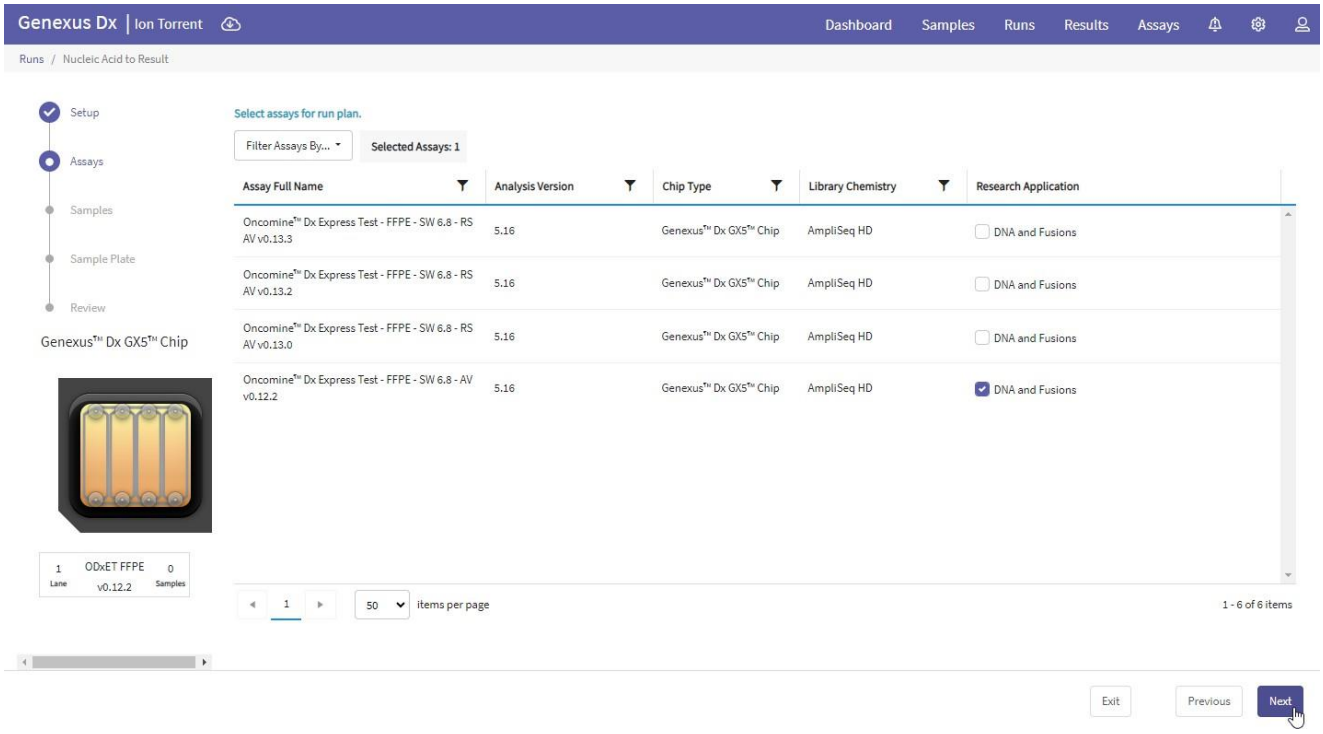

   b. *(Optional)* In the **Reporting (Optional)** section, select **Generate Report**, if needed.

      Note: Leave **Generate Report** enabled to generate a variant report using the default report template. To create a report template, click **Assays►Manage Presets**, then in the **Report Templates** tab, click + **Add New**.

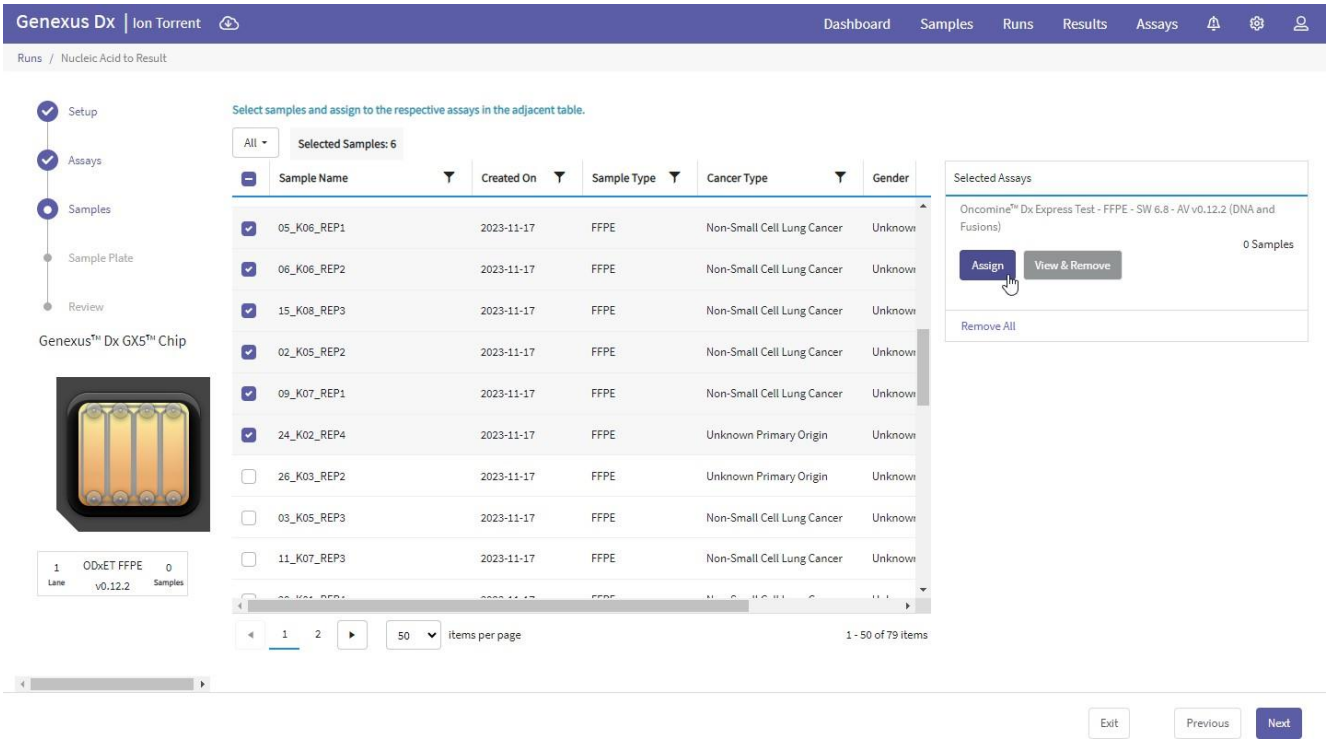
   c. Click **Next**.

      If a chip is installed in the sequencer, the chip view graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, in the **Research Application** column, select the assay that you want to include in the run, then click **Next**.



4. In the **Samples** step, select the samples that you want to run with the assay.
   a. Select the checkbox next to each sample that you want to include in the run, then in the **Selected Assays** pane, for the assay that you want to assign to the selected sample or samples, click **Assign**.




The chip view updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including positive and no-template controls) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity. If the

*minimum read count per sample × the number of samples*

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the chip view updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.

Green lane color indicates lane usage and sample assignment within lane capacity. Red lane color indicates sample assignment that exceeds lane capacity.

   b. If needed, edit samples in one of the following ways, then click **Next**.
      - Click **View & Remove**, make the selections, then click **Update**.
      - Click **Remove All**, make the selections, then click **Assign**.
   c. Click **Next**.

5. In the **Sample Plate** step, review position assignments in the sample plate.

   Appropriate controls are automatically included in the sample plate list:

   - Oncomine™ Dx Express Test FFPE DNA Control
   - Oncomine™ Dx Express Test FFPE RNA Control
   - Oncomine™ Dx Express Test NTC Control

   ---

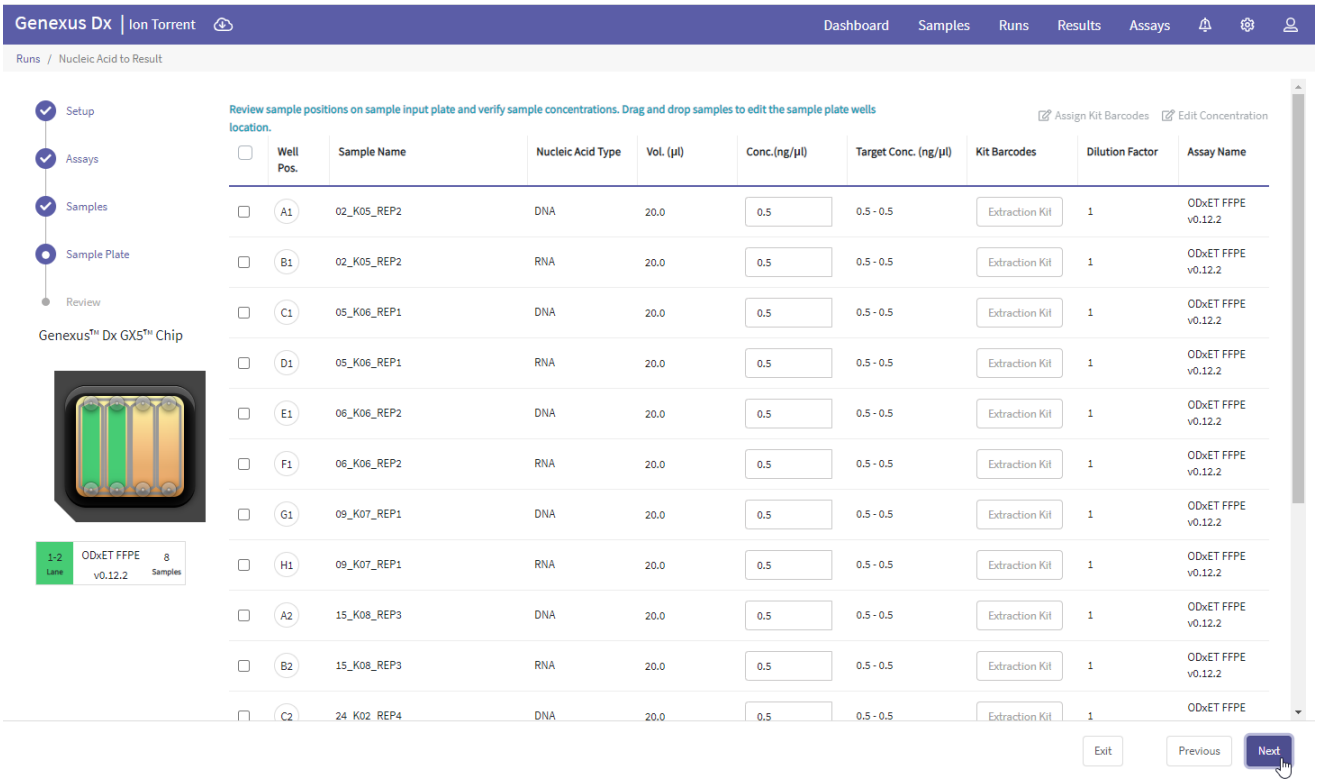
   Note: Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

   ---

   a. Use the connected barcode scanner to enter the kit barcode for one or more samples or controls. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples or controls, then click **Assign Kit Barcodes**. In the **Assign Kit Barcodes** dialog box, enter the kit barcode.

      If needed, you can enter the barcodes manually.

b. Modify the concentration of samples, if needed. For a single sample, in the row of the sample of interest, in the **Conc. (ng/µl)** column, edit the concentration.




To modify the concentration of multiple samples, select the samples of interest, then click **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration. Click **Submit** to return to the **Sample Plate** screen.

If a sample concentration is ≤1,024X of the target concentration for the assay, which is displayed as a default value for each sample in the **Sample Plate** screen in the **Target Conc. (ng/µL)** column, the sequencer automatically dilutes the sample to the target concentration during the run. If a sample concentration is greater than this value, you need to manually dilute the sample to the target concentration, or to a value within range for automated dilution before loading on the sample plate.

---

Note: The sample volume that is needed for library preparation is not adjustable. For more information, see “Load the sample plate—Nucleic Acid to Result run” for specific sample volumes to load on the sample plate.

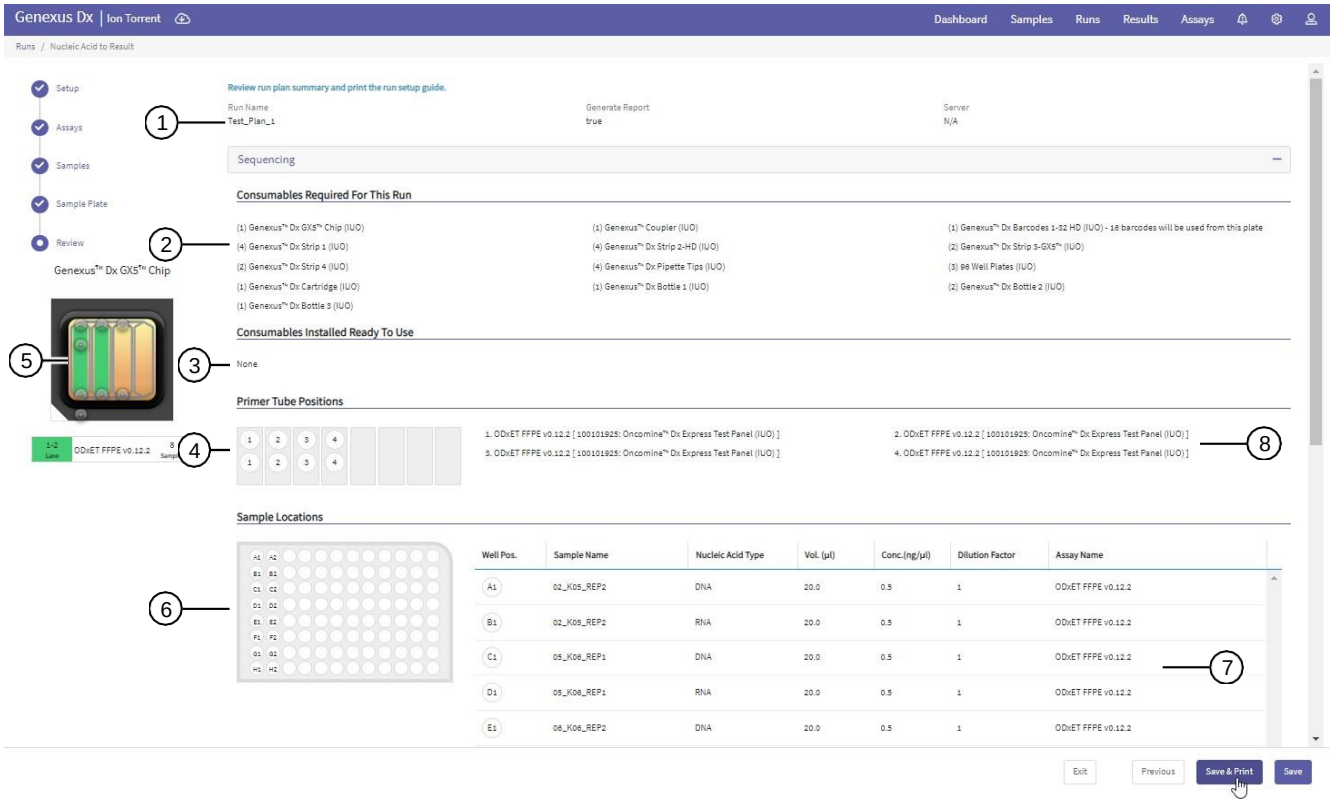
---

c. Ensure that sample plate information is correct, then click **Next**.

6. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.

   The run plan summary lists the following details:

- the consumables that are needed for this run
- how much sample volume to load
- where to load samples and primer pool tubes
- details about the assay.




1. Run information.
2. List of consumables needed for the run.
3. List of consumables that are installed on the sequencer and available for the run.
4. Positions to load primer pool tubes.
5. Chip view showing the lanes to be used in the run.
6. Positions in the sample plate to load the samples.
7. Table listing the sample plate position, sample type, volume to load, concentration, dilution factor, and assay for each sample.
8. Description of each primer pool and its position.

The run appears in the run list on the **Manage Runs** screen with the name you specified. Select the run on the sequencer screen during run setup.

# Plan a Library to Result run

You can plan a run that starts with prepared libraries that have been recovered from a previous Oncomine™ Dx Express Test run. This type of run is called a Library to Result sequencing run. The volume of library recoverable from a previous run depends on the number of samples that were included in the run.

---

IMPORTANT! Before planning a Library to Result run using pooled libraries recovered from a previous run, confirm that the run failed due to a templating or sequencing problem, and that a sufficient volume of pooled sample library is available in well E4 of the PCR amplification plate. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 217.

---

Before you can plan a Library to Result run, you need to first prepare a library batch that associates an assay to be used in the run with the library batch. For more information, see “Prepare or import a library batch” on page 41.

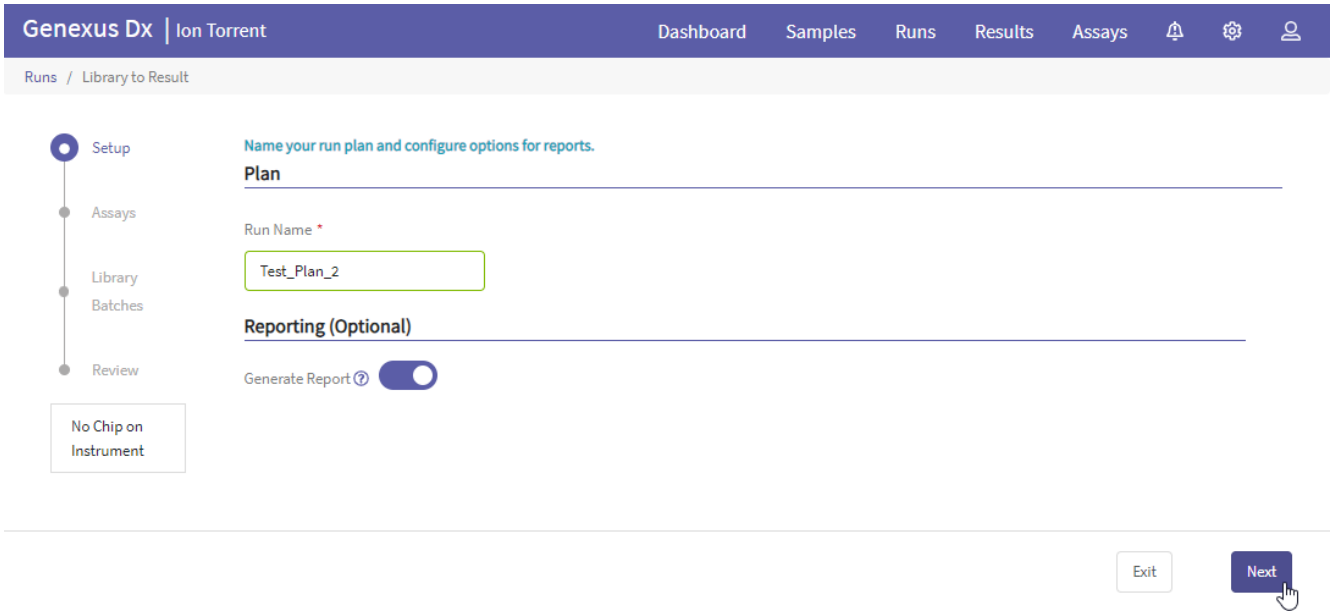
Genexus™ Dx Software guides you through the four steps of planning a Library to Result run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs►Library to Result**.

   Note: You can also click + **Library to Result** in the **Runs / Manage Runs** screen.



2. In the **Setup** step, enter a name for the run, then configure the reporting options.
   a. In **Run Name**, enter a unique name.
      The name is limited to 50 characters and no spaces are allowed.



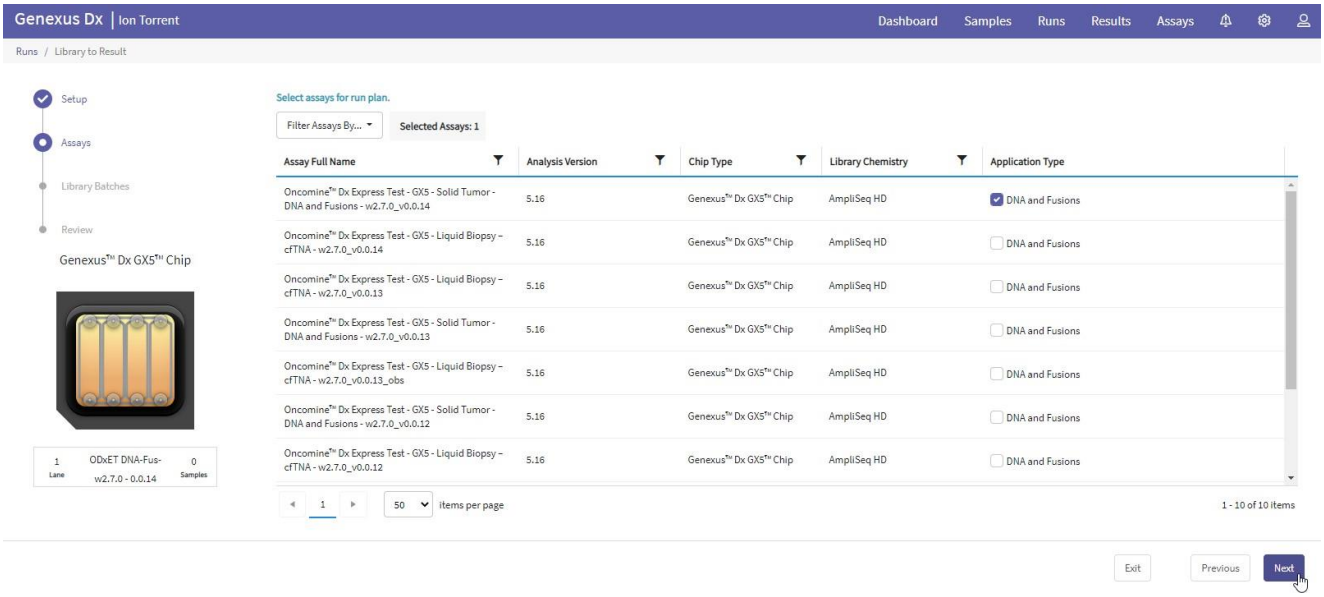

   b. *(Optional)* In the **Reporting (Optional)** section, select **Generate Report**, if needed.

      Note: Leave **Generate Report** enabled to generate a variant report using the default report template. To create a report template, click **Assays**►**Manage Presets**, then in the **Report Templates** tab, click + **Add New**.

   c. Click **Next**.
      If a chip is installed in the sequencer, the chip view graphic in the lower left corner indicates the lanes that are available for sequencing.

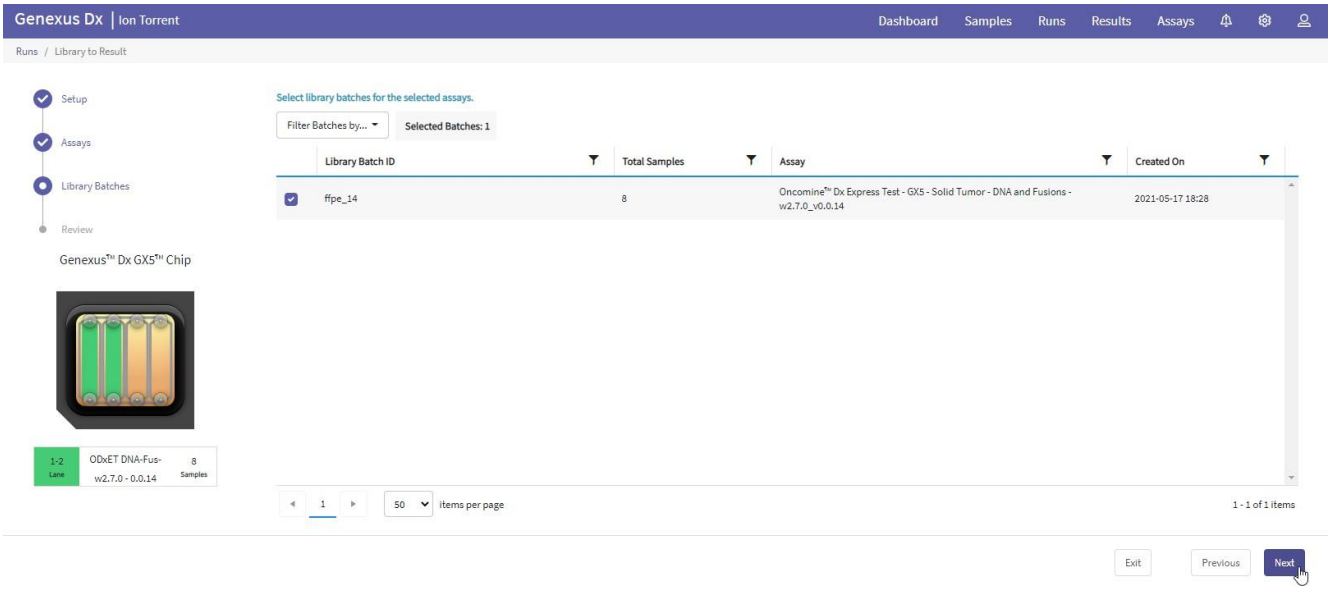
3. In the **Assays** step, select the assay that you want to use in the run, then click **Next**.

   For the assay to be selectable at this step, you need to have prepared a library batch that assigns the assay to the batch.




4. In the **Library Batches** step, select the library batch that you want to use in the run.

   Note: Only one library batch can be selected per assay.




   The chip view updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum read counts per sample for the assay.

   Green lane color indicates lane usage and sample assignment within lane capacity. Red lane color indicates sample assignment that exceeds lane capacity.

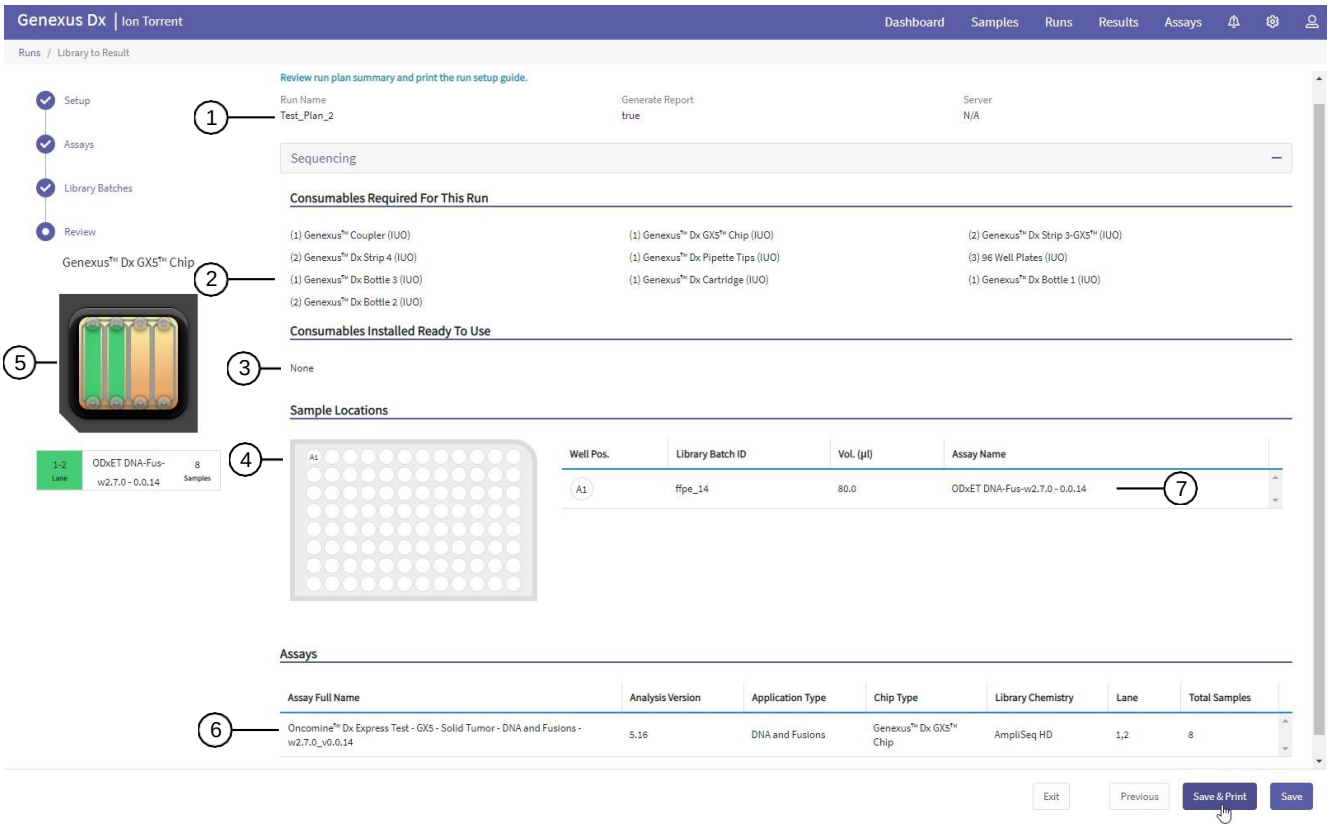
If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the chip view updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not obtain the required reads per sample to pass QC metrics.

5. Click **Next**.

6. In the **Review** step, review the run setup guide.

   - Click **Save and Print** to print the run setup guide
   - Click **Save** to save the run without printing.

   The run plan summary lists the consumables that are needed for the run, where to load the library batch on the sample plate, and how much library volume to load.




① Run information.
② List of consumables needed for the run.
③ List of consumables installed on the sequencer and available for the run.
④ Position or positions in the sample plate to load the library batch.
⑤ Chip view showing the lanes to be used in the run.
⑥ Table listing assay and run information.
⑦ Table listing the well position, library batch ID, volume to load, and assay for each library batch.

The run appears in the run list on the **Manage Runs** screen with the name you specified. Select the run on the sequencer screen during run setup.

# Plan a Genexus™ Dx FFPE DNA and RNA Purification run—standalone configuration

If you are purifying DNA and RNA from FFPE samples on the Genexus™ Dx Purification System in a standalone configuration, create a plan for the run using the touchscreen of the purification instrument. In the standalone instrument configuration, all purified sample DNA and RNA is loaded in an archive plate.

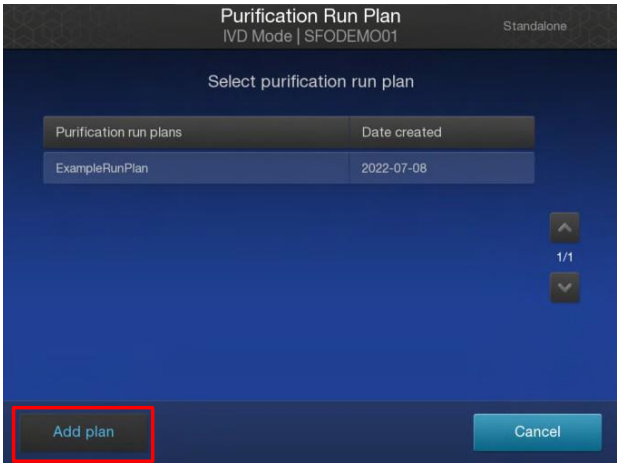
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Note: We recommend that you plan the run before preparing the samples and loading into the Genexus™ Dx FFPE DNA and RNA Purification Plate 1. However, experienced users can save time by planning the purification run during the protease digestion step of sample preparation.

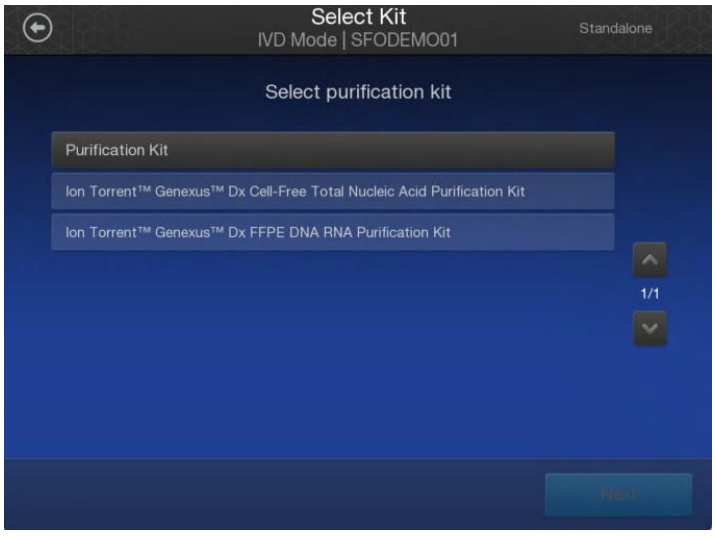
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1. Enter your username and password to sign in to the instrument.
2. Tap **Run**, then tap **Add plan**.

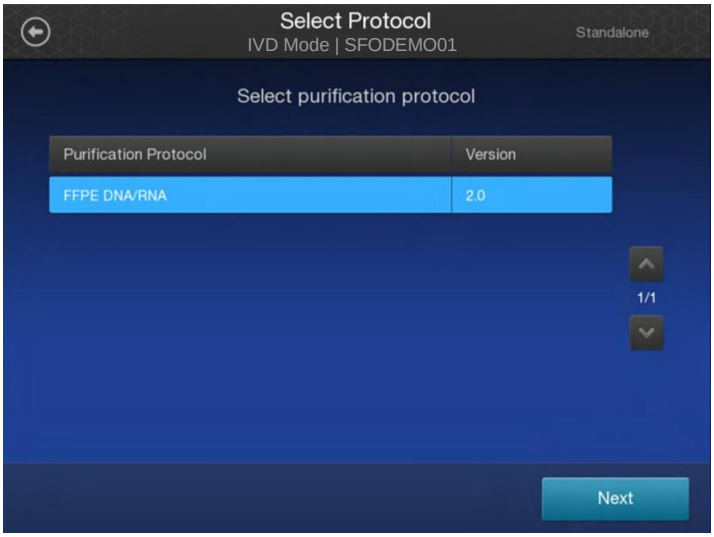



3. Tap in the entry box, enter a unique name for the run plan, then tap **Done►Next**.
4. Select the Ion Torrent™ Genexus™ Dx FFPE DNA and RNA Purification Kit, then tap **Next**.



5. Select the **FFPE DNA/RNA** purification protocol, then tap **Next**.



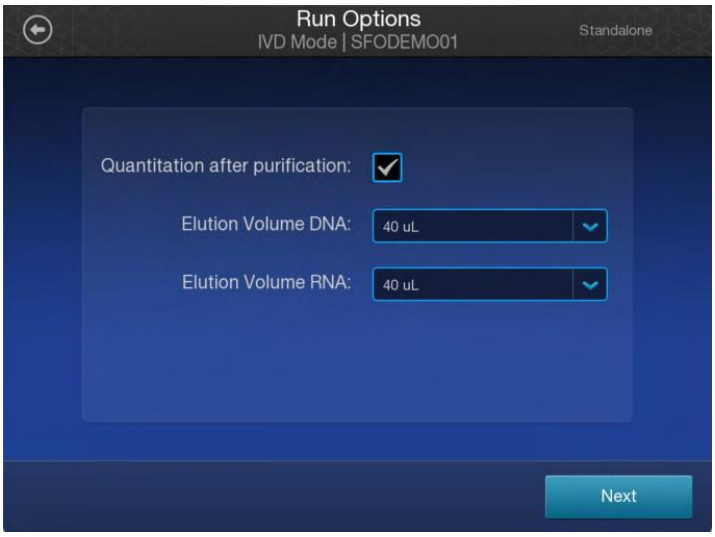

6. Enable or disable **Quantitation after Purification**.

---

Note:

- The Quantitation Plate is required even if **Quantitation after Purification** is disabled.
- Disabling **Quantitation after Purification** can reduce the purification run time by up to 2.5 hours.

---




7. Accept the default elution volume. If needed, select the desired elution volume from the dropdown list, then tap **Next**.

8. *(Optional)* Change the number of samples and the sample details.
   a. In the **Manage Samples** screen, deselect extra samples (for example, if you are only running 10 samples, deselect samples 11 and 12).

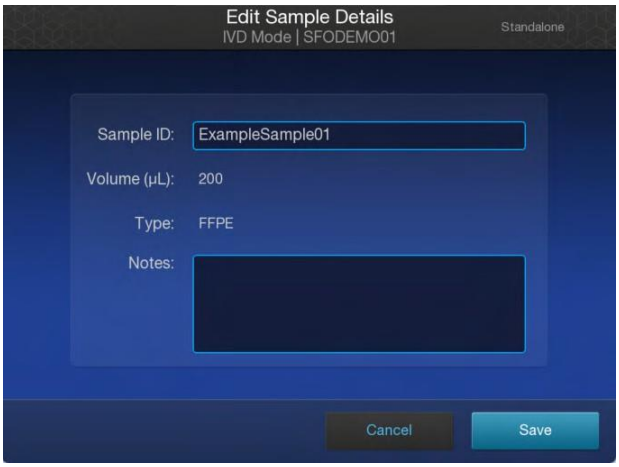
b. Tap on a sample ID to select the sample.

c. Tap **Edit**, enter a new **Sample ID** and any **Notes**, then tap **Save**.

d. Repeat substep 8b and substep 8c for each additional sample.

e. Click **Next**.




9. *(Optional)* Import samples and the sample details.

---

Note:

- Importing sample information overwrites the existing sample information in the run plan.
- In the standalone run configuration, you must prepare a CSV sample import file and save it to a USB drive to import sample information. For more information, see page 208.

---

a. In the **Manage Samples** screen, select the samples for which sample information is being imported, then tap **Import**.

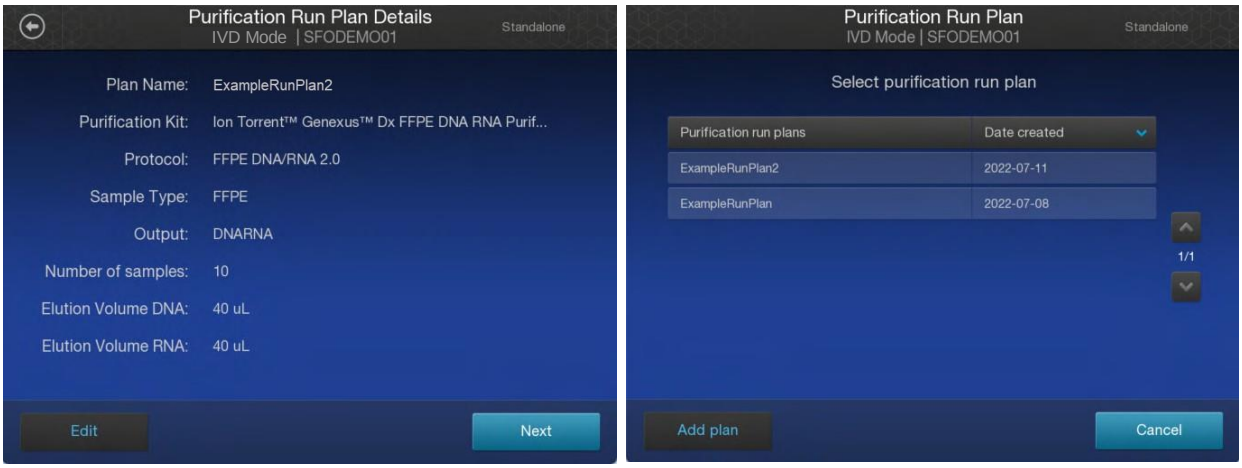
b. The **Sample Import** screen opens. Tap **Import** to proceed.

c. Insert the USB drive containing the sample import CSV file into the USB port on the front of the purification instrument. In the **Sample Import** screen, select the USB drive, then navigate to and select the sample import file.

d. *(Optional)* Tap **Details** to view the CSV file listing the sample names to be imported.

e. Tap **Import**, then in the **Import Successful** screen, tap **OK**.
The imported sample information is shown in the **Manage Samples** screen. Edit imported sample information if needed as described in step 8.

10. Review the **Purification Run Plan Details**. Tap **Edit** to change any of your selections, otherwise tap **Next**.




The new run plan appears in the list of available purification run plans on the Genexus™ Dx Purification System. Run plans can be edited and reused.

To delete an existing run plan, see “Delete a run plan in the Genexus™ Dx Purification System touchscreen (standalone configuration)” on page 209.

# 5 Start a purification run for FFPE samples


- Prepare FFPE samples ..... 71
- Load the instrument and start the run ..... 78
- Unload and UV clean the instrument ..... 87
- View and export quantitation results ..... 90


This chapter describes how to prepare FFPE samples and perform a purification run on the Genexus™ Dx Purification System in both integrated and standalone configurations. In an integrated run configuration, the sample purification and sequencing workflows are integrated in a Sample to Result run that you plan in Genexus™ Dx Software (see "Plan a Sample to Result run" on page 48). After purification is complete, sample DNA and RNA that is ready for sequencing is loaded by the instrument in a sample output plate.

In a standalone run configuration, the purification only workflow is set up in a run plan that you create in the touchscreen of the Genexus™ Dx Purification System (see "Plan a Genexus™ Dx FFPE DNA and RNA Purification run—standalone configuration" on page 67). After purification is complete, all purified sample DNA and RNA is loaded in an archive plate for storage or for use in a Nucleic Acid to Result sequencing run.

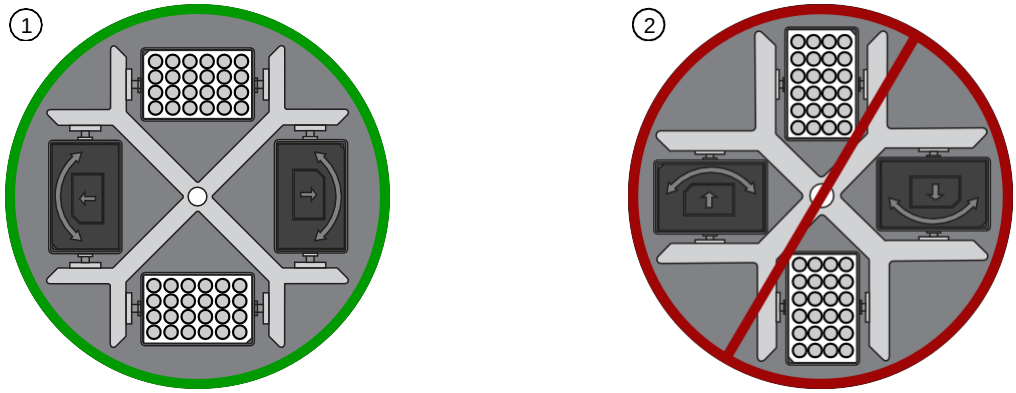
## Prepare FFPE samples

### Procedural guidelines

IMPORTANT! Store all kit components containing liquid in the upright orientation.

- Perform all steps at room temperature (15–30°C) unless otherwise noted.
- Thawing or storing on ice can be substituted with thawing or storing at 4°C (2–8°C refrigerator or prechilled benchtop cold block).
- When mixing samples by pipetting up and down, avoid creating bubbles.
- Wear clean gloves and a clean laboratory coat.
- Change gloves whenever you suspect that they are contaminated.
- Open and close all sample tubes carefully. Avoid splashing or generating aerosols of the samples.

- When working with RNA:
  - Use a positive-displacement pipettor and RNase-free pipette tips.
  - Clean laboratory benches and equipment periodically with an RNase decontamination solution, such as RNase*Zap*™ RNase Decontamination Solution (Cat. No. AM9780) or RNase *AWAY*™ Surface Decontaminant (Cat. No. 7005-11).
  - Store RNA at –90°C to –70°C.
- Volumes for reagent mixes are given per sample. We recommend that you prepare master mixes for larger sample numbers. To calculate volumes for master mixes, see the per-well volume and add 5–10% overage.
- We recommend using a plate centrifuge that holds the AutoLys M Tube Rack in "landscape" orientation.




1 Landscape orientation—recommended

2 Portrait orientation—not recommended

Note: Place AutoLys M Tube Rack in the centrifuge with the arrow on the cover pointing outward as shown.

- The plate chiller on the Genexus™ Dx Purification System shuts off 60 minutes after run completion. Remove the 96-Well Nucleic Acid Output Plate and Genexus™ Dx 48-Well Nucleic Acid Archive Plate with purified nucleic acids from the instrument within 1 hour of run completion. Proceed immediately to sequencing or properly store the nucleic acids until use.
- The Genexus™ Dx Quantitation Plate requires equilibration to room temperature for at least 30 minutes before use.

## Before each use of the kit

- Preheat one incubator to 60°C and one incubator to 90°C.
- Prepare Protease Digestion and DNase Digestion solutions immediately before use.
- Centrifuge purification plates for 30 seconds at 1,000 × *g* to collect the contents.

## Materials required

- Genexus™ Dx FFPE DNA and RNA Purification (Part. No. A51076)
  - Genexus™ Dx Proteinase K (red cap)
  - Genexus™ Dx FFPE Protease Buffer
- AutoLys M TubeLifter or Pliers
- AutoLys M Tubes and Caps
- AutoLys M Tube Rack
- Plate centrifuge
- Two incubators—one set at 60°C, and one set at 90°C

## Recommended input amount

| Sample type | Recommended input |
|---|---|
| FFPE tumor resections | 2 × 5-µm curls/slides or equivalent amount |
| FFPE core needle biopsy (CNB) | 9 × 5-µm curls/slides or equivalent amount |
| FFPE fine needle aspirate (FNA) | 7 × 5-µm curls/slides or equivalent amount |

## Prepare 1X Protease Digestion Master Mix

Prepare the 1X Protease Digestion Master Mix immediately before use.

1. Invert the Genexus™ Dx FFPE Protease Buffer and Genexus™ Dx Proteinase K tubes supplied in the kit 5X each, then briefly centrifuge.

2. In a 5–10-mL tube, prepare a 1X Protease Digestion Master Mix as indicated, where $n$ is the number of tissue samples.

| Component | Volume per reaction |
|---|---|
| Genexus™ Dx FFPE Protease Buffer | $(n + 1) \times 225$ µL |
| Genexus™ Dx Proteinase K (red cap) | $(n + 1) \times 10$ µL |
| Total volume | $(n + 1) \times 235$ µL |

3. Vortex for ~5 seconds to mix, then briefly centrifuge to collect the contents.

# Prepare FFPE curls with AutoLys M Tubes

## Digest with Protease in AutoLys M Tubes

Note: To minimize the amount of time between protease digestion and starting the purification run on the instrument, prepare the reagents and consumables that are required by the instrument during the 90°C incubation (step 6).

1. Label an AutoLys M Tube for each FFPE tissue sample.
2. Transfer each FFPE section curl from its storage tube to a separate labeled AutoLys M Tube.
   a. Invert a labeled AutoLys M Tube over an uncapped storage tube containing the curl, then carefully invert the 2 tubes together so that the storage tube is on top.
   b. Tap or flick the storage tube so that the curl falls into the AutoLys M Tube.

   If the curl adheres to the storage tube, gently grasp an edge of the curl with forceps, then transfer it to the AutoLys M Tube. Clean the forceps between samples by rinsing with nuclease-free water, followed by ethanol or wiping with ethanol wipes. Do not reuse the forceps without cleaning. Alternatively, use sterile disposable forceps.
3. Cap the AutoLys M Tubes tubes, place the tubes in an AutoLys M Tube Rack, then centrifuge at 2,000 × *g* for 1 minute to collapse the curl before the addition of buffer.
4. Pipet 235 µL 1X Protease Digestion Master Mix into each labeled tube.

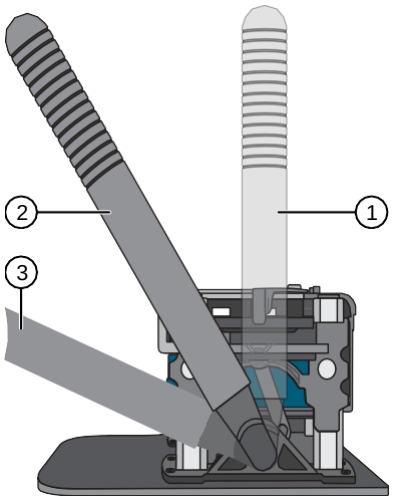
   Ensure that the samples are submerged in the Protease Digestion Master Mix. If tissue adheres to the side of the tube, use a sterile pipette tip to push the sample into the solution.
5. Securely recap each tube to limit evaporation, then incubate the capped tubes at 60°C for ≥60 minutes in an AutoLys M Tube Rack.

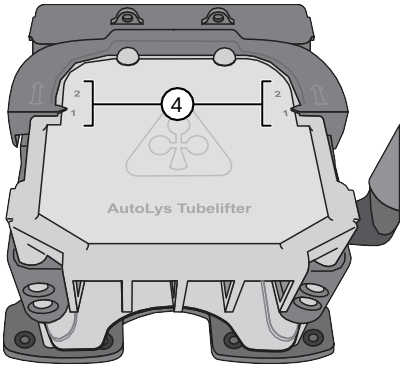
   Incubation at 60°C can be extended to 2 hours to increase DNA yields. We recommend that you incubate low yielding samples such as core needle biopsy and fine needle aspirate samples for 2 hours to maximize yield.
6. Incubate at 90°C for 60 minutes.
   - Prepare the reagents and consumables that are required by the instrument during the incubation. See “Prepare the consumables” on page 79.
   - Equilibrate the Quantitation Plate to room temperature during the incubation.
7. Allow samples to cool to room temperature for at least 5 minutes and up to 10 minutes before proceeding to lift the tubes.
8. Lift the tubes. The following steps describe use of the AutoLys M TubeLifter to process up to 24 samples simultaneously. Alternatively, AutoLys M Tube Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys M TubeLifter lever is in Position A (straight up) and the slider is in Position 1.

b. Slide the 24-well AutoLys M Tube Rack containing the lysed samples into the AutoLys M TubeLifter.

c. Press the lever down from Position A to Position B, then remove the rack from the lifter.

For more information about use of the AutoLys M TubeLifter see the *AutoLys M TubeLifter User Guide* (Pub. No. MAN0017676).




① AutoLys M TubeLifter lever Position A
② AutoLys M TubeLifter lever Position B
③ AutoLys M TubeLifter lever Position C
④ AutoLys M TubeLifter slider positions 1 & 2

9. Slide the AutoLys M Tube Locking Lid onto the rack, then centrifuge the samples at 2,000 × *g* for 10 minutes.

IMPORTANT! Ensure that the embossed arrow of the AutoLys M Tube Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge. See “Procedural guidelines” on page 71.

10. Separate the filter from the outer tube.
    a. Adjust the position of the AutoLys M TubeLifter slider to Position 2.
    b. Remove the AutoLys M Tube Locking Lid, then slide the rack into the AutoLys M TubeLifter.
    c. Press the lever down from Position B to Position C.

    Keep the samples on ice or at 4°C.

Proceed to “Add samples to Genexus™ Dx FFPE DNA and RNA Purification Plate 1” on page 80.

STOPPING POINT If needed, transfer samples to a separate labelled 1.5-mL Eppendorf™ LoBind™ tube and store at 4°C for up to 4 days or frozen at –30°C to –10°C for up to 7 days.

# Prepare FFPE slide sections with AutoLys M Tubes

## Collect the tissue

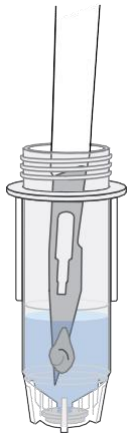
1. Label an AutoLys M Tube for each FFPE tissue sample.

   Label each tube (cap and side) with its Sample ID using a marker that is resistant to xylene and ethanol.

2. Pipet 235 µL 1X Protease Digestion Master Mix into each labeled tube.

3. Use a sterile disposable scalpel with a flat blade to scrape the tissue in a single direction, then collect the tissue into a cohesive mass on the tip of the scalpel blade.

4. Carefully insert the scalpel blade with the tissue mass into the 1X Protease Digestion Master Mix in the AutoLys M Tube. Rinse the tissue from the blade into the buffer, then ensure that the entire mass is in solution.



5. Remove and inspect the blade to ensure that no tissue remains on it.

6. Inspect the slide to ensure that all the tissue is removed (the slide should be translucent). Discard the scalpel in a waste container for sharp objects.

7. Cap each tube securely, then gently flick the tube to mix and to immerse the tissue.

   If the tissue adheres to the side of the tube, use a sterile pipette tip to push the tissue into the solution.

## Digest with protease

To minimize the amount of time between protease digestion and starting the purification run on the instrument, prepare the reagents and consumables that are needed by the instrument during the 90°C incubation (step 2).

1. Incubate at 60°C for ≥60 minutes in an AutoLys M Tube Rack.

   Incubation at 60°C can be extended to 2 hours to increase DNA yields. We recommend that you incubate low yielding samples such as core needle biopsy and fine needle aspirate samples for 2 hours to maximize yield.

   ---

   Note: Ensure that the incubator loaded with sample tubes has sufficient space to allow the rapid transfer of heat within the incubator—do not overcrowd with tubes.

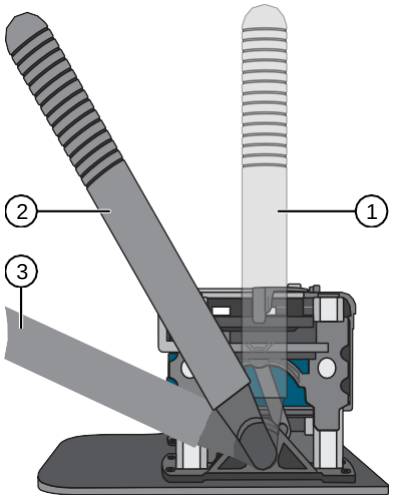
   ---

2. Incubate at 90°C for 60 minutes.

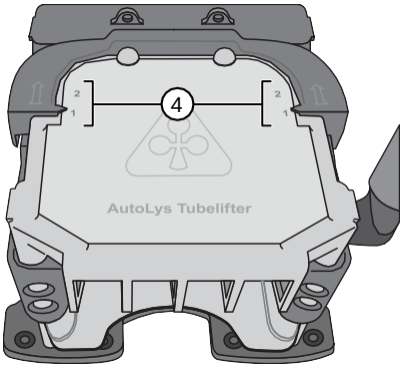
   - Prepare the reagents and consumables that are needed by the instrument during the incubation. See “Prepare the consumables” on page 79.
   - Equilibrate the Genexus™ Dx Quantitation Plate to room temperature during the incubation.

3. Allow samples to cool to room temperature for at least 5 minutes and up to 10 minutes before proceeding to lift the tubes.

4. Lift the tubes. The following steps describe use of the AutoLys M TubeLifter to process up to 24 samples simultaneously. Alternatively, AutoLys M Tube Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys M TubeLifter lever is in Position A (straight up) and the slider is in Position 1.
   b. Slide the 24-well AutoLys M Tube Rack that contains the lysed samples into the AutoLys M TubeLifter.
   c. Press the lever down from Position A to Position B, then remove the rack from the lifter.




① AutoLys M TubeLifter lever Position A
② AutoLys M TubeLifter lever Position B
③ AutoLys M TubeLifter lever Position C




④ AutoLys M TubeLifter slider positions 1 & 2

5. Slide the AutoLys M Tube Locking Lid onto the rack, then centrifuge the samples at 2,000 × *g* for 10 minutes.

   IMPORTANT! Ensure that the embossed arrow of the AutoLys M Tube Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge (see “Procedural guidelines” on page 71).

6. Separate the filter from the outer tube.
   a. Adjust the position of the AutoLys M TubeLifter slider to Position 2.
   b. Remove the AutoLys M Tube Locking Lid, then slide the rack into the AutoLys M TubeLifter.
   c. Press the lever down from Position B to Position C.

   Keep the samples on ice or at 4°C.

Proceed to “Add samples to Genexus™ Dx FFPE DNA and RNA Purification Plate 1” on page 80.

STOPPING POINT If needed, transfer samples to a separate labelled 1.5-mL Eppendorf™ LoBind™ tube and store at 4°C for up to 4 days or frozen at –30°C to –10°C for up to 7 days.

# Load the instrument and start the run

This section describes how to perform the following procedures.

- Set up the instrument for use by loading all of the required reagents and consumables.
- Start a run.

Note: Do NOT load any consumables onto the instrument until after the instrument has performed the prerun UV cleaning.

## Materials required

- Genexus™ Dx FFPE DNA and RNA Purification (Part. No. A51076)
  - Genexus™ Dx FFPE DNA and RNA Purification Plate 1
  - Genexus™ Dx FFPE DNA and RNA Purification Plate 2
  - Genexus™ Dx DNase (yellow cap)
  - Genexus™ Dx DNase Buffer (blue cap)
  - Genexus™ Dx 12-Well Tip Comb
- Genexus™ Dx Nucleic Acid Quantitation (Part. No. A51078)
  - Genexus™ Dx Quantitation Plate
  - Genexus™ Dx Quantitation Tube
- Genexus™ Dx Purification Supplies 2 (Part. No. A51081)
  - 2 Genexus™ Dx Purification Tip Cartridges
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal
- 96-Well Nucleic Acid Output Plate
- P200 pipette and filtered tips

# Prepare the consumables

Note: Consumables can be prepared during the protease digestion 90°C incubation step to save time.

Remove all cartridges and consumables from their packaging, then place them on the bench at room temperature. Prepare the following cartridges and consumables.

- Genexus™ Dx Purification Supplies 2
  - 2 Genexus™ Dx Purification Tip Cartridges
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal
- Genexus™ Dx 12-Well Tip Comb

## Equilibrate the Genexus™ Dx Quantitation Plate

Note: The Genexus™ Dx Quantitation Plate can be equilibrated to room temperature during the protease digestion to save time.

1. Remove the Genexus™ Dx Quantitation Plate from storage at 2°C–8°C and equilibrate the plate to room temperature for at least 30 minutes.
2. Place the plate and Genexus™ Dx Quantitation Tube on the bench next to the Genexus™ Dx Purification System.

   IMPORTANT! Protect the Genexus™ Dx Quantitation Plate from light to prevent photobleaching of the preloaded reagents.

## Add 1X DNase Digestion Master Mix to the Genexus™ Dx FFPE DNA and RNA Purification Plate 2

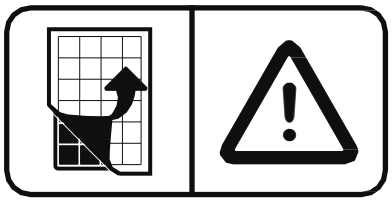
The Genexus™ Dx FFPE DNA and RNA Purification Plate 2 contains magnetic beads in row H.

1. Vortex the Genexus™ Dx DNase Buffer and Genexus™ Dx DNase supplied in the kit for ~5 seconds each, then briefly centrifuge to collect the contents.
2. In a 1.5-mL low-retention microcentrifuge tube, prepare a 1X DNase Digestion Master Mix as indicated, where `n` is the number of tissue samples.

| Component | Volume per reaction |
|---|---|
| Genexus™ Dx DNase Buffer (blue cap) | (n + 1) × 99 µL |
| Genexus™ Dx DNase (yellow cap) | (n + 1) × 1.0 µL |
| Total volume | (n + 1) × 100 µL |

3. Vortex for ~5 seconds to mix, then briefly centrifuge to collect the contents.
4. Centrifuge the Genexus™ Dx FFPE DNA and RNA Purification Plate 2 at 1,000 × *g* for 30 seconds to collect the contents.

5. Carefully remove the plate seal without disturbing the contents.



6. Pipet 100 µL 1X DNase Digestion Master Mix into each well that is used in Row A of the Genexus™ Dx FFPE DNA and RNA Purification Plate 2.

## Add samples to Genexus™ Dx FFPE DNA and RNA Purification Plate 1

The Genexus™ Dx FFPE DNA and RNA Purification Plate 1 contains magnetic beads in row B.

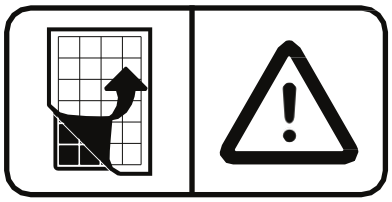
Note: You can add the samples to the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 and load the plate as the final steps of loading the instrument. This can be done to help ensure that other components are successfully loaded and accepted by the instrument before adding samples of possibly limited supply to the purification plate.

1. Centrifuge the plate at 1,000 x *g* for 30 seconds to collect the contents.

   IMPORTANT! Do not create bubbles when preparing the plate.

2. Inspect the plate to ensure that the contents of all rows are at the bottom of the wells.

3. Carefully remove the plate seal without disturbing the contents.



4. Transfer 200 µL of each sample to an individual well in row A of the prefilled Genexus™ Dx FFPE DNA and RNA Purification Plate 1.

   Add samples to consecutive wells starting with sample 1 in well A1, through sample 12 in well A12 as defined in the run plan. Do not skip wells.

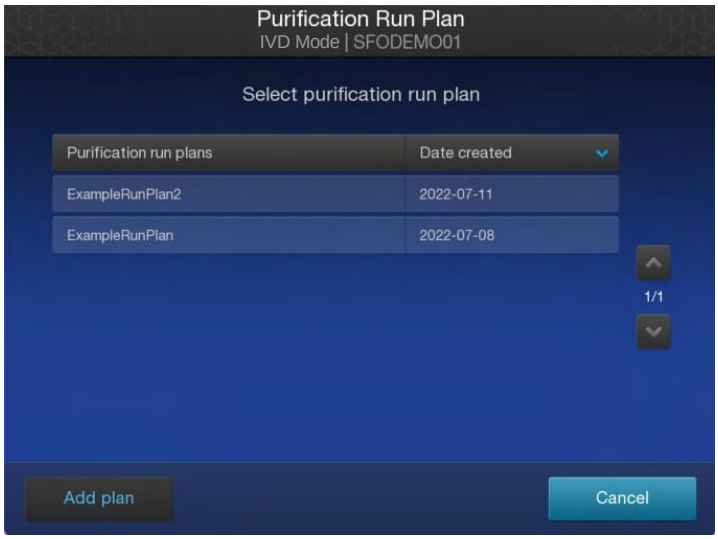
   A precipitate can form, but this does not interfere with the DNA binding. Proceed directly to the next step.

   Reagent consumables cannot be reused.

   IMPORTANT! Load the plate in the instrument (see page 82) within 15 minutes of adding samples to the plate.

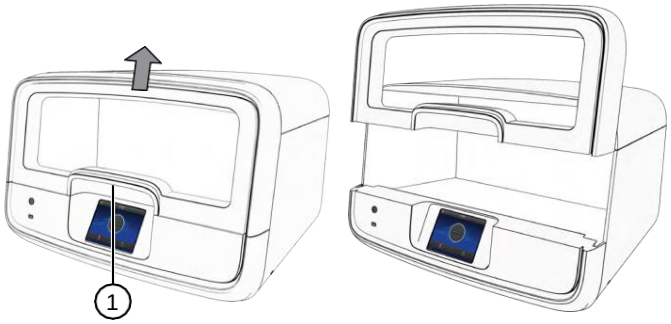
## Start the purification run

1. In the instrument touchscreen, tap **Run**, then tap to select the run plan that you created for this run.




2. Ensure that the correct run plan is selected, then proceed through a series of screens to review and confirm purification batch, run plan details, and sample assignment. At each screen, tap **Next** to proceed.
   After you proceed through the final screen, the instrument performs a 2-minute UV cleaning, then unlocks the door.

3. Lift the instrument door to the stop.



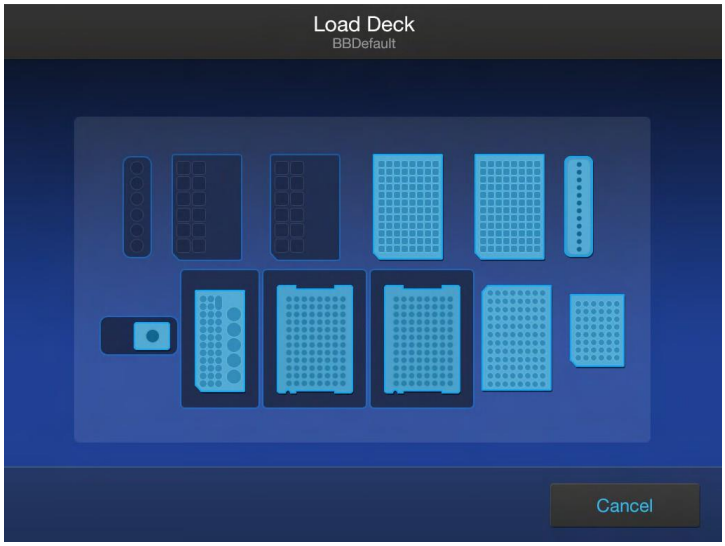

① Hold here, then lift.

### Load the Genexus™ Dx Purification System

IMPORTANT!

- Ensure that all components are clean and dry before loading them onto the Genexus™ Dx Purification System.
- Ensure that the Reagent and Quantitation station compartments are free of condensate before loading components. If needed, use a lint-free wipe to dry the compartment.

Follow the on-screen prompts to load the Genexus™ Dx Purification System.

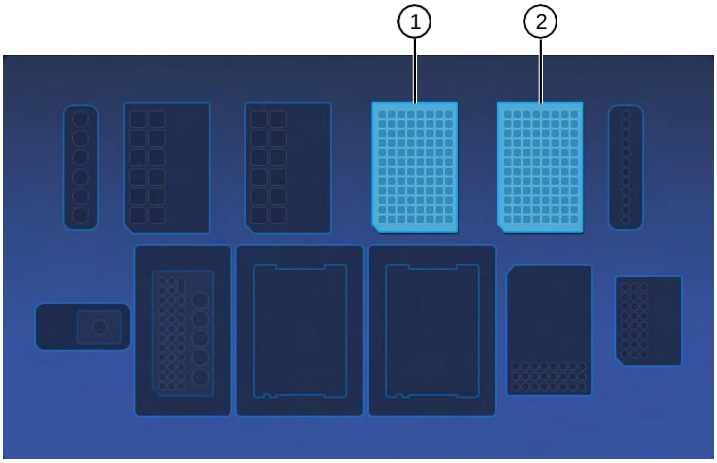



Screen display for an instrument fully loaded for an FFPE sample purification run (integrated configuration).

## Load Genexus™ Dx FFPE DNA and RNA Purification Plate 1 & 2

1. Load the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 (DNA plate) prepared in step 4 of "Add samples to Genexus™ Dx FFPE DNA and RNA Purification Plate 1" on page 80.

2. Load the Genexus™ Dx FFPE DNA and RNA Purification Plate 2 (RNA plate) prepared in step 6 of "Add 1X DNase Digestion Master Mix to the Genexus™ Dx FFPE DNA and RNA Purification Plate 2" on page 79.




① Genexus™ Dx FFPE DNA and RNA Purification Plate 1 position
② Genexus™ Dx FFPE DNA and RNA Purification Plate 2 position

## Load the Genexus™ Dx 12-Well Tip Comb, Genexus™ Dx Purification Tip Cartridges, 96-Well Nucleic Acid Output Plate, and Genexus™ Dx 48-Well Nucleic Acid Archive Plate

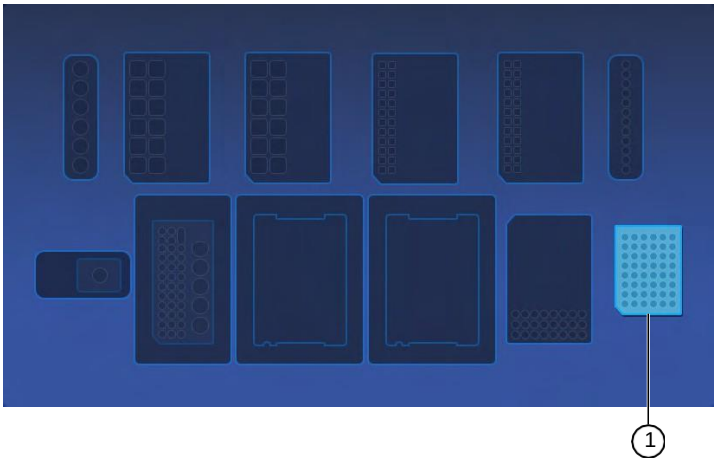
1. Unwrap, then load a new Genexus™ Dx 12-Well Tip Comb.

Note: Ensure the Genexus™ Dx 12-Well Tip Comb is straight and that the tabs are not bent or broken. If needed, gently bend the tip comb in the opposite direction to the curvature to straighten the tip comb before installing it.




1 Genexus™ Dx 12-Well Tip Comb position

2. Unwrap, then load a new Genexus™ Dx 48-Well Nucleic Acid Archive Plate.

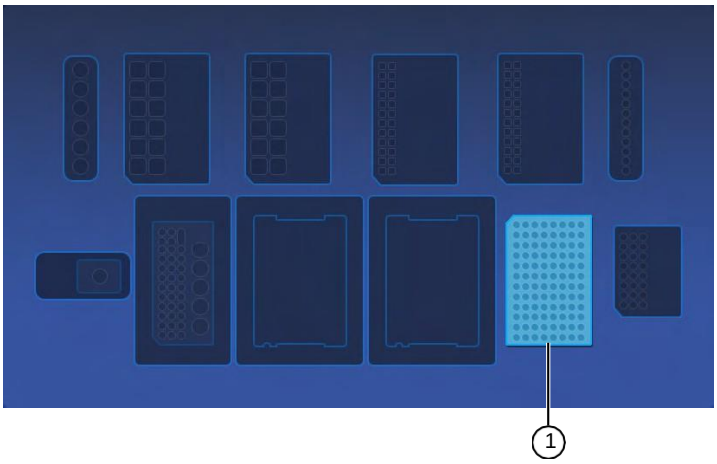



1 Genexus™ Dx 48-Well Nucleic Acid Archive Plate position

Note: Ensure that the archive plate is seated flat in the deck position. If the plate is curved, gently bend the plate in the opposite direction to the curvature to straighten it before loading.

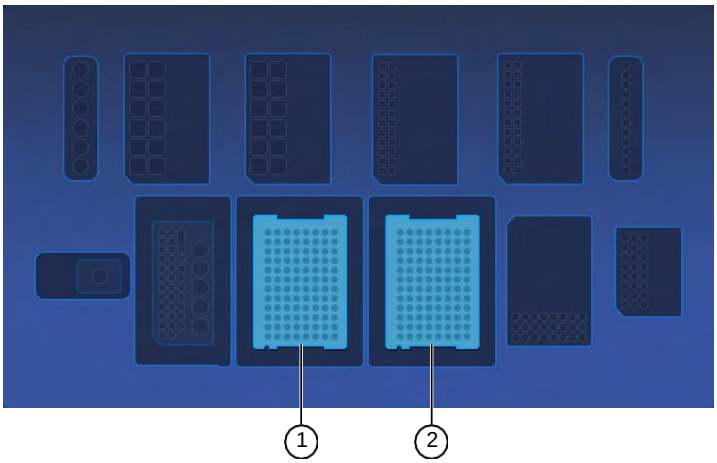
3. *(Integrated run configuration only)* Load a new 96-Well Nucleic Acid Output Plate (MicroAmp™ EnduraPlate™ Optical 96-Well Blue Reaction Plates with Barcode) into the output plate position.

Note: After a Sample to Result purification run, the 96-Well Nucleic Acid Output Plate becomes the Sample plate to be loaded in the Genexus™ Dx Integrated Sequencer.




1. 96-Well Nucleic Acid Output Plate position

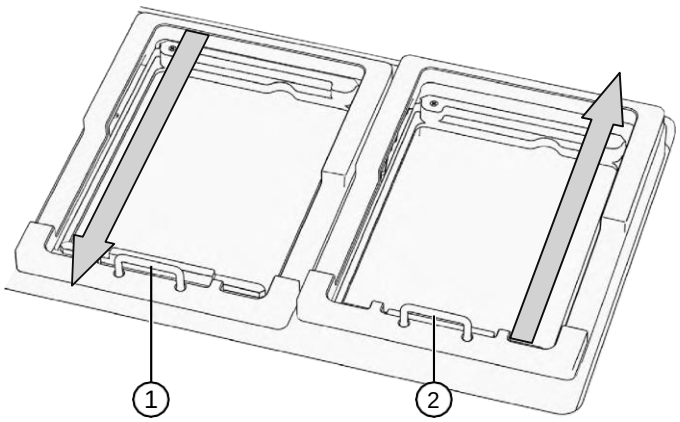
4. Unwrap two new Genexus™ Dx Purification Tip Cartridges and remove the cover to expose the pipette tips, then load them in positions 1 and 2.




1. Genexus™ Dx Purification Tip Cartridges position 1
2. Genexus™ Dx Purification Tip Cartridges position 2

a. Pull the locking mechanism handle forward (callout 1), then place the tip box in the open position.

b. Push the locking mechanism handle back (callout 2) to lock the tip box in place.

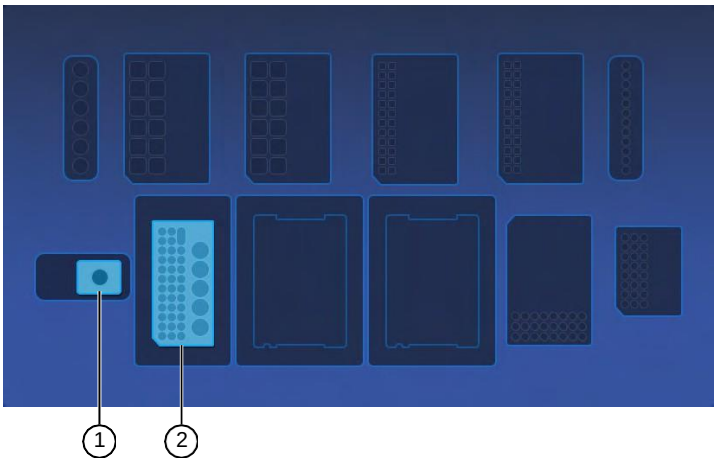



1. Unlocked (forward)
2. Locked (back)

## Load the quantitation reagents and consumables

Note: Protect the Genexus™ Dx Quantitation Plate from direct bright light to prevent photobleaching of the preloaded reagents.

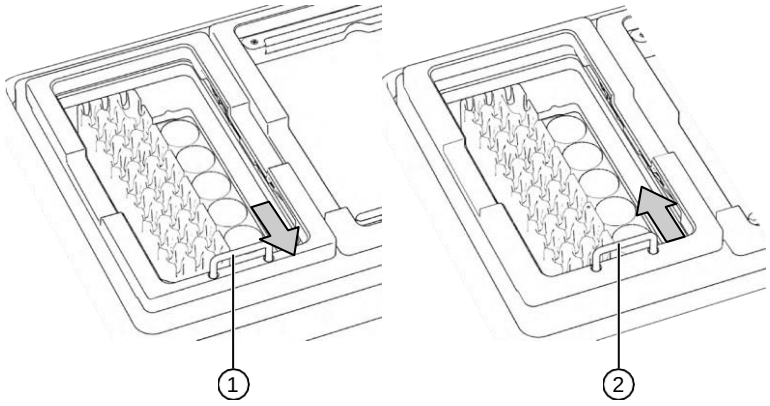
1. Centrifuge the Genexus™ Dx Quantitation Plate at 1,000 × *g* for 30 seconds to collect the contents.
2. Load the Quantitation Plate in position 2.




① Quantitation Tube position
② Quantitation Plate position

   a. Pull the locking mechanism handle forward, then place the Quantitation Plate in the open position.

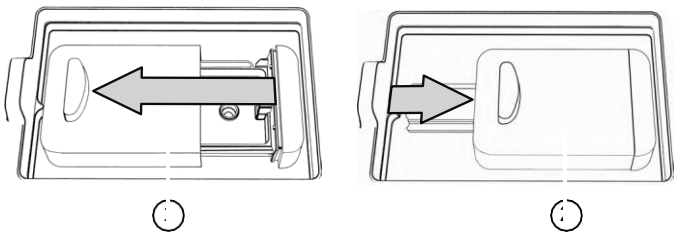
   b. Push the locking mechanism handle back to lock the plate in place.




① Unlocked (forward)
② Locked (back)

3. Slide and hold the quantitation module cover to the left, then insert the Quantitation Tube. **Press down firmly** to properly seat the tube, then allow the module cover to close.

WARNING! Do not force the module cover closed. Forcing the module cover closed can damage the instrument.



1 Open
2 Closed

## Confirm that consumables are installed correctly

IMPORTANT! To support correct and safe instrument operation, confirm that all consumables are installed correctly on the deck before you start a run. The instrument vision system confirms that required reagents are in place, no reagents are expired, and foil seals are removed. The vision system does not verify all aspects of the consumable setup before beginning each run.

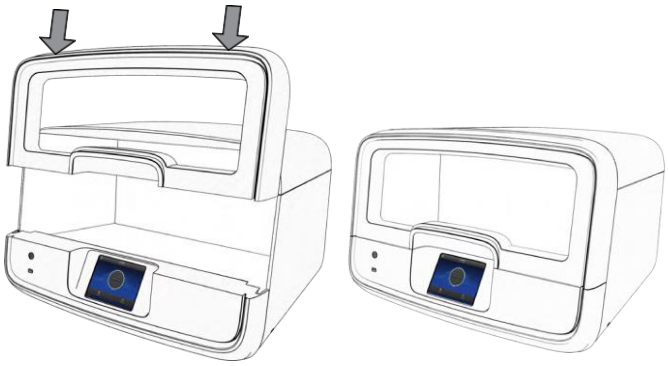
Confirm the following.

- Foil seals are removed from the purification plates. Do not remove foil seal from the Genexus™ Dx Quantitation Plate.
- Each component is at the correct location and in the correct orientation. Press down on all plates and cartridges to ensure that they are firmly seated in place.
- The Genexus™ Dx 12-Well Tip Comb is in place.
- The Genexus™ Dx Quantitation Plate is in the correct location, is in the correct orientation, and is locked in place.
- The Genexus™ Dx Quantitation Tube is firmly seated in the quantitation module.
- Each Genexus™ Dx Purification Tip Cartridge is in the correct location, in the correct orientation, and locked in place.

If the vision system detects an error, the location indicator does not turn gray in the touchscreen.

## Start the run

1. When all reagents and consumables are loaded in the purification instrument, tap **Next**.
2. Close the instrument door by pressing down on both top corners. Ensure that the door is locked after closing it.



The instrument vision system confirms that all reagents are in place and are not expired.

3. Tap **Start**.

   The time remaining until the purification is complete is displayed, and the interior lighting turns green.

   - If you need to stop the run for any reason, tap **Cancel**, then tap **Yes** to confirm the cancellation. A canceled run cannot be resumed. You must restart a run from the beginning.
   - The interior lighting turns off during quantitation, then turns blue when the run is complete.
   - If the instrument encounters a problem during the run, it aborts the run and displays the error on the instrument touchscreen. The interior lighting turns red.

When the run is complete, the interior light turns blue, and the touchscreen displays **Run Complete**. Quantitation results are available immediately. See “View and export quantitation results” on page 90.

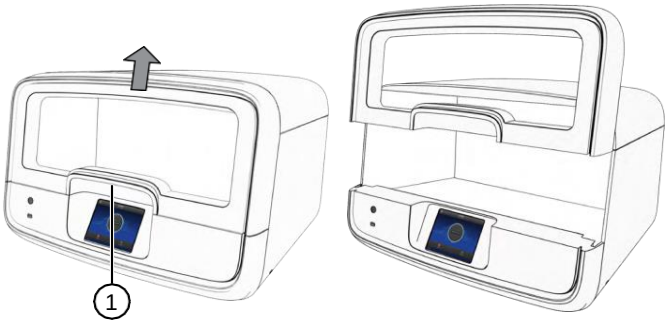
# Unload and UV clean the instrument

Note: Do not allow purified nucleic acids to sit on the instrument. Store as directed or use in sequencing reactions within 60 minutes.

1. In the touchscreen, tap **Unload**. The door unlocks.

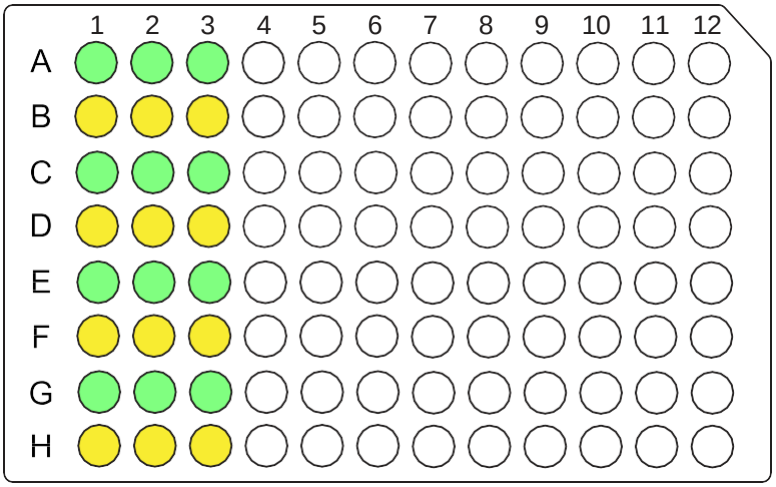


2. Lift the instrument door to access the instrument deck.




① Hold here, then lift.

3. *(Integrated run configuration only)* Remove the 96-Well Nucleic Acid Output Plate, containing the purified sample DNA and RNA, ready for the addition of controls. If not immediately loading controls, seal the plate, then store on ice or at 4°C. To determine the sample concentrations, see “View and export quantitation results” on page 90.




In the 96-Well Nucleic Acid Output Plate, DNA and RNA for each sample are loaded pair-wise starting at wells A1 and B1

- DNA for 12 samples
- RNA for 12 samples

4. *(Integrated run configuration only)* For short term storage if you are not using the output plate immediately in a sequencing run, seal the plate with an Adhesive PCR Plate Foil (Cat. No. AB0626). Store the plate at –20°C to –10°C for up to 1 week.

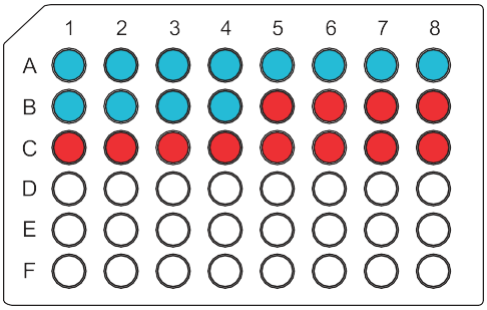
   For long term storage (>1 week), do not seal the plate. Instead, transfer the samples to labeled low-retention tubes (Eppendorf™ DNA LoBind™ Tubes; Cat. No. 13-698-791), then store the DNA samples at –30°C to –10°C and the RNA samples at –90°C to –65°C for up to 6 months.

5. Remove the Genexus™ Dx 48-Well Nucleic Acid Archive Plate containing the remainder of sample DNA and RNA.

---

Note: In the standalone configuration, the Genexus™ Dx 48-Well Nucleic Acid Archive Plate contains all the sample DNA and RNA.

---




DNA is in wells A1–B4 (12 samples)
RNA is in wells B5–C8 (12 samples)

6. Seal the archive plate with a Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal. Store the archive plate at –20°C to –10°C for up to 6 months.

Note: If the archive plate is thawed during storage, transfer the DNA and RNA into labeled low-retention tubes (Eppendorf™ DNA LoBind™ Tubes; Cat. No. 13-698-791), then store the DNA samples at –30°C to –10°C and the RNA samples at –90°C to –65°C for up to 6 months after purification. Do not reseal the archive plate with the used plate seal.

7. Remove and discard the deep-well sample input plates.
   a. Remove the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 from the instrument.
   b. Dispose of the liquid waste by tipping the deep-well plate on one corner and pouring the waste into an appropriate liquid biohazardous waste container.

WARNING! Liquid waste contains guanidine thiocyanate, dispose of properly.

   c. Dispose of the deep-well plate in an appropriate waste container.
   d. Repeat substep 7a through substep 7c to discard the Genexus™ Dx FFPE DNA and RNA Purification Plate 2.

8. Unlock, then remove and dispose of each Genexus™ Dx Purification Tip Cartridge in an appropriate waste container. Used tip cartridges contain a small amount of quantitation kit reagents.

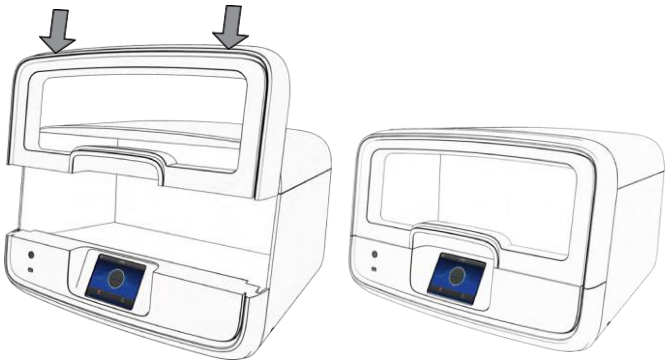
9. Unlock, then remove and dispose of the Genexus™ Dx Quantitation Plate.
   a. Dispose of the liquid waste by tipping the deep-well plate on one corner and pouring the waste into an appropriate liquid biohazardous waste container.

WARNING! No data are currently available that address the mutagenicity or toxicity of the Qubit™ RNA BR Reagent (Component A). This reagent is known to bind nucleic acid and is provided as a solution in DMSO. Handle the Qubit™ RNA BR Reagent with the same safety precautions as all other potential mutagens and dispose of the dye in accordance with local regulations.

   b. Dispose of the deep-well plate in an appropriate waste container.

10. Open the Qubit quantitation module cover, remove and discard the Genexus™ Dx Quantitation Tube, then allow the module cover to close.

11. Close and lock the instrument door by pressing down on both top corners, then tap **Start UV Clean**.



The time remaining in the UV Clean is displayed. When complete, the instrument is ready to start a new purification run.

# View and export quantitation results

Purification runs that include sample quantitation produce sample concentration results that can be accessed after the run is complete. Results can be accessed from the **Run Complete** screen or the home screen.

1. In the **Run Complete** screen, tap **View report**.

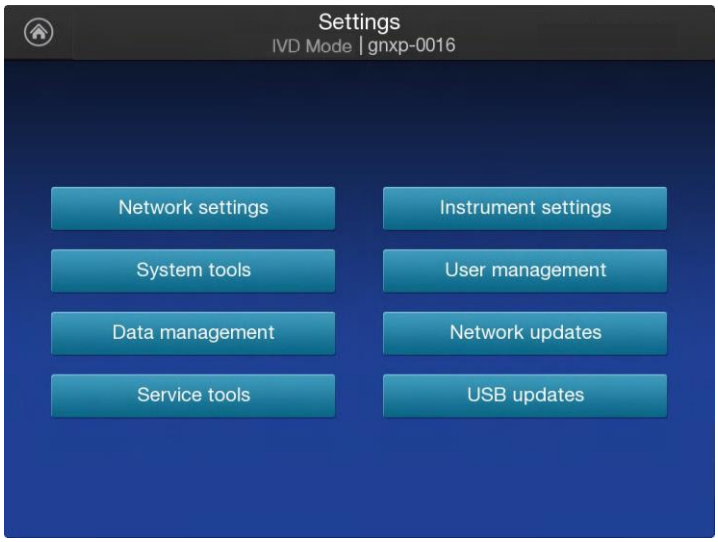



The **Saved Experiment Reports** screen opens. See step 4.

2. At any time after unloading and UV cleaning the instrument, sample concentration results can be accessed through the home screen. Tap ⚙ (**Settings**).



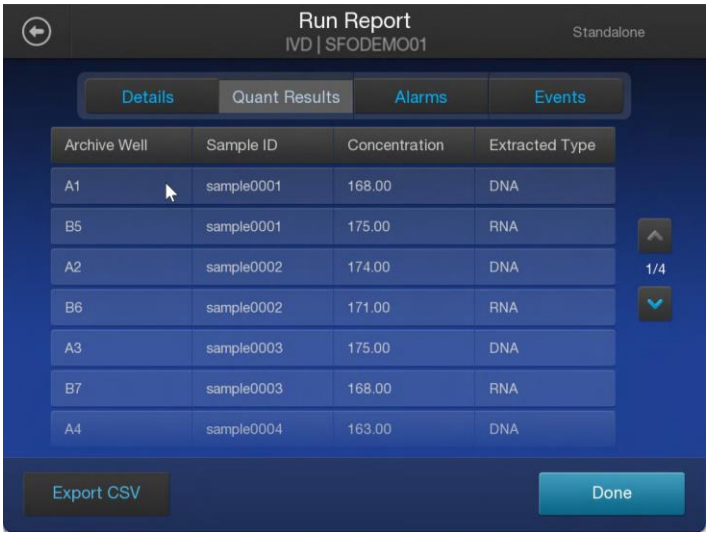

3. In the **Settings** screen, tap **Data Management**.



4. In the **Saved Experiment Reports** screen, tap ⌄ or ⌃ to page through the list. Locate the **Experiment Name** of interest, tap in the row to select the experiment, then tap **View Report**.




5. In the **Run Report** screen, tap **Quant Results** to view the sample concentration results.




Note: The concentration units are ng/µL.

6. Insert a USB drive into the USB port on the front of the instrument, then tap **Export Report**. Navigate to the file destination, then tap **Save**.

In Sample to Result runs, sample concentrations can also be viewed in Genexus™ Dx Software in the **Run Summary** tab of the run results.

**Purification Samples**

| Sample Name | Sample Type | Nucleic Acid Type | Conc.(ng/µl) | QC Conc. Range (ng/µl) | Batch Status | Archive Position | Library Prep |
|---|---|---|---|---|---|---|---|
| 01_Sample_rep1 | FFPE | DNA | 9.74 | 0.5 - 512 | Completed | A1 | ✓ |
| 01_Sample_rep1 | FFPE | RNA | 40.26 | 0.5 - 512 | Completed | B5 | ✓ |
| 02_Sample_rep2 | FFPE | DNA | 9.86 | 0.5 - 512 | Completed | A2 | ✓ |
| 02_Sample_rep2 | FFPE | RNA | 39.94 | 0.5 - 512 | Completed | B6 | ✓ |
| 03_Sample_rep3 | FFPE | DNA | 9.91 | 0.5 - 512 | Completed | A3 | ✓ |
| 03_Sample_rep3 | FFPE | RNA | 38.19 | 0.5 - 512 | Completed | B7 | ✓ |

# 6 Add Oncomine™ Dx Express Test controls to the sample plate


- Add controls to the sample plate—Sample to Result run.....94
- Dilute the samples, if needed—Nucleic Acid to Result run..... 96
- Load the sample plate—Nucleic Acid to Result run..... 98


In a Sample to Result run, purified samples are loaded in an output sample plate by the Genexus™ Dx Purification System in a format that meets the sample format requirements of the Genexus™ Dx Integrated Sequencer. You next need to add Oncomine™ Dx Express Test positive and no-template controls to the sample plate manually according to the run setup guide. For a Nucleic Acid to Result run, you need to add purified samples and controls to the sample plate. For a Library to Result run, you need to add previously prepared libraries to a sample plate.

## Add controls to the sample plate—Sample to Result run

Sample plates created by the Genexus™ Dx Purification System for Sample to Result runs have the correct volume of samples loaded in the positions specified in the run setup guide. Before loading the plate in the sequencer, you must manually add positive controls and no-template control (NTC) to the sample plate.

Thaw control tubes ahead of time on ice or at 4°C for up to 2 hours. After thawing, vortex to mix, then centrifuge to collect tube contents.

---

IMPORTANT! Ensure that contents of the control tubes are completely thawed before adding to the sample plate.

---

1. Add controls to the sample plate with the volume and in the well positions that are specified in the run setup guide.

   ---

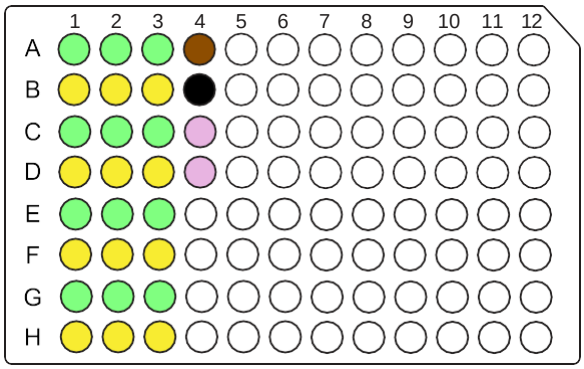
   Note:

   - Do not adjust control volume.
   - Add controls to sample plate wells in the following order:
     a. Positive controls
     b. NTC (add to 2 wells)

- Before adding the NTC, replace your gloves with a new pair of gloves.
- Use a PCR hood (if available) when adding controls to the plate to reduce the risk of contamination.

Controls volume

| Control | Cap color | Volume |
|---|---|---|
| FFPE samples | | |
| Oncomine™ Dx Express Test FFPE DNA Control | Brown | 20 µL |
| Oncomine™ Dx Express Test FFPE RNA Control | White | 20 µL |
| Oncomine™ Dx Express Test NTC Control (for DNA and RNA) | Purple | 20 µL |




Example sample plate for FFPE DNA and RNA samples (12 samples) showing default positions of samples and controls. DNA and RNA for a sample are loaded pair-wise. If you change sample and control positions from the default order during run planning or have a different number of samples, the run setup guide shows the loading positions needed for controls.

DNA sample

RNA sample

Oncomine™ Dx Express Test FFPE DNA Control

Oncomine™ Dx Express Test FFPE RNA Control

Oncomine™ Dx Express Test NTC Control

Note: The plate diagram above is only an example. Follow the sample positions assigned in the run plan summary when setting up a sample plate for a sequencing run.

2. Seal the plate with an Adhesive PCR Plate Foil (Thermo Fisher Scientific Cat. No. AB0626). If available, press the seal tightly onto the plate using an adhesive film applicator.

Note: The use of other plate seals can affect performance.

3. After sealing, centrifuge the plate at 300 × *g* for 15 seconds to collect contents at the bottom of plate wells.

   Note: Ensure that the centrifuge has been wiped down and cleaned before centrifuging the plate.

4. Keep the plate on ice or at 4°C until you are ready to load it in the sequencer.

# Dilute the samples, if needed—Nucleic Acid to Result run

Nucleic acid samples with concentrations up to 1,024X of the target concentration for an assay (displayed as default values in the **Sample Plate** step screen in run planning) are in range for automated dilution and require no manual dilution. Alternatively, DNA and RNA from FFPE samples can be manually diluted to 0.5 ng/µL. Enter the concentrations during sample run planning at the **Sample Plate** step (see step 5).

Note: After a sample is diluted on the system, the diluted sample cannot be used in future runs. If sample quantity is limited, manual dilution of the sample is recommended to allow for potential repeat runs, if needed.

For samples with concentrations greater than the upper range for automated dilution, when setting up a Nucleic Acid to Result run, or when processing previously purified nucleic acid, you can manually dilute the sample with nuclease-free water to the target concentration of 0.5 ng/µL DNA and RNA for FFPE samples. For samples that are in range, proceed to “Load the sample plate—Nucleic Acid to Result run”. For samples that need manual dilution, proceed to “Procedural guidelines for manual dilution of samples” on page 97.

| Sample type | If the sample concentration is | Action |
|---|---|---|
| FFPE DNA and RNA | ≥0.5 ng/µL, but ≤512 ng/µL | No manual dilution is necessary. The sequencer dilutes the sample to the target concentration during the run. If you are setting up a Nucleic Acid to Result run, or preparing sample for multiple assays, samples can be manually diluted to the target concentration (0.5 ng/µL). |
| | >512 ng/µL | Manually dilute to the target concentration (0.5 ng/µL), or to a concentration in range for automated dilution by the sequencer. |

Note:

- If you enter a concentration <0.05 ng/µL or >10,000 ng/µL, a warning that the concentration is out of range appears, and you are not allowed to proceed to the next step.
- If the concentration is >0.05 ng/µL but less than the target concentration of the assay, you can proceed. However, because the sequencer cannot concentrate samples, the sample concentration will be less than the target concentration.
- If the concentration is ≤10,000 ng/µL, but >1,024X of the target concentration, you can proceed. However, because the sequencer cannot dilute samples more than 1,024-fold, the diluted sample concentration will be greater than the target concentration.

## Procedural guidelines for manual dilution of samples

- Do not pipet volumes less than 2.0 µL. If sample concentration is above 5 ng/µL for FFPE samples, first dilute each sample to an intermediate dilution (for example, 5 ng/µL), using the relationship C1 × V1 = C2 × V2 (C1: initial concentration, V1: Initial volume, C2: target concentration, V2: target volume).
- Unless the test protocol dictates that multiple libraries are prepared from a single sample (for example, replicate libraries), dilute only sufficient sample to prepare a single library (20 µL total).
- If samples were previously frozen, thaw on ice or at 4°C, then vortex and pulse centrifuge to ensure samples are fully resuspended and homogeneous.
- For manual sample quantification, if not using the on-board quantification by the Genexus™ Dx Purification System in standalone configuration, we recommend using the Qubit™ Fluorometer or equivalent fluorometer with a nucleic acid-specific fluorometric kit. Sample concentration determined by UV absorbance is not recommended because UV absorbance is not specific to DNA and RNA.

## Dilute the samples manually, if needed

For samples that need manual dilution, dilute DNA and RNA from FFPE samples to a final concentration of 0.5 ng/µL.

IMPORTANT! Do not perform the following dilution procedure until you are ready to proceed to loading the sample plate. Avoid freeze-thaw cycles of samples that have been diluted to target concentrations.

1. Label a new 1.5-mL nuclease-free, low-retention microcentrifuge tube for each sample being diluted. Place tubes on ice or in a prechilled benchtop cold block until needed.

   Note: When diluting DNA and RNA samples from the same patient, we recommend that you label tubes using different color markers (for example, blue for DNA and red for RNA) to avoid mixing nucleic acid types during manual plate loading.

2. Use the sample concentrations to calculate the volume (X) of each sample needed for 10 ng of FFPE DNA and RNA. Use the following formula: (ng sample input) / (sample concentration, ng/µL) = X µL of sample.

3. Calculate the volume (Y) of nuclease-free water needed to bring FFPE sample volume to 20 µL, using the following formula:

   (10 ng / 0.5 ng/µL) – X µL of sample = Y µL of nuclease-free water

   Note: Dead volume is included in the 20 µL sample volume. However, 10% excess volume can be added if needed by multiplying the X and Y values by 1.1.

4. For each sample, pipet the calculated Y µL of nuclease-free water into the appropriate labeled 1.5-mL microcentrifuge tube.

5. Add the calculated X µL of sample into the appropriate tube, pipet up and down 3 times and vortex to mix, then pulse centrifuge.

6. Keep samples on ice, in a chilled cold block, or in a 2–8°C refrigerator until ready for use.

Proceed to “Load the sample plate—Nucleic Acid to Result run”.

# Load the sample plate—Nucleic Acid to Result run

Thaw control tubes ahead of time on ice or at 4°C for up to 2 hours. After thawing, vortex to mix, then centrifuge to collect tube contents.

---

IMPORTANT! Ensure that contents of the control tubes are completely thawed before adding to the sample plate.

---

1. Add samples, positive controls, and no-template control (NTC) to the sample plate with the volume and in the well positions that are specified in the run setup guide.

   ---

   Note:

   - Do not adjust sample and control volume.
   - Load sample plate wells in the following order:
     a. Samples
     b. Positive controls
     c. NTC (add to 2 wells)
   - Before adding the NTC, replace your gloves with a new pair of gloves.

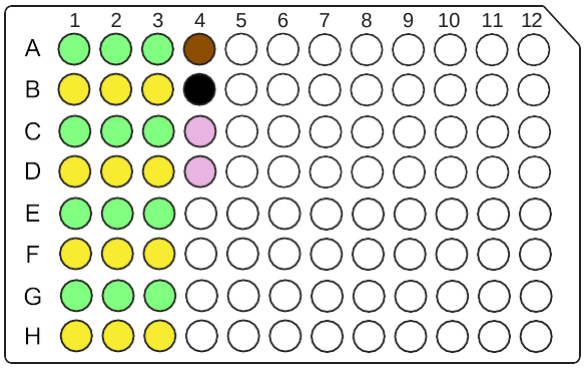
   ---

   Sample volume

| Sample type | Volume |
|---|---|
| DNA | 20 µL |
| RNA | 20 µL |

   Controls volume

| Control | Cap color | Volume |
|---|---|---|
| Oncomine™ Dx Express Test FFPE DNA Control | Brown | 20 µL |
| Oncomine™ Dx Express Test FFPE RNA Control | White | 20 µL |
| Oncomine™ Dx Express Test NTC Control (for DNA and RNA) | Purple | 20 µL |




Example sample plates for FFPE DNA and RNA samples (12 samples) showing locations of controls and samples. Load controls directly from control tubes without diluting. For clinical samples, you can either allow the sequencer to perform on-board dilution if the sample concentration is within range, or manually dilute to 0.5 ng/µL (FFPE samples). Add DNA and RNA for samples pair-wise starting at positions A1 and B1 for sample 1, positions C1 and D1 for sample 2, and so on.

- Oncomine™ Dx Express Test FFPE DNA Control
- Oncomine™ Dx Express Test FFPE RNA Control
- DNA sample
- RNA sample
- Oncomine™ Dx Express Test NTC Control

2. Seal the plate with an Adhesive PCR Plate Foil (Cat. No. AB0626). If available, press the seal tightly onto the plate using an adhesive film applicator.

---

Note:

- The use of other plate seals can affect performance.
- If you dilute samples on the sample plate and vortex the plate after sealing, apply the foil seal carefully between wells with an applicator before vortexing to help ensure that the seal is complete and contamination between wells does not occur.

---

3. After sealing, centrifuge the plate at 300 × *g* for 15 seconds to collect contents at the bottom of plate wells.

---

Note: Ensure that the centrifuge has been wiped down and cleaned before centrifuging the plate.

---

4. Keep the sample plate on ice or at 4°C until you are ready to load it in the sequencer.

# 7 Start a sequencing run


- Materials required ..... 100
- Before you begin ..... 101
- Load the sequencer and start a run ..... 103
- Clear the instrument deck and perform a UV Clean ..... 111
- Options for an expired sequencer initialization ..... 114


## Materials required

Genexus™ Dx Integrated Sequencer consumables

- Genexus™ Dx Strip 1
- Genexus™ Dx Strip 2-HD
- Genexus™ Dx Strip 3-GX5™
- Genexus™ Dx Strip 4
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Sequencing Kit
  - Genexus™ Dx Cartridge
  - Genexus™ Dx Bottle 2
  - Genexus™ Dx Bottles 1 and 3
- Genexus™ Dx Pipette Tips
- MicroAmp™ EnduraPlate™ Optical 96-Well Clear Reaction Plates with Barcode (2)

Other items

- Oncomine™ Dx Express Test Panel
- Genexus™ Strip Centrifuge Adapter
- Sample plate from a Genexus™ Dx Purification System run

# Before you begin

Before setting up a sequencing run, review general procedural guidelines for handling panels, reagents, and samples to minimize the chance of contamination and help ensure the success of the run. See “Guidelines for panel and reagent use and handling” on page 29 and “Guidelines for preventing contamination” on page 30.

1. Remove Oncomine™ Dx Express Test Panel tubes that are needed for the run from the freezer, then thaw on ice or at 4°C for at least 30 minutes. After thawing, centrifuge at 300 × *g* for 15 seconds using the Genexus™ Strip Centrifuge Adapter to collect contents at the bottom of the tubes. Keep the panel tubes on ice or at 4°C until you load them in the sequencer.

2. If you are installing a new Genexus™ Dx Cartridge, thaw the cartridge at room temperature for 30 minutes before loading in the sequencer.

3. Remove the library and templating strips from the refrigerator or freezer.
   a. Equilibrate Genexus™ Dx Strip 1, Genexus™ Dx Strip 2-HD, and Genexus™ Dx Strip 3-GX5™ at room temperature for 30 minutes before loading in the sequencer.

   b. Thaw Genexus™ Dx Strip 4 by laying the strip on ice or incubating at 4°C for at least 30 minutes, or until the tube contents are fully dissolved and ready for loading in the sequencer.

   IMPORTANT!
   - Ensure that strip contents are completely thawed before proceeding.
   - If loading is delayed, keep thawed Genexus™ Dx Strip 2-HD and Genexus™ Dx Strip 4 on ice or at 4°C until you are ready to load the sequencer.

4. Vortex each strip at maximum speed for 5–10 seconds while rocking the strips from side to side.

   Note: Vortexing helps dislodge magnetic beads that can be trapped in keyholes of Genexus™ Dx Strip 1 and Genexus™ Dx Strip 3-GX5™, and to dissolve precipitate in Genexus™ Dx Strip 2-HD, if present.

   IMPORTANT! Visually check the bottom of tube 3 of Genexus™ Dx Strip 2-HD for white precipitate by doing a pulse-vortex to resuspend and expose precipitate. If precipitate is present, continue to pulse-vortex strips every 10 minutes until precipitate fully dissolves, keeping the strips at room temperature. If precipitate fails to dissolve after 3 rounds of pulse-vortexing, replace the strip with a new Genexus™ Dx Strip 2-HD and repeat the thawing procedure.

Note: Load the Genexus™ Dx Integrated Sequencer within 2 hours of thawing reagents.

Proceed to “Centrifuge the library and templating reagent strips”.

## Centrifuge the library and templating reagent strips

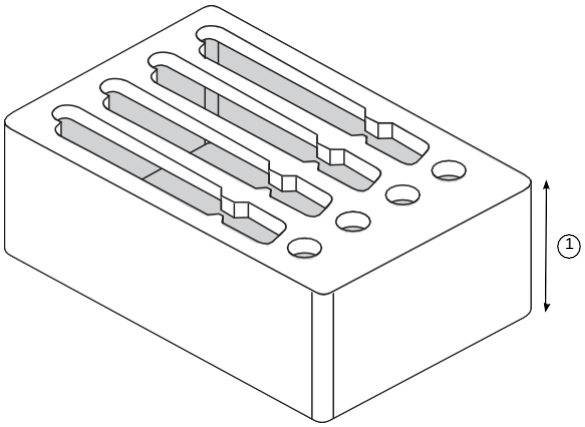
Use the Genexus™ Strip Centrifuge Adapter as a holder for centrifuging library and templating strips to collect contents in the bottom of strip tubes after vortexing. The adapter is provided at sequencer installation.

Use of the strip adapter requires a centrifuge with buckets that support the height of the holder at 44 mm so that buckets swing freely in the centrifuge rotor when loaded with strips. For more information, see “Required materials and equipment not supplied” on page 15. To use the Genexus™ Strip Centrifuge Adapter, follow these steps.

---

Note: The Genexus™ Strip Centrifuge Adapter can also be used for centrifuging primer pool tubes to collect tube contents.

---




Genexus™ Strip Centrifuge Adapter

① 44 mm height

1. After thawing and vortexing, load the strips in the adapters in a balanced orientation. The centrifuge buckets must support the height of the strip adapters loaded with strips.
2. Place each adapter loaded with strips in the centrifuge.
3. Centrifuge the strips at 300 × *g* for 15 seconds.
4. Remove strips from the adapters, then inspect the strips to ensure that contents have been collected and air bubbles are not present. Line up similar strips against each other and ensure that the reagent volume in each well is consistent between the two strips. Repeat this process until all strips are inspected. If volume inconsistencies are observed, do not use the strip.

   ---

   Note: Alternatively, verify well reagent volumes by comparing fill levels with the Genexus™ Dx strip figures provided on page 222.

   ---

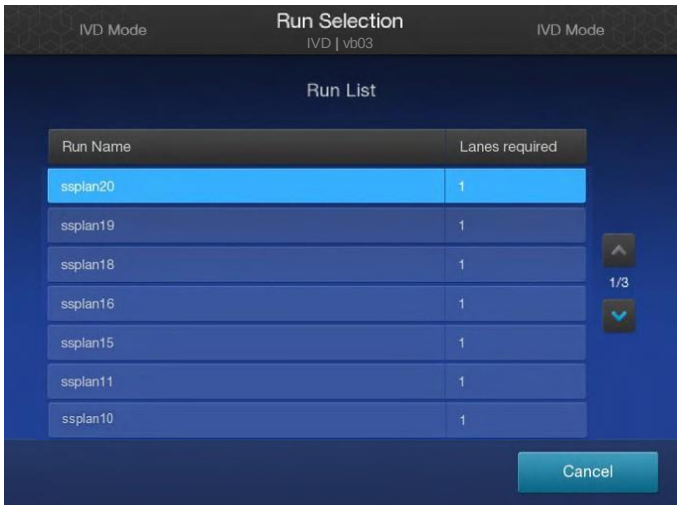
5. Repeat step 1 through step 4 for the remaining library and templating strips to be loaded for a sequencing run, if needed.
6. After centrifugation, return Genexus™ Dx Strip 4 to ice or 4°C until you are ready to load in the sequencer.
7. To verify correct panel volume of primer pool tubes, hold two sets next to each other at an angle above eye-level. If volume inconsistency is suspected, compare against the Genexus™ Dx primer pool tube figures provided on page 222.

# Load the sequencer and start a run

1. Tap **Run** on the sequencer home screen to start the setup and loading procedure.




2. In the **Run Selection** screen, select the run that you want to use from the list.

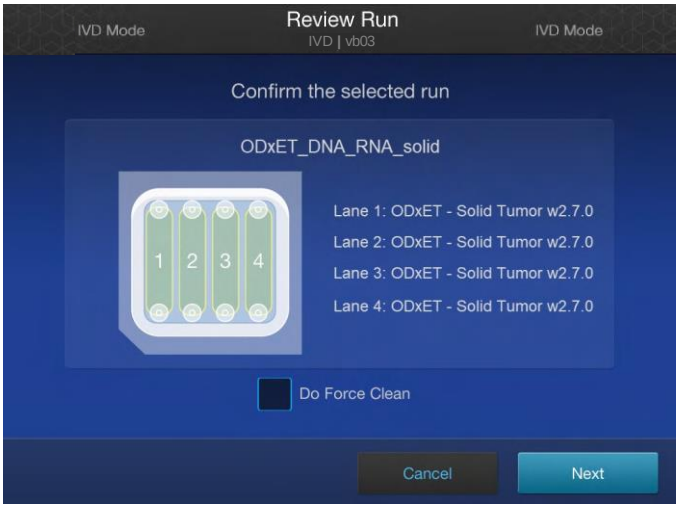



---

Note: If you select a run that requires more lanes than are available on a currently installed chip, a dialog appears giving you the option to install a new chip, or cancel. If you proceed with a new chip, a post-chip clean needs to be performed to remove the installed chip, then the sequencer prompts you to perform the Clear Deck, UV Clean, Load Deck, Clear Sequencing Reagents, and Load Sequencing Reagents steps.

---

3. In the **Review Run** screen, confirm the run and assay selections, then tap **Next**.




The deck door opens automatically.

---

Note:

- If the instrument vision system detects consumables loaded on the deck, the sequencer prompts you to remove the consumables, then starts a UV Clean.
- If all 4 lanes of the installed chip were previously used, the used chip shuttles to the install position. You are prompted to remove it before the UV Clean starts.
- Select the **Do Force Clean** checkbox if there will be 1–3 unused lanes on the installed chip after the run, but you want to start the next run on a new chip after the current run. A force clean automatically cleans the instrument after the run, eliminating the need for an operator to manually perform the **Clean instrument** procedure between the completion of the current run and the next run. Selecting **Do Force Clean** makes all lanes of the chip and installed sequencing reagents unusable after the run.
- If all 4 lanes will be used, the sequencer automatically performs a clean at the end of the run, and you do not need to select the **Do Force Clean** option.

---

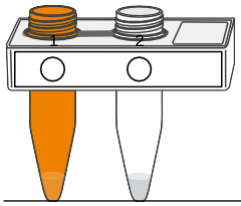
4. In the **Load Deck** screen, the sequencer instructs you step by step to load each required consumable and sample plate in a highlighted position on the deck. The sequencer detects the loading of each consumable in real time and advances to the next component automatically.




IMPORTANT!

- Remove the primer pool tube caps before installing the tube carrier on the deck.
- Load the correct type of barcode plate, library strip, or template strip for the type of run you are setting up. The sequencer displays a warning if you have installed consumables that are incompatible with the run you have selected. For example, a warning appears if an RUO-labelled barcode plate or library strip is loaded in an IVD-mode run.
- Remove the lids from all the boxes of Genexus™ Dx Pipette Tips before you load the boxes in the sequencer.
- After removing a tip box lid, visually inspect the tip box to ensure that all tips are seated level in the tip rack before and after placing on the instrument deck. If a bent or damaged tip is suspected, replace the tip box with a new tip box. Reracking tips from partially used boxes is not supported and can cause some tips to seat at an angle, which can affect run performance.
- Ensure that all tip boxes sit level on the deck by visually inspecting all boxes across the tip station. Reseat the boxes before starting the run, if needed.
- Do not use third-party pipette tips or repackage third-party tips into Genexus™ Dx pipette tip boxes. Use of third-party tips can result in run failure.
- Load instrument plates, including PCR plate, barcode plate, enrichment plate, and sample plate, into position by pressing down firmly and evenly on all sides and corners of each plate.

Note: A primer pool tube carrier can only be installed with the position 1 tube in the back row of the Genexus™ Dx Primer Pool Tube station. Follow the guidance in the run setup guide for loading the primer pool tube carrier or carriers in the correct position and order in the station.



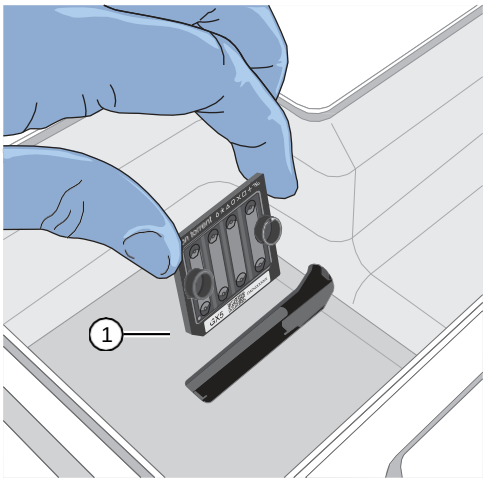

① Position 1
② Position 2

5. Ensure that all the primer pool tube carrier barcodes are visible and are accurately shown as loaded in the instrument software.

IMPORTANT! The instrument cannot detect if the Genexus™ Primer Pool Tube Holder is improperly placed. Ensure that the tube holder is correctly positioned before proceeding.

6. If prompted, insert a new Genexus™ Dx GX5™ Chip and Genexus™ Coupler. Insert the chip into the chip install slot with the chip notch oriented down and toward the front of the instrument.

IMPORTANT! Insert only a new chip and coupler when prompted. Do not insert a partially used chip.






① Notched corner of chip

Note: A chip shuttle under the deck moves the installed chip to loading and sequencing positions during the run.

IMPORTANT! Insert the Genexus™ Coupler so that it is level to help ensure it will properly align with the Genexus™ Dx GX5™ Chip. A coupler that is installed at an angle or is not level will not align properly to the chip and can result in a failed run.

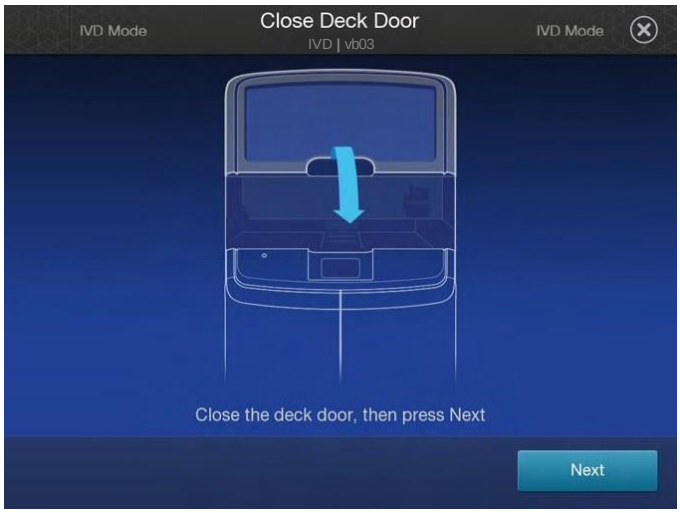
7. When the deck consumables and sample plate have been loaded, lock the library and templating strips in place by sliding the latches toward the rear of the deck.

Note: Load the sample plate last.




If a chip is detected and the strip latches are closed, the **Close Deck Door** screen appears.

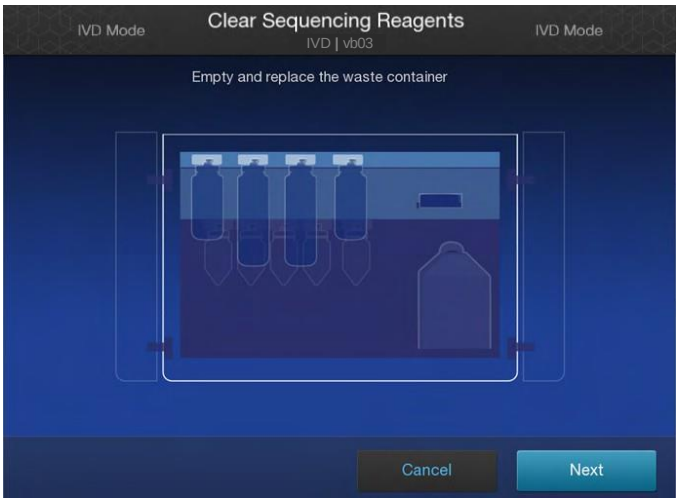
8. Close the deck door, then tap **Next**.




- If you installed a new chip in the sequencer, the sequencer prompts you to open the sequencing reagents bay doors to empty the waste and remove used sequencing reagents bay consumables. Proceed to step 9.
- If you are using a chip that was previously installed and has sufficient lane capacity for the run, the sequencer prompts you to start the run.

IMPORTANT! The cartridge and bottles in the sequencing reagents bay must be replaced every time that a new chip is installed, regardless of how many lanes were used in the previous chip.

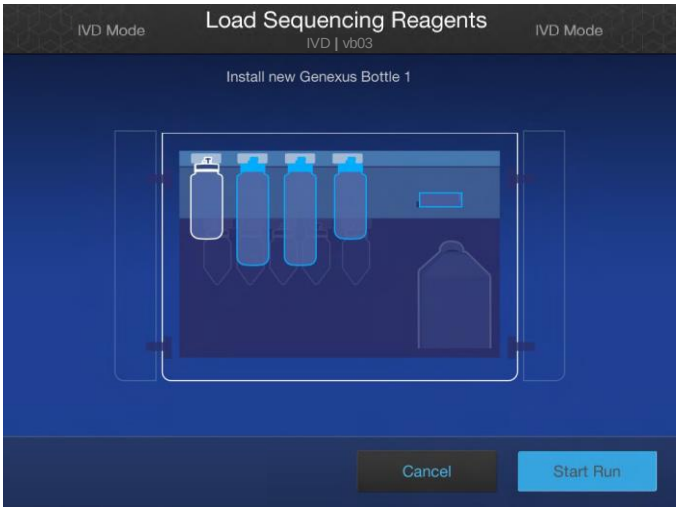
9. Follow on-screen instructions to empty the waste in the Waste carboy, remove waste pipette tips, remove the used Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2, Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge, then tap **Next**.




IMPORTANT!

- Empty and replace the Waste carboy and the waste pipette tip bin.
- After replacing the emptied Waste carboy, ensure that you reinsert the waste tube into the carboy.
- Follow all applicable local, state/provincial, and/or national regulations when recycling or disposing of consumables and liquid waste.

10. Install a new Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2 (two required), Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge.

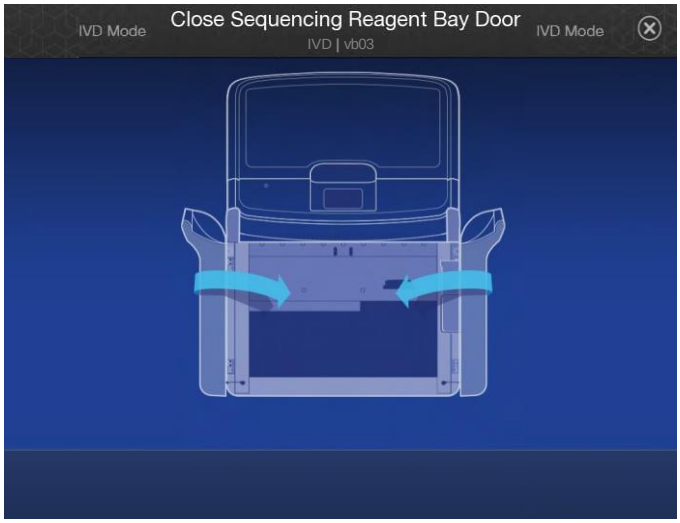


IMPORTANT!

- Before installing, *gently* invert each capped Genexus™ Dx Bottle 2 five times to mix—avoid vigorous mixing. Inspect the plastic nozzles for any pinches or deformations. To avoid pinching or folding of the nozzle, install the bottles straight-on, not at an angle.
- The installed reagents can be used for up to 14 days on the sequencer. The installed reagents on the sequencer should be used within 14 days of installation to avoid run failure, although the sequencer permits use for up to 28 days

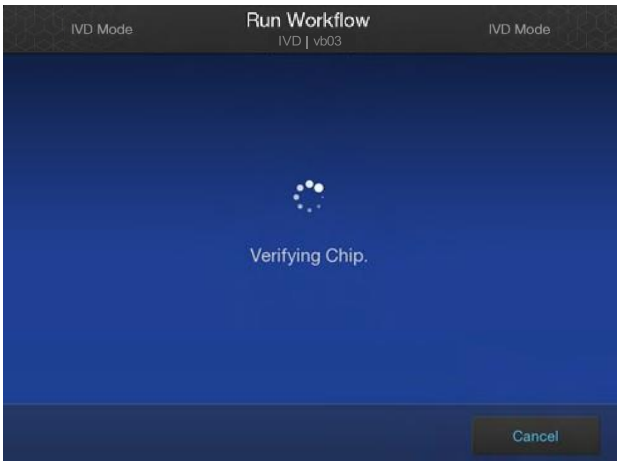
After reagents have been installed, the **Close Sequencing Reagent Bay Door** screen appears.

11. Close the sequencing reagents bay doors.




After the doors are closed, the sequencer automatically starts the run.

At the beginning of the run, the instrument verifies the chip, checks for leaks, then calculates run time.










A sequencing run encompasses the following stages:

1. Starting
2. Initializing
3. Library Prep
4. Templating
5. Presequencing
6. Sequencing
7. Cleaning

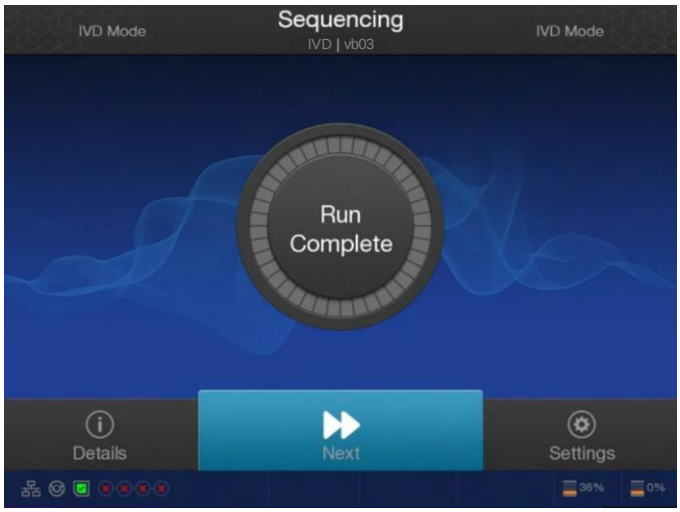
At each stage, the instrument shows the time remaining on the touchscreen.

---

Note:

- The time remaining shown on the screen does not include run analysis time.
- The leak check takes approximatively 22 minutes. Ensure that the leak check has completed and the run has started before leaving the laboratory.

---




When the run finishes, the sequencer displays the **Run Complete** screen.

Note: If all the lanes of a chip are used in a run, the used chip remains engaged in the sequencer until you start a new run. The chip then shuttles to the install position, and you are prompted to remove it before loading the deck with new consumables.

# Clear the instrument deck and perform a UV Clean

After a run completes, remove used consumables from the deck and perform a **UV Clean** to ready the instrument for the next run.

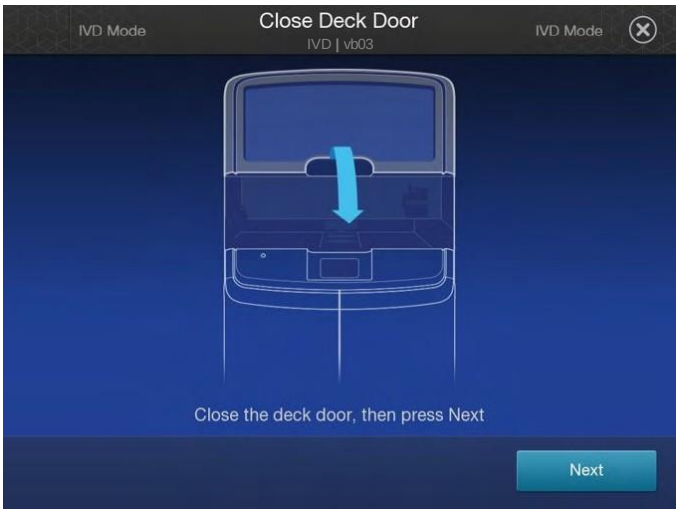
1. In the **Run Complete** screen, tap **Next** to start removal of used consumables.
   The deck door opens.

2. In the **Clear Deck** screen, the sequencer provides step-by-step instructions by highlighting the components to be removed. Unlock the library and templating strips by sliding the latches toward the front of the deck, then remove the used strips. Remove the remaining deck components specified by the sequencer.

   Note: Seal the PCR amplification plate with a foil seal before removing the plate from the PCR amplification station. Sealing the plate before removal helps prevent contamination of libraries if libraries are recovered for reuse. Store the sealed library plate at 4–8°C in a post-amplification area.



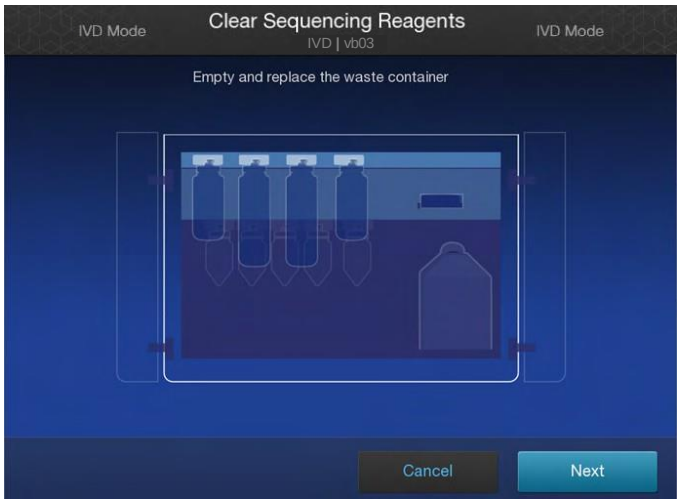

3. Inspect the Genexus™ Dx Filter in the liquid waste disposal port and verify that no standing liquid or debris is present.
   - If no standing liquid is found but debris is present, replace it with a new filter. For more information, see “Replace the Genexus™ Dx Filter” on page 211.
   - If standing liquid is present, contact Technical Support and report a possible filter or deck liquid waste line clog.
4. When finished, close the deck door, then tap **Next**.




A two-minute **UV Clean** starts.




5. After UV cleaning, if all the chip lanes were used, the sequencing reagents bay doors unlock. Open the doors, remove used components from the bay and empty the Waste carboy and waste pipette tip bin, then tap **Next**.



IMPORTANT! Do **not** discard or remove the conical bottles, unless alerted by the sequencer to replace the bottles after a conical bottle flow rate test. A conical bottle change may occur at the end of the run before the cleaning is performed and the instrument is ready for the next run. This process can add approximately 2 hours to when the sequencer can be loaded for the next run. For more information, see “Replace the Genexus™ Dx Conical Bottles” on page 211.

IMPORTANT! Follow all applicable local, state/provincial, and/or national regulations when recycling or disposing of Genexus™ Integrated Sequencer consumables and liquid waste.

CAUTION! The Genexus™ Dx Bottle 1 (small waste bottle) contains small amounts of formamide. Dispose of this waste appropriately.

6. After removal of used components, close the sequencing reagents bay doors, then tap **Next**.
   The sequencer returns to the home screen.

# Options for an expired sequencer initialization

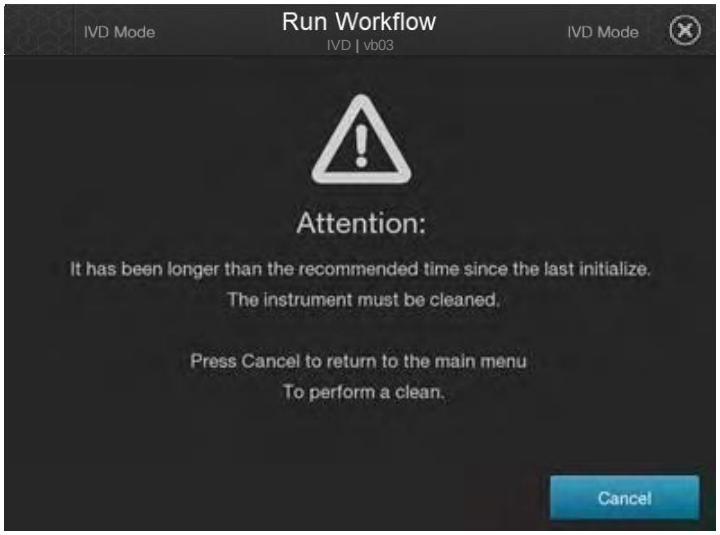
A sequencer initialization is defined by the installation of a new chip, chip coupler, sequencing bottles, and reagent cartridge on the sequencer before a run. Reagents are stable on the sequencer for 14 days, after which you can experience reduced performance. After 14 days, the following on-instrument warning appears:




We recommend that you not bypass the warning by tapping **Ignore**, but instead tap **Cancel** to proceed with an instrument clean. After tapping **Cancel**, select **Settings►Clean instrument** to clean the instrument before starting a run with a new chip. Lane assignment for a new run starts with lane 1.

IMPORTANT! Reagents remain stable on the sequencer for up to 14 days. After this period, performance can decrease, potentially leading to run failure. We recommend that you not bypass the warning by tapping **Ignore**, but instead tap **Cancel** to proceed with an instrument clean.

After 28 days, you can no longer proceed with an expired initialization. The following message appears:



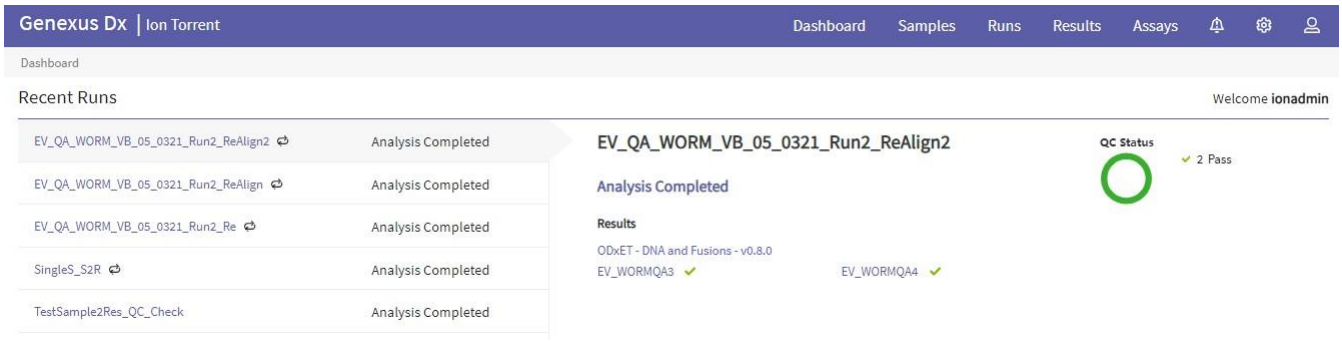

Tap **Cancel** to return to the main menu to perform an instrument clean.

# 8 Monitor the run

## View run progress on the instrument

You can view details for the five most recent and current runs in the **Dashboard** in Genexus™ Dx Software.

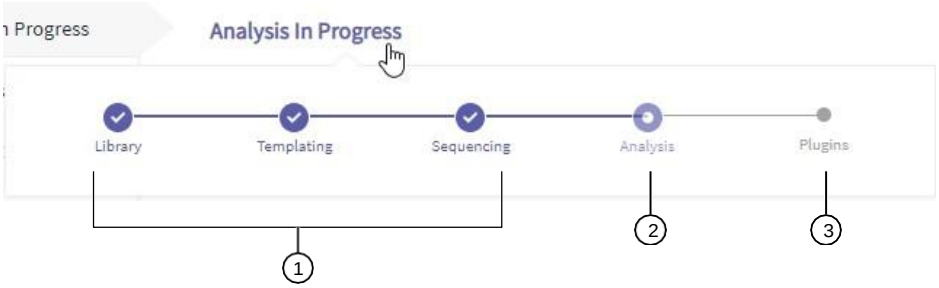
Runs that are reanalyzed are listed with ↻ **(Reanalysis)** after the run name. For information on reanalyzing runs, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or the software help system.




1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

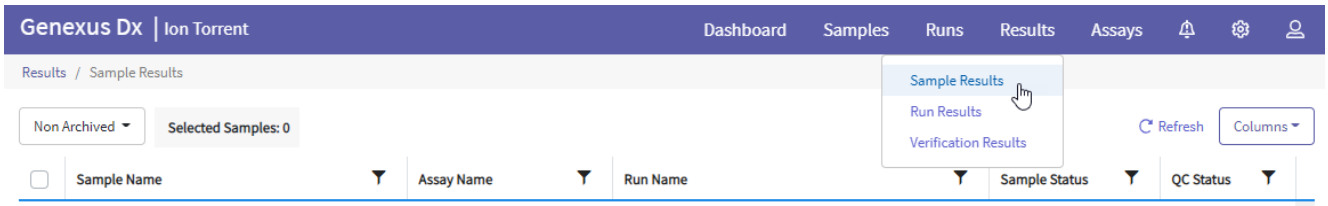
| View option | Description |
|---|---|
| View detailed run result information for completed runs. | In the left side of the screen, click the run name of interest.<br>The Run Summary tab opens. For more information, see “The run summary” on page 147.<br>In the menu bar, click Dashboard to return to the Dashboard. |
| View the quality control information. | In the left side of the screen, click the row of the run of interest.<br>The QC status for the run and for the samples is shown in the details pane in the right side of the screen. For more information, see “QC results” on page 140. |
| View assay-level metrics and results for the run. | 1. In the left side of the screen, click the row of the run of interest.<br>2. In the run details pane in the right side of the screen, click the assay name of interest.<br>The Assay Metrics tab opens. For more information, see “Assay metrics and the run report” on page 150.<br>In the menu bar, click Dashboard to return to the Dashboard. |

*(continued)*

| View option | Description |
|---|---|
| View the sample results. | 1. In the left side of the screen, click the row of the run of interest.<br>2. In the run details pane in the right side of the screen, click the sample name of interest.<br><br>The Key Findings tab opens. For more information, see "Key Findings" on page 122.<br>In the menu bar, click Dashboard to return to the Dashboard. |
| View the progress of the run. | 1. In the left side of the screen, click the row of the run of interest.<br>2. In the run details pane in the right side of the screen, place the pointer over the status for the run.<br><br>A progress bar for the run is shown. Steps in the run that are complete are listed with a check mark. Steps that are in progress are shown with a flashing circle. Steps that remain to be completed are listed in grey. If any failures occur, failed steps are also listed in grey.<br>  <br><br>① Completed steps are shown with a check mark. In this example, Library preparation, Templating, and Sequencing are complete.<br>② Steps that are in progress are shown with a flashing circle. In this example, Analysis is in progress.<br>③ Steps that remain to be completed are shown in grey. In this example, the Plugins step remains to be started after Analysis is complete. |

# 9 Review data and results


- Results navigation bar overview....118
- Review sample results....120
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- Clinical Test Report and Clinical Laboratory Report....154
- Reanalyze a run or sample....163


Use the **Results** menu to review results and data analysis, and to perform data management tasks. You can view results sorted by sample or by run.

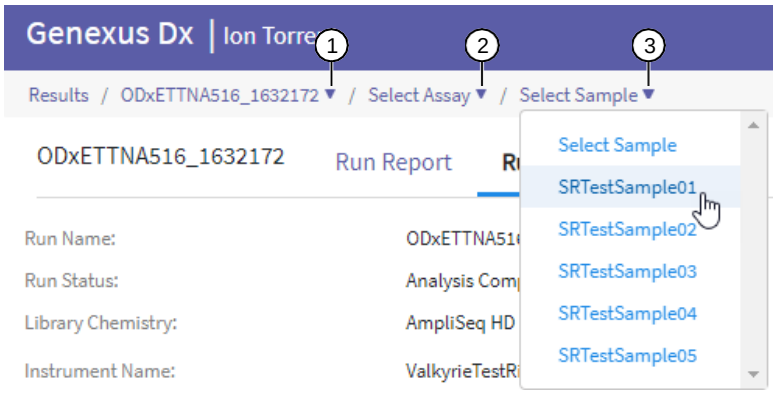
| Selection | Description |
|---|---|
| Click Results▶Sample Results | Select this option to review completed sample results and reports. |
| Click Results▶Run Results | Select this option to review completed run results and reports by assay. |
| Click Results▶Verification Results | Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification. |

## Results navigation bar overview

After you select a run result or a sample result, a results navigation bar appears and allows you to easily toggle between different results screens for each run. You can quickly switch between run-level results, assay-level results, and sample-level results for the run. In addition, you can easily find samples or assays in the run with the search field that appears in each dropdown list.

Assay-level results include assay metrics, such as final read data.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.

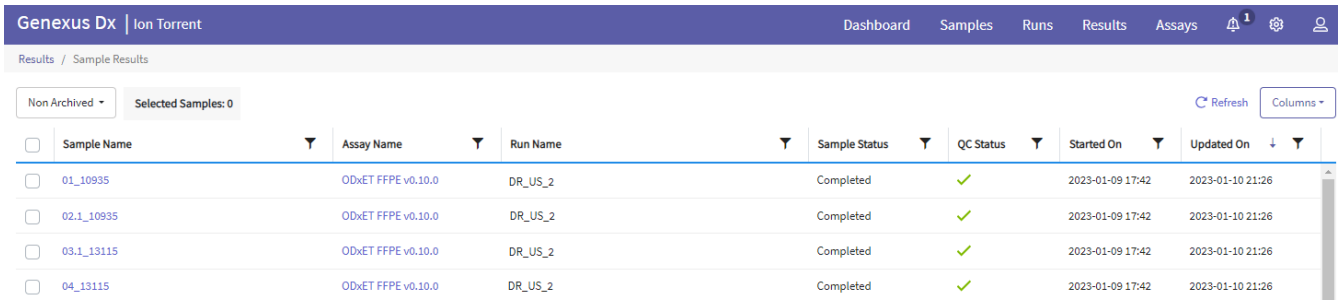



| Callout | Dropdown menu | Description |
|---|---|---|
| 1 | Run name | The run name is listed. Multiple runs are listed only if the run has been reanalyzed. |
| 2 | Select Assay | You can click the assay name of interest to view assay metrics for the run.<br>You can search for or toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed.<br>You can select Select Assay from the dropdown menu to remove the assay selection. If you remove the assay and sample selections, the Run Summary tab opens with a summary of results for the run selected in the Run Results dropdown menu. Assay metrics are also shown in the Run Report tab. For more information, see “View assay metrics and the run report” on page 150. |
| 3 | Select Sample | You can click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in Select Assay. The search field allows you to find samples in the run.<br>You can select Select Sample from the dropdown menu to remove the sample selection. If you remove the sample selection, the Assay metrics tab for the selected assay opens. If you select Select Assay with Select Sample shown, the Run Summary tab opens. Assay metrics are also shown in the Run Report tab. For more information, see “View assay metrics and the run report” on page 150. |

# Review sample results

In the **Results / Sample Results** screen, samples that have been sequenced are listed by sample name.

You can filter the list of results by clicking **(Filter)** in a column of interest, then entering a full or partial sample, assay, or run name.




The following information appears in the **Sample Results** screen. You can add columns to the Sample Results table to view more information. For example, you can add a column for **Electronic Signatures** to view signature and lock information for variant reports. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Results screen for the sample. |
| Assay Name | The name of the assay used in the run plan. |
| Run Name | The unique name of the run given when it was created in the software. |
| Sample Status | The status of the run or sample (for example: Completed, Running, Failed, Terminated, Pending, Stalled). |
| QC Status | The QC status of a completed run.<br>• ✔ (Passed) indicates the sample passed all QC metrics.<br>• ✘ (Failed) indicates the sample failed a QC metric.<br>• — (Not Calculated) indicates a sample did not undergo QC analysis. |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |
| Tags | Tags that are assigned to the sample are listed. |

You can also perform the following actions in the **Sample Results** screen.

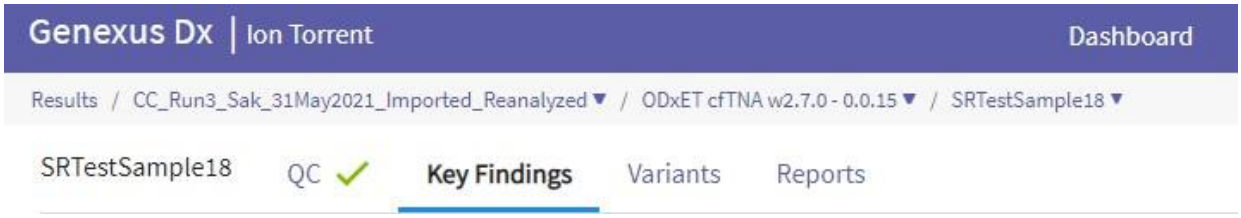
| Option | Description |
|---|---|
| Actions | Available action links for a sample are shown when you place the pointer over the row of a sample.<br>• Report—Download the report (available only for samples with a sample status of completed).<br>• Assign Tags—Add or remove the tags assigned to a sample result.<br>• Audit—View the audit trail for the run.<br>• Notes—View, add, or delete notes for sample result.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>• Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. |
| Actions that you can perform when you select samples | Assign Tags—Assign a tag or tags to selected sample results in one action. The Assign Tags command appears above the Sample Results table after you select one or more samples.<br>Note: For more information on using the Assign Tags feature, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910), or the software help system. |

## View key findings

You can view the **Key Findings** screen for a sample starting from either sample results or run results.

In the menu bar:

- Click **Results►Sample Results**, then click a sample name.
- Click **Results►Run Results** to open the **Results / Run Results** screen, then then click a run name in the **Run Name** column, click a run name to open the **Run Summary** screen, You can then select an assay from the **Select Assay** dropdown list and a sample name from the **Select Sample** dropdown list.




You can toggle between different assays used in a run with the **Assays** dropdown list at the top of the screen.

## Key Findings

The **Key Findings** table shows details about the sample, a list of biomarkers reported for the sample, and a summary of key metrics for genes assayed in the run.

Note: The **Key Findings** table is by default the first view that is shown for sample results.

The information that is displayed depends on the assay that was used in the run.

Sample Details

| Sample Detail | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Collection Date | The date that the sample was collected. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Sample Type | A term that describes the sample, for example, FFPE, DNA, DNA & RNA |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. "Percent Cellularity" refers to the same measurement as "Percent Tumor Content." |

Biomarker Results

| Result category[1] | Description |
|---|---|
| Level 1: Companion Diagnostics (CDx) Biomarkers | Gene variants that have companion diagnostic claims in the corresponding cancer type that are associated with an FDA-approved therapy as listed in Table 1 of the intended use. |
| Level 2: Cancer Mutations with Evidence of Clinical Significance | Gene variants with evidence of clinical significance that are not prescriptive or conclusive for labeled use of a specific therapeutic product, and for which clinical validation has not been performed, but which have curated clinical evidence associated with FDA-approved therapies and/or National Comprehensive Cancer Network (NCCN) clinical guidelines (Category 1 and 2A) in the sample cancer type. |
| Level 3: Cancer Mutations with Potential Clinical Significance | Gene variants not considered Level 1 or Level 2 biomarkers that can be described as cancer mutations with potential clinical significance. These variants are not prescriptive or conclusive for labeled use of a specific therapeutic product, and clinical validation has not been performed. They have curated clinical evidence associated with NCCN clinical guidelines Category 2A in a different cancer type or Category 2B in any cancer type. They may also have curated evidence as an enrollment criterion in clinical trials or biomarkers with evidence in peer-reviewed literature. |

[1] For more information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 158.

Key Metrics

| Metric | Description |
|---|---|
| Amplicon Summary | |
| Average Base Coverage Depth | The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage. |
| Uniformity Of Base Coverage | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths. |
| Percent Reads On Target | The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified. |
| Uniformity Of Amplicon Coverage | The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts. |

## View variant results

You can view detailed variant results in the **Variants** table. In the default view, all variants are listed. You can download the information for the variants in XLS or TSV format.

You can also limit the types of variants that are shown and toggle between different variant results: SNVs/Indels, Fusions, and CNVs.

Variant Tables

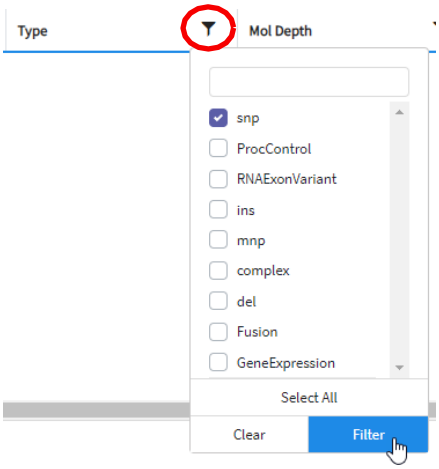
| Variant type | Description |
|---|---|
| SNVs/Indels | • SNVs: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included.<br>• Indels: insertion and deletion variants.<br>For more information, see “View SNV/Indel results” on page 124. |
| Fusions | Translocations of genetic material and splicing variants.<br>For more information, see “View fusion results” on page 126. |
| CNVs | Copy number variations (CNVs) are variations of the number of copies of a given gene.<br>For more information, see “View CNV results” on page 131. |

## Search and filter the list of variants

You can search and filter to immediately narrow the list of results that are shown in the variant tables for the selected sample. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name in the **Sample Name** column.
3. In the **Results** screen, click the **Variants** tab.
4. Select the variant class to display the results: **SNVs/Indels**, **Fusions**, or **CNVs**.
5. In the table of variants, in the column heading of interest, click **(Filter)**.
   - In the search field, enter at least 3 characters, then click **Filter**.
   - Select the checkbox in the row of each filter that you want to apply, then click **Filter**.

   The options that are available depend on the column and variant class. For example, you can filter data in the **Type** column to show one specific variant type.
6. Click **Clear Filters** to remove all filters and view the full list of run results.




The column or columns to which you applied a filter change to reflect the filter and selected options.

## View SNV/Indel results

The **SNVs/Indels** table lists the calls and other information for the SNV and INDEL variants that are analyzed in each sample in a run.

To view the **SNVs/Indels** table for a sample, click **Results►Sample Results** in the menu bar, then in the **Sample name** column, click the name of the sample of interest. In the **Variants** tab, click **SNVs/Indels** to display the data. To export the data in tabular format, click **Export** in the upper right corner of the screen.

## SNVs/Indels table

The data displayed in the **SNVs/Indels** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 124.

| Column | Description |
|---|---|
| Variant ID | The name of the hotspot as defined in the Browser Extensible Data (BED) file.<br>Click the link to view more annotation information. |
| Variant Name | The name of the variant. |
| Locus | The chromosome and position of the detected variant. |
| Gene | The gene name. |
| AA Change | Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature. |
| Ref | The reference base or bases at that locus. |
| Alt | The alternate base or bases at that locus. |
| Type | The type of variant that is detected.<br>• snp (single nucleotide polymorphism)<br>• mnp (multi-nucleotide polymorphism)<br>• ins (insertion)<br>• del (deletion)<br>• complex |
| Test Result | Indicates the presence or absence of an SNV/Indel variant.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—Indicates the absence of a variant that differs from the reference allele at a given position. |
| Call Details | The reason why a variant is reported as NO CALL. |
| Phred QUAL Score | The relative probability of either the "reference" hypothesis interval [0,cutoff], or the "variant" hypothesis interval [cutoff,1], Phred-scaled (-10*log10). A higher score means more evidence for the variant call. |
| Raw Read Depth | Total read coverage across amplicon containing SNV/Indel hotspot locations. Count of chip-level reads aligned at this locus that participate in variant calling. |
| Effective Read Depth | The number of reads covering the position. |

*(continued)*

| Column | Description |
|---|---|
| Alt Allele Read Counts | The number of reads containing the alternate allele. |
| Nuc Change | The position and identity of the nucleic acid change. |
| Allele Frequency | The number of variant read counts divided by the total number of read counts for the sample. |
| Allele Frequency (%) | The allele frequency, represented as a percentage. |
| Mut/WT | The ratio of mutant allele to wild-type allele. |
| PPA | Possible Polyploidy Alleles (PPA).<br>A value of Yes indicates variants that are PPA alleles. A value of No indicates variants that are not PPA alleles. |

## View fusion results

You can view calls and other information for the fusions that are analyzed within each sample in a run. To view the **Fusions** table for a sample, click **Results►Sample Results** in the menu bar, then in the **Sample name** column, click the name of the sample of interest. In the **Variants** tab, click **Fusions** to display the data. To export the data in tabular format, click **Export** in the upper right corner of the screen.

## Fusions table

The data displayed in the **Fusions** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 124.

| Column | Description |
|---|---|
| Variant ID | The name of the fusion target as defined in the BED file. |
| Variant Name | The name of the variant. |
| Locus | The chromosome positions in the reference genome that define the fusion junction. |
| Genes (Exon) | The name of fusion target and representative acceptor and donor exons. |
| Read Counts | The frequency of the fusion detected in the sample. |
| Type | Assay type (for example, Fusion, RNAExonVariant (exon skipping), RNAExonTiles, ProcControl). |

*(continued)*

| Column | Description |
|---|---|
| Test Result | Indicates the presence or absence of a fusion or RNA exon variant.<br>• POSITIVE—indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—although some evidence for the presence of a fusion exists, the call does not pass one or more filters that are required for a high confidence fusion call.<br>• NEGATIVE—indicates the absence of a fusion due to the variant call not passing read or call thresholds. |
| Call Details | The reason for reporting a fusion as NO CALL or NEGATIVE. |
| Read Counts Per Million | The number of fusion read counts detected per million total reads. |
| Gene Isoform | The name of the fusion target as defined in the BED file.<br>Note: For fusion assays, the value for Gene Isoform is identical to that of Variant ID, |
| Mol Cov. Mutant | The median molecular coverage across a fusion amplicon. |
| Imbalance Score[1] | Each fusion gene exhibits a characteristic Imbalance Score threshold. Scores that exceed the threshold value indicate a high likelihood of the presence of the fusion in the sample. Imbalance score calculation starts with the normalization of reads in the exon-tiling amplicons of the gene, followed by correction with a baseline that represents expression values in normal samples. Reads for observed imbalance scores come from samples. Baseline scores come from normal, fusion-negative samples and are stored in the exon tile assay baseline file for the assay.<br>Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline)<br>where<br>Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples)<br>• 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample.<br>• 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample.<br>Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline)<br>• 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene.<br>• 5' of breakpoint is the total normalized baseline values of the amplicons for the gene. |
| Imbalance P-Value[1] | The statistical significance of measure of imbalance relative to a control gene. |
| Predicted Break-point Range[1] | The exonic range for predicted fusion break point in exon tiling assays. |

*(continued)*

| Column | Description |
|---|---|
| Ratio To Wild Type[1] | The molecular ratio for exon skipping assays relative to wild type control amplicons. |
| Norm Count Within Gene[1] | Exon skipping assay coverage normalized to molecular coverage of wild type (WT) MET control amplicons. |

[1] Column appears only in analyses that use the exon tiling fusion detection method.

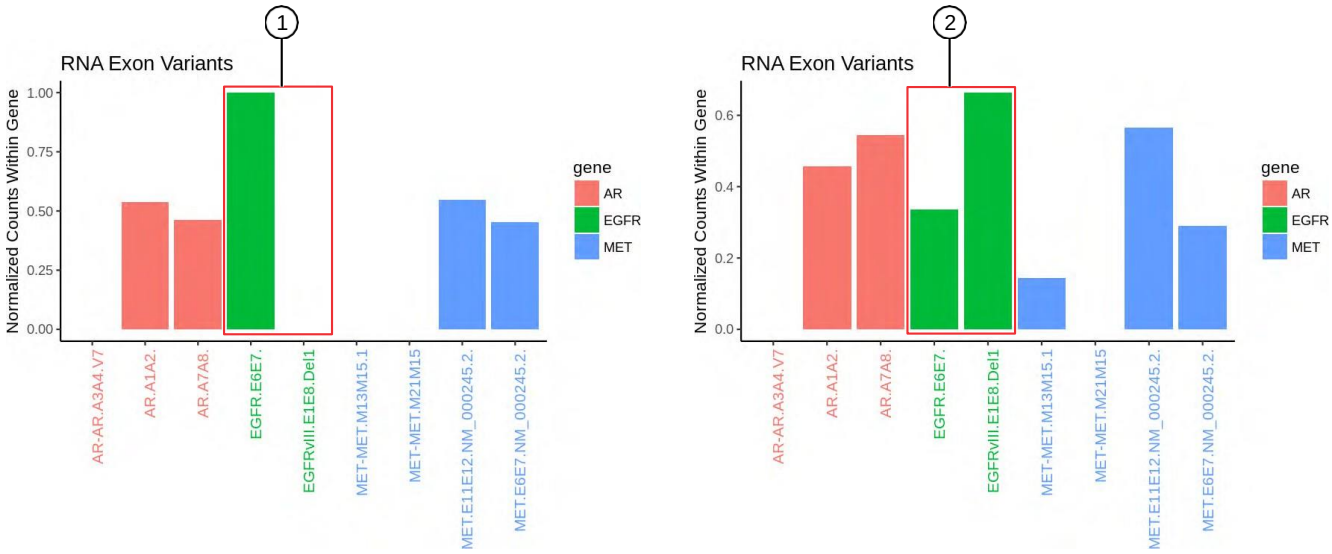
## View RNA Exon Variants

The **RNA Exon Variant** data view displays a bar graph summary of intragenic exon rearrangements or fusions. The displayed RNA exon variants are defined in the BED file that is associated with an assay. The **RNA Exon Variant** data view is available for all RNA and fusion assays.

Note: To review **RNA Exon Tile Fusion Imbalance** analysis plots, see the user guide for your assay.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 126.
4. In the top right corner of the screen, click **Visualization►RNA Exon Variant**, then review the **RNA Exon Variants** plot.

Representative RNA Exon Variant plots




The X-axis represents specific exon variants, where each variant is labeled with a gene ID followed by a sequence of adjacent exons. The Y-axis measures the read counts for each variant, normalized to the wild type.

(1) Example result where only the wild type EFGR (EFGR.E6E7) was detected.

(2) Example result where RNA exon 2–7 deletion occurred in the EFGR gene. The deletion of exons 2–7 resulted in an increase of normalized read counts for the EFGR variant that contains the intragenic fusion of exon 1 and exon 8 (EFGR.E1E8.Del1) and a decrease of normalized read counts for the wild type EFGR (EFGR.E6E7).

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View RNA Exon Tile Fusion Imbalance

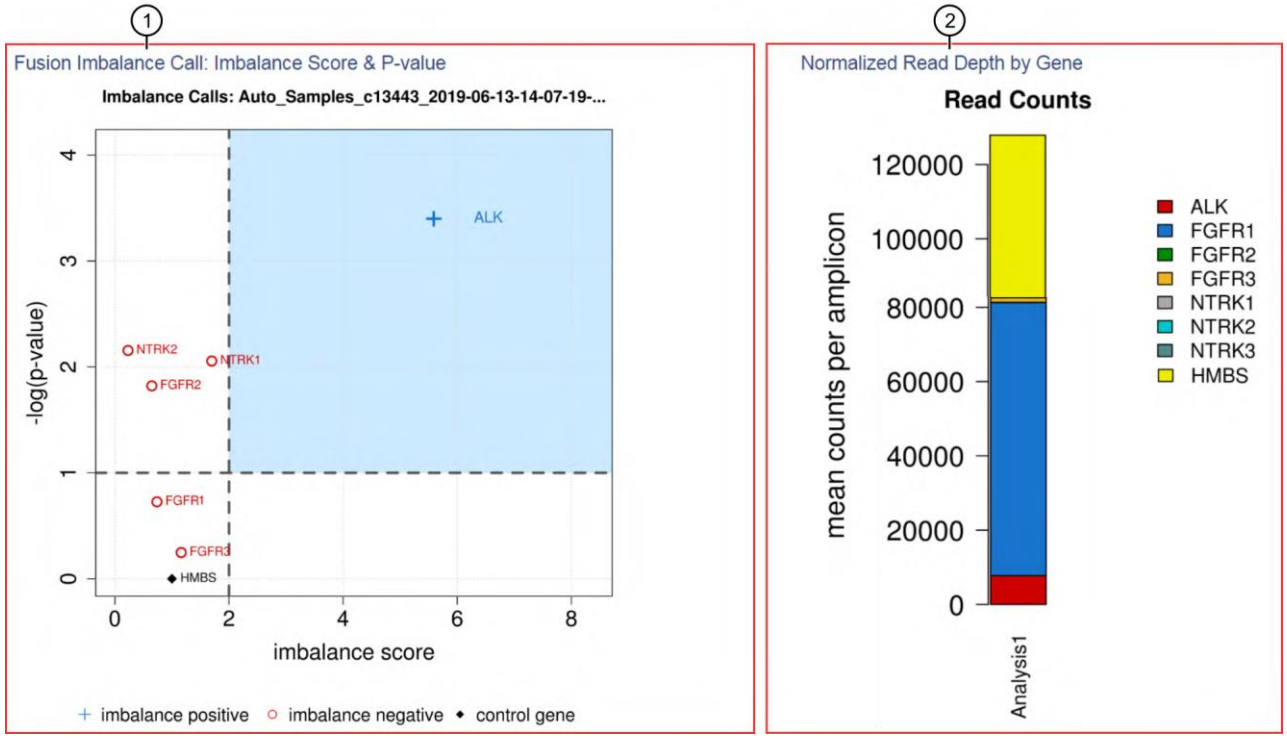
The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

Driver genes, for example ALK, FGFR2, and NTRK1, have amplicons that span exon-exon junctions to probe the expression difference between the 3' and the 5' regions of the gene.

An imbalance score is calculated for each driver gene in a sample to quantify the magnitude of the expression imbalance change between the two parts of the gene. For example, the 3':5' ratio in ALK; and the 5':3' ratio in FGFR2.

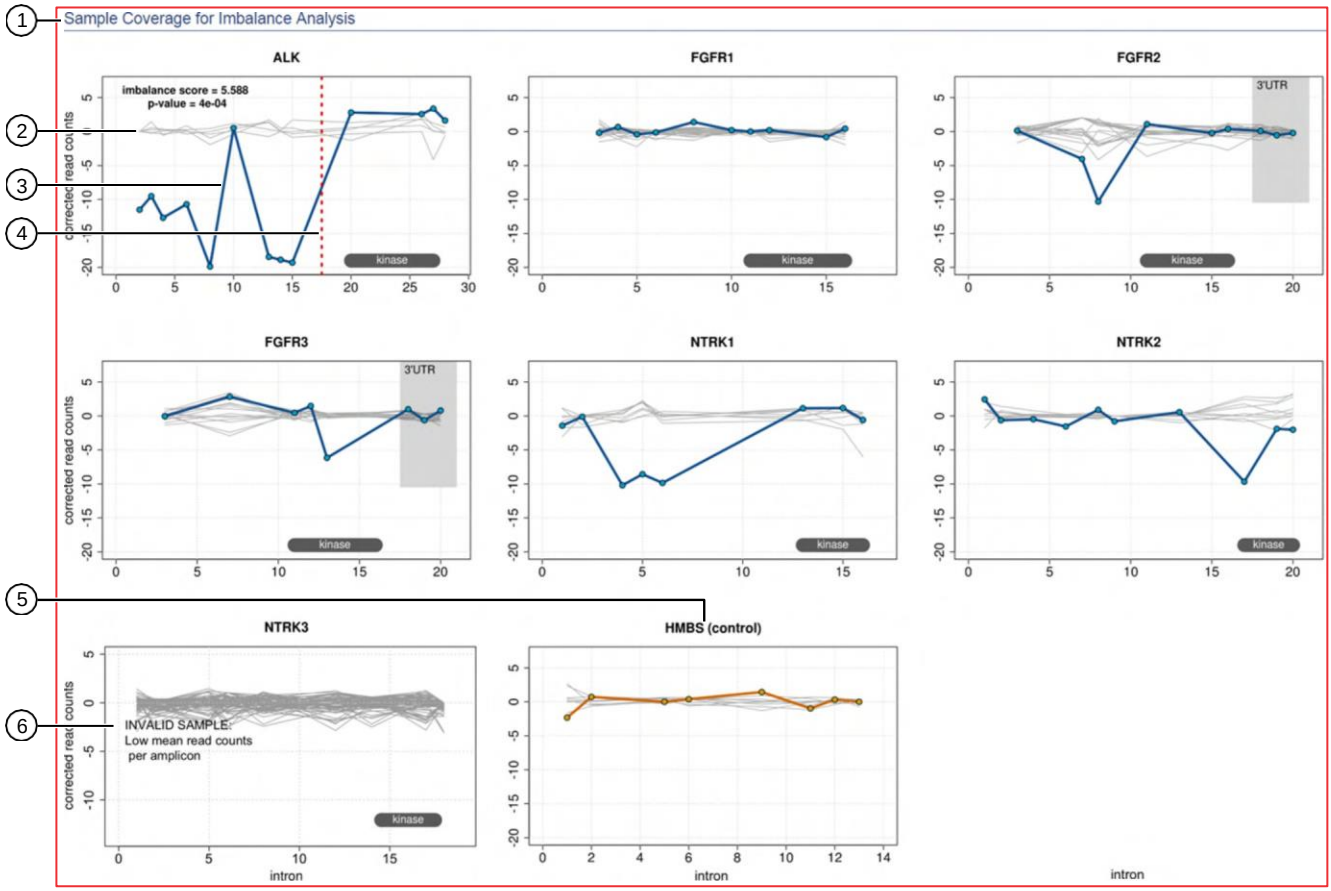
1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see "Fusions table" on page 126.
4. In the top right corner of the screen, click **Visualization►RNA Exon Tile Fusion Imbalance**, then review the **RNA Exon Tile Fusion Imbalance** plots.

## Representative RNA Exon Tile Fusion Imbalance plots




① The **Fusion Imbalance Call: Imbalance Score & P-value** plot shows the imbalance scores and p-values for all the genes in the selected sample. The dashed gray lines mark the threshold for an imbalance call, which is applied to all genes across all samples. Points that fall within the blue shaded area of the plot represent fusion-positive genes (✚). All other points that are outside of the blue shaded area represent fusion-negative genes (◯). Control genes are marked with ◆.

② The **Normalized Read Depth by Gene** plot shows the mean read counts of each gene that is captured on the chip for the selected sample. For each gene, the read counts are normalized to the number of amplicons.




1. The **Sample Coverage for Imbalance Analysis** plots show the expression profile for each exon-exon tiling amplicon for each gene. The Y-axis represents the corrected molecular counts. The X-axis represents individual exon-exon junctions, which are listed from 5′ to 3′. The **imbalance score** and **p-value** are listed in the panel of each gene that was called positive for fusion.
2. Baseline (a cluster of gray lines), generated from a fusion-negative sample.
3. Test sample corrected read coverage (blue line), normalized to the baseline. Each point on the line represents a unique exon-exon junction that was covered by the assay and normalized to the baseline.
4. Predicted range for the fusion break point for a fusion-positive gene (dashed red line).
5. Sample coverage profile for the control gene (orange line).
6. If the collected data are insufficient to determine an imbalance score, the **INVALID SAMPLE** message appears in the panel for that gene.

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View CNV results

The **CNVs** table lists the calls and other information for the copy number variants (CNVs) analyzed in each sample in a run.

To view the **CNVs** table for a sample, click **Results►Sample Results** in the menu bar, then in the **Sample name** column, click the name of the sample of interest. In the **Variants** tab, click **CNVs** to display the data. To export the data in tabular format, click **Export** in the upper right corner of the screen.

## CNVs table

The data displayed in the table depends on the assay that was used in the run.

Results in the table can be filtered using the filtering tools. For more information, see “Search and filter the list of variants” on page 124.

IMPORTANT! *(FFPE samples only)* If the **%Cellularity** value for a sample is set to <100, then the magnitude of copy number gain can be decreased.

| Column | Description |
|---|---|
| Variant ID | The identifier of the CNV variant.<br>Click the link to view more annotation information. |
| Variant Name | The name of the variant. |
| Locus | The starting position of the first amplicon covering the CNV gene. |
| Gene | The gene name. |
| Copy Number | The copy number of a CNV gene locus per genome. This column is available when a positive call is made. |
| Test Result | Indicates the presence or absence of a CNV.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—The absence of a variant; result is below the detection threshold for a CNV call. |
| P-Value | The statistical significance of the CNV ratio measurement. |
| Call Details | The reason for reporting a CNV as NO CALL. |
| CNV Confidence | The CNV confidence interval associated with the call. The 5% lower confidence bound value is the ploidy estimate, where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate, where it is 95% certain that the true ploidy is below that value. |
| CNV Ratio | The ratio of measured CNV gene locus coverage relative to coverage of non-CNV loci. |
| Valid CNV Amplicons | The number of amplicons spanning the CNV call. |

## Review the variant pileup view

You can review variant tracks with the pileup view in Genexus™ Dx Software. The pileup view is a figure that the software generates to show each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup view can be used to identify potential variants.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

The pileup view is shown below the variants table.

## Change the view of the variant pileup

You can zoom and pan the variant pileup view and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 134.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

| Action | Procedure |
|---|---|
| Track the position as you place the pointer over the variant pileup. | Click **Cursor Guide**. |
| Mark the center of the variant pileup. | Click **Center Line**. |
| Remove the track labels. | Click **Track Labels**. |
| Zoom in to increase the magnification of the image. | Click ⊕ **(Zoom in)** one or more times, or double-click the image. |
| Zoom out to decrease the magnification of the image. | Click ⊖ **(Zoom out)** one or more times. |
| Pan across the image to move the view to another location on the screen with its current magnification. | Click and drag the pointer over the image. |

## Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Dx Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

Note: For best results, click the **Expand** option to view details about variants and base calls that are shown in each read coverage track

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to a track, then select an option to adjust the view of the track.

| Option | Description |
|---|---|
| **Expand** | Select this option to view the tracks with the maximum visible height. |
| **Squish** | Select this option to view the tracks with the minimal height for each track. |
| **Collapse** | Select this option to view the tracks with overlapping transcripts shown along a single line. When **Collapse** is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown. |

## Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Dx Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the reference track.

| Option | Description |
|---|---|
| View the reverse strand | Click **Reverse**.<br>This option is available only when the forward strand is shown. |
| View the forward stand | Click **Forward**.<br>This option is available only when the reverse strand is shown. |
| View the reference sequence translation | Click **Three-Frame Translate**.<br>This option is available only when the amino acid sequence is not shown. |
| Hide the reference sequence translation | Click **Close Translation**.<br>This option is available only when the amino acid sequence is shown. |

## Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Dx Software to dynamically customize the view of the data.

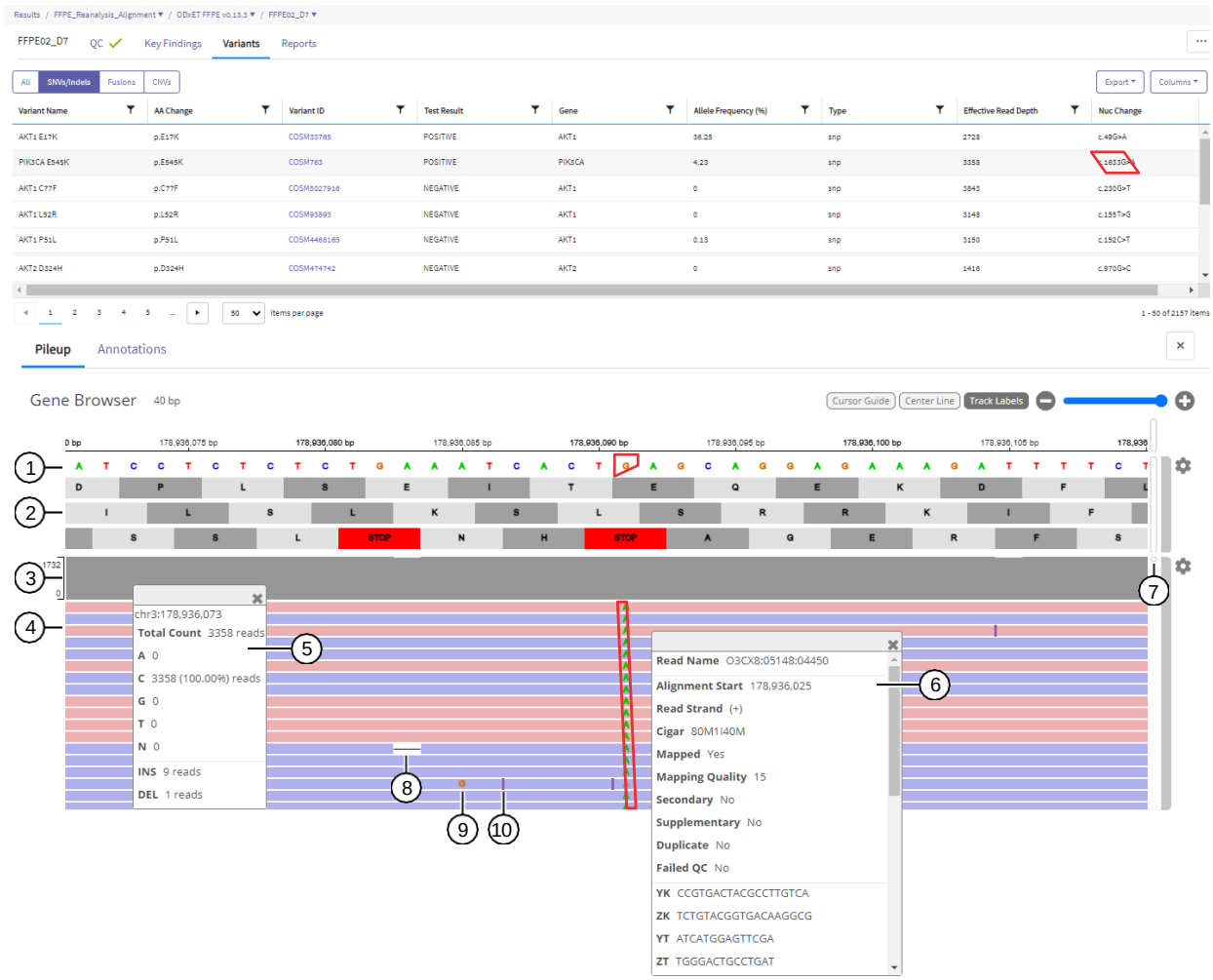
1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙ (Actions) next to the BAM tracks.

| Option | Description |
|---|---|
| View all BAM tracks in monochrome gray | In **Color By**, deselect the checkmark next to **Read Strand**.<br>This option is available only when the BAM tracks are colored by read strand. |
| Distinguish between the direction of the BAM tracks with color | In **Color By**, click **Read Strand**.<br>This option is available only when the BAM tracks are not colored by read strand. |
| Expand, squish or collapse the BAM tracks | For more information, see “Adjust pileup tracks” on page 134. |
| Show all the bases in the BAM tracks | Click **Show all bases**.<br>This option is available only when all bases in the BAM tracks are not shown. |
| Change the minimum threshold for the visibility of BAM tracks | a. Click **Set visibility window**.<br>b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks.<br>When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see “Change the view of the variant pileup” on page 133. You can adjust the visibility window according to the scale shown in the screen. |

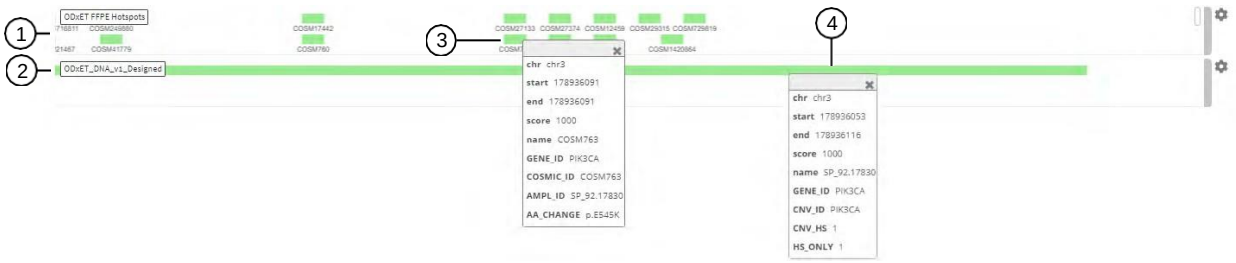
## Examples of a variant pileup view

The following figure is an example of a variant pileup view, shown in the **Gene Browser** in the **Variant** tab. The variant pileup view graphically represents the sequencing tracks, the reference track, and the annotation tracks, and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant are indicated in the image, both in the variant table and the in the pileup view. You can customize your pileup view. Reorder the tracks displayed by clicking a gray bar on the right edge of a track, then dragging the track to a different location. You can also click-drag the cursor at any position in the image to move the image left or right in the screen. For more information, see “Adjust pileup tracks” on page 134.




1. The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
2. The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 134.
3. The coverage track.
4. The BAM track.
5. Click the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
6. Click a BAM track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
7. Use the slider to view more tracks.
8. Deletion.
9. Alternate base in the BAM read coverage track.
10. Insertion.

The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.




① The hotspot track.
② The target region track.
③ Click a hotspot track for additional details, such as the start and end positions.
④ Click the target region track for additional details, such as the start and end positions.

## Coverage histogram

When you zoom in on the variant pileup view, you see the coverage histogram and reads from the .bam track. The histogram in gray shows read depth at that location and reads from the .bam track show read direction and location of variants.




① Read depth
② Read location and direction of variants

## View the variants from an Oncomine™ Dx Express Test run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. Review detailed variant data and read coverage tracks.




**Example visualization**

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate a family.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" indicates insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

| Option | Description |
|---|---|
| View the read coverage tracks sorted by family | Click ⚙ **(Actions)** next to the read coverage track, then select **Color►Molecule**. |
| Adjust the BAM tracks | Click ⚙ **(Actions)** next to the read coverage track, then select an option to adjust the view of the track. For more information, see “Adjust pileup tracks” on page 134. |
| Review detailed data about a BAM read | Single-click the read track to obtain information such as the mapping quality, the strand, and the read base. |
| Review distribution of base calls at a selected position | Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions. |

## View the QC results

You can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Dx Software.

Each run includes NTC and positive controls that are added automatically to each run.

1. To view the **QC** screen, in the **Results / Sample Results** screen, click a sample name in the **Sample Name** column. In the **Results** screen, click the **QC** tab. The QC status for each metric is indicated beneath each QC test (Run QC, Templating Control QC–CF–1, Sample QC–DNA, and Sample QC–RNA).

2. Review the QC metrics.

   For more information, see “QC results” on page 140.

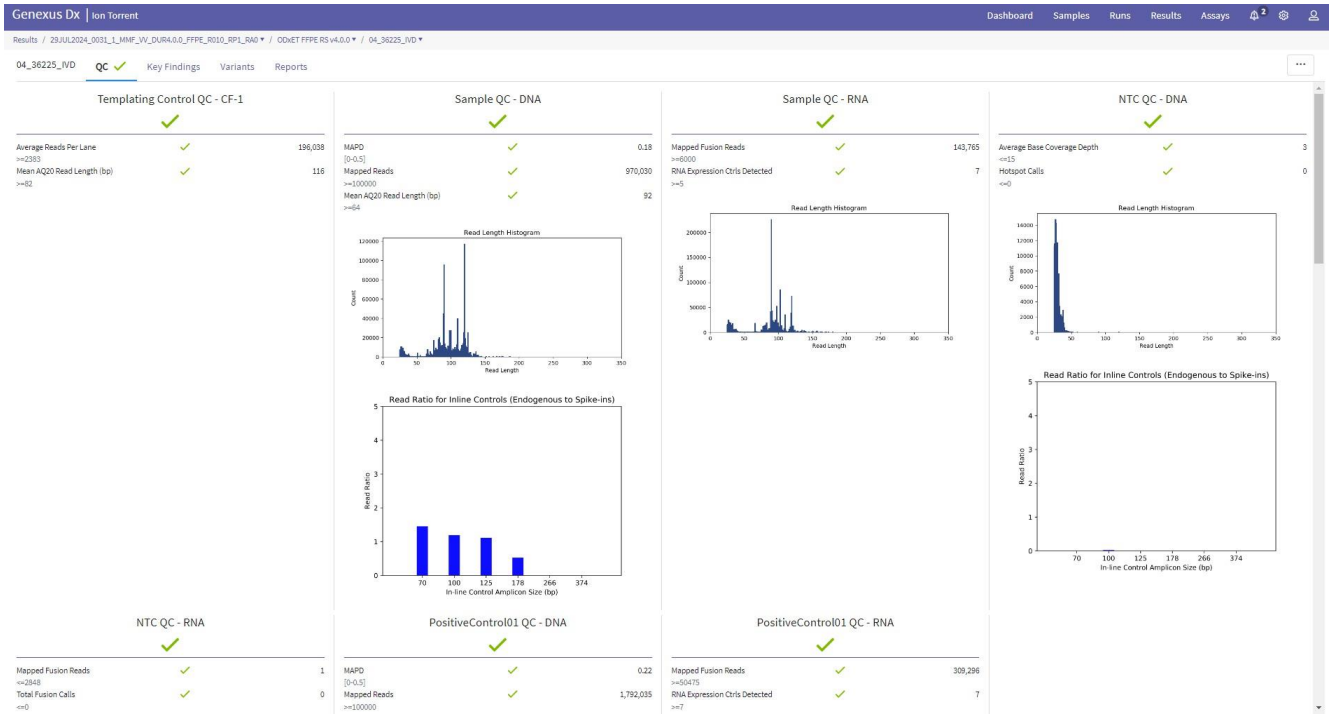
## QC results

The quality control (QC) metrics for each sample that was sequenced in a run are displayed in Genexus™ Dx Software.

If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test in order to pass. The QC status is divided into the following categories.

- ✔ (Passed) indicates the sample passed all QC metrics.
- ✘ (Failed) indicates the sample failed a QC metric.
- — (Not Calculated) indicates a sample did not undergo QC analysis.

The data shown in the screen depend on the assay that was used in the run. Not all metrics are shown for every assay.

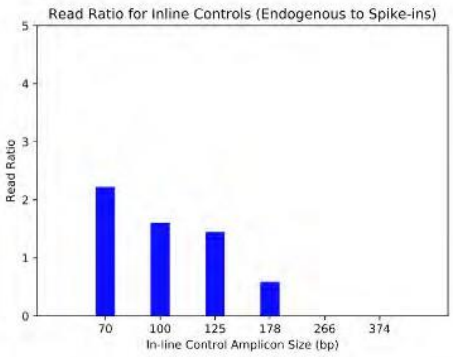



| Metric | Description |
|---|---|
| Purification QC | Quality control information for nucleic acid extraction that is performed on a Genexus™ Dx Purification System. |
| Sample Concentration RNA | The concentration of extracted RNA after purification. |
| Sample Concentration DNA | The concentration of extracted DNA after purification. |
| Run QC | General run quality control information. |
| Key Signal | The average signal after software processing for library ISPs that identically match the library key (TCAG). |

*(continued)*

| Metric | Description |
|---|---|
| Percent Loading | The number of wells with ISPs divided by the number of the total addressable wells in a run. |
| Raw Read Accuracy | The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as 100 * (1 - (sum(per base error)/sum(per base depth))). |
| Templating QC—CF-1 Control | Sequencing quality metrics of the control fragment. These metrics indicate templating success. |
| Average Reads Per Lane | The number of CF-1 reads divided by the number of chip lanes used in the run. |
| Mean AQ20 Read Length (bp) | Average length, in base pairs, at which the accuracy rate is ≥99% for CF-1 reads. |
| Sample QC—DNA | Sequencing quality metrics of the sample DNA library. |
| MAPD | MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of ≤0.5 for FFPE samples indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls. |
| Mapped Reads | The total valid mapped reads. |
| Mean AQ20 Read Length (bp) | The average length, in base pairs, at which the accuracy rate is ≥99% for all aligned reads of a library. |
| Mean Mol Cov | Uniformity of molecular coverage for all amplicons. |
| Read Length Histogram | The histogram presents all filtered and trimmed DNA library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |

*(continued)*

| Metric | Description |
|---|---|
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of genomic sample reads to control reads for each inline control amplicon.<br>  <br><br>Note: The Read Ratio for each inline control amplicon is expected to be 1–3 with 10 ng DNA input (high quality DNA inputs) for Ion AmpliSeq™ HD chemistries . |
| Sample QC – RNA | Sequencing quality metrics of the sample RNA library. |
| Mapped Fusion Reads | The total valid mapped reads. That is, the mapped reads that pass the thresholds and filters across all of the RNA targets in the reference file. This number is a subset of the mapped reads shown in the Run Samples table in the Assay Metrics. |
| RNA Expression Ctrls Detected | The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. |
| Read Length Histogram | The histogram presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| NTC QC – DNA | Sequencing quality metrics of the no-template control. |
| Hotspot Calls | The number of positive hotspots detected for the no-template control. |
| Average Base Coverage Depth | The average number of DNA reads of all targeted reference bases. |
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of no-template control sample reads to control reads for each DNA inline control amplicon. |
| NTC QC – RNA | Sequencing quality metrics of the no-template control. |
| Mapped Fusion Reads | The total number of valid mapped reads for the NTC. |
| Total Fusion Calls[1] | The number of fusion variants detected for the no-template control. |

*(continued)*

| Metric | Description |
|---|---|
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| PositiveControl01 QC - DNA | Sequencing quality metrics of the DNA control. |
| MAPD | MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of ≤0.5 for FFPE samples indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls. |
| Mapped Reads | The total valid mapped reads for DNA control. |
| Mean AQ20 Read Length (bp) | The average length, in base pairs, at which the accuracy rate is ≥99% for aligned reads of the DNA control. |
| <Variant ID> | Listing of each reported DNA control variant and its corresponding allele frequency or CNV copy number, and its positive or negative status. |
| Read Length Histogram | The histogram presents all filtered and trimmed DNA control library reads that are reported in the output BAM file for DNA positive controls (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of DNA control reads to inline control reads for each inline control amplicon. |
| PositiveControl01 QC - RNA | Sequencing quality metrics of the RNA control. |
| Mapped Fusion Reads | The total valid mapped reads for the RNA control. |
| RNA Expression Ctrls Detected | The number of expression control genes detected for the RNA control. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. |
| <Fusion Targeted Isoform or Expression Imbalance> | Listing of each reported fusion targeted isoform and fusion expression imbalance in the RNA control, and its positive or negative status. |
| Read Length Histogram | The histogram presents all filtered and trimmed RNA control library reads that are reported in the output BAM file for RNA controls (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |

[1] Counts for this metric include fusions, and non-wild type RNAExonVariant and RNAExonTiles variants.

### MAPD copy number QC metric

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on runs that detect CNVs.

MAPD is one of the metrics that we define to assess whether the panel data are useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log2 ratios per tile for a given run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log2 ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log2 ratio belongs to two pairs.

Formally, if `xi` is the log2 ratio at position `i`, with `i` ordered by genomic position:

```
MAPD = median ( | x(i - 1) - x(i) | )
```

MAPD is a per-sequencing run estimate of copy number variability, similar to standard deviation (SD). If one assumes the log2 ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log2 ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

## Review run results

In the **Results / Run Results** screen, runs that are pending, in progress, or complete are listed. Runs with a status of failed, aborted, or stalled are also listed.

You can filter the list of run results by clicking **(Filter)** in a column of interest, then entering a full or partial run or assay name, or other applicable filter term.

The following run information appears in the **Results / Run Results** screen.

| Column | Description |
|---|---|
| Run Name | The unique name of the run given when it was created in the software. Click a run name to open the Run Summary.<br>Runs that are reanalyzed are listed with (Reanalysis) after the run name. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910). |
| Assay Name | The name of the assay selected for the run. You can view the Assay Name and corresponding Assay Full Name for all assays in the Assays►Manage Assays screen. |
| Run Status | The status of the run. For example, Analysis In Progress, Executing Plugins, Analysis Completed, Terminated, Archival: In Progress, Purification In Progress, or Purification Completed. |
| Total Samples | The total number of samples in a run. |

*(continued)*

| Column | Description |
|---|---|
| PCR Plate Number | A unique identifier for the 96-well plate used for library preparation and templating. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910). |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |

You can also perform the following actions in the **Results / Run Results** screen.

| Option | Description |
|---|---|
| Actions | Available action links for a run are shown when you place the pointer over the row of a run. The actions that are available depend on the type of run.<br>• Delete—Delete the run.<br>• Audit—View the audit trail for the run.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>• Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. |
| Actions | These actions are available only for Sample to Result runs.<br>• View Plan—View detailed run plan information.<br>• Assign Kit Barcodes—<br>• Abort—Enables you to abort a run after purification, but before sequencing. This action is available when the run status is Purification Review Required, or when the run status is Purification Completed and some purification samples have been excluded from sequencing. |
| Actions | Available action links for a run are shown when you select the checkbox to the left of the Run Name.<br>• Clear—Deselect the selected run or runs.<br>• Retain Data—Click to keep run data regardless of disk cleanup settings. To undo, select Release Data.<br>• Archive Data—Click to archive run data. Select run data options (Sequencing Output, Intermediate Files) in the Archive Confirmation dialog box. Analysis Results are selected by default and are not optional.<br>• Delete—Delete the run data of the selected run or runs. |

## View the run summary

The run summary provides an overview of the run. The information that is displayed includes the name of the assay used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information. Metrics from sample purification are also provided, if applicable.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Name** column, click the run name of interest.
   The **Run Summary** tab opens.
3. Review the run summary.

| Action | Procedure |
|---|---|
| View the assay metrics. | In the Assays section, click the assay name of interest. |
| View the sample locations in an image of a 96-well sample plate. | In the Sample Locations section, click PCR Plate View. |
| Reanalyze a run with a new assay. | In the upper right corner of the screen, click ••• (More Options)►Reanalyze.<br>For more information, see the software help system or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910). |
| Download customer support archive (CSA) log files for the run to help with troubleshooting. | In the upper right corner of the screen, click ••• (More Options)►CSA. |
| View the history of variant classifications. | In the upper right corner of the screen, click ••• (More Options)►Variant Audit.<br>The information is available only for results that include variant classifications. |
| View the run report. | Click the Run Report tab. |

## The run summary

The run summary displays run information, assays used in the run, sample locations, metrics from sample purification, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

| Item | Description |
|---|---|
| Run Name | The name of the run. |
| Started On | The date and time when the run was started. |
| Completed On | The date and time when the run was completed. |
| Run Status | The status of the run. |
| Report Template | If the option to generate a report was enabled in the run plan, the report template used is listed. |
| Chip Type | The semiconductor sequencing chip used in the run, such as the Genexus™ Dx GX5™ Chip. |
| Library Chemistry | The type of library chemistry used in the run, such as Ion AmpliSeq™ HD. |

Sequencing instrument information

| Item | Description |
|---|---|
| Instrument Name | The name of the sequencer. |
| Instrument Serial Number | The serial number of the sequencer. |
| Status | The status of the sequencing portion of the run. |
| Operator | The name of the person who was signed in the Genexus™ Dx Integrated Sequencer when the sequencing run was performed. |
| Start Date | The date and time when the run was started on the sequencer. |
| Completion Date | The date and time when the run was completed on the sequencer. |
| Sequencing Flows | The number of flows performed by the sequencer. |

Purification instrument information

| Item | Description |
|---|---|
| Purif. Instrument | The name of the purification instrument. |
| Instrument Serial Number | The serial number of the purification instrument. |
| Status | The status of the purification portion of the run for the batch. |
| Operator | The name of the person who was signed in the purification instrument when purification was performed. |
| Start Date | The date and time when the batch was started on the purification instrument. |
| Completion Date | The date and time when the batch was completed on the purification instrument. |

Assays table

| Item | Description |
|---|---|
| Assay Full Name | The name of the assay used in the run. Click the assay name to display the details of the assay. |
| Assay Name | An abbreviated name of the assay or assays used in the run. |
| Analysis Version | The version of the assay used for analysis. |
| Application Type | The application type for the assay, such as DNA and Fusions. |
| Lane | The chip lane or lanes used in the sequencing run for the assay. |
| Total Samples | The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample. |

Purification Samples table

| Item | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Sample Type | A term that describes the sample, for example, FFPE. |
| Nucleic Acid Type | DNA, RNA, or DNA+RNA. |
| Conc. (ng/µl) | The concentration of the sample measured by the purification instrument.<br>In some instances, such as when purification is in progress or when the sample is a no-template control, the values is listed as N/A.<br>If the concentration is not within the QC concentration range specified by the assay, a Quantity Not Sufficient (QNS) alert is displayed. |
| QC Conc. Range (ng/µl) | The QC concentration range for the assay. |

Purification Samples table *(continued)*

| Item | Description |
|---|---|
| Batch Status | The run status of a purification batch.<br>The status can be N/A, Planned, Review, Started, Aborted, or Completed. |
| Archive Position | The sample positions in the archive plate.<br>Sample positions can be in rows A to D and in columns 1 to 8. |
| Library Prep | An indicator of whether the sample is selected for sequencing.<br>• ✔(Checkmark) – indicates that the sample is selected for sequencing.<br>• ⊘– (No icon) indicates that the sample is not selected for sequencing.<br>• N/A – indicates that purification has not started or is in progress. |

Sample Locations table

| Item | Description |
|---|---|
| PCR Plate View | Click to view the sample locations in an image of a 96-well sample plate. |
| Well Pos. | The well position indicates the location of the sample on the plate. |
| Library Batch ID | The unique identifier created for the library batch. This information is available only for Library to Result runs. |
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA+RNA. |
| Vol. (µl) | The volume of the sample. |
| Conc. (ng/µl) | The concentration of the sample. |
| Dilution Factor | The dilution factor of the sample. |
| Kit Barcodes | The barcode for the kit used for nucleic acid extraction, if applicable. |
| Barcode | The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101. |
| Assay Name | The assay that was used to sequence the sample for the indicated well position. |
| Library Position | The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run. |

Reagents table

| Item | Description |
|---|---|
| Consumable | Consumables used in the run, such as the PanelKit and LibraryKit, are listed. |
| Barcode | The consumable barcode, if applicable. |
| Part # | The consumable part number, if applicable. |
| Expiration Date | The consumable expiration date, if applicable. |
| Lot # | The consumable lot number, if applicable. |

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

## View assay metrics and the run report

The run report provides detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run.

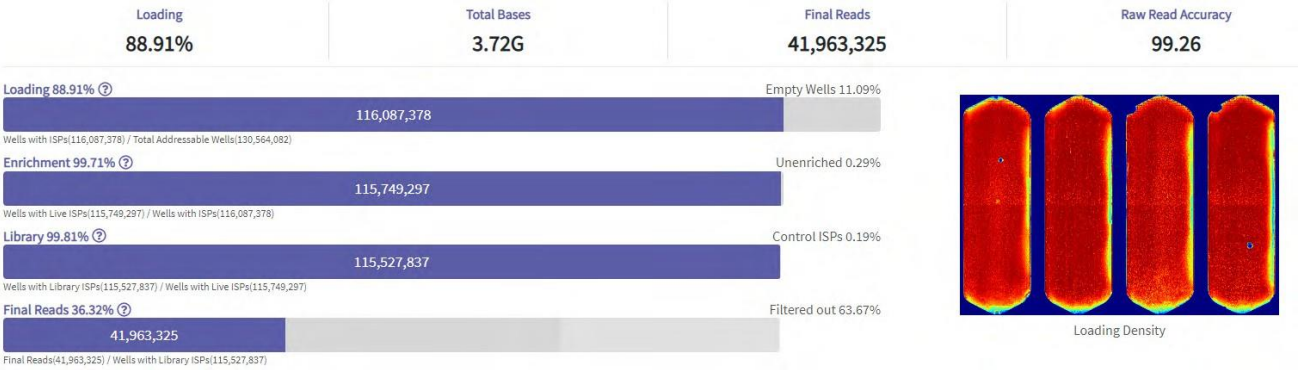
For runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

To view the **Run Report**, in the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest. In the **Run Summary** screen, click the **Run Report** tab.

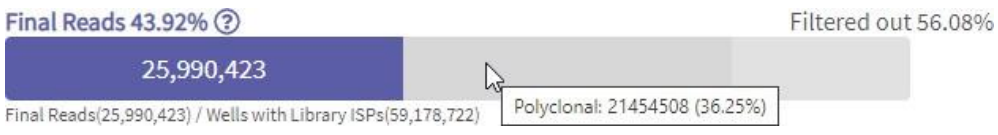
The **Run Report** is assay-specific and cannot be viewed within the sample results screens. To view the **Run Report**, ensure that **Select Sample** is selected in the **Select Sample** dropdown menu.

### Assay metrics and the run report

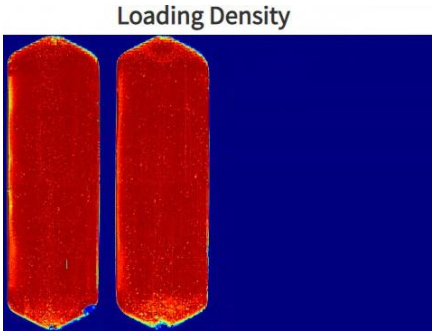
The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.



Run Report

| Metric | Description |
|---|---|
| Loading | The number and percentage of total addressable wells on the chip that contain an ISP. |
| Enrichment | The number and percentage of wells ISPs that contain live ISPs. |
| Library | The number and percentage of wells with live ISPs that contain Library ISPs. |
| Final Reads | Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement. |
| Total Bases | The number of filtered and trimmed base pairs that are reported in the output BAM file. |
| Raw Read Accuracy | The raw read accuracy across each individual base position in a read calculated as, (1−[total errors in the sequenced reads]/[total bases sequenced]) × 100. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is *not* based on consensus accuracy across multiple reads for the same base position. |
| Wells with ISPs | The number of wells that contain an ISP. |
| Unenriched | The number and percentage of wells with ISPs that do not contain live ISPs. |
| Total Addressable Wells | Wells on the chip that can be physically reached by a library. |
| Empty Wells | The percentage of total addressable wells on the chip that do not contain an ISP. |
| Wells with Live ISPs | Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key. |
| Wells with Library ISPs | Loaded wells with live ISPs with a key signal that is identical to the library key signal. |
| Control ISPs | Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal. |
| Filtered out | The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads. |
| Polyclonal | Wells with a live ISP that carries clones from two or more templates.<br>To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot.<br>  |

Run Report *(continued)*

| Metric | Description |
|---|---|
| Low Quality | Loaded wells with a low or unrecognizable signal.<br>To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot.<br>  |
| Adapter Dimer | Loaded wells with a library template of an insert size less than 8 bases.<br>Adapter dimer reads represent a small proportion of total reads and can be seen by hovering the pointer over the right end of the Final Reads bar.<br>  |
| Loading Density | A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs.<br>  |

## Run Samples

The Run Samples table lists read data for each individual sample in the assay.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered in the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA+RNA. |
| Barcode | The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample. |
| Total Reads | The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. |
| Mean Read Length | The average length, in base pairs, of usable library reads for each sample. |
| ≥Q20 Bases | The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample. |

Run Samples *(continued)*

| Column | Description |
|---|---|
| Mapped Reads | The number of reads that are mapped to the reference file. |
| On Target Reads | The percentage of sequencing reads mapped to any target region of the reference. |
| Mean Depth | The average number of reads of all targeted reference bases. |
| Uniformity | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. |
| Read Length Histogram | A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).   |

### Download a run report

You can download a run report summary in PDF format. The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “View assay metrics and the run report” on page 150. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
   Multiple panes including a **Run Report** pane, a **Variant Report** pane, and any panes for customized reports that have been generated are shown.
4. In the **Run Report** pane, click **Download Report** to download a run report summary in PDF format.

## Assign PCR Plate

Genexus™ Dx Software lets you track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

Note: You should assign a PCR plate to a run if there are leftover pooled libraries that you want to use at a later time to perform a Library to Result run. You can look up the PCR plate number on completed runs to get the library and barcode mapping, then enter the same Barcode information into the Library to Result run.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.

   The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

# Clinical Test Report and Clinical Laboratory Report

The Clinical Test Report and Clinical Laboratory Report are PDF variant reports generated by the Oncomine™ Dx Express Test for each sample in a sequencing run. Each system-installed assay for the test provides Clinical Test Report and the Clinical Laboratory Report templates, set as default templates, that you can use without change to generate reports. Alternatively, you can modify the customizable sections of the layout and contents of the default report templates to create user-defined templates. Some sections of the report templates are locked and cannot be customized. For more information about creating custom report templates, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

To make Clinical Test and Laboratory Reports available for each sample result on completion of a sequencing run, enable **Generate Report** in the **Setup** step when you plan the run. For more information, see Chapter 4, “Plan a run in Genexus™ Dx Software”. If you need to generate a report after a run is complete, see “Generate a variant report” on page 160.

When Clinical Test and Laboratory Reports have been generated for a sample result, they are available for download in three places:

- In the **Results** screen for the sample when you click ••• **(More Options)►Download Files**. In the dialog, select the **Reports** checkbox to enable download of both reports.
- In the pane for the Clinical Test Report or Clinical Laboratory Report in the **Reports** tab when you click ⋮ **(More)►Download Report**.
- In the **Results / Sample Results** screen when you place the pointer over the row for that sample, then click **Report** to download the reports.

The Clinical Test Report and Clinical Laboratory Report can be electronically or manually signed by users. Electronic signatures are shown in the **Report Signed By** section of the report.

## Sections of the Clinical Test Report

The Clinical Test Report contains the following sections and information.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered in the software. You can customize the format of the Sample Details section when you create a new report template. To create a new report template, click Assays►Manage Presets, then in the Report Templates tab, click + Add New. |
| Tumor Mutation Profiling Results: Summary | A summary of results for variants detected in the sample classified in 3 biomarker levels:<br>• Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use<br>• Level 2: Cancer Mutations with Evidence of Clinical Significance<br>• Level 3: Cancer Mutations with Potential Clinical Significance<br>If a detected variant is listed in the Companion Diagnostics (CDx) Biomarkers level, the associated therapy or therapies are listed after the variant. For further information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 158. |
| Tumor Mutation Profiling Results: Detected | A list of variants detected in the sample grouped by SNVs/Indels, Fusions, and CNVs. Variant ID, amino acid change, and fusion information is also provided in this section. This section is customizable. Additional columns from the Variants tab can be included in a custom report template if you would like to show more information for positive calls in the report. |
| Intended Use | Intended use statement for the Oncomine™ Dx Express Test, including a table that lists the current Companion Diagnostic Indications. |
| Test Description | General description of the test, including a table listing the types of genetic alternations detected by the Oncomine™ Dx Express Test, and the genes targeted by the test for each type of alteration. |
| Tumor Mutation Profiling Results: No Calls | A list of No Calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. A No Call is defined as a call for which some evidence for the presence of a variant exists, but the call does not pass one or more filters that are required for a high confidence variant call. This section is customizable. Additional columns from the Variants tab, for example, Call Details, can be included in a custom report template if you would like to show in the report the possible reason or reasons why a No Call was made. |
| Tumor Mutation Profiling Results: Negative Calls | A list of negative calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. This section is customizable or deletable. Additional columns from the Variants tab can be included in a custom report template if you would like to show in the report the reason why a negative call was made. |
| Report Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |

*(continued)*

| Section | Description |
|---|---|
| Manual Signatures | *(Optional)* A section for physical signatures of the printed report. |
| Custom Text | *(Optional)* A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Footer | The footer is repeated on each page of the report. |

## Sections of the Clinical Laboratory Report

The Clinical Laboratory Report contains all the information in the Clinical Test Report, and in addition includes the **QC Metrics**, **Sequencing Run Details**, **Reagents**, and **Kit Barcodes** sections.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered in the software. You can customize the format of the Sample Details section when you create a new report template. To create a new report template, click Assays►Manage Presets, then in the Report Templates tab, click + Add New. |
| Tumor Mutation Profiling Results: Summary | A summary of results for variants detected in the sample classified in 3 biomarker levels:<br>• Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use<br>• Level 2: Cancer Mutations with Evidence of Clinical Significance<br>• Level 3: Cancer Mutations with Potential Clinical Significance<br>If a detected variant is listed in the Companion Diagnostics (CDx) Biomarkers level, the associated therapy or therapies are listed after the variant. For further information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 158. |
| Tumor Mutation Profiling Results: Detected | A list of variants detected in the sample grouped by SNVs/Indels, Fusions, and CNVs. Variant ID, amino acid change, and fusion information is also provided in this section. This section is customizable. Additional columns from the Variants tab can be included in a custom report template if you would like to show more information for positive calls in the report. |
| Intended Use | Intended use statement for the Oncomine™ Dx Express Test, including a table that lists the current Companion Diagnostic Indications. |
| Test Description | General description of the test, including a table listing the types of genetic alternations detected by the Oncomine™ Dx Express Test, and the genes targeted by the test for each type of alteration. |

*(continued)*

| Section | Description |
|---|---|
| Tumor Mutation Profiling Results: No Calls | A list of No Calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. A No Call is defined as a call for which some evidence for the presence of a variant exists, but the call does not pass one or more filters that are required for a high confidence variant call. This section is customizable. Additional columns from the Variants tab, for example, Call Details, can be included in a custom report template if you would like to show in the report the possible reason or reasons why a No Call was made. |
| Tumor Mutation Profiling Results: Negative Calls | A list of negative calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. This section is customizable or deletable. Additional columns from the Variants tab can be included in a custom report template if you would like to show in the report the reason why a negative call was made. |
| QC Metrics | • NTC QC – DNA—A summary of the DNA no-template control quality control metrics.<br>• NTC QC – RNA—A summary of the RNA no-template control quality control metrics.<br>• Sample QC – RNA—A summary of RNA sample quality control metrics.<br>• Sample QC – DNA—A summary of DNA sample quality control metrics.<br>• Templating Control QC – CF-1—A summary of the CF-1 quality control metrics.<br>• Positive Control01 QC – DNA—A summary of the DNA positive control quality control metrics.<br>• Positive Control01 QC – RNA—A summary of the RNA positive control quality control metrics.<br>• Purification QC—A summary of QC metrics for the purification run.<br>This section is locked and cannot be customized. |
| Sequencing Run Details | Contains the following subsections:<br>• Assay Name—The assay used in the run<br>• Panel—The panel used in the run<br>• Started On—The date and time that the run was started<br>• Completed On—The date and time when the run was completed<br>• Operator—The user who sent the run to the instrument<br>• Instrument—The name of the instrument<br>• Serial Number—The serial number of the instrument<br>This section is locked and cannot be customized. |
| Reagents | Details about run consumables, including part numbers, expiration dates, and lot numbers.<br>This section is locked and cannot be customized. |
| Kit Barcodes | Details about the kits used for each sample sequenced in the run, including kit barcode information, nucleic acid type, and kit type. |
| Report Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |

*(continued)*

| Section | Description |
|---|---|
| Manual Signatures | *(Optional)* A section for physical signatures of the printed report. |
| Custom Text | *(Optional)* A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Footer | The footer is repeated on each page of the report. |

### FDA's 3-Level biomarker classification-based variant reporting

Oncomine™ Dx Express Test results are reported in alignment with an FDA's 3-Level biomarker classification-based system. Evidence of clinical relevance is obtained from a curated database called Oncomine™ Knowledgebase. Variants are classified into clinical relevance categories as defined below:

- Level 1: Companion Diagnostics (CDx) Biomarkers—gene variants that have companion diagnostic claims in the corresponding cancer type that are associated with an FDA-approved therapy as listed in Table 1 of the intended use.
- Level 2: Cancer Mutations with Evidence of Clinical Significance—gene variants that are not prescriptive or conclusive for labeled use of a specific therapeutic product, and for which clinical validation has not been performed, but which have curated clinical evidence associated with FDA-approved therapies and/or National Comprehensive Cancer Network (NCCN) clinical guidelines (Category 1 and 2A) in the sample cancer type.
- Level 3: Cancer Mutations with Potential Clinical Significance—gene variants not considered Level 1 or Level 2 biomarkers that can be described as cancer mutations with potential clinical significance. These variants are not prescriptive or conclusive for labeled use of a specific therapeutic product, and clinical validation has not been performed. These variants have curated clinical evidence associated with NCCN clinical guidelines Category 2A in a different cancer type or Category 2B in any cancer type. They may also have curated evidence as an enrollment criterion in clinical trials or biomarkers with evidence in peer-reviewed literature.

Variant category files are updated with every new assay definition file.

## View a Clinical Test Report or Clinical Laboratory Report

A variant report must be generated before you can view the report. If a report template is set as the default template for the assay associated with the run, one or more variant reports are generated after a run completes successfully. Alternatively, you can generate one or more variant reports after a run completes successfully. For more information, see “Generate a variant report” on page 160.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.

3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown. The pane for a variant report includes the following information.
   - Report name
   - Report template name
   - Date that the report was generated
   - Username for the user who generated the report
   - Language used for the report
   - If a variant report has been electronically signed at least once, the signature information is listed in the pane for the variant report. If multiple users have signed the report, you can place the pointer over the electronic signature information to view more details. If the electronic signature that is used is associated with a lock-level signature type, the report is locked.
   - If a variant report is locked, **(Lock)** is shown in the upper right corner of the pane.
4. In the pane of the report of interest, click **(More)►View Report**.

The report opens in a new browser tab. You can download variant reports as PDF files.

## Download a Clinical Test Report or Clinical Laboratory Report

You can download a Clinical Test Report or Clinical Laboratory Report for a sample result of interest from the **Results / Sample Results** screen, from the pane that shows the report, or as part of a download of results files.

1. In the menu bar, click **Results►Sample Results**.
2. Download the report with one of the following options.

| Option | Procedure |
|---|---|
| Download a ZIP file that contains all variant reports for a sample result. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click **(More Options)►Download Files**. In the **Download Files** dialog, select the **Reports** checkbox, then click **Download**. |
| Download the variant report in PDF format from the **Reports** tab for the sample results. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click the **Reports** tab. In the pane of the report of interest, click **(More)►Download Report**. |
| Download a ZIP file that contains all variant reports for a sample result from the **Sample Results** screen. | In the **Sample Results** screen, place the pointer over the row of the sample of interest, then click **Report**. |

   A ZIP file that contains the PDF report is downloaded to the computer if you download reports from the **Sample Results** screen or as part of the results files. A report in PDF format downloads if you use the **Reports** tab for the download.
3. Extract the downloaded files, then open the PDF file in an appropriate viewer.

## Generate a variant report

You can generate a new variant report for sample results after a run is complete. A **(Lock)** in the variant report indicates that the electronic signature option for the report is locked. After a variant report is locked, the report cannot be electronically signed by any other user.

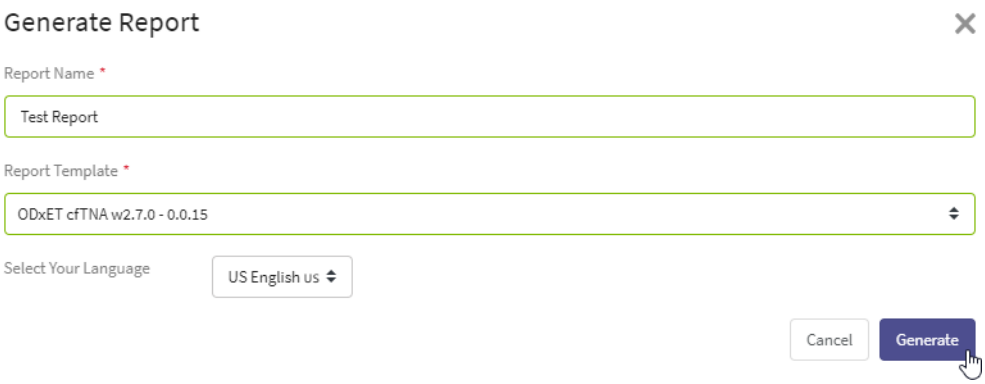
You can generate multiple reports for a sample result, if each report is named uniquely, and is generated in a different language, or uses a different report template.

When generating a customized report, you can update any report template selections. You can use this procedure to generate multiple reports for a sample, if a unique report name is entered for each report. For example, you may want to generate reports for different languages, or reports that use different templates.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Results / Sample Results** screen, click the sample of interest in the **Sample Name** column.
3. Select the **Reports** tab, then click **+ Generate Variant Report**.
4. In the **Generate Report** dialog box, change the name of the report that is generated by the software, if needed.
   a. Select the report template and language of the report.
   b. If the option to make custom text **Editable on Report Generation** was selected when the report template was created, enter a title in the **Custom Text** section and if needed, a description.

   The **Report Template** list includes the report templates that are associated with the assay that was used in the run. For information about creating report templates, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

   If you select the same report template that was used to generate a variant report, and have not locked that report, the new selections you make override the previous variant report.




5. Click **Generate**.
   A draft version of the report is added to the **Reports** tab.

A pane for the new report is added next to the Run Report pane in the **Reports** tab. Reports that have been generated are available for download in the **Reports** tab, in the **Sample Results** screen, and in the ZIP package that contains results files.

## Sign off variant reports

Manager, administrator, operator, and report-level users can electronically sign a variant report for sample results in Genexus™ Dx Software. A sample result can have multiple variant reports only if each report uses a different report template, and a different language.

---

IMPORTANT! If you use the same template and language to regenerate an existing report, the existing report will be overwritten.

---

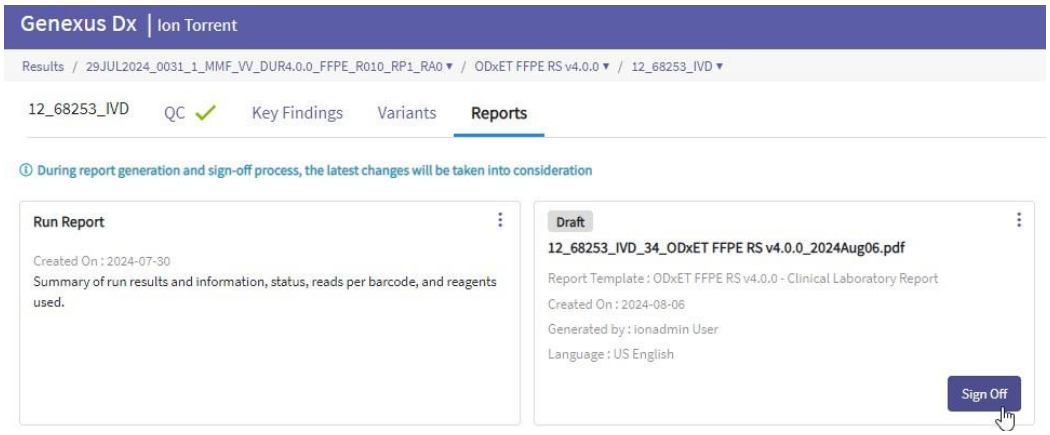
Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign the report, you will be the last person to sign the report before it is locked.

The signature information appears in the variant report PDF file or a user-created report, if selected. For more information, see “Clinical Test Report and Clinical Laboratory Report” on page 154. For information about how to create a report template, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN0024910).

Multilanguage support for PDF report generation is provided.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample result of interest in the **Sample Name** column.
3. In the **Reports** tab, in a variant report pane that is labeled as **Draft**, click **Sign Off**.



The **Sign Off Report** dialog box opens and shows the name of the report, the template used for the report, and the following information, which is based on the profile of the user who signs the report.

- **Electronic Signature**
- **User Name**
- **Language**

4. In **Password**, enter the password.
5. In **Sign Off Comments**, enter a comment.
6. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.

   If you entered footer information in the **Footer Field** when you created a report template, or if text was entered during a previous sign off, the same footer information is shown in the **Sign Off Report** dialog box. You can edit the text in the **Footer Field** to change the information that will be shown in the footer of the PDF report.
7. Click **Sign Off** to confirm the electronic signature.

   The pane for the signed variant report is updated to include the signature information for the variant report. If the electronic signature that is associated with a lock-level signature type is used, a **(Lock)** is shown in the variant report pane and the report is locked.
8. In the menu bar, click **Results►Sample Results** to return to the **Results / Sample Results** screen.

   If the **Electronic Signatures** column is shown in the **Sample Results** screen, the electronic signature types for users who signed the report are listed, and a **(Lock)** is shown for reports that are locked.
9. In the row of the sample of interest, click **Report** to download the report.

The report is signed. The **Electronic Signatures** section of the report pane is updated to include your username and the signature type for your account. You can hover over the signature type to show the list of signature types that are completed for the report. If multiple electronic signatures are added to the report, an electronic signature type for each signer is shown in the **Electronic Signatures** column of the sample results table. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

You cannot generate new reports from the variant results if a report from those results is signed off and locked. If you must change the report, reanalyze the sample and generate a report for the reanalyzed sample.

You can send notifications for report sign-offs to other users. For more information, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

# Reanalyze a run or sample

If a sequencing analysis fails, you can reanalyze a run or sample using the same assay. For information about how to reanalyze a run or sample, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

# A Troubleshooting


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## Customer Support Archive (CSA)

You can download an archive that a Technical Support representative can use to diagnose Genexus™ Dx Software issues. The Customer Support Archive (CSA) contains log files and other technical data about Genexus™ Dx Software and other files from runs on a Genexus™ Dx Purification System or Genexus™ Dx Integrated Sequencer.

You can access customer support archive files from the **Results / Sample Results** and the **Results / Run Results** screens.

### Download a Customer Support Archive

A Customer Support Archive (CSA) file is a downloadable archive that a support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Dx Software. The archive contains log files and other technical data about the software and other files from sequencing runs.

CSA files are generated for sequencing runs in which the CustomerSupportArchive plugin is run. The plugin can be run automatically during the sequencing run, or manually after a sequencing run is complete.

1. Click **Results►Run Results**, then click a run name of interest.
2. In the **Run Results** screen, click ••• **(More Options)►CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to your computer and is ready to be shared with a support representative.

Your support representative might recommend one of the alternative ways to download a CSA file.

- In the **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.
- In the **Sample Results** screen, click a sample name, then from any tab in the **Results** screen, click ••• **(More Options)►CSA**.
- Click **Results►Run Results**, then place the pointer over the row of the run result of interest, then click **CSA**.

# Genexus™ Dx Purification System

## General troubleshooting

| Observation | Possible cause | Recommended action |
|---|---|---|
| Run Fails | Instrument detected an error or user aborted the run. | 1. Record the error message displayed on the instrument touchscreen.<br>2. Export the Genexus™ Dx Purification System CSA file from the Run Report.<br>3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting.<br>Note: Depending on the point during the purification run that the abort occurred, the consumables and reagents may not be suitable for reuse.<br>4. Restart the run beginning from “Prepare samples”. If the run fails again, contact Technical Support to troubleshoot the problem. |
| Quantitation failure during purification run | Instrument detected an error, or user aborted the run. | 1. Record the error message displayed on the instrument touchscreen.<br>2. Export the Genexus™ Dx Purification System CSA file from the Run Report.<br>3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting.<br>4. Manually quantify samples present in the 48-well archive plate (reference archive plate maps for FFPE) using the Qubit™ Fluorometer, or equivalent fluorometric method. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Unexpected sample concentration (negative or greater than normal concentration values for multiple samples) | • Erroneous standard measurements were made.<br>• Pipetting error occurred during liquid transfers.<br>• Tip blockage occurred during liquid transfer.<br>• Bubble in Genexus™ Dx 48-Well Nucleic Acid Archive Plate sample well. | Manually quantify samples present in the 48-well archive plate (reference archive plate maps for FFPE) using the Qubit™ Fluorometer or equivalent fluorometric method. |
| Magnetic beads in Genexus™ Dx 48-Well Nucleic Acid Archive Plate or output plate wells | Carry over of magnetic beads due to inefficient elution. | This can lead to lower observed quantitation value but may not affect sequencing. |
| | | Separate the eluate from beads using a magnetic holder or plate. If the eluate can be separated from the beads, proceed with sequencing. Otherwise, re-extract the sample. |
| | Proteinase K digestion was inefficient due to suboptimal sample preprocessing. | Follow the guidelines and procedures for sample extraction and use of the AutoLys M Tube. |
| | Proteinase K digestion was inefficient due to sample overload. | Ensure that the recommended tissue input is used, and tissue sections do not include excessive amounts of paraffin. |

## Genexus™ Dx FFPE DNA and RNA Purification

| Observation | Possible cause | Recommended action |
|---|---|---|
| Lower than expected yield | Protease digestion incubation temperatures were below 60°C and 90°C. | Use a calibrated thermometer to verify incubation oven temperature. Reset oven temperature, if needed. |
| | | Ensure that incubators have reached target temperature before starting incubation. |
| | | Increase the duration of incubation at 60°C by up to 1 hour. |
| | Restricted airflow limited the heat transfer to the sample tubes during incubation. | Incubate samples in a 4-way rack. Do not use a rack with a solid bottom. |
| | The volume of DNase or Proteinase K solution added was not correct. | Ensure that the correct volume of 1X DNA Digestion Master Mix, Genexus™ Dx FFPE Protease Buffer, and Genexus™ Dx Proteinase K solution is added. |
| | Lysate recovery from AutoLys M Tube was low. | Ensure that AutoLys M Tube caps are securely closed before incubation. |
| | | Allow AutoLys M Tube to cool to room temperature before centrifugation. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Lower than expected yield *(continued)* | AutoLys M Tube collapsed during centrifugation. | Ensure that AutoLys M Tube caps are securely held by the AutoLys M Tube Locking Lid before starting centrifugation. |
| | The start of the purification run was delayed after preparing the Genexus™ Dx FFPE DNA and RNA Purification Plate 1. | Start the purification run within 15 minutes after addition of samples. |
| | The sample input amount was not correct or insufficient, or the amount of material in the sample was insufficient. | Use the recommended tissue input amount. See "Recommended input amount" on page 73. |
| | | Increase the tissue input if needed as follows:<br>• FFPE tumor resections—up to 14 × 5-µm curls/slides, or equivalent amount<br>• FFPE FNA or CNB—up to 14 × 5-µm curls/slides, or equivalent amount |
| | Sample quantitation was not correct. | Avoid prolonged exposure of the Genexus™ Dx Quantitation Plate to light. |
| | | Equilibrate the Genexus™ Dx Quantitation Plate to room temperature for at least 30 minutes before starting the purification run. |
| | | Ensure that reagents have been brought to room temperature. |
| | | Avoid introducing bubbles into samples. |
| | Loss of sample occurred during preprocessing (deparaffinization). | Follow FFPE sample preparation with AutoLys M Tube protocol closely. |
| Unexpected sample concentration (negative or greater than normal concentration values for multiple samples)<br>Details: FFPE purification run | No eluate present in the 48-well archive plate. | 1. Check the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 row G for any remaining DNA eluate and Genexus™ Dx FFPE DNA and RNA Purification Plate 2 row B for any remaining RNA eluate. Order of samples in the 96-well plate is similar to the sample order in the run plan. If eluate is present, transfer to the 48-well archive plate according to the plate map (reference archive plate maps for FFPE).<br>2. Manually quantify samples present in the 48-well archive plate (reference archive plate maps for FFPE) using the Qubit™ Fluorometer or equivalent fluorometric method. |
| Cloudy samples in some Genexus™ Dx 48-Well Nucleic Acid Archive Plate and output plate wells | Deparaffinization was incomplete. | Follow FFPE sample preparation with AutoLys M Tube protocol closely. This situation may not affect sequencing. |

# Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls

You can use quality control results to troubleshoot Genexus™ Dx Integrated Sequencer runs to help identify the cause of performance problems. Inclusion of a set of six control amplicons (covering a range of amplicon length) and spike-in nucleic acid into sample library preparation reactions helps determine whether poor performance is due to insufficient sample input and/or poor sample quality, or is unrelated to sample input and quality. With 10 ng sample input, the read ratio of endogenous sample reads to spike-in control reads is expected to be ~1–3. Using more than 10 ng sample input results in a proportionally higher read ratio. For example, if you load 20 ng of sample, the read ratio should be ~2–6.

The CF-1 templating control serves as a check on templating and sequencing performance that is independent of library preparation.

Use the following table as a guide to help identify the source of performance problems. For recommended actions, see the troubleshooting topics under "Genexus™ Dx Integrated Sequencer—general and QC troubleshooting" on page 169.

| QC category | Run diagnostic | | | |
|---|---|---|---|---|
| | Successful run | Sample input and/or quality problem | Library preparation problem unrelated to sample | Templating or Sequencing problem |
| Sample QC (endogenous sample reads) | Passed | Failed | Failed | Failed |
| Read ratio for inline controls (endogenous to spike-in reads) | Normal<br>Read ratio ~1–3 | Low<br>Read ratio <<1–3 | Normal or variable | — |
| Templating Control QC - CF-1 | Passed | Passed | Passed | Failed |

# Genexus™ Dx Integrated Sequencer—general and QC troubleshooting

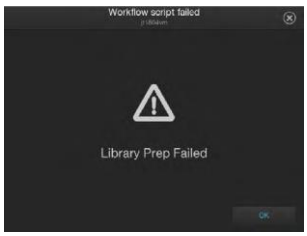
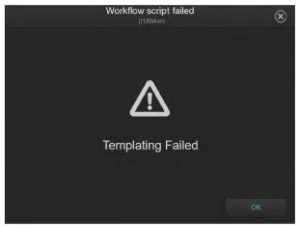
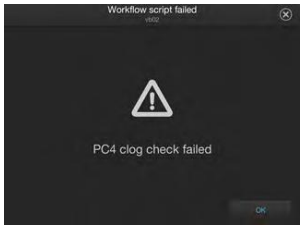
| Observation | Possible cause | Recommended action |
|---|---|---|
| A consumable is not recognized by the sequencer after loading on the deck | The consumable (for example, a strip, pipette tip box) is correctly placed but is not completely inserted into its position, causing it to be misaligned with its expected position. | Ensure that the consumable is pressed completely into place. Apply firm pressure on the item so that it fits snugly into its deck position. |
| | Consumable version does not match the Genexus™ Dx Software version. For example, a consumable compatible with RUO mode is installed in a sequencer running in IVD mode. | Ensure that you are using consumables compatible with the software mode running on the sequencer. |
| | The expiration date of the consumable has passed. | Ensure that you load consumables that have not exceeded their expiration dates. |
| | The instrument camera is not functioning properly. | If no other cause can be found for the issue, there may be a problem with the camera. Contact Technical Support. |
| Run Status = Failed<br><br>Details: In the Genexus™ Software **Run Result** screen, the **Run Status** for a completed run is listed as "**Failed**". In the **Sample Results** screen, the **Sample Status** is listed as "**BaseCallingActor FAILED**". | Chip calibration failed due to a chip problem, or an instrument problem. | Repeat the run with a new chip. If the problem persists, contact Technical Support. |
| A lane that has been used is not crossed out in the sequencer screen<br><br>Details: After completion of a run, the lane used in the run was not crossed out, so that the next run could reuse the lane. | A chip problem caused a datacollect failure to read efuse. | In the sequencer screen, tap **Settings►Clean instrument** to perform a clean instrument. For details, see “Perform a Clean instrument procedure” on page 194. After cleaning, start a new run.<br><br>Note: The **Clean instrument** procedure renders remaining sequencing reagents and unused lanes on the installed chip unusable in a sequencing run after the cleaning. |

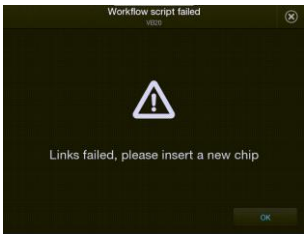
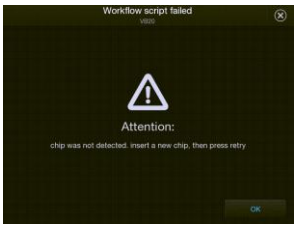
| Observation | Possible cause | Recommended action |
|---|---|---|
| The number of sample reads is low, CF-1 metrics pass QC, but read ratio of inline controls is low<br><br>Details: If CF-1 reads per lane, accuracy, and mean AQ20 read length are good, and read ratio of inline controls (endogenous vs. spike-in) is low (<< 1–3), a problem with sample input is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 168. | Nucleic acid input may have been insufficient, and/or the nucleic acid was degraded. | For a sample run, requantify nucleic acid samples and/or perform sample QC to ensure that the expected nucleic acid input and size was loaded. |
| | | If needed, reisolate and purify nucleic acid samples. |
| The number of sample reads is low, but CF-1 metrics pass QC, and read ratio of inline controls is normal<br><br>Details: If CF-1 metrics passed QC, and read ratio of inline controls is normal (~1–3), a problem in library preparation unrelated to sample input or quality may be indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 168. | One or more of the Genexus™ Dx Strip 1 strips used in the run had magnetic beads trapped in the tube 5 keyhole. | Repeat the run with strips that you have verified have no trapped beads. For more information, see "Before you begin" on page 101. |
| | An incorrect assay was selected for the run, or library amplification parameters were not optimal. | Ensure that you have selected the correct assay and reviewed assay parameters. |
| | Library strips were inadequately equilibrated to room temperature (Genexus™ Dx Strip 1), or incompletely thawed (Genexus™ Dx Strip 2-HD) before loading in the sequencer. | Ensure that Genexus™ Dx Strip 1 strips are fully equilibrated to room temperature, and Genexus™ Dx Strip 2-HD strips are completely thawed before loading in the sequencer. |
| The number of sample reads is low, and CF-1 metrics fail QC<br><br>Details: If CF-1 metrics failed QC, a problem in templating or sequencing is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 168. | One or more of the Genexus™ Dx Strip 3-GX5™ strips used in the run may have had an excessive amount of magnetic beads trapped in the tube 6 or 7 keyhole. | Repeat the run with strips that you have verified have no trapped beads. For more information, see "Before you begin" on page 101. |

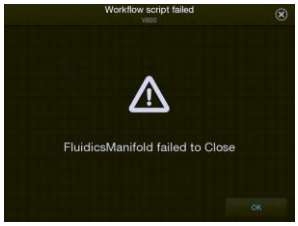
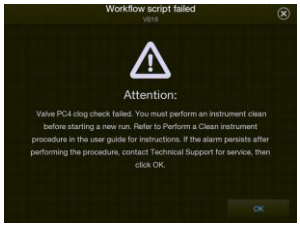
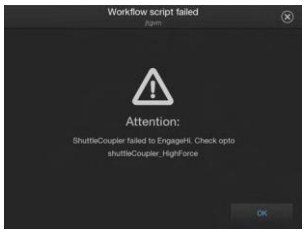
| Observation | Possible cause | Recommended action |
|---|---|---|
| The number of sample reads is low, and CF-1 metrics fail QC<br><br>Details: If CF-1 metrics failed QC, a problem in templating or sequencing is indicated. For more information, see "Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls" on page 168.<br>*(continued)* | Template strips were inadequately equilibrated to room temperature (Genexus™ Dx Strip 3-GX5™), or incompletely thawed (Genexus™ Dx Strip 4) before loading in the sequencer. | Ensure that Genexus™ Dx Strip 3-GX5™ strips are fully equilibrated to room temperature, and Genexus™ Dx Strip 4 strips are completely thawed before loading in the sequencer. |
| | The sequencing chip or coupler was defective or leaky. | Repeat the run with new chip and coupler. If low performance continues, contact Technical Support. |
| | The run was started >14 days after the last initialization was performed, or on an expired initialization. | Perform a Clean instrument procedure (**Settings►Clean instrument**). For more information, see "Perform a Clean instrument procedure" on page 194. After the Clean instrument procedure, install new a chip, and new sequencing reagent bottles and cartridge in the sequencing reagents bay, then repeat the run.<br><br>Note: Reagents are stable on the sequencer for 14 days, after which you may experience decreased performance. For more information, see "Options for an expired sequencer initialization" on page 114. |

<table>
<tr><th>Observation</th><th>Possible cause</th><th>Recommended action</th></tr>
<tr><td>CF-1 AQ20 Mean Read Length QC fails, but positive control/NTC QC passes<br><br>Details: Alternatively, CF-1 AQ20 Mean Read Length QC fails, or is borderline passing, and one or more positive control metrics fail (for example, DNA Control AQ20 Mean Read Length, and DNA or RNA mapped reads).</td><td>Reads were filtered out due to increased low quality ISPs caused by an excess of templated beads generated in the templating reaction.</td><td>Recover the unused pooled sample library from the run, then plan and perform a Library to Result run with diluted library.<br><br>1. Follow the procedure described in “Recover the pooled sample libraries” on page 220 to recover the pooled sample libraries from well E4 of the PCR amplification plate after the run.<br>2. For the number of chip lanes used in the original run, add the volume of recovered pooled libraries and nuclease-free water or Low TE indicated in the following table to well A1 of the sample plate.<br><br><table><tr><th></th><th></th><th></th><th></th></tr><tr><td>1</td><td>17.5 µL</td><td>32.5 µL</td><td>50 µL</td></tr><tr><td>2</td><td>28 µL</td><td>52 µL</td><td>80 µL</td></tr><tr><td>3</td><td>38.5 µL</td><td>71.5 µL</td><td>110 µL</td></tr><tr><td>4</td><td>49 µL</td><td>91 µL</td><td>140 µL</td></tr></table><br>3. Pipet the contents of well A1 up and down 4–5 times to mix.<br>4. Plan and perform a Library to Result run with the diluted library.<br><br>For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 217.</td></tr>
<tr><td>Some sample QC categories show N/A, and the Sample Status in the Sample Results screen shows "AnnotatorActor: FAILED"<br><br>Details: Reanalysis does not rescue the sample.</td><td>A rare error occurred in the variant caller and caused the variant caller and downstream annotator actor to fail.</td><td>Repeat sequencing with the same nucleic acid sample (Nucleic Acid to Result run).<br><br>Alternatively, if sufficient sample library has been recovered from the PCR amplification plate after the run, the operator can repeat sequencing from the same sample library (Library to Result run). For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 217.</td></tr>
<tr><td>Pipette tips remain in the used tip rack</td><td>Tips were unable to be picked up by the pipettor.</td><td>In Genexus™ Dx Software 6.6 and later, if the pipettor cannot pick up a tip, it leaves the problematic tip in its place and moves to the next available tip. No action is needed.</td></tr>
</table>

# Genexus™ Dx Integrated Sequencer error and warning messages

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error message: Library Prep Failed<br>  | • The sequencer failed xy homing, *or*<br>• The sequencing chip failed to engage at pipette position. | Confirm that no analyses are in progress, then reboot the instrument. Repeat the run. If the failure continues, contact Technical Support. |
| Error message: Templating Failed<br>  | • The sequencer failed xy homing, *or*<br>• The Genexus™ Coupler was misaligned. | 1. Confirm that no analyses are in progress, then reboot the instrument. Repeat the run, and ensure that the coupler is installed with proper alignment. If the failure continues, contact Technical Support.<br>2. If the volume of leftover pooled library in well E4 of the PCR amplification plate is sufficient, perform a Library to Result run using the leftover pooled library. For more information, see "Recover pooled sample libraries and plan a Library to Result run" on page 217.<br>Note: Diluting leftover pooled library more than 2-fold can result in low reads. |
| Error message: Initialization Failed: XXX did not reach fill<br>  | Genexus™ Bottle 2 flow was restricted, or a flow sensor malfunctioned.<br>OR<br>During a post-chip clean, the Genexus™ Bottle 3 was empty, or Genexus™ Bottle 3 flow was restricted.<br>Note: XXX is the name of the conical tube that did not reach fill. | Allow the run to complete, then select **Settings►Clean instrument** to do a clean. After cleaning completes, start a new run. If the error message appears again, contact Technical Support for service.<br>Note: After the run completes, you can recover pooled libraries from the amplification plate, if desired. For more information, see "Recover pooled sample libraries and plan a Library to Result run" on page 217. |
| Error message: XXX clog check failed<br>  | Clog check following Clean Instrument failed. Incomplete fluid flush or debris has caused a clog.<br>Note: XXX refers to the failed waste lines, chip, or main 1-4. | Contact Technical Support for recommended action. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error message: Flow rate from W3 failed – below 50 µL/s<br>  | During the post-chip clean the Genexus™ Dx Bottle 3 was empty, or Genexus™ Dx Bottle 3 flow was restricted. | Select **Settings►Clean instrument** to do a clean. Ensure that the Genexus™ Dx Bottle 3 is installed correctly, and the bottle contains sufficient volume. Replace as needed. For details, see “Perform a Clean instrument procedure” on page 194. If the failure persists, contact Technical Support. |
| Error message: PostChipClean failed | The post-chip clean aborted due to a chip error. | In the sequencer screen, tap **Settings►Clean instrument** to re-do a Clean instrument procedure. For details, see “Perform a Clean instrument procedure” on page 194. You may or may not need to replace the chip during deck set up. |
| Error message: Links failed, please insert a new chip<br>  | A chip failed to come up when shuttled to the sequencing position during run setup. | Replace the chip, then try to proceed again. If the error message appears again, contact Technical Support. |
| Error message: Chip was not detected. Insert a new chip, then press retry<br>  | Chip connection was lost and sequencing failed. | 1. Insert a new chip, then press **Retry**.<br>2. If this does not resolve the problem, remove the chip from the instrument. An instrument clean is required before a new run can proceed.<br>3. Force an instrument clean by navigating to **Settings►Clean instrument**.<br>4. After the clean, repeat the run with a new chip and reagents. |
| Error message: Vacuum Clean Failed: Instrument Service might be required | XY motion failed, OR a leak was detected in the vacuum system. | Contact Technical Support for recommended action. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Error Message: FluidicsManifold failed to close<br>  | The sequencing clamp hardware did not operate properly. | Instrument service is required to proceed.<br>Note: Pooled libraries can be recovered if the error occurred after library preparation was complete. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 217. |
| Error message: Valve PC4 clog check failed<br>  | A clog was detected during the post-chip clean. | 1. Perform an instrument clean by navigating to **Settings►Clean instrument**.<br>2. After cleaning completes, begin a new run.<br>3. If the error message appears again, contact Technical Support for service. |
| Error message: ShuttleCoupler failed to EngageHi<br>  | Chip shuttle hardware did not operate properly. | Instrument service is required to proceed.<br>Note: Pooled libraries can be recovered if the error occurred after library preparation was complete. For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 217. |
| Warning message: Pressure Leak Detected in Vac: vac pressure drop leakrate<br>**Press Ok to open doors and fix the leak.** | Genexus™ Dx Bottle 1 leak was detected. | Remove the Genexus™ Dx Bottle 1 and ensure that the cap is tight, then replace the bottle securely on the instrument. Continue with the run. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Vac: vac time to pressure<br>**Press Ok to open doors and fix the leak.** | A Genexus™ Dx Bottle 1 leak or over-fill was detected. | Ensure that a new Genexus™ Dx Bottle 1 is installed, and the cap is tight and the bottle is secure on the instrument. Continue with the run. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Vac: vac pressure baseline<br>**Press Ok to open doors and fix the leak.** | Genexus™ Dx Bottle 1 leak was detected. | Remove the Genexus™ Dx Bottle 1 and ensure that the cap is tight, then replace the bottle securely on the instrument. Continue with the run. If the pressure leak message appears again, replace Genexus™ Dx Bottle 1 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Warning message: Pressure Leak Detected in Seq: W2P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 2 at the left position. | Remove Genexus™ Dx Bottle 2 installed at the left position, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run. If the pressure leak message is seen again, replace the left Genexus™ Dx Bottle 2 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: W22P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 2 installed at the right position. | Remove the Genexus™ Dx Bottle 2 installed at the right position, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run. If the pressure leak message appears again, replace the right Genexus™ Dx Bottle 2 with a new bottle, then continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: W3P **Press Ok to open doors and fix the leak.** | A leak was detected in the Genexus™ Dx Bottle 3. | Remove the Genexus™ Dx Bottle 3, ensure that the cap is tight, then reinstall the bottle securely. Continue with the run, or repeat the clean if this message appears in a post-chip clean. If the pressure leak message appears again, replace the Genexus™ Dx Bottle 3 with a new bottle. Continue with the run, or repeat the clean if this message appears in a post-chip clean. If the failure persists, contact Technical Support. |
| Warning message: Pressure Leak Detected in Seq: Conicals & Reagent Cartridge **Press Ok to open doors and fix the leak.** | A leak was detected in one or more of the Genexus™ Dx Conical Bottles or the Genexus™ Dx Cartridge. | Tighten all Genexus™ Dx Conical Bottles one bottle at a time to prevent accidental position changes. Inspect the Genexus™ Dx Cartridge for defects. Continue with the run. If the failure persists, contact Technical Support. |
| Warning message: Liquid detected in conicals **Press Ok to open doors and fix the issue.** | The previous post-chip clean did not remove all of the liquid in the Genexus™ Dx Conical Bottles, causing failure of the Conicals empty test and Conical volume test. | Replace the Genexus™ Dx Conical Bottles with new bottles, then tap **Settings►Clean instrument** to clean the instrument. After cleaning, start a new run. |

# Genexus™ Dx Software

| Observation | Possible cause | Recommended action |
|---|---|---|
| Cannot sign in to Genexus™ Dx Software | You either entered an incorrect password or you are signed out due to several failed login attempts. | Contact the Genexus™ Dx Software system administrator. After 3 failed sign in attempts, the user is locked out for 1 hour. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Batch sample import fails | One or more entries in the sample-import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the sample-import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample-import template file, and be careful to copy only those rows that contain actual data. |
| | The sample import spreadsheet contains a nonunique sample name. | Every sample name in the software must be unique. Ensure that the spreadsheet does not contain any duplicate sample names, then repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1. |
| | The headings in the sample import spreadsheet do not match the sample attributes in the software. | The headings must match the sample attributes in the software exactly. Check the headings for spelling or other errors. |
| Library batch import fails | One or more entries in the library batch import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the library batch import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template file, and be careful to copy only those rows that contain actual data. |
| | The library batch import spreadsheet contains a nonunique **Library Batch ID**. | Each **Library Batch ID** in the software must be unique. Ensure that the spreadsheet does not contain any duplicate IDs, then repeat the import. The system check is not case-sensitive. For example, a **Library Batch ID** of ABC1 conflicts with abc1. |
| | A sample name entered in the library batch import spreadsheet does not match a sample name listed in the **Manage Samples** screen. | Ensure that the sample names entered into the spreadsheet are correct and match an existing sample name added to the software. |
| | The Barcode ID name format does not exactly match the format that is used in the **Prepare Library Batch** dialog box. | Use the name format following the Barcode ID name format found in the Barcode Set reference lists (**Settings►References►Barcode Set**), for example: IonDual_0101 through IonDual_0196, or IonHDdual_0101 to IonHDdual_0132. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Library batch import fails *(continued)* | An invalid library, control, or panel kit barcode has been entered in the spreadsheet. | Ensure that you have correctly entered a valid kit barcode in the appropriate cell of the spreadsheet. |
| The results of the run do not appear in the Results / Run Results screen | The instrument disk space is full. | Clear disk space on the sequencer. For more information, see "Manually delete run data" on page 204. |
| Cannot download run result files | The run failed. Links to run files are not available for runs that fail QC. | Repeat the run. |

# B Touchscreen reference


- Genexus™ Dx Purification System ..... 179
- Genexus™ Dx Integrated Sequencer ..... 190


## Genexus™ Dx Purification System

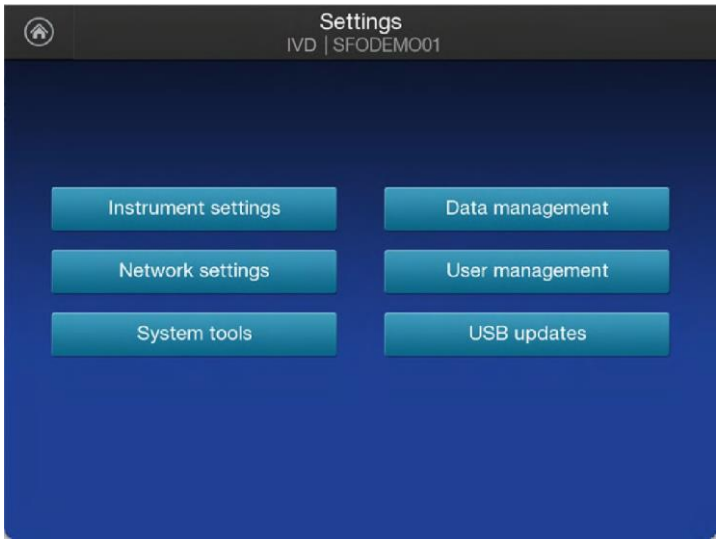
### Touchscreen icons




| Number | Icon | Description |
|---|---|---|
| 1 | | User profile |
| 2 | | USB – available |
| | | USB – not available |
| 3 | | Network connectivity – connected |
| | | Network connectivity – not connected |

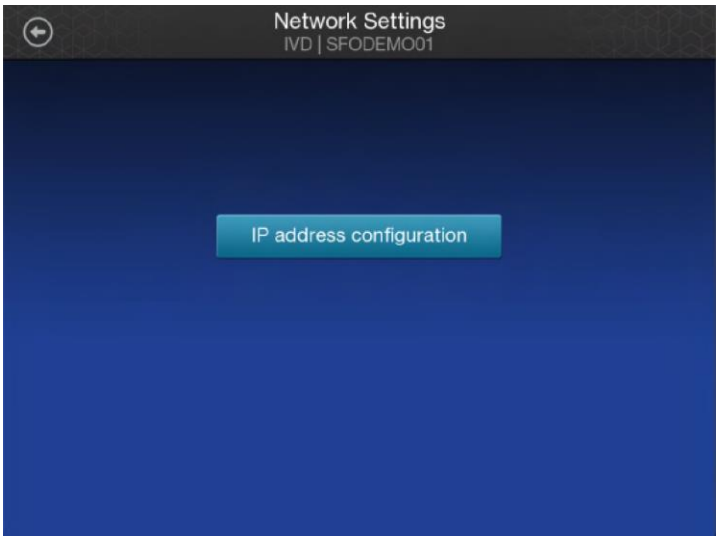
# Settings

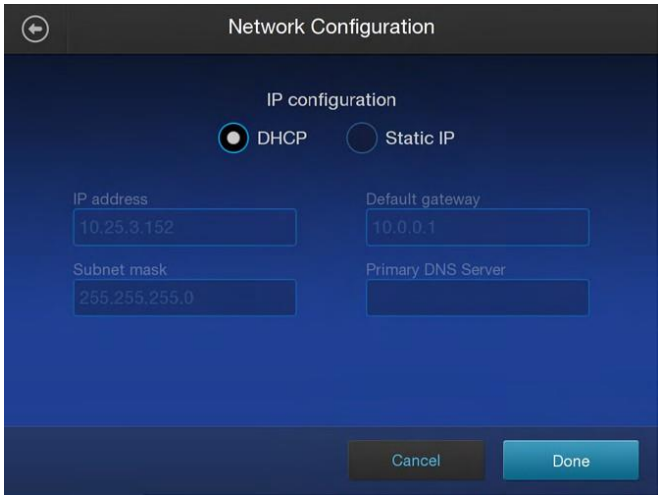
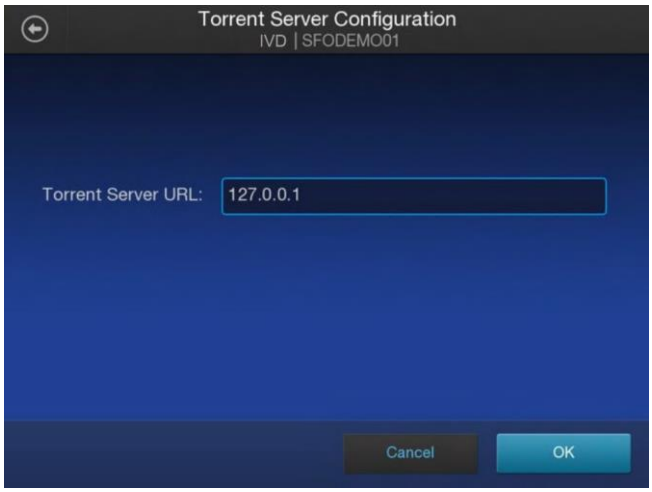
Use the **Settings** menu to view and change instrument settings, and to manage data.




## Network Settings

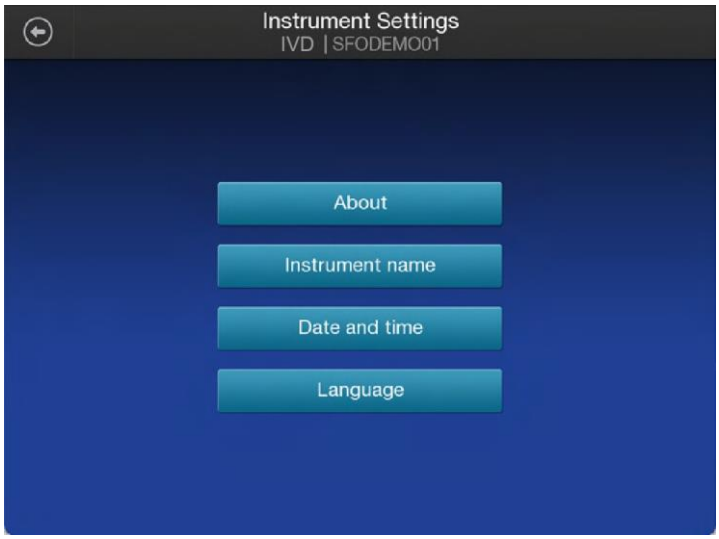
The **Network Settings** menu enables you to configure IP address and Ion Torrent™ Server settings, and update the server.

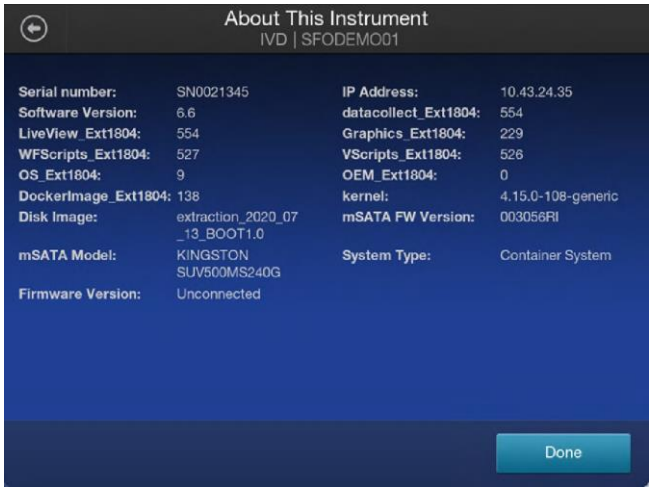


| Item | Description | When/How to use |
|---|---|---|
| IP Address Configuration | Enables users to set or change the IP configuration (DHCP or Static IP).<br>  | 1. Select either the DHCP or Static IP option.<br>2. Touch the screen in the field you want to edit to activate the field. A virtual keyboard appears.<br>3. Enter the new information, then press Done. |
| Ion Torrent™ Server Configuration | Enables users to change the Ion Torrent™ Server IP address and user information.<br>  <br>Enter the following IP address in the Torrent Server URL field for your instrument: 127.0.0.1 | When a change to the Ion Torrent™ Server IP address or user information is needed.<br>Note: Do not change address unless directed to do so by a Thermo Fisher Scientific field service engineer. |

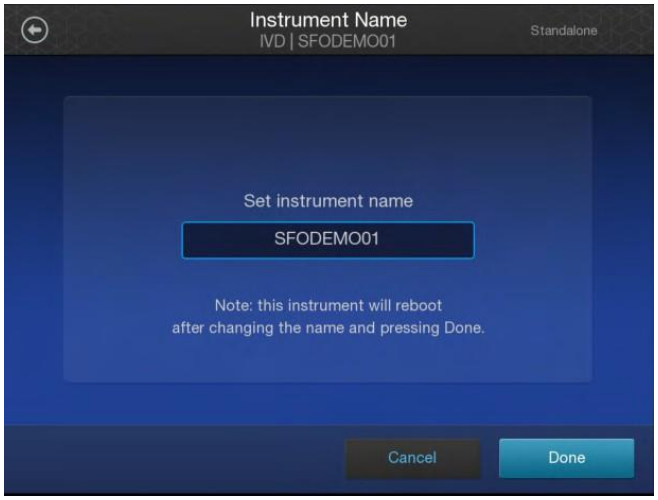
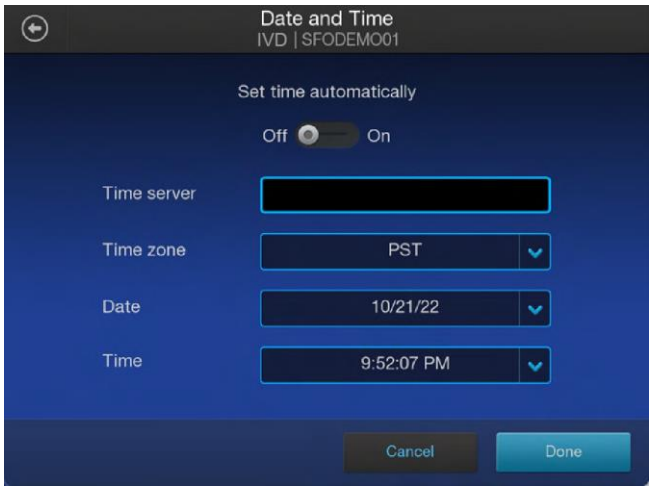
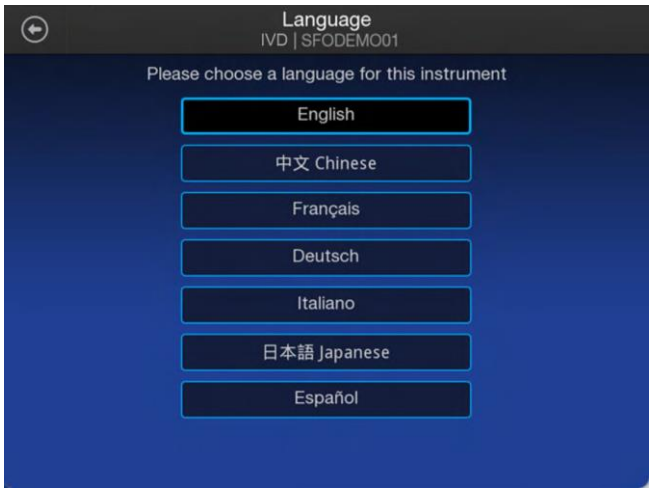
## Instrument settings

Use the **Instrument Settings** menu to obtain information about the instrument, to set the instrument name, date and time, and language.



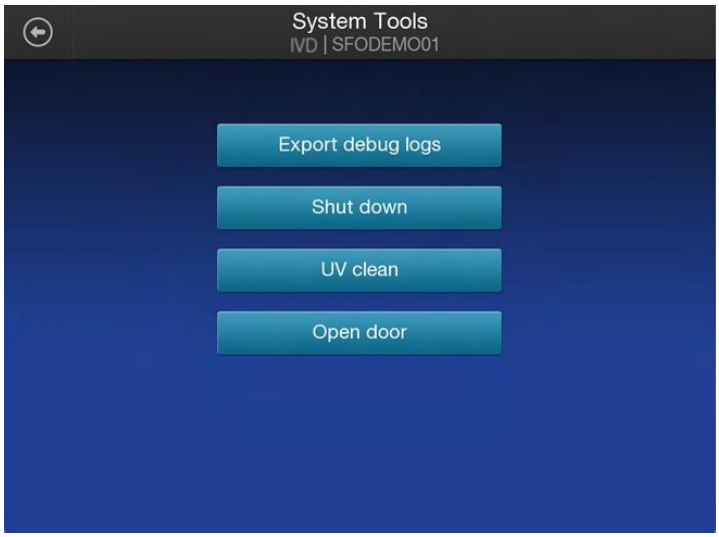

| Item | Description | When and how to use |
|---|---|---|
| About | Provides instrument details.<br>  | Use to view instrument reference information and performance qualification status. |

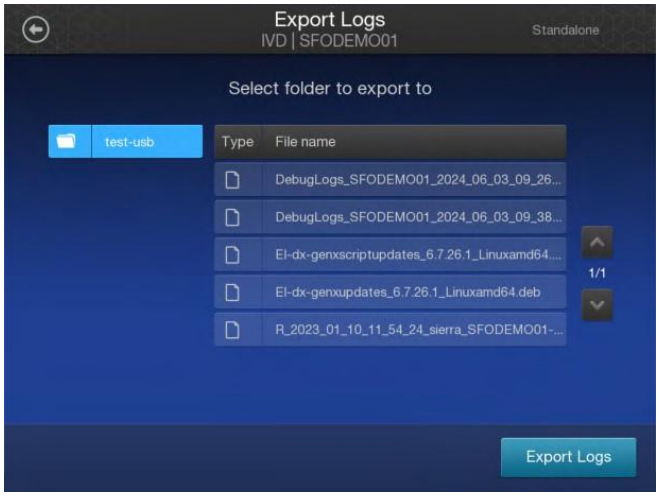
*(continued)*

| Item | Description | When and how to use |
|---|---|---|
| Instrument Name |  | Use to change the instrument name. |
| Date and time |  | The Time server is set by the field service engineer during installation. Enter the desired Time zone, Date, and Time to manually set the date and time.<br><br>IMPORTANT! Do not leave the Time server field blank. This will cause the instrument to stop responding and require a reboot to restore functionality. |
| Language | Lists available user interface languages.<br>  | If changing the language. Tap to select a language, then tap OK. The instrument will reboot in the selected language. |

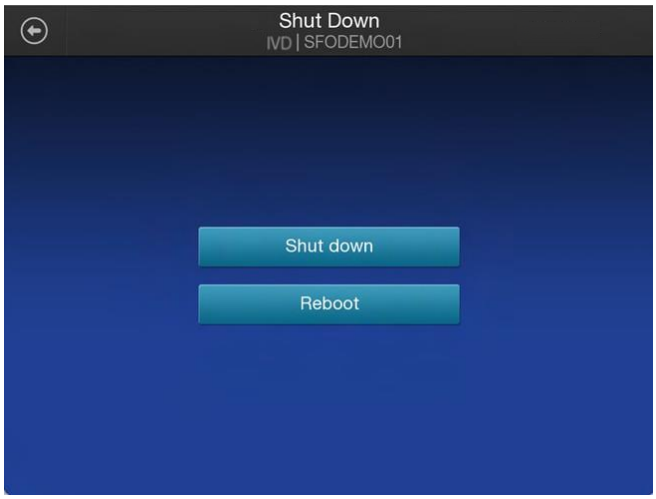
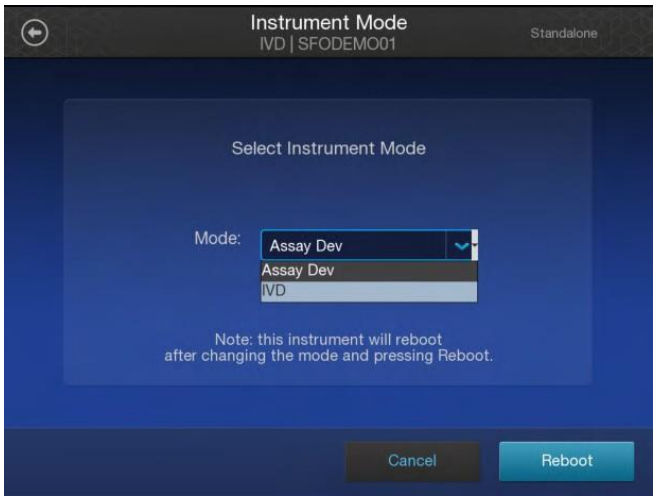
## System Tools

Use the **System Tools** menu to upload instrument diagnostics, manage data, and shut down or reboot the instrument.



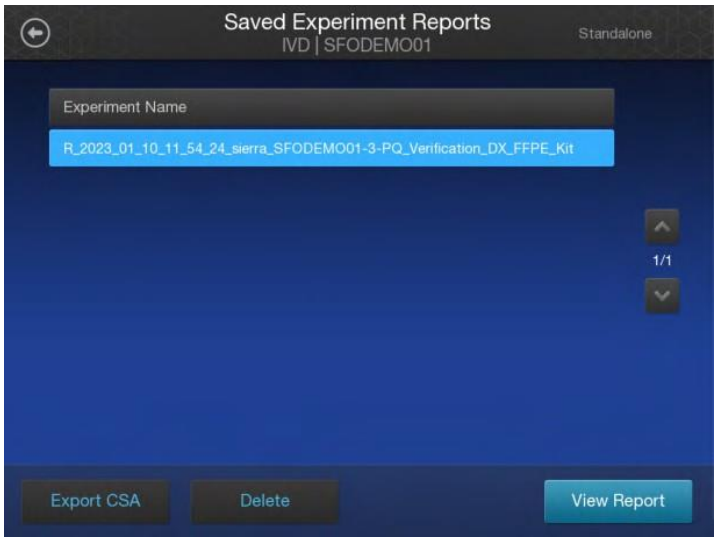

| Item | Description | When and how to use |
|---|---|---|
| Export debug logs | Provides instrument error configuration information.<br>  | Use for troubleshooting if directed to do so by Technical Support.<br>1. Tap ⚙ (Settings)►System Tools►Export debug logs.<br>2. In the Export Logs screen, tap the folder to export to.<br>3. From the file list, select a file to export.<br>4. Insert a USB drive into the USB port on the front of the instrument, then tap Export Logs.<br>5. When prompted, tap Continue.<br>6. When the export is complete, tap Done. |

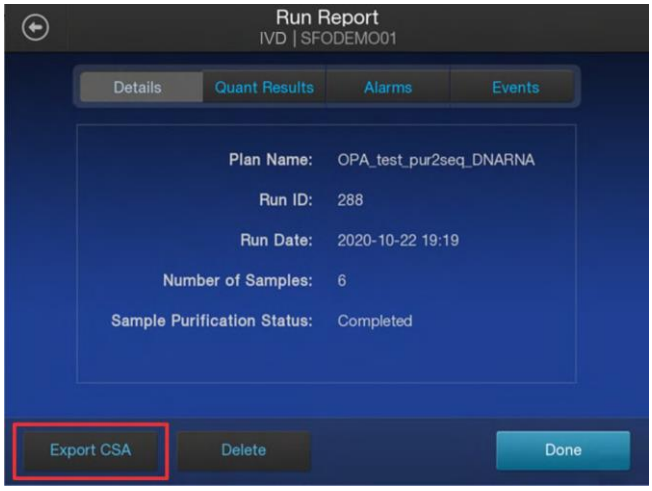
*(continued)*

| Item | Description | When and how to use |
|---|---|---|
| Shut down | Provides access to Shut down and Reboot commands.<br>  | Use if directed to do so by Technical Support as part of a troubleshooting procedure, or if the instrument will not be used for more than 3 days.<br>It is not necessary or recommended to shut down the instrument overnight or over the weekend. To power on the instrument after a shut down, see “Power on” on page 34. |
| UV clean | Irradiates the instrument deck with ultraviolet (UV) light for 2 minutes. | If a reagent or sample spills on the instrument deck, use to irradiate the instrument deck. See “Clean and decontaminate the Genexus™ Dx Purification System” on page 205. |
| Open door | Unlocks and opens the instrument door. | If a run is aborted, use to open the instrument door to remove consumables in preparation for a subsequent run. |
| Instrument mode | Sets the mode of the instrument to either IVD or Assay Dev (Assay Develoment).<br>  | If changing the mode, select the mode from the dropdown list, then click Reboot. |

## Data management

Use the **Data management** screen view run purification report information, export CSA information, or delete purification run reports.



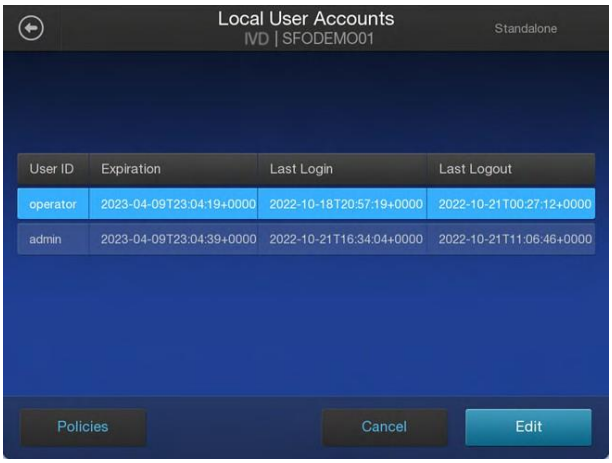
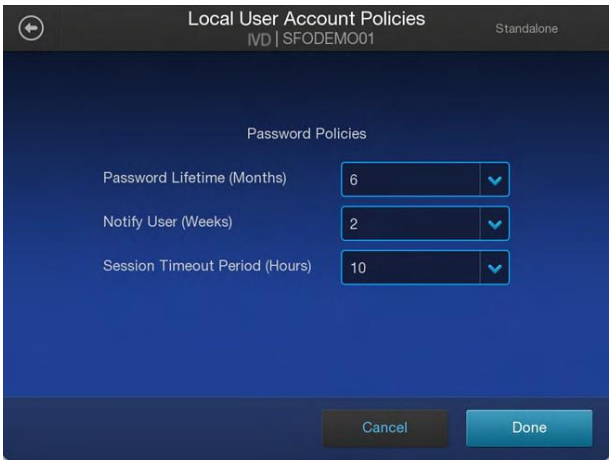

| Item | Description | When and how to use |
|---|---|---|
| View Report | Purification runs that include sample quantitation produce sample concentration results that can be accessed after the run is complete. | To view reports, see “View and export quantitation results” on page 90. |
| Export CSA | Provides instrument error configuration information.<br>  | Use for troubleshooting if directed to do so by Technical Support.<br>1. Tap ⚙ (Settings)►Data management, then tap in the row of the Experiment Name of interest.<br>2. Tap Export CSA<br>3. Insert a USB drive into the USB port on the front of the instrument. Navigate to the file destination on the USB drive, then tap Export CSA.<br>4. When prompted, tap Continue.<br>5. When the export is complete, tap Done. |

*(continued)*

| Item | Description | When and how to use |
|---|---|---|
| Delete | Delete purification run reports that are no longer needed.<br><br>IMPORTANT! You must export purification run reports before you delete them. An error message appears if you attempt to delete a report that has not been exported.To export a run report, see "View and export quantitation results" on page 90. | **1.** Tap ⚙ (Settings)►Data management, then tap in the row of the Experiment Name of interest.<br>**2.** Tap Delete.<br>**3.** When prompted, tap Delete again. |

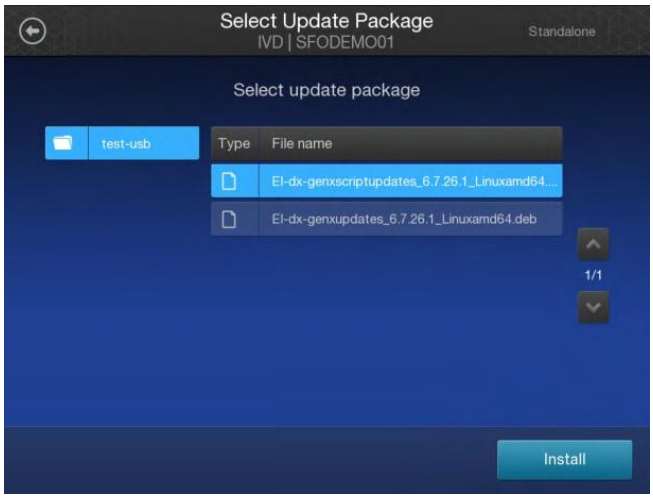
## User management

Administrators use this screen to reset user account passwords and policies.

| Item | Description | When and how to use |
|---|---|---|
| User Accounts |  | **1.** To update a user account password, tap ⚙ (Settings)►User management, then tap in the row of the User ID.<br>**2.** Tap Edit, then enter then new password, then tap Done.<br>**3.** Tap Done, then when prompted, tap Close. |
| Policies |  | **1.** To update password policies, tap ⚙ (Settings)►User management, then tap Policies.<br>**2.** Change the following settings as needed, then tap Done.<br>• Password Lifetime—Set the number of months until a user password expires.<br>• Notify User—Set the number of weeks before a user account password expires that the user receives a notification to reset their password.<br>• Session Timeout Period—Set the number of hours before a session times out and a user must login to their user account again |

## USB updates

Administrators use the **USB update** screen to perform software updates.

| Item | Description | When and how to use |
|---|---|---|
| USB updates | Used to perform software updates.<br>  | Administrators perform software updates as needed.<br>**1.** Insert the USB drive with the software upgrade package into the USB port on the front of the instrument.<br>**2.** Tap ⚙ (Settings)►USB Updates.<br>**3.** Navigate to the file destination on the USB drive, then tap Install.<br>**4.** Follow the prompts on the screen to complete the install. |

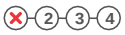
# Genexus™ Dx Integrated Sequencer

## Sequencer touchscreen icons




| Number | Icon | Description |
|---|---|---|
| 1 | | Network connectivity – connected |
| | | Network connectivity – not connected |
| 2 | | Instrument idle |
| | | Sequencing in progress |
| | | Instrument ready |
| | | Error |
| 3 | | Chip status – Absent[1] |
| | | Chip status – Standby |
| | | Chip status – Connecting |
| | | Chip status – Ready |
| | | Chip status – Imaging |

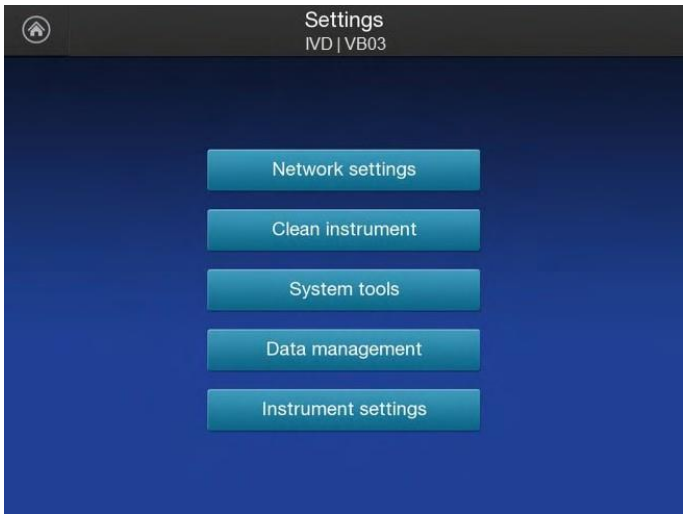
*(continued)*

| Number | Icon | Description |
|---|---|---|
| 3 |  | Chip status – Error |
| 4 |  | Chip lane status – 4 lanes available |
| |  | Chip lane status – 1 lane in use or used, 3 lanes available |
| |  | Chip lane status – 2 lanes in use or used, 2 lanes available |
| |  | Chip lane status – 3 lanes in use or used, 1 lane available |
| |  | Chip lane status – 4 lanes in use or used, 0 lanes available |
| 5 |  | Instrument File System Space – the percent of file space used is indicated. The instrument checks for sufficient disk space before each run, and notifies the operator if there is insufficient disk space for a run.<br>The indicator turns yellow when disk space is ≥67% full. The indicator turns red when ≥90% full. |
| 6 |  | Instrument Server File System Space – the percent of file space used is indicated.<br>If the indicator turns red, archive data from the server to free up disk space. See the software help system for information about archiving data. |

[1] If Chip status is Absent, always insert a new chip.

## Settings

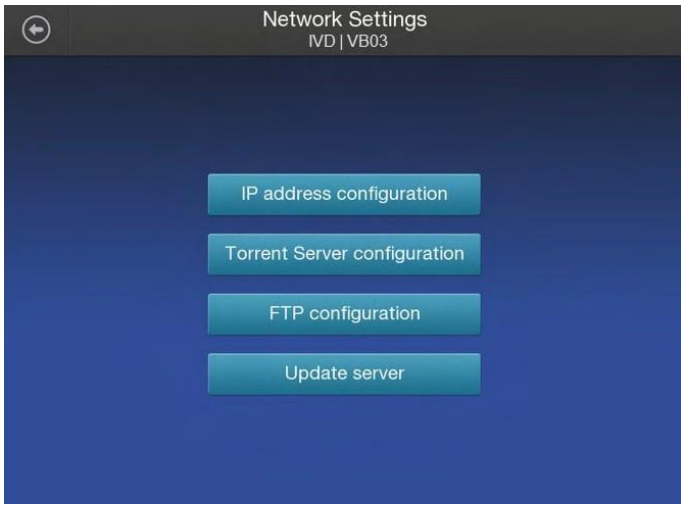
Use the **Settings** menu to view and/or change instrument settings, manage data and network configurations, and clean the instrument.

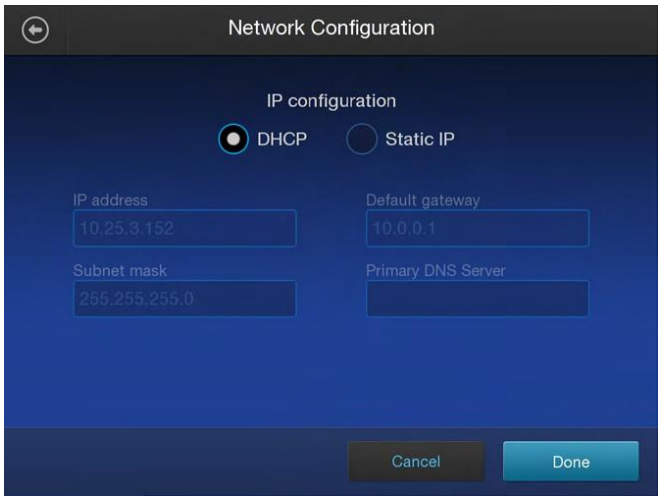



Note: The **System tools** option is for use only by trained service personnel.

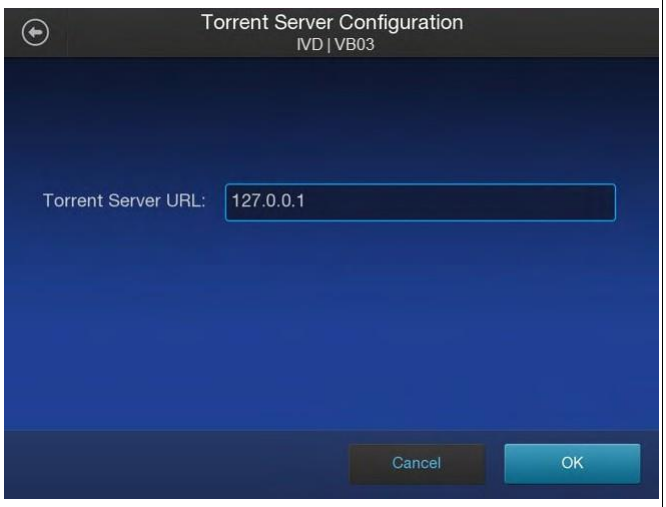
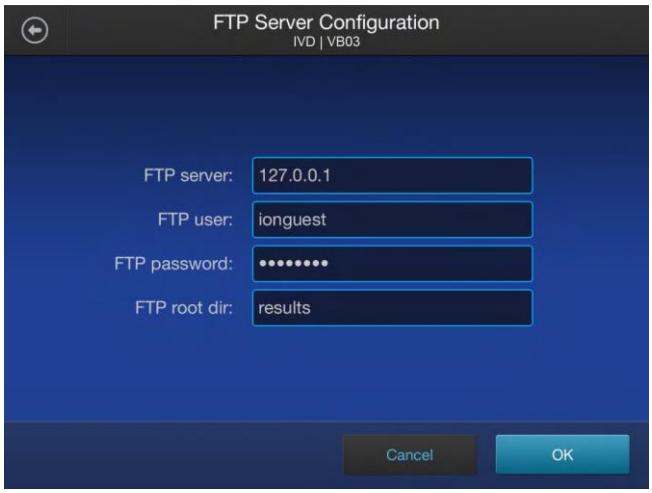
## Network Settings

Use the **Network Settings** menu to configure IP address, Ion Torrent™ Server, and FTP settings.




| Screen | Description | When/How to use |
|---|---|---|
| IP Address Configuration | Set or change the IP configuration (DHCP or Static IP).<br>  | 1. Select either the DHCP or the Static IP option.<br>2. Touch the screen in the field you want to edit to activate the field. A virtual keyboard appears.<br>3. Enter the new information, then press Done. |

*(continued)*

| Screen | Description | When/How to use |
|---|---|---|
| Ion Torrent™ Server Configuration | Change the Ion Torrent™ Server IP address and user information.<br>  <br>Enter the following IP address in the Torrent Server URL field for your instrument: 127.0.0.1 | Use this screen when a change to the Ion Torrent™ Server IP address or user information is required.<br><br>IMPORTANT! Do not change the address unless directed to do so by a Thermo Fisher Scientific field service engineer. |
| FTP Server Configuration | Change the FTP Server IP address and user information.<br>  <br>Enter the following IP address in the FTP server field for your instrument: 127.0.0.1 | Use the screen when a change to the FTP Server IP address or user information is required.<br><br>IMPORTANT! Do not change the address unless directed to do so by a Thermo Fisher Scientific field service engineer. |

## Perform a Clean instrument procedure

UV cleaning is normally performed at the end of a sequencing run. Perform a **Clean instrument** procedure from the **Settings** menu if:

- A sequencing run has been aborted, or a power failure occurred during a run.

  IMPORTANT! If the run stoppage or power failure occurred during the first run on a new chip, contact Technical Support to open a service call.

- The normal post-sequencing run cleaning was not complete.
- The sequencer has been idle for ≥2 weeks after a run that used the remaining lanes or all 4 lanes of a chip.
- The sequencer has been powered on after a long-term shutdown as described in the procedure in “Power on after a long-term shutdown” on page 216.

The **Clean instrument** command starts the following cleaning procedures:

- Vacuum clean—Cleaning of the four chip vacuum lines and robotic waste line.
- Post-chip clean—Cleaning of sequencer fluidic lines with a conical flow rate test.
- UV clean—Irradiation of the deck surface with UV light. For more information, see “Clear the instrument deck and perform a UV Clean” on page 111.

IMPORTANT! The **Clean instrument** procedure renders remaining sequencing reagents and unused lanes on the installed chip unusable in a sequencing run after the cleaning.

The following items are needed if the items are not already loaded in the sequencer:

- New Genexus™ Dx Bottle 3
- Used and empty Genexus™ Dx Bottle 2 (2)
- Used Genexus™ Dx Bottle 1
- Used Genexus™ Dx Cartridge
- Used chip
- Used coupler

IMPORTANT! Keep used sequencing reagent bottles and supplies on hand to perform the **Clean instrument** procedure.

1. In the **Settings** menu, tap **Clean instrument**.

2. Follow the on-screen instructions, then tap **Proceed** to go to the following sequence of screens: **Clear Deck** if there are items to be cleared, **Load Deck**, **Clear Sequencing Reagents**, and **Load Sequencing Reagents**. For more information, see “Clear the instrument deck and perform a UV Clean” on page 111.

   Note: If a sequencing run completes with 1–3 unused lanes remaining on a chip, and the operator had selected **Do Force Clean** during run setup, the sequencer proceeds directly to the **Clean instrument** procedure after the run. The sequencing reagents bay doors remain closed and no prompts appear on the sequencer screen.

3. Follow the on-screen instructions in the **Clear Deck** and **Clear Sequencing Reagents** screens. After sequencing reagents are cleared, tap **Next** to return to the home screen.

Note: The **Clean instrument** procedure takes about 2 hours to complete.

## Replace Genexus™ Dx Conical Bottles during a Clean instrument

If the sequencer detects restricted flow from the Genexus™ Dx Conical Bottles during a **Clean instrument** procedure, the following alert is displayed.



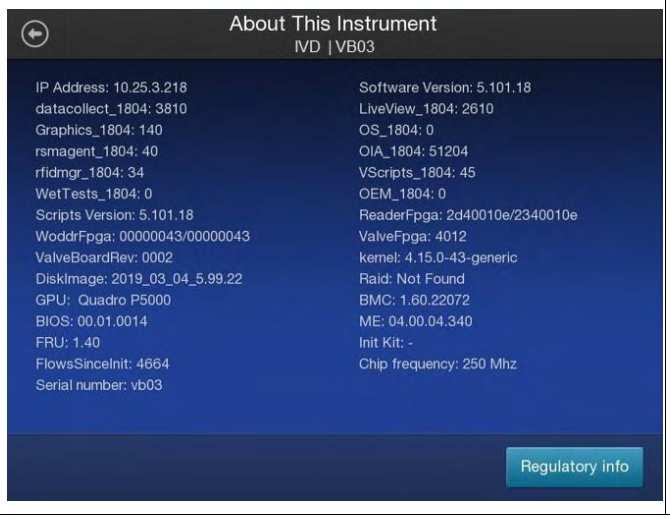

If you see this alert, follow these steps to replace the 5 Genexus™ Dx Conical Bottles.

1. Tap **OK**. The screen returns to the run screen.
2. Tap **Settings►Clean instrument** again.
3. Follow the screen instructions to clear the deck, then load the deck.
4. When the sequencing reagents bay doors open, do the following.
   a. Remove, but do not discard, the Genexus™ Dx Bottle 1, the 2 Genexus™ Dx Bottle 2 bottles, and the Genexus™ Dx Bottle 3 to gain easier access to the Genexus™ Dx Conical Bottles.
   b. Remove the 5 used Genexus™ Dx Conical Bottles, then install 5 new conical bottles.
   c. Replace the 4 reagent bottles that you removed in their original positions.
5. Close the sequencing reagents bay doors.
   The **Clean instrument** procedure begins automatically.
6. After the cleaning finishes, follow the screen instructions to clear the deck and remove the sequencing reagents.
   For further information, see “Replace the Genexus™ Dx Conical Bottles” on page 211.

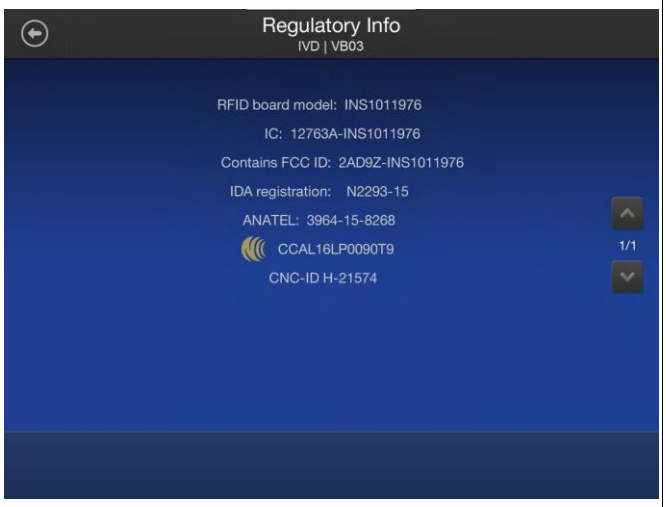
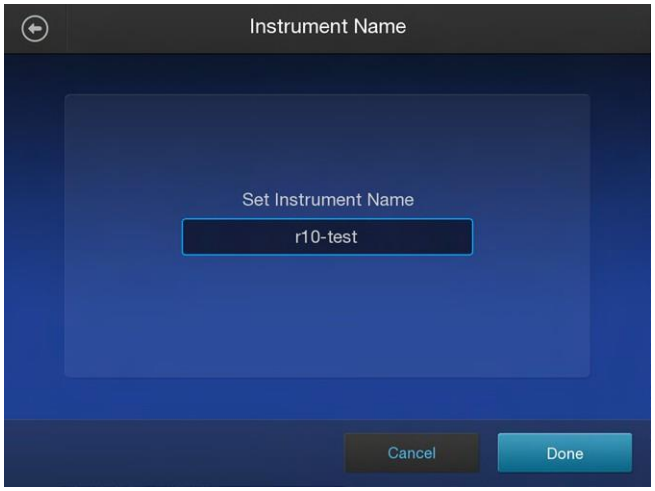
## Instrument settings

The **Instrument Settings** buttons access information about the instrument and allow you to set the instrument name and calibrate the touchscreen.




| Button | Description | When/How to use |
|---|---|---|
| About | Provides instrument details.<br>  | Use the screen to view instrument reference information or access regulatory information. Click Regulatory info to view the following screen. |

*(continued)*

| Button | Description | When/How to use |
|---|---|---|
| Regulatory info | Lists instrument-specific regulatory information.<br>  | To view instrument RFID regulatory information. See “Radio compliance standards” on page 234 for compliance information. |
| Instrument name |  | To change the instrument name. |

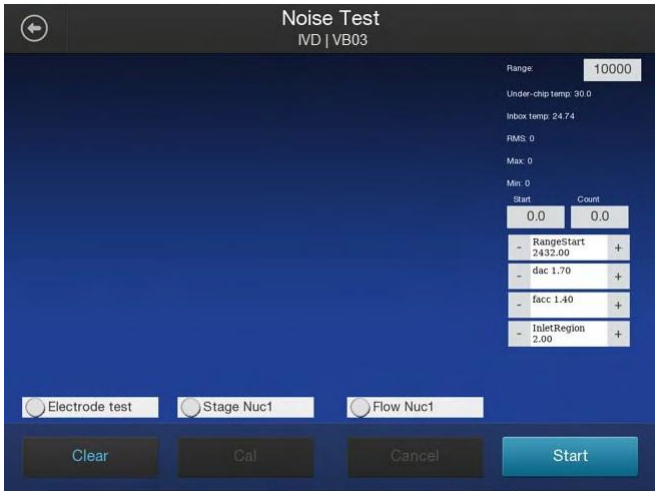
*(continued)*

| Button | Description | When/How to use |
|---|---|---|
| Screen calibration |  | For troubleshooting if directed to do so by Technical Support.<br>Touch the red cross with your finger or a stylus each time it appears. In total, you touch the screen 4 times, one time in each corner. |
| Smart Monitor Enabled | — | Select the checkbox to enable remote monitoring of instrument runs by Thermo Fisher Scientific for troubleshooting. |

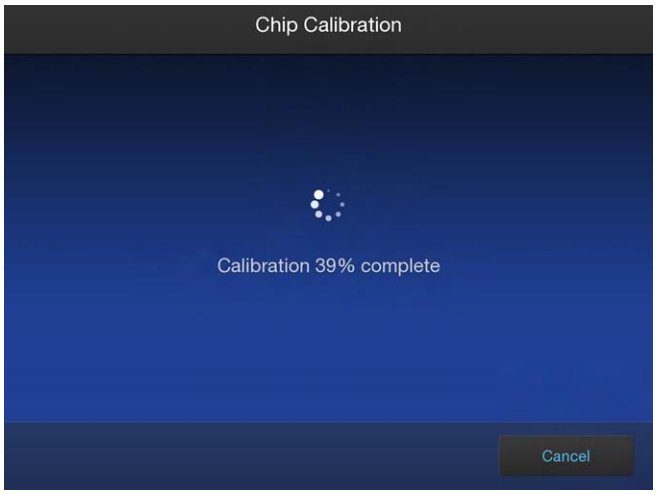
## System Tools

The **System Tools** menu enables you to upload instrument diagnostics, manage data, and shut down or reboot the instrument.

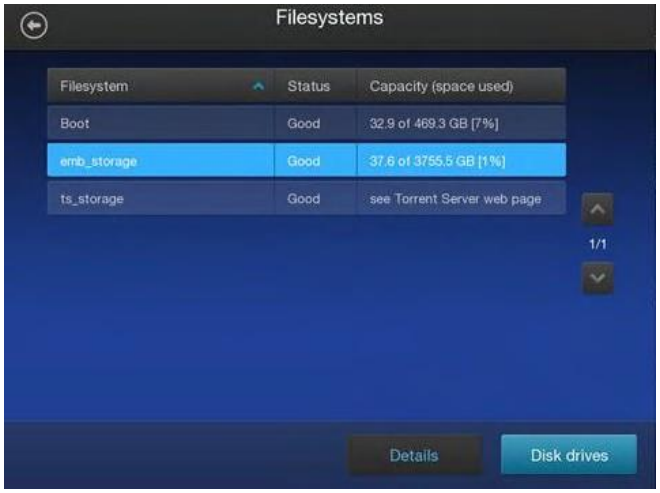
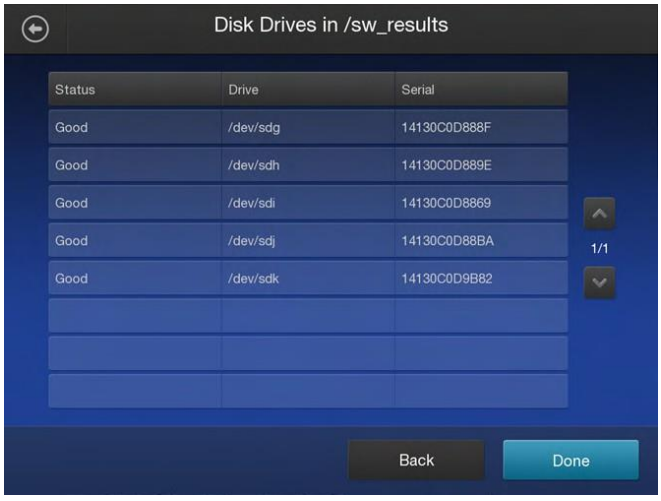
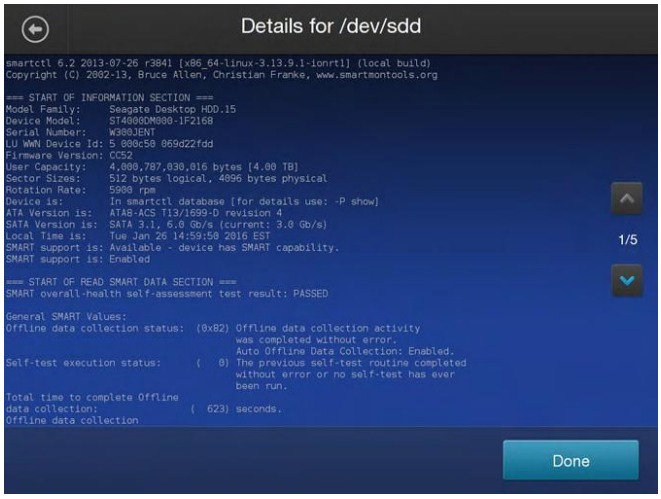


| Tool | Description | When/How to use |
|---|---|---|
| RFID | Lists the product, product expiration date, and remaining uses of the product.<br>  | To determine if reagents that were left on an instrument during a period of instrument shutdown have expired.<br>1. Tap the screen within the row of the product to view an RFID Tag Detail page.<br>2. Tap Done to return.<br>Note: You can also use the RFID Tags screen to track remaining days of reagent use before a sequencer initialization expires. We recommend that you use reagents within 14 days of initialization. After 28 days, the sequencer displays a warning at run start with options to proceed to a clean and new initialization, ignore to continue without clean, or cancel the run. For further information, see “Options for an expired sequencer initialization” on page 114. |
| Noise test | Provides real-time measurement of electrical noise readings on the chip.<br>  | For troubleshooting if directed to do so by Technical Support. |

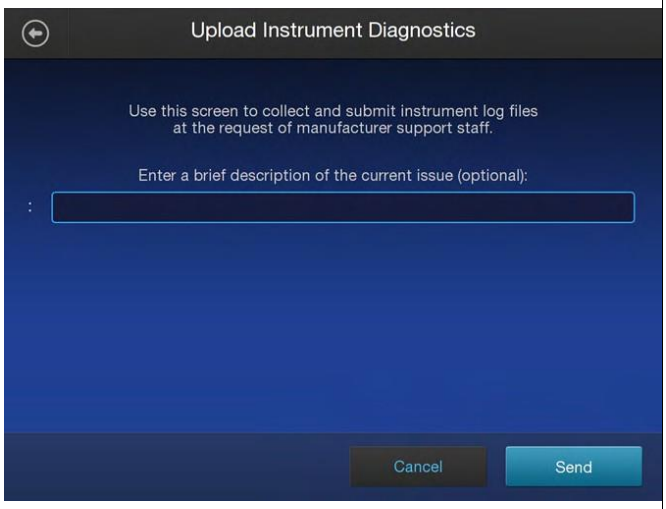
*(continued)*

| Tool | Description | When/How to use |
|---|---|---|
| Chip calibration | Checks the status of a chip.<br><br>IMPORTANT! Do not perform a chip calibration unless instructed to do so by a field service engineer.<br><br>  | **1.** Insert a new chip into the chip installation slot.<br>**2.** In the System Tools menu, tap Chip Calibration to start the chip calibration.<br>**3.** In the dropdown lists, select the Image to display and the type of Calibration to perform.<br>**4.** After a chip calibration, perform a Clean instrument before removing the chip.<br><br>Note: Tap Cancel to quit the chip calibration. |

*(continued)*

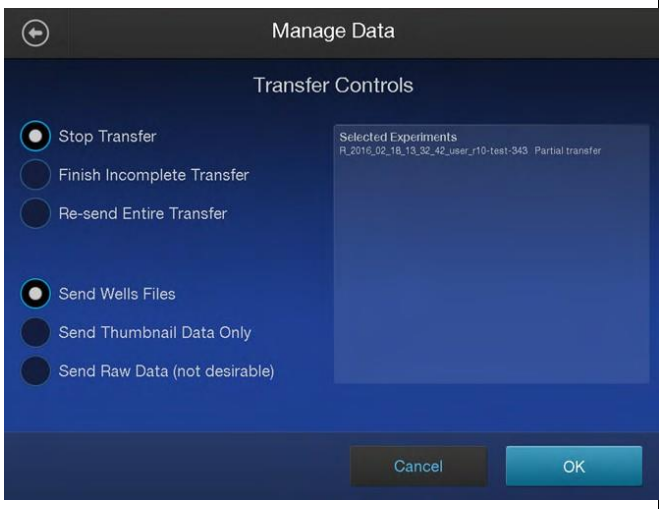
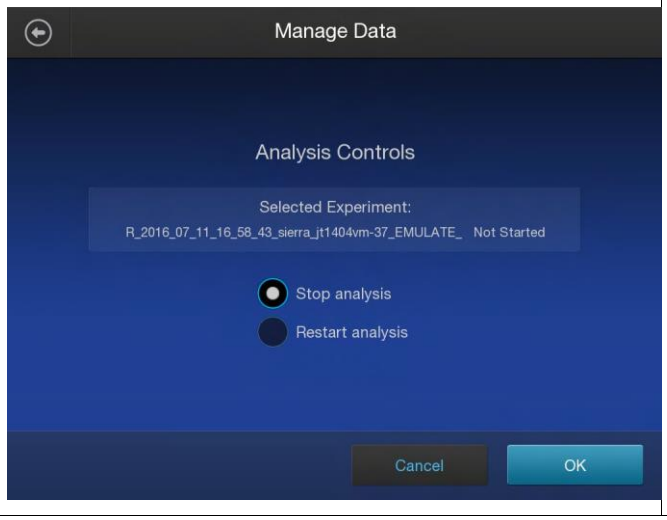
| Tool | Description | When/How to use |
|---|---|---|
| Filesystems and disks | Provides real-time drive activity status and disk space usage.<br>  <br><br>  <br><br>  <br> | Use if directed to do so as part of a troubleshooting procedure:<br>**1.** In the System Tools menu, tap Filesystems and disks. The Filesystems screen opens.<br>**2.** Tap the screen within the row of the Filesystem that you want to view, then tap Details. The Disk Drives in <Filesystem> screen opens.<br>**3.** Tap the screen within the row of the Drive to view details of the disk drive. The Details for <Drive> screen opens.<br>**4.** Tap Done to return to the previous screen. |

*(continued)*

| Tool | Description | When/How to use |
|---|---|---|
| Upload diagnostics | Upload instrument diagnostics files for review by support personnel.<br>  | Upload an instrument diagnostics file after an aborted run, or if otherwise directed to do so by Technical Support.<br>Note: The file is pushed to Genexus™ Dx Software. To retrieve the file using Genexus™ Dx Software, navigate to ⚙ (Settings)►About, click Instrument Diagnostics, then follow the on-screen instructions. |
| Shut down | Provides access to "Shut down" and "Reboot" commands.<br>  | Use these commands if directed to do so by Technical Support as part of a troubleshooting procedure, or if the instrument will not be used for more than 3 days.<br>Note: It is not necessary or recommended to power off the instrument overnight or over the weekend. To power on the instrument after a power off, see “Power on” on page 34. |

## Data Management

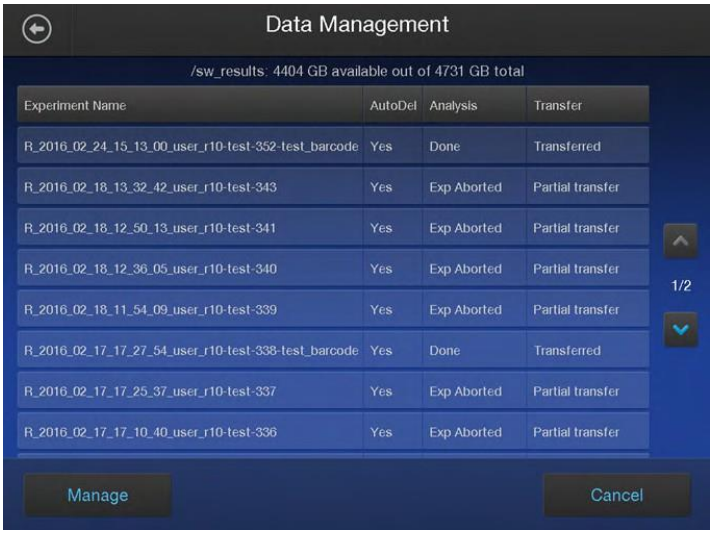
The **Data Management** function allows you to delete run data manually, or transfer data in the event of a failure of automatic transfer. Under normal conditions, run data are automatically transferred to the analysis server, then deleted from the instrument hard drive.

| Item | Description | When/How to use |
|---|---|---|
| Delete data | Manually delete run data from the instrument. | If the instrument hard drive becomes full, see “Manually delete run data” on page 204. |
| Transfer Controls | Transfer run data from the instrument hard drive to the Ion Torrent™ Server.<br>  | 1. Tap the screen in the Experiment Name row of the individual experiment to be transferred.<br>2. Tap Transfer.<br>3. Select the transfer action to be performed.<br>4. Select the file transfer option.<br>5. Tap OK. |
| Analysis Controls | Stop the analysis of the data of individual runs.<br>  | 1. Tap the screen in the Experiment Name row of the individual experiment to be analyzed, then tap Analysis.<br>2. Select Stop Analysis, then tap OK.<br>Note: Restart analysis is currently not supported. |

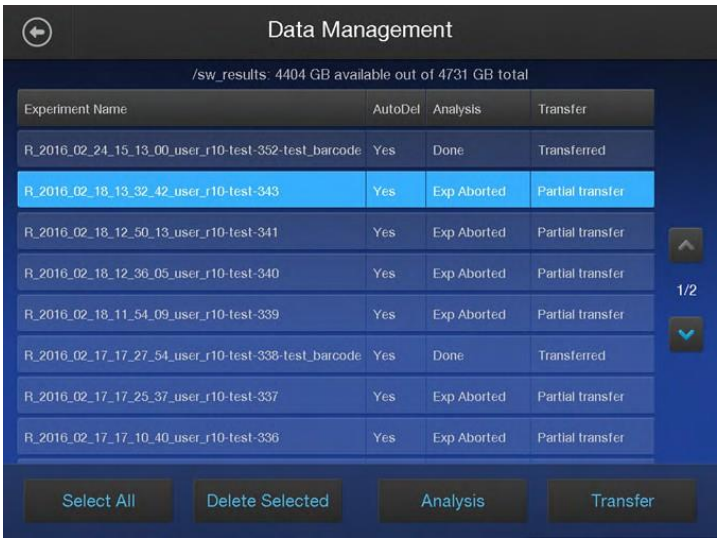
## Manually delete run data

To troubleshoot data management problems, use the **Data Management** function to delete run data manually, or transfer the data to an external server.

1. In the **Settings** menu, tap **Data Management** to access the **Data Management** screen, then tap **Manage**.




2. Tap **Select All** to select all the available experiments, or tap the screen in the **Experiment Name** row of the individual experiment to be managed.




3. Tap **Delete Selected**.

# C Supplemental information


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## Clean and decontaminate the Genexus™ Dx Purification System

Perform the decontamination procedure if accidental spills or leaks of samples or reagents occur during a run.

Each instrument run includes an automated ultraviolet (UV) light irradiation step (**UV clean**) that must be performed after every run. The **UV clean** is initiated from the instrument touchscreen and is designed to minimize potential contamination. After all consumables are removed from the instrument, the deck is irradiated with UV light for 2 minutes.

---

IMPORTANT! Although the Genexus™ Dx Purification System **UV clean** provides some protection against contamination, it is not a substitute for good laboratory techniques or precautions. When preparing samples for use or when preparing the instrument, ensure that you always observe sterile laboratory procedures to facilitate minimal contamination.

---

## Decontamination protocol

Perform the decontamination protocol if accidental spills or leaks of samples or reagents occur during a run.

The Genexus™ Dx Purification System includes an automated UV cleaning function that must be performed after every run. The cleaning routine is initiated from the instrument touchscreen and is designed to minimize potential contamination. After all consumables are removed from the instrument, the cleaning routine irradiates the deck with ultraviolet light for 2 minutes.

---

IMPORTANT! Although the Genexus™ Dx Purification System cleaning routine provides some protection against contamination, it is not a substitute for good laboratory techniques or precautions. When preparing samples for use or when preparing the instrument, ensure that you always observe sterile laboratory procedures to ensure minimal contamination.

---

## Materials required

- Laboratory coat
- Gloves, powder-free nitrile
- Protective safety glasses
- Deionized water
- Isopropanol, 70% solution
- Wipes, lint-free

## Decontaminate the Genexus™ Dx Purification System



WARNING! The samples can be potentially infectious. Dispose of all potentially contaminated consumables and wipes as biohazardous waste according to your local regulations.

The decontamination procedure must be completed before any instrument service is performed or before relocation of the instrument.

In the following scenarios, begin the decontamination procedure at the indicated step.

- If the sample or reagent spill occurred while loading the instrument and the instrument door is open. Begin from step 1.
- If the sample or reagent spill occurred while unloading the instrument and the instrument door is open. Begin from substep 2a or the appropriate substep for the next consumable that needs to be removed.
- If decontaminating the instrument in preparation for instrument service (no spill). Begin from step 5.

1. In the **Home** screen, tap **Cancel►OK**.
2. Remove and discard reagent plates and all consumables from the instrument deck.

   IMPORTANT! If needed, the **UV clean** irradiation step can be performed before removing and discarding reagent plates and consumables. For more information, see step 6.

   a. Remove the purification plate, pour the liquid waste into an appropriate liquid biohazardous waste container by tipping the deep-well plate on one corner, then discard the empty plate in a solid biohazardous waste container.

   WARNING!
   - Liquid waste contains guanidine thiocyanate. Dispose of liquid waste properly.
   - The samples can be potentially infectious. Dispose of all potentially contaminated consumables and wipes as biohazardous waste.

   b. Unlock, then remove and dispose of the Genexus™ Dx Purification Tip Cartridges in an appropriate biohazardous waste container.

   c. Unlock, then remove and empty the Genexus™ Dx Quantitation Plate by tipping the deep-well plate on one corner and pouring the waste into an appropriate liquid biohazardous waste container. Discard the empty plate in a solid biohazardous waste container.

   WARNING! No data are currently available that address the mutagenicity or toxicity of the Qubit™ RNA BR Reagent (Component A). This reagent is known to bind nucleic acid and is provided as a solution in DMSO. Treat the Qubit™ RNA BR Reagent with the same safety precautions as all other potential mutagens, and dispose of the dye in accordance with local regulations.

   d. Open the quantitation module cover, remove and discard the Genexus™ Dx Quantitation Tube, then allow the module cover to gently close.

   e. Remove, then dispose of the combs and archive plates into an appropriate solid biohazardous waste container.

3. Use lint-free wipes to soak up as much liquid as possible, then dispose of all liquid and solid waste in the appropriate biohazardous waste containers.
4. Wipe the affected surfaces with wipes moistened with deionized water.
5. Wipe the affected surfaces with wipes moistened with 70% isopropanol. Wipe surfaces with a clean wipe, then allow to air-dry.
6. Close the instrument door, then perform a UV cleaning.
   a. Close the instrument door.

   b. In the **Home** screen, tap **Settings►System Tools►UV clean**.

   c. Tap **Yes**.

d. When the UV irradiation of the deck is complete, tap **Open Door**.

The instrument is now ready to perform a new purification run.

# Create a template for importing samples in standalone configuration purification run plans

To use the **Import** function to import sample information from a USB drive into a standalone configuration run plan for the Genexus™ Dx Purification System, add the sample information to a CSV file with a specific format. Follow these steps to create an import template file with a plain text editor, or with Microsoft™ Excel™ software.

1. Use the format in the follow examples to set up two columns in a CSV file for **Sample Name** and **Notes**. Populate the sample rows with sample name and notes.

```
Sample Name,Notes
CSV Sample 1,CSV notes for sample 1
CSV Sample 2,CSV notes for sample 2
CSV Sample 3,CSV notes for sample 3
CSV Sample 4,CSV notes for sample 4
CSV Sample 5,CSV notes for sample 5
CSV Sample 6,CSV notes for sample 6
CSV Sample 7,CSV notes for sample 7
CSV Sample 8,CSV notes for sample 8
CSV Sample 9,CSV notes for sample 9
CSV Sample 10,CSV notes for sample 10
CSV Sample 11,CSV notes for sample 11
CSV Sample 12,CSV notes for sample 12
```

① Example file if in a plain text editor.

| | A | B |
|---|---|---|
| 1 | Sample Name | Notes |
| 2 | CSV Sample 1 | CSV notes for sample 1 |
| 3 | CSV Sample 2 | CSV notes for sample 2 |
| 4 | CSV Sample 3 | CSV notes for sample 3 |
| 5 | CSV Sample 4 | CSV notes for sample 4 |
| 6 | CSV Sample 5 | CSV notes for sample 5 |
| 7 | CSV Sample 6 | CSV notes for sample 6 |
| 8 | CSV Sample 7 | CSV notes for sample 7 |
| 9 | CSV Sample 8 | CSV notes for sample 8 |
| 10 | CSV Sample 9 | CSV notes for sample 9 |
| 11 | CSV Sample 10 | CSV notes for sample 10 |
| 12 | CSV Sample 11 | CSV notes for sample 11 |
| 13 | CSV Sample 12 | CSV notes for sample 12 |

② Example file in Microsoft™ Excel™ software.

The **Sample Name** field populates the **Sample ID** field in the instrument screen.

2. Save the file using a CSV file format to a USB drive.

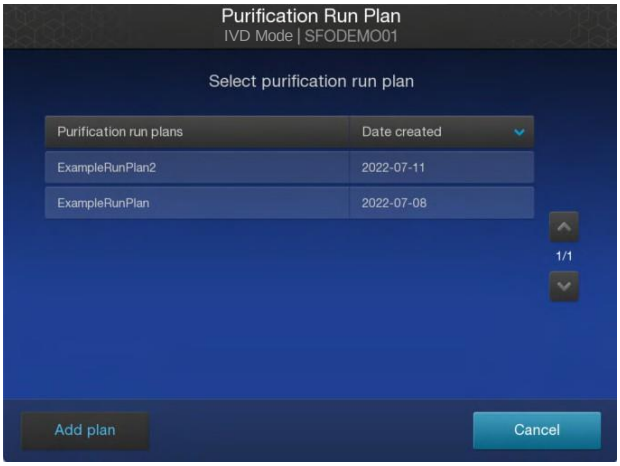
IMPORTANT!

- Do not add extra spaces in the headings, or add other columns in either file type.
- If you transfer files between macOS™ and Windows™ computers, extra characters that can cause errors can be added to files. Check for the presence of hidden characters, then delete if found.
- An alert is shown if fewer samples are in the CSV file than are selected for the run, or if two samples have the same name in the CSV file. No samples are imported until the error is corrected.

# Delete a run plan in the Genexus™ Dx Purification System touchscreen (standalone configuration)

1. Tap **Run**, then tap the plan to be deleted.




2. In the **Purification Run Plan Details** screen, tap **Edit**, then tap **Delete**.
3. Tap **Delete** to confirm deletion of the selected purification run plan.

Note: Deletion of a purification run plan is irreversible and cannot be undone.

# Maintain the Genexus™ Dx Integrated Sequencer

## Materials required

Cleaning

- Lint-free wipes
- Deionized water
- 70% isopropanol

Liquid waste filter replacement

- Genexus™ Dx Filter (ordered separately [Cat. No. A50868])

Conical bottle replacement

- Genexus™ Dx Conical Bottles (from the Genexus™ Dx Integrated Sequencer Install Kit, or ordered separately [Cat. No. A50869])

## Clean or decontaminate the sequencer

If a spill or leak occurs inside the instrument, perform the following steps to clean or decontaminate the sequencer.

Note: Dispose of all waste in appropriate liquid or solid waste containers.

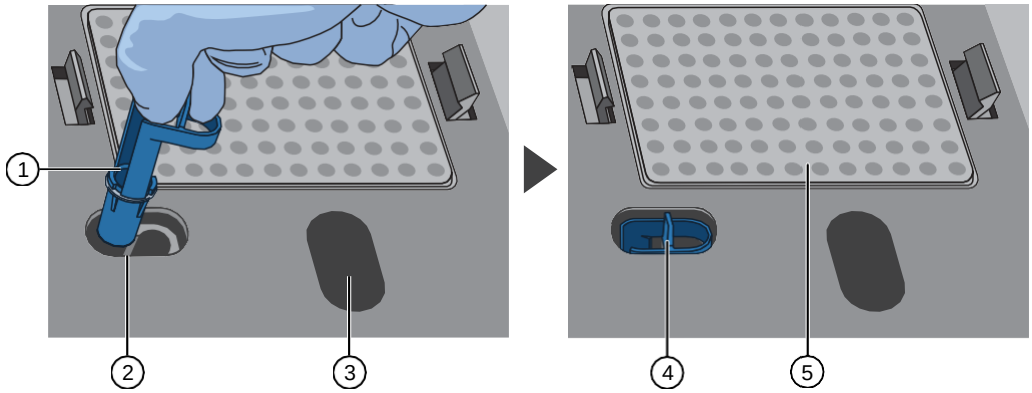
1. Remove each Genexus™ Dx Bottle 2, then remove and empty the Genexus™ Dx Bottle 1 and Waste carboy.
2. Remove the Genexus™ Dx Cartridge.
3. Inspect the floor of the sequencing reagents bay and Genexus™ Dx Cartridge port for liquid.
4. Using lint-free wipes, soak up as much liquid as possible, then clean the affected area with wipes moistened with deionized water.
5. Wipe the affected surfaces with 70% isopropanol, then allow to air-dry.

## Replace the Genexus™ Dx Filter

The Genexus™ Dx Filter captures particulate matter in the liquid waste to prevent blockage of the waste line over time. The filter needs to be replaced with a new filter after one year of regular instrument use as part of annual preventive maintenance, or if a clog is present. For more information, see “Clear the instrument deck and perform a UV Clean” on page 111.

1. Remove the old filter from the liquid waste disposal port on the instrument deck by grasping the filter firmly and pulling up. Discard the filter as regular waste.

2. Insert a new Genexus™ Dx Filter into the liquid waste disposal port, then press firmly to seat the filter O-ring securely.




1. Genexus™ Dx Filter
2. Liquid waste disposal port
3. Used pipette tip disposal port
4. Installed filter
5. Sample plate station

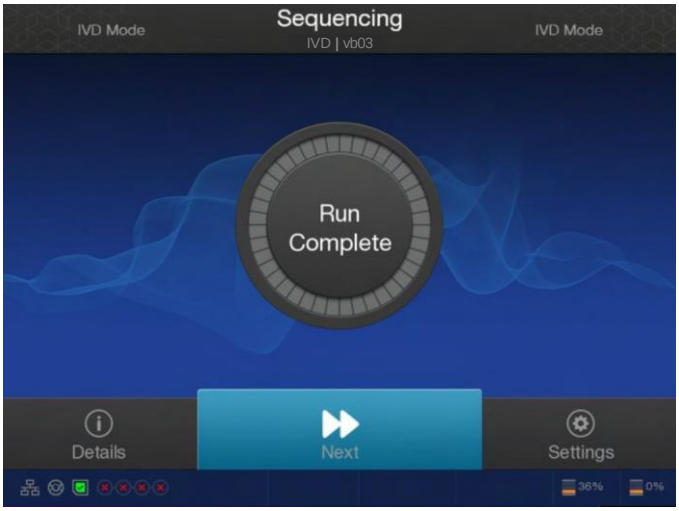
## Replace the Genexus™ Dx Conical Bottles

The Genexus™ Dx Conical Bottles need to be replaced after one year of regular instrument use as part of annual preventive maintenance or when filters in the bottles become partially clogged, causing decreased flow rate. The Genexus™ Dx Integrated Sequencer measures flow rate in a Conical Flow Rate test that is performed during every post-chip clean. If the conical flow rate test fails, the sequencer automatically redirects to the conical bottle replacement procedure. During a normal run, a post-chip clean is performed in three situations:

- At the end of the run if all four lanes have been used.
- At the end of the run if you selected **Do Force Clean** at run setup.
- At run setup when a run is selected but insufficient unused lanes in the installed chip are available for the run. If you continue with the run, a post-chip clean is performed before the run can proceed.

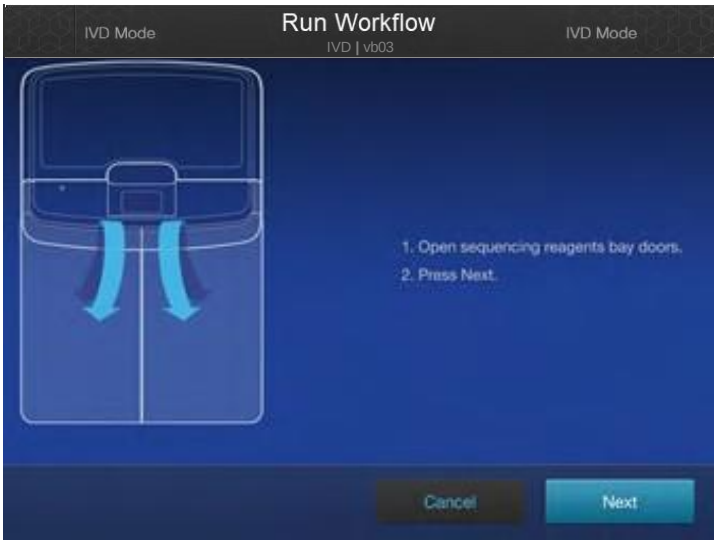
Follow these steps to replace the Genexus™ Dx Conical Bottles.

1. After a run, in the **Run Complete** screen, tap **Next**.




If the Conical Flow Rate test failed, a screen appears directing you to open the sequencing reagents bay doors.

2. Open the sequencing reagents bay doors, then tap **Next**.




The **Remove Consumables** screen appears.

3. Remove, but do not discard, the Genexus™ Dx Bottle 1 (position 1), the two Genexus™ Dx Bottle 2 bottles (positions 2 and 3), and the Genexus™ Dx Bottle 3 (position 4), then tap **Next**.




4. Remove the five used Genexus™ Dx Conical Bottles, install five new conical bottles, then tap **Next**.




Note: Discard the used conical bottles appropriately. Inspect the plastic nozzles on the new bottles for any pinches or deformations. To avoid pinching or folding of the plastic nozzle, install the bottles straight-on, not at an angle.

5. Replace the four reagent bottles that you removed in step 3 in their original positions.




Note: Inspect the plastic nozzles for any pinches or deformations. To avoid pinching or folding of the plastic nozzle, install the bottles straight-on, not at an angle.

6. After replacing the bottles, close the sequencing reagents bay doors, then tap **Next** in the **Insert Consumables** screen.

   After you tap **Next**, the sequencer starts a **Leak Test**.



Note: If you tap **Next** while the sequencing reagents bay doors are open, an alert appears asking you to close the doors before proceeding.

- If the sequencer passes the **Leak Test**, no action is needed. The sequencer proceeds to a cleaning cycle.




- If the sequencer fails the **Leak Test** a notification appears. Tap **OK** to return to step 2 to fix the leak. Open the sequencing reagents bay doors, check the tightness of each conical bottle that you installed, then ensure that the four reagent bottles are installed correctly. Close the sequencing reagents bay doors, then tap **Next** to repeat the **Leak Test**. If the **Leak Test** fails again, contact Technical Support.

After completion of a successful **Leak Test** and cleaning, the sequencer moves to a normal workflow.

- If the sequencer is at the end of the run, the next screen is a **Clear Deck** screen, followed by a **Clear Sequencing Reagents** screen.
- If the sequencer is at the start of the run, the next screen is a **Clear Deck** screen, followed by a **Load Deck** screen.

## Power off and power on the sequencer before and after a long-term shutdown

Follow these procedures to power off the sequencer if it will not be used for more than 28 days, and to power on after a long-term shutdown.

### Power off before a long-term shutdown

- If all four lanes of a chip have been used, and a post-chip clean has completed successfully, follow these steps to power off the sequencer, as described in “Power off” on page 34.
  a. In the home screen, tap **Settings►System Tools►Shut down**.
  b. Select **Shutdown**.
     A confirmation message appears. Select **Yes** to power off the instrument.

c. Turn the power switch located at the back of the instrument to the off ( O ) position.

- If a partially used chip is installed on the instrument, follow these steps to perform a Clean instrument procedure as described in page 194, then power off the instrument.

  a. In the home screen, tap **Settings►Clean Instrument**.

  b. Follow the on-screen instructions to start the cleaning.

  Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

  c. After the cleaning completes, power off the system in the home screen by tapping **Settings►System Tools►Shut down**.

  d. Select **Shutdown**.
  A confirmation message appears. Select **Yes** to power off the instrument.

  e. Turn the power switch located at the back of the instrument to the off ( O ) position.

## Power on after a long-term shutdown

After a long-term shutdown (>28 days), follow these steps to power on the sequencer, as described in “Power on” on page 34.

1. Turn the power switch on the back of the sequencer to the on ( | ) position.
2. Press the power button on the front of the instrument.
   The button illuminates.
3. Sign in with your user name and password.
   After the home screen appears, the sequencer is ready for use. Check for instrument alarms, if any.
4. Perform a Clean instrument procedure as described in “Perform a Clean instrument procedure” on page 194. A Clean instrument procedure must be performed even if the procedure was performed before the long-term shutdown.

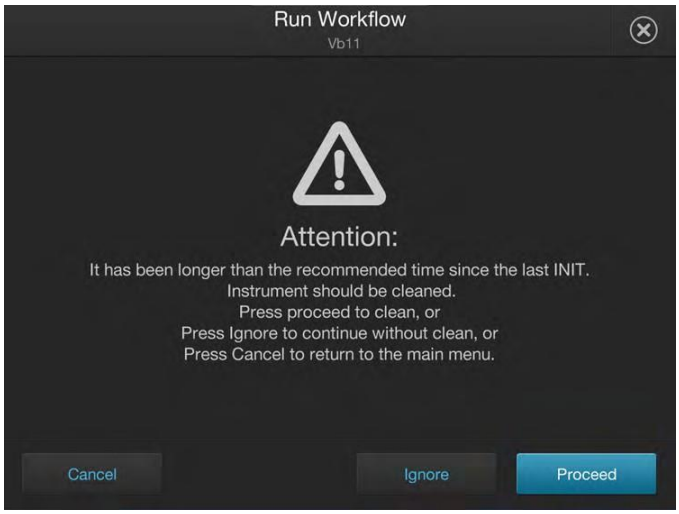
   a. In the home screen, tap **Settings ►Clean Instrument**.

   b. Follow the on-screen instructions to start the cleaning.

   Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

   c. After the cleaning is complete, the instrument is ready to use.

5. If the instrument was in an initialized state before the system was powered off, and shows the following alarm for an expired initialization, perform the following steps to clean the instrument as described in “Options for an expired sequencer initialization” on page 114.




   a. Select **Cancel** to return to the home screen, then tap **Settings ►Clean Instrument**.

   b. Follow the on-screen instructions to start the cleaning.

      Note: If an alarm appears, follow the on-screen prompts, or contact technical support for further help. Do not continue until the problem is resolved.

   c. After the cleaning is complete, the instrument is ready to use.

# Instrument planned maintenance

The Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer need annual planned maintenance. The instruments display a notification when planned maintenance is due. To schedule a planned maintenance visit, contact your Thermo Fisher Scientific Field Service Engineer.

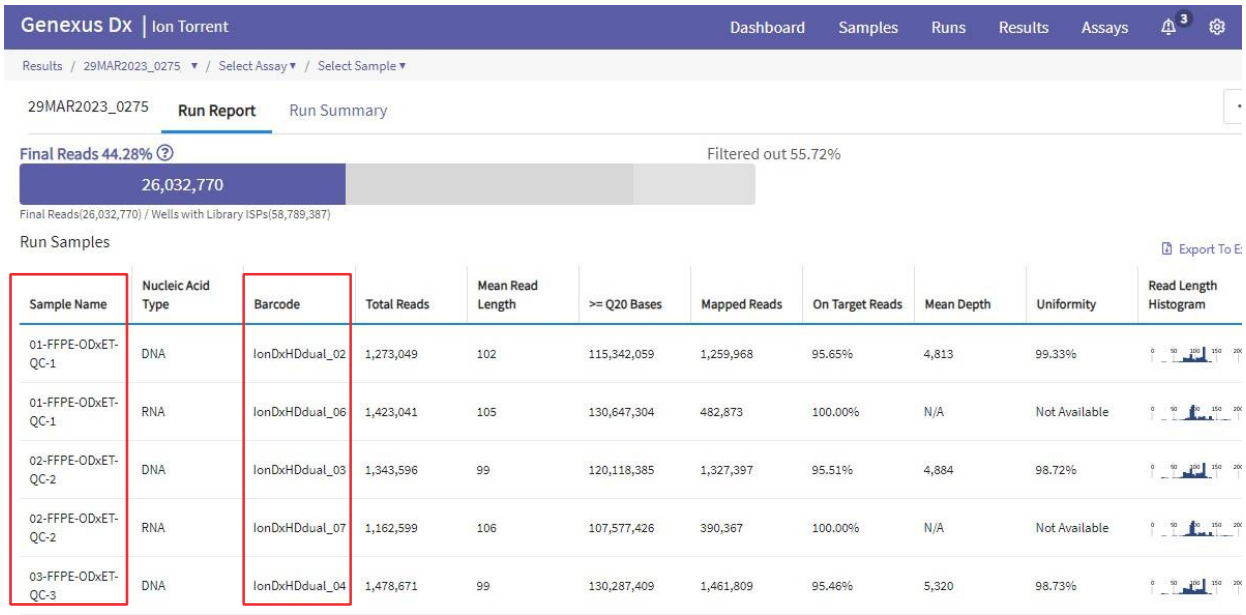
# Recover pooled sample libraries and plan a Library to Result run

If a sequencing run fails due to a templating or sequencing error, you can recover pooled sample libraries from well E4 of the PCR amplification plate from the run and use them in a subsequent Library to Result run. Follow these steps to plan a run and recover the pooled sample libraries.

## Prepare a library batch

If you are planning a run starting from pooled libraries that you have recovered from a previous sequencing run, you need to first prepare a library batch in Genexus™ Dx Software from samples that you have entered. For more detailed instructions, see “Prepare a library batch” on page 41.

To prepare a library batch, have at hand the sample names, HD barcodes, and assay used in the run that you want to repeat. This information can be found in the run report.




1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select all the samples included in the failed run, or contained in the E4 pool, by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.
3. In the **Create Library batch** screen, select the assay used in the previous run from the **Select Assay** dropdown list.
4. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.
5. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.
6. Select the DNA and RNA barcodes used for each sample as specified in the run report.
7. For the FFPE DNA and RNA Positive Controls and NTC, enter or scan with a barcode scanner the barcode from the control kit in the DNA and RNA **Control Kit Barcode** fields. For the NTC, enter or scan the barcode from the NTC kit in the DNA and RNA **Control Kit Barcode** fields.
8. Click **Submit** to create the library batch.
   The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Plan the Library to Result run

After creating the library batch, plan a new Library to Result run using the library batch with its associated assay. For more detailed instructions, see “Plan a Library to Result run” on page 63.

For a 4-lane FFPE DNA/RNA run (12 samples + 2 positive controls + 2 NTCs), a Library to Result run requires 140 µL of pooled library. If the volume recovered from this run is <140 µL, use nuclease-free water or low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required 140 µL.

For a 2-lane FFPE DNA/RNA run (6 samples + 2 positive controls + 2 NTCs), a Library to Result run requires 80 µL of pooled library. If the volume recovered from this run is <80 µL, use nuclease-free water or low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required 80 µL.

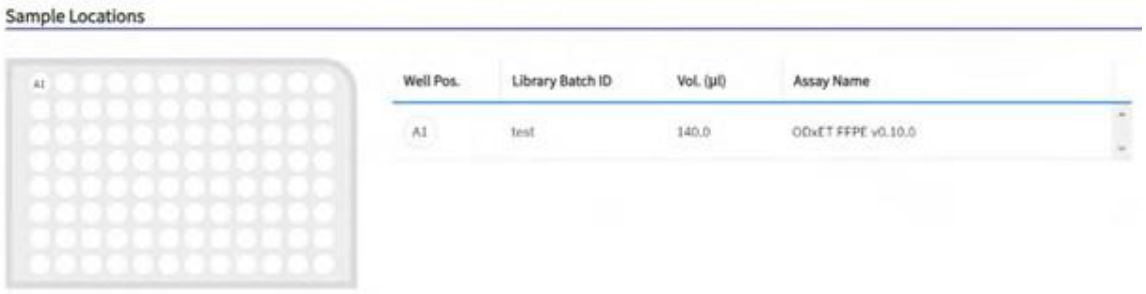
---

IMPORTANT! Run failure due to low loading can occur if insufficient pooled library is recovered and is too diluted. We recommend that you dilute recovered libraries no more than 2-fold when adjusting to the required volume.

---

1. In the menu bar, click **Runs►Library to Result**.
2. In the **Setup** step, enter a name for the run, configure the reporting options, then click **Next**.
3. In the **Assays** step, select the assay that you selected for the library batch, then click **Next**.
4. In the **Library Batches** step, select the library batch that you created, then click **Next**.
5. In the **Review** step, review the run setup guide generated for the sample library pool.

   IMPORTANT! Run failure due to low loading can occur if insufficient library pool is recovered from well E4 and is too diluted.




Proceed to “Recover the pooled sample libraries” to recover the sample library pool from well E4 of the PCR amplification plate.

## Recover the pooled sample libraries

After a run, pooled libraries are in well E4 of the PCR amplification plate for all workflows. The pooled sample libraries in well E4 are covered with a layer of oil. All the oil must be removed to avoid interference with downstream reactions.

IMPORTANT! The following procedure should be performed in a dedicated post-PCR laboratory space and/or laminar flow hood to decrease the possibility of spreading amplicon contamination.

1. After the run is complete, unload the plate from the sequencer deck. Seal the plate with an adhesive PCR plate foil (Cat. No. AB0626), then centrifuge the plate at 1,000 × *g* for 15–30 seconds.
2. Remove the foil, then collect the entire contents of well E4 with a pipette. Transfer the volume to a clear 0.5-ml or 1.5-ml microcentrifuge tube (low nucleic acid-binding).

   Note: A narrow tube is recommended to see the top layer of oil more easily.
3. Centrifuge the tube at 1,000 × *g* for 15–30 seconds, then carefully remove the top oil layer with a pipette. Repeat the centrifugation if needed until all the oil has been removed.
4. Determine the volume of the sample library pool with a pipette. If needed, add nuclease-free water or Low TE buffer (10 mM Tris, 0.1 mM EDTA, pH 8.0) to bring the volume up to the required volume shown in the run setup guide, then add to well A1 of a new sample plate.

The sample plate is ready to load in the sequencer for a Library to Result run. Proceed to Chapter 7, “Start a sequencing run”.

# Genexus™ Dx Install Kit

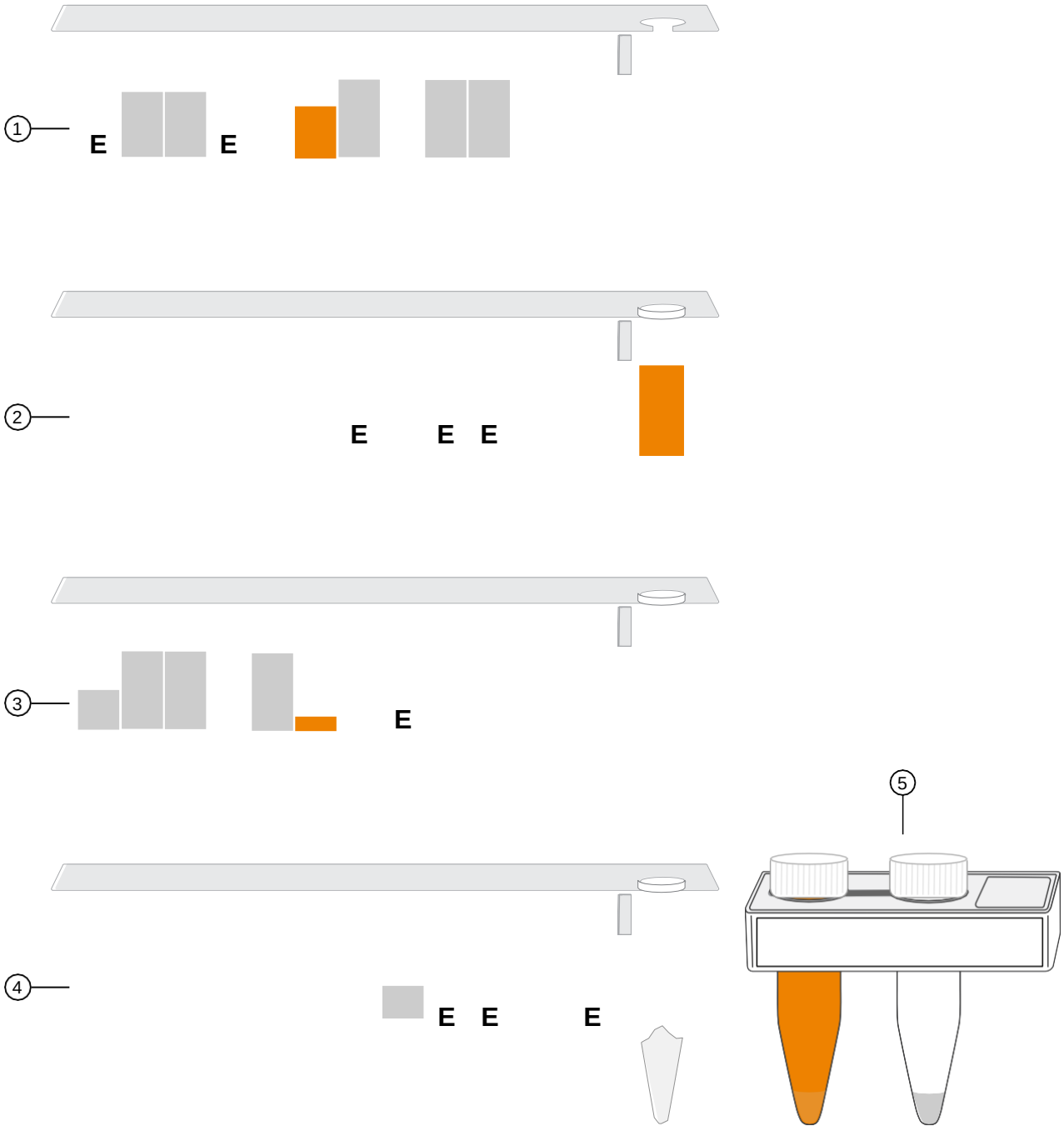
The Ion Torrent™ Genexus™ Dx Install Kit (Cat. No. A52172) is provided to first-time owners of a Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer. The kit contains the following reagents, supplies, and controls that are used during the installation, training, and operation of the instruments.

Genexus™ Dx Install Kit

| Contents | Part No. | Quantity | Storage |
|---|---|---|---|
| Genexus™ Dx Purification Install Kit | A52169 | 1 kit | −30°C to −10°C |
| Genexus™ Purification Integrated Workflow PQ Controls | A49291 | 1 each | 2°C to 8°C |
| Genexus™ Dx FFPE DNA and RNA Purification | A51076 | 1 kit | 15°C to 30°C |
| Genexus™ Dx Nucleic Acid Quantitation | A51078 | 1 kit | 2°C to 8°C |
| Genexus™ Dx Purification Supplies 2 | A51081 | 1 kit | 15°C to 30°C |
| Genexus™ Dx Integrated Sequencer PQ Kit<br>• Genexus™ Dx PQ Panel-HD<br>• Genexus™ Dx DNA PQ Control<br>• Genexus™ Dx PQ Library-HD | A51336 | 1 kit | −30°C to −10°C |
| Genexus™ Dx Barcodes 1–32 HD | A50410 | 1 plate | 15°C to 30°C |
| Genexus™ Dx Strip 1 | A50862 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 2-HD | A50861 | 8 strips | −30°C to −10°C |
| Genexus™ Dx Strip 3-GX5™ | A50864 | 8 strips | 2°C to 8°C |
| Genexus™ Dx Strip 4 | A50863 | 8 strips | −30°C to −10°C |
| Genexus™ Dx Cartridge | A50865 | 2 cartridges | −30°C to −10°C |
| Genexus™ Dx GX5™ Chip and Genexus™ Coupler | A54106 | 2 each | 15°C to 30°C |
| Genexus™ Dx Bottles 1 and 3 | A50867 | 2 bottles each | |
| Genexus™ Dx Bottle 2 | A50866 | 4 bottles | |
| Genexus™ Dx Pipette Tips | A50426 | 12 racks | |
| Genexus™ Dx Filter | A50868 | 1 filter | |
| Genexus™ Dx Conical Bottles | A50869 | 2 sets of 5 bottles | |
| Adhesive PCR Plate Foils | AB0626 | 1 box (100 foils) | |

# Genexus™ Dx reagent strip and panel tube fill volume guide

You can use the figures below to check for appropriate fill volume of library and template strip tubes, and Oncomine™ Dx Express Test primer pool tubes. Print this page at 100% and keep as a reference. If a fill volume appears inaccurate, contact Technical Support to report the problem. "E" indicates an empty tube.




1. Genexus™ Dx Strip 1
2. Genexus™ Dx Strip 2-HD
3. Genexus™ Dx Strip 3-GX5™
4. Genexus™ Dx Strip 4
5. Assay primer pool tubes

# D Performance characteristics

For performance characteristics of the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

# Safety

WARNING! **GENERAL SAFETY.** Using this product in a manner not specified in the user documentation may result in personal injury or damage to the instrument or device. Ensure that anyone using this product has received instructions in general safety practices for laboratories and the safety information provided in this document.

- Before using an instrument or device, read and understand the safety information provided in the user documentation provided by the manufacturer of the instrument or device.
- Before handling chemicals, read and understand all applicable Safety Data Sheets (SDSs) and use appropriate personal protective equipment (gloves, gowns, eye protection, and so on). To obtain SDSs, visit **thermofisher.com/support**.

## Symbols on this instrument

Symbols may be found on the instrument to warn against potential hazards or convey important safety information. In this document, the hazard symbol is used along with one of the following user attention words.

- **CAUTION!**—Indicates a potentially hazardous situation that, if not avoided, may result in minor or moderate injury. It may also be used to alert against unsafe practices.
- **WARNING!**—Indicates a potentially hazardous situation that, if not avoided, could result in death or serious injury.
- **DANGER!**—Indicates an imminently hazardous situation that, if not avoided, will result in death or serious injury.

### Standard safety symbols

| Symbol and description |
|---|
| CAUTION! Risk of danger. Consult the manual for further safety information. |
| CAUTION! Risk of electrical shock. |
| CAUTION! Hot surface. |

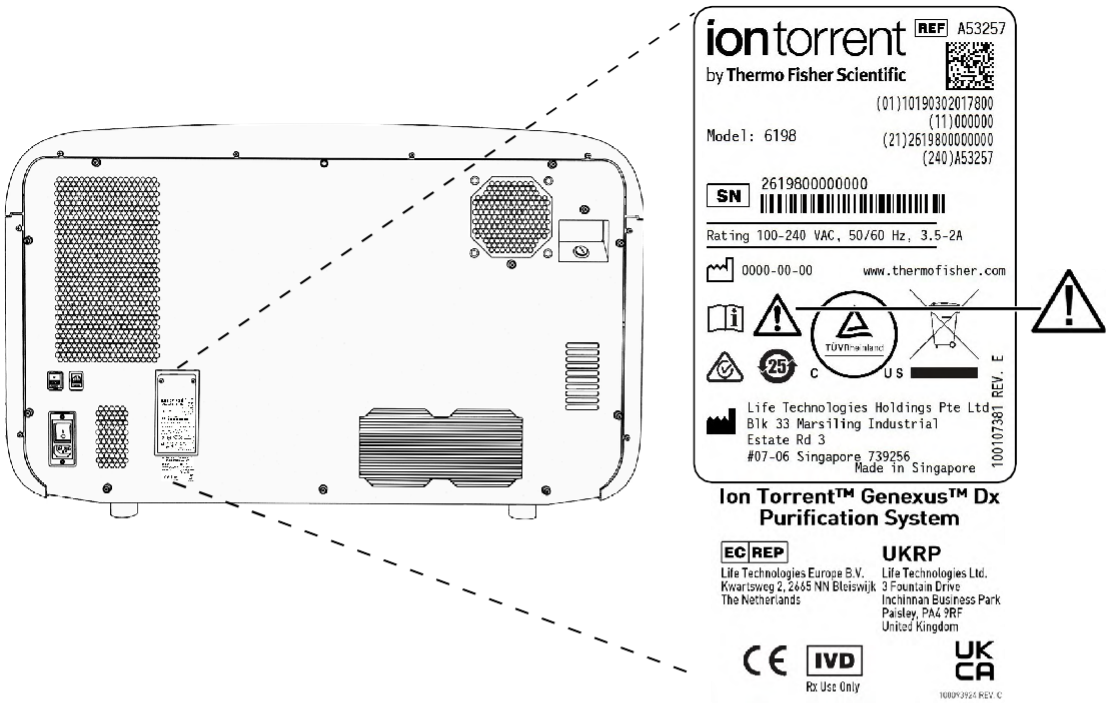
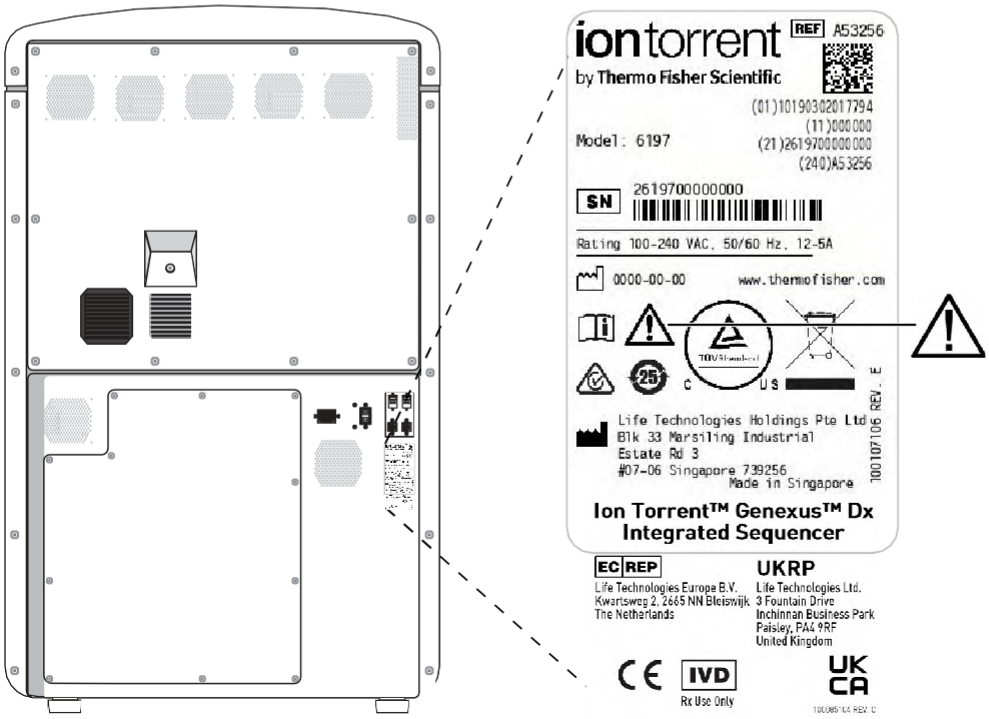
*(continued)*

| Symbol and description |
|---|
|  CAUTION! Potential biohazard. |
|  CAUTION! Ultraviolet light. |

## Additional safety symbols

| Symbol and description |
|---|
|  CAUTION! Moving parts. |
|  CAUTION! Piercing hazard. |

# Location of safety labels

| Label and location |
| --- |
|  <br>Purification instrument rear panel |
|  <br>Sequencer rear panel |

## Control and connection symbols

| Symbol | Description |
|---|---|
|  | On (Power) |
|  | Off (Power) |
|  | Protective conductor terminal (main ground) |
|  | Alternating current |

## Conformity symbols

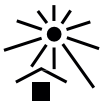
| Conformity mark | Description |
|---|---|
|  | Indicates conformity with safety requirements for Canada and U.S.A. |
|  | Indicates conformity with China RoHS requirements. |
|  | Indicates conformity with European Union requirements. |
|  | Indicates conformity with Australian standards for electromagnetic compatibility. |
|  | INDICATES CONFORMITY WITH THE WEEE DIRECTIVE 2012/19/EU.<br><br>CAUTION! To minimize negative environmental impact from disposal of electronic waste, do not dispose of electronic waste in unsorted municipal waste. Follow local municipal waste ordinances for proper disposal provision and contact customer service for information about responsible disposal options. |

# Definition of symbols on product labels or product documents

The following table describes symbols that are present on product labels or product documents. Symbols conform to applicable international regulatory standards.

| Symbol | Description |
|---|---|
|  | MANUFACTURER |
|  | DATE OF MANUFACTURE (YYYY-MM-DD) |
|  | CONSULT ELECTRONIC INSTRUCTIONS FOR USE |
|  | CATALOGUE NUMBER |
|  | UNIQUE DEVICE IDENTIFIER |
|  | BATCH CODE |
|  | SERIAL NUMBER |
|  | CONTAINS SUFFICIENT FOR <n> TESTS |
|  | DO NOT REUSE |

*(continued)*

| Symbol | Description |
|---|---|
|  | USE-BY DATE |
|  | TEMPERATURE LIMIT |
|  | UPPER LIMIT OF TEMPERATURE |
|  | LOWER LIMIT OF TEMPERATURE |
|  | UPPER AND LOWER LIMITS OF HUMIDITY |
|  | KEEP AWAY FROM SUNLIGHT, KEEP AWAY FROM HEAT |
|  | IN VITRO DIAGNOSTIC MEDICAL DEVICE |
|  | PRESCRIPTION USE ONLY |
|  | READ SAFETY DATA SHEET (SDS) |
|  | BIOLOGICAL RISKS |
|  | OBSERVE PRECAUTIONS FOR HANDLING ELECTROSTATIC SENSITIVE DEVICES |

*(continued)*

| Symbol | Description |
|---|---|
|  | CAUTION, CONSULT ACCOMPANYING DOCUMENTS |
|  | FRAGILE, HANDLE WITH CARE |

# Instrument safety

## General



CAUTION! **Do not remove instrument protective covers.** If you remove the protective instrument panels or disable interlock devices, you may be exposed to serious hazards including, but not limited to, severe electrical shock, laser exposure, crushing, or chemical exposure.

## Physical injury



CAUTION! **Moving and Lifting Injury.** The instrument is to be moved and positioned only by the personnel or vendor specified in the applicable site preparation guide. Improper lifting can cause painful and permanent back injury.

Things to consider before moving the instrument:

- Depending on the weight, moving may require two or more persons.
- If you decide to move the instrument after it has been installed, do not attempt to do so without the assistance of others, the use of appropriate moving equipment, and proper lifting techniques.
- Make sure that the path from where the object is to where it is being moved is clear of obstructions.



CAUTION! **Moving Parts.** Moving parts can crush, pinch and cut. Keep hands clear of moving parts while operating the instrument. Disconnect power before servicing.

## Electrical safety

WARNING! **Ensure appropriate electrical supply.** For safe operation of the instrument:

- Plug the system into a properly grounded receptacle with adequate current capacity.
- Ensure the electrical supply is of suitable voltage.
- Never operate the instrument with the ground disconnected. Grounding continuity is required for safe operation of the instrument.

WARNING! **Power Supply Line Cords.** Use properly configured and approved line cords for the power supply in your facility.

WARNING! **Disconnecting Power.** To fully disconnect power either detach or unplug the power cord, positioning the instrument such that the power cord is accessible.

## Cleaning and decontamination

CAUTION! **Cleaning and Decontamination.** Use only the cleaning and decontamination methods that are specified in the manufacturer user documentation. It is the responsibility of the operator (or other responsible person) to ensure that the following requirements are met:

- No decontamination or cleaning agents are used that can react with parts of the equipment or with material that is contained in the equipment. Use of such agents could cause a HAZARD condition.
- The instrument is properly decontaminated a) if hazardous material is spilled onto or into the equipment, and/or b) before the instrument is serviced at your facility or is sent for repair, maintenance, trade-in, disposal, or termination of a loan. Request decontamination forms from customer service.
- Before using any cleaning or decontamination methods (except methods that are recommended by the manufacturer), confirm with the manufacturer that the proposed method will not damage the equipment.

## Instrument component and accessory disposal

To minimize negative environmental impact from disposal of electronic waste, do not dispose of electronic waste in unsorted municipal waste. Follow local municipal waste ordinances for proper disposal provision and contact customer service for information about responsible disposal options.

# Safety and electromagnetic compatibility (EMC) standards

The instrument design and manufacture complies with the following standards and requirements for safety and electromagnetic compatibility.

## Safety standards

| Reference | Description |
|---|---|
| EU Directive 2014/35/EU | European Union low voltage directive |
| IEC 61010-1<br>EN 61010-1<br>UL 61010-1<br>CAN/CSA C22.2 No. 61010-1<br>GB 4793.1 (China) | *Safety requirements for electrical equipment for measurement, control, and laboratory use—Part 1: General requirements* |
| IEC 61010-2-010<br>EN 61010-2-010<br>GB 4793.6 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-010: Particular requirements for laboratory equipment for the heating of materials* |
| IEC 61010-2-020<br>EN 61010-2-020<br>GB 4793.7 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-020: Particular requirements for laboratory centrifuges* |
| IEC 61010-2-081<br>EN 61010-2-081<br>GB 4793.9 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-081: Particular requirements for automatic and semi-automatic laboratory equipment for analysis and other purposes* |
| IEC 61010-2-101<br>EN 61010-2-101<br>YY 0648 (China) | *Safety requirements for electrical equipment for measurement, control and laboratory use—Part 2-101: Particular requirements for in vitro diagnostic (IVD) medical equipment* |

## EMC standards

| Reference | Description |
|---|---|
| EU Directive 2014/30/EU | European Union EMC directive. |
| EN 61326-1<br>IEC 61326-1<br>GB/T 18268.1 (China) | *Electrical Equipment for Measurement, Control and Laboratory Use. EMC Requirements—Part 1: General Requirements* |
| EN 61326-2-6<br>IEC 61326-2-6 | *Electrical equipment for measurement, control and laboratory use—EMC requirements—Part 2-6: Particular requirements—In vitro diagnostic (IVD) medical equipment* |
| GB/T 18268.26 (China) | *Electrical equipment for measurement, control and laboratory use. EMC requirements. Part 26. Particular requirements. In vitro diagnostic (IVD) medical equipment* |
| FCC Part 18 (47 CFR) | *U.S. Standard for Industrial, Scientific, and Medical Equipment* |
| AS CISPR 11 | Industrial, scientific and medical equipment—Radio frequence disturbance characteristics—Limits and methods of measurement |
| ICES-001, Issue 4 | *Industrial, Scientific and Medical (ISM) Radio Frequency Generators* |
| FCC Part 15 Subpart B (47 CFR) | *U.S. Standard for Radio Frequency Devices*<br>This equipment has been designed and tested to CISPR 11 Class A. In a domestic environment it may cause radio interference, in which case, you may need to take measures to mitigate the interference.<br>Do not use this device in close proximity to sources of strong electromagnetic radiation (for example, unshielded intentional RF sources), because these can interfere with proper operation. |

## Environmental design standards

| Reference | Description |
|---|---|
| Directive 2012/19/EU | European Union WEEE Directive on waste electrical and electronic equipment. |
| Directive 2011/65/EU | European Union RoHS Directive on restriction of hazardous substances in electrical and electronic equipment. |
| SJ/T 11364-2014 | China RoHS Standard—Marking for the restriction of the use of hazardous substances in electronic and electrical products<br>For instrument specific certificates, visit our customer resource page at **www.thermofisher.com/us/en/home/technical-resources/rohs-certificates.html**. |

## Radio compliance standards

| Reference | Description |
|---|---|
| Directive 2014/53/EU | European Union: *RE Directive*—Radio equipment |
| RFID | FCC Notice (for U.S. Customers):<br>This device complies with Part 15 of the FCC Rules.<br>Operation is subject to the following conditions:<br>1. This device may not cause harmful interference, and<br>2. This device must accept any interference received, Including interference that may cause undesired operation.<br>Changes and modifications not expressly approved byThermo Fisher Scientific can void your authority to operate this equipment under Federal Communications Commissions rules. |
| RFID | Canada:<br>This device complies with Industry Canada license-exempt RSS standard(s). Operation is subject to the following two conditions:<br>(1) this device may not cause interference, and (2) this device must accept any interference, including interference that may cause undesired operation of the device. |
| RFID | Canada (Français québécois):<br>Le présent appareil est conforme aux CNR d'Industrie Canada applicables aux appareils radio exempts de licence. L'exploitation est autorisée aux deux conditions suivantes :<br>(1) l'appareil ne doit pas produire de brouillage, et (2) l'utilisateur de l'appareil doit accepter tout brouillage adioélectrique subi, même si le brouillage est susceptible d'en compromettre le fonctionnement. |
| Notice 423 [2005]<br>Industrial and Information Department Radio, BRR[1] | China:<br>Notice on issuing the technical requirements for micropower (short range) radio equipment. |
| GB/T 12572-2008 | China:<br>*Universal requirements and measurement methods of parameters for radio transmitting equipment* |
| ETSI EN 300 330-1 (2015-03) | China:<br>*Electromagnetic compatibility and Radio spectrum Matters (ERM); Short Range Devices (SRD); Radio equipment in the frequency range 9 kHz to 25 MHz and inductive loop systems in the frequency range 9 kHz to 30 MHz; Part 1 Technical characteristics and test methods* |

*(continued)*

| Reference | Description |
|---|---|
| No 1 [2014]1 of Industrial and Information Department Radio, BRR[1] | China:<br>*Methods for the administration of type approval of radio transmitting modules used in independent operation* |
| China: The Ministry of Industry and Information Technology (MIIT) issued a notice on micro-power short-range radio transmitting equipment. | |

[1] Bureau of Radio Regulation of the Ministry of Industry and Information Technology, State Radio Office

# Chemical safety

WARNING! **GENERAL CHEMICAL HANDLING.** To minimize hazards, ensure laboratory personnel read and practice the general safety guidelines for chemical usage, storage, and waste provided below. Consult the relevant SDS for specific precautions and instructions:

- Read and understand the Safety Data Sheets (SDSs) provided by the chemical manufacturer before you store, handle, or work with any chemicals or hazardous materials. To obtain SDSs, see the "Documentation and Support" section in this document.
- Minimize contact with chemicals. Wear appropriate personal protective equipment when handling chemicals (for example, safety glasses, gloves, or protective clothing).
- Minimize the inhalation of chemicals. Do not leave chemical containers open. Use only with sufficient ventilation (for example, fume hood).
- Check regularly for chemical leaks or spills. If a leak or spill occurs, follow the manufacturer cleanup procedures as recommended in the SDS.
- Handle chemical wastes in a fume hood.
- Ensure use of primary and secondary waste containers. (A primary waste container holds the immediate waste. A secondary container contains spills or leaks from the primary container. Both containers must be compatible with the waste material and meet federal, state, and local requirements for container storage.)
- After emptying a waste container, seal it with the cap provided.
- Characterize (by analysis if needed) the waste generated by the particular applications, reagents, and substrates used in your laboratory.
- Ensure that the waste is stored, transferred, transported, and disposed of according to all local, state/provincial, and/or national regulations.
- **IMPORTANT!** Radioactive or biohazardous materials may require special handling, and disposal limitations may apply.

WARNING! **HAZARDOUS WASTE (from instruments).** Waste produced by the instrument is potentially hazardous. Follow the guidelines noted in the preceding General Chemical Handling warning.

WARNING! **4L Reagent and Waste Bottle Safety.** Four-liter reagent and waste bottles can crack and leak. Each 4-liter bottle should be secured in a low-density polyethylene safety container with the cover fastened and the handles locked in the upright position.

# Biological hazard safety



WARNING! **Potential Biohazard.** Depending on the samples used on this instrument, the surface may be considered a biohazard. Use appropriate decontamination methods when working with biohazards.



WARNING! **BIOHAZARD.** Biological samples such as tissues, body fluids, infectious agents, and blood of humans and other animals have the potential to transmit infectious diseases. Conduct all work in properly equipped facilities with the appropriate safety equipment (for example, physical containment devices). Safety equipment can also include items for personal protection, such as gloves, coats, gowns, shoe covers, boots, respirators, face shields, safety glasses, or goggles. Individuals should be trained according to applicable regulatory and company/ institution requirements before working with potentially biohazardous materials. Follow all applicable local, state/provincial, and/or national regulations. The following references provide general guidelines when handling biological samples in laboratory environment.

- U.S. Department of Health and Human Services, *Biosafety in Microbiological and Biomedical Laboratories (BMBL)*, 6th Edition, HHS Publication No. (CDC) 300859, Revised June 2020
  **cdc.gov/labs/bmbl**
- Laboratory biosafety manual, fourth edition. Geneva: World Health Organization; 2020 (Laboratory biosafety manual, fourth edition and associated monographs)
  **who.int/publications/i/item/9789240011311**

# F Instrument warranty

For new Ion Torrent™ instruments, Life Technologies warrants to and only to buyer for twelve (12) months from the date of shipping, that the Ion Torrent™ software and Ion Torrent™ instruments are free from defects in material and workmanship and conform to Life Technologies' published specifications in all material respects. Where a valid and timely claim in respect of breach of Ion Torrent™ warranty is submitted to Life Technologies, Life Technologies may, at its discretion, replace, repair or modify the Ion Torrent™ instrument. Any agreed replacement shall be at 1:1, like-kind basis, at no cost to the buyer. For Ion Torrent™ chips or reagents reasonably determined by Life Technologies to be defective, independent of user error, shall be replaced by Life Technologies on a 1:1, like-kind basis at no cost to buyer, provided that such defective Ion Torrent™ chips or reagents were used by buyer prior to their expiration date, or if there is no expiration date, the Ion Torrent™ chips or reagents were used within six (6) months of receipt, and the defect was promptly reported with appropriate detail to Life Technologies' technical support.

NO OTHER WARRANTIES SHALL BE APPLICABLE TO ION TORRENT PRODUCTS (WHETHER OR NOT ANY FURTHER WARRANTY DOCUMENTATION MAY BE INCLUDED IN THE SHIPMENT), WITH THE EXCEPTION OF THIRD PARTY WARRANTIES WITH RESPECT TO THIRD PARTY PRODUCT. ANY THIRD PARTY PRODUCTS ARE NOT COVERED BY THIS SECTION AND ANY WARRANTIES FOR THIRD PARTY PRODUCTS ARE PROVIDED BY THE ORIGINAL MANUFACTURER OF THE THIRD PARTY PRODUCT. Warranties are made only to buyer purchasing the Ion Torrent™ Product directly from Life Technologies, are not transferable and do not extend to the benefit of any other person or entity, unless otherwise expressly stated in writing by Life Technologies. ANY PRODUCT NOT COVERED BY AN EXPRESS WRITTEN WARRANTY IS SOLD AND PROVIDED "AS IS," WITHOUT WARRANTY OF ANY KIND, STATUTORY, EXPRESS OR IMPLIED. Any description of Ion Torrent™ Product recited in Life Technologies' quotation is for the sole purpose of identifying Ion Torrent™ Product, and any such description is not part of any contract between Life Technologies and buyer and does not constitute a warranty that Ion Torrent™ Product shall conform to that description. Any sample or model used in connection with Life Technologies' quotation is for illustrative purposes only, and is not part of any contract between Life Technologies and buyer and does not constitute a warranty that Ion Torrent™ Product will conform to the sample or model. No affirmation of fact or promise made by Life Technologies, whether or not in Life Technologies' quotation, shall constitute a warranty that Ion Torrent™ Product will conform to the affirmation or promise. Unless otherwise specified in writing in documentation shipped with Ion Torrent™ Product or otherwise agreed by Life Technologies in writing. Life Technologies does not provide service or support for custom products or other products made to buyer's specifications. THE WARRANTIES IDENTIFIED IN THIS CLAUSE ARE LIFE TECHNOLOGIES' SOLE AND EXCLUSIVE WARRANTIES WITH RESPECT TO Ion Torrent™ PRODUCT AND ARE IN LIEU OF ALL OTHER WARRANTIES, STATUTORY, EXPRESS OR IMPLIED, ALL OF WHICH OTHER WARRANTIES ARE EXPRESSLY DISCLAIMED, INCLUDING WITHOUT LIMITATION ANY IMPLIED WARRANTY OF MERCHANTABILITY, FITNESS FOR A PARTICULAR PURPOSE, NON-INFRINGEMENT, OR REGARDING RESULTS OBTAINED THROUGH THE USE OF ANY PRODUCT (INCLUDING, WITHOUT LIMITATION, ANY CLAIM OF INACCURATE, INVALID OR INCOMPLETE RESULTS), WHETHER ARISING FROM A STATUTE OR OTHERWISE IN LAW OR FROM A COURSE OF PERFORMANCE, DEALING OR USAGE OF TRADE.



# Customer and technical support

Visit **thermofisher.com/support** for the latest in services and support, including:

- Worldwide contact telephone numbers
- Product support
- Order and web support
- Safety Data Sheets (SDSs; also known as MSDSs)

Additional product documentation, including user guides and Certificates of Analysis, are available by contacting Customer Support.

## Obtaining Certificates of Analysis

The Certificate of Analysis provides detailed quality control and product qualification information for each product. Certificates of Analysis are printed and shipped with the product.

## Obtaining Certificates of Conformance

The Certificate of Conformance provides information on conformance testing of each instrument provided with the system. Certificates of Conformance are shipped with the instrument, and are also available by contacting Customer Support at **thermofisher.com/support**.

## Cybersecurity  incidents

Report any cybersecurity incident that has occurred in relation to the device to Thermo Fisher Scientific. Contact Customer Support at **thermofisher.com/support** or **Contact Us**.

ion torrent

# Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference

## USER GUIDE

Publication Number MAN0024910

Revision A

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
#07-06, Singapore 739256

Products manufactured at this site:

- Genexus™ Dx Purification System
- Genexus™ Dx Integrated Sequencer
- Genexus™ Dx Software

Life Technologies Corporation |
7335 Executive Way |
Frederick, Maryland 21704 USA

Products manufactured at this site:

- Oncomine™ Dx Express Test
- Oncomine™ Dx Express Test FFPE DNA and RNA Control Kit
- Genexus™ Dx FFPE DNA/RNA Purification Kit
- Genexus™ Dx Library Strips 1 and 2-HD
- Genexus™ Dx Templating Strips 3-GX5™ and 4
- Genexus™ Dx Sequencing Kit
- Genexus™ Dx Barcodes 1–32 HD
- Genexus™ Dx GX5™ Chip and Genexus™ Coupler
- Genexus™ Dx Pipette Tips
- Genexus™ Dx Filter
- Genexus™ Dx Conical Bottles

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.

Revision history: MAN0024910 A (English)

| Revision | Date | Description |
|---|---|---|
| A | 25 June 2025 | New IVD-labeled part 3 user guide for the Oncomine™ Dx Express Test. |

The information in this guide is subject to change without notice.

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# About this guide

## Purpose of this guide

This user guide provides detailed instructions for sample entry and run planning in Genexus™ Dx Software, and reviewing results obtained with the Oncomine™ Dx Express Test. This guide also provides information on system administration, how to create reports, troubleshooting, and other reference information.

## Oncomine™ Dx Express Test user guides

This user guide is part of a three-guide set.

- *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide*
- *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*
- *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*

All three guides are required to complete the entire Oncomine™ Dx Express Test workflow.

# 1 Product information

## Oncomine™ Dx Express Test

The Ion Torrent™ Oncomine™ Dx Express Test (Cat. No. A54103) is ordered as a set of primer panel tubes sufficient for sequencing up to 32 libraries starting from FFPE tissue sections, or whole blood.

For a complete description of the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

## Theory of operation

For a complete description of the theory of operation, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

## Software overview

Ion Torrent™ Genexus™ Dx Software supports the Ion Torrent™ Genexus™ Dx Integrated Sequencer workflow from sample preparation through library preparation, template preparation, and sequencing. During and after sequencing, the software generates base calls, trims reads, and determines quality values (primary analysis), then aligns reads, calls variants, and generates reports (secondary analysis).

The software also supports the Ion Torrent™ Genexus™ Dx Purification System Instrument for nucleic acid isolation, followed by sequencing on the Genexus™ Dx Integrated Sequencer.

This documentation provides instructions for using the software to perform the following functions:

- Create and manage samples, libraries, and runs in the software, and manage assays.
- Monitor the status of runs on the Genexus™ Dx Integrated Sequencer, which provides all-in-one library preparation, chip loading, template preparation, and sequencing.
- Monitor runs on the Genexus™ Dx Purification System Instrument.
- View data, results, and quality control (QC) reports after a run.
- Download data files.
- Export and print audit records.
- Create and manage user accounts.

## Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software 6.8.2 or later. Version-specific information is provided in the software release notes for your version of the software. The software version is shown in the bottom footer of each screen in the software.

Genexus™ Dx Software is supported on Google Chrome™ browser version 90 and later and is best viewed with 1440 × 900 screen resolution. Google Chrome™ browser is recommended for use with the software.

English must be the primary language that is set in the web browser in which you use Genexus™ Dx Software when you download a template file for sample creation.

The operating system of the sequencer is Ubuntu™ 18.04.1 LTS.

## Installation and verification run

IMPORTANT! The Genexus™ Dx Integrated Sequencer and the Genexus™ Dx Software provide the option to perform a performance qualification (PQ) verification run.

Notifications are sent when the PQ run is expired. Contact your Field Support Engineer (FSE) or Technical Support if you have questions about an expired PQ status.

## Network and password security requirements

### Network configuration, security, and data backup

The network configuration and security settings (for example, firewalls, antivirus software, network passwords) of your laboratory or facility are the sole responsibility of your facility administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions.

If external or network drives are connected to the sequencer, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss.

If a Laboratory Information Management System (LIMS) connection is set up, you can use the LIMS API to obtain the list of files generated for the assay. For more information, see “LIMS introduction” on page 193.

Note: If a LIMS system is configured to retrieve files from the sequencer, you can manually set up an FTP directory or drive mapping. This configuration is not provided as part of sequencer installation, and must be set up by your LIMS system integration or IT group.

You must have an established setup for regularly backing up and archiving your data. Data backup is solely your responsibility. In the event your system needs repair, Thermo Fisher Scientific is not responsible for data backup or any loss of data.

## Password security

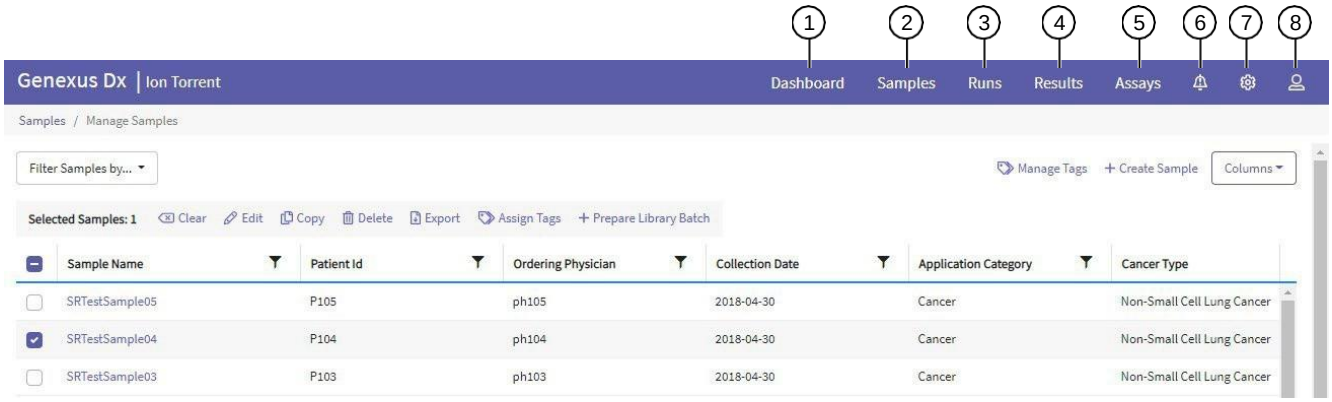
Best practice is to maintain unique passwords for all accounts in use in Genexus™ Dx Software. All passwords must be reset the first time a user signs into the software. Change passwords according to your organization's password policy.

It is the sole responsibility of your IT personnel to develop and enforce secure use of passwords. This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# 2 Get started with Genexus™ Dx Software

## About Genexus™ Dx Software

Genexus™ Dx Software provides menus to help you add, select, and manage samples, libraries, runs, and assays. You can also view and manage sequencing results, monitor Genexus™ Dx Integrated Sequencer runs in progress, and manage software settings.




1. **Dashboard**—View recent run history, and current purification or sequencing run status.
2. **Samples**—Add new samples, import samples, prepare library batches, import library batches and manage attributes.
3. **Runs**—Plan a run starting from a sample, a nucleic acid sample, or a library. View, edit, and manage runs.
   Sample to result runs are for nucleic acid isolation on a Genexus™ Dx Purification System followed by sequencing on a Genexus™ Dx Integrated Sequencer.
4. **Results**—View sample results, run results, and verification results.
5. **Assays**—View and manage assays.
6. **Notifications**—Receive alerts and messages for password expiration, system critical service failures, system backup failures, available software updates, and full disk space.
7. **Settings**—Access audit records and run logs, configure network settings, manage backup settings, restore runs, link to Connect user accounts, check for software updates, and manage data archiving, disk space, and users.
8. **Profile**—Access the Help system, manage and edit user profile settings, configure an SSH key (system administrator only), and sign out.

## Access the End User License Agreement (EULA)

A link to the End User License Agreement (EULA) is located at the bottom of the screen.

1. Scroll to the bottom of the screen.
2. Click the **EULA** link to open a PDF file of the EULA.

## Contact Support

You can access Genexus™ Dx Software technical support from the software.

1. Scroll to the bottom of the screen.
2. Click **Support** to open **https://www.thermofisher.com/contactus**, where you can request help and technical support.

# User-access levels

| Users at this level | Can perform these actions |
|---|---|
| Report | • Access the Sample Results and Run Results screens to view results<br>• Generate, view, and sign variant reports<br>• Send notifications<br>• Download results files<br>• View and edit notes for the sample result<br>• View the audit trail for sample results<br>• View notifications |
| Operator | Report functions, plus:<br>• Add, import, and export sample files<br>• Edit sample files<br>• Prepare library batches<br>• Plan, save, audit, and delete runs<br>• Monitor runs<br>• View results and reports<br>• View the audit trail for samples, library batches, runs, and assays<br>• Reanalyze runs |

*(continued)*

| Users at this level | Can perform these actions |
|---|---|
| Manager | Operator functions, plus:<br>• Create, edit, and obsolete sample attributes<br>• Import assays<br>• Create report templates<br>• Edit and delete sample files<br>• Add or edit Thermo Fisher accounts<br>• Restart a stalled or failed run<br>• Access services information |
| Administrator | Operator and manager functions, plus:<br>• View, export, and print audit records<br>• Configure network settings<br>• View and manage software updates<br>• Configure backup settings and restore runs<br>• Manage sequencer and software log files<br>• Add and manage user accounts |

# System tracking

The system tracks and checks auditable user activities for samples, assays, QC metrics and information related to reagents.

If the software detects an error at any step—for example, a scanned barcode is inconsistent with the information given for the run—the software alerts the user and does not proceed with the run.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

# Reagent management

Put in place a reagent management system to inventory and track reagent use.

Proper use of a reagent management system promotes regulatory compliance and enables optimal use of kits, chips, and reagents.

# View notifications

You can view notifications for files that you upload and import in Genexus™ Dx Software.

1. In the menu bar, click **(Notifications)**.
   If you have any unread notifications, the number of unread notifications is shown to the upper right of the notification button.
2. View the list of notifications in the dialog box that appears.
3. To delete a notification, click **(Remove)** in the section of the notification.

# Sign in to a new user account

Users must request a new user account from administrators before you can sign in to Genexus™ Dx Software. Administrators receive the request and create the user account. After the administrator creates a new account, the software sends an email to the new user with a user name and temporary password. Your user name and password must be unique and must not be shared with other users.

1. Obtain your user name and temporary password that is supplied by your administrator in an email message.
2. Browse to the Genexus™ Dx Software sign in page (for example, *https://<your server address>*), then enter your user name and the temporary password.
3. Press **Enter**, or click **Sign In**.
4. In the **End User License Agreement** that appears at first sign on, read the agreement, then click **Accept**.
5. In the **Change Password** screen, enter your temporary password, type a new password, confirm the password, then click **Change**.

The software opens to the **Dashboard**.

# Sign in

Before you can sign in to Genexus™ Dx Software, you must have a registered account. See “Sign in to a new user account” on page 17.

1. Browse to the Genexus™ Dx Software sign in page (for example, *https://<your server address>*).
2. *(Optional)* Click **Switch Language**, then select the preferred language from the list.
3. Enter your user name and password, then press **Enter** or click **Sign In**.
   If a message appears that indicates that there is a problem with the security token, refresh the browser, then enter the user name and password again.

The software opens to the **Dashboard**.

## Mode switch with a partially used chip installed

In Genexus™ Dx Software you can switch from IVD mode to Assay Development mode, or from Assay Development mode to IVD mode, when you sign in to the software. If you want to switch modes to plan and perform a run in a new mode, and you have a partially used chip installed in the sequencer, you cannot use the partially used chip and sequencing consumables in the new mode. A dialog box appears asking you to confirm the mode switch. You can cancel the mode switch, but if you proceed, you must perform an instrument clean and install a new chip and consumables. You will lose the unused chip lanes and remaining sequencing consumables.

If you want to switch modes to review run data or reanalyze data in the other mode, and do not perform a sequencing run, an instrument clean is not needed. You can proceed with the mode switch and you do not need to replace the installed chip and consumables.

## Switch the software mode

You can switch between two separate software modes in Genexus™ Dx Software:

- Assay Development mode
- IVD mode

1. Go to the Genexus™ Dx Software sign in screen.
2. In **Choose System Mode**, select the mode that you want to use.
3. Click **Switch Language**, then select the preferred language from the dropdown list that appears, if desired.
4. Enter the user name and password, then press **Enter** or click **Sign In**.

Note: If a message appears that indicates that there is a problem with the security token, refresh the browser and enter the user name and password again.

# Edit a user profile

After your user profile has been established, you can view user profile information, change a password, or change email contact information.

1. In the menu bar, click **(Profile)►My Profile**.
2. In the **Profile** screen, view the profile information.

| To | Do this |
|---|---|
| Change a password | a. Click **Change Password**.<br>b. In the **Change Password** dialog box, enter the current password and a new password. Confirm the password, then click **Change**. |
| Change an email address | a. Click **Change Email**.<br>b. In the **Set Email** dialog box, enter a new email address, and a password, then click **Save**. |
| Edit security questions and answers | a. Click **Edit Security questions and answers**.<br>b. In the **Edit Security questions and answers** dialog box, select a security question from the dropdown list, then enter an answer.<br>c. Enter the password for the account that is associated with the profile.<br>d. Click **Submit**. |

The changes are applied immediately. After the password is changed, you must sign in with the new password.

# Genexus™ Dx Software concepts

The following terms are frequently used in Genexus™ Dx Software.

For more definitions, see “Genexus™ Dx Software definitions” on page 231.

| Term or concept | Description |
|---|---|
| Sample | The information that describes the material from a single source. Each sample is defined by a set of attributes, which are categories of information that describe important aspects of each sample, such as date of sample collection, gender, or a sample type.<br>In the software, a sample can be isolated nucleic acid or a sample can be a specimen that requires integrated nucleic acid purification before sequencing. |
| Sample name | A unique identifier for a particular sample. |
| Assay | A collection of settings and parameters for library preparation, templating, sequencing, and sequencing data analysis. |
| Library | A sample associated with an assay, including barcode information and sample input quantity. Libraries are associated with an assay during library batch creation. |

*(continued)*

| Term or concept | Description |
|---|---|
| Library batch | A collection of libraries that are prepared and sequenced in the same run under the same conditions on the Genexus™ Dx Integrated Sequencer. |
| Library batch ID | A unique identifier for a library batch. |

# Workflows

| Type of workflow | More information |
|---|---|
| Sample to Result | “Sample to Result workflow” on page 21 |
| Nucleic Acid to Result<br>Library to Result | “Nucleic Acid to Result or Library to Result workflow” on page 22 |

## Sample to Result workflow

### Workflow for a Sample to Result purification and sequencing run

**View and manage assays** (page 143)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Dx Software. You can use the system-installed assays in a run without change.

**Enter and manage samples and library batches** (page 23)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as sample collection date, gender, sample type, and application category.



**Plan a Sample to Result run** (page 53)

Runs created in Genexus™ Dx Software contain the settings that are used in purification, library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

**Prepare samples and load the Genexus™ Dx Purification System Instrument (see the *Genexus™ Dx Purification System User Guide* )**

Prepare samples, then follow the step-by-step instructions in the purification instrument touchscreen for automated purification of sequencing-ready DNA and RNA samples from FFPE sections.



**Load the Genexus™ Dx Integrated Sequencer and start a run (see the *Genexus™ Dx Purification System User Guide* )**

Follow the step-by-step instructions in the sequencer touchscreen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



**Monitor the run** (page 66)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



**Review results in Genexus™ Dx Software** (page 70)

Review data and results and generate a report for each sample.

## Nucleic Acid to Result or Library to Result workflow

### Workflow for a Nucleic Acid to Result or Library to Result sequencing run

**View and manage assays** (page 143)

System-installed assays that are specifically configured for each sample type are available in Genexus™ Dx Software. You can use the system-installed assays in a run without change.

**Enter and manage samples and library batches** (page 23)

Enter samples in Genexus™ Dx Software to assign sample names and provide other information such as sample collection date, gender, sample type, and application category.



**Plan a Nucleic Acid to Result run** (page 56) **or Plan a Library to Result run** (page 59)

Runs created in Genexus™ Dx Software include the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes.

**Dilute samples and load the sample plate (see the *Genexus™ Dx Purification System User Guide* )**

Quantify and dilute the nucleic acid samples, then load the sample plate.



**Load the Genexus™ Dx Integrated Sequencer and start a run (see the *Genexus™ Dx Purification System User Guide* )**

Follow the step-by-step instructions in the sequencer touchscreen to load the sample plate and consumables in the Genexus™ Dx Integrated Sequencer.



**Monitor the run** (page 66)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.

**Review results in Genexus™ Dx Software** (page 70)

Review data and results and generate a report for each sample.



# 3 Samples and library batches

## About samples and library batches

A sample can be isolated nucleic acid or a specimen that requires nucleic acid isolation. Before you can plan a run on the Genexus™ Integrated Sequencer, you must enter samples into Genexus™ Dx Software. Samples in Genexus™ Dx Software include the data and attributes that characterize the genomic data from a single source. You add this sample information to run plans when Genexus™ Dx Software guides you through the steps to set up a run plan.

A library batch is a group of samples that are sequenced in a library run. You can create library batches from samples you have previously added or uploaded to the software. During library batch preparation, you identify the barcode adapters that were used to prepare the libraries. You add library batches to run plans when Genexus™ Dx Software guides you through the steps to plan a library run.

## Enter and manage samples

There are two ways to add samples in Genexus™ Dx Software.

- Enter information for a single sample or multiple samples into the software.
- Create a file of information for a group of samples and import that file.

If you enter the information for samples into the software, you can select from system-installed sample attributes that are available in the software. For more information, see “Create a new sample” on page 28 and “System-installed sample attributes” on page 221.

System-installed and custom sample attributes are also available in an example file that you can use to import samples. See “Download a template file for sample creation” on page 32 for more information.

---

IMPORTANT! The cancer type attribute is included in the Assay that is added to Genexus™ Dx Software, and is not available as a system attribute. Before you can create a sample in Genexus™ Dx Software, you must upload the assay that includes the cancer type attribute.

---

Samples in Genexus™ Dx Software are listed in the **Manage Samples** screen.

## Customize the samples table

The samples table is organized into columns of sample attributes. You can customize the table with table preferences to determine which columns appear. When you save the table customizations, the changes you make are saved only for your user profile.

1. In the menu bar, click **Samples►Manage Samples**.
2. Click **Columns**.
   - Select the checkbox next to each attribute you would like to display.
   - Deselect the checkbox next to each attribute you would like to remove from the table.
3. Scroll to the bottom of the list.
   - Click **Save** to save your new selections. Your selections persist when you reopen the table.
   - Click **Restore default** to restore the table to the default column settings.

## Find samples

You can sort, search, and filter information to refine and narrow the samples that are listed in the **Samples / Manage Samples** screen in Genexus™ Dx Software. The sort, search, and filter tools help you organize samples and find a samples of interest.

For information to search for a sample by a unique identifier, such as the sample name, see “Search for samples” on page 24.

For information to filter samples by a common trait, such as the date the sample was created, see “Filter the list of samples by the date of sample creation” on page 26.

### Search for samples

Samples are listed in the **Samples / Manage Samples** screen in Genexus™ Dx Software.

You can search for samples with keywords or text that are included in the sample attributes, such as sample name, tags, or application category. You can also search for a specific date or date range for the **Created On** attribute.

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IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

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1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, use the options to filter the list of sample attributes.

| Option | Steps |
|---|---|
| Limit the list of sample attributes that is shown. | Click **(Filter)** in the column heading for a sample attribute of interest, then do one or more of the following.<br>• In the search field, enter 3 or more characters for the sample attribute.<br>• Select a checkbox next to one or more sample attributes.<br>• Click **Select All** to select all sample attributes.<br>• Click **Clear** to close the filter dialog box. |
| Limit the list of samples based on a creation date, or a range of dates. | To set the criteria for the date or date range, click **(Filter)** in the **Created On** column heading, and select **Is equal to**, **Is before or equal to**, or **is after or equal to**.<br>Select a date in the calendar.<br>*(Optional)* Select **And**, or **Or** to include a second set of criteria and further narrow the search.<br>For more details, see “Filter the list of samples by the date of sample creation” on page 26. |

3. Click **Filter**.

   In the column heading, changes color to indicate that a filter is applied.

The filtered list of samples is shown in the **Manage Samples** screen.

Click **Clear Filters** at the top of the **Samples / Manage Samples** screen to remove all filters and view the full list of samples.

## Sort the list of samples

You can sort the list of samples to make it easier to find a sample in Genexus™ Dx Software.

By default, samples are listed in the **Samples / Manage Samples** screen by the date that the sample was created. The most recently created sample is at the top of the list.

You can change the order that the samples are listed in the table.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, click a column heading of interest.

   The list of samples is sorted by the sample attribute, in alphabetical or chronological order of the attribute, depending on the heading.
3. Click the column heading again to reverse the order.

## Filter the list of samples by the date of sample creation

You can apply filters to narrow search results, or shorten the list of samples when you view the list of samples in Genexus™ Dx Software.

In addition to the filter tools in the column headings, you can also filter the list of samples with a dropdown list and search by the sample name or other column headings. For more information, see “Refine the list of samples” on page 27 and “Search for samples” on page 24.

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Manage Samples** screen, select one or more filters.

| Option | Description |
|---|---|
| List only samples created on an exact date. | a. In the **Created On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| List samples created within a date range. | a. In the **Created On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date that the samples of interest were created.<br>d. In the Boolean operator dropdown list, select **And**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date that the samples of interest were created.<br>g. Click **Filter**.<br>Records are listed for samples created on dates that meet both conditions. |
| List samples according to a date that meets either one condition or a second condition. | a. In the **Created On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date that the samples of interest were created.<br>d. In the Boolean operator dropdown list, select **Or**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date that the samples of interest were created.<br>g. Click **Filter**.<br>Records are listed for samples created on dates that meet at least one of the conditions. |

| Option | Description |
|---|---|
| List only samples that were created before a date that you indicate. | a. In the **Created On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is before or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| List only samples that were created after a date that you indicate. | a. In the **Created On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column heading, changes color to indicate that a filter is applied.

The list of samples is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of samples.

## Refine the list of samples

You can shorten the list of samples that are shown in Genexus™ Dx Software.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, select the type of sample you want to view.

| Option | Description |
|---|---|
| List only the samples that have not been prepared as a library batch. | Select **To Be Prepared**. |
| List all samples. | Select **All**. |

Only the selected type of samples are listed.

## Create a new sample

You can enter information individually for each sample that you add in the software. Complete this procedure to make a sample available for use in a run, or to add the sample to a library batch. If you have many samples to add, see instead “Create multiple samples” on page 28 and “Import samples” on page 29.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Samples/Create Sample** screen, enter a name for the sample, select an **Application Category**, then complete the required fields, and optional fields if needed.
   - In the **Tags** field, enter a full or partial tag name of at least 3 characters, then select one or more tags of interest to assign a tag to the sample.
   - If the tag that you want to assign to the sample does not exist in the software, a manager-or administrator-level user can enter a new tag name in the **Tags** field to create a new tag and assign it to the sample.

   The fields for sample attributes change based on the **Application Category**. Attributes identified with a red asterisk (*) are required.

   Note: Users are recommended to add custom attributes such as "Specimen Collection Site" and "Specimen Type" (for example, resection, CNB, and FNA). For information about adding custom attributes, see “Add a custom sample attribute (manager/administrator)” on page 33.
3. Click **Save**.
   The new sample is listed in the **Manage Samples** screen and is available to use in a run plan.

## Create multiple samples

There are two ways you can create multiple new samples in Genexus™ Dx Software.

- You can enter sample information for multiple samples in a table format in the software.
- You can import a file that contains sample data. For more information, see instead “Import samples” on page 29.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.
   A table with columns that are specific for the selected application category appears.
4. *(Optional)* Click **Columns** in the upper right corner of the screen to customize the optional attributes for the samples that you want to create.
   - Select the checkbox for a sample attribute to add a column to the table. Click the row to view fields into which you can enter attribute information.
   - Deselect the checkbox for a sample attribute to remove the attribute from the table.

5. Enter the information for a sample.
   Attributes identified with a red asterisk (*) are required.
6. Click **Add Row**, then enter the information in the new row for each new sample.
7. Repeat step 6 for each new sample.
8. Select the checkbox in the row for each sample in the list that you want to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample that you will create.
9. Click **Save**.
   The new samples are listed in the **Manage Samples** screen and are available to use in a run plan.

   Note: Samples are listed in the sample plate numerically and alphabetically. If you add 01, 02, 03, and so on as sample name prefixes according to the order you need, samples are automatically arranged in the desired order in the sample plate.

## Import samples

You can enter sample information for multiple samples directly in Genexus™ Dx Software. When you want to create more than a few samples, an easy and fast way to add multiple samples in the software is to create a file of information for a group of samples and import that file.

Sample data files can be used to capture, manage, and edit sample data. You can import sample data files in TSV, XLS, XLSX, or CSV file formats. For a list of the sample attributes that are included in the import file, see “System-installed sample attributes” on page 221. For ease of use, you can download a Microsoft™ Excel™ template file to create an import file.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In the **Application Category** dropdown list, select the application category for the samples.

   Tip: Use the search field to search for the application category of interest.

4. Set up a sample file using one of two options.

| Option | Description |
|---|---|
| Download a template file, then edit it to create a new file. | For more information, see "Download a template file for sample creation" on page 32. Upload the edited file using the **Browse** button.<br><br>Note: When you select **Other** as the **Application Category**, you must enter text in the **Application Category** text box, then click anywhere in the screen in order for the template file to be available. |
| Upload the sample data from an existing file. | a. Click **Browse**.<br>b. Navigate to the file, then click **Open**.<br><br>The data contained in the file populates the table in the screen. |

5. In the sample file, edit the sample table and data, if needed.

| Option | Description |
|---|---|
| Remove an attribute column. | Click **Columns**, then deselect the column name. |
| Add an attribute column to the table. | Click **Columns**, then select the column name. The column names that are listed are **Application Category**-specific. |
| Edit sample data. | Click the sample row of interest, then edit the text field or dropdown list for the sample. |
| Add more samples to the table. | Click **Add Row**, then enter the sample data. |
| Remove a sample from the table. | Select the checkbox for the row, then click **Remove Row**. |

6. Select the checkbox in the row for each sample to create. To select all samples, select the checkbox in the column heading row.

   IMPORTANT! The information for samples that are not selected is not retained by the software. Ensure that you select the checkbox for every sample you intend to create.

7. Click **Save**.

   You can place the pointer over ⓘ **(alert)** to view more information if needed.

The new samples are listed in the **Manage Samples** screen and are available to assign to a run plan.

# View sample summary

You can view a summary of the details for the attributes of a sample. The sample summary lists the information for sample attributes for the sample and the date the sample was created in the software. For a list of the information that is shown in the sample summary, see "Key Findings" on page 74.

For more information about sample attributes, see "System-installed sample attributes" on page 221.

To view the sample summary:

1. In the menu bar, click **Samples►Manage Samples**.
2. In the **Sample Name** column, click the sample name of interest.
   Details for the sample are shown in the sample summary pane.
3. When you are finished viewing the sample details, click another sample name to view details for another sample, or click ✕ **(Close)** to return to the **Manage Samples** screen.

## Sample summary

You can view details for its attributes when you select a single sample that is listed in the **Samples/Manage Samples** screen in Genexus™ Dx Software. The sample summary lists details that describe sample attributes, such as gender and the date that the sample was created in the software.

Notes for a sample are not shown in the sample summary pane. For more information, see "View notes or add a note to a sample" on page 38.

The sample attributes that are shown in the sample summary pane depend on the application category for the sample. The following table lists the system-installed sample attributes for a sample that has an application category of cancer. Custom sample attributes are also shown in the sample summary pane, but are not listed in the following table.

| Sample attribute | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Patient Id | An identifier for the patient. |
| Date of Birth | The date of birth for the patient. |
| Ordering Physician | The name of the physician who created the order. |
| Collection Date | The date that the sample was collected. |
| Created On | The date the sample was imported or created in the software. |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Sample Type | A term that describes the sample, for example, RNA. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. 'Percent Cellularity' refers to the same measurement as 'Percent Tumor Content'. |

*(continued)*

| Sample attribute | Description |
|---|---|
| %Necrosis | The percentage of cellular necrosis in the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected.<br>Stage 0–IV, or Primary, Unknown, or Other. |

## Download a template file for sample creation

You can download a template file, add sample information to it, then use the file to import the sample data for multiple samples.

Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headings in the **Specimen Format** tab. Use the **Specimen Format** tab to enter samples.

Template files contain both the system-installed and the custom sample attributes as column headings.

1. In the menu bar, click **Samples►Create Sample**.
2. In the **Create Sample** screen, click the **Multiple (.xls, .csv, .tsv)** tab.
3. In **Application Category**, select the application category for the samples.
   Template files are specific for each application category.
4. Click **Download Template** to download the Microsoft™ Excel™ template file.
   The template file contains default sample attributes as columns. If custom sample attributes have been configured in the software, the custom attributes are added to the template file.
5. Save the file to the computer, then open the file, enter sample data in the **Sample Details** tab.
6. When you are finished adding sample information to the file, save the file.

You can now import the file to Genexus™ Dx Software. For more information, see “Import samples” on page 29.

## Add a custom sample attribute (manager/administrator)

Samples attributes in Genexus™ Dx Software are categories of information that describe important aspects of each sample, such as patient ID, date of birth, date of sample collection, gender, or a sample type. The values for each attribute define the data that describes each sample, such as RNA or DNA for the nucleic acid type attribute.

Manager- and administrator-level users can add custom attributes and add or change values of those attributes.

1. To create a custom attribute, navigate to the **Samples►Manage Attributes** screen, then click **+ Add New Attribute**.

2. In the **Add New Attribute** dialog, define the attribute in the **Attribute Name**, **Attribute Category**, and **Data Type** fields, then click **Save**. The user can also determine if the attribute is required or optional.

Custom and predefined sample attributes can be viewed in the Samples table. Each column in the Samples table represents a single sample attribute.

For more information, see “Create a new sample” on page 28, “Create multiple samples” on page 28, “Import samples” on page 29, “Edit a sample” on page 38, and “System-installed sample attributes” on page 221.

## Sample tags

A sample tag is a label that you add to a sample to help you group and track samples in Genexus™ Dx Software. For example, you can assign a tag to each sample in a study or project so that you can easily find that group of samples in the software. A manager- or administrator-level user firsts creates a tag, then any user can assign that tag to a single sample or a group of samples. You can assign multiple tags to each sample.

You can assign a tag when you create a sample or you can add, edit, or remove the tags that are assigned to a sample later. You can apply sample tags to multiple samples at one time. Later, you can filter the table of samples with that tag to list only the samples that have the tag. The tags that you assign to samples are also shown in the list of sample results so sample tags can be used to group and find sample results that are part of a study or project.

If you edit a sample and make a change to the tags associated with the sample, the changes you make are tracked in the software and can be reviewed. For more information, see “Edit a sample” on page 38 and “Review sample edit history” on page 40.

Tags are preserved with the sample when you archive a run that includes a sample with one or more assigned tags.

You can also add tags to sample results. For more information, see “Assign sample tags to a sample result” on page 101 and “Assign sample tags to multiple sample results” on page 102.

## Create a sample tag (manager/administrator)

A manager-or administrator-level user can create sample tags in Genexus™ Dx Software that can be used to label and group samples.

You can create parent tags to nest the labels if desired. For example, if a large project is divided into sections, you can create a parent tag called *Master project* and a child tag called *New variant of concern*. You can create multiple layers of nested tags to help sort projects and sections within projects. When you assign nested tags to a sample, the parent tag and all child tags are listed as tags for the sample. Later, when you search or filter for nested tags, the parent tags are also shown to help you group and track the samples.

1. In the menu bar, click **Samples**►**Manage Samples**.

   Alternatively, you can create a tag when you create a sample. For more information, see “Create a new sample” on page 28 and “Import samples” on page 29.

2. In the **Manage Samples** screen, click **Manage Tags**.

   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.

3. Click + **Create Tag**.

4. In the **Create Tag** dialog box, enter a name for the tag.

   - Select a **Parent Tag** from the list to create a hierarchical tag that is nested below the parent tag, if desired.

5. Click **Save**.

The tag is available to assign to samples.

## Assign sample tags

You can assign tags to samples in Genexus™ Dx Software to label and group samples.

1. In the menu bar, click **Samples**►**Manage Sample**.

2. In the **Manage Samples** screen, place the pointer over the row of the sample that you want to tag, then click **Assign Tags**.

3. In the **Assign Tags** dialog box, search for or scroll to a tag.

   If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click + **Create Tag** to create a new sample tag.

4. Select one or more tags that you want to assign to the sample, then click **Save**.

The tag is assigned to the sample and is listed in the row for the sample in the samples table.

## Assign a sample tag to multiple samples

You can assign tags to multiple samples at once in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, click the checkbox to the left of each sample of interest, then click **Assign Tags** above the list of samples.

   **Assign Tags** is not shown until you select at least one sample.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.

   If the tag that you want to assign to a sample does not exist in the software, a manager-or administrator-level user can click **Create Tag** to create a new sample tag.
4. Select one or more tags that you want to assign to the samples, then click **Save**.

The tag is assigned to the selected samples and is listed in the row for each sample in the samples table.

## Change or remove the tags that are assigned to a sample

You can change or remove the tags that are assigned to a sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Assign Tags**.
3. In the **Assign Tags** dialog box:
   - Deselect the checkbox next to each tag that you want to remove from the sample.
   - Select the checkbox next to each tag that you want to add to the sample.
4. Click **Save**.

## Edit a sample tag (manager/administrator)

A manager-or administrator-level user can rename a tag and change the parent tag associated with a tag in Genexus™ Dx Software to change the hierarchy of the tags. When a tag is edited, the change is applied to all samples assigned with the tag.

1. In the menu bar, click **Samples►Manage Sample**.
2. In the **Manage Samples** screen, click **Manage Tags**.

   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click **(Edit)**.

4. Change the name of the tag and parent tag assignment if needed, then click **Save**.
5. In the confirmation dialog box that appears, click **Yes** to confirm that you want to edit the sample tag.

All samples that have the tag that you edit are updated with the change. If the name of the tag is changed, the new name is shown for all samples and sample results with the tag. If the hierarchy of the tag is changed, that change is also applied to all samples and sample results with the tag.

### Delete a sample tag (manager/administrator)

A manager-or administrator-level user can delete sample tags in Genexus™ Dx Software. When a tag is deleted, the tag is also removed from any samples assigned with the tag. Additionally, when you delete a parent tag, all tags that are nested under the parent tag are also deleted.

1. In the menu bar, click **Samples**►**Manage Sample**.
2. In the **Manage Samples** screen, click **Manage Tags**.
   The **Manage Tags** screen opens and lists tags, if any, that have been created in the software.
3. In the **Manage Tags** dialog box, click **Delete**.
4. In the **Confirm** dialog box, click **Yes** to confirm the changes.

The tag is no longer listed in the **Manage Tags** list and is removed from all samples and sample results.

## Manage samples

You can find tools for creating, searching, sorting, editing, deleting, and exporting samples, and for viewing the sample history in the **Samples / Manage Samples** screen.

### Copy a sample

You can copy a sample and change the information for the sample, if needed, to create a new sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to copy, then click **Copy**.
   The **Copy Sample** screen opens. The sample information for the source sample is shown. The **Sample Name** for the new sample is the sample name of the sample that was copied, appended with _**Copy**.
3. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample name, if needed.
4. Click **Save**.

The new sample is listed in the **Manage Samples** table and is available to assign to a run plan.

## Copy multiple samples

You can copy multiple samples at once in Genexus™ Dx Software. You can only copy multiple samples that have not been assigned to a run plan. To copy samples that have been assigned to a run plan, you must copy one sample at a time. For more information, see “Copy a sample” on page 36.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
   The actions that you can apply to multiple samples appear.
3. Click **Copy**.
   The **Copy Sample** screen opens. The sample information for each source sample is shown. The **Sample Name** for each new sample is the sample name of the sample that was copied, appended with _**Copy**.
4. In the **Copy Sample** screen, edit the sample information and sample tags, and change the sample names, if needed.
5. Click **Save**.

The new samples are listed in the **Manage Samples** table and are available to assign to a run plan.

## Export samples

The **Export** function generates an XLS file that contains details about the selected sample.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to export. To select all samples, select the checkbox in the column heading row.
3. Click **Export**.
   An XLS file is created that contains the details of the selected samples. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.
4. Open the XLS file in an appropriate viewer to review or print.

## View notes or add a note to a sample

You can view existing notes or add new notes to a sample in Genexus™ Dx Software. Use notes to capture time-stamped written comments and observations for a sample. A sample can contain multiple notes from different users.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.

| Action | Description |
|---|---|
| View existing notes for a sample. | In the **Add Notes** dialog box, notes that are associated with the sample are listed by user, date, and time. |
| Add a new note to a sample. | a. In the **Add Notes** dialog box, click + **Add Note**.<br>b. Enter the note, then click **Save**.<br>The note is added to the sample and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time. |

## Delete a sample note

You can delete notes that have been added to a sample in Genexus™ Dx Software.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Notes**.
3. In the **Add Notes** dialog box, in the row of the note that you want to delete, click **(Delete)**.

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

## Edit a sample

You can edit the information that describes a sample in Genexus™ Dx Software. When you edit a sample, you can change the application category, update sample data, and remove data that are not required.

You can edit a sample until a run that uses that sample is started. You cannot edit a sample during a run. After a run is complete, you can edit a sample. However, when you edit a sample after the run is complete, the sample status for the result is changed to obsolete and the report and other files associated with the sample are no longer available.

If you edit a sample after the run and the sample result is obsolete, you must reanalyze the sample to generate sample results and a new variant report for the sample. For more information, see “Reanalyze a run” on page 119.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample that you want to edit, then click **Edit**.
3. In the **Edit Sample** screen, modify the sample information.
   See “System-installed sample attributes” on page 221 for more information about system-installed sample attributes.
4. Click **Save**.

## Edit multiple samples

You can edit multiple samples at once in Genexus™ Dx Software.

You can edit samples until a run that uses that sample is started. You cannot edit samples during a run. After a run is complete, you can edit samples. However, when you edit samples after the run is complete, the status for the sample results is changed to obsolete and the report and other files associated with the samples are no longer available.

If you edit samples after the run and the sample results are obsolete, you must reanalyze the samples to generate sample results and a new variant report for the samples. For more information, see “Reanalyze a run” on page 119.

Only samples that have the same sample application category can be edited together.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select two or more samples.
   The options that you can apply to multiple samples appear.
3. Click **Edit**.
4. In the **Edit Sample** screen, modify the sample information.
   For more information about edit options, see “Create multiple samples” on page 28.
5. Click **Save**.

The changes to the samples are saved and the edited samples are available to assign to run plans.

You can view a history of changes that have been made to each sample. For more information, see “Review sample edit history” on page 40.

## Review sample edit history

The entire history of defining a sample is available to review and export in Genexus™ Dx Software. The history shows the original sample values and any new values or activities. The following actions generate an audit record:

- Create a new sample
- Edit a sample, including changes to sample tags
- Delete a sample

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, place the pointer over the row of a sample of interest, then click **Audit**.
3. In the **Audit Details** screen:

| Option | Description |
|---|---|
| Review the audit trail details. | Each action that was performed for the sample, such as sample creation or edits, is listed. |
| Export the audit record. | Click **Download** to generate a PDF file. Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file. |
| Close the **Audit Details** screen. | Click **Back** to return to the list of samples. |

## Delete samples

You can delete samples that have not been assigned to a run. Samples that are assigned to a run or library batch cannot be deleted.

1. In the menu bar, click **Samples**►**Manage Samples**.
2. In the **Manage Samples** screen, select the checkbox in the row of each sample that you want to delete.
   The options that you can apply to multiple samples appear.
3. Click **Delete**.
   The **Delete Sample** dialog box opens with the message ***Are you sure that you want to delete selected sample(s)?***
4. Click **Yes** to delete the selected samples.

The samples are removed from the software.

# Enter and manage library batches

You can prepare a library batch manually or import library batch information from a file. To change sample or library batch information, you can edit the information in the samples, update library batches, or delete samples or library batches.

## Prepare or import a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Dx Software from samples. You can enter samples in the software or import a sample file. For more information see, “Enter and manage samples” on page 23.

Select the library batch when you plan the run. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 56.

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Note:

- Each library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

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### No-template and positive controls

A no-template control (NTC) sample and a positive sample control are included in runs that are planned in Genexus™ Dx Software. The controls are added automatically after you assign samples to a run plan, or when you create a library batch. The configuration of each control is based on the assay that is used. For more information, see the user guide for the panel that you use.

### Prepare a library batch

A library batch is a group of prepared libraries that are sequenced in the same Library to Result run. If you are planning a run starting from libraries that you have already prepared manually, you must first create a library batch in Genexus™ Dx Software from samples that you have added. If you are planning a run starting from nucleic acid samples, skip this step and proceed to “Plan a Nucleic Acid to Result run” on page 56.

Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software to prepare the library batch and verify that the barcode scanner is compatible with the computer.

1. In the menu bar, click **Samples►Manage Samples**.

2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown menu, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

   For more information about how to filter the list of samples, see “Find samples” on page 24.

3. Select samples in the list by clicking the checkbox to the left of each sample, then click + **Prepare Library Batch**.

   Note: Two control samples are included in each run. The controls are added automatically to a run plan after you assign one or more samples, or to a library batch when you create the library batch.

4. In the **Create Library batch** screen, select the assay that you want to use in the run plan from the **Select Assay** dropdown list.

   The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library preparation type, the **Library Prep Type:** automatically fills with the library preparation type for the nucleic acid type that is specified by the assay you selected. For example, **DNA+RNA**.

5. In the expanded screen, in **Library Batch ID**, enter a unique identifier for the library batch.

   Note: Library Batch IDs can contain only alphanumeric characters (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-). Required fields are indicated with a red asterisk (*).

6. *(Optional)* Enter the **Library Kit Barcode** and the **Panel Kit Barcode**.

7. Select the barcode ID of the adapter used to prepare each library. If appropriate, swap the default barcodes in the dialog box between DNA, RNA, and Fusions by clicking the **Swap Barcodes** swap image.

   For example, click DNA ⇄ Fusions to swap barcodes.

   Each library in a library batch must have a different barcode ID. When preparing the physical libraries, best practice is to swap barcodes between DNA and RNA libraries in consecutive sequencing runs to prevent carryover contamination. The barcodes that are listed in the **DNA Barcode** or **RNA Barcode** dropdown list belong to the barcode set that was selected when the assay was created.

   IMPORTANT! Ensure that the barcodes that you used to create the libraries match the barcodes that you enter in the **Create Library batch** screen.

8. *(Optional)* Enter the **Input Quantity** of each DNA+RNA library.

9. Use the connected barcode scanner to scan the barcode from the control kit into the fields in the **DNA Kit Barcodes**, and **RNA Kit Barcodes** columns for the positive controls and the no-template controls, and for each sample, if applicable.

   If needed, you can enter the barcodes manually.

10. Click **Submit** to save and submit your selections.

    The **Samples / Manage Library Batches** screen opens, listing the library batch that you created. Libraries that are prepared in the same batch have the same **Library Batch ID**.

## Import a library batch

You can import library batch information in the form of an .XLS or .XLSX file. The import file must include all required library and kit information.

1. In the menu bar, click **Samples►Manage Libraries**.
2. In the **Manage Libraries** screen, click **Import Library Batch**.
3. In the **Import Library Batch** dialog box, click **Click here** to select an assay for which the libraries are prepared, and to download an example file for import.

   If the assay includes more than one library preparation type, a library preparation type dropdown list appears.

   a. Select the library preparation type, such as **DNA+RNA** for the library batch.
4. Select an assay from the list, then click **Download**.

   The assay name is auto-populated in the Microsoft™ Excel™ template file that downloads to your drive.
5. In the template file, enter or confirm the library batch information.

| Template item | Description |
|---|---|
| Reagents tab | |
| Assay Name | Auto-populated when assay is selected in step 4 (required) |
| Extraction Method Type | Auto-populated when assay and library prep type, if applicable, is selected in step 4 (required) |
| Library Batch ID | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required) |
| Library Kit Barcode | For example, barcode (optional) |
| Panel Kit Barcode | For example, Oncomine™ Dx Express Test barcode (optional) |
| Libraries tab | |
| Sample Name | Must be alphanumeric (0–9, Aa–Zz), period (.), underscore (_), and hyphen (-) (required) |
| Barcode | Barcodes used for each sample and control library preparation (required) |
| Nucleic Acid Type | DNA+RNA (required) |
| Input Quantity | Library input quantity (optional) |
| Control Kit Barcode | The barcode for the kit used for nucleic acid extraction of no template controls (required) |
| Extraction Kit Barcode | The barcode for the kit used for nucleic acid extraction of a sample (required) |

IMPORTANT! For DNA and Fusions assays, the DNA library and RNA libraries must be listed in sequential order per pool for each sample. For example, for a 1-pool DNA and Fusions assay, order should be DNA, RNA for sample 1, DNA, RNA for sample 2. For a 2-pool DNA and Fusions assay, library order should be DNA, RNA (pool 1), DNA, RNA (pool 2) for sample1, then DNA, RNA (pool 1), DNA, RNA (pool 2) for sample 2.

6. Save the file.
7. In the **Import Library Batch** dialog box, click **Select Library batch file**, navigate to the saved file, then select it.
8. Click **Upload**.

## Find library batches

You can sort, search, and filter information to narrow the library batches that are listed in the **Samples / Manage Library Batches** screen in Genexus™ Dx Software. The sort, search, and filter tools help you organize the library batches and find a specific library batch.

For information to search for a library batch by a unique identifier, such as the library batch ID, see “Search for library batches” on page 44.

For information to filter libraries by a common trait, such as the type of nucleic acid in the library batch, see “Filter the list of library batches” on page 46.

### Search for library batches

Library batches are listed in the **Samples / Manage Library Batches** screen in Genexus™ Dx Software. The most recently created library batch is at the top of the list.

You can search for library batches with full or partial keywords of the following items that are associated with the library batches.

- **Library Batch ID**
- **Assay Full Name**
- **Nucleic Acid Type**
- **Sample Name**

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, in the column heading of interest, click **(Filter)**.

3. In the search field, enter a full or partial name or ID of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.

4. In the filter dialog box, select from the limited list.
   - Select a checkbox next to the name or ID to select one or more items.
   - Click **Select All** to select all of the items listed in the filter dialog box.
   - Click **Clear** to close the filter dialog box.

5. Click **Filter**.
   In the column heading, changes color to indicate that a filter is applied.

The filtered list of libraries is shown in the **Manage Library Batches** screen.

Click **Clear Filters** to remove all filters and view the full list of library batches.

## Sort the list of library batches

You can sort the list of library batches to make it easier to find a library batch in Genexus™ Dx Software.

By default, library batches are listed in the **Samples / Manage Library Batches** screen by the date the library batch was created. The most recently created library batch is at the top of the list.

You can change the order that the library batches are listed and sort the library batches alphabetically or reverse alphabetically by the **Library Batch ID** or **Assay Full Name**.

1. In the menu bar, click **Samples►Manage Library Batches**.

2. In the **Manage Library Batches** screen, click the column heading of interest.
   The list of library batches is sorted alphabetically.

3. Click the column heading again to reverse the order.

## Filter the list of library batches

You can apply filters to narrow search results, or shorten the list of library batches when you view the list of library batches in Genexus™ Dx Software.

You can also search for library batches with keywords or text. For more information, see “Search for library batches” on page 44.

In addition to the filter tools in the column headings, you can also filter the list of library batches with a dropdown list. For more information, see “Manage library batches” on page 47.

1. In the menu bar, click **Samples►Manage Library Batches**.
2. In the **Manage Library Batches** screen, select one or more filters.

| Option | Description |
|---|---|
| List all library batches that are associated with a sample. | In the **Total Samples** column heading, click **(Filter)** then select one or more samples of interest. |
| Filter and search by the **Library Batch ID** or other column headings. | In the **Library Batch ID** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter by the type of nucleic acid in the library. | In the **Nucleic Acid Type** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter by the full assay name in the library batch. | In the **Assay Full Name** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Filter the list of all samples that are included in library batches. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column heading, changes color to indicate that a filter is applied.

The list of library batches is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of libraries.

## Manage library batches

You can view library batch information, edit, delete, add notes, and review the history of changes to library batches in Genexus™ Dx Software.

You can also search, sort, and filter the list of libraries batches. For more information, see “Find library batches” on page 44.

1. In the menu bar, click **Samples**▶**Manage Library Batches**.
2. In the **Manage Library Batches** screen, perform one or more actions.

| Option | Description |
|---|---|
| View more library batch information, such as the list of samples included in the library batch. | Click a Library Batch ID in the library batches list. Alternatively, in the Total Samples column, click the number of samples in the library batch.<br>In the pane that shows details for the library batch, you can click ••• (More Options) to perform more actions, such as edit, audit, or add notes to the library batch. |
| View all library batches. | In the Filter Library Batches By dropdown menu, select All. |
| View libraries that are not in a run plan. | In the Filter Library Batches By dropdown menu, select To Be Planned. |
| View multiple pages of libraries. | Click the page numbers and arrows at the bottom of the screen. |
| Delete a library batch. | Select the checkbox next to one or more library batch IDs, then click Delete. |
| Edit a library batch. | 1. Place the pointer over the row of a library batch that you want to edit, then click Edit.<br>2. Modify the information in the Edit Library batch screen, then click Submit.<br>Alternatively, in the library batch details pane, click the library batch ID to perform this action.<br>You can edit a library batch only if none of the libraries in the library batch have been assigned to a run. |
| View or export the audit trail of a library batch. | 1. Place the pointer over the row of a library batch that you want to audit, then click Audit.<br>The Audit Records screen opens with time-stamped audit records that include details and changes for each action performed on the library batch.<br>2. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions.<br>3. Click Download in the upper right corner of the screen to download the record as a PDF file. |

*(continued)*

| Option | Description |
|---|---|
| View, add, or delete notes for a library batch. | **1.** Place the pointer over the row of a library batch of interest, then click Notes.<br>The Add Notes dialog box opens and notes that are associated with the library batch are listed by user, date, and time.<br>**2.** In the Add Notes dialog box, click + Add Note.<br>**3.** Enter the note, then click Save.<br>**4.** If needed, in the Add Notes dialog box, in the row of the note that you want to delete, click (Delete). |

# 4 Plan and manage runs

Runs created in Genexus™ Dx Software contain the settings that are used in library preparation, templating, sequencing, and analysis, including sample information and plate location, assays, and barcodes. Runs are used to track samples, consumables, and chips throughout the library preparation, templating, sequencing, and data analysis workflow.

There are two types of runs that you can plan in Genexus™ Dx Software.

Types of runs in Genexus™ Dx Software

| Run type | Description |
|---|---|
| Sample to Result | An integrated run for sequential and automated nucleic acid purification and sequencing. This run type requires the Genexus™ Dx Purification System. |
| Nucleic Acid to Result | A sequencing run that uses nucleic acid samples as input. |
| Library to Result | A sequencing run that uses libraries that you have previously prepared as input. |

Genexus™ Dx Software guides you step-by-step through the process to set up a run. The software prompts you to select required information and consumables, then provides a printed run setup guide to help you load consumables on the Genexus™ Dx Integrated Sequencer and the Genexus™ Dx Purification System, if applicable.

You can plan sequencing runs that use nucleic acid samples with a **Nucleic Acid to Result** run or that use libraries prepared in a previous Genexus™ Dx Integrated Sequencer run with a **Library to Result** run.

You can plan runs for sequential and automated nucleic acid purification and sequencing with a **Sample to Result** run. You must connect at least one Genexus™ Dx Purification System to execute a **Sample to Result** run. For more information, see “Integrated nucleic acid isolation in Sample to Result runs” on page 52.

# Before you plan a run

The following conditions must be met before you plan a run in Genexus™ Dx Software.

- Sample information is accurate in the software and the name assigned to each sample is unique. For more information, see “Enter and manage samples” on page 23.
- A Genexus™ Dx Purification System is set up for use in an integrated configuration before you plan a **Sample to Result** run.
- Ensure that the following conditions are met for a **Library to Result** run.
  - Library batches are prepared and each batch uses a unique library batch ID.
  - Each sample library in a library batch is prepared with and assigned a unique barcode or barcode pair.

  For more information, see “Enter and manage library batches” on page 41.

The software returns an error message when any of the conditions are not met.

# Find a run plan

You can sort, search, and filter information to narrow the run plans that are listed in the **Runs / Manage Runs** screen. The sort, search, and filter tools help you organize runs and find a specific run plan in Genexus™ Dx Software. You can also apply the search, sort, and filter tools in the **Assays** step when you plan a run. Use the tools to limit the assays that are listed so that you can easily identify one or more assays to include in the run plan.

For information to search for a run plan by a unique identifier, such as the name of the run plan or the assay applied to the run plan, see “Search for a run plan” on page 50.

For information to filter run plans by a common trait, such as the run plans that include purification, see “Filter the list of run plans” on page 52.

## Search for a run plan

Run plans are listed in the **Runs / Manage Runs** screen in Genexus™ Dx Software. The most recently created run plan is on the top of the list.

You can search for run plans with keywords or text that are included in the name of the run plan or the name of an assay applied to the run plan.

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IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

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1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, in the **Run Name** column heading or **Assay Name** column heading, click **(Filter)**.
3. In the search field, enter a full or partial assay or run name of at least 3 characters.
   When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
4. In the filter dialog box, select from the limited list.
   - Select a checkbox next to a run or assay name to select one or more items.
   - Click **Select All** to select all items listed in the filter dialog box.
   - Click **Clear** to close the filter dialog box.
5. Click **Filter**.
   In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of run plans is shown in the **Manage Runs** screen.

Click **Clear Filters** to remove all filters and view the full list of run plans.

## Sort the list of run plans

You can sort the list of run plans to make it easier to find run plans in Genexus™ Dx Software.

By default, run plans are listed in the **Runs / Manage Runs** screen by the date the run plan was created. The most recently created run plan is at the top of the list.

You can change the order that the run plans are listed and sort the run plans alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, click the **Run Name** heading.
   The list of runs is sorted alphabetically.
3. Click the column heading again to reverse the order.

## Filter the list of run plans

You can apply filters to narrow search results, or shorten the list of run plans when you view the list of run plans in Genexus™ Dx Software. You can also apply filters in the **Assays** step when you plan a run to help find one or more assays to include in the run plan.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the run name or the assay selected in the run. | For more information, see “Search for a run plan” on page 50. |
| Filter by the number of samples in the run. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run. |
| List runs based on whether the run includes purification. | In the **Run Status** column heading, click **(Filter)** then select a status.<br>• Select **Planned** to list all **Nucleic Acid to Result**, **Library to Result**, and **BAM to Result** planned runs.<br>• Select **Purification Planned** to list planned **Sample to Result** runs only. |
| Remove all selections and close the filter dialog box. | In the filter dialog box, click **Clear**. |

3. In the filter dialog box, click **Filter** to apply the filtered selections.
   In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The list of run plans is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of run plans.

# Integrated nucleic acid isolation in Sample to Result runs

A **Sample to Result** run is an integrated run for sequential and automated nucleic acid purification and sequencing. Administrators, managers, and operators can create **Sample to Result** runs for the formalin-fixed, paraffin-embedded (FFPE) tissue sample type.

Core needle biopsy (CNB) and fine needle aspiration (FNA) samples are considered FFPE sample types in the software and can be grouped with FFPE tissue sections in a nucleic acid isolation batch. The Genexus™ Dx Purification System can run one nucleic acid isolation batch at a time.

For more information, see the *Genexus™ Dx Purification System User Guide* (Pub. No. MAN1000137).

## Plan a Sample to Result run

This procedure applies only if a Genexus™ Dx Purification System is used with the Genexus™ Dx Integrated Sequencer.

Ensure that the following prerequisites are complete before you plan a **Sample to Result** run.

- Integrate a Genexus™ Dx Purification System with the system.
- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 28, or “Import samples” on page 29.
- Connect the barcode scanner to the computer on which you use Genexus™ Dx Software to plan runs and verify that the barcode scanner is compatible with the computer.

Planning a **Sample to Result** run is organized into five steps: **Setup**, **Assays**, **Samples**, **Purification**, and **Review**. Progress through the steps is tracked in the upper left corner of the **Runs / Sample to Result** screen.

1. In the menu bar, click **Runs**►**Sample to Result**.

   Note: You can also click + **Sample to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.
      The name is limited to 50 characters and no spaces are allowed.
   b. *(Optional)* In the **Reporting (Optional)** section, enable **Generate Report** to generate a variant report that uses the default report template.
   c. Click **Next**.

3. In the **Assays** step, select the assay that you want to include in the run.
   Only assays that are configured for **Sample to Result** runs are listed. If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.
   a. Use the ▼ **(Filter)** tools in table column headings to find assays of interest, if desired.
   b. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include the research application for the assay in the run plan.
   c. Click **Next**.

4. In the **Samples** step, select the samples that you want to run with each research application of each assay.
   a. Select the checkbox next to each sample that you want to assign to the research application of an assay, then in the **Selected Assays** pane, for the assay and research application that you want to use for the selected samples, click **Assign**.

      Note: Two control samples are included for each assay. The controls are added automatically to a run plan after you assign one or more samples.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including a no template control, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

If the

*minimum reads per sample* × *number of samples*

exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not achieve the required reads per sample to pass QC metrics.




Green lane color denotes lane usage and sample assignment within lane capacity.




Red lane color denotes sample assignment that exceeds lane capacity.

b. If needed, edit samples in one of the following ways.
   - Click **View & Remove**, make the selections, then click **Update**.
   - Click **Remove All**, to remove all sample assignments for all assays.

c. Click **Next**.

5. In the **Purification** step, review and edit selections.

   a. Review and edit purification selections.

| Option | Description |
|---|---|
| Protocol Selection | Select a protocol from the dropdown list for each purification kit, if more than one is available. |
| Elution Vol. (µL) | Modify the elution volume within the allowable range, if needed. |

   b. Use the connected barcode scanner to enter the control kit barcodes in the **Kit Barcodes** fields.

   If needed, you can enter the barcodes manually.

   c. Click **Next**.

6. In the **Review** step, review the run plan summary, then click **Save**.

   Note: A run can include multiple purification batches.
   For more information, see “Integrated nucleic acid isolation in Sample to Result runs” on page 52.

   The run plan summary lists the following details:

   - the consumables that are required for purification
   - how much volume to load
   - where to load samples
   - the consumables that are required for sequencing
   - details about the assay or assays.

   Tip: Click **Sequencing** to expand the sequencing section of the run plan summary.

   IMPORTANT! The consumables required for sequencing can change. After purification, review the run plan in the **Runs / Manage Runs** screen for updated sequencing consumable information. For more information, see “Print a run plan” on page 62.

   The run plan appears in the **Manage Runs** screen with the name that you specified.

After you select the run and load the Genexus™ Dx Purification System, the run is started on the purification instrument screen.

When purification and sequencing are complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*.

## Review samples for Sample to Result runs

A **Sample to Result** run integrates nucleic acid purification and sequencing. When you create a **Sample to Result** run plan, you can include an option that allows samples to be reviewed before library preparation starts. Samples that do not meet a specified concentration threshold after purification have a run status of **Purification Review Required**.

For more information about **Sample to Result** runs, see “Integrated nucleic acid isolation in Sample to Result runs” on page 52.

1. In the menu bar, click **Results►Run Results**.

   **Sample to Result** runs that require review after purification have a **Run Status** of **Purification Review Required**.

2. In the **Run Results** screen, place the pointer over the row of the run plan of interest, then click **Review**.

3. In the **Purification Samples** screen, review the concentration for each sample, then select the checkbox in the row for each sample that you want to sequence.

4. Click **Submit**.

The samples are ready for sequencing.

## Abort a Sample to Result run

When you create a **Sample to Result** run plan, you can include an option that allows samples to be reviewed before library preparation starts. When you enable the review of samples after purification, you can end the run before sequencing if needed.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, place the pointer over the row of the run that you want to abort, then click **Abort**.
3. In the confirmation dialog box that appears, click **Yes** to confirm that you want to abort the run.

In the Run Summary, you can view information about the purification run. The status of the run in the Run Summary is Aborted. For more information, see “View the run summary” on page 104.

# Plan a Nucleic Acid to Result run

You can plan a run for sequencing that starts with nucleic acid samples. A run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

Planning a **Nucleic Acid to Result** run is organized into steps: **Setup**, **Assays**, **Samples**, **Sample Plate**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Nucleic Acid to Result** screen.

Ensure that the following prerequisites are complete before you plan a **Nucleic Acid to Result** run.

- Before you begin this procedure, connect the barcode scanner to the computer on which you use Genexus™ Dx Software and verify that the barcode scanner is compatible with the computer.
- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 28, or “Import samples” on page 29.
- Ensure that the assay that you want to use in the run exists in the software. For more information, see “Assays in Genexus™ Dx Software” on page 143.
- Quantify sample concentration, if you are not diluting sample concentrations manually to the target concentration of the assay. For more information, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide* (Pub. No. MAN0024909).

1. In the menu bar, click **Runs**►**Nucleic Acid to Result**.

   Note: Alternatively, you can click **+Nucleic Acid to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.

   b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.

   c. Click **Next**.

3. In the **Assays** step, select the assay that you want to include in the run.
   a. Use the **(Filter)** tools in table column headings to find assays of interest, if desired.
   b. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include the research application for the assay in the run plan.
   c. Click **Next**.
4. In the **Samples** step, select the samples that you want to run with each research application of each assay.
   a. Select the checkbox next to each sample that you want to assign to the research application of an assay, then in the **Selected Assays** pane, for the assay and research application that you want to use for the selected samples, click **Assign**.

      The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples (including a no-template control, if selected) and the minimum read counts per sample for the assay. Green denotes a chip lane in the run containing assigned samples within lane capacity.

      If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Assign** asking you to confirm that you want to continue. After confirmation, the **Chip View** updates and shows the lane color as red instead of green. The run is allowed if the lane capacity is exceeded, but you may not obtain the reads needed per sample to pass QC metrics.




      Green lane color denotes lane usage and sample assignment within lane capacity. Red lane color denotes sample assignment that exceeds lane capacity.

   b. If needed, edit samples in one of the following ways.
      - Click **View & Remove**, make the selections, then click **Update**.
      - Click **Remove All**, to remove all sample assignments for all assays.
   c. Click **Next**.

5. In the **Sample Plate** step, review position assignments in the sample plate. Drag-and-drop samples and no-template controls to edit the location of samples and controls, if applicable.
   a. Use the connected barcode scanner to enter the kit barcode for one or more samples or controls. For a single sample, in the row of the sample of interest, in the **Kit Barcodes** column, enter the extraction kit barcode or control kit barcode, if applicable. For multiple samples or controls, select the samples or controls, then click **Assign Kit Barcodes**. In the **Assign Kit Barcodes** dialog box, enter the kit barcode.

      If needed, you can enter the barcodes manually.

   b. Modify the concentration of samples, if needed. For a single sample, in the row of the sample of interest, in the **Conc. (ng/µl)** column, edit the concentration.

      To modify the concentration of multiple samples, select the samples of interest, then click **Edit Concentration**. In the **Bulk Edit** dialog box, enter the concentration for all selected samples, then click **Submit**. The concentration for each selected sample is updated to the new value.

      If a sample concentration is ≤1,024X the target concentration for the assay, which is displayed as a default value for each sample in the **Sample Plate** screen, the sequencer automatically dilutes the sample to the target concentration during the run. If a sample concentration is greater than this value, you need to manually dilute the sample to the target concentration, or to a value within range for automated dilution before loading on the sample plate.

      ---

      Note: The sample volume that is required for library preparation is not adjustable. The volume depends on the number of primer pools in the assay, sample type, and library chemistry.

      ---

      ---

      IMPORTANT! A warning symbol (⚠) with a message that states “Sample concentration out of range” can be shown in error for automated sample dilution in the **Run Plan** screen when you plan a run, or view the **Run Summary** screen, in Genexus™ Dx Software.

      - If the sample concentration is below the required minimum concentration and shows a **Dilution Factor** of 1.00 (that is, no dilution), the message is valid.
      - If a sample that includes the warning message shows a **Dilution Factor** >1.00 and <1,024.00, the message is *not valid* and can be ignored.

      ---

   c. Ensure that sample plate information is correct, then click **Next**.

6. In the **Review** step, review the run plan summary, then click **Save & Print** to print the run setup guide, if desired. Click **Save** to save the run without printing.

   ---

   Note: To print the run setup guide, you need to allow pop-ups in the browser.

   ---

   The run plan summary lists the following details.

   - the consumables that are required for this run
   - how much sample volume to load
   - where to load samples and primer pool tubes
   - details about the assay.

   After saving, the run appears in the **Manage Runs** screen in the run list with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

When the sequencing run is complete, you can recover the leftover volume from each library that was prepared in the run. For more information to find, recover, and purify the leftover library preparations, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide* (Pub. No. MAN0024909).

# Plan a Library to Result run

You can plan runs for sequencing that start with libraries. A run that starts with prepared libraries is called a **Library to Result** run. Before you can plan a **Library to Result** run, you must first enter sample information and prepare a library batch that associates an assay with the library batch. The library batch must be created with the assay to be used in the run. The assay specifies the barcode set that was used to prepare the sample libraries.

For more information, see “Enter and manage library batches” on page 41.

Genexus™ Dx Software guides you through the four steps of planning a **Library to Result** run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

1. In the menu bar, click **Runs**►**Library to Result**.

   Note: You can also click + **Library to Result** in the **Runs / Manage Runs** screen.

2. In the **Setup** step, enter a name for the run, then configure the reporting options.
   a. In **Run Name**, enter a unique name.

      The name is limited to 50 characters and no spaces are allowed.

   b. *(Optional)* In the **Reporting (Optional)** section, ensure that **Generate Report** is enabled to generate a variant report using the default report template.

   c. Click **Next**.

      If a chip is installed in the sequencer, the **Chip View** graphic in the lower left corner indicates the lanes that are available for sequencing.

3. In the **Assays** step, select the assay that you want to include in the run.

   For the assay to be selectable at this step, you need to have prepared a library batch that assigns the assay to the batch. The assay specifies the barcode set that was used to prepare the sample libraries.
   a. Use the ▼ **(Filter)** tools in table column headings to find assays of interest, if desired.

   b. In the **Research Application** column for the assay of interest, select one or more research applications, such as **DNA and Fusions**, to include each selected research application for the assay in the run plan.

   c. Click **Next**.

4. In the **Library Batches** step, select the library batch that you want to use in the run, then click **Next**.

   Note: Only one library batch can be selected per assay. However, you can plan a library run that uses multiple assays if you select multiple compatible assays in the **Assays** step.

   The **Chip View** updates to show the lanes to be used in the run as green. Lane usage is calculated based on the number of samples and minimum reads per sample entered at assay setup.




   Green lane color denotes lane usage and sample assignment within lane capacity. Red lane color denotes sample assignment that exceeds lane capacity.

   If the *minimum reads per sample* × *number of samples* exceeds the chip or lane well capacity, a dialog box appears after you click **Next** asking you to confirm that you want to continue. After clicking **Yes**, the **Chip View** updates and shows the lane color as red instead of green. In the example shown at right, eight samples were included in a library batch instead of five. The run is allowed if the lane capacity is exceeded, but you may not obtain the required reads per sample to pass QC metrics.

5. In the **Review** step, review the run plan summary.

   - Click **Save and Print** to print the run setup guide.
   - Click **Save** to save the run without printing.

   Note: To print the run setup guide, you need to allow pop-ups in the browser.

   The run plan summary lists the consumables that are required for the run, where to load the library batch on the sample plate, and how much library volume to load.

   The run appears in the run list on the **Manage Runs** screen with the name you specified.

After you select the run and load the sequencer, the run is started on the sequencer screen.

# Manage runs

Run plans are listed in the **Runs / Manage Runs** screen. The most recently created run is on the top of the list. Only runs that have not started are listed.

In this screen, you can:

- Plan a run.
- Sort and search the list of run plans.
- Review and print a run plan.
- View the audit trail of a run plan.
- Edit a run plan.
- Delete a run plan.
- View, add, or delete the notes for a run plan.

## View a run plan

You can view detailed run plan information in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, click the run name of interest.
   Run plan details are shown in the run plan summary pane. For more information, see “Run plan details” on page 61.
3. When you are finished viewing the run plan details, click another run name to view details for another run plan, or click ✕ **(Close)** to return to the **Manage Runs** screen.
4. *(Optional)* Click **Print** to print the information about the run.
   To print the run plan, you must allow pop-ups in the browser.

You can click ••• **(More Options)** to perform more actions, such as edit, add notes, or audit the run plan. The actions for each run plan are also available in the **Manage Runs** screen when you place the pointer over the row of a run name. For more information, see “Edit a run” on page 64, “Add a note to a run” on page 64, “Review and download a run history” on page 63.

### Run plan details

Detailed run plan information is shown in the run plan summary pane.

| Run plan detail | Description |
|---|---|
| Generate Report | Indicates if a report is automatically generated for the run. |
| Run Status | The run status. For example, Planned or Purification Planned. |
| Run Start Date | This information is available when the run is started. |
| Run Completion Date | This information is available when the run is completed. |
| Sequencing[1] | |
| Consumables Required For This Run | A list of the required consumables, for example, the chip and barcodes. The quantity of each consumable is shown in parentheses before the name of the consumable. |

*(continued)*

| Run plan detail | Description |
|---|---|
| Consumables Installed Ready To Use | A list of consumables installed on the sequencer and available for the run. |
| Primer Tube Positions | An image that shows the locations of the primer pool tubes. |
| Sample Locations | Detailed sample information and an image that shows sample locations on the 96-well plate. For more information, see “The run summary” on page 105. |
| Assays | |
| Assay | Detailed assay information for each assay in the run. For more information, see “The run summary” on page 105. |

[1] The Sequencing section is collapsible and expandable.

Additional information for Sample to Result runs

| Run plan detail | Description |
|---|---|
| Purification[1,2] | |
| Consumables Required For Purification Batch X | A list of the required consumables. For example, the Quantitation Plate and Quantitation Tubes. The quantity of each consumable is shown in parentheses before the name of the consumable. Information for each purification batch is listed in a separate table. |
| Input Sample Locations For Purification Batch X | Detailed sample information and an image that shows sample locations on the 96-well plate. Information for each purification batch is listed in a separate table. |

[1] The Purification section is collapsible and expandable.
[2] For information about purification batches, see “Integrated nucleic acid isolation in Sample to Result runs” on page 52.

## Print a run plan

You can print detailed run plan information in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, click a run name in the list.
3. Click **Print** to print the information about the run plan.
   To print the run plan, you must allow pop-ups in the browser.

Run plan details are shown in the run plan summary pane. You can review the information that was entered or generated when you created the run. For more information, see “Run plan details” on page 61.

## Review and download a run history

The entire history of a run plan is available to review, export, or print in Genexus™ Dx Software. The history shows the original run plan values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Audit**.
   The **Audit Details** screen opens with time-stamped audit records that include run plan details and changes for each action performed on the run plan. Details for the most recent action are shown.
3. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions.
4. Review the audit record.

   Details in the audit record

| Item | Description |
|---|---|
| User name | The name of the person who was signed in when the action was performed. |
| Run Plan | The name of the run plan. |
| Data Object Name | The data object that is affected by the action. |
| Action Performed | Actions performed on the run plan include:<br>• Create<br>• Edit |
| Time Stamp | The time and date of the action. |
| ID | An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier. |
| New Values | A list of all values in the run plan. If a value changed as a result of the action, the new value is shown. |
| Old Values | A list of the previous values in the run plan. |

5. Click **Download** to generate a PDF file, if needed.
   Depending on the browser settings, the software automatically downloads the file or prompts you to open or save the file.
6. Click **Back** to return to the list of runs.

## Edit a run

You can edit runs that have a run status of **Planned** or **Purification Planned**. Parameters that can be edited include reporting options, assays used in the run, samples included in the run, chip assignment, and sample concentration.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Edit**.
3. In the **Edit Sample Run** workflow, change the desired settings by navigating through the steps clicking **Next**, then click **Save**.

   For more information about the settings, see “Plan a Sample to Result run” on page 53, “Plan a Nucleic Acid to Result run” on page 56 and “Plan a Library to Result run” on page 59.

## Delete a run

You can delete runs from the software.

You must select at least one run in the **Manage Runs** screen before the option to delete a run appears.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, select one or more runs to delete by selecting the checkbox next to the run name.

   Select all of the runs in the screen by selecting the checkbox above the column.

   If needed, click **Clear** to deselect all of the runs.
3. Click **Delete**.
4. In the **Delete Plan** message box, click **Yes** to delete the selected run.

## Add a note to a run

You can add a note or notes to **Nucleic Acid to Result** and **Library to Result** runs that have a run status of **Planned**, or **Sample to Result** runs that have a run status of **Purification Planned** in Genexus™ Dx Software. A note is any written comment or observation that you want to add to the run.

1. In the menu bar, click **Runs**►**Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, click **Add Note**.
4. In the **Add Notes** dialog box, enter the note.
5. Click **Save**.

The note appears in the **Add Notes** pane of a selected run in the list of runs.

## View a run note

You can view the notes that have been added to a run. Run notes are time-stamped written notes to capture comments or observations for a run that is planned or completed. A run can contain multiple notes from different users.

You cannot edit run notes. However, you can add multiple notes to a run.

1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. Review the note or notes information.

## Delete a run note

You can delete a run note in Genexus™ Dx Software.

Notes record any written comments or observations for a run plan.

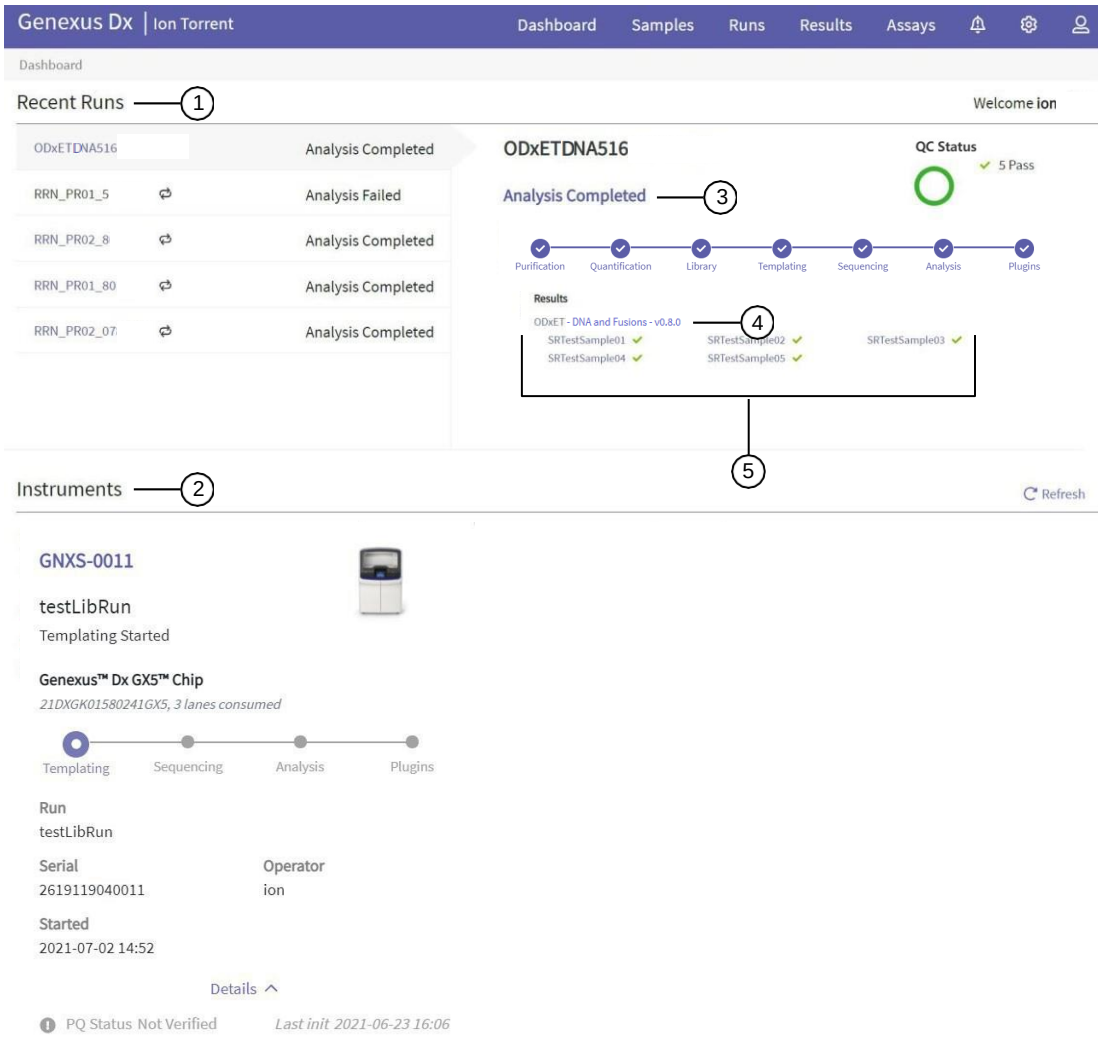
1. In the menu bar, click **Runs►Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, in the row of the note you want to delete, click **(Delete)**.

The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

# 5 Genexus™ Dx Software Dashboard

In the **Dashboard** screen you can view instrument status and details for recent and current runs.




① The five most recent runs are listed in the left pane.

- You can click the row of a run of interest to view details for the run in the right pane.
- You can click the name of the run to leave the dashboard and view the results for the run.

② Instruments that are integrated with the software are shown.

③ In the details pane on the right, you can place the pointer over the status for the run to view the progress of the run.

④ For the recent run that is selected in the left pane, each assay for the run is shown. You can click the name of an assay to leave the dashboard and open the results for the run and view assay metrics.

⑤ For the recent run that is selected in the left pane, each sample in the run is shown. You can click a sample name to leave the dashboard and open the sample results.

# View run progress on the instrument

You can view details for the five most recent and current runs in the **Dashboard** in Genexus™ Dx Software.

Runs that are reanalyzed are listed with ⟲ **(Reanalysis)** after the run name. For more information, see “Reanalyze a run” on page 119.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

| Option | Description |
|---|---|
| View detailed run result information for completed runs. | In the left side of the screen, click the run name of interest.<br>The **Run Summary** tab opens. For more information, see “The run summary” on page 105.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the quality control information. | In the left side of the screen, click the row of the run of interest.<br>The QC status for the run and for the samples is shown in the details pane in the right side of the screen. For more information, see “QC results” on page 93. |
| View assay-level metrics and results for the run. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, click the assay name of interest.<br>The **Assay Metrics** tab opens. For more information, see “Assay metrics and the run report” on page 109.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the sample results. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, click the sample name of interest.<br>The **Key Findings** tab opens. For more information, see “Key Findings” on page 74.<br>In the menu bar, click **Dashboard** to return to the **Dashboard**. |
| View the progress of the run. | a. In the left side of the screen, click the row of the run of interest.<br>b. In the run details pane in the right side of the screen, place the pointer over the status for the run.<br>A progress bar for the run is shown. Steps in the run that are complete are listed with a check mark. Steps that are in progress are shown with a flashing circle. Steps that remain to be completed are listed in grey. If any failures occur, failed steps are also listed in grey. |

Tip: If the recent run includes many samples, limit the samples that are listed. For more information, see “Filter the list of samples shown in the Dashboard” on page 68.

## Filter the list of samples shown in the Dashboard

You can apply filters to shorten the list of samples shown in the **Dashboard** in Genexus™ Dx Software.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard**, in the **Recent Runs** section, click the row of the run of interest.
   Assays and samples in the run are listed in the details pane in the right side of the screen. In addition, the QC status and number of samples that passed or failed QC are shown.
3. In the details pane, filter the list of samples.

| Option | Description |
|---|---|
| Shorten the list of samples. | Click **(Filter)**, select the samples of interest, scroll to the bottom of the filter dialog box, then click **Filter**.<br>**(Filter)** changes color to indicate that a filter is applied. |
| Refine the samples shown by the QC status of the sample result. | In the details pane, in the **QC Status** section, click **Pass** to list only samples that passed QC metrics or click **Fail** to list only samples that failed QC metrics. |

The list of samples is filtered according to the selections.

Click **Clear Filters** to remove all filters and view the full list of samples.

# View the status of an instrument

In the **Dashboard**, when a run is in progress, you can view the status of the Genexus™ Dx Integrated Sequencer and any Genexus™ Dx Purification System that are connected with Genexus™ Dx Software. You can see if a run is in progress, and view details of the run, except for the run name if the run is not within your organization.

1. In the menu bar, click **Dashboard**.
2. In the **Dashboard**, in the **Instruments** section, view the status of an instrument.
   The following information is provided for all connected instruments.
   - Instrument name
   - Run status of the current or most recent run
   - PQ status—Indicates whether the performance qualification (PQ) status of the instrument is **Verified**, **Not Verified** or **Expired**.
   - **(Alarms)**—Instrument alarms, if any
     If the instrument displays one or more alarms, the number of alarms is shown in parentheses. You can place the pointer over **Alarms** to view more information about an alarm.

3. If a run is in progress, view the details of the run. In the **Instruments** section, in an instrument pane, click **Details**.

   Tip: Check the status of a purification instrument for **Run completed waiting user input** to know when you must perform an action to proceed through the run.

4. Click **Refresh** to update the information in the **Instruments** section.

# Information about connected instruments

When a run is in progress, information about the instruments that are connected to Genexus™ Dx Software is available in the **Dashboard**. You can expand the **Details** section for each instrument to view more information about a run in progress.

| Item | Description |
|---|---|
| Run | The name of the run that is in progress or the last completed run on the instrument.<br>A string of asterisks (*) for the run name indicates that the instrument is in use by another organization. |
| Serial | The serial number of the instrument. |
| Operator | The name of the operator who started the run on the instrument. |
| Started | The date and time that the run started on the instrument. |
| Completed | The date and time that the run completed on the instrument. |
| PQ Status Not Verified | This message appears if PQ has not been performed. |
| Last Init | The last initialization completion date and time. |

# 6 Review data and results

Use the **Results** menu to review results and data analysis, and to perform data management tasks. You can view results sorted by sample or by run.

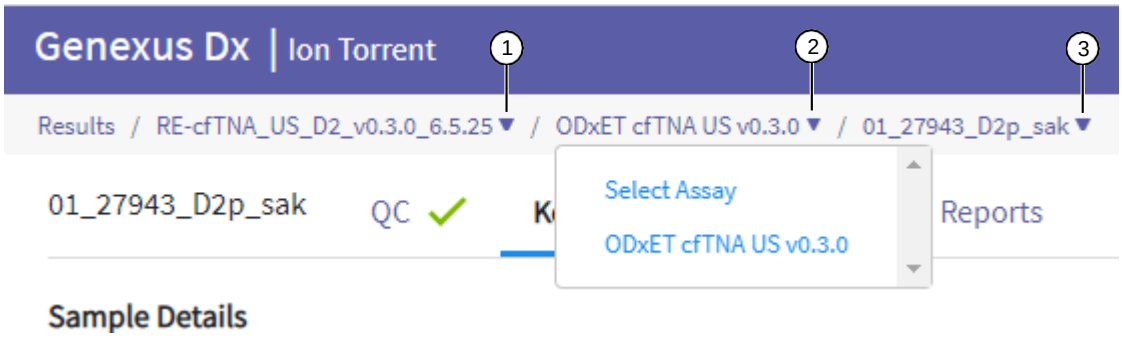
| Selection | Description |
|---|---|
| Click Results▶Sample Results | Select this option to review completed sample results and reports. |
| Click Results▶Run Results | Select this option to review completed run results and reports by assay. |
| Click Results▶Verification Results | Select this option to review data from completed verification runs that were performed during sequencer installation or performance qualification. |

## Results navigation bar overview

After you select a run result or a sample result, a results navigation bar appears and allows you to easily toggle between different results screens for each run. You can quickly switch between run-level results, assay-level results, and sample-level results for the run. In addition, you can easily find samples or assays in the run with the search field that appears in each dropdown list.

Assay-level results include assay metrics, such as final read data.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.



| Callout | Dropdown menu | Description |
|---|---|---|
| 1 | Run name | The run name is listed. Multiple runs are listed only if the run has been reanalyzed. For more information, see “Reanalyze a run” on page 119. |
| 2 | Select Assay | You can click the assay name of interest to view assay metrics for the run.<br>You can search for or toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed.<br>You can select Select Assay from the dropdown menu to remove the assay selection. If you remove the assay and sample selections, the Run Summary tab opens with a summary of results for the run selected in the Run Results dropdown menu. Assay metrics are also shown in the Run Report tab. For more information, see “Assay metrics and the run report” on page 109. |
| 3 | Select Sample | You can click a sample name to view the sequencing results for the sample. You can click a different sample name to view other sample results for other samples in the run for the assay that is selected in Select Assay. The search field allows you to find samples in the run.<br>You can select Select Sample from the dropdown menu to remove the sample selection. If you remove the sample selection, the Assay metrics tab for the selected assay opens. If you select Select Assay with Select Sample shown, the Run Summary tab opens. Assay metrics are also shown in the Run Report tab. For more information, see “Assay metrics and the run report” on page 109. |

# Review sample results

You can review completed sample results and reports in Genexus™ Dx Software.

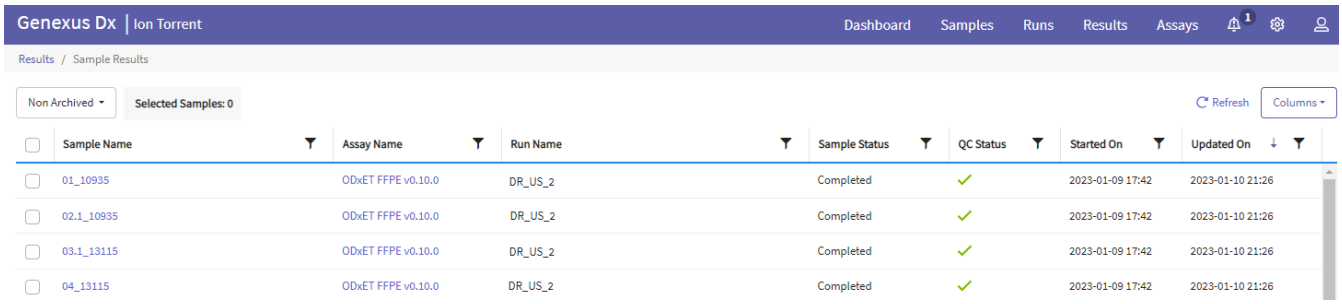
See “Find sample results” on page 96 for information about how to find sample results of interest.

After you open a sample result, you can view different results screens for the run that allow you to toggle between run-level results, assay-level results, and sample-level results. For more information, see “Results navigation bar overview” on page 70.

Assay-level results include assay metrics, such as final read data. Assay metrics are also shown in the **Run Report** tab. For more information, see “Assay metrics and the run report” on page 109.

Samples that are run with the same assay in the same run share the same run report. All other results are specific to the sample and are shown in the sample results.

1. Click **Results►Sample Results** to view sequencing results, including variant and fusion calls, for a selected sample.
   In the **Results / Sample Results** screen, samples that have been sequenced are listed by sample name.




The following information appears in the **Sample Results** screen. You can add columns to the **Sample Results** table to view more information. For example, you can add a column for **Electronic Signatures** to view signature and lock information for variant reports. For more information, see “Customize the samples table” on page 24.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. Click the Sample Name to open the Sample Results screen for the sample. |
| Assay Name | The name of the assay used in the run plan. |
| Run Name | The unique name of the run given when it was created in the software. |
| Sample Status | The status of the run or sample (for example: Completed, Running, Failed, Terminated, Pending, Stalled). |
| QC Status | The QC status of a completed run.<br>• ✔ (Passed) indicates that the sample passed all QC metrics.<br>• ✘ (Failed) indicates that the sample failed a QC metric.<br>• — (Not Calculated) indicates that a sample did not undergo QC analysis. |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |
| Tags | Tags that are assigned to the sample are listed. |

2. In the **Sample Name** column, click a sample name.

   The **Results** screen opens to the **Key Findings** tab, showing the key findings. You can click other tabs to view QC metrics, variants, and reports. For more information, see “View key findings” on page 73, “View the QC results” on page 92, “View variant results” on page 75, and Chapter 7, “Create, view, and sign reports”.

You can also perform the following actions in the **Sample Results** screen.

| Option | Description |
|---|---|
| Actions | Available action links for a sample are shown when you place the pointer over the row of a sample.<br>• Report—Download the report (available only for samples with a sample status of completed).<br>For more information, see "Clinical Test Report and Clinical Laboratory Report" on page 126.<br>• Assign Tags—Add or remove the tags assigned to a sample result.<br>For more information, see "Sample tags" on page 33.<br>• Audit—View the audit trail for the run.<br>For more information, see "Review and download a run history" on page 63.<br>• Notes—View, add, or delete notes for sample result.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>For more information, see "Download a Customer Support Archive" on page 225.<br>• Reanalyze—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step.<br>For more information, see "Reanalyze a sample" on page 120. |
| Actions that you can perform when you select samples | Assign Tags—Assign a tag or tags to selected sample results in one action. The Assign Tags command appears above the Sample Results table after you select one or more samples. |

## View key findings

You can view sample details, biomarker results, and a summary of key metrics for genes that were assayed in a selected run in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.

Review the key findings. For more information, see "Key Findings" on page 74.

## Key Findings

The **Key Findings** table shows details about the sample, a list of biomarkers reported for the sample, and a summary of key metrics for genes assayed in the run.

Note: The **Key Findings** table is by default the first view that is shown for sample results.

The information that is displayed depends on the assay that was used in the run.

Sample Details

| Sample Detail | Description |
|---|---|
| Sample Name | A unique identifier representing the sample. |
| Collection Date | The date that the sample was collected. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Sample Type | A term that describes the sample, for example, FFPE, DNA, DNA & RNA |
| Application Category | The application type of the sample. |
| Cancer Type | The type of cancer that is represented by the sample. |
| Cancer Stage | The stage of the cancer from which the sample was collected. |
| %Cellularity | The percentage of tumor cells out of the total number of nucleated cells in the sample. "Percent Cellularity" refers to the same measurement as "Percent Tumor Content." |

Biomarker Results

| Result category[1] | Description |
|---|---|
| Level 1: Companion Diagnostics (CDx) Biomarkers | Gene variants that have companion diagnostic claims in the corresponding cancer type that are associated with an FDA-approved therapy as listed in Table 1 of the intended use. |
| Level 2: Cancer Mutations with Evidence of Clinical Significance | Gene variants with evidence of clinical significance that are not prescriptive or conclusive for labeled use of a specific therapeutic product, and for which clinical validation has not been performed, but which have curated clinical evidence associated with FDA-approved therapies and/or National Comprehensive Cancer Network (NCCN) clinical guidelines (Category 1 and 2A) in the sample cancer type. |
| Level 3: Cancer Mutations with Potential Clinical Significance | Gene variants not considered Level 1 or Level 2 biomarkers that can be described as cancer mutations with potential clinical significance. These variants are not prescriptive or conclusive for labeled use of a specific therapeutic product, and clinical validation has not been performed. They have curated clinical evidence associated with NCCN clinical guidelines Category 2A in a different cancer type or Category 2B in any cancer type. They may also have curated evidence as an enrollment criterion in clinical trials or biomarkers with evidence in peer-reviewed literature. |

[1] For more information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 130.

Key Metrics

| Metric | Description |
|---|---|
| Amplicon Summary | |
| Average Base Coverage Depth | The average number of reads of all targeted reference bases. This is the total number of base reads on target divided by the number of targeted bases, and therefore includes any bases that had no coverage. |
| Uniformity Of Base Coverage | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. Cumulative coverage is linearly interpolated between the nearest integer base read depths. |
| Percent Reads On Target | The percentage of filtered reads that are mapped to any targeted region relative to all reads mapped to the reference. A read is considered on target if at least one aligned base overlaps at least one target region. If no target regions (file) was specified, this value will be the percentage of reads passing uniquely mapped and/or non-duplicate filters, or 100% if no filters were specified. |
| Uniformity Of Amplicon Coverage | The percentage of amplicons that had at least 20% of the average number of reads per amplicon. Cumulative coverage is linearly interpolated between nearest integer read depth counts. |

## View variant results

You can view detailed variant results in the **Variants** table. In the default view, all variants are listed. You can download the information for the variants in XLS or TSV format.

You can also limit the types of variants that are shown and toggle between different variant results: SNVs/Indels, Fusions, and CNVs.

Variant Tables

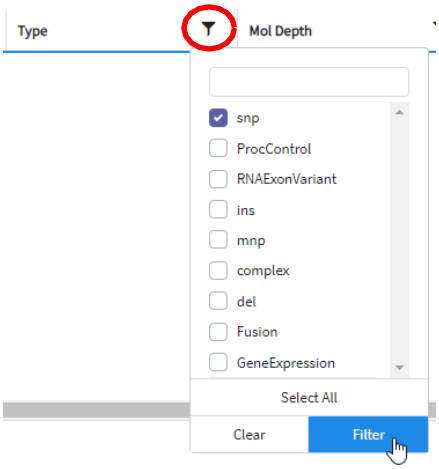
| Variant type | Description |
|---|---|
| SNVs/Indels | • SNVs: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included.<br>• Indels: insertion and deletion variants.<br>For more information, see “View SNV/Indel results” on page 77. |
| Fusions | Translocations of genetic material and splicing variants.<br>For more information, see “View fusion results” on page 79. |
| CNVs | Copy number variations (CNVs) are variations of the number of copies of a given gene.<br>For more information, see “View CNV results” on page 84. |

## Search and filter the list of variants

You can search and filter to immediately narrow the list of results that are shown in the variant tables for the selected sample. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters are applied.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name in the **Sample Name** column.
3. In the **Results** screen, click the **Variants** tab.
4. Select the variant class to display the results: **SNVs/Indels**, **Fusions**, or **CNVs**.
5. In the table of variants, in the column heading of interest, click **(Filter)**.
   - In the search field, enter at least 3 characters, then click **Filter**.
   - Select the checkbox in the row of each filter that you want to apply, then click **Filter**.

   The options that are available depend on the column and variant class. For example, you can filter data in the **Type** column to show one specific variant type.
6. Click **Clear Filters** to remove all filters and view the full list of run results.




The column or columns to which you applied a filter change to reflect the filter and selected options.

## Customize the view of the variants table

The **Variants** table includes detailed variant results in Genexus™ Dx Software. You can customize the table to view the information of your choice.

## Variants table column options

When you view the **Variants** table in Genexus™ Dx Software, a default set of columns is included. The table columns are based on various factors, including the samples that are sequenced and the assay used for sequencing.

## Add or remove columns in the variants table

You can add columns to the **Variants** table that you view in Genexus™ Dx Software.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to view results for the sample.

3. Click **Columns**, then select the columns that you want to add to the **Variants** table, or deselect the columns that you want to remove from the table.
   The selected columns are added to the **Variants** table until you close the table.
4. To save the custom table so that the changes to the columns persist the next time the table is viewed, click **Save**.

## Change the width of columns in the variants table

You can change width of columns in the **Variants** table in Genexus™ Dx Software. Not all columns are visible on the screen. You can use the scrollbar under the table to view more columns.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Change the width of each column, as desired.

## Restore default settings for the variants table

You can restore default column headings in the **Variants** table in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click **Columns►Restore default**.

The default columns shown in the table are restored.

## View SNV/Indel results

The **SNVs/Indels** table lists the calls and other information for the SNV and INDEL variants that are analyzed in each sample in a run.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **SNVs/Indels** to view the **SNVs/Indels** table.
   For more information about the data shown in the table, see “SNVs/Indels table” on page 78.
5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the SNV/Indels variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the SNV/Indels variant table, and information for table columns that are available but not currently shown in the SNV/Indels table.

## SNVs/Indels table

The data displayed in the **SNVs/Indels** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 76.

| Column | Description |
|---|---|
| Variant ID | The name of the hotspot as defined in the Browser Extensible Data (BED) file.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 86. |
| Variant Name | The name of the variant. |
| Locus | The chromosome and position of the detected variant. |
| Gene | The gene name. |
| AA Change | Identification of the amino acid change using Human Genome Variation Society (HGVS) nomenclature. |
| Ref | The reference base or bases at that locus. |
| Alt | The alternate base or bases at that locus. |
| Type | The type of variant that is detected.<br>• snp (single nucleotide polymorphism)<br>• mnp (multi-nucleotide polymorphism)<br>• ins (insertion)<br>• del (deletion)<br>• complex |
| Test Result | Indicates the presence or absence of an SNV/Indel variant.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—Indicates the absence of a variant that differs from the reference allele at a given position. |
| Call Details | The reason why a variant is reported as NO CALL. |
| Phred QUAL Score | The relative probability of either the "reference" hypothesis interval [0,cutoff], or the "variant" hypothesis interval [cutoff,1], Phred-scaled (-10*log10). A higher score means more evidence for the variant call. |
| Raw Read Depth | Total read coverage across amplicon containing SNV/Indel hotspot locations. Count of chip-level reads aligned at this locus that participate in variant calling. |

*(continued)*

| Column | Description |
|---|---|
| Effective Read Depth | The number of reads covering the position. |
| Alt Allele Read Counts | The number of reads containing the alternate allele. |
| Nuc Change | The position and identity of the nucleic acid change. |
| Allele Frequency | The number of variant read counts divided by the total number of read counts for the sample. |
| Allele Frequency (%) | The allele frequency, represented as a percentage. |
| Mut/WT | The ratio of mutant allele to wild-type allele. |
| PPA | Possible Polyploidy Alleles (PPA).<br>A value of Yes indicates variants that are PPA alleles. A value of No indicates variants that are not PPA alleles. |

## View fusion results

You can view calls and other information for the fusions that are analyzed within each sample in a run.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. Click **Fusions** to view the **Fusions** table.
   For more information about the data displayed in the table, see “Fusions table” on page 80.
5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the Fusions variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the Fusions variant table, and information for table columns that are available but not currently shown in the Fusions table.

## Fusions table

The data displayed in the **Fusions** table depends on the assay that was used in the run.

Results in the table can be filtered using filtering tools. For more information, see “Search and filter the list of variants” on page 76.

| Column | Description |
|---|---|
| Variant ID | The name of the fusion target as defined in the BED file.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 86. |
| Variant Name | The name of the variant. |
| Locus | The chromosome positions in the reference genome that define the fusion junction. |
| Genes (Exon) | The name of fusion target and representative acceptor and donor exons. |
| Read Counts | The frequency of the fusion detected in the sample. |
| Type | Assay type (for example, Fusion, RNA exon variant (exon skipping), RNAExon Tile, Proc Control). |
| Test Result | Indicates the presence or absence of a fusion or RNA exon variant.<br>• POSITIVE—indicates a high confidence call that passes all filter thresholds at a given variant position.<br>• NO CALL—although some evidence for the presence of a fusion exists, the call does not pass one or more filters that are required for a high confidence fusion call.<br>• NEGATIVE—indicates the absence of a fusion due to the variant call not passing read or call thresholds. |
| Call Details | The reason for reporting a fusion as NO CALL or NEGATIVE. |
| Read Counts Per Million | The number of fusion read counts detected per million total reads. |
| Gene Isoform | The name of the fusion target as defined in the BED file.<br>Note: For fusion assays, the value for Gene Isoform is identical to that of Variant ID, |
| Mol Cov. Mutant | The median molecular coverage across a fusion amplicon. |

*(continued)*

| Column | Description |
|---|---|
| Imbalance Score[1] | Each fusion gene exhibits a characteristic Imbalance Score threshold. Scores that exceed the threshold value indicate a high likelihood of the presence of the fusion in the sample. Imbalance score calculation starts with the normalization of reads in the exon-tiling amplicons of the gene, followed by correction with a baseline that represents expression values in normal samples. Reads for observed imbalance scores come from samples. Baseline scores come from normal, fusion-negative samples and are stored in the exon tile assay baseline file for the assay.<br>Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline)<br>where<br>Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples)<br>• 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample.<br>• 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample.<br>Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline)<br>• 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene.<br>• 5' of breakpoint is the total normalized baseline values of the amplicons for the gene. |
| Imbalance P-Value[1] | The statistical significance of measure of imbalance relative to a control gene. |
| Predicted Break-point Range[1] | The exonic range for predicted fusion break point in exon tiling assays. |
| Ratio To Wild Type[1] | The molecular ratio for exon skipping assays relative to wild type control amplicons. |
| Norm Count Within Gene[1] | Exon skipping assay coverage normalized to molecular coverage of wild type (WT) MET control amplicons. |

[1] Column appears only in analyses that use the exon tiling fusion detection method.

## View RNA Exon Variants

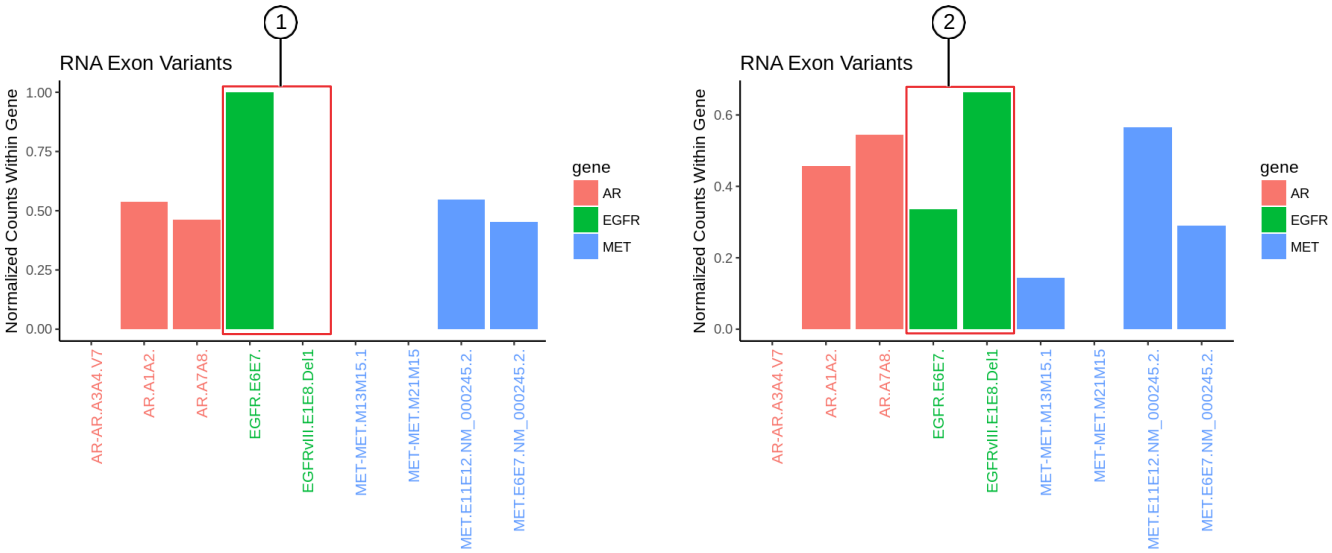
The **RNA Exon Variant** data view displays a bar graph summary of intragenic exon rearrangements or fusions. The displayed RNA exon variants are defined in the BED file that is associated with an assay. The **RNA Exon Variant** data view is available for all RNA and fusion assays.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.

3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 80.

4. In the top right corner of the screen, click **Visualization►RNA Exon Variant**, then review the **RNA Exon Variants** plot.

Representative RNA Exon Variant plots




The X-axis represents specific exon variants, where each variant is labeled with a gene ID followed by a sequence of adjacent exons. The Y-axis measures the read counts for each variant, normalized to the wild type.

① Example result where only the wild type EFGR (EFGR.E6E7) was detected.

② Example result where RNA exon 2–7 deletion occurred in the EFGR gene. The deletion of exons 2–7 resulted in an increase of normalized read counts for the EFGR variant that contains the intragenic fusion of exon 1 and exon 8 (EFGR.E1E8.Del1) and a decrease of normalized read counts for the wild type EFGR (EFGR.E6E7).

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View RNA Exon Tile Fusion Imbalance

The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

Driver genes, for example ALK, FGFR2, and NTRK1, have amplicons that span exon-exon junctions to probe the expression difference between the 3’ and the 5’ regions of the gene.

An imbalance score is calculated for each driver gene in a sample to quantify the magnitude of the expression imbalance change between the two parts of the gene. For example, the 3’:5’ ratio in ALK; and the 5’:3’ ratio in FGFR2.

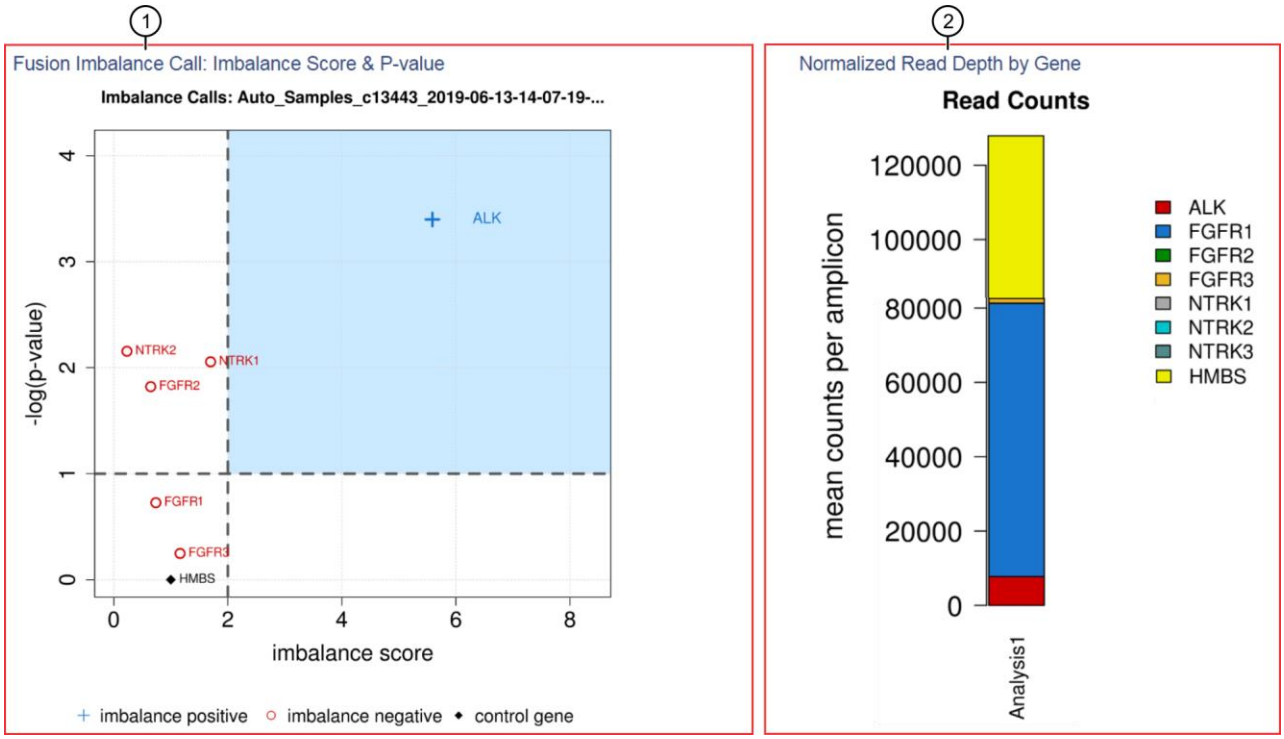
1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample name** column, click the name of the sample of interest.

3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 80.

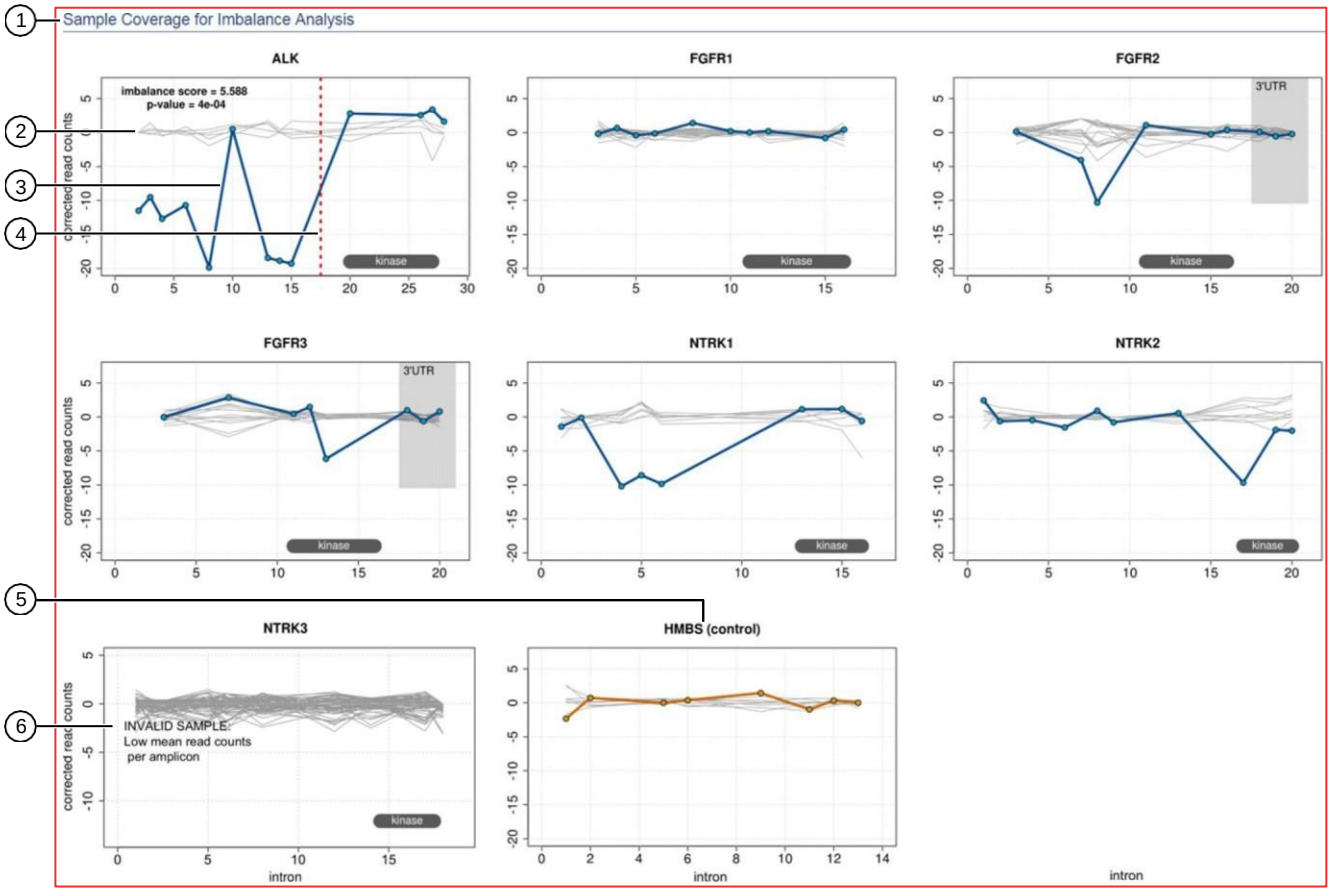
4. In the top right corner of the screen, click **Visualization►RNA Exon Tile Fusion Imbalance**, then review the **RNA Exon Tile Fusion Imbalance** plots.

Representative RNA Exon Tile Fusion Imbalance plots




① The **Fusion Imbalance Call: Imbalance Score & P-value** plot shows the imbalance scores and p-values for all the genes in the selected sample. The dashed gray lines mark the threshold for an imbalance call, which is applied to all genes across all samples. Points that fall within the blue shaded area of the plot represent fusion-positive genes ( ✚ ). All other points that are outside of the blue shaded area represent fusion-negative genes ( ◯ ). Control genes are marked with ◆.

② The **Normalized Read Depth by Gene** plot shows the mean read counts of each gene that is captured on the chip for the selected sample. For each gene, the read counts are normalized to the number of amplicons.




1. The **Sample Coverage for Imbalance Analysis** plots show the expression profile for each exon-exon tiling amplicon for each gene. The Y-axis represents the corrected molecular counts. The X-axis represents individual exon-exon junctions, which are listed from 5′ to 3′. The **imbalance score** and **p-value** are listed in the panel of each gene that was called positive for fusion.
2. Baseline (a cluster of gray lines), generated from a fusion-negative sample.
3. Test sample corrected read coverage (blue line), normalized to the baseline. Each point on the line represents a unique exon-exon junction that was covered by the assay and normalized to the baseline.
4. Predicted range for the fusion break point for a fusion-positive gene (dashed red line).
5. Sample coverage profile for the control gene (orange line).
6. If the collected data are insufficient to determine an imbalance score, the **INVALID SAMPLE** message appears in the panel for that gene.

To return to the table view of fusions, click ✕ **(Remove)** next to the **Visualization** dropdown menu.

## View CNV results

The **CNVs** table lists the calls and other information for the copy number variants (CNVs) analyzed in each sample in a run.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.

4. Click **CNVs** to view the **CNVs** table.

   For more information about the data displayed in the table, see “CNVs table” on page 85.

5. Click **Export** in the upper right corner of the screen to export the results data in XLS or TSV format, if needed.
   - The file in XLS format contains the information that is shown in the CNVs variant table.
   - The file in tab-separated value format (TSV) format contains the information that is shown in the CNVs variant table, and information for table columns that are available but not currently shown in the CNVs table.

## CNVs table

The data displayed in the table depends on the assay that was used in the run.

Results in the table can be filtered using the filtering tools. For more information, see “Search and filter the list of variants” on page 76.

IMPORTANT! *(FFPE samples only)* If the **%Cellularity** value for a sample is set to <100, then the magnitude of copy number gain can be decreased. For more information, see “System-installed sample attributes” on page 221.

| Column | Description |
|---|---|
| Variant ID | The identifier of the CNV variant.<br>Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 86. |
| Variant Name | The name of the variant. |
| Locus | The starting position of the first amplicon covering the CNV gene. |
| Gene | The gene name. |
| Copy Number | The copy number of a CNV gene locus per genome. This column is available when a positive call is made. |
| Test Result | Indicates the presence or absence of a CNV.<br>• POSITIVE—Indicates a high confidence call that passes all filter thresholds.<br>• NO CALL—Although some evidence for the presence of a variant exists, the call does not pass one or more filters that are required for a high confidence variant call.<br>• NEGATIVE—The absence of a variant; result is below the detection threshold for a CNV call. |
| P-Value | The statistical significance of the CNV ratio measurement. |
| Call Details | The reason for reporting a CNV as NO CALL. |
| CNV Confidence | The CNV confidence interval associated with the call. The 5% lower confidence bound value is the ploidy estimate, where there is a 5% chance that the true ploidy is below that value. The 95% upper confidence bound is the ploidy estimate, where it is 95% certain that the true ploidy is below that value. |

*(continued)*

| Column | Description |
|---|---|
| CNV Ratio | The ratio of measured CNV gene locus coverage relative to coverage of non-CNV loci. |
| Valid CNV Amplicons | The number of amplicons spanning the CNV call. |

## View more annotations and annotation sources

In addition to the annotations that are included with Genexus™ Dx Software, you can view annotations and annotation source information from public websites for each variant.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click the sample name in the **Sample Name** column in the row of a sample of interest.
3. Click the **Variants** tab.
4. In the **Variants** table, click the **Variant ID** of the variant of interest.
   You can click the row even if a **Variant ID** is not listed.
   The **Pileup** view is shown below the variants table. For more information, see “Review the variant pileup view” on page 86.
5. Click the **Annotations** tab.
6. Review annotation information in the **Annotations** tab, and if available, click a link in the row of an annotation to open a website with information about the annotation.

## Review the variant pileup view

You can review variant tracks with the pileup view in Genexus™ Dx Software. The pileup view is a figure that the software generates to show each BAM track represented by a single bar, with the sequence variation or variations denoted in each track. The BAM tracks are aligned against a reference track. The variant pileup view can be used to identify potential variants.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

The pileup view is shown below the variants table.

## Change the view of the variant pileup

You can zoom and pan the variant pileup view and add or remove guides and track labels to more easily understand the data. You can also adjust the view of the individual pileup tracks. For more information, see “Adjust pileup tracks” on page 87.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

| Action | Procedure |
|---|---|
| Track the position as you place the pointer over the variant pileup. | Click **Cursor Guide**. |
| Mark the center of the variant pileup. | Click **Center Line**. |
| Remove the track labels. | Click **Track Labels**. |
| Zoom in to increase the magnification of the image. | Click ⊕ **(Zoom in)** one or more times, or double-click the image. |
| Zoom out to decrease the magnification of the image. | Click ⊖ **(Zoom out)** one or more times. |
| Pan across the image to move the view to another location on the screen with its current magnification. | Click and drag the pointer over the image. |

## Adjust pileup tracks

You can adjust the BAM read coverage track, the reference track, hotspot tracks, the target region track, and annotation tracks in Genexus™ Dx Software to make it easier to view the data. When you adjust a track, it changes the amount of data that is shown in the plot. For example, you can see more tracks when you use the **Squish** setting.

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Note: For best results, click the **Expand** option to view details about variants and base calls that are shown in each read coverage track

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1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙(Actions) next to a track, then select an option to adjust the view of the track.

| Option | Description |
|---|---|
| **Expand** | Select this option to view the tracks with the maximum visible height. |
| **Squish** | Select this option to view the tracks with the minimal height for each track. |
| **Collapse** | Select this option to view the tracks with overlapping transcripts shown along a single line. When **Collapse** is selected for BAM tracks, the BAM tracks are not shown; only the coverage density is shown. |

## Adjust the view of the reference track

You can adjust the view of the reference track in Genexus™ Dx Software to view or hide the three-frame translation of the reference track sequence. You can also view the forward or reverse reference track sequence.

1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.
5. In the **Variants** tab, in the **Pileup** subtab, click ⚙(Actions) next to the reference track.

| Option | Description |
|---|---|
| View the reverse strand | Click **Reverse**.<br>This option is available only when the forward strand is shown. |
| View the forward stand | Click **Forward**.<br>This option is available only when the reverse strand is shown. |
| View the reference sequence translation | Click **Three-Frame Translate**.<br>This option is available only when the amino acid sequence is not shown. |
| Hide the reference sequence translation | Click **Close Translation**.<br>This option is available only when the amino acid sequence is shown. |

## Adjust the view of the BAM tracks

You can adjust the view of the BAM tracks in Genexus™ Dx Software to dynamically customize the view of the data.

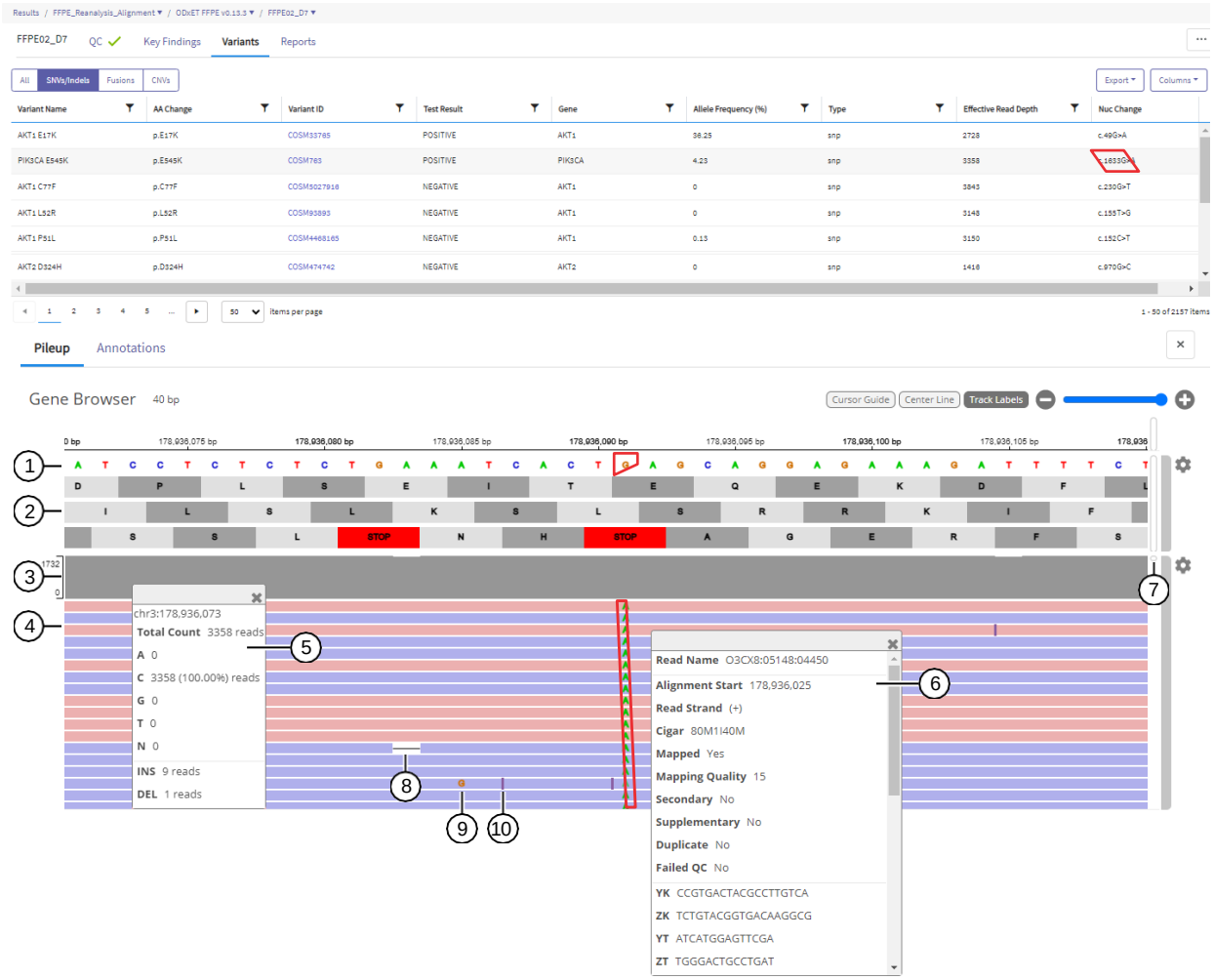
1. In the menu bar, click **Results**►**Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. In the **Variants** tab, in the **Pileup** subtab, click ⚙(Actions) next to the BAM tracks.

| Option | Description |
|---|---|
| View all BAM tracks in monochrome gray | In **Color By**, deselect the checkmark next to **Read Strand**.<br>This option is available only when the BAM tracks are colored by read strand. |
| Distinguish between the direction of the BAM tracks with color | In **Color By**, click **Read Strand**.<br>This option is available only when the BAM tracks are not colored by read strand. |
| Expand, squish or collapse the BAM tracks | For more information, see “Adjust pileup tracks” on page 87. |
| Show all the bases in the BAM tracks | Click **Show all bases**.<br>This option is available only when all bases in the BAM tracks are not shown. |
| Change the minimum threshold for the visibility of BAM tracks | a. Click **Set visibility window**.<br>b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks.<br>When you zoom in and out to view the variant pileup, the scale of the tracks shown in the screen changes. For more information, see “Change the view of the variant pileup” on page 87. You can adjust the visibility window according to the scale shown in the screen. |

## Examples of a variant pileup view

The following figure is an example of a variant pileup view, shown in the **Gene Browser** in the **Variant** tab. The variant pileup view graphically represents the sequencing tracks, the reference track, and the annotation tracks, and can help you understand the variant data and analyze results. The reference allele and the alternate base for the selected variant are indicated in the image, both in the variant table and the in the pileup view. You can customize your pileup view. Reorder the tracks displayed by clicking a gray bar on the right edge of a track, then dragging the track to a different location. You can also click-drag the cursor at any position in the image to move the image left or right in the screen. For more information, see “Adjust pileup tracks” on page 87.




1. The reference track. When zoomed out, the nucleotide sequence is no longer displayed, but the sequence is represented by colored bars.
2. The three-frame translation is shown in this example (not by default). For more information, see “Adjust the view of the reference track” on page 88.
3. The coverage track.
4. The BAM track.
5. Click the coverage track to see detailed information, such as the total count, the number of reads, and the number of molecules.
6. Click a BAM track to see detailed information, such as the **Mapping Quality** and **Base Quality**.
7. Use the slider to view more tracks.
8. Deletion.
9. Alternate base in the BAM read coverage track.
10. Insertion.

The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.




① The hotspot track.
② The target region track.
③ Click a hotspot track for additional details, such as the start and end positions.
④ Click the target region track for additional details, such as the start and end positions.

## Coverage histogram

When you zoom in on the variant pileup view, you see the coverage histogram and reads from the .bam track. The histogram in gray shows read depth at that location and reads from the .bam track show read direction and location of variants.




① Read depth
② Read location and direction of variants

## View the variants from an Oncomine™ Dx Express Test run

You can view a summary of data about the identified variants, and toggle to other views that provide more details about the same variants.

1. In the menu bar, click **Results►Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

5. Review detailed variant data and read coverage tracks.




**Example visualization**

Ion AmpliSeq™ HD sequencing runs group consensus reads into families. A family is a group of reads that are associated with the same DNA molecule before library amplification. Each family is identified using the molecular tags, and consensus reads with the same molecular tags are grouped into the same family. The color of the consensus reads is used to indicate a family.

Within each read track, each nucleotide variant is indicated by a different color. T, A, C, and G are red, green, blue, and orange, respectively. An "I" indicates insertion, and white color with a dash indicates deletion.

6. You can sort, adjust, and view details about variants and base calls that are visualized in each read coverage track.

| Option | Description |
|---|---|
| View the read coverage tracks sorted by family | Click ⚙ **(Actions)** next to the read coverage track, then select **Color►Molecule**. |
| Adjust the BAM tracks | Click ⚙ **(Actions)** next to the read coverage track, then select an option to adjust the view of the track. For more information, see “Adjust pileup tracks” on page 87. |
| Review detailed data about a BAM read | Single-click the read track to obtain information such as the mapping quality, the strand, and the read base. |
| Review distribution of base calls at a selected position | Click the density plot (the gray bar at the top of the read coverage tracks) to view information about the total count, total reads, and total number of molecules, the distribution of single nucleotides at that position, and the number of insertions and deletions. |

## View the QC results

You can view the quality control (QC) metrics for each sample that was sequenced in a run in Genexus™ Dx Software.

Each run includes NTC and positive controls that are added automatically to each run.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Name** column, click the sample name of interest.
3. Click the **QC** tab.
   The QC metrics are shown.

Review the QC data. For more information, see “QC results” on page 93.

## QC results

The quality control (QC) metrics for each sample that was sequenced in a run are displayed in Genexus™ Dx Software.

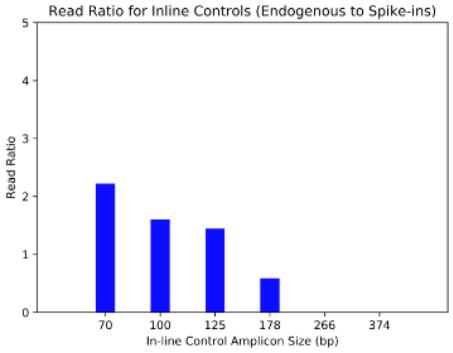
If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test in order to pass. The QC status is divided into the following categories.

- ✔ (Passed) indicates the sample passed all QC metrics.
- ✖ (Failed) indicates the sample failed a QC metric.
- — (Not Calculated) indicates a sample did not undergo QC analysis.

The data shown in the screen depend on the assay that was used in the run. Not all metrics are shown for every assay.

| Metric | Description |
|---|---|
| Purification QC | Quality control information for nucleic acid extraction that is performed on a Genexus™ Dx Purification System. |
| Sample Concentration RNA | The concentration of extracted RNA after purification. |
| Sample Concentration DNA | The concentration of extracted DNA after purification. |
| Run QC | General run quality control information. |
| Key Signal | The average signal after software processing for library ISPs that identically match the library key (TCAG). |
| Percent Loading | The number of wells with ISPs divided by the number of the total addressable wells in a run. |
| Raw Read Accuracy | The average raw accuracy across each individual base position in a read, where raw read accuracy is calculated as 100 * (1 - (sum(per base error)/sum(per base depth))). |
| Templating QC—CF-1 Control | Sequencing quality metrics of the control fragment. These metrics indicate templating success. |
| Average Reads Per Lane | The number of CF-1 reads divided by the number of chip lanes used in the run. |
| Mean AQ20 Read Length (bp) | Average length, in base pairs, at which the accuracy rate is ≥99% for CF-1 reads. |
| Sample QC—DNA | Sequencing quality metrics of the sample DNA library. |
| MAPD | MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of ≤0.5 for FFPE indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls. |

*(continued)*

| Metric | Description |
|---|---|
| Mapped Reads | The total valid mapped reads. |
| Mean AQ20 Read Length (bp) | The average length, in base pairs, at which the accuracy rate is ≥99% for all aligned reads of a library. |
| Mean Mol Cov | Uniformity of molecular coverage for all amplicons. |
| Read Length Histogram | The histogram presents all filtered and trimmed DNA library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of genomic sample reads to control reads for each inline control amplicon.<br>  <br><br>Note: The Read Ratio for each inline control amplicon is expected to be 1–3 with 10 ng DNA input (high quality DNA inputs) for Ion AmpliSeq™ HD chemistries . |
| Sample QC – RNA | Sequencing quality metrics of the sample RNA library. |
| Mapped Fusion Reads | The total valid mapped reads. That is, the mapped reads that pass the thresholds and filters across all of the RNA targets in the reference file. This number is a subset of the mapped reads shown in the Run Samples table in the Assay Metrics. |
| RNA Expression Ctrls Detected | The number of expression control genes detected for the sample. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. |
| Read Length Histogram | The histogram presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| NTC QC – DNA | Sequencing quality metrics of the no-template control. |
| Hotspot Calls | The number of positive hotspots detected for the no-template control. |
| Average Base Coverage Depth | The average number of DNA reads of all targeted reference bases. |

*(continued)*

| Metric | Description |
|---|---|
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of no-template control sample reads to control reads for each DNA inline control amplicon. |
| NTC QC – RNA | Sequencing quality metrics of the no-template control. |
| Mapped Fusion Reads | The total number of valid mapped reads for the NTC. |
| Total Fusion Calls[1] | The number of fusion variants detected for the no-template control. |
| Read Length Histogram | The histogram presents all filtered and trimmed reads for the no-template control that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis). |
| PositiveControl01 QC - DNA | Sequencing quality metrics of the DNA control. |
| MAPD | MAPD (Median of the Absolute values of all Pairwise Differences) is a quality metric that estimates coverage variability between adjacent amplicons in CNV analyses. A MAPD value of ≤0.5 for FFPE indicates an acceptable level of coverage variability. High MAPD value typically translates to a lower coverage uniformity. Lower coverage uniformity can result in missed or erroneous CNV calls. If the MAPD QC threshold is not met, CNVs are not called. The MAPD metric does not affect SNVs/INDEL calls. |
| Mapped Reads | The total valid mapped reads for DNA control. |
| Mean AQ20 Read Length (bp) | The average length, in base pairs, at which the accuracy rate is ≥99% for aligned reads of the DNA control. |
| <Variant ID> | Listing of each reported DNA control variant and its corresponding allele frequency or CNV copy number, and its positive or negative status. |
| Read Length Histogram | The histogram presents all filtered and trimmed DNA control library reads that are reported in the output BAM file for DNA positive controls (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |
| Read Ratio for Inline Controls (Endogenous to Spike-ins) | The ratio of DNA control reads to inline control reads for each inline control amplicon. |
| PositiveControl01 QC - RNA | Sequencing quality metrics of the RNA control. |
| Mapped Fusion Reads | The total valid mapped reads for the RNA control. |
| RNA Expression Ctrls Detected | The number of expression control genes detected for the RNA control. This metric measures the RNA input integrity, input amount, and the fidelity of the reverse transcriptase that was used in library preparation. |

*(continued)*

| Metric | Description |
|---|---|
| <Fusion Targeted Isoform or Expression Imbalance> | Listing of each reported fusion targeted isoform and fusion expression imbalance in the RNA control, and its positive or negative status. |
| Read Length Histogram | The histogram presents all filtered and trimmed RNA control library reads that are reported in the output BAM file for RNA controls (Y-axis) and the mean read length in base pairs (X-axis). The shape of the histogram should closely resemble the library size distribution trace without the adapter sequences. |

[1] Counts for this metric include fusions, and non-wild type RNAExonVariant and RNAExonTiles variants.

### MAPD copy number QC metric

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on runs that detect CNVs.

MAPD is one of the metrics that we define to assess whether the panel data are useful for copy number variation (CNV) run results.

MAPD is defined as the Median of the Absolute values of all Pairwise Differences between log2 ratios per tile for a given run. Tiles roughly correspond to amplicons in an Ion AmpliSeq™ assay. Each pair is defined as adjacent in terms of genomic distance. Tiles corresponding to copy number amplicons and other amplicons are being treated equally as no differences in variability are seen between these types. Then, any two log2 ratios that are adjacent on the genome are a pair. Except at the beginning and the end of a chromosome, every log2 ratio belongs to two pairs.

Formally, if `xi` is the log2 ratio at position `i`, with `i` ordered by genomic position:

```
MAPD = median ( | x(i - 1) - x(i) | )
```

MAPD is a per-sequencing run estimate of copy number variability, similar to standard deviation (SD). If one assumes the log2 ratios are distributed normally with mean 0 and a constant SD, then MAPD/0.95 is approximately equal to SD. However, unlike SD, using MAPD is robust against high biological variability in log2 ratios induced by known conditions such as cancer.

Regardless of the source of the variability, increased variability decreases the quality of CNV calls.

## Find sample results

You can sort, search, and filter information to narrow the sample results that are listed in the **Results / Sample Results** screen. The sort, search, and filter tools help you organize sample results and find a specific sample result in Genexus™ Dx Software.

For information to search for a sample result by a unique identifier, such as the name of a run or the name of a sample, see “Search for sample results” on page 97.

For information to filter sample results by a common trait, such as QC status, see “Filter the list of sample results” on page 98.

## Search for sample results

Sample results are listed in the **Results / Sample Results** screen in Genexus™ Dx Software. The most recent sample result is on the top of the list.

You can search for sample results with keywords or text that are included in the name of the sample, run name, or the name of an assay applied to the run.

---

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

---

1. In the menu bar, click **Results**▶**Sample Results**.

2. In the **Sample Results** screen, in the **Sample Name**, **Run Name**, or **Assay Name** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay, sample, or run name of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to a sample, run, or assay name to select one or more items.
      - Click **Select All** to select all items listed in the filter dialog box.

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of sample results is shown in the **Sample Results** screen.

Click **Clear Filters** to remove all filters and view the full list of sample results.

## Sort the list of sample results

You can sort the list of sample results to make it easier to find sample results in Genexus™ Dx Software.

By default, sample results are listed in the **Results / Sample Results** screen by date. The most recent sample result is at the top of the list.

You can change the order that the sample results are listed in the table.

For example, you can sort the **Call** column alphabetically so that the **Absent** calls appear first. This allows you to easily view hotspots that did not pass thresholds for a variant call, such as when the allele fraction is zero.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, click a column heading of interest.
   The list of sample results is sorted in alphabetical or chronological order depending on the entries in the column.
3. Click the column heading again to reverse the order.

## Filter the list of sample results

You can apply filters to narrow search results, or shorten the list of sample results when you view the list of sample results in Genexus™ Dx Software.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the name of the sample, the run name or the assay selected in the run. | For more information, see "Search for sample results" on page 97. |
| Show or hide sample results that are archived | In the dropdown list in the upper left corner, select an archive status.<br>• Archived<br>• Non Archived<br>Select All to list all sample results. |
| Filter by the status of the sample result. | **1.** In the Sample Status column heading, click ▼ (Filter) then select at least one status.<br>• Completed<br>• Running<br>• Failed<br>• Terminated<br>• Stalled<br>• Aborted<br>• Pending<br>**2.** Click Filter.<br>**3.** Records are listed for sample results that meet the sample status criteria. |

*(continued)*

| Option | Description |
|---|---|
| Filter by the QC status. | 1. In the QC Status column heading, click (Filter) then select at least one QC status.<br>• Failed<br>• Not Calculated<br>• Passed<br>2. Click Filter.<br>Records are listed for sample results that meet the QC status criteria. |
| List only sample results from runs that were started on or updated on an exact date. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or updated on the date entered. |
| List only sample results from runs that were started on or updated within a date range. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the earliest date of the range.<br>4. In the Boolean operator dropdown list, select And.<br>5. In the second dropdown list, select Is before or equal to.<br>6. In the calendar that appears, select the latest date of the range.<br>7. Click Filter.<br>Records are listed for sample results from runs started on or updated on dates that meet both conditions. |
| List sample results according to a date that meets either one condition or a second condition | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the earliest date for the sample results of interest.<br>4. In the Boolean operator dropdown list, select Or.<br>5. In the second dropdown list, select Is before or equal to.<br>6. In the calendar that appears, select the latest date for the sample results of interest.<br>7. Click Filter.<br>Records are listed for sample results from runs that were started on or updated on dates that meet at least one of the conditions. |

*(continued)*

| Option | Description |
|---|---|
| List only sample results for runs that were started on or before or updated on or before a date that you indicate. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is before or equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or before the date entered. |
| List only sample results for runs that were started on or after or updated on or after a date that you indicate. | 1. In the Started On or Updated On column heading, click (Filter).<br>2. In the first dropdown list, select Is after or equal to.<br>3. In the calendar that appears, select the date of interest.<br>4. Click Filter.<br>Records are listed for sample results from runs that were started on or after, or updated on or after, the date entered. |

In the column heading, **(Filter)** changes color to indicate that a filter is applied.

Click **Clear Filters** to remove all filters and view the full list of sample results.

## Customize the sample results table

By default, the following information is listed in the sample results table in Genexus™ Dx Software.

- Name of the sample, assay, and run.
- Status of the sample result, such as completed.
- QC status.
- Date and time that the run was started and updated.

More information is available to show in the sample results table. You can customize the table to reflect the information that is most relevant.

### Change the columns in the sample results table

The sample results table in Genexus™ Dx Software includes a default set of columns. You can add columns to the table, or remove columns from the table. The columns that are available depend on the assays that are installed in the software. Table columns are based on sample attributes. You can also add or remove a column for information about **Electronic Signatures**.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, click **Columns**.

3. Select the columns that you want to add to the sample results table, or deselect the columns that you want to remove from the table.
   The selected columns are added to the table or are removed from the table until you close the table.

4. Click **Save** to save the changes to the table columns.
   The changes that you have made to the table appear each time that you view the sample results table. The changes are saved until you add or remove other columns, then save those changes.

To restore the table to the default view, see “Restore the default columns in the sample results table” on page 101.

### Restore the default columns in the sample results table

You can restore the default view of the columns in the sample results table in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample Results** screen, click **Columns**.

3. At the end of the list of columns that appears, click **Restore default**.

The default columns in the sample results table are restored.

## Sample tags in sample results

You can assign tags to sample results in Genexus™ Dx Software. Tags are used to label and group sample results. A tag can be assigned to a single sample or to multiple samples at once.

### Assign sample tags to a sample result

You can assign tags to sample results in Genexus™ Dx Software to label and group sample results. For more information, see “Sample tags” on page 33.

1. In the menu bar, click **Results►Sample Results**.

2. In the **Sample Results** screen, place the pointer over the row of the sample result that you want to tag, then click **Assign Tags**.

3. In the **Assign Tags** dialog box, search for or scroll to a tag.
   If the tag that you want to assign to a sample does not exist in the software, a manager- or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 34.

4. Select one or more tags that you want to assign to the sample result, then click **Save**.

The tag or tags are assigned to the sample result and are listed in the row for the sample result in the list of sample results.

### Assign sample tags to multiple sample results

You can assign one or more tags to multiple sample results in Genexus™ Dx Software. For more information about sample tags, see “Sample tags” on page 33.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the checkbox to the left of each sample result of interest, then click **Assign Tags** above the list of sample results.

   **Assign Tags** is not shown until you select at least one sample result.
3. In the **Assign Tags** dialog box, search for or scroll to a tag.

   If the tag that you want to assign to a sample result does not exist in the software, a manager- or administrator-level user can create a new sample tag. For more information, see “Create a sample tag (manager/administrator)” on page 34.
4. Select one or more tags that you want to assign to the sample results, then click **Save**.

The tag or tags are assigned to the selected sample results and are listed in the row for each sample result in the list of sample results.

## Review run results

You can review completed sample results and reports in Genexus™ Dx Software.

See “Find a run result” on page 112 for information about how to find run results of interest.

1. Click **Results►Run Results** to view run results.
   A list of runs that are pending, in progress, or completed are shown in the **Results / Run Results** screen. Runs with a status of failed, aborted, or stalled are also listed.

   The following run information appears in the **Results / Run Results** screen.

| Column | Description |
|---|---|
| Run Name | The unique name of the run given when it was created in the software. Click a run name to open the Run Summary. For more information, see “The run summary” on page 105.<br>Runs that are reanalyzed are listed with ⟲ (Reanalysis) after the run name. For more information, see “Reanalyze a run” on page 119. |
| Assay Name | The name of the assay selected for the run. You can view the Assay Name and corresponding Assay Full Name for all assays in the Assays►Manage Assays screen. |

*(continued)*

| Column | Description |
|---|---|
| Run Status | The status of the run. For example, Analysis In Progress, Executing Plugins, Analysis Completed, Terminated, Backup: In Progress, Purification In Progress, or Purification Completed).<br>For purification runs that have a status of Purification In Progress, Purification Complete, or for failed or aborted purification runs, you can place the pointer over the shaded number, in the Status column to view the status of each purification batch for the run. The shaded number, such as 1 or 2, represents a purification batch for the run. For example, a status can be 1 Completed, 2 Started, and 3 Planned, which indicates that the first batch is complete, the second batch has started, and the third purification batch has not yet started, but is planned. For more information about purification batches, see “Integrated nucleic acid isolation in Sample to Result runs” on page 52. |
| Total Samples | The total number of samples in a run. |
| PCR Plate Number | A unique identifier for the 96-well plate used for library preparation and templating. For more information, see “Assign PCR Plate” on page 116. |
| Started On | The date and time when the run was started. |
| Updated On | The date and time when the last action was completed on the run. |

2. In the **Run Name** column, click a run name.
   The **Results** screen opens to the **Run Summary** table.

3. View the run results.

   - The **Run Summary** table contains details about the run, including the assays used in the run, samples, reagents and primer tube locations, and whether the run was reanalyzed. For more information, see “View the run summary” on page 104.
   - Click **Run Report** to view detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run. For more information, see “View assay metrics and the run report” on page 108.

You can also perform the following actions in the **Results / Run Results** screen.

| Option | Description |
|---|---|
| Actions[1] | Available action links for a run are shown when you place the pointer over the row of a run.<br>• Delete—Delete the run.<br>• Audit—View the audit trail for the run.<br>For more information, see "Review and download a run history" on page 63.<br>• CSA—Download customer support archive (CSA) log files for the run to help with troubleshooting.<br>For more information, see "Download a Customer Support Archive" on page 225.<br>• Assign PCR Plate—Enter a unique identifier for the 96-well plate used for library preparation and templating. For more information, see "Assign PCR Plate" on page 116. |
| Actions[2] | • View Plan—View detailed run plan information.<br>For more information, see "Run plan details" on page 61.<br>• Assign Kit Barcodes—Assign purification kit barcodes used in the run.<br>• Abort—Allows you to abort a run after purification, but before sequencing. This action is available when the run status is Purification Review Required, or when the run status is Purification Completed and some purification samples have been excluded from sequencing. |

[1] The actions that are available depend on the type of run.
[2] Actions that are available for Sample to Result runs only.

## View the run summary

The run summary provides an overview of the run. The information that is displayed includes the name of the assay used in the run, sample locations, information about the reagents used in the run, primer tube positions, and instrument information. Metrics from sample purification are also provided, if applicable.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Name** column, click the run name of interest.
   The **Run Summary** tab opens.
3. Review the run summary.

| Action | Procedure |
|---|---|
| View the assay metrics. | In the Assays section, click the assay name of interest. |
| View the sample locations in an image of a 96-well sample plate. | In the Sample Locations section, click PCR Plate View. |
| Reanalyze a run with a new assay. | In the upper right corner of the screen, click ••• (More Options)►Reanalyze.<br>For more information, see "Reanalyze a run" on page 119. |

*(continued)*

| Action | Procedure |
|---|---|
| Download customer support archive (CSA) log files for the run to help with troubleshooting. | In the upper right corner of the screen, click ••• (More Options)►CSA.<br>For more information, see “Customer Support Archive (CSA)” on page 225. |
| View the history of variant classifications. | In the upper right corner of the screen, click ••• (More Options)►Variant Audit.<br>The information is available only for results that include variant classifications. |
| View the run report. | Click the Run Report tab. |

## The run summary

The run summary displays run information, assays used in the run, sample locations, metrics from sample purification, information about the reagents used in the run, primer tube positions, and instrument information.

Depending on the type of the run and the selections made in the run plan, the run information can include the following items.

Run information

| Item | Description |
|---|---|
| Run Name | The name of the run. |
| Started On | The date and time when the run was started. |
| Completed On | The date and time when the run was completed. |
| Run Status | The status of the run. |
| Report Template | If the option to generate a report was enabled in the run plan, the report template used is listed. |
| Chip Type | The semiconductor sequencing chip used in the run, such as the Genexus™ Dx GX5™ Chip. |
| Library Chemistry | The type of library chemistry used in the run, such as Ion AmpliSeq™ HD. |

Sequencing instrument information

| Item | Description |
|---|---|
| Instrument Name | The name of the sequencer. |
| Instrument Serial Number | The serial number of the sequencer. |
| Status | The status of the sequencing portion of the run. |
| Operator | The name of the person who was signed in the Genexus™ Dx Integrated Sequencer when the sequencing run was performed. |
| Start Date | The date and time when the run was started on the sequencer. |

Sequencing instrument inf ormation *(continued)*

| Item | Description |
|---|---|
| Completion Date | The date and time when the run was completed on the sequencer. |
| Sequencing Flows | The number of flows performed by the sequencer. |

Purification instrument information

| Item | Description |
|---|---|
| Purif. Instrument | The name of the purification instrument. |
| Instrument Serial Number | The serial number of the purification instrument. |
| Status | The status of the purification portion of the run for the batch. |
| Operator | The name of the person who was signed in the purification instrument when purification was performed. |
| Start Date | The date and time when the batch was started on the purification instrument. |
| Completion Date | The date and time when the batch was completed on the purification instrument. |

Assays table

| Item | Description |
|---|---|
| Assay Full Name | The name of the assay used in the run. Click the assay name to display the details of the assay. |
| Assay Name | An abbreviated name of the assay or assays used in the run. |
| Analysis Version | The version of the assay used for analysis. |
| Application Type | The application type for the assay, such as DNA and Fusions. |
| Lane | The chip lane or lanes used in the sequencing run for the assay. |
| Total Samples | The total number of samples sequenced for the assay. A single sample can correlate with multiple wells on the plate for some assays when multiple nucleic acid types are contained within the sample. |

Purification Samples table

| Item | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Sample Type | A term that describes the sample, for example, FFPE. |
| Nucleic Acid Type | DNA, RNA, or DNA+RNA. |

Purification Sample table *(continued)*

| Item | Description |
|---|---|
| Conc. (ng/µl) | The concentration of the sample measured by the purification instrument.<br>In some instances, such as when purification is in progress or when the sample is a no-template control, the values is listed as N/A.<br>If the concentration is not within the QC concentration range specified by the assay, a Quantity Not Sufficient (QNS) alert is displayed. |
| QC Conc. Range (ng/µl) | The QC concentration range for the assay. |
| Batch Status | The run status of a purification batch.<br>The status can be N/A, Planned, Review, Started, Aborted, or Completed. |
| Archive Position | The sample positions in the archive plate.<br>Sample positions can be in rows A to D and in columns 1 to 8. |
| Library Prep | An indicator of whether the sample is selected for sequencing.<br>• ✔(Checkmark) – indicates that the sample is selected for sequencing.<br>• ⊘– (No icon) indicates that the sample is not selected for sequencing.<br>• N/A – indicates that purification has not started or is in progress. |

Sample Locations table

| Item | Description |
|---|---|
| PCR Plate View | Click to view the sample locations in an image of a 96-well sample plate. |
| Well Pos. | The well position indicates the location of the sample on the plate. |
| Library Batch ID | The unique identifier created for the library batch. This information is available only for Library to Result runs. |
| Sample Name | The unique identifier created when the sample was entered into the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA+RNA. |
| Vol. (µl) | The volume of the sample. |
| Conc. (ng/µl) | The concentration of the sample. |
| Dilution Factor | The dilution factor of the sample. |
| Kit Barcodes | The barcode for the kit used for nucleic acid extraction, if applicable. |
| Barcode | The name of the barcode adapter or adapters that are associated with the sample, for example, IonHDdual_0101. |

Sample Locations table *(continued)*

| Item | Description |
|---|---|
| Assay Name | The assay that was used to sequence the sample for the indicated well position. |
| Library Position | The plate location of libraries. For completed runs that were planned using samples, the Library Position indicates the location of unused libraries that can be sequenced by planning and initiating a library run. |

Reagents table

| Item | Description |
|---|---|
| Consumable | Consumables used in the run, such as the PanelKit and LibraryKit, are listed. |
| Barcode | The consumable barcode, if applicable. |
| Part # | The consumable part number, if applicable. |
| Expiration Date | The consumable expiration date, if applicable. |
| Lot # | The consumable lot number, if applicable. |

The **Primer Tube Positions** table shows an image that indicates the locations of the primer pool tubes.

## View assay metrics and the run report

The run report provides detailed information about a run, such as the **Total Bases** and **Final Reads**. For runs with multiple assays, metrics are provided for each assay in the run. The run report contains the assay metrics for all assays in the run.

For runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

1. Click **Results►Run Results** to view sequencing results for a particular sample.
2. In the **Run Name** column, click the run name of interest.

| Action | Procedure |
|---|---|
| View the run report | Click the **Run Report** tab. |
| View the assay metrics for a single assay | In the **Select Assay** dropdown list at the top of the screen, select the assay of interest. |

For more information, see “Assay metrics and the run report” on page 109.

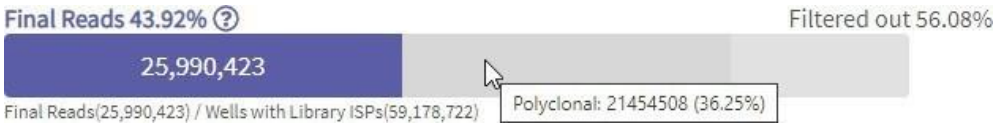
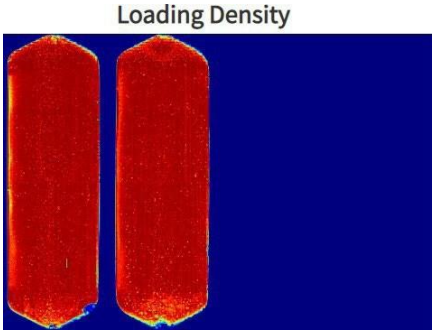
## Assay metrics and the run report

The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics in the **Run Samples** table. Read data for individual samples for the assay are listed in the **Run Samples** table.

Run Report

| Metric | Description |
|---|---|
| Loading | The number and percentage of total addressable wells on the chip that contain an ISP. |
| Enrichment | The number and percentage of wells ISPs that contain live ISPs. |
| Library | The number and percentage of wells with live ISPs that contain Library ISPs. |
| Final Reads | Library reads passing all filters that are recorded in the output BAM files. This value can be different from the total number of reads due to technicalities associated with read trimming beyond a minimal requirement. |
| Total Bases | The number of filtered and trimmed base pairs that are reported in the output BAM file. |
| Raw Read Accuracy | The raw read accuracy across each individual base position in a read calculated as, (1−[total errors in the sequenced reads]/[total bases sequenced]) × 100. Raw read accuracy is measured at each base across the length of the read and is based on 1x sequencing coverage; raw read accuracy is *not* based on consensus accuracy across multiple reads for the same base position. |
| Wells with ISPs | The number of wells that contain an ISP. |
| Unenriched | The number and percentage of wells with ISPs that do not contain live ISPs. |
| Total Addressable Wells | Wells on the chip that can be physically reached by a library. |
| Empty Wells | The percentage of total addressable wells on the chip that do not contain an ISP. |
| Wells with Live ISPs | Loaded wells with ISPs with a signal of sufficient strength and composition to be associated with the library or control fragment key. |
| Wells with Library ISPs | Loaded wells with live ISPs with a key signal that is identical to the library key signal. |
| Control ISPs | Loaded wells with live ISPs with a key signal that is identical to the control fragment key signal. |
| Filtered out | The total percentage of filtered reads, or the sum of the percentages of polyclonal, low quality, and adapter dimer reads. |

Run Report *(continue )*

| Metric | Description |
|---|---|
| Polyclonal | Wells with a live ISP that carries clones from two or more templates.<br>To view polyclonal metrics, mouse over the first low quality portion (gray) of the Final Reads bar plot.<br>  |
| Low Quality | Loaded wells with a low or unrecognizable signal.<br>To view polyclonal metrics, mouse over the second low quality portion (gray) of the Final Reads bar plot.<br>  |
| Adapter Dimer | Loaded wells with a library template of an insert size less than 8 bases.<br>Adapter dimer reads represent a small proportion of total reads and can be seen by hovering the pointer over the right end of the Final Reads bar.<br>  |
| Loading Density | A visual representation of chip loading. Red color indicates areas of higher density of loading. Blue color indicates areas of lower density of loading. The following example shows a sequencing experiment where two lanes on the chip are uniformly loaded with ISPs.<br>  |

### Run Samples

The Run Samples table lists read data for each individual sample in the assay.

| Column | Description |
|---|---|
| Sample Name | The unique identifier created when the sample was entered in the software. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA+RNA. |
| Barcode | The unique identifiers of the dual barcode pair assigned to the DNA and/or RNA library for a sample. |
| Total Reads | The total number of filtered and trimmed reads with the listed dual barcodes assigned to the sample. |
| Mean Read Length | The average length, in base pairs, of usable library reads for each sample. |
| ≥Q20 Bases | The total number of called bases that have ≥99% accuracy (or less than 1% error rate) aligned to the reference for the sample. |
| Mapped Reads | The number of reads that are mapped to the reference file. |
| On Target Reads | The percentage of sequencing reads mapped to any target region of the reference. |
| Mean Depth | The average number of reads of all targeted reference bases. |
| Uniformity | The percentage of bases in all targeted regions (or whole genome) with a depth of coverage ≥20% of the mean read coverage. |
| Read Length Histogram | A histogram that presents all filtered and trimmed library reads that are reported in the output BAM file (Y-axis) and the mean read length in base pairs (X-axis).   |

## Download information about run samples

Information about each sample in a run, such as the barcode for the sample and the total reads, are shown in the run samples table. You can download the information shown in the run samples table.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. Click the **Run Report** tab, then scroll to the **Run Samples** table.
4. In the upper right corner above the **Run Samples** table, click **Export to Excel**.

An XLSX file that contains the information shown in the run samples table is downloaded.

Read length histograms are not included in the file.

### View resequencing metrics

For assays that include resequencing, you can view resequencing metrics when a run is complete.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In **Select Assay**, select the assay name of interest.
   The assay metrics for the sequencing run are shown.
4. Click **Resequencing Metrics**.
   The resequencing assay metrics are shown. For more information, see “Assay metrics and the run report” on page 109.

## Find a run result

You can sort, search, and filter information to narrow the run results that are listed in the **Results / Run Results** screen. The sort, search, and filter tools help you organize run results and find a specific run result in Genexus™ Dx Software.

For information to search for a run result by a unique identifier, such as the name of a run, see “Search for run results” on page 112.

For information to filter run results by a common trait, such as run results that have a status of analysis completed, see “Filter the list of run results” on page 114.

### Search for run results

Run results are listed in the **Results / Run Results** screen in Genexus™ Dx Software. The most recent run result is on the top of the list.

You can search for run results with keywords or text that are included in the name of the run name, the name of an assay applied to the run, or by the PCR Plate number. For more information about how to use the PCR Plate number to track libraries, see “Assign PCR Plate” on page 116.

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IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

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1. In the menu bar, click **Results►Run Results**.

2. In the **Run Results** screen, in the **Run Name**, **Assay Name**, or **PCR Plate Number** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay or run name or PCR plate number of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to a run or assay name to select one or more items.
      - Click **Select All** to select all items listed in the filter dialog box.

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

   The filtered list of run results is shown in the **Run Results** screen.

3. Click **Clear Filters** to remove all filters and view the full list of run results.

## Sort the list of run results

You can sort the list of run results to make it easier to find a run result of interest in Genexus™ Dx Software.

By default, run results are listed in the **Runs / Run Results** screen by date. The most recent run result is at the top of the list.

You can change the order that the run results are listed and sort the run results alphabetically or reverse alphabetically.

1. In the menu bar, click **Runs►Run Results**.

2. In the **Run Results** screen, click the **Run Name** heading.
   The list of run results is sorted alphabetically.

3. Click the column heading again to reverse the order.

## Filter the list of run results

You can apply filters to narrow search results, or shorten the list of run results when you view the list of run results in Genexus™ Dx Software.

1. In the menu bar, click **Runs►Run Results**.
2. In the **Run Results** screen, select one or more filters.

| Option | Description |
|---|---|
| Filter and search by the run name or the assay selected in the run. | For more information, see "Search for run results" on page 112. |
| Show or hide run results that are archived. | a. In the dropdown list in the upper left corner, select an archive status.<br>• **Archived**<br>• **Non Archived**<br>b. Select **All** to list all run results. |
| Filter by the status of the run result. | a. In the **Run Status** column heading, click **(Filter)** then select at least one status.<br>• **Analysis Completed**<br>• **Analysis In Progress**<br>• **Analysis Failed**<br>• **Analysis Aborted**<br>• **Analysis Stalled**<br>• **User Aborted**<br>• **Run Aborted**<br>• **Purification Completed**<br>• **Purification Run Aborted**<br>• **Purification Failed**<br>• **Purification Review Required**<br>• **Purification In Progress**<br>• **Selected on Purification Instrument**<br>• **Backup In Progress**<br>• **Executing Plugins**<br>• **Library Preparation Aborted**<br>• **Library Preparation Completed**<br>• **Library Preparation Started**<br>• **Selected on Instrument**<br>• **Sequencing Completed**<br>• **Sequencing Started**<br>• **Sequencing Aborted**<br>• **Templating Aborted**<br>• **Templating Completed**<br>• **Templating Started**<br>• **Kit Barcodes Required**<br>b. Click **Filter**.<br>Records are listed for run results that meet the run status criteria. |
| Filter by the number of samples in the run. | In the **Total Samples** column heading, click **(Filter)** then use the options to filter the list in the filter dialog box based on the number or span of numbers of samples in the run. |
| List only run results from runs that were started on or updated on an exact date. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is equal to**. |

| Option | Description |
|---|---|
| | c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for run results that were started on or updated on the date entered. |
| List only run results from runs that were started on or updated within a date range. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date of the range.<br>d. In the Boolean operator dropdown list, select **And**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date of the range.<br>g. Click **Filter**.<br>Records are listed for sample results started on or updated on dates that meet both conditions. |
| List run results according to a date that meets either one condition or a second condition. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the earliest date for the sample results of interest.<br>d. In the Boolean operator dropdown list, select **Or**.<br>e. In the second dropdown list, select **Is before or equal to**.<br>f. In the calendar that appears, select the latest date for the sample results of interest.<br>g. Click **Filter**.<br>Records are listed for sample results for runs that were started on or updated on dates that meet at least one of the conditions. |
| List only run results for runs that were started on or before or updated on or before a date that you indicate. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is before or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for run results that were started on or before the date entered. |
| List only run results for runs that were started on or after, or updated on or after, a date that you indicate. | a. In the **Started On** or **Updated On** column heading, click **(Filter)**.<br>b. In the first dropdown list, select **Is after or equal to**.<br>c. In the calendar that appears, select the date of interest.<br>d. Click **Filter**.<br>Records are listed for runs that were started on or after, or updated on or after, the date entered. |

In the column heading, **(Filter)** changes color to indicate that a filter is applied.

Click **Clear Filters** to remove all filters and view the full list of run results.

## Retain run results

You can retain run results in Genexus™ Dx Software. When you retain a run result, run data, including the run result, .wells files, and intermediate files, are retained and cannot be deleted. Users with any level of permissions can retain run results. When a run result is retained, it overrides the automatic deletion settings in the software. For more information, see “Set a schedule to permanently delete files (administrator)” on page 179.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to retain.
3. Click **Retain Data**.

Run results that have been retained are denoted with after the run name in the **Run Results** screen. The run result is retained in the system and cannot be deleted. For information to revert the run to a run that is not retained, see “Release run results” on page 116.

## Release run results

Only run results that have been retained in Genexus™ Dx Software can be released. For more information, see “Retain run results” on page 116.

When you release run results, you can delete the run results. The run results, the associated .wells files, and intermediate files are available to be auto-deleted according to system settings. For more information, see “Set a schedule to permanently delete files (administrator)” on page 179.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, click **Release Data** in the row of the run of interest.
   Run results that have been retained are denoted with (retained) after the run name.
3. Click **Yes** to confirm that you want to proceed with the data release.

The run result is released. The run result can be deleted or auto-deleted.

## Assign PCR Plate

Genexus™ Dx Software lets you track and associate a run with the PCR plate used in the run. The PCR plate is the 96-well plate that is used for library preparation and templating. You can assign a unique identifier, a **PCR Plate Number**, to runs that have a status of **Library Preparation Completed**, **Sequencing Completed**, or **Run Completed**. The PCR plate number that you enter is shown in the **Run Results** screen and if needed, can help you track libraries and troubleshoot. Later, if you sequence the remaining libraries in a different sequencing run and assign the PCR plate number to the run, you can easily search for and find all run results associated with the libraries in the PCR plate.

Note: You should assign a PCR plate to a run if there are leftover libraries that you want to use at a later time to perform a Library to Result run. You can look up the PCR plate number on completed runs to get the library and barcode mapping, then enter the same Barcode information into the Library to Result run.

For more information to find a run with a search of the PCR Plate Number, see “Search for run results” on page 112.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Assign PCR Plate number**.
3. In the **Assign PCR Plate Number** dialog box, confirm, edit, or enter the **PCR Plate Number**.

   The PCR plate number must be between 1 and 10 characters. Only alphanumeric characters (numbers 0 to 9 and letters A to Z), period (.), underscore (_), or hyphen (-) are allowed. Spaces are not allowed.
4. Click **Submit** to associate the PCR plate with the run.

# View verification run results

Manager- and administrator-level users can view verification run details, including the run summary, QC summary, and the reagents used in the run. Verification runs are performed during installation to validate the performance of the instrument or instruments. Sequencing verification runs are performed to validate the Genexus™ Dx Integrated Sequencer.

If a Genexus™ Dx Purification System is also connected, a verification run that validates both the purification instrument and sequencer is performed.

Note: Field Service Engineers use system-installed verification templates to validate the installation of the Genexus™ Dx Integrated Sequencer. Field Service Engineers can view the template information but cannot modify it.

1. In the menu bar, click **Results►Verification Results**.
2. In the **Verification Results** screen, view or download information for the verification run.

| Option | Description |
|---|---|
| View verification results details | In the **Run Name** column, click the verification run name of interest to open the **Verification Results Details** screen.<br>In the **QC** section of the **Verification Results Details** screen, click the **>** symbol to the left of the **Lane** column in the lane of interest to expand the results details pane. |
| Download a verification report that has been signed | Place the pointer over the row of the verification run of interest, then click **Report**. |

3. You can perform the following actions for **Verification Results**. Action links are available when you place the pointer over the row of a verification result.

| Option | Description |
|---|---|
| View Plan | If the run has not started on an instrument, you can view the run plan for the verification run. |
| Audit | Displays the list of users who created and edited the run. You can export and print information from the list from the Audit Trail dialog box. |
| CSA | CSA—Customer Support Archive. Click this link to download the sequencer log files. Log files contained within the CSA can be useful when troubleshooting problems with the sequencer. For more information, see Chapter 10, “Customer Support Archive”. |
| Report | Download a verification report that has been signed. |

## Verification runs

The following information is available in the **Results / Verification Results** screen.

| Column | Description |
|---|---|
| Run Name | The name of the run, created when the run was planned. Click the name to open the Verification Results Details screen. |
| Assay Full Name | The name of the assay used in the run. |
| Field Engineer Name | The name of the support specialist who performed the run. |
| Instrument Name | The name of the sequencer that was verified. |
| Run Status | The current status of the full sequencing run, including analysis. |
| QC Status | Indicates whether a sequencing run passed or failed, based on the sequencing QC metrics selected for the assay. |
| Started On | The date and time that the run was started. |
| Updated On | The date and time that the run was last updated. |

## Sign verification run reports (manager/administrator)

Manager- and administrator-level users can sign results reports for verification runs. However, we recommend that only qualified support specialists sign performance qualification (PQ) reports.

Verification reports can be electronically signed only when all samples pass QC analyses.

1. Click **Results►Verification Results**.
2. In the **Verification Results** screen, click the name of the verification run of interest.
3. In the **Verification Results Details** screen, click **Sign Off**.
4. In the **Sign Off Report** dialog box, enter or make the following selections.
   a. Enter the password.

b. In **Electronic Signature**, select **Approval**.

c. In **Sign off Comments**, enter a comment.

d. In **Laboratory Comments** and **Footer Field**, enter optional information.

5. Click **Sign Off.**

The verification run report is electronically signed.

You can view, print, or download the report in a PDF file. For more information, see “Verification runs” on page 118.

# Reanalyze a run

Runs can be reanalyzed using the same assay in Genexus™ Dx Software. Reanalysis of runs can start from the alignment, basecalling, or signal processing steps. When you reanalyze a run, the reanalysis is applied to all samples in the assay.

| Reanalysis from the | Description |
|---|---|
| Signal processing step | The option to reanalyze a run from the signal processing step is available only when the following conditions are met.<br>• When a run fails during the basecalling step or earlier.<br>• When the run has not already been successfully reanalyzed.<br>Reanalysis at the signal processing step uses DAT files. |
| Basecalling step | The option to reanalyze a run from the basecalling step is available only when a run or reanalysis successfully completes the signal processing step.<br>Reanalysis at the basecalling step uses .wells files. |
| Alignment step | The option to reanalyze a run from the alignment step is available only when a run or reanalysis successfully completes the basecalling step.<br>Reanalysis at the alignment step uses BAM files. |

Note:

- Manager- and administrator-level users can reanalyze a sequencing run only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the run cannot be reanalyzed.
- If a sample fails QC, you can sequence the sample library again. For information to find, recover, and purify the leftover library preparations, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*. Alternatively, you can prepare a new library for sequencing.

The files needed for the stage of reanalysis that you select must be present in the software. For example, if the .wells files for the run have been removed from the software, you cannot reanalyze from basecalling. Instead, reanalyze the BAM files at alignment. Administrator-level users can manage the settings and schedule to backup and delete files and data. For more information, see “Manage backup settings and restore runs (administrator)” on page 168.

You can reanalyze a run with any compatible assay that exists in the software.

1. In the menu bar, click **Results►Run Results**.
2. In the **Results / Run Results** screen, in the **Run Name** column, click the run name of interest.
3. In the upper right corner of the screen, click ⋯**(More Options)►Reanalyze**.
4. In the **Reanalyze** dialog box, enter or select the following information.
   a. In **Reanalysis Run Name** field, enter a reanalysis run name.
   b. In **Start Reanalysis from** dropdown list, select **Alignment**, **Basecalling** or **Signal Processing**.
   c. In the **Current Assay in Run** column, select the checkbox in the row of the assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for the reanalysis from the dropdown list.
5. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Run Results** screen in the **Run Status** column, and in the **Results / Sample Results** screen in the **Sample Status** column. When reanalysis is complete, the new results can be viewed by clicking the run name corresponding to the reanalysis assay in the **Results / Run Results** screen. Runs that have been reanalyzed are appended with ⟲ after the run name.

# Reanalyze a sample

Samples can be reanalyzed using the same assay in Genexus™ Dx Software. A sample can be reanalyzed only when starting from the alignment step. You can reanalyze all samples in a run from the basecalling or signal processing steps. For more information, see “Reanalyze a run” on page 119.

Reanalysis at the alignment step uses BAM files.

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Note: Manager- and administrator-level users can reanalyze a sample only if the run completed without any critical alarms or errors. If the run aborted or produced major alarms or errors, the sample cannot be reanalyzed.

---

The option to reanalyze a sample from the alignment step is available only when a sample or reanalysis successfully completes the basecalling step.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Sample Results** screen, place the pointer over the row of a sample of interest, then click **Reanalyze**.

   Alternatively, you can reanalyze a sample when you click a sample name, then click ••• **(More Options)**►**Reanalyze**.
3. In the **Reanalyze** dialog box, enter or select the following information.
   a. In **Reanalysis Run Name** field, enter a reanalysis run name.
   b. In the **Current Assay in Run** column, select the checkbox in the row of the assay that you want to reanalyze, then in the **Choose Assay to Reanalyze** column, select an assay that you want to use for the reanalysis from the dropdown list.
4. Click **Reanalyze**.

Follow the progress of the reanalysis in the **Results / Sample Results** screen in the **Sample Status** column. When reanalysis is complete, the new results can be viewed by clicking the sample name corresponding to the reanalysis assay in the **Results / Sample Results** screen. Samples that have been reanalyzed are listed in the table of sample results with the run name appended with ⟲ .

# Results files

The following files can be downloaded for each sample. The files that are available for download vary depending on the assay. Results files include the sequencing data, results from the analyses, such as variant files, and audit and log files.

For instructions to download results files, see “Download results files for a sample result” on page 123.

| Option | File name | Description |
|---|---|---|
| Variants | | |
| Filtered Variants (.vcf) | `<Sample_name>_<LibPrepID>_filtered.vcf` | Summary of filtered variant results in variant call format (VCF). |
| Variant Summary (.tsv) | `Summary.tsv` | File that lists SNV/INDEL, copy number, and fusion results in tab-separated value format (TSV). |
| Snvindel (.tsv) | `Snvindel.tsv` | File that lists SNV/INDEL variant results in tab-separated value format (TSV). |
| Fusion (.tsv) | `Fusion.tsv` | File that lists fusion results in tab-separated value format (TSV). |
| CNV (.tsv) | `Cnv.tsv` | File that lists copy number variant results in tab-separated value format (TSV). |

*(continued)*

| Option | File name | Description |
|---|---|---|
| Sequencing Results | | |
| DNA Unmapped Bam File (.bam) | `<barcode>_rawlib.basecaller.bam` | Unmapped DNA barcode BAM file; output after mapping reads to reference. |
| DNA Mapped Bam File (.bam) | `merged.bam` | Mapped BAM file of combined barcode reads. |
| DNA Mapped Bam Index File (.bai) | `merged.bam.bai` | Mapped BAM Index file. |
| DNA Basecaller FASTAQ File (.fastq) | `<barcode>_rawlib.basecaller.fastq` | FASTQ file of the DNA barcodes used. |
| DNA Processed Bam File | `merged.bam.ptrim.bam` | Mapped BAM file of combined barcode reads. |
| DNA Processed Bam Index | `merged.bam.ptrim.bam.bai` | Mapped BAM index file. |
| RNA Unmapped Bam File (.bam) | `<barcode>_rawlib.basecaller.bam` | Unmapped RNA BAM file; output of base calling, contains unmapped reads. |
| RNA Mapped Bam File (.bam) | `<barcode>_rawlib.basecaller_alignments.bam` | Mapped BAM file of combined barcode reads. |
| RNA Mapped Bam Index File (.bai) | `<barcode>_rawlib.basecaller_alignments.bam.bai` | Mapped BAM index file. |
| RNA Basecaller FASTAQ File (.fastq) | `<barcode>_rawlib.basecaller.fastq` | FASTQ file generated from the unmapped BAM file of the RNA barcodes used. |
| Test Fragment Basecaller FASTAQ File (.fastq) | `rawtf.basecaller.fastq` | FASTQ file for the test fragment. |
| Audit and Log | | |
| Analysis Log File | `analysis.log` | Analysis log file. |
| Run Audit | `PlannedRun-AuditTrail.pdf` | Contains all audit records pertaining to the run. |
| Reports | | |
| Report | `<language>_<sample name>_Dx_<report template name>_<assay name>_<date>.pdf` | A PDF report that contains sample-specific results. For more information, see “Clinical Test Report and Clinical Laboratory Report” on page 126. |

*(continued)*

| Option | File name | Description |
|---|---|---|
| Sample Summary | `Info.csv` | Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. |
| Troubleshooting Files | | |
| Log Files[1] | `analysis.log` | Analysis log file. |
| | `summary.<timestamp>.log` | Start and end time for each time an assay module is executed for the analysis. |
| | Various other files with `.err` `.out` file extensions | Analysis pipeline logs used by field service engineers for troubleshooting. |
| | `Info.csv` | Contains information about the run and analysis, including software version details, sample details, library details, run details, assay details, reagent and consumable information, run and sample QC metrics, and instrument summary. |
| | `PlannedRun-AuditTrail.pdf` | Contains the audit trail of the run plan in PDF format. |
| VCF Files | `analysis.vcf` | Summary of variant results for sample, NTC and positive control in variant call format (VCF). |

[1] Separate folders are generated for each sample. If included in the run, separate folders are also generated for an NTC and positive control.

## Download results files for a sample result

You can download results files for a sample result in Genexus™ Dx Software.

1. In the menu bar, click **Results►Sample Results**, or **Results►Run Results**, then do one of the following procedures.

| Option | Selection |
|---|---|
| Download files for a sample | |
| Download results files for a sample from the list of sample results. | In the Results / Sample Results screen, place the pointer over the row of a sample of interest, then click Download Files. |
| Download results files for a selected sample from the Results screen. | **1.** In the Results / Sample Results screen, in the Sample Name column, click the sample name of interest.<br>**2.** Click ••• (More Options)►Download Files. |

*(continued)*

| Option | Selection |
|---|---|
| Download files for a run or assay | |
| Download results files for a run from the list of run results. | In the Results / Run Results screen, place the pointer over the row of a run of interest, then click Download Files. |
| Download results files for a selected run or assay from the Results screen. | 1. In the Results / Run Results screen, in the Run Name column, click the run name of interest.<br>2. Click the run or assay of interest from the results navigation bar.<br>3. Click ••• (More Options)►Download Files. |

2. In the **Download Files** dialog box, select the files to download, then click **Download**.
   For information about the files, see “Results files” on page 121.

The selected results files are downloaded in one ZIP folder.

## Download results files for multiple sample results

You can download results files for multiple sample results in Genexus™ Dx Software. If the run included more than one assay or more than one research application for the assay, you can download the results for each assay or research application at once.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over the row of a run of interest, then click **Download**.
   Alternatively, in the **Run Results** screen, click the name of a run, then in the **Run Summary** tab, click ••• **(More Options)►Download Files** to download the results files.
3. Select the assay if the run included more than one assay or more than one research application for the assay.
4. In the **Download Files** dialog box, select the files to download, then click **Download**.
   For information about the files, see “Results files” on page 121.

The selected results files are downloaded in one ZIP folder named `Runname_assayname`. The folder contains subfolders for each sample result except for any NTC and positive control samples.

Control sample information is in the `SmallVariants.vcf`. For more information, see “Results files” on page 121.

# Data files and flow

During a Genexus™ Dx Integrated Sequencer run, raw data (DAT) files from the first flows are available. Genexus™ Dx Software starts processing the data, producing a 1.wells file. Basecalling is performed on the 1.wells file data, producing an unmapped BAM (uBAM) file. Subsequent analysis produces mapped reads (BAM) and variant calls (VCF) files.

The following table shows the flow of data and typical file size generated as the Genexus™ Dx Software processes data.

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IMPORTANT! Data from approximately 200 sequencing runs can be accommodated on the sequencer before disk space becomes a limiting factor and data archiving is required for runs that use a GX5™ Chip if 1.wells files and intermediate files are marked for deletion, after a cleanup threshold for disk space is reached. You can perform approximately 45 to 50 whole chip runs without deleting any wells files on the system. For more information about data archival of the analysis files, see “Manage backup settings and restore runs (administrator)” on page 168.

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| Process | File type | File size |
|---|---|---|
| Sequencing[1] | DAT | 2 TB |
| ▼ | | |
| Signal Processing [2] | Wells | 250 GB |
| ▼ | | |
| Base Calls (reads) | uBAM | 15 GB |
| ▼ | | |
| Mapped Reads | BAM | 15–20 GB |
| ▼ | | |
| Variant Calls | VCF | 120 Kb |

[1] The sequencing raw data (DAT) files are deleted from the sequencer after run completion.

[2] A configurable disk space cleanup process is executed nightly to free up space for future runs and to help ensure that sufficient hard disk space is maintained. If the system memory available for storing run analysis files reaches the threshold that is configured, the system auto-deletes 1.wells files for the oldest completed runs. A minimum of 1TB of space is required to start new runs. If the system approaches the 1TB threshold, an alert notifies you when you sign in. New run cannot be started if the 1TB threshold is reached. If desired, you can back up 1.wells files to an external drive.

# 7 Create, view, and sign reports

The Oncomine™ Dx Express Test provides two types of reports in Genexus™ Dx Software.

- Variant reports such as the Clinical Test Report and the Clinical Laboratory Report

  The Clinical Test Report and the Clinical Laboratory Report are PDF variant reports of the results for a sample in a sequencing run. You can use the system-installed report templates to generate variant reports, or you can customize the layout and contents in a user-defined variant report. The Clinical Test Report and Clinical Laboratory Report can be electronically signed off by more than one user, including users who have different roles. One user can be designated to lock reports after reports are signed off.

- Run reports (see “Run report” on page 141)

  A run report is a PDF report of the information that is shown in the **Run Report** tab. The run report includes assay metrics and the record of reagents that were used in a run. For more information, see “Assay metrics and the run report” on page 109.

To save or distribute a variant report or a run report, you can download the report.

Administrators and managers create user-defined report templates for their organization in Genexus™ Dx Software. For more information, see “Report templates (manager/administrator)” on page 135.

## Clinical Test Report and Clinical Laboratory Report

The Clinical Test Report and Clinical Laboratory Report are PDF variant reports generated by the Oncomine™ Dx Express Test for each sample in a sequencing run. Each system-installed assay for the test provides Clinical Test Report and the Clinical Laboratory Report templates, set as default templates, that you can use without change to generate reports. Alternatively, you can modify the customizable sections of the layout and contents of the default report templates to create user-defined templates. Some sections of the report templates are locked and cannot be customized. For more information about creating custom report templates, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide*.

To make Clinical Test and Laboratory Reports available for each sample result on completion of a sequencing run, enable **Generate Report** in the **Setup** step when you plan the run. For more information, see Chapter 4, “Plan and manage runs”. If you need to generate a report after a run is complete, see “Generate a variant report” on page 132.

When Clinical Test and Laboratory Reports have been generated for a sample result, they are available for download in three places:

- In the **Results** screen for the sample when you click ··· **(More Options)** ▶ **Download Files**. In the dialog, select the **Reports** checkbox to enable download of both reports.
- In the pane for the Clinical Test Report or Clinical Laboratory Report in the **Reports** tab when you click ⋮ **(More)** ▶ **Download Report**.
- In the **Results / Sample Results** screen when you place the pointer over the row for that sample, then click **Report** to download the reports.

The Clinical Test Report and Clinical Laboratory Report can be electronically or manually signed by users. Electronic signatures are shown in the **Report Signed By** section of the report.

## Sections of the Clinical Test Report

Manager- and administrator-level users can create custom report templates that can include various sections. Each section contains details about a result, so you can include information to meet laboratory requirements.

The Clinical Test Report contains the following sections and information.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered in the software. You can customize the format of the Sample Details section when you create a new report template. For more information, see “Create a report template (manager/administrator)” on page 136. |
| Tumor Mutation Profiling Results: Summary | A summary of results for variants detected in the sample classified in 3 biomarker levels:<br>• Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use<br>• Level 2: Cancer Mutations with Evidence of Clinical Significance<br>• Level 3: Cancer Mutations with Potential Clinical Significance<br>If a detected variant is listed in the Companion Diagnostics (CDx) Biomarkers level, the associated therapy or therapies are listed after the variant. For further information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 130. |
| Tumor Mutation Profiling Results: Detected | A list of variants detected in the sample grouped by SNVs/Indels, Fusions, and CNVs. Variant ID, amino acid change, and fusion information is also provided in this section. This section is customizable. Additional columns from the Variants tab can be included in a custom report template if you would like to show more information for positive calls in the report. |
| Intended Use | Intended use statement for the Oncomine™ Dx Express Test, including a table that lists the current Companion Diagnostic Indications. |
| Test Description | General description of the test, including a table listing the types of genetic alternations detected by the Oncomine™ Dx Express Test, and the genes targeted by the test for each type of alteration. |

*(continued)*

| Section | Description |
|---|---|
| Tumor Mutation Profiling Results: No Calls | A list of No Calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. A No Call is defined as a call for which some evidence for the presence of a variant exists, but the call does not pass one or more filters that are required for a high confidence variant call. This section is customizable. Additional columns from the Variants tab, for example, Call Details, can be included in a custom report template if you would like to show in the report the possible reason or reasons why a No Call was made. |
| Tumor Mutation Profiling Results: Negative Calls | A list of negative calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. This section is customizable or deletable. Additional columns from the Variants tab can be included in a custom report template if you would like to show in the report the reason why a negative call was made. |
| Report Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |
| Manual Signatures | *(Optional)* A section for physical signatures of the printed report. |
| Custom Text | *(Optional)* A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Footer | The footer is repeated on each page of the report. |

## Sections of the Clinical Laboratory Report

The Clinical Laboratory Report contains all the information in the Clinical Test Report, and in addition includes the **QC Metrics**, **Sequencing Run Details**, **Reagents**, and **Kit Barcodes** sections.

| Section | Description |
|---|---|
| Header | Information that repeats on each page of the report. Includes the title of the report, the organization address, the date that the report was generated, an optional logo, and other fields about the sample, such as patient ID and date of birth. |
| Sample Details | The sample information that is entered in the software. You can customize the format of the Sample Details section when you create a new report template. For more information, see “Create a report template (manager/administrator)” on page 136. |
| Tumor Mutation Profiling Results: Summary | A summary of results for variants detected in the sample classified in 3 biomarker levels:<br>• Level 1: Companion Diagnostics (CDx) Biomarkers Noted in Table 1 of the Intended Use<br>• Level 2: Cancer Mutations with Evidence of Clinical Significance<br>• Level 3: Cancer Mutations with Potential Clinical Significance<br>If a detected variant is listed in the Companion Diagnostics (CDx) Biomarkers level, the associated therapy or therapies are listed after the variant. For further information about the biomarker levels, see “FDA's 3-Level biomarker classification-based variant reporting” on page 130. |

*(continued)*

| Section | Description |
|---|---|
| Tumor Mutation Profiling Results: Detected | A list of variants detected in the sample grouped by SNVs/Indels, Fusions, and CNVs. Variant ID, amino acid change, and fusion information is also provided in this section. This section is customizable. Additional columns from the Variants tab can be included in a custom report template if you would like to show more information for positive calls in the report. |
| Intended Use | Intended use statement for the Oncomine™ Dx Express Test, including a table that lists the current Companion Diagnostic Indications. |
| Test Description | General description of the test, including a table listing the types of genetic alternations detected by the Oncomine™ Dx Express Test, and the genes targeted by the test for each type of alteration. |
| Tumor Mutation Profiling Results: No Calls | A list of No Calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. A No Call is defined as a call for which some evidence for the presence of a variant exists, but the call does not pass one or more filters that are required for a high confidence variant call. This section is customizable. Additional columns from the Variants tab, for example, Call Details, can be included in a custom report template if you would like to show in the report the possible reason or reasons why a No Call was made. |
| Tumor Mutation Profiling Results: Negative Calls | A list of negative calls for the sample grouped by SNVs/Indels, Fusions, and CNVs. This section is customizable or deletable. Additional columns from the Variants tab can be included in a custom report template if you would like to show in the report the reason why a negative call was made. |
| QC Metrics | • NTC QC – DNA—A summary of the DNA no-template control quality control metrics.<br>• NTC QC – RNA—A summary of the RNA no-template control quality control metrics.<br>• Sample QC – RNA—A summary of RNA sample quality control metrics.<br>• Sample QC – DNA—A summary of DNA sample quality control metrics.<br>• Templating Control QC – CF-1—A summary of the CF-1 quality control metrics.<br>• Positive Control01 QC – DNA—A summary of the DNA positive control quality control metrics.<br>• Positive Control01 QC – RNA—A summary of the RNA positive control quality control metrics.<br>• Purification QC—A summary of QC metrics for the purification run.<br>This section is locked and cannot be customized. |
| Sequencing Run Details | Contains the following subsections:<br>• Assay Name—The assay used in the run<br>• Panel—The panel used in the run<br>• Started On—The date and time that the run was started<br>• Completed On—The date and time when the run was completed<br>• Operator—The user who sent the run to the instrument<br>• Instrument—The name of the instrument<br>• Serial Number—The serial number of the instrument<br>This section is locked and cannot be customized. |

*(continued)*

| Section | Description |
|---|---|
| Reagents | Details about run consumables, including part numbers, expiration dates, and lot numbers.<br>This section is locked and cannot be customized. |
| Kit Barcodes | Details about the kits used for each sample sequenced in the run, including kit barcode information, nucleic acid type, and kit type. |
| Report Signed By | A section with information about one or more electronic signatures for the report, including the name of the user who signed the report, the signature type, the date, and comments.<br>This section is locked and cannot be customized. |
| Manual Signatures | *(Optional)* A section for physical signatures of the printed report. |
| Custom Text | *(Optional)* A section for custom text. You can allow the text to be edited when the report is generated if desired. |
| Footer | The footer is repeated on each page of the report. |

## FDA's 3-Level biomarker classification-based variant reporting

Oncomine™ Dx Express Test results are reported in alignment with an FDA's 3-Level biomarker classification-based system. Evidence of clinical relevance is obtained from a curated database called Oncomine™ Knowledgebase. Variants are classified into clinical relevance categories as defined below:

- Level 1: Companion Diagnostics (CDx) Biomarkers—gene variants that have companion diagnostic claims in the corresponding cancer type that are associated with an FDA-approved therapy as listed in Table 1 of the intended use.
- Level 2: Cancer Mutations with Evidence of Clinical Significance—gene variants that are not prescriptive or conclusive for labeled use of a specific therapeutic product, and for which clinical validation has not been performed, but which have curated clinical evidence associated with FDA- approved therapies and/or National Comprehensive Cancer Network (NCCN) clinical guidelines (Category 1 and 2A) in the sample cancer type.
- Level 3: Cancer Mutations with Potential Clinical Significance—gene variants not considered Level 1 or Level 2 biomarkers that can be described as cancer mutations with potential clinical significance. These variants are not prescriptive or conclusive for labeled use of a specific therapeutic product, and clinical validation has not been performed. They have curated clinical evidence associated with NCCN clinical guidelines Category 2A in a different cancer type or Category 2B in any cancer type. They may also have curated evidence as an enrollment criterion in clinical trials or biomarkers with evidence in peer-reviewed literature.

Variant category files are updated with every new assay definition file.

## View a Clinical Test Report or Clinical Laboratory Report

A variant report must be generated before you can view the report. If a report template is set as the default template for the assay associated with the run, one or more variant reports are generated after a run completes successfully. Alternatively, you can generate one or more variant reports after a run completes successfully. For more information, see “Generate a variant report” on page 132.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.
3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown. The pane for a variant report includes the following information.
   - Report name
   - Report template name
   - Date that the report was generated
   - Username for the user who generated the report
   - Language used for the report
   - If a variant report has been electronically signed at least once, the signature information is listed in the pane for the variant report. If multiple users have signed the report, you can place the pointer over the electronic signature information to view more details. If the electronic signature that is used is associated with a lock-level signature type, the report is locked.
   - If a variant report is locked, **(Lock)** is shown in the upper right corner of the pane.
4. In the pane of the report of interest, click **(More)►View Report**.

The report opens in a new browser tab. You can download variant reports as PDF files.

## Download a Clinical Test Report or Clinical Laboratory Report

You can download a Clinical Test Report or Clinical Laboratory Report for a sample result of interest from the **Results / Sample Results** screen, from the pane that shows the report, or as part of a download of results files.

1. In the menu bar, click **Results**►**Sample Results**.
2. Download the report with one of the following options.

| Option | Procedure |
|---|---|
| Download a ZIP file that contains all variant reports for a sample result. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click ••• **(More Options)**►**Download Files**. In the **Download Files** dialog, select the **Reports** checkbox, then click **Download**. |
| Download the variant report in PDF format from the **Reports** tab for the sample results. | In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column, then click the **Reports** tab. In the pane of the report of interest, click ⋮ **(More)**►**Download Report**. |
| Download a ZIP file that contains all variant reports for a sample result from the **Sample Results** screen. | In the **Sample Results** screen, place the pointer over the row of the sample of interest, then click **Report**. |

   A ZIP file that contains the PDF report is downloaded to the computer if you download reports from the **Sample Results** screen or as part of the results files. A report in PDF format downloads if you use the **Reports** tab for the download.

3. Extract the downloaded files, then open the PDF file in an appropriate viewer.

## Generate a variant report

You can generate a new variant report for sample results after a run is complete. A 🔒 **(Lock)** in the variant report indicates that the electronic signature option for the report is locked. After a variant report is locked, the report cannot be electronically signed by any other user.

You can generate multiple reports for a sample result, if each report is named uniquely, and is generated in a different language, or uses a different report template.

1. In the menu bar, click **Results**►**Sample Results**.
2. In the **Results / Sample Results** screen, click the sample of interest in the **Sample Name** column.
3. Select the **Reports** tab, then click **+ Generate Variant Report**.
4. In the **Generate Report** dialog box, change the name of the report that is generated by the software, if needed.
   a. Select the report template and language of the report.

b. If the option to make custom text **Editable on Report Generation** was selected when the report template was created, enter a title in the **Custom Text** section and if needed, a description.

Report templates that are associated with the assay that was used in the run are included in the **Report Template** list. For more information, see “Create a report template (manager/ administrator)” on page 136.

If you select the same report template that was used to generate a variant report, and have not locked that report, the new selections you make override the previous variant report.

5. Click **Generate**.
   A draft version of the report is added to the **Reports** tab.

A pane for the new report is added next to the Run Report pane in the **Reports** tab. Reports that have been generated are available for download in the **Reports** tab, in the **Sample Results** screen, and in the ZIP package that contains results files.

## Sign off variant reports

Manager, administrator, operator, and report-level users can electronically sign a variant report for sample results in Genexus™ Dx Software. A sample result can have multiple variant reports only if each report uses a different report template, and a different language.

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IMPORTANT! If you use the same template and language to regenerate an existing report, the existing report will be overwritten.

---

Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign the report, you will be the last person to sign the report before it is locked.

The signature information appears in the variant report PDF file or a user-created report, if electronic signatures are included in the template that is used to generate the report. For more information, see “Clinical Test Report and Clinical Laboratory Report” on page 126.

Multilanguage support for PDF report generation is provided.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

You must have electronic permissions enabled to sign reports. Reports that can be electronically signed must be labeled as **Draft**, and not be locked.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample result of interest in the **Sample Name** column.

3. In the **Reports** tab, in a variant report pane that is labeled as **Draft**, click **Sign Off**.
   The **Sign Off Report** dialog box opens and shows the name of the report, the template used for the report, and the following information, which is based on the profile of the user who signs the report.
   - **Electronic Signature**
   - **User Name**
   - **Language**
4. In **Password**, enter the password.
5. In **Sign Off Comments**, enter a comment.
6. In **Footer Field**, enter any text that you want to appear in the footer of the PDF report pages.
   If you entered footer information in the **Footer Field** when you created a report template, or if text was entered during a previous sign off, the same footer information is shown in the **Sign Off Report** dialog box. You can edit the text in the **Footer Field** to change the information that will be shown in the footer of the PDF report.
7. Click **Sign Off** to confirm the electronic signature.
   The pane for the signed variant report is updated to include the signature information for the variant report. If the electronic signature that is associated with a lock-level signature type is used, a **(Lock)** is shown in the variant report pane and the report is locked.
8. In the menu bar, click **Results►Sample Results** to return to the **Results / Sample Results** screen.
   If the **Electronic Signatures** column is shown in the **Sample Results** screen, the electronic signature types for users who signed the report are listed, and a **(Lock)** is shown for reports that are locked.
9. In the row of the sample of interest, click **Report** to download the report.

The report is signed. The **Electronic Signatures** section of the report pane is updated to include your username and the signature type for your account. You can hover over the signature type to show the list of signature types that are completed for the report. If multiple electronic signatures are added to the report, an electronic signature type for each signer is shown in the **Electronic Signatures** column of the sample results table.

You cannot generate new reports from the variant results if a report from those results is signed off and locked. If you must change the report, reanalyze the sample and generate a report for the reanalyzed sample.

You can send notifications for report sign-offs to other users. For more information, see “Send a notification to another user”. For information to download the signed report, see “Download a Clinical Test Report or Clinical Laboratory Report” on page 132.

## Send a notification to another user

You can send notifications to other users in Genexus™ Dx Software. For example, you can notify another user when you have signed a variant report.

To receive email notifications, the recipient must have an email address that is configured in the software. For more information, see “Configure email notifications (manager/administrator)” on page 165.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample result of interest.
3. In the upper right corner of the screen, click ⋯**(More Options)►Send Notification**.
   The **Send Notification** dialog box opens.
4. In **Send Notification to**, select one or more administrator-level users of interest.
5. In **Comments** enter the message.
6. Click **Send** to send the message.

An email message with the text that you entered is sent to the users that you selected. In addition, when a recipient signs in to the software, an unread notification, (Notifications), is shown. For more information, see “View notifications” on page 17.

# Report templates (manager/administrator)

Manager- and administrator-level users create and manage custom report templates that are used to generate PDF reports after a run. Users select a report template when they plan a run.

## Preview a report template (manager/administrator)

Manager- and administrator-level users can view custom and system-installed report templates in Genexus™ Dx Software.

For more information about system-installed report templates, see “Default report templates” on page 220.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
   Existing report templates are listed in this tab, including system-installed report templates. For more information, see “The report templates table” on page 136.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to view, then click **Preview**.
   The **Report Template Preview** opens.

4. In the **Report Template Preview** screen, click **Open** to download a PDF file of the report template.
    a. Click **Close** to close the **Report Template Preview** and return to the list of report templates.

You can place the pointer of the row of a report template to perform more actions, such as lock, edit, delete, or add notes to a report template. The options that are available depend on whether the report template is a draft or is locked.

### The report templates table

You can view details about report templates in Genexus™ Dx Software.

Details about report templates

| Item | Description |
|---|---|
| Name | The name of the template. System-installed report templates are included with the software. Custom report templates are named by the person who creates the template. |
| Application Category | The application category that is associated with the report template. |
| Assay Name | The name of assay that is associated with the template. |
| Status | Status of the report template.<br>• Locked<br>• Draft |
| Default | Indicates if the report template is designated as the default report template. |
| Created On | Date on which the report template was created. |

## Create a report template (manager/administrator)

Manager- and administrator-level users create custom report templates that are used to generate PDF reports after a run. As you create the report template, you can reorder sections, and add or remove sections. For more information, see “Sections of the Clinical Laboratory Report” on page 128.

To create a new report template, you must start from an existing report template, and then make changes to the layout and content of the sections if needed.

IMPORTANT! For a report template to be used in a run, it must first be locked.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Manage Presets** screen, click the **Report Templates** tab.
   Existing report templates are listed in this tab, including system-installed templates.
3. Click ┼ **Add New**.

4. In the **Report Template** screen, enter a unique report template name, then complete the remaining sections.

   IMPORTANT! When you enter a name for the report template, use only alphanumberic characters (A-Z, a-z, 0-9), and the special characters period (.), hyphen (-), underscore (_), and space.

   If other special characters are used in the report template name, you cannot view or download variant reports that generated with the template.

   Note: The application category determines the sample attributes that are available to add to the report template.

   a. In **Application Category** select the application category that you want to associate with the report.

   b. In **Select Assay**, select the assay that you want to associate with the report.

      Note: After you associate an assay and application category with a report template, that report template becomes available for use with samples for that application category that are processed with the associated assay.

   c. In **Start From Existing Report Template**, select a report template to start from an existing report template if desired.

      Only report templates that are associated with the assay and application category that you select are listed.

   d. In **Languages Setting**, select the language for the report.

   e. Enable or disable the option to set the report template as the default template.

      When a report template is set as default, a report is generated from the report template each time you complete a run with an assay that is associated with the report template. Each assay can be associated with 3 default report templates in various languages. A report pane for each report is visible in the **Reports** tab of the sample results table. If you do not designate a report template as default, you can generate reports from the report template after a run is complete in the **Reports** tab. For more information, see “Generate a variant report” on page 132.

      - To set the new report template as default, keep the **Set as Default** toggle switch enabled.
      - If you do not want the report template to be set as default, disable the **Set as Default** toggle switch.

5. Add information to the **Header** section.

   The **Header** and **Footer** sections are required. All other sections are optional. For more information about the sections that can be included in a report, see “Sections of the Clinical Laboratory Report” on page 128.

   a. Enter a report title and the organization address.

   b. To include a logo on the report, in the **Logo** field, click **Select File** to browse to the logo file.

      Acceptable formats are PNG, GIF, and JPG.

c. In **Header Fields**, click the field, then in the list that appears, select a term to add it to the **Header**.

d. Enable or disable the **Report Date** and **Assay Name** checkboxes if needed.

6. In the **Configuration** pane on the left, you can add new sections, then move or delete sections from the template if needed.

   a. In **Add Section**, select the section, then click **+ (Add)**.
   The new section is added at the bottom of the list.

   b. Use the following options to move or delete sections from the template.

| Option | Description |
|---|---|
| Add multiple sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, for each section to include, click the section name of interest, then click » **(Add)**.<br>Repeat for each section to include in the report template.<br>c. Click **Apply**. |
| Remove a section. | In the box for the section, click ✕ **(Remove)**. |
| Remove multiple sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click the section name of interest, then click « **(Remove)**.<br>Repeat for each section to remove from the report template.<br>c. Click **Apply**. |
| Add all sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, click **Select All**.<br>c. Click **Apply**. |
| Remove all sections. | a. Click **Bulk Add**.<br>b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click **Select All**.<br>c. Click **Apply**. |
| Rearrange sections of the report template. | Click-drag the section boxes to rearrange the order. |

You can add the following items and sections to a report template multiple times. Each section, except for a page break, can be uniquely customized.

- **Variant Details**
- **Custom Text**

  You can add text that can be edited when you generate a report. For more information, see “Generate a variant report” on page 132.
- **Page Break**

The following items and sections can appear in the report template in one place.

- **Key Metrics**
- **Manual Signatures**
- **Sample Details**
- **QC**
- **Sequencing Run Details**
- **Reagents**
- **Electronic Signatures**

7. Enter and customize the information for each section of the report template.
   a. For each unlocked section of the template, in the **Configuration** pane on the left, click the section name of interest.
   b. In the **Section** pane on the right, add or remove information for the section.

      You can click any customizable field to populate a list of available options for the field.

   Sections that are locked and cannot be customized are designated with (locked).
8. To preview the report layout, click **Preview Report**.

   You must enter information in the required fields before you can preview a report template.
9. When you are finished, click **Save**.

   The new report template appears in the **Report Templates** list.

## Edit a draft report template (manager/administrator)

You can edit a variant report that is not locked for electronic sign-offs if you have account permissions to electronically sign reports. A locked report template cannot be edited.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to edit, then click **Edit**.
4. Edit the sections of the report template as desired, then click **Save**.

The report template remains a draft until you lock the report template. For more information, see “Lock a draft report template (manager/administrator)” on page 140.

## Delete a draft report template (manager/administrator)

When a report template is in draft status, it can be deleted from the software. Only draft report templates can be deleted.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to delete, then click **Delete**.
4. In the **Delete Report Template** dialog box, click **Yes** to confirm the deletion.

The draft report template is removed from the software.

## Lock a draft report template (manager/administrator)

Manager- and administrator-level users can lock draft report templates in the **Assays / Manage Presets** screen under the **Report Templates** tab to manage report templates.

When a manager or administrator first creates an report template, the status of the report template is **Draft**. After a report template is locked, it cannot be edited or deleted.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template that you want to lock, then click **Lock**.

The report template is now locked and cannot be edited or deleted.

## View or add notes to a report template (manager/administrator)

Manager- and administrator-level can view existing notes or add new notes to draft or locked report templates in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Presets**
2. In the **Manage Presets** screen, click the **Report Templates** tab.
3. In the **Report Templates** table, place the pointer over the row of the report template of interest, then click **Notes**.

| Action | Description |
|---|---|
| View existing notes for a report template. | In the **Add Notes** dialog box, notes that are associated with the report template are listed by user, date, and time. |
| Add a new note to a report template. | a. In the **Add Notes** dialog box, click + **Add Note**.<br>b. Enter the note, then click **Save**.<br>The note is added to the report template and includes the username for the account of the person who entered the note, and a timestamp that records the note entry date and time. |
| Delete a note from a report template | In the **Add Notes** dialog box, in the row of the note that you want to delete, click 🗑 **(Delete)**. |

Click **Close** to return to the list of report templates.

# Run report

The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Assay metrics and the run report” on page 109. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

## View a run report

You can view or download a run report in Genexus™ Dx Software.

For information to download a run report, see “Download a run report” on page 142.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, click the sample name of interest in the **Sample Name** column.
3. Click the **Reports** tab.
   A pane for the run report and each variant report that has been generated is shown. The name of the report is listed at the top of the pane.
4. In the **Run Report** pane, click **View Report**.

The run report opens in a new browser tab.

## Download a run report

You can download a run report summary in PDF format. The run report includes assay metrics and the record of reagents that were used in a run. For information about the contents of the run report, see “Assay metrics and the run report” on page 109. If you entered extraction kit barcodes for samples when you prepared library batches or when you planned the run, the extraction kit barcodes are listed in the run report.

1. In the menu bar, click **Results►Sample Results**.
2. In the **Sample Results** screen, in the **Sample Name** column, click the sample name of interest.
3. Click the **Reports** tab.
   Multiple panes including a **Run Report** pane, a **Variant Report** pane, and any panes for customized reports that have been generated are shown.
4. In the **Run Report** pane, click **Download Report** to download a run report summary in PDF format.

# 8 View and manage assays (manager/administrator)

## Assays in Genexus™ Dx Software

Assays contain the settings and parameters for library preparation, templating, controlling the sequencing run, analyzing, and reporting the results. Assays also define the panels, kits, and chips that are used in a run, and specify the reference files and threshold values for quality control and variant detection. The software files that contain the assay settings and parameters are packaged in a ZIP file called an assay definition file (ADF).

Assays can contain system-installed annotation sets, copy number baselines, sequence variant baselines and exon tile fusion baselines.

An assay is a reusable experimental design that contains predefined settings appropriate for use with common types of applications. An assay can be used to plan many runs and plays an important role in enabling rapid throughput across the sequencing instrument. Assays help reduce the chance of errors, because information is stored and then applied to runs instead of entered manually for each run.

## Find an assay

You can sort, search and filter information to narrow the list of assays shown in the **Assays / Manage Assays** screen, then find a specific assay in Genexus™ Dx Software.

For information to search for an assay by a unique identifier, such as the name of the assay, see “Search for assays” on page 144.

For information to filter assays by a common trait, such as the name of the creator of the assay, see “Filter and search in the list of assays” on page 145.

## Search for assays

You can search for assays in Genexus™ Dx Software with keywords or text that are included in the name or full name of an assay.

IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

1. Click **Assays►Manage Assays**.
2. In the **Manage Assays** screen, in the **Assay Full Name** column heading or **Assay Name** column heading, click **(Filter)**.
   a. In the search field, enter a full or partial assay name of at least 3 characters.
      When you have entered at least 3 characters, the filter dialog box limits the list in the filter dialog box based on the entry.
   b. In the filter dialog box, select from the limited list.
      - Select a checkbox next to the name of an assay to select one or more assays.
      - Click **Select All** to select all assays listed in the filter dialog box.

      Note: If you click **Clear**, all selections are removed and the filter dialog box closes.

   c. In the filter dialog box, click **Filter**.
      In the column heading, **(Filter)** changes color to indicate that a filter is applied.

The filtered list of assays is shown in the **Manage Assays** screen.

Click **Clear Filters** to remove all filters and view the full list of assays.

## Sort the list of assays

You can sort the list of assays to make it easier to find assays in Genexus™ Dx Software.

1. Click **Assays►Manage Assays** to open the **Manage Assays** screen.
2. Click a column heading in the list of assays.
   If the column is sortable, the contents of the column is sorted alphabetically, or chronologically.
3. Click the column heading again to reverse the order.

## Filter and search in the list of assays

You can apply filters to narrow search results, or shorten the list of assays when you view the list of assays in Genexus™ Dx Software.

You can also search for assays in Genexus™ Dx Software with keywords or text that are included in an assay name.

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IMPORTANT! Search terms must adhere to the following rules.

- An asterisk (*) is not allowed in the search field for use as a wildcard.
- Searches are case-insensitive, that is, both upper-case and lower-cases letters are found regardless the case of search term letters.
- Searches find every occurrence of a continuous string. For example, a search for *demo* in analysis names returns a list of all analyses with a name that includes *demo*. For example, demo1, demo2, and so on.
- Spaces are removed during searches, and therefore are not recommended. For example, a search for *demo 1* would return results only for data that includes the string *demo1*.

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1. Click **Assays►Manage Assays** to open the **Manage Assays** screen.
2. Click the **Filter Assays By** dropdown menu, then apply the filters.
   - Select **Status** to expand the list of options for status, then select a status:
     - Obsolete
     - Locked
   - Select **All** to return to the complete list of assays.

3. Select from the filter and search options in the column heading of interest.

| Option | Description |
|---|---|
| Filter and search by name of an object. | Click **(Filter)** in the column heading of interest, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial name for the object. Select one or more matches from the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen. |
| Filter and search by application type. | Click **(Filter)** in the **Application Type** column, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial name of the application type. Select one or more matches from the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen.<br>• Select from the application types shown, then click **Filter**. |
| Filter and search by the creator of the assay. | Click **(Filter)** in the **Created By** column, then select one or more of the search options.<br>• Enter at least 3 characters to search by the full or partial assay name. Select one or more matches listed in the filter dialog box. To select all matches, click **Select All**. Next, click **Filter**.<br>The filtered list of assays is shown in the **Manage Assays** screen.<br>• Select from the user names that are shown, then click **Filter**. |
| Filter by the exact or approximate date on which assays were created. | Click **(Filter)** in the **Created On** column, then use the options to filter by a date or range of dates to filter the list based on the assay creation date. |

4. *(Optional)* Click **Clear Filters** to return to the entire list of assays.

The list of assays is filtered according to the selections.

# View annotation set details

You can view details about an annotation set in Genexus™ Dx Software.

1. Click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, in the **Annotation Sets** tab, click the annotation set name.

The following details for the selected annotation set are shown.

Annotation Set Details

| Item | Description |
|---|---|
| Analysis Pipeline Version | The analysis pipeline version that is used by the annotation set. |
| Last Modified By | For custom annotation sets, the sign-in name of the person who created or modified a custom annotation set. |
| Last Modified On | Date on which a custom annotation set was last modified. |
| Created By | Custom annotation sets show the sign-in name of the person who created the annotation set.<br>Annotation sets that are included with the software are labeled as System Installed. |
| Created On | Date on which a custom annotation set was created. |
| Status | Status of the annotation set, including:<br>• Locked |
| Reference | Genome reference that is available in the annotation set.<br>• hg19<br>• GRCh38 |
| Annotation Versions | A list of the annotations included in the annotation, and the version of each annotation. |

# Copy number baselines (manager/administrator)

A copy number baseline is a set of control samples that are used to create a baseline for the detection of copy number variants (CNVs). A copy number baseline is associated with a panel file in the software. CNV analysis is based on reads counts for each amplicon.

Known sources of variability include pool imbalance (when an assay has more than one pool of amplicons), total number of reads, per-amplicon attributes of GC proportion, and length of the amplicon insert. In practice, we observe other variability that does not associate with known attributes yet is systematic.

Copy number estimates are made using a proprietary algorithm.

Genexus™ Dx Software includes predefined copy number baselines.

## View copy number baseline details (manager/administrator)

Manager- and administrator-lever users can view details for the system-installed copy number baselines.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Copy Number Baselines** tab.
   The **Copy Number Baselines** table lists both system-installed copy number baselines.
3. Click the name of a copy number baseline.
4. Review the details in the **Copy Number Baseline Details** pane.
   The details that are shown depend on the setup of the copy number baseline.

Copy Number Baseline Details

| Item | Description |
|---|---|
| Created By | Sign-in name of the person who created the baseline. System-installed baselines are included with the software. |
| Created On | Date on which the baseline was created. |
| Number of Normal Samples | The number of normal samples that are included in the baseline. |
| Gender | The biological sex of the sample: Female, Male, or Unknown. |
| Number of Samples | The total number samples that are included in the baseline. |
| Amplification Type | Library amplification technology that is used in the baseline (Ion AmpliSeq™ or Ion AmpliSeq™ HD). |
| Panel Name | Name of the panel that is included in the baseline. |
| Reference | The reference genome for the baseline. |
| Reference File Name | The name of the reference genome file that is used in the baseline. |

# Exon tile assay baselines

An exon tile assay baseline is used to model the amplicon expression variation within a gene. When added to an assay in Genexus™ Dx Software, the exon tile fusion baseline normalizes a test sample, corrects the coverage in exon-tile amplicons, and identifies the presence or absence of expression imbalance in a gene. Exon tile assay baselines that you have created can only be incorporated into some assays. Contact a Field Bioinformatics Specialist (FBS) for guidance.

An exon tile assay baseline is a TXT file that contains the following items.

- The normalized expression values of exon-tile amplicons for genes with imbalance assays.
- The median normalized expression for each amplicon.

Exon tile assay baselines are associated with a panel BED file in the software.

Genexus™ Dx Software includes predefined exon tile assay baselines.

## View exon tile assay baseline details (manager/administrator)

Manager- and administrator-lever users can view details for the system-installed exon tile assay baselines.

1. In the menu bar, click **Assays►Manage Presets**.
2. In the **Assays / Manage Presets** screen, click the **Exon Tile Assay Baselines** tab.
   The **Exon Tile Assay Baselines** table lists the system-installed exon tile assay baselines.
3. Click the name of an exon tile assay baseline.

Review the details in the **Exon Tile Assay Baselines** pane.

The details that are shown depend on the setup of the exon tile assay baseline.

Exon Tile Assay Baselines

| Item | Description |
|---|---|
| Created By | Sign-in name of the person who created the baseline. System-installed baselines are included with the software. |
| Created On | Date on which the baseline was created. |
| Number of Samples | The total number samples that are included in the baseline. |
| Amplification Type | Library amplification technology that is used in the baseline. |
| Panel Name | Name of the panel that is included in the baseline. |
| Reference | The file name of the fusion reference for the baseline. |

# Manage assays

You can view assay details, and view and download the audit trail of an assay in Genexus™ Dx Software.

Manager- and administrator-level users can also import an assay into the software.

## View assay details

You can view assay details in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Assays**.
2. In the **Manage Assays** screen, click the assay name of interest.

The assay details are shown.

If desired, click **(More Options)►Audit** to audit the assay. For more information, see “Review and export an assay history” on page 151.

## Assay details

When you view assay details in Genexus™ Dx Software, you can view information about the components of the assay. The information that is available depends on the type of assay.

You can view the following assay creation details.

- **Application Type**—The type of application for the assay, such as **DNA and Fusions**.
- **Created By**—The sign-in name of the person who created the assay.

  Assays that are included with the software are labeled as **System Installed**.
- **Analysis Pipeline Version**—The version of the analysis pipeline used for the assay.
- **Created On**—The date on which the assay was created.
- **Assay Name**—The name and version of the assay.

  Note: For system-installed assays or assays that you install in the software, the end of the assay name, for example w5.0.0, is the version number of the assay. The assay version number is not the same as the **Version** that is shown in the assay creation details.
- **Version**—The revision number of the assay.

| Assay detail[1] | Description |
|---|---|
| Panel | In addition to other components, the panel section lists the reference files that are used in an assay.<br>One item that is listed is the Minimum Read Count Per Sample, which is the minimum number of reads per sample that are needed to achieve the coverage depth required for the assay. The minimum read counts per sample is used to calculate the number of samples that are allowed in a chip lane when you plan a run. |
| Reagents | In addition to other components, the reagents section lists the kits and chips for the assay. |
| • NTC QC – RNA<br>• CF-1 Control<br>• Purification QC<br>• NTC QC – DNA<br>• Run QC<br>• Sample QC | The QC sections list the Quality Control (QC) metric values for the assay. |
| Plugins | The plugins sections lists the plugins that are associated with the assay. |
| Description | A short description of the assay, similar to the assay full name. |

[1] The details that are shown depend on the assay.

## Review and export an assay history

You can review, export, or print the entire history of changes for an assay. The history shows the original values and any new values or activities.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **Assays►Manage Assays**.
2. In the **Manage Assays** screen, place the cursor in the row for the assay of interest, then click **Audit**.
   The **Audit Details** screen opens with time-stamped audit records that include assay details and changes for each action performed on the assay.
3. *(Optional)* Review the audit record.

   Details in the audit record

| Item | Description |
|---|---|
| User name | The name of the person who was signed in when the action was performed. |
| Assay Full Name | The name of the assay. |
| Data Object Name | The data object that is affected by the action. |
| Action Performed | Actions performed on the assay include:<br>• Create<br>Note: The create action includes imports of assays. |
| Time Stamp | The time and date of the action. |
| ID | An identifier to represent a specific action in the audit record. Each action in the audit record includes a unique identifier. |

4. *(Optional)* Click **Download** to download a print-ready PDF file of the audit record.
   You can open the PDF file in an appropriate viewer, then print the record from within the open document.
5. Click **Back** to return to the **Manage Assays** screen.

## Import an assay (manager/administrator)

A manager- or administrator-level user can import an assay from another Genexus™ Dx Integrated Sequencer if the assay has been first exported to a local drive.

Ensure that the name of the assay, the barcode kit, and the extraction method type for the source system and the target system are identical.

1. In the menu bar, click **Assays**►**Manage Assays**.
2. In the **Manage Assays** screen, click **Import Assay**.
3. In the **Import Assay** dialog box, click **Select Assay file**, then go to the folder on the computer that contains the exported assay and select the ZIP file for the assay.
4. Click **Upload**.

The assay appears in the list of assays in the **Assays**►**Manage Assays** screen.

# 9 System administration and management (manager/administrator)

Manager-level users use tools in the **(Settings)** menu to:

Administrator-level users use tools in the **(Settings)** menu to:

- View and export audit records.
- Configure user email settings, set up laboratory information, and audit these settings.
- Create and manage account settings, and audit these accounts.
- Manage software updates.
- Manage data backup, disk cleanup settings, and disk space usage.
- Manage log files.
- Manage services for servers, services, and active jobs.
- Create and manage user accounts.

## Accounts for use with Thermo Fisher™ Connect Platform in Genexus™ Dx Software (manager/administrator)

Manager- and administrator-level users can configure Genexus™ Dx Software to link to one or more Connect Platform accounts.

The first Connect Platform account that is added in the software is designated as the administrator (Admin) account and cannot be unlinked or removed.

Active Connect Platform accounts can perform the following tasks in the Genexus™ Dx Software:

- Download the latest software updates
- Download assay definition files
- Download configuration packages

Note: Configuration packages contain software updates, such as updates for kits and chips, documents such as an updated End-user License Agreement (EULA), Help documentation, and so on.

# Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)

Manager- and administrator-level users can link their Connect account at Genexus™ Dx Software with a QR code generated in the software.

When a Connect account at **thermofisher.com/connect** is configured and active, administrator-level users can perform the following tasks in the Genexus™ Dx Software:

- Download the latest software updates
- Download assay definition files
- Download software configuration packages

Before you link a Connect account to create a **Thermo Fisher Connect** account, you must have a valid Connect account. You must also have the InstrumentConnect mobile app to link the account with a QR code that is scanned on a mobile device. For more information, see the *Thermo Fisher™ Connect Platform Help System* (Pub. No. MAN0010954).

1. Click **(Settings)►Thermo Fisher Account**.
2. Click **Link Account to Device**.
   The **Link Account to Device** dialog box opens and a QR code image and QR code text are shown.
3. Link the Connect account with the software.

| Option | Description |
|---|---|
| Link a Connect account with the InstrumentConnect mobile app | a. Open the InstrumentConnect app, then sign in.<br>b. In the app, click +.<br>c. Click QR Code, then scan the QR code shown in the **Link Account to Device** dialog box in Genexus™ Dx Software. |
| Link the account through the Connect website | a. Click the link in the QR Code to open the Thermo Fisher Connect website.<br>b. In the Connect website, click **InstrumentConnect** in the left sidebar.<br>c. Click **Link instrument** in the upper right corner of the screen.<br>d. Enter the QR code text shown in the **Link Instrument** screen.<br>e. Click **Link**. |

The linked account is listed in the **Settings / Thermo Fisher Account** screen. A successfully authenticated account has **Active** listed in the **Status** column.

When the **Thermo Fisher Account** is active, software updates are available in Genexus™ Dx Software. Administrator-level users can download and install the updates. For more information, see “Software updates” on page 165.

## Unlink a Thermo Fisher™ Connect Platform account (manager/administrator)

Manager- and administrator-level users can unlink Thermo Fisher™ Connect Platform accounts in Genexus™ Dx Software.

The first Thermo Fisher™ Connect Platform account that is added is designated as the administrator (Admin) account and cannot be unlinked or removed in the software.

1. Click **(Settings)**►**Thermo Fisher Account**.
2. Place the pointer over the row of the account that you want to unlink, then click **Unlink**.

The Thermo Fisher™ Connect Platform account is no longer linked in Genexus™ Dx Software.

## View the changes made to a connected account (manager/administrator)

Manager- and administrator-level users can view the changes made to connected accounts in Genexus™ Dx Software.

1. Click **(Settings)**►**Thermo Fisher Account**.
2. In the **Thermo Fisher Account Settings** screen, place the pointer over the row of the account that you want to audit, then click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected account.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made to the selected account.
5. *(Optional)* Click **Download** to export the record of the single change to a PDF file.

# User accounts (administrator)

Administrator-level users can perform user management actions in the **Settings / User Management** screen.

- Add new user accounts
- Assign user privileges (roles)
- Edit user information
- View user account audit trails
- Manage user account security policies

## Add a new user account (administrator)

Administrator-level users add new user accounts to the software, and grant some users permissions to electronically sign reports.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 165.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, click **Add New**.
3. In the **Create User** dialog box, enter the user account information.
   User names can contain alphanumeric characters and underscores, periods, and hyphens.
4. From the **Role** list, select the appropriate user-access level (**Administrator**, **Manager**, or **Operator**.
   The **Report** role has electronic signature permissions enabled by default.
5. Grant permissions for a manager, administrator, or report user to electronically sign reports.
   a. Enable the **Electronic Signature** toggle switch.
   b. Select an **Electronic Signature Type** to associate with the user account. For example, **Approved**. A **(Lock)** is shown when you select the **Electronic Signature Type** that is configured to lock a report upon sign off.
6. Click **Save**.
   The new account is added to the list in the **User Management** screen. The software sends an email notification containing a temporary password to the email address of the new user.

## Sort the user list (admin)

Administrator-level users can sort the user list by user name, first name, and last name.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, click the column for **User Name**, **First Name**, or **Last Name** to sort the column alphabetically.
3. Click the column name a second time to reverse the order.

## Edit user details (administrator)

Administrator-level users can edit user details in the **Settings / User Management** screen. An administrator can also suspend or disable user accounts. A history of user account changes is maintained by the audit feature.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, place the pointer over the row of the user account, then click **Edit**.
3. In the **Edit User** dialog box, enter changes in the appropriate account information fields.
   - In the **Role** list, select the user-access level (**Administrator**, **Manager**, or **Operator**.
     The **Report** role has electronic signature permissions enabled by default.
   - To grant permissions for administrator, manager or report users to sign reports, enable the **Electronic Signature** toggle switch, and select an **Electronic Signature Type**.
     A **(Lock)** is shown when you select the **Electronic Signature Type** that is configured to lock a report upon sign off.
   - In the **State** list, select the appropriate state (**Active**, **Suspended**, or **Disable**).
4. Click **Save** to make the changes.

Changes that are made to the user account are shown in the **User Management** screen, and shown in the **Audit Trail** screen for the user.

## Reset a user password (Administrator)

An administrator-level user resets passwords for user accounts. A history of user password changes is maintained by Genexus™ Dx Software.

Periodically, the software sends a system-generated email notification to a user that their password is about to expire. The account holder can follow the instructions in the email to change a password. A user can also request that an administrator reset their password.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

The email settings in the software must be configured before new users can receive email notifications. For more information, see “Configure email notifications (manager/administrator)” on page 165.

Users with administrator accounts can change a password immediately, or send an email to a user who requests to have a password changed.

1. To change a password immediately, do one of the following actions, depending on the account with which you sign in and the type of user account requires a password change.

| Sign-in requirements for password change | Action |
|---|---|
| To change the password for any user account, except for the system adminstrator account, you must be signed in with an administrator account. | • Click **(Settings)►User Management**, then place the pointer over the row of the account for which the password will be changed, then click **Reset Password**.<br>• In the **Change Password** dialog box, enter a new password in the **New Password** field and re-enter the new password in the **Confirm Password** field, then click **Change**. |
| To change the password for any user account, including the system administrator account, you must be signed in with the account for which the password will be changed. | • Go to **(Profile)►My Profile**, then click the **Change Password** link.<br>• In the **Change Password** dialog box, enter the information in the **Current Password**, **New Password**, and **Confirm Password** fields, then click **Change**. |

   The confirmation message "Password changed successfully" is shown to indicate the password reset.

2. To send an email to a user who requests to have a password changed, click **(Settings)►User Management**, then place the pointer over the row of the account for which the password will be changed, and click **Reset Password**. Then in the **Change Password** dialog box, click **Send Password in Email** to send a software-generated password in an email message to the user.
   A confirmation message is shown to indicate the email was sent to the user.

## View user account history (administrator)

An administrator-level user can view the user account history in Genexus™ Dx Software. The user account audit trail is a record of when the user account was created and modified.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, place the pointer over the row of the user of interest, then click **Audit**.
3. In the **Audit Details** screen, view the audit trail details for the selected user. For more information, in the **Audit Record Details** column, click the time stamp of interest.
4. *(Optional)* In the **User Management** screen, click **Download** to export the record to a PDF file.

## Set user account security policies (administrator)

An administrator-level user sets security policies for Genexus™ Dx Software user accounts. User account security policies include the permissible number of failed sign-in attempts, password lifetime, and the length of inactivity before users are automatically signed out of the software. These settings apply to all users.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►User Management**.
2. In the **User Management** screen, click **Policies**.
3. In the **Policies** dialog box, select the appropriate values to set the user-account suspension period, password, and session timeout policies.
4. Confirm that the **Enabled** toggle switch is set to enable the session policy settings.
5. Click **Save**.

## Configure an SSH key (system administrator)

System administrator-level users can set up an SSH key in their user profile in the **Profile / My Profile** screen. SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives administrator-level users a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

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IMPORTANT! Ensure that you use only an SSH key that is password protected. The software does not enforce the use of password protected SSH keys, but the use of a password for the SSH keys is strongly recommended.

Do not share the SSH private key or the credentials for the SSH private key. The SSH private key should be used only by the user who creates the SSH private key.

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Configuring an SSH key is optional, but best practice is to configure one or more SSH keys. The ionadmin and ionservice users may need to configure SSH keys so that they can access the system at the operating system level to configure some system-level settings, for example mounting an archive directory on the system. A user can have more than one SSH key.

Before configuring an SSH key, you must first generate keys by using the Keygen command on your local system. When the public and private keys are generated, copy the text string from the public key to use in this procedure.

1. In the menu bar, click **(Profile)►My Profile**.
2. In the **SSH Keys** section, click **Add SSH Key**.
   The **SSH Keys** section is available only to ionadmin and ionservice users.
3. Complete the **Add new SSH Key** dialog box.
   a. In **Label**, type a unique name for this SSH key.

b. In **SSH Key**, paste or type the SSH key string generated by using the Keygen command on your local system.

c. Click **Save**.

# Audit records (administrator)

Administrator-level users can use the tools in the **Settings / Audit Records** screen to configure and manage audit records. Information that can be audited includes activities by users on samples, libraries, assays, planned runs, system setup of the sequencer, accounts, and software updates.

The **Audit Records** screen lists actions that are performed on any data object, including the user who performed the action, the action performed, the data object name, and the date and time of the action. Additional details concerning the action are available in an accompanying audit record.

The actions can be searched and sorted, and the audit records can be exported and printed.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

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Note: Library batches and runs that are created in a batch with other objects in a single LIMS transaction do not have a "Create" action that is listed in the **Settings / Audit Records** screen. However, you can view complete library batch audit records from the **Samples / Manage Library Batches** and **Runs / Manage Runs** screens. Any subsequent actions that are performed on library batches are listed in the **Audit Records** screen. A library batch or run that is created individually in a LIMS transaction is listed.

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## Search audit records (administrator)

An administrator-level user can search for an existing audit record with the search tools in the **Audit Records** screen.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.

2. In the **Audit Records** screen, select the search parameters.
   a. To select a date range, click **Calendar** next to **Start Date** and **End Date**.

      If an end date is not selected, the search results include all records from the start date through the current date.

   b. To limit the results list to actions that are performed by a specific user, select the user from the **Select User** list.

   c. To limit the results to specific actions, select from the **Select Action** list. If you leave **Select Action** as the selection, all audited actions are shown in the search results.

d. To list other items in the table, select from the data object list.

   The list is set to **Sample** by default and all items listed in the **Audit Records** screen have the data object of **Sample** in the **Data Object Name** column. You can change the type of objects that are shown in the table. For example, you can search for audit records of run plans when you select **Planned Runs** in the data object list.

3. Click **Search**.

4. Click **Clear** to return to the default list of records, with the data object of **Sample** in the **Data Object Name** column.

## Sort audit records (administrator)

An administrator-level user can sort the columns in the **Settings / Audit Records** screen to display the list of audit records in a different order. The default order is the newest record on top.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.

2. In the **Audit Records** screen, click the column heading of interest.
   The list of records is reordered based on the heading selected. **User**, **Action Performed**, and **Data Object Name** sort alphabetically.

3. Click the column heading a second time to reverse the order of records displayed.

4. Click **Clear** to return to the sorted column to the default order.

5. Click the **Timestamp** heading to return to the default order, with the most recent record on top.

## Export audit records (administrator)

An administrator-level user can export a print-ready PDF file of selected audit records from the **Audit Records** screen.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings)►Audit Records**.

2. In the **Audit Records** screen, select the record or records to be exported by clicking the checkbox next to the record of interest. Select the checkbox above the column to select all audit records.

3. Click **Export**.
   A PDF file is generated. Depending on your browser settings, the software automatically downloads the file or prompts you to open or save the file.

4. Open the PDF file in an appropriate viewer to review and print the audit record or records.

## Update the audit configuration (administrator)

An administrator-level user configures the audit details by requiring that a reason for a change be included in audit records when a user changes information. When a reason is required, the user must supply a reason for change when editing the information, and the reason is included in the audit record.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. In the menu bar, click **(Settings) ►Audit Records**.
2. In the **Audit Records** screen, click **Audit Configuration**.
3. In the **Audit Configuration** dialog box, select the **Require Reason** checkbox next to the **Data Object Name** for which a reason for change is required.
4. Click **Save**.

   To remove a reason for change requirement, open the **Audit Configuration** dialog box, deselect the checkbox, then click **Save**.

# Configure electronic signatures (administrator)

Administrator-level users configure electronic signatures in Genexus™ Dx Software. For more information about electronic signatures, see “Sign off variant reports” on page 133.

## Manage electronic signature types (administrator)

Signature types in Genexus™ Dx Software are associated with electronic signature permissions to designate how a signature is used. Different types of signatures, such as review, approve, and lock, can be required to sign off on a variant report.

Administrator-level users can add new electronic signature types. Each user account can be associated with just one signature type. Signature types can be renamed, made obsolete, restored, or deleted. Changes to electronic signature types are shown in the audit trail for the signature type.

Designate one of the signatures to lock the report during sign-off.

1. Click **(Settings)►Configuration**.
2. In the **Configuration** screen, in the **Electronic Signature** section, manage signatures.

| Option | Description |
|---|---|
| Add new types of electronic signatures. | In the Bulk add list of comma delimited meaning of signature field, enter new types of signatures, separated by commas. For example, enter *Review,Inconclusive* to add two new signature types.<br>Signature types that are added can be associated with users who are granted permission to electrically sign reports. For more information, see “Add a new user account (administrator)” on page 156 and “Edit user details (administrator)” on page 157. |

*(continued)*

| Option | Description |
|---|---|
| Rename a type of signature. | Click Edit, then enter a new name for the signature type. |
| Associate locking a report with a type of signature. A report becomes locked when a user whose account is associated with the selected type of signature electronically signs the report. | In the Select a type of signature to lock report when signing off section, select the radio button next to the type of signature that you want to use to lock a report during report sign-off. |
| Obsolete a type of signature. | Click Obsolete to remove the signature type as an option during variant report sign-off.<br>If an obsolete signature type is associated with a user account, an alert is added to the User Management screen next to Electronic Signature Type. An alert is also shown if no signature type is associated with a user account that includes permissions for electronic signatures. The signature type must be edited by an administrator before the user can electronically sign reports. For more information, see “Edit user details (administrator)” on page 157. |
| Restore a type of signature. | Click Reactivate to reactivate a type of signature that has been made obsolete. |
| Delete a custom type of signature. | Click Delete to delete a type of signature. A custom electronic signature type can be deleted if is not associated with a user, is not set as lock-level, and is not used in electronic sign-offs for any reports. System-installed signature types cannot be deleted. |
| Review the changes made to a type of signature. | 1. Click Audit.<br>2. In the Audit Trail - Electronic Signature screen:<br>3. • Review the audit trail details for the most recent change.<br>• Click a time stamp of interest to view the details for other changes that have been made to the selected signature.<br>• Click Download to export the record of the single change to a PDF file.<br>• Click Back to return to the Configuration screen. |

# Configure email settings (manager/administrator)

Manager- and administrator-level users configure email settings. Administrator-level users can configure support contact information, such as the lab contact and the IT contact for the organization.

# Lab information

The **Lab Information** tab in the **Settings / Configuration** screen provides a reference for support personnel, listing points of contact if a problem with the connected Genexus™ Dx Integrated Sequencer occurs.

- **Lab Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer.
- **IT Contact**—This is the person in your organization who should be notified during a support request of problems related to the sequencer hardware or the network environment.

IMPORTANT! Administrator and Manager-level users should review the contact information periodically to ensure that it is current and accurate.

## Edit the reference lab information (manager/administrator)

Manager- and administrator-level users can edit the reference lab information in Genexus™ Dx Software. You can view the history of changes to the contact information. For more information, see "View the changes made to the lab information (manager/administrator)" on page 164.

1. Click **(Settings)►Configuration**.
2. In the **Lab Information** section, below the **Lab Contact** or **IT Contact** pane, click **Edit**.
   The fields for the **Lab Contact** or **IT Contact** are now available to edit.
3. Enter the updated contact information.
4. Click **Save** to save the new selections or click **Reset** to revert to the original information.

## View the changes made to the lab information (manager/administrator)

Manager- and administrator-level users can view the changes made to the lab information in Genexus™ Dx Software.

1. Click **(Settings)►Configuration**.
2. In the **Lab Information** section, in the **Lab Contact** or **IT Contact** pane, click **Audit**.
3. In the **Audit Details** screen, view the most recent change for the selected contact.
4. In the **Audit Record Details** pane, click a time stamp of interest to view the details for other changes that have been made for the selected contact.
5. *(Optional)* Click **Download** to export the record of the single change to a PDF file.

# Configure email notifications (manager/administrator)

Manager- and administrator-level users configure the Genexus™ Dx Software to send email notifications to a specified administrator.

Administrator-level users receive email notifications when there is a restore system failure. Administrator-level users can also select to receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. For more information, see “Change email notification settings (administrator)” on page 182.

1. Obtain the SMTP server URL and port information from your IT department.
2. Click **(Settings)►Configuration**.
3. In the **Configuration** screen, in the **Email Settings** section, enter the server and port information supplied by IT, and the Genexus™ Dx Software user name, password, and email address of the administrator.
   - Keep the **Use TLS** toggle switch enabled to send email notifications with transport layer security.
   - Click the **Use TLS** toggle switch to send email notifications without transport layer security.
4. Click **Update**.
5. Click **Send Test Email** to send a test email to the email address provided in the settings.

# Software updates

An administrator-level user can select **Software Updates** in the **(Settings)** menu to:

- See the currently installed software versions for each software module, assay definition files, plugin, and configuration package installed on the system under the **Installed Software** link.
- Check for new software updates that are available to be downloaded and installed under the **Software Updates** link.

For users who have set up a Thermo Fisher™ Connect Platform account, **(Notifications)** appears in the Genexus™ Dx Software banner on the top of the screen upon login when updated software packages are available for download from the Connect Platform.

Updated software packages are available from the App Store at **apps.thermofisher.com**. Software packages can include assay definition files, for example. Administrator-level users can download and install software directly from the App Store if a Connect Platform account has been configured. Alternatively, a USB drive can be used to install and update the software if the instrument is not connected to the internet.

| Option | For more information, see this section |
|---|---|
| Download and install software directly | "Download and install software from the App Store (administrator)" on page 166 |
| Download and install software with a USB drive | "Format a USB drive for a software update" on page 167<br>"Download and install software with a USB drive (administrator)" on page 167 |

Note: For first-time update of system software from the 5.100.2 version, contact a Field Service Engineer (FSE) to update the system software.

IMPORTANT! Before updating the software, ensure that the instrument is idle, no runs are in progress or are queued to run, and the screen on the instrument is the main menu screen.

## Download and install software from the App Store (administrator)

In order to download and install software updates from the App Store on **thermofisher.com/connect**, you must first:

- Configure a Thermo Fisher Connect account. For instructions, see "Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)" on page 154.
- Ensure that the Genexus™ Dx Integrated Sequencer has the latest software version, Genexus™ Dx Software 6.8.2.
- Connect the Genexus™ Dx Integrated Sequencer to the internet.

1. In the menu bar, click **(Settings)►Software Updates**, then click **Software Updates** in the upper right corner.
   The **Software Updates** screen opens with **App Store** enabled.
2. *(Optional)* Click the desired package name to view the installation details and version information in the **View Package Details** pane.
3. Click **Download** in the **Actions** column in the row of the software package of interest.
   After the download is complete, the **Download** link changes to an **Install** link.
4. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install.
   If you are performing a system software module installation, a command window opens to show the progress of the installation.

   Note: There may be a delay before the command window opens.
5. When installation of the software package is complete, click **Installed Software** to verify the installation.

## Format a USB drive for a software update

Prerequisites:

- An Ubuntu Linux desktop computer with disk utility software.
- A USB drive with a capacity of 128 GB or more.

IMPORTANT! Ensure that the USB drive does not contain any viruses or malware before you insert the USB drive into the instrument.

1. Insert the USB drive into the Ubuntu Linux computer.
2. With Ubuntu running, open the disk utility by navigating to **System ►Administration►Disk Utility**.
3. If the USB drive has partitions, delete the partitions.
   a. Under **Peripheral Devices from Storage Devices**, click the **USB Flash Drive (SMI USB DISK)** to select the USB drive.
   b. Highlight the volumes (New Volume FAT >60 GB).
   c. Click **Unmount Volume**, then click **Delete Partition**.
   d. Click **Delete** to confirm.
4. Select the free USB volume, then click **Create Partition**.
   The **Create partition on SMI USB DISK** window opens.
5. Deselect the **Take ownership of file system** checkbox.
6. Select **ext3** from the **Type** dropdown menu, then select the entire size of the partition. Name the partition *usbinstall*.
   The partition name must be completely lowercase.
7. Click **Mount**.
   The mount point of the USB drive is **/media/usbinstall**.

Proceed to “Download and install software with a USB drive (administrator)”.

## Download and install software with a USB drive (administrator)

In order to download and install software updates, you must first:

- Create or access a Thermo Fisher™ Connect Platform account. For instructions, see “Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)” on page 154.
- Format a USB drive for the software update. For more information, see “Format a USB drive for a software update” on page 167.

1. In the Ubuntu Linux computer where the USB is mounted, sign in to the Thermo Fisher App Store on Connect at **apps.thermofisher.com**.
2. On the left side of the App Store screen, click **(AppConnect)**.
3. On the right side of the screen, under **Resource Libraries**, click **Ion Torrent™ Genexus™**.
4. Click **Software Update**.
5. In the Ion Torrent™ Genexus™ Software table, select the row that contains the software package for the version that you want to install.
6. Click **(Download)** to download the package.
7. Copy the downloaded package (.zip) to the `/media/usbinstall` folder in the USB drive.
8. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the Genexus™ Integrated Sequencer.
9. In the menu bar of Genexus™ Dx Software, click **(Settings)►Software Updates**, then click **Software Updates** in the upper right corner.
10. Select **USB**.
11. Click **Install**. When the confirmation window appears, click **Yes** to confirm that you want to install.

When installation of the software package is complete, click **Installed Software** to verify the installation.

# Manage backup settings and restore runs (administrator)

Administrator-level users can view and manage backup settings and restore runs in Genexus™ Dx Software with the following actions:

- Monitor system storage space usage.
- Manage settings to backup and delete files and data.
- View an audit trail of any changes made.
- View and change email notification settings.
- Restore runs.

## Disk space usage and backup settings (administrator)

An administrator-level user can monitor disk space usage for the Genexus™ Dx Integrated Sequencer and can change data backup settings in Genexus™ Dx Software. System disk space usage information is available in the **Settings / Backup & Restore** menu. You can view the date and time of the last successful backup for data and analysis results and the date and time of the last successful backup for additional files such as sequencing output and intermediate files, based on the backup settings.

A best practice to maintain sufficient disk space is a systematic plan that archives run data, results files, variant information, and signed reports that you want to store to an external file storage system. A system administrator or Field Service Engineer (FSE) sets up a data backup strategy and disk cleanup settings. An administrator-level user then configures the backup schedule. For more information, see “Change the backup settings (administrator)” on page 177.

Before data can be backed up or archived, you must add a mount point. When files are backed up, the files are copied to an external storage device or to a cloud system. The files remain on both the sequencer and the external storage system. After data backup, files are saved in a folder in the external storage that is named `dx`. If multiple sequencers are connected to the software, the software generates a parent folder for each instrument.

When files are cleaned up, the files are deleted from the sequencer, but remain on the external storage system.

Backup and cleanup settings

| Option | Description |
|---|---|
| Back up system data and run results | Backup occurs daily, at approximately midnight, after an administrator-level user adds a mount point.<br>After backup, you can view the results for runs that are backed up in the Run Results and Sample Results screens. |
| Clean up system data and run results | If enabled, run results are deleted from the sequencer after a specified interval of time. For example, 12 months after a run is completed. Runs that are backed up and deleted are marked as Archived.<br>Cleanup settings do not apply for run results that you retain. When you retain run results, the run data, including the run results, 1.wells files, and intermediate files, are retained on the instrument and in the storage system. Runs that have been retained are shown in the software and are not deleted.<br>Archived runs are listed in the Run Results screen with the archived symbol ( ) after the run name. By default, archived runs are not listed. To see the archived runs, select Archived in the Filter runs by list. An administrator-level user can restore the runs to access the data. |

Backup and cleanup settings *(continued)*

| Option | Description |
|---|---|
| Back up intermediate files and 1.wells files | If enabled, intermediate files and 1.wells files are backed up at a specified time. For example, every Friday at 10 pm.<br>Because 1.wells files are 10 times larger than the run results files (BAM files), more time is needed to back up 1.wells files than to back up system data and run results. |
| Clean up intermediate files and 1.wells files | If enabled, intermediate files and 1.wells files are deleted when the system disk space falls below a specified threshold, such as 4 TB. If you have not configured the mount point, or if you have not configured the backup settings for additional files, files are cleaned up when the space threshold is met.<br>Cleanup settings do not apply for run results that you retain. When you retain run results, the run data, including the run results, 1.wells files, and intermediate files, are retained on the instrument and in the storage system. Runs that have been retained are shown in the software and are not deleted.<br>Because 1.wells files are 10 times larger than the run results files (BAM files), more time is needed to back up 1.wells files than to backup system data and run results. |

The software ensures that disk space is available for runs. If the system has less than 1 terabyte (TB) of unused disk space, no new runs can be started on the sequencer. In this case, an alert notifies the user that there is insufficient disk space when a run is set up on the sequencer. The run cannot proceed until data are backed up and deleted.

An administrator-level user can perform the following actions to free up system disk space, if needed.

- In the **Backup & Restore** screen, you can click **Start Now** to start a backup and cleanup at any time.
- In the **Run Results** screen, you can delete one or more run results.
- In the **Run Results** screen, you can archive one or more run results if you have added a mount point. The settings in the **Backup and Restore** screen are not applied when you archive runs in the **Run Results** screen.

Contact a local system administrator to backup and delete data manually. An administrator-level user can also change the schedule to backup and delete data.

Results and reports that have been backed up can be restored in Genexus™ Dx Software. For more information, see “Restore run results for a single run (administrator)” on page 183.

## Types of files for backup

There are three categories of files for backup in Genexus™ Dx Software.

| Category | Types of files | Cumulative size per run | Reanalysis use |
|---|---|---|---|
| Sequencing output | 1.wells | 250–500 GB | Reanalysis from the basecalling step |
| Intermediate files | Troubleshooting | 150–180 GB | Not applicable |
| Analysis results | • BAM<br>• VCF<br>• Reports | 150–180 GB | Reanalysis from the alignment step |

## Specifications and disk space usage

The amount of disk space that the Genexus™ Dx Integrated Sequencer uses for a run depends on the nucleic acid sample type and the assay that is used for sequencing. The system can store 40 to 50 whole chip data runs. If the system has less than 1 terabyte (TB) of available storage space, no new runs can be started. For more information, see “Disk space usage and backup settings (administrator)” on page 169.

The following table lists chip specifications and data usage for a run with the Ion Torrent™ GX5™ chip.

| Assay | Chip specifications and data usage |
|---|---|
| Sample type | FFPE |
| Number of reads per chip (4 lanes) | 48–60 million |
| Number of reads per lane | 12–15 million |
| Signal processing input size per chip[1] | ~2 terabytes |
| Basecalling input size per chip[2] | 200–300 gigabytes |
| Data size of intermediate and results files and logs[3] | 120–180 gigabytes |
| Data size per lane of intermediate and results files and logs | 30–45 gigabytes |
| Data size of backed up files and logs | 30–45 gigabytes |
| Data size of results files per lane[4] | ~8–24 gigabytes |
| Recommended number of samples per lane | 4 samples per lane |
| Data size of the results per sample | ~2–6 gigabytes |

[1] The signal processing input on the sequencer is deleted after analysis is complete and successful.

[2] Administrator- level users can schedule when basecalling input files are deleted. By default, these files are not retained. For more information, see “Set a schedule to permanently delete files (administrator)” on page 179.

[3] Includes QC, plugins, logs, results, and other assay and variant database files.

[4] For more information about results files, see “Results files” on page 121.

# Configure backup settings (administrator)

An administrator-level user views and configures sequencer backup settings in the **Settings / Backup & Restore** screen.

Update backup settings for various reasons, including:

- The backup location has limited free disk space remaining.
- The backup location is not accessible when the system tries to back up a run.

Consult a local Field Service Engineer to set up a backup system. For more information, see “Set up a backup server (administrator)” on page 176.

The following options are available only after a backup directory has been configured:

- View the backup disk space.
- Change the backup settings.

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup Settings** tab, edit the backup settings.

| Option | Description |
|---|---|
| View the system disk space | View the chart that shows the percentage of system disk space that is used.<br>View the total amount of disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes. |
| View the backup disk space | View the chart that shows the percentage of backup disk space that is used.<br>View the total amount of backup disk space, the amount of free disk space, and the amount of used disk space, shown in terabytes. |
| Change the backup directory | Change the backup directory.<br>For more information, see “Add or change the backup directory (administrator)” on page 173. |
| Change the email notification settings | Select when administrator-level users receive email notifications.<br>For more information, see “Change email notification settings (administrator)” on page 182. |
| Change the backup settings | Backup .wells files and intermediate files.<br>For more information, see “Change the backup settings (administrator)” on page 177. |
| Change the disk cleanup settings | Set the schedule to permanently delete analysis results files.<br>Set the conditions to permanently delete other files.<br>For more information, see “Set a schedule to permanently delete files (administrator)” on page 179. |
| Enable backup and cleanup | Click the Backup & Cleanup Enabled toggle switch to the enabled position to start the backup and cleanup process at the scheduled time. |
| Disable backup and cleanup | Click the Backup & Cleanup Enabled toggle switch to the disabled or off position for temporary instances, such as when the backup server is down.<br>This setting overrides all other backup settings. |

*(continued)*

| Option | Description |
|---|---|
| Start backup | Click Start Now to start:<br>• the backup of system data and analysis results to the selected backup directory.<br>• if enabled, the backup of sequencing output and intermediate files to the selected backup directory.<br>• data clean up based on cleanup settings.<br>Backup can take a considerable amount of time when the backup is set up for first time on the system. |

3. Click **Save**.

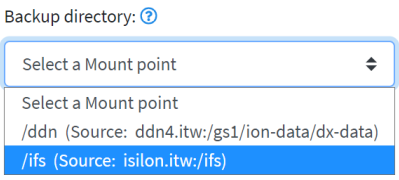
## Add or change the backup directory (administrator)

Administrator-level users can use a Network File System (NFS) to set up an external storage device that is a backup server to store system data. When the current backup directory has limited free disk space remaining, change the backup directory used by the system.

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, select either **Network** or **Cloud** as the connection type.

3. Edit the backup settings.

| Option | Description |
|---|---|
| Add a new mount point | a. In the **Backup Disk Space** section, click **Add Mount**.<br>b. In the **Add Mount** dialog box:<br>• Enter the name of the mount directory. For example, *ddn*.<br>Note: Do not include information from a hostname in the mount directory name. For example, if the host name is *gnx001*, do not use *gnx001* in the mount directory name.<br>You can type a new name to create a new directory.<br>• Enter the storage source. For example, *ddn.itw:/gs1/ion-data/dx-data*<br>c. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is restarted.<br>When the option is unselected, you must mount a directory after a restart. |
| Change the existing backup directory | In the **Backup Disk Space** section, select the directory that you want to use for backups from the **Backup Directory** dropdown menu.<br>You must add a mount point before it can be selected in the dropdown.<br>  |
| Remove a backup directory that is not in use, for example a directory that is full or is no longer available | a. Click **Remove Mount**.<br>b. In the **Remove Mount** dialog box, select the mounted drive you want to remove.<br>c. Click **Remove**.<br>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory. |

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

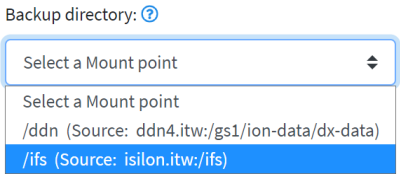
## Add or change the cloud backup directory (administrator)

You can change the cloud backup directory that is used by the system. For example, you might change the directory if the current backup directory has limited storage space. You can also add a new directory, or remove a directory.

1. In the menu bar, click **(Settings)►Backup & Restore**.
   The **Backup & Restore** screen opens.

2. Under **Backup Disk Space** select **Cloud** as the connection type.

3. Edit the backup settings.

| Option | Description |
|---|---|
| Add a new mount point | **1.** In the Backup Disk Space section, click Add Mount.<br>**2.** In the Add Mount dialog box:<br>• Enter the name of the Mount Point directory. For example, *ddn*.<br>You can type a new name to create a new directory.<br>• Enter the Cloud Provider. For example, *AWS-S3*.<br>**3.** Enter the Configuration information.<br>• Nick Name—A name to uniquely identify the directory and mount point on the directory. Do not use spaces or special characters in the name.<br>Note: The following information must be entered correctly to ensure that the mount works correctly. Information should be provided by an administrator for the network or Amazon™ Web Services (AWS).<br>• Bucket Name—The name of the AWS S3 bucket.<br>• Access Key Id—A key identifier that, similar to a username, provides programmatic and CLI access to AWS resource, such as the S3 bucket<br>• Secret Access Key—An access key that, similar to a password, provides programmatic and CLI access to AWS resource, such as the S3 bucket.<br>• Region—The AWS region in which for the S3 bucket resides. For example, *us-east-1*, *us-west-1*.<br>**4.** *(Optional)* Select Automount to allow the system to mount backup directories automatically when the system is rebooted.<br>When the option is unselected, you must mount a directory after a reboot.<br>**5.** Click Save. |
| Change the existing backup directory | In the Backup Disk Space section, select the directory that will be used for backups from the Backup directory drop-down list.<br>You must add a mount point before for the backup directory can be selected.<br>  |
| Remove a backup directory that is not in use, for example a directory that is full or is no longer available | **1.** Click Remove Mount.<br>**2.** In the Remove Mount dialog box, select the mounted drive you want to remove.<br>**3.** Click Remove.<br>**4.** Click Save.<br>If you remove a directory that is in use for backup, backups cannot not be performed until you select a new backup directory. |

4. In the **Backup & Restore** screen, click **Save** to save the new selections.

The changes that you made are shown in the **Backup Disk Space** section of the **Backup Settings** screen.

## Set up a backup server (administrator)

Administrator-level users can set up an external storage device to be a backup server to store system data with this procedure. Alternatively, administrator-level users can directly set up a backup server in Genexus™ Dx Software. For more information, see “Add or change the backup directory (administrator)” on page 173.

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IMPORTANT! We recommend Network File System (NFS) version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

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1. Mount the NFS to the sequencer.

   The following example is for users who have an existing NFS with read and write access. Mount the NFS to the sequencer using the *fstab* file so that the mount point is persistent (NFS mounts automatically after rebooting).

   **Example:**
   For NFS share: `cqtsop4:/md1200`
   Mount point: `nfs1`

2. Create a mount point to mount the external device on nfs share.
   a. Create the directory /nfs1 using the following command:

      ```
      sudo mkdir /nfs1
      ```

   b. Append the mount point entry in /etc/fstab.

      Command example:

      ```
      sudo vi /etc/fstab
      cqtsop4:/md1200 /nfs1 nfs defaults 0 0
      ```

   c. Save, then exit by pressing **Esc** on your keyboard, then type *:wq*

3. Create a permanent or temporary mount on nfs share:
   - Permanent mount command: `sudo mount -a`
   - Temporary mount command (example): `sudo mount cqtsop4:/md1200 /nfs1`

4. Verify that the NFS share is mounted using the df -kh command.

   ```
   ionadmin@eternity1:~$ df -kh
   Filesystem Size Used Avail Use% Mounted on
   /dev/sda9 253G 5.9G 235G 3% /
   none 4.0K 0 4.0K 0% /sys/fs/cgroup
   udev 63G 4.0K 63G 1% /dev
   tmpfs 13G 1.3M 13G 1% /run
   none 5.0M 0 5.0M 0% /run/lock
   none 63G 0 63G 0% /run/shm
   none 100M 0 100M 0% /run/user
   ```

```
/dev/sda5 19G 2.9G 15G 17% /var
/dev/sda1 938M 37M 854M 5% /boot
/dev/sda6 9.1G 1.7G 7.0G 20% /home
/dev/mapper/raid-serverdata 8.4T 4.3T 4.1T 52% /serverdata
/dev/mapper/raid-servershare 1.0T 9.0G 1015G 1% /servershare
/dev/mapper/raid-scratch 1.0T 838M 1023G 1% /scratch
/dev/sda7 92G 61M 87G 1% /tmp
pluto:/md1200 37T 1.1T 36T 3% /nfs2
penguin-ts1:/local 11T 9.3T 978G 91% /nfs
ddn4.itw:/gs1/ion-data/dx-data 1.1P 1001T 115T 90% /ddn
cqtsop4:/md1200 37T 37T 77G 100% /nfs1
```

The mounted directory is listed in the **Backup Directory** dropdown list in the **Backup & Restore** screen.

For more information, see **help.ubuntu.com/lts/serverguide/network-file-system-configuration**.

## External storage devices

IMPORTANT! We recommend NFS version 3 (NFSv3) or NFS version 4 (NFSv4) with Linux Operating System external storage devices for Genexus™ Dx Software.

## Change the backup settings (administrator)

A backup directory must be set up before an administrator-level user can change the backup settings. For more information, see “Disk space usage and backup settings (administrator)” on page 169.

The following types of data are backed up daily at midnight and cannot be changed:

- **System Data**–Includes the system database, reference files, assay definition files, annotation content, and system-installed panel files, hotspot files, fusion reference files, and CNV baseline files.
- **Analysis Results**–The final results generated after sequencing and analysis. Analysis results include run details, all BAM files, filtered results, graphs, run reports, metrics, annotated variant data, and VCF files.

IMPORTANT! The first time you back up the system, it takes a considerable amount of time. The amount of time required for system backup depends on the number of runs that require backup and the amount of disk space used. For more information, see “Specifications and disk space usage” on page 171.

A system restart also takes time. When the system restarts, it resumes the backup where it left off.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, in the **Backup Settings** tab, edit the backup settings.

| Option | Procedure |
|---|---|
| Back up .wells files | Select the Sequencing Output checkbox.<br>Sequencing output backup takes time. |
| Back up intermediate files, such as intermediate BAM files and log files | Select the Intermediate Files checkbox. |
| Change the frequency or timing of the .wells and intermediate files backup | Select Daily or Weekly, then select the time and day, if applicable, for the backup.<br>Intermediate file backup takes time. Consider scheduling the backup of intermediate file on a weekend or at a time when the system is not in use. |

3. Click **Save**.

## View and export the audit trail for backup settings (administrator)

An administrator-level user can view the audit trail for backup settings, disk cleanup settings, and email notification settings in Genexus™ Dx Software.

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, view the audit trail.

| Option | Procedure |
|---|---|
| View the audit trail for the backup disk space | In the Backup Disk Space section, click audit to review changes made to the backup directory. |
| View the audit trail for the backup settings | In the Backup Settings section, click audit to review changes made to the backup settings. |
| View the audit trail for email notification settings | In the Email Notification Settings section, click audit to review changes made to the email notification settings. |
| View the audit trail for disk cleanup settings | In the Disk Cleanup Settings section, click audit to review changes made to the disk cleanup settings. |

3. In the **Audit Trail - Archive Settings** screen:

| Option | Procedure |
|---|---|
| Export the record to a PDF file | Click Download. |
| Return to the Backup Settings tab. | Click Back. |

## Set a schedule to permanently delete files (administrator)

Results files and `.wells` files, and intermediate files that have been backed up can be scheduled to be deleted by an administrator-level user in Genexus™ Dx Software.

Intermediate and .wells files include intermediate BAM files, log files, and Sequencing output files. These files are not backed up by default. When you set a schedule to delete these files, by default, the files are deleted when they are not backed up and the system disk space falls below 4 terabytes (TB). You can change the system disk space threshold to delete these files.

Run results that are retained are never deleted in the software, even when you set a schedule to permanently delete run results in the software. For more information, see “Retain run results” on page 116.

---

IMPORTANT!

- By default, when the **Disk Cleanup Settings** are enabled, the software is set to delete results 12 months after the run was last updated and backed up. You can change this setting.
- Intermediate and .wells files are not backed up unless an administrator-level user sets a schedule to back up these files. By default, these files are auto-deleted when the free disk space threshold is met, even when these files are not backed up. If you select to keep the default auto-delete setting for intermediate and .wells files and the files are deleted, the impact is minimal.

  You cannot perform the following actions when these files are deleted.

  - Reanalyze a run from basecalling.

---

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, in the **Disk Cleanup Settings** section:

| Option | Procedure |
|---|---|
| Do not auto-delete analysis results at any time. | In Delete Analysis Results, click No. |
| Change the schedule to permanently delete analysis results. | Analysis results are deleted only if the results have been backed up.<br>**1.** In Delete Analysis Results, click Yes.<br>**2.** In months after completion, set the schedule to delete the files:<br>• 3 months<br>• 6 months<br>• 9 months<br>• 12 months<br>• 24 months – default setting<br>• 36 months |
| Do not auto-delete .wells files and intermediate files at any time. | In Delete additional files, click No. |
| Change the schedule to permanently delete .wells files and intermediate files. | In Delete additional files, click Yes, then select the threshold of free disk space on the instrument, from 1 to 10 TB, that triggers the system to delete these files.<br>The system automatically deletes .wells and intermediate files when the disk space falls below the selected threshold.<br>If you have enabled the backup of intermediate files and .wells files, these additional files must be backed up before they can be deleted. |

3. Click **Save**.

## Archive a run result (administrator)

In addition to system cleanup, in which all backed up runs that are older than a specified length of time are archived, an administrator-level user can archive one to five run results at any time. Archived run results are copied to the external storage device or to a cloud system that you have set up. Then the archived run results are removed from the sequencer.

When you archive a run result, the analysis results files are archived. In addition, you can archive the intermediate and 1.wells files, if needed. You can archive run results that are no longer needed to ensure that there is sufficient disk space on the sequencer.

If you archive a run result at the time of a scheduled system archive, the system cleanup is skipped for that instance.

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, place the pointer over one or more run result that you want to archive, then click **Archive Data**.
3. In the **Confirm archive data for selected result(s)** dialog box, select **Sequencing Output** and **Intermediate files** to include 1.wells and intermediate files, if needed.
4. Click **Archive**.

Archived runs results are listed in the **Run Results** screen with the archived symbol ( ) after the run name. An administrator-level user can restore the run result to access the data.

## Delete a run result (administrator)

An administrator-level user can delete a run result in Genexus™ Dx Software. When you delete a run result, files for the run result are deleted from the sequencer.

---

IMPORTANT! This action cannot be reversed if you proceed. You can delete a run result even if it is not backed up. Ensure that you do not need the run result before you delete the run result from the sequencer and the software if the run result has not been backed up.

---

1. In the menu bar, click **Results►Run Results**.
2. In the **Run Results** screen, select the checkbox in the row of one or more run results that you want to delete.
3. In the **Confirm delete data for selected result(s)** dialog box, confirm that the data are backed up, if needed, and select the files to delete.
   - **Sequencing Output**
   - **Intermediate Files**
   - **Analysis Results**

   If you delete the analysis results files, the sequencing output and intermediate files are also deleted.
4. Click **Delete**.
5. Click **Yes** to confirm the deletion.

The files for the run result that you select are deleted from the sequencer and are no longer available in the software. If the run results are backed up before the deletion, an administrator-level user can restore the run result to access the data.

You can view the audit trail for a run result that you have deleted in the **Settings / Audit Records** screen. Deleted run results are listed as **Planned Runs**. For more information, see “Search audit records (administrator)” on page 160.

## Data output directory

The data output directory is the primary location on the hard drive where data generated by the Genexus™ Dx Integrated Sequencer is stored.

The directory location is locked and cannot be changed. Users should configure their laboratory information management systems (LIMS) to access this folder location to receive sequencing output files.

After a sequencing run and data analysis are complete, the software creates a separate results folder in the data output directory for each sample, and uses the following naming conventions:

```
AssayDev_<Sample Name>_<Assay Name>_<RunPlanShortCode>_<RunPlanShortCode>
```

## Email notifications

Administrator-level users receive email notifications when there is a restore run failure. Administrator-level users can also receive email notifications when other conditions are met, such as when the system has limited free disk space remaining. To review or update these settings, see “Change email notification settings (administrator)” on page 182.

Before administrator-level users can receive email notifications, an administrator or manager must first set up notifications for valid Genexus™ Dx Software user accounts. For more information, see “Configure email notifications (manager/administrator)” on page 165.

### Change email notification settings (administrator)

Administrator-level users can select when to receive email notifications, such as when the system has limited disk space remaining. Administrator-level users receive an email message when conditions occur according to the selections in the email notification settings.

1. In the menu bar, click **(Settings)►Backup & Restore**
2. In the **Backup & Restore** screen, in the **Email Notifications Settings** section, select or deselect the checkbox next to one or more of the following options. Administrator-level users receive email notifications for options that are selected. They do not receive email messages for options that are deselected.
   - Backup fails or has insufficient storage space.
   - Backup is successfully completed.
   - Genexus™ Dx Integrated Sequencer has insufficient system disk space. Select the amount of sequencer disk space that must be available in the dropdown list.
   - Backup fails or has insufficient disk space. Select the amount of backup disk space that must be available in the dropdown list.
3. Click **Save**.

Changes to the email notifications take effect immediately.

# Restore run results (administrator)

An administrator-level user can configure Genexus™ Dx Software to automatically back up older run data, results files, and signed reports to external data storage or a cloud storage space. For more information, see “Disk space usage and backup settings (administrator)” on page 169 and “Add or change the backup directory (administrator)” on page 173. In addition, an administrator-level user can set a schedule to automatically delete backed up runs after a specified amount of time. For more information, see “Set a schedule to permanently delete files (administrator)” on page 179. Runs, for which analysis results have been backed up and deleted in the software, are listed in the **Run Results** screen and are denoted with the archived symbol ( ) after the run name.

An administrator-level user can later restore the run results, files, and reports in the software. There are two ways to restore run results in Genexus™ Dx Software.

- For information about how to restore run results for a single run, see “Restore run results for a single run (administrator)” on page 183.
- For information about how to restore or import multiple run results, see “Restore or import multiple archived run results (administrator)” on page 183.

## Restore run results for a single run (administrator)

An administrator-level user can restore run results, files, and reports that have been archived in Genexus™ Dx Software.

Archived runs that can be restored are listed in the **Results / Run Results** screen with the **Restore** link active when you place the pointer over the row of run. Runs, for which analysis results have been backed up and deleted (archived) from the system, are listed in the **Run Results** screen and are denoted with the archived symbol ( ) after the run name.

1. In the menu bar, click **Results**►**Run Results**.
2. In the **Run Results** screen, click the run name of interest.
3. In •••**(More Options)**, click **Restore**.
4. Click **OK** in the confirmation dialog box.
5. Click the run name in the **Run Results** screen to view run results.

## Restore or import multiple archived run results (administrator)

An administrator-level user can:

- Restore run results, files, and reports that have been archived.
- Import run results, files and reports from external data storage or another Genexus™ Dx Integrated Sequencer.

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Note: Do not attempt to import verification runs because they are specific to a single system. **Sample to Result** run details for the Genexus™ Dx Purification System are not displayed in the target system (the system results are imported to) if the target system does not have a connected purification instrument.
After a run is imported, you can access **Run Results** and **Sample Results** in the target system. Genexus™ Integrated Sequencer details are also listed with the instruments in the target system.

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Results that have been backed up, then deleted in the software, are listed in the **Results / Run Results** screen with (archived) after the run name.

Before you can import a run from external data storage or another Genexus™ Dx Integrated Sequencer:

- The run data must be archived to external data storage, and the same external data storage must be connected to the Genexus™ Dx Integrated Sequencer to which you will import the run.
- The samples for the run or runs that you want to import must have been created in the software with identical names. For more information about how to import a sample from another system, see “Import samples” on page 29.
- The assay for the run or runs that you want to import must exist in the software. The name of the assay, the barcode kit, and the extraction method type, must be identical between the source system and the target system. For information about how to import an assay, see “Import an assay (manager/administrator)” on page 151.

You can search the list of runs that can be restored by the run name or a sample tag name. However, if you restore a run that has an assigned sample tag name that has been deleted in the software, the tag is not shown in the software when you restore the run. In this case, you can reassign the tag if needed. For more information, see “Sample tags” on page 33.

1. In the menu bar, click **(Settings)►Backup & Restore**.
2. In the **Backup & Restore** screen, click the **Restore Runs** tab and select from the following options.

| Option | Description |
|---|---|
| Restore archived run results | **1.** In the type of runs dropdown list, keep the selection of Archived runs.<br>**2.** In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>**3.** In the Select backup folder to list runs dialog box, navigate to the folder with the name of the system that you use, then select the subfolder that contains the runs you want to restore and click Select.<br>**4.** In the Restore Runs tab, select the checkbox in the row for each run that you want to restore, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>**5.** *(Optional)* After the archived run is restored, click Check Log to view if the process was successful or if there are problems. |

*(continued)*

| Option | Description |
|---|---|
| Import run results from another system | 1. In the type of runs dropdown list, keep the selection of Archived runs.<br>2. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>3. In the Select backup folder to list runs dialog box, navigate to the folder for a different system, then click Select.<br>4. In the Restore Runs tab, select the checkbox in the row for each run that you want to import, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>5. *(Optional)* After the archived run is imported, click Check Log to view if the process was successful or if there are any problems, such as if a sample has not been created in the software. |
| Restore runs if there is a system crash | Use this procedure only if there is a system crash and you have worked with a Field Service Engineer (FSE) to restore the data onto a new system. When you select All runs in any other instance, the software overwrites existing data with identical data.<br>1. In the type of runs dropdown list, select All runs.<br>2. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click Browse.<br>3. In the Select backup folder to list runs dialog box, navigate to the folder that contains the runs that you want to restore, then click Select.<br>4. In the Restore Runs tab, select the checkbox in the row for each run that you want to restore, then click Restore Runs.<br>Use the Search By dropdown list and the search field to search for samples by a run name or by sample tags.<br>The process to restore runs takes time. After the archived run is restored, you can view the run results for the run in the software.<br>5. *(Optional)* After the archived run is restored, click Check Log to view if the process was successful or if there are problems. |

# Log files (administrator)

Administrator-level users use the **Logs** screen to view log files generated by the software and connected purification and sequencing instruments. Viewing log files is useful for troubleshooting activities.

Administrator-level users can also use the **Logs** screen to manage the storage of log data by setting the retention period for the audit trail function and enabling auto-deletion rules for logs.

## View log files (administrator)

Administrator-level users can view software and instrument log files in Genexus™ Dx Software.

1. Click **(Settings)►Logs**.
2. In the **Logs** screen, select an option in the software or connected instrument dropdown menu.

| Option | Description |
|---|---|
| View software log files. | Click **Ion Torrent Genexus Software**. |
| View log files for a connected purification or sequencing instrument. | Click **Instrument**. |

3. Click the log file name of interest.

| File name | Description |
|---|---|
| `ApplicationLog_<date>.log` | A log file with information related to some actions in the Genexus™ Dx Software, including user sign-ins and sign-outs and failed attempts to sign-in or sign-out. |
| `InstrumentGX5_<date>.log` | A log of activity on the sequencer instrument. |
| `InstrumentPurif_<date>.log` | A log of activity on the purification instrument. |

The log file opens in a new tab in the browser.

## Manage log settings (administrator)

An administrator-level user manages the length of time that log files are kept and configures the software to allow log files to be automatically deleted.

This feature allows you to meet Title 21 CFR Part 11 of Federal Regulations that establishes the United States Food and Drug Administration regulations on electronic records and signatures, password policies, and user activity auditing.

1. Click **(Settings)►Logs**.
2. In the **Logs** screen, click **Manage Logs**.
3. In the **Manage Logs** dialog box, in **Retention Period**, select the number of months that logs are to be retained on the sequencer.
4. Select **Enable Auto Deletion** to delete log files automatically after the designated retention period.
5. Click **Save**.

## View the log settings history

Administrator-level users can view the history of changes made to the settings for the retention of log files in Genexus™ Dx Software.

1. Click **(Settings)►Logs**.
2. In the **Logs** screen, click **Audit Trail**.
   The **Audit Trail - Log Settings** screen opens.
3. Click **Back** to return to the list of log files.

# View panel details

You can view details about system-installed and panels that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. View the panel details.

# View hotspot file details

You can view details about system-installed hotspot files and hotspot files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Hotspots** tab.
3. In the **Hotspots** table, view the details for the hotspot files, including the panel with which the the file is associated, whether the file is **Locked**, if the file is **Active**, who created the file, and the date on which the file was created. You can also view the genomic coordinates and alleles that are included in the hotspot file.

# View fusion reference file details

You can view details for system-installed fusion reference files and fusion reference files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Fusion References** tab.
3. In the **Fusion References** table, view the details for the fusion reference files.

# View reference genome file details

You can view details for system-installed reference genome files and reference genome files that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **References** tab.
3. In the **References** table, view the details for the reference genome files.

# View barcode set details (manager/administrator)

You can view details for system-installed barcode sets and barcode sets that have been added in Genexus™ Dx Software.

1. In the menu bar, click **Assays►Manage Panels**.
   The **Panels** tab in the **Manage Panels** screen opens.
2. Click the **Barcode Sets** tab.
3. In the **Barcode sets** table, view the list of barcode sets.
4. Click the name of a barcode set in the list to view the barcode IDs, sequences, and other details for the barcode set.
5. Click ✕ **(Remove)** to return to the complete list of barcode sets.

# Control fragments (manager/administrator)

Control fragment sequences are system-installed in Genexus™ Dx Software and are included in reagents. These internal controls are predefined and cannot be modified by users.

Manager- and administrator-level users can view control fragment information in the **Assays / Manage Panels** screen, in the **Control Fragments** tab.

| Column | Description |
|---|---|
| Name | The control fragment name. |
| Sequence | Single-letter nucleotide sequence of the control fragment. |
| Control | The process in which the control fragment is used. |
| Status | Indicates whether the control fragment information is locked and cannot be modified by the user. |

# System services (manager/administrator)

Manager- and administrator-level users can view information about system services using the **Settings / Services** screen. The information can be useful when troubleshooting error messages that are received during an active job or why a run failed.

| Service | Function |
|---|---|
| Jobs Server | Shows a list of server information and active (running) software. |
| Services | Shows the service name, description, and status. The Services list can be used for troubleshooting. If a service has stopped during a run, an option to start the service will be visible for manager- and administrator-level users.<br>For more information, see “Start a stopped service (manager/administrator)” on page 190. |
| Active Jobs | • Shows a list of runs in progress that are active and queued on the sequencer.<br>• The message “No records available” indicates that no jobs are currently active.<br>• Allows manager- and administrator-level users to stop a job.<br>For more information, see “Terminate an active job (manager/administrator)” on page 190. |
| ionCrawler Service Details | Provides information about the data transfer process from the Genexus™ Dx Integrated Sequencer. The information includes whether the service was started or stopped, which user completed the action, and a timestamp for the action. |
| Smart Monitoring Service Details | Allows remote monitoring of the Genexus™ Dx Integrated Sequencer. Allows manager- and administrator-level users to start or stop the service. For more information, see “Start a stopped service (manager/ administrator)” on page 190. |

*(continued)*

| Service | Function |
|---|---|
| Download Instrument Diagnostics | Contains the initialization logs of the Genexus™ Dx Integrated Sequencer, and allows you to download the diagnostic archive file in ZIP format. |
| System Check Report | Manager and administrator-level users can click Run System Check to view the system check report. |

## Terminate an active job (manager/ administrator)

If an active job is not going to be useful or if a different job is urgent, you can terminate an active job. A manager- or administrator-level user can stop an active job in the **Settings / Services** screen.

1. In the menu bar, click ⚙ **(Settings)►Services**.
2. In the **Services** screen, in the **Active Jobs** section, find the active job that you want to stop, then click **Terminate**.
   A confirmation message opens, **Are you sure you want to terminate `<job name>` ?**.
3. Click **Terminate** to terminate the job.

## Start a stopped service (manager/ administrator)

A manager- or administrator-level user can use the **Settings / Services** screen to restart a stopped service if a service has stopped running for some reason. The two database services, **ntp** and **deeplaser**, are the only services that can be restarted.

1. In the menu bar, click ⚙ **(Settings)►Services**.
2. In the **Services** screen, in the **Services** section, find the service that you want to restart, then click **Start**.
   A confirmation message opens, **Are you sure you want to Start `<service name>`?**.
3. Click **Yes** to restart the service.

## Disable or enable the Smart Monitoring service (manager/ administrator)

The Smart Monitoring service allows Thermo Fisher Scientific personnel to monitor the status of the Genexus™ Dx Integrated Sequencer remotely through an internet connection. Smart Monitoring employs multiple layers of security, including a Secure Sockets Layer (SSL) and Lightweight Directory Access Protocol (LDAP) authentication, to provide real-time troubleshooting and problem resolution for the Genexus™ Dx Integrated Sequencer.

The Smart Monitoring service is active by default and can be disabled by manager- or administrator-level users, if required.

- Disable the service.
  a. In the menu bar, click ⚙ **(Settings)►Services**.

b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Stop**.

c. In the confirmation message, click **Yes** to disable the service.
The **Stop** option changes to **Start**.

- Enable the service.
  a. In the menu bar, click **(Settings)►Services**.

  b. In the **Services** screen, in the **Smart Monitoring Service Details** section, click **Start**.

  c. In the confirmation message, click **Yes** to enable the service.
  The **Start** option changes to **Stop**.

# Sequencer decommissioning and data sanitization

Contact a Thermo Fisher Scientific Field Service Engineer for any request to decommission a Genexus™ Dx Integrated Sequencer. Decomissioning and data sanitization must be carried out by a Field Service Engineer.

# 10 Customer Support Archive

The Customer Support Archive is a downloadable archive that a technical support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Dx Software. The archive contains log files and other technical data about the software and other files from sequencing runs on a Genexus™ Dx Integrated Sequencer.

For more information, see “Download a Customer Support Archive” on page 225.

# A Genexus™ Dx Software web services API

Genexus™ Dx Software has a web services API that allows you to automate the retrieval of key information from the system.

This section provides examples to show you how to leverage the API.

## LIMS introduction

Genexus™ Dx Software integrates with laboratory information management systems (LIMS) to help with sample, library batch, and run plan creation. The interaction is through the LIMS calling a set of RESTful APIs provided by the software (see the following table). The LIMS connects to the software by making an HTTP call to the URL of an API, and sending a user name and a password in the HTTP header. The user name and password must be for a registered active user account of the software. The LIMS passes the input information in JSON format within the HTTP body, while the response from the software is returned in JSON format as well. Users must configure their LIMS to create JSON files containing the required attributes for each LIMS request file.

Outlined in the following table are the API names, functions, and URLs of the five RESTful APIs that are provided.

| API | Function | API URL |
|---|---|---|
| Get report templates<br>For more information, see “Get report templates” on page 205. | Returns report templates. | `https://<host>/genexus/api/lims/v2/report-templates` |
| Get a list of assays<br>For more information, see “Get assay list” on page 196. | Returns a list of the assays in the system. PQ assays are excluded. | `https://<host>/genexus/api/lims/v2/assays` |
| Get a list of samples<br>For more information, see “Get samples” on page 197. | Returns a list of the samples in the system. | `https://<host>/genexus/api/lims/v2/samples` |
| Get a list of library batches<br>For more information, see “Get library batches” on page 198. | Returns a list of the library batches in the system. | `https://<host>/genexus/api/lims/v2/library-batches` |

*(continued)*

| API | Function | API URL |
|---|---|---|
| Get a list of run plans<br>For more information, see "Get run plans" on page 201. | Returns a list of available run plans in the system. | `https://<host>/genexus/api/lims/v2/plans` |
| Get results<br>For more information, see "Get results" on page 206. | Returns results of the run and samples in JSON file format. | `https://<host>/genexus/api/lims/v2/results` |
| Get a list of signed off samples<br>For more information, see "Get signed off samples" on page 211. | Returns a list of the samples that are designated with *(Signed Off)*. | `https://<host>/genexus/api/lims/v2/signedOffSamples` |
| Download files for completed runs<br>For more information, see "Download files for completed runs" on page 212. | Returns a compressed file in ZIP format that contains the requested files for the sample. | `https://<host>/genexus/api/lims/v2/download` |
| Create samples<br>For more information, see "Create samples" on page 212. | Create samples. | `https://<host>/genexus/api/lims/v2/samples` |
| Create a library batch<br>For more information, see "Create a library batch" on page 214. | Create a library batch from existing samples. | `https://<host>/genexus/api/lims/v2/library-batches` |
| Create a Sample to Result (purification) run<br>For more information, see "Plan a Sample to Result run" on page 53. | Create a Sample to Result run from one or more samples and an assay. | `https://<host>/genexus/api/lims/v2/plan-by-purification` |
| Create a Nucleic Acid to Result run<br>For more information, see "Create a Nucleic Acid to Result run" on page 215. | Create a Nucleic Acid to Result run from one or more samples and an assay. | `https://<host>/genexus/api/lims/v2/plan-by-samples` |
| Create a library run<br>For more information, see "Create a library run" on page 216. | Create a library run from one or more library batches and an assay. | `https://<host>/genexus/api/lims/v2/plan-by-library` |

## LIMS system FTP setup or drive mapping

To enable the LIMS system to retrieve data analysis files from the sequencer, a manual FTP setup or drive mapping is required. This configuration is not provided as part of the sequencer installation, and must be set up as part of the LIMS system by the system integration or laboratory IT group. See the sections in this appendix for file paths and other setup information.

# API common parameters

If the JSON download process converts `@` symbols to `%40`, do not change the text. Instead, copy the JSON files as is into the browser.

| Parameter | Entry |
|---|---|
| BASE_URL | `https://<host>/genexus/api/lims/v2` |
| Request | `Authorization:Basic`<br>`Accept`:application/json |
| Response | `Content-Type`:application/json |

Response fields

| Code | Status |
|---|---|
| 200 | Successful response |
| 400 | Bad request<br>Error response:<br>`{ "errors":[ { "field":"validation error" } ] }` |
| 401 | Unauthorized |
| 500 | Internal Server Error |

Get requests

| Type of request | Entry |
|---|---|
| Get list of resources | `BASE_URL/resource_url/` |
| Get single object | `BASE_URL/resource_url?id=object_id` |

Common request parameters

| Parameter | Entry | Value |
|---|---|---|
| Starting page number | `pageNumber` | The default value is "1". |
| Number of items to fetch[1] | `pageSize` | The default value is "20". |
| A filed based on which, the result set needs to be ordered | `orderBy` | Only specific values are allowed. If no value is provided, the result set is ordered by ID. |
| Order direction of the result set | `sortOrder` | Valid values are "asc" or "desc".<br>The default value is "asc". |

[1] You can limit the number of items that are fetched when you add parameters to the request. For example, add "status" to the get assay list request.

# Get assay list

| assays | |
|---|---|
| URL | `BASE_URL/assays` |
| Description | Returns a list of the assays in the system. PQ assays are excluded. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/assays` |

*(continued)*

| assays | |
|---|---|
| Supported parameters | • `assayName`<br>String. *Optional*.<br>Assay name.<br>• `status`<br>String, *Optional*.<br>Valid values are "draft", "obsolete", or "locked". |
| Example response | {<br>"total": 1,<br>"objects": [<br>{<br>"id": 18,<br>"assayName": "Generic_DNA_and_Fusions_EU_v0.3.0",<br>"assayShortName": "Generic DNA-Fus EU v0.3.0",<br>"assayDisplayedName": "Generic - DNA and Fusions - EU v0.3.0",<br>"application": "DNA+RNA",<br>"createdBy": "System Installed",<br>"createdDate": "2021-07-14T00:05:12.000+0000",<br>"isReanalysisAssay": false,<br>"genomeReference": "hg19",<br>"targetRegions": "Ronin_w2.7.0_v0.3.0_dnafusion_FFPE_DNA_v1.20210604.designed.bed",<br>"hotspots": "Ronin_w2.7.0_dnafusion_FFPE_DNA.v1.20210608.hotspots_0.3.0.bed",<br>"fusionReference": "Ronin_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.fasta",<br>"fusionTargetRegions":<br>"Ronin_w2.7.0_dnafusion_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.bed",<br>"chipType": "Genexus™ Dx GX5™ Chip",<br>"panelKit": "Generic Assay Panel (IVD)",<br>"controlKit": "",<br>"extractionKit": "",<br>"libraryKit": "Genexus™ Dx Library Strips 1 and 2-HD (IVD)",<br>"templatingKit": "Genexus™ Dx Templating Strips 3-GX5™ and 4 (IVD)",<br>"barcodeKit": "Genexus™ Dx Barcodes 1-32 HD (IVD)",<br>"sequencingKit": "Genexus™ Dx Sequencing Kit (IVD)",<br>"flows": 500,<br>"templatingSize": 200,<br>"status": "Locked",<br>"tumorCellularity": "1",<br>"reanalysisAssay": false<br>}<br>]<br>} |

# Get samples

| samples | |
|---|---|
| URL | `BASE_URL/samples` |
| Description | Returns a list of the samples in the system. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/samples` |

*(continued)*

| samples | |
|---|---|
| Request query parameters | • `sampleId`<br>String. *Optional*.<br>Sample name. |
| Example response | {<br>"total": 1,<br>"objects": [<br>{<br>"otherAttributes": {<br>"Patient Id": "MRND685",<br>"Date of Birth": "2000-01-01",<br>"Ordering Physician": "Dr Fred"<br>},<br>"id": 163,<br>"sampleId": "Test_Sample1_224",<br>"collectionDate": "2018-10-02",<br>"gender": "Male",<br>"sampleType": "DNA",<br>"tumorCellularity": "70",<br>"origin": "Non-Small Cell Lung Cancer",<br>"cancerStage": "Stage 0",<br>"diseaseCategory": "Cancer",<br>"tags": [<br>"LIMS"<br>]<br>}<br>]<br>} |

# Get library batches

| library-batches | |
|---|---|
| URL | `BASE_URL/library-batches` |
| Description | Returns a list of the library batches in the system. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/library-batches` |

*(continued)*

| library-batches | |
|---|---|
| Request query parameters | • `id`<br>String. *Required*.<br>Library batch ID. |

*(continued)*

| library-batches | |
|---|---|
| Example response | {<br>"total": 1,<br>"objects": [<br>{<br>"libraries": [<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_05",<br>"libraryName": "NTC_DNAST_LibBatch_DNA_RNA_3",<br>"inputQuantity": 87.0,<br>"sampleId": "NTC",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165751724060110123456724 0A51339",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_06",<br>"libraryName": "NTC_RNAST_LibBatch_DNA_RNA_3",<br>"inputQuantity": 55.3,<br>"sampleId": "NTC",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165751724060110123456724 0A51339",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_03",<br>"libraryName": "PositiveControl01_DNAST_LibBatch_DNA_RNA_1",<br>"inputQuantity": 87.0,<br>"sampleId": "PositiveControl01",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165441725093010260745524 0A51337",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_04",<br>"libraryName": "PositiveControl01_RNAST_LibBatch_DNA_RNA_1",<br>"inputQuantity": 55.3,<br>"sampleId": "PositiveControl01",<br>"tumorCellularity": null,<br>"controlKitBarcode": "01101903020165441725093010260745524 0A51338",<br>"extractionKitBarcode": null<br>},<br>{<br>"libraryType": "DNA",<br>"barcodeID": "IonDxHDdual_01",<br>"libraryName": "LIMS_Sample_55_DNAST_LibBatch_DNA_RNA_2",<br>"inputQuantity": 57.0,<br>"sampleId": "LIMS_Sample_55",<br>"tumorCellularity": null,<br>"controlKitBarcode": null,<br>"extractionKitBarcode": "91100090559101234567172110 19"<br>},<br>{<br>"libraryType": "RNA",<br>"barcodeID": "IonDxHDdual_02",<br>"libraryName": "LIMS_Sample_55_RNAST_LibBatch_DNA_RNA_2",<br>"inputQuantity": 51.3,<br>"sampleId": "LIMS_Sample_55",<br>"tumorCellularity": null,<br>"controlKitBarcode": null,<br>"extractionKitBarcode": "91100090560101234567172112 31"<br>}<br>],<br>"id": 13,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",<br>"libraryBatchId": "ST_LibBatch_DNA_RNA",<br>"libraryKitBarcode": "",<br>"panelKitBarcode": "",<br>"libraryPrepType": "DNA+RNA",<br>"ampliSeqHd": false<br>}<br>]<br>} |

# Get run plans

| plans | |
|---|---|
| URL | `BASE_URL/plans` |
| Description | Returns a list of available run plans in the system. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/plans` |
| Request query parameters | • `planName`<br>String. *Optional*.<br>Name of the run plan.<br>• `viewType`<br>String. *Optional*.<br>Valid values are "all" or "toBeProcessed". |
| Example response | {<br>"objects": [<br>{<br>"id": 1,<br>"planName": "EK_Generic_FFPE_EU_Ronin_D5_0.3.0_6.5.26",<br>"planShortId": "EQ55D",<br>"type": "sample",<br>"createdBy": "System Installed",<br>"updatedOn": "2021-07-17T20:33:48.000+0000",<br>"noOfSamples": 8,<br>"noOfLibraries": 16,<br>"samples": [<br>{<br>"id": 8,<br>"sampleId": "NTC",<br>"assayName": "Generic - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_28"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_32"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 5,<br>"sampleId": "SRTestSample64",<br>"assayName": "Generic - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_25"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_29"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}, |

*(continued)*

| plans | |
|---|---|
| Example response, continued | {<br>"id": 6,<br>"sampleId": "SRTestSample65",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_26"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_30"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 2,<br>"sampleId": "SRTestSample61",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_18"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_22"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 7,<br>"sampleId": "SRTestSample66",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_27"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_31"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}, {<br>"id": 3,<br>"sampleId": "SRTestSample62",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_19"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_23"<br>],<br>"libraryBatchId": "1",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 4,<br>"sampleId": "SRTestSample63",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_20" |

*(continued)*

| plans | |
|---|---|
| Example response, continued | 
```
],
          "rnaBarcodes": [
            "IonDxHDdual_24"
          ],
          "libraryBatchId": "1",
          "libraryPrepType": "DNA+RNA",
          "pauseForReview": false
        }
      ],
      "runAssays": [
        "Generic Assay - DNA and Fusions - EU v0.3.0"
      ]
    },
    {
      "id": 9,
      "planName": "Re_Ali_EK_ODxET_FFPE_EU_Ronin_D5_0.3.0_6.5.26",
      "planShortId": "CBLG5",
      "type": "sample",
      "createdBy": "System Installed",
      "updatedOn": "2021-07-15T04:16:31.000+0000",
      "noOfSamples": 8,
      "noOfLibraries": 16,
      "samples": [
        {
          "id": 49,
          "sampleId": "PositiveControl01",
          "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
          "dnaBarcodes": [
            "IonDxHDdual_17"
          ],
          "rnaBarcodes": [
            "IonDxHDdual_21"
          ],
          "libraryBatchId": "9",
          "libraryPrepType": "DNA+RNA",
          "pauseForReview": false
        },
        {
          "id": 52,
          "sampleId": "SRTestSample63",
          "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
          "dnaBarcodes": [
            "IonDxHDdual_20"
          ],
          "rnaBarcodes": [
            "IonDxHDdual_24"
          ],
          "libraryBatchId": "9",
          "libraryPrepType": "DNA+RNA",
          "pauseForReview": false
        },      {
          "id": 55,
          "sampleId": "SRTestSample66",
          "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
          "dnaBarcodes": [
            "IonDxHDdual_27"
          ],
          "rnaBarcodes": [
            "IonDxHDdual_31"
          ],
          "libraryBatchId": "9",
          "libraryPrepType": "DNA+RNA",
          "pauseForReview": false
        },
}
```
 |

*(continued)*

| plans | |
|---|---|
| Example response, continued | {<br>"id": 50,<br>"sampleId": "SRTestSample61",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_18"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_22"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 53,<br>"sampleId": "SRTestSample64",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_25"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_29"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 54,<br>"sampleId": "SRTestSample65",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_26"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_30"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 51,<br>"sampleId": "SRTestSample62",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_19"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_23"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>},<br>{<br>"id": 56,<br>"sampleId": "NTC",<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"dnaBarcodes": [<br>"IonDxHDdual_28"<br>],<br>"rnaBarcodes": [<br>"IonDxHDdual_32"<br>],<br>"libraryBatchId": "9",<br>"libraryPrepType": "DNA+RNA",<br>"pauseForReview": false<br>}<br>],<br>"runAssays": [<br>"Generic Assay - DNA and Fusions - EU v0.3.0"<br>]<br>}<br>],<br>"total": 2<br>} |

# Get report templates

| report-templates | |
|---|---|
| URL for a list of resources | `BASE_URL/report-templates` |
| URL for a single object | `BASE_URL/report-templates` |
| Description | Returns report templates. API supports multiple filters. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/report-templates` |
| Request query parameters | `assayName`<br>String. *Optional*.<br>Get the report template for the given assay.<br>Use the Assay Name. Do not use the Assay Full Name. |
| Example response | {<br>"mode": "dx",<br>"reportingTemplate": [<br>{<br>"name": "LAB_ODxET_DNA_QC_6.8_v1.0.0",<br>"id": 22,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - DNA QC v1.0.0",<br>"assayId": 41,<br>"description": "ODxET DNA QC v1.0.0 - Lab Report",<br>"locale": "en"<br>},<br>{<br>"name": "ev_template",<br>"id": 21,<br>"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",<br>"assayId": 40,<br>"description": "EV_Template",<br>"locale": "en"<br>}<br>],<br>"total": 2<br>} |

# Get results

| results | |
|---|---|
| URL | `BASE_URL/results` |
| Description | Returns results of the run and samples in JSON file format. |
| Method | `GET` |
| Example URL | `https://<host>/genexus/api/lims/v2/results` |

*(continued)*

| results | |
|---|---|
| Request query parameters | • `id`<br>String. *Optional*.<br>Results for the selected result id.<br>Example: *123*<br>• `assay_id`<br>String. *Optional*.<br>Results for the assay id.<br>Example: *123*<br>• `plan_id`<br>String. *Optional*.<br>Results for selected run plan id.<br>Example: *123*<br>• `sampleId`<br>String. *Optional*.<br>Results for the selected sample name.<br>Example: *MySample*<br>• `planName`<br>String. *Optional*.<br>Results for selected run plan name.<br>Example: *MyPlan*<br>• `analysisComplete`<br>String. *Optional*.<br>Results for completed runs. Results included for failed and successfully completed runs.<br>Example: *true*<br>• `updatedBefore`<br>String. *Optional*.<br>Returns results updated before the selected date. Use date format: `yyyy-MM-dd HH:MM`.<br>Example: *2019-01-15 10:30*<br>• `updatedAfter`<br>String. *Optional*.<br>Returns results updated after the selected date. Use date format: `yyyy-MM-dd`<br>Example: *2019-01-15*<br>• `orderBy`<br>String. *Optional*.<br>Parameter to sort results by selected field. The default sort is by `updatedBefore` or `updatedAfter`. Results are listed in descending order, with the most recent results listed first. |

| Example response |
|---|
| Output is as follows. |

```
{
    "objects": [
        {
            "id": 328,
            "planName":  "EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-T2.0.0-2",
            "sampleId": "SRTestSample61",
            "assay": {
                "id": 18,
                "value": "Generic Assay - DNA and Fusions - EU v0.3.0",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "planRun": {
                "id": 59,
                "value":  "EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-T2.0.0-2",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "sample": {
                "id": 45,
                "value": "SRTestSample61",
                "accountType": null,
                "isGreaterVersion": false,
                "runType": null,
                "greaterVersion": false
            },
            "planShortCode": "YYRU2",
            "resultName":
"Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_328",
            "libraryNames": "SRTestSample61_RNA831, SRTestSample61_DNA831",
            "runStartDate": "2021-07-23T00:00-0400",
            "runCompleteDate": "2021-07-24T00:45-0400",
            "updatedDate": "2021-07-24T00:44-0400",
            "libraryprepId": 428,
            "analysisComplete": true,
            "analysisVersion": "6.5.26",
            "isThumbnail": false,
            "isArchived": false,
            "isReanalysis": false,
            "runStatus": {
                "id": null,
                "name": "Analysis",
                "value": "Completed"
            },
            "qcStatus": "Passed",
            "instrumentName": "ValkyrieTestRig",
            "outputsLink": "/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/outputs",
            "outputsDir": "/serverdata/dxdata/IR/data/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/outputs",
            "loadingMetrics": [
                {
```

```
                "name": "Total Addressable Wells",
                "count": 64847855
            },
            {
                "name": "Wells With ISPs",
                "count": 60145720,
                "percentage": 92.75
            },
            ...
        ],
        "filteringMetrics": [
            {
                "name": "Filtered: Polyclonal",
                "count": 22931697,
                "percentage": 38.27
            },
            {
                "name": "Filtered: Primer-Dimer",
                "count": 9955,
                "percentage": 0.02
            },
            ...
        ],
        "runQCMetrics": [
            {
                "data": [
                    {
                        "metricName": "Key Signal",
                        "value": "75",
                        "referenceRange": "Not Set",
                        "qCStatus": "Not Calculated",
                        "percentValue": false,
                        "iscosmic": false,
                        "isPQControl": false,
                        "uniqueName": "RunQC_KeySignal",
                        "isDisplayMetric": false,
                        "isDisplayQc": false,
                        "libraryType": "NA",
                        "dataType": "INT",
                        "pqcontrol": false,
                        "displayMetric": false,
                        "displayQc": false
                    },
                    ...
                ],
                "sequence": 0,
                "localizationKey": "RunQC",
                "inlineControlQc": false,
                "customApp": "",
                "images": []
            }
        ],
        "purificationQCMetrics": [...],
        "controlQCMetrics": [...],
        "sampleQCMetrics": [...],
        "sampleInlineControlQCMetrics": [],
        "ntcQCMetrics": [],
        "plugins": [
            {
                "results": {
                    "Library Type": "AmpliSeq HD - DNA and Fusions
```

```
(Single Library)",
                        "Min Mapping Quality": "0",
                        "Percent end-to-end reads": "91.62%",
                        "Use Only Non-duplicate Reads": "No",
                        "Sample Tracking": "No",
                        "barcodes": {
                            "IonDxHDdual_18": {
                                "Alignments":
"IonDxHDdual_18_EK_ODxET_FFPE_EU_D5_0.3.0_6.5.26-NA-
T2.0.0-2_Auto_R_2021_05_19_14_48_13_ion",
                                "Amplicon base composition bias": "1.141",
                                "Amplicons reading end-to-end": "98.80%",
                                "Amplicons with at least 1 read": "100.00%",
                        ...
                            },
                            ...
                        },
                        "Amplicons reading end-to-end": "86.40%",
                        "Min Aligned Length": "0",
                        "Targeted Regions": "Barcode specific",
                        "Tier 2 Coverage Depth": "350",
                        "Reference Genome": "Barcode specific",
                        "Use Only Uniquely Mapped Reads": "No",
                        "Target Padding": "0",
                        "Launch Mode": "Autostart with default
configuration",
                        "barcoded": "true",
                        "Tier 3 Coverage Depth": "500",
                        "Tier 1 Coverage Depth": "100"
                    },
                    "id": 455,
                    "pluginName": "coverageAnalysis",
                    "pluginVersion": "5.16.0.4",
                    "updatedOn": "2021-07-24T01:58:19.000+0000",
                    "status": "COMPLETED",
                    "path": "/serverdata/dxdata/IR/data/analysis_output/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59/
Auto_R_2021_05_19_14_48_13_ionservice_GNXS-0128-37-19MAY2021_AEG_59_202107232
15902373/ChipLane1_2/SRTestSample61_LibPrep428/plugins/
coverageAnalysis_5.16.0.4/1627091942777",
                    "runType": "SAMPLE"
                },
                ...
            ],
            "biomarkers": [
                {
                    "Variant": "ALK fusion,   AR fusion",
                    "level": "Level 3"
                }
            ]
        }
    ],
    "total": 1
}
```

# Get signed off samples

| signedOffSamples | |
|---|---|
| URL | `BASE_URL/signedOffSamples` |
| Description | Returns a list of the samples that are designated with *(Signed Off)*. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/signedOffSamples` |
| Request query parameters | • `start_date`<br>String. *Optional*.<br>Provide the start date as yyyy-mm-dd.<br>• `end_date`<br>String, *Optional*.<br>Provide the end date as yyyy-mm-dd.<br>• `signedOff`<br>String, *Required*<br>A Boolean data type. Valid values are "true" or a "false".<br>The default value is "true". |
| Example response | {<br>"meta": {<br>"pageSize": 20,<br>"next": null,<br>"pageNumber": 0,<br>"previous": null,<br>"total_count": 76<br>},<br>"objects": [<br>{<br>"sample": {<br>"assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0",<br>"base_url":<br>"AssayDev_PQSample1_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS",<br>"checksum_file": "CHECKSUM",<br>"created_date": "2021-06-23",<br>"sample_file": "Info.csv",<br>"sample_id": "92",<br>"sample_name": "PQSample1",<br>"planName": "IQ_Lib"<br>}<br>},<br>{<br>"sample": {<br>"assay_name": "qualification_ampliseq_genexus_pq_template516_4.0.0",<br>"base_url":<br>"AssayDev_PQSample2_qualification_ampliseq_genexus_pq_template516_4.0.0_A9GHS_A9GHS",<br>"checksum_file": "CHECKSUM",<br>"created_date": "2021-06-23",<br>"sample_file": "Info.csv",<br>"sample_id": "93",<br>"sample_name": "PQSample2",<br>"planName": "IQ_Lib"<br>}<br>}<br>]<br>} |

# Download files for completed runs

| download | |
|---|---|
| URL | `BASE_URL/download` |
| Description | Returns a compressed file in ZIP format that contains the requested files for the sample. |
| Method | `GET` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/download` |
| Request query parameters | • `file_list`<br>String. *Required*.<br>Provide the file name in comma separated format.<br>• `path`<br>String, *Required*.<br>Provide the LIMS folder path of the analyzed sample. You can request the folder path when you get signed off samples. The folder path is listed as the "base_URL". For more information, see “Get signed off samples” on page 211. |
| Example request | **1.** Perform a checksum function to query for the list of files for a sample that has been sequenced and analyzed.<br>`https://BASE_URL/download?file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q`<br>**2.** Parse the checksum to get the file name, then request that file.<br>`https://BASE_URL/download?file_list=Info.csv,BaseCaller.json,analysis.log&path=AssayDev_Sample_ C1_ qualification_ ampliseq_ Genexus_ pq_template516_1.3.2_3MI8Q_3MI8Q`<br>**3.** Ensure that the name of the downloaded file matches the checksum requested in step 1. |

# Create samples

| samples | |
|---|---|
| URL | `BASE_URL/samples` |
| Description | Create 1 to *n* samples. "All" or "None" is followed for all samples in a request. If the creation of any one sample fails, creation of the entire batch fails. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |

*(continued)*

| samples | |
|---|---|
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/samples` |
| Attribute definitions | • `Patient Id`. String. *Required*. A unique identifier for the patient.<br>• `Date of Birth`. String. *Required*. The date of birth for the patient.<br>• `Ordering Physician`. String. *Required*. The name of the physician who created the order for the sample.<br>• `sampleId`. String. *Required*. Sample name.<br>• `sampleType`. String. *Required*. Examples of valid values are "DNA" or "RNA".<br>• `collectionDate`. String. *Required*. The date that the sample was collected.<br>• `gender`. String. *Required*. The biological sex of the sample: Female, Male, or Unknown.<br>• `diseaseCategory`. String. *Required*. The sample application category.<br>• `cancerStage`. String. *Required*. Cancer stage.<br>• `origin`. String. *Required*. Cancer type.<br>• `notes`. String. *Required*. An open-entry field for more sample information.<br>• `necrosis`. String. *Optional*. The percentage of cellular necrosis in the sample.<br>• `tumorCellularity`. String. *Optional*. The percentage of tumor cells over normal cells in the sample.<br>• `condition`. String. *Optional*. The condition of the sample. |
| Example request | {<br>"objects":[<br>{<br>"sampleId":"Sample_NSCLC_99",<br>"diseaseCategory":"Cancer",<br>"sampleType":"DNA & RNA",<br>"origin":"Non-Small Cell Lung Cancer",<br>"cancerStage":"Unknown",<br>"necrosis":100,<br>"tumorCellularity":31,<br>"gender":"Unknown",<br>"collectionDate":"2018-10-02",<br>"condition":"good",<br>"tags":["NSCLC"],<br>"notes":"LIMS API created Cancer sample and existing Tag NSCLC",<br>"otherAttributes":<br>{<br>"Patient Id":"MRND12121",<br>"Ordering Physician":"Dr Fred",<br>"Date of Birth": "2000-01-01"<br>}<br>}<br>]<br>} |

# Create a library batch

| library-batches | |
|---|---|
| URL | `BASE_URL/library-batches` |
| Description | Create a library batch from one or more existing samples. If any requested sample does not exist in the software, the creation of the entire library batch fails. |
| Method | `POST` |
| Example URL | `https://<host>/genexus/api/lims/v2/library-batches` |
| Example request | 
```
{
  "objects":[
    {
    "libraryBatchId": "LibBatch_DNA_RNA_99",
    "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
    "libraryPrepType": "DNA+RNA",
    "libraryKitBarcode": "01ABC00123000000017221123102-321AB234240A51153",
    "panelKitBarcode": "01ABC00123000000017221123102-321AB234240A51345",
    "libraries": [
      {
        "barcodeID": "IonDxHDdual_01",
        "inputQuantity": 57.0,
        "libraryType": "DNA",
        "libraryName": "DNA_Lib1_01",
        "sampleId": "Test_Sample_01",
        "extractionKitBarcode": "A51000"
      },
      {
        "barcodeID": "IonDxHDdual_02",
        "inputQuantity": 51.3,
        "libraryType": "RNA",
        "libraryName": "RNA_Lib1_01",
        "sampleId": "Test_Sample_01",
        "extractionKitBarcode": "A51000"
      },
      {
        "barcodeID": "IonDxHDdual_03",
        "inputQuantity": 87.0,
        "libraryType": "DNA",
        "libraryName": "",
        "sampleId": "PositiveControl01",
        "controlKitBarcode": "A51337"
      },
      {
        "barcodeID": "IonDxHDdual_04",
        "inputQuantity": 55.3,
        "libraryType": "RNA",
        "libraryName": "",
        "sampleId": "PositiveControl01",
        "controlKitBarcode": "A51338"
      },
      {
        "barcodeID": "IonDxHDdual_05",
        "inputQuantity": 87.0,
        "libraryType": "DNA",
        "libraryName": "",
        "sampleId": "NTC",
        "controlKitBarcode": "A51339"
      },
      {
        "barcodeID": "IonDxHDdual_06",
        "inputQuantity": 55.3,
        "libraryType": "RNA",
        "libraryName": "",
        "sampleId": "NTC",
        "controlKitBarcode": "A51339"
      }
    ]
  }
]
}
```
 |

# Create a Sample to Result (purification) run

| plan-by-purification | |
|---|---|
| URL | `BASE_URL/plan-by-purification` |
| Description | Create a Sample to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/plan-by-purification` |
| Example request | <pre>{<br>"reportTemplateName": "",<br>"samples":[<br>{<br>"assayName": "Oncomine™ Dx Express Test – DNA and Fusions – US v0.3.0",<br>"sampleId":"Test_Sample4",<br>"purificationProtocol": "FFPE DNA/RNA",<br>"elutionVolumeDNA": 40,<br>"elutionVolumeRNA": 40,<br>"pauseForReview": false<br>}<br>],<br>"notes":"Sample to Result run created by LIMS API",<br>"planName":"PurificationPlan_DNF_US_1Sample_01"<br>}</pre> |

# Create a Nucleic Acid to Result run

| plan-by-samples | |
|---|---|
| URL | `BASE_URL/plan-by-samples` |
| Description | Create a Nucleic Acid to Result run from one or more samples and an assay. The sample or samples and the assay must exist in the system. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |

*(continued)*

| plan-by-samples | |
|---|---|
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/plan-by-samples` |
| Example request | {<br>"reportTemplateName": "",<br>"samples":[<br>{<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"sampleId":"Test_Sample1"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample2"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample3"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample4"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample5"<br>},<br>{<br>"assayName": "{{DNA_RNA_Assay}}",<br>"sampleId":"Test_Sample6"<br>}<br>],<br>"notes":"Nucleic Acid Run created by LIMS API",<br>"planName":"NucleicAcidRun_DNF_6Samples_Run"<br>} |

# Create a library run

| plan-by-library | |
|---|---|
| URL | `BASE_URL/plan-by-library` |
| Description | Create a library run from one or more library batches and an assay. The library batch or batches and the assay must exist in the system. |
| Method | `POST` |
| Header | `curl -k --header 'username:<user name>' --header 'password: <user password>` |
| Example URL | `curl -k --header 'username:<user name>' --header 'password: <user password>'https://<host>/genexus/api/lims/v2/plan-by-library` |
| Example request | {<br>"reportTemplateName": "",<br>"samples":[<br>{<br>"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",<br>"libraryBatchId":"DNA_RNA_LibBatch_01"<br>}<br>],<br>"notes":"Library plan run created by LIMS API",<br>"planName":"PlanByLib_DNF_1L_01"<br>} |

B

# Reference information

## Variant calling criteria

In addition to the run and clinical sample validity criteria listed above, several variant calling criteria established separately for different biomarkers are used to determine which biomarkers are reportable in a passing sample in a successful run. Primary parameters evaluated to produce each variant call include number of reads supporting a variant call, allele frequency, chromosomal strand coverage, and ultimately, alignment with known and targeted hotspots by the panel included in the assay definition file (ADF). The variant caller algorithm calls variants using either read-based evidence or molecular family-based evidence.

**Variant Calling Criteria for Companion Diagnostic Biomarker**

A targeted EGFR exon 20 insertion mutation is called 'positive' if it meets the variant calling criteria specified in the assay definition file. The Oncomine™ Dx Express Test covers codons E762 to R776 and Q787 to L814 of EGFR exon 20. The EGFR exon 20 insertion annotation is applied only to in-frame insertions (or in-frame block substitutions with a net nucleotide gain) affecting codons 762–775.

**Variant Calling Criteria for Tumor Profiling Variants**

The variant calling approach for tumor profiling incorporates multiple algorithms tailored to each variant class. Single nucleotide variants (SNVs), insertions, and deletions are identified by the variant caller algorithm using criteria such as read depth, quality score, and allele frequency. Copy number variants (CNVs) are evaluated using a separate algorithm that considers sample-specific tumor cellularity. Gene fusions and RNA splice variants are detected based on minimum thresholds for unique molecule counts or reads. In certain cases, variant-specific criteria are also applied.

## Rules for variant names in Genexus™ Dx Software

Ion Torrent™ Genexus™ Dx Software provides standard naming conventions for genomic variants and other biomarkers. Standard variant names are derived from sample and results data and properties that are available in the software for each variant, such as variant or biomarker type, gene name, and amino acid change. The conventions are used for both de novo variants and variants that are included in a hotspots file. Variant names are shown in the **Variant Name** column of data results tables in Genexus™ Dx Software, and in reports generated by the software.

## Summary of variant types and examples of standardized variant names

Genexus™ Dx Software applies a set of rules to it a variant, then assigns or returns a name for the variant that best describes variant details. Rules that are used to name variants in the software are as follows.

| Variant or biomarker type | Description | Variant name examples |
|---|---|---|
| Variants with protein change | Variants that provide an amino acid change syntax. | BRAF V600E<br>BRAF p.(V600E) c.1799A>C |
| Variants with coding change but no protein change | Variants that provide a coding change, but for which an amino acid is missing or is `p.?`. | MET c.2942-29_2942-28delinsGG |
| Oncomine class-based variants | Any variant with a variant type of `SNV` or `INDEL` at the locus based on Oncomine™ annotations. These changes are typically complex and are frequently referenced in scientific literature. | EGFR exon 19 deletion<br>MET exon 14 skipping |
| EGFR exon 19 insertion variants | Variant with `Function="nonframeshiftInsertion"` in exon 19 of EGFR gene. | EGFR exon 19 insertion |
| Copy number variants | Copy number gain or loss, signified by a variant type of `CNV`. | ALK amplification<br>PTEN deletion |
| RNA exon variants | Specific RNA exon variants, identified by variant ID. | MET exon 14 skipping<br>EGFRvIII |
| Long deletion Variants | Oncomine Variant Class equals `LongDel` and length attribute is a positive integer. | BRCA1 40 base deletion |
| Exon deletions | A special type of CNV that impacts BRCA genes only. These variants are identified by a type of `CNV`, a subtype of `BigDel`, and call details that specify the exon range affected. | BRCA1 exon 1 deletion |
| Exon duplications | A special type of CNV that impacts BRCA genes only. These variants are identified by a type of `CNV`, a subtype of `BigDup`, and call details that specify the exon range affected. | BRCA1 exon 1 duplication |
| Targeted fusions | Fusions detected by targeting a specific breakpoint. | SLC34A2-ROS1 fusion |
| Non-targeted fusions | Non-targeted fusions | SLC45A3-ETV1 fusion |

| | | |
|---|---|---|
| Expression imbalance variants | Variants for which the Oncomine Variant Class is `"ExpressionImbalance";`. This is measured by the partner-agnostic detection of fusions in driver genes using imbalance in exonic expression. | MET fusion |
| All other mutations | Mutation variants that do not match the criteria of other variants in this table. | ADAMTSL4-AS1 mutation |
| Sample MSI | Sample-level microsatellite stability/instability. This biomarker type is a measure of the entire sample rather than an individual variant. | Microsatellite instability-High<br>Microsatellite stable |
| Expression control variants | Variants with `SVTYPE=ExprControl`. | LMNA expression control |
| Gene expression variants | Variants with `SVTYPE=GeneExpression`. | MET expression |
| Sample TMB | Sample-level tumor mutational burden. This biomarker type is a measure of the entire sample rather than an individual variant. | Tumor Mutational Burden |
| Sample LOH | Sample-level loss of heterozygosity. This biomarker type is a measure of the entire sample rather than an individual variant. | Loss of Heterozygosity |
| Sample TAI | Sample-level telomeric allelic imbalance. This biomarker type is a measure of the entire sample rather than an individual variant. | Telomeric Allelic Imbalance |
| Sample LST | Sample-level large-scale state transition. This biomarker type is a measure of the entire sample rather than an individual variant. | Large-Scale State Transition |
| Gene-level LOH Variants | Oncomine Variant Class<br>equals `"LOH"`. | BARD1 LOH |

# System-installed barcode sets

The Genexus™ Dx Software includes system-installed barcode sets as presets to use in assays. System-installed barcode sets are locked and cannot be changed.

| System-installed barcode set | Description |
|---|---|
| Genexus™ Dx Barcodes 1–32 HD | A unique set of 32 barcode adaptors designed and validated for optimal performance with the Genexus™ Dx Integrated Sequencer. |

# System-installed plugins

Genexus™ Dx Software includes system-installed plugins that expand the analysis capabilities of the software. System-installed plugins are locked and cannot be changed.

These plugins are run automatically in sequencing runs that use Oncomine™ Dx Express Test. Plugin results are included as part of the sample results.

You can have results from these plugins automatically included as part of the sample results.

| System-installed plugin | Description |
|---|---|
| coverageAnalysis | This plugin provides statistics and graphs that describe the level of sequence coverage that is produced for targeted genomic regions.<br>This plugin generates QC metrics. For more information, see “QC results” on page 93. |
| molecular Coverage Analysis | This plugin generates QC metrics. For more information, see “QC results” on page 93. |

# System-installed reference genome files

The Genexus™ Dx Software includes system-installed reference genome files as presets to use in assays. System-installed reference genome files are locked and cannot be changed.

| System-installed reference genome file | Description |
|---|---|
| hg19 | Human reference genome. |

# Default report templates

Genexus™ Dx Software supports the default report templates that are imported with assay definition files. The report templates are pre-configured to generate variant reports and can be used as a template to generate new report templates that include additional customizations. The default report templates are locked and cannot be changed.

# System-installed sample attributes

The following table lists and describes system-installed sample attributes. System-installed sample attributes cannot be edited.

| Sample attribute | Description |
|---|---|
| Sample Name[1] | A unique identifier representing the sample.<br>The sample name can contain only alphanumeric characters (0–9, Aa–Zz), periods (.), underscores (_), or hyphens (-), cannot contain spaces, and is limited to a maximum of 20 characters.<br>IMPORTANT! To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample.<br>Note:<br>• Samples that have been used in a run cannot be deleted.<br>• To prevent duplication, the software checks all sample names and returns an error message if a non-unique sample name is detected. |
| Patient Id[1] | A unique identifier for the patient. |
| Date of Birth[1] | The date of birth for the patient. |
| Ordering Physician[1] | The name of the physician who created the order for the sample. |
| Collection Date[1] | The date that the sample was collected.<br>Click Calendar to select the date in the correct format. |
| Gender[1] | The biological sex of the sample: Female, Male, or Unknown.<br>IMPORTANT! Male or Female must be selected for correct measurement of AR CNV. |
| Sample Type[1] | A term that describes the sample, for example, RNA. You can also select Other, then enter a custom sample type. |
| Nucleic Acid Type | The sample nucleic acid type, such as DNA, or RNA. |
| Application Category[1] | The sample application category, such as Cancer (Germline) or Solid Tumor.<br>Note: If you select an oncology application category in this list, the Cancer Stage, Cancer Type, % Cellularity, and % Necrosis attributes listed below become available in the Add New Sample dialog box. |
| Cancer Type[1] | The type of cancer that is represented by the sample.<br>Select the type of solid or hematologic cancer. If cancer type is unknown, select Unknown Primary Origin. |
| Cancer Stage[1] | The stage of the cancer from which the sample was collected.<br>Select Stage 0–IV, or Primary, Unknown, or Other. |

*(continued)*

| Sample attribute | Description |
|---|---|
| % Cellularity | The percentage of tumor cells over normal cells in the sample. This is a whole number between 1 and 100.<br><br>IMPORTANT!<br>• If this value is not set, % Cellularity is assumed to be 100% in calculations that use this attribute.<br>• % Cellularity is a required attribute for CNV analyses. Do not leave the field blank.<br>• If % Cellularity value is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see "CNVs table" on page 85. |
| % Necrosis | The percentage of cellular necrosis in the sample. This is a whole number between 1 and 100. |
| Notes | An open-entry field for more sample information. |

[1] Required attribute

# System-installed verification templates

Field Service Engineers use the system-installed verification templates during Genexus™ Integrated Sequencer installation.

IMPORTANT! The **Assays / Verification Templates** screen lists the verification templates that are used only by service engineers during Genexus™ Dx Integrated Sequencer installation and performance qualification testing. These templates should not be used or modified in any way.

Details about each verification template is available when you click the name of the template in the software.

- **Performance Qualification Assay**
- **Integrated Purification Performance Qualification Assay**

# C Troubleshooting

## Download Instrument Diagnostics

You can download diagnostics files for troubleshooting purposes. Instrument diagnostics files include:

- Diagnostic archive file (ZIP format)
- Init.log file (TXT format)
- Installation Acceptance Report (PDF format)

You must upload the instrument diagnostics files from the Genexus™ Dx Integrated Sequencer before you can download them in Genexus™ Dx Software.

For more information, see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide*.

1. In the menu bar, click **(Settings)►Services**.
2. Scroll to the bottom of the screen.
3. In the **Download Instrument Diagnostics** section, in the **Select Instrument** dropdown menu, select the instrument.
4. Click **Download**.

# Customer Support Archive (CSA)

You can download an archive that a Technical Support representative can use to diagnose Genexus™ Dx Software issues. The Customer Support Archive (CSA) contains log files and other technical data about your Genexus™ Dx Software and other files from sequencing runs on a Genexus™ Dx Integrated Sequencer.

You can access customer support archive files from the **Results / Sample Results** and the **Results / Run Results** screens.

## Download a Customer Support Archive

A Customer Support Archive (CSA) file is a downloadable archive that a support representative can use to troubleshoot and diagnose issues with sequencing runs or with Genexus™ Dx Software. The archive contains log files and other technical data about the software and other files from sequencing runs.

CSA files are generated for sequencing runs in which the CustomerSupportArchive plugin is run. The plugin can be run automatically during a sequencing run, or manually after a sequencing run.

1. Click **Results►Sample Results**.
2. Download a CSA file using one of the following options.
   - In the **Sample Results** screen, place the pointer over the row of the sample result of interest, then click **CSA**.
   - Click **Results►Sample Results**. Select the sample names of interest for a single sequencing run. Then, place the pointer over the row of the sample result of interest, then click **CSA**.
   - Click **Results►Run Results**. Click the run name of interest. Place the pointer over the run of interest, then click **CSA**. In the **Download CSA** dialog box that appears, select the assay of interest, then click **Download**.

An XZ compressed TAR archive (TXZ) file is downloaded to your computer and is ready to be shared with Support.

# Troubleshoot Genexus™ Dx Software

| Observation | Possible cause | Recommended action |
|---|---|---|
| Cannot sign in to the Genexus™ Dx Software | You have either forgotten your password or are signed out due to several failed login attempts. | Contact the Genexus™ Dx Software system administrator. |
| Batch sample import fails | One or more entries in the sample-import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| Batch sample import fails *(continued)* | Blank rows were copied into the sample-import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean sample-import template file, and be careful to copy only those rows that contain actual data. |
| | The sample import spreadsheet contains a nonunique sample name. | Every sample name in the software must be unique. Ensure that the spreadsheet does not contain any duplicate sample names, then repeat the import. Note that the system check is not case-sensitive, so a sample name of ABC1 conflicts with abc1. |
| | The headings in the sample import spreadsheet do not match the sample attributes in the software. | The headings must match the sample attributes in the software exactly. Check the headings for spelling or other errors. |
| Library batch import fails | One or more entries in the library batch import spreadsheet contains special characters, lines breaks, unexpected spaces, incorrect entry length, incorrect date formatting, or other formatting errors. | Check each entry for correct formatting, correct any errors, and repeat the import. |
| | Blank rows were copied into the library batch import template file from a different source. | Rows that appear empty can contain hidden formatting that conflicts with the import function. Start with a clean library batch import template file, and be careful to copy only those rows that contain actual data. |
| | The library batch import spreadsheet contains a nonunique **Library Batch ID**. | Each **Library Batch ID** in the software must be unique. Ensure that the spreadsheet does not contain any duplicate IDs, then repeat the import. The system check is not case-sensitive. For example, a **Library Batch ID** of ABC1 conflicts with abc1. |
| | A sample name entered in the library batch import spreadsheet does not match a sample name listed in the **Manage Samples** screen. | Ensure that the sample names entered into the spreadsheet are correct and match an existing sample name added to the software. |
| | The Barcode ID name format does not exactly match the format that is used in the **Prepare Library Batch** dialog box. | Use the name format following the Barcode ID name format found in the Barcode Set reference lists (**Settings►References►Barcode Set**), for example: IonDual_0101 through IonDual_0196, or IonHDdual_0101 to IonHDdual_0132. |
| | An invalid library, control, or panel kit barcode has been entered in the spreadsheet. | Ensure that you have correctly entered a valid kit barcode in the appropriate cell of the spreadsheet. |

| Observation | Possible cause | Recommended action |
|---|---|---|
| No information for my loci of interest in the results | The wrong hotspot or BED file is associated with the assay. | 1. Check the hotspot and BED files associated with the assay. If either is incorrect, create a new assay.<br>2. Plan a new run for the sample or sample library with the correct assay.<br>3. Repeat sequencing of the sample or sample library. |
| Cannot download run result files | The run failed. Links to run files are not available for runs that fail QC. | Repeat the run. |

# D Performance characteristics

For performance characteristics of the Oncomine™ Dx Express Test, see the *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* (Pub. No. MAN0024908).

# Documentation and support

## Customer and technical support

Visit **thermofisher.com/support** for the latest service and support information.

- Worldwide contact telephone numbers
- Product support information
  - Product FAQs
  - Software, patches, and updates
  - Training for many applications and instruments
- Order and web support
- Product documentation
  - User guides, manuals, and protocols
  - Certificates of Analysis
  - Safety Data Sheets (SDSs; also known as MSDSs)

  Note: For SDSs for reagents and chemicals from other manufacturers, contact the manufacturer.

## Related documentation

| Document | Publication number |
|---|---|
| *Genexus™ Dx Integrated Sequencer User Guide* | MAN1000073 |
| *Genexus™ Dx Software 6.8 User Guide* | MAN1000074 |
| *Genexus™ Dx Integrated Sequencer Site Preparation Guide* | MAN0025692 |
| *Genexus™ Dx Purification System User Guide* | MAN1000137 |
| *Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide* | MAN0024908 |
| *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide* | MAN0024909 |

# Limited product warranty

Life Technologies Corporation and its affiliates warrant their products as set forth in the Life Technologies' General Terms and Conditions of Sale at **www.thermofisher.com/us/en/home/global/terms-and-conditions.html**. If you have questions, contact Life Technologies at **www.thermofisher.com/support**.

# Glossary

## Genexus™ Dx Software definitions

### adapter dimer (ISPs)

ISPs that carry DNA insert length of less than 8 bp.

### aligned read length

The aligned length of a read at a given accuracy threshold is defined as the greatest position in the read at which the accuracy in the bases up to and including the position meets the accuracy threshold. For example, the AQ17 length of a read is the greatest length at which the read error rate is 2% or less. The "perfect" length is the longest perfectly aligned segment. For all of these calculations, the alignment is constrained to start from position 1 in the read—that is, no 5′ clipping is allowed.

### aligned reads

The number of bases covered by reads aligned to the reference sequence.

### AQ score

An alignment quality (AQ) score defines read accuracy when compared to the reference sequence. The discrepancy can be biological (real variant) or technical (sequencing error). For example, an AQ score of 17 represents 2% aligned read error rate, while an AQ score of 20 represents 1% aligned read error rate when compared to the reference sequence.

### average base coverage depth

The average number of reads of all targeted reference bases.

### average base read depth

The average number of reads of all targeted reference bases that were read at least once.

### BAM file

A BAM (binary alignment map) file (.bam) is the binary version of a SAM (sequence alignment map) file (.sam). A SAM file is a tab-delimited text file that contains sequence alignment data. A BAM file contains aligned reads sorted by reference location.

### barcode

A barcode is a machine-readable code in the form of numbers and a pattern of parallel lines of varying widths, printed on and identifying a product.

There are several applications for barcodes. Libraries can be molecularly barcoded with unique nucleic acid sequence identifiers. Library barcodes are used during data analysis to sort the sequencing results from sequencing reactions that contain combined libraries. Chips and sample tubes also contain unique numeric barcodes that aid in the setup of the experimental analysis workflow.

### base calling

Base calling is the process by which raw data from the sequencing instrument is converted to nucleotide sequences. This is performed by base calling software that is usually run from the instrument itself.

### basecalling input file

Signal processing input files are converted to a single condensed basecalling input file that represents the processed signal. Basecalling input files are required files for basecalling.

### bead loading

The percentage of chip wells that contain live ISPs. The percentage value considers only potentially addressable wells. Bead loading is calculated as number of loaded ISPs divided by the number of potentially addressable wells.

### bp

Abbreviation for "base pair(s)".

### cellularity (%)

The percentage of tumor cells in a given sample.

### BED file

Browser Extensible Data file—BED file—defines chromosome positions or regions.

### CNV

Copy number variation (CNV) is the variation in copy number of any given gene between two samples. CNV is a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals in the human population.

### contig

A contig (from contiguous) is a set of overlapping DNA segments that together represent a consensus region of DNA.

### CSV file

A comma-separated values (CSV) file is a delimited text file in which each line represents a data record with information fields separated by a comma. A CSV file stores tabular data (numbers and text) in plain text. Each line of the file is a data record.

CSV files are easily opened using spreadsheet software, such as Microsoft™ Excel™ or Apache® OpenOffice™ Calc, where each comma-separated field is listed in a separate column.

DAT file

A DAT file is a generic data file. It contains data that may be opened or referenced by a specific application. While some DAT files can be opened directly, most contain program data that is referenced the program when it is running. Therefore, most DAT files are not meant to be opened manually.

empty wells

Wells on a chip that do not contain an ISP.

enrichment (%)

Predicted number of live ISPs that have a key signal identical to the library key signal. The percent enrichment value reported is the number of loaded ISPs that are library ISPs, after taking out the test fragment ISPs.

FASTA file

A FASTA file is a text-based format for representing either nucleotide sequences or peptide sequences, in which base pairs or amino acids are represented using single-letter codes. A sequence in FASTA format begins with a single-line description, followed by lines of sequence data.

FASTQ file

A FASTQ file is a text-based format for storing both a biological sequence (usually nucleotide sequence) and its corresponding quality scores. Both the sequence letter and quality score are each encoded with a single ASCII character for brevity.

FD (flow disruptiveness)

A data filtering parameter that is used instead of INDEL, SNP, and MNP.

final library reads

Number (and percentage) of reads, passing all filters that are recorded in the unmapped BAM file. This value may be different from the total number of reads located in the Library Summary Section due to specifications associated with read trimming beyond a minimal requirement resulting in total number of reads being slightly less than Final Library Reads.

flow

A flow is the event of exposing a chip to one particular dNTP ( deoxyribonucleotide triphosphate), followed by a washing step.

flow order

The order in which a chip is exposed to each particular dNTP. The default Samba flow order consists of a 32-base sequence, repeated. This flow order resists phase errors by providing opportunities for out-of-phase molecules to catch up and is designed to sample all dimer (nucleotide pair) sequences efficiently. Samba is the default flow order because it improves sequencing accuracy for longer reads by resisting phase errors.

### flow transfer

Progress of the sequencing run expressed as number of total flows completed. For example, a sequencing run set to 500 flows shows 250/500 flows in the status column of the **Runs in Progress** table when the sequencing run is half completed.

### fusions

A target technique used for detection and annotation of gene fusions (or translocation of genetic material) in samples.

### hotspots file

A BED or a VCF file that defines regions in the gene that typically contain variants. Specifying a hotspots file to use in a run enables the Torrent Variant Caller (TVC) module module to identify if a specific variant is present or absent. A hotspots file instructs the Torrent Variant Caller (TVC) module module to include these positions in its output files, including evidence for a variant and the filtering thresholds that disqualified a variant candidate. A hotspots file does not affect other parts of the analysis pipeline.

### IA

Isothermal application (IA) is a template preparation technique that uses nonemulsion isothermal reaction to clonally amplify DNA onto an ISP surface.

### INDEL

INDEL is an abbreviation used to designate an insertion or deletion of bases in the genome of an organism.

### intermediate files

Intermediate files contain information used for debugging runs.

### ISPs

Ion Sphere™ Particles (ISPs) are particles that contain bound copies of a single (ideally) DNA fragment amplified during template preparation.

### key signal

Average 1-mer signal in the library key.

### library ISPs

Live ISPs that have a key signal identical to the library key signal.

### LIMS metadata

Laboratory Information Management System (LIMS) software is used for recording sequencing metadata. Entered text is associated with the Planned Run and can be extracted using APIs for LIMS consumption.

live ISP

An ISP with a signal of sufficient strength and composition to be associated with the library or test fragment key.

low quality ISP

An ISP with a low or unrecognizable signal.

library key

A short known sequence of bases used to distinguish a library fragment from a test fragment (for example, "TCAG").

MAPD

Acronym for median absolute pairwise difference. Assuming that adjacent amplicons in the genome most likely have the same underlying copy number in a sample, the difference between the log2(read count ratio) values against the reference baseline for all adjacent amplicons contains information for the noise level of the data. The median of the absolute values of all such difference in log2(read count ratio) is the measure for how informative the results fare for copy number estimates.

mapped reads

Sequencing reads that have been mapped to the reference sequence.

mean raw accuracy

Average raw accuracy of 1-mers at a specific position in the read.

missense SNV

A point mutation that changes the amino acid of the respective protein. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

MNP

Multiple nucleotide polymorphism (MNP) is a genetic mutation in an allele that differs from the reference allele of the same length by >1 nucleotide.

nonsense SNV

A point mutation that changes one of the 20 amino acids into a stop codon, hence a shorter or unfinished protein product. SNV is an acronym for single nucleotide variation, which means at one base there is a difference.

on target reads

Sequencing reads mapped to any target region of the reference. A read is considered to be on target if at least one aligned base overlaps a target region. A read that overlaps a targeted region but where only flanking sequence is aligned, for example, due to poor matching of 5′ bases of the read, is not counted.

### primer dimer

A primer dimer (PD) is a potential by-product in PCR, a common biotechnological method. As its name implies, a PD consists of primer molecules that have attached (hybridized) to each other because of strings of complementary bases in the primers

### primer dimer ISP

An ISP that carries an insert length of less than 8 base pairs.

### read length

The length of called reads measured in base pairs.

### read

The sequence of a section of a unique fragment obtained after the end of the sequencing process.

### reference library

A consensus nucleotide sequence that represents the genome of a particular species. The results from a sequencing run are compared to the reference library to identify sequence variants.

### SAM file

A SAM (sequence alignment map) file (.sam) is a tab-delimited text file that contains sequence alignment data. It stores biological sequences aligned to a reference sequence.

### sample

Genetic material from one source (for example, DNA from one individual).

### signal processing input files

Signal processing input files consist of the raw voltage measurement data collected during the sequencing run. These files are required to reanalyze the run from signal processing.

### SNP

Single nucleotide polymorphism (SNP) is a genetic mutation in an allele that differs from the reference allele of the same length by one nucleotide.

### SSH

SSH, also known as Secure Shell or Secure Socket Shell, is a network protocol that gives users, particularly system administrators, a secure way to access a computer over an unsecured network. SSH also refers to the suite of utilities that implement the SSH protocol.

### structural variants

Genetic mutations that cause a change in the organism's chromosome structure, such as insertions, deletions, copy number variations, duplications, inversions, and translocations.

### target base coverage

Summary statistics for targeted base reads of the reference. A base covered by multiple target regions is counted only once per sequencing read.

### target regions file

A BED file that specifies all of the regions that a panel represents such as the amplified regions that are used with target sequencing. The complete software analysis pipeline is restricted to include only these specified regions instead of the entire reference library.

### test fragment

A test fragment (TF) is a known nucleotide sequence that is used to monitor system characteristics.

### test fragment ISPs

Live ISPs with a key signal that is identical to the test fragment key signal.

### test fragment key

A test fragment key (TF key) is the nucleotide sequence that is used to identify test fragment reads.

### TF key peak counts

Signal strength of the first three bases of the TF key.

### total reads

Total number of filtered and trimmed reads independent of length reported in the output BAM file.

### TSV file

A tab-separated values (TSV) file is a tab-delimited file that is used with spreadsheet software. TSV files are essentially text files, and the raw data can be viewed by text editors, though they are often used when moving raw data between spreadsheets. See also *VCF file*.

### uBAM file

An unmapped BAM (uBAM) file is a variant form of the BAM file format in which the read data does not contain mapping information. This is basically an "off-label" use of the BAM format (which was specifically designed to contain mapping information) that is used for data management reasons. It allows you to attach metadata to the reads from as early on in the analysis process as possible.

### unaligned reads

Nucleotide bases covered by reads that are not aligned to the reference.

### uniformity of base coverage

The percentage of bases in all targeted regions (or whole genome) covered by at least 0.2X the average base coverage depth.

### usable sequence / usable reads

Usable reads consist of library ISPs that pass the polyclonal, low quality, and primer dimer filter.

### VCF file

A variant call format (VCF) file specifies a variant of interest and its location. This file stores the differences between the BAM file and the reference file.

### wells with ISPs

Number of wells that were determined to be positive for the presence of an ISP within the well. Wells containing ISPs have a delayed pH change due to the presence of an ISP slowing the detection of the pH change from the solution.

### XLS file

Files that use the .xls extension. XLS files can be created by Microsoft™ Excel™ and other similar spreadsheet programs.