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 MAN1001774

Revision A

IVD

For In Vitro Diagnostic Use. | Rx Only

ThermoFisher SCIENTIFIC

Life Technologies Holdings Pte Ltd |
Block 33 |
Marsiling Industrial Estate Road 3 |
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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: MAN1001774 A (English)

Revision	Date	Description
A	9 September 2026	New IVD user guide for the Oncomine™ Dx Express Test 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 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 a companion diagnostic to identify patients who may benefit from treatment with the targeted therapies 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)
	ERBB2/*HER2*	*ERBB2*/*HER2* activating mutations (SNVs in exons 18–21 within the tyrosine kinase domain and exon 20 insertions)	HYRNUO® (sevabertinib)

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—FFPE samples

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. For FFPE samples, remaining nucleic acid is loaded into a separate archive plate for storage and future use. Other IVD-labeled sample extraction methods are acceptable for FFPE samples.

The recommended number of slide-mounted 5-µm FFPE sections or curls used in extraction varies depending on the sample collection method (see Table 3).

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	0.5 ng/µL	0.5–512 ng/µL

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, RNA variant, and copy number variant (CNV) calls.
Run Report	A summary of run results and information, status, reads per barcode, reagents used, and kit barcodes.
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: • 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	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* and *ERBB2* 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).
30. For *MET* Exon 14 skipping results for cancer types other than non-small cell lung cancer, confirmatory testing using a clinically validated assay is recommended.

31. For NSCLC, the Oncomine™ Dx Express Test assay definition file includes prevalent, but not all rare or newly identified, *EGFR* exon 20 insertions, *ERBB2*/*HER2* exon 20 insertions, and *ERBB2*/*HER2* SNVs in exons 18-21 within the tyrosine kinase domain. The Oncomine™ Dx Express Test may miss the following rare or newly identified variants:
    - *EGFR* exon 20 insertions carried by a subset of patients who may derive benefit from sunvozertinib.
    - *ERBB2*/*HER2* activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions) carried by a subset of patients who may derive benefit from sevabertinib.

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 6.8.2.1 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. MAN1001776).

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. MAN1001775).

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

Table 6 Oncomine Dx Express Test components *(continued)*

✓	Component	Catalog or Part No.
	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
	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

Table 7 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 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 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.

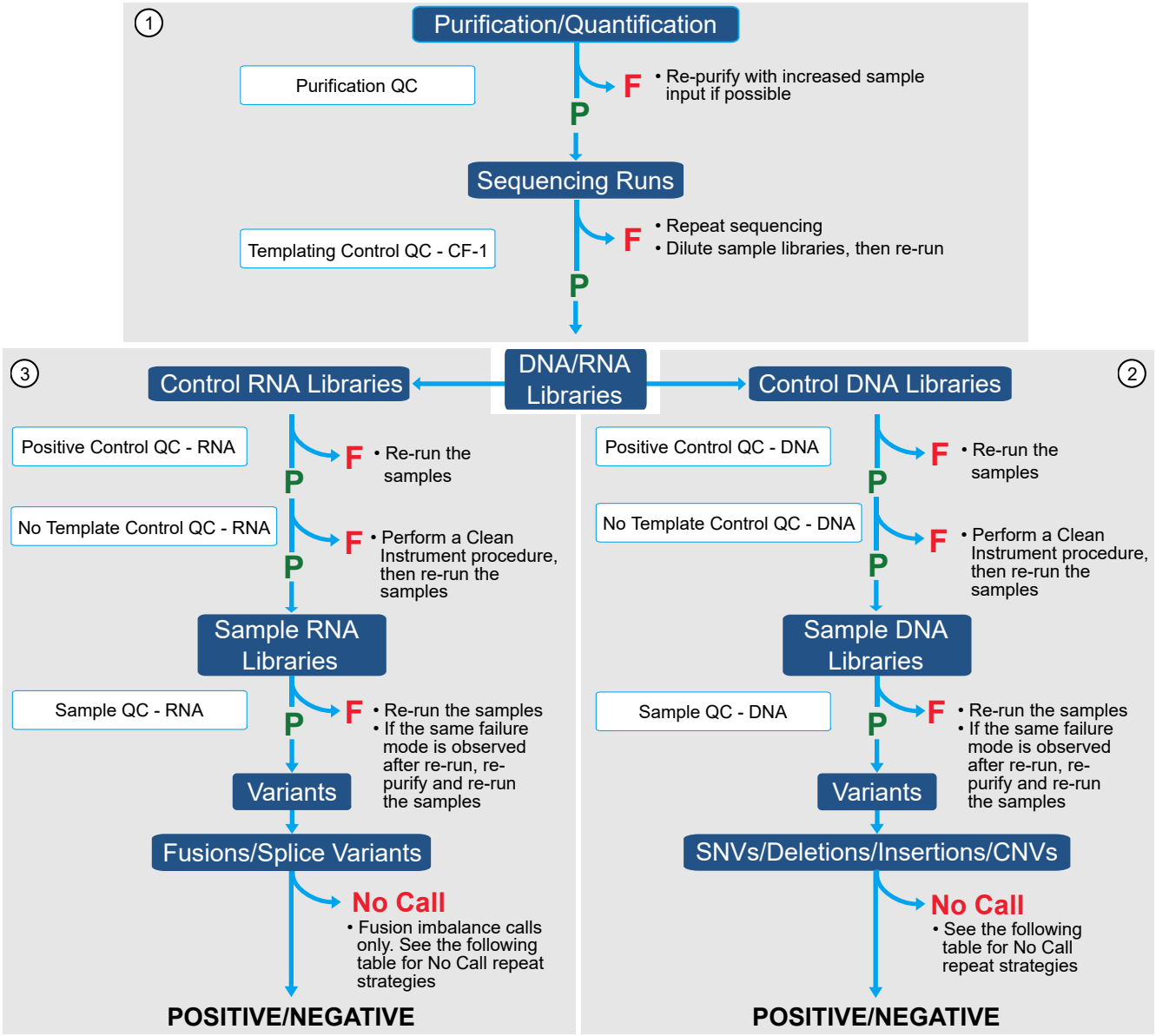


Figure 1 Repeat strategy for FFPE samples (DNA and RNA)

Table 9 Retest strategy

Point of failure	Specification type	Passing criteria	Repeat strategy
1	Purification QC	The following minimum concentrations for DNA, RNA, (FFPE) are required: • DNA ≥0.5 ng/µL • RNA ≥0.5 ng/µL	If the minimum concentration requirement is not met for DNA or RNA, the samples must be repurified 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: • CF-1 Mean AQ20 Read Length (bp) must be ≥82 • 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: NTC QC - DNA • Total hotspot calls must = 0 • Average base coverage depth must be ≤15 NTC QC - RNA • Mapped reads must be ≤2848 • 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*). **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 *(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: • MAPD ≤0.5 • Mapped reads ≥100,000 • AQ20 Mean Read Length (bp) ≥76 • 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. • ERBB2 copy number ≥5.0 • MET copy number ≥7.0	If any of these specifications fail, the operator should rerun 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. **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: • Targeted isoforms in the RNA control sample must be called "Positive". • Expression imbalance controls in the RNA control sample must be called "Positive". • RNA expression controls detected =7 • Mapped fusion reads ≥50,475	If any of these specifications fail, the operator should rerun 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. **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. • Mapped reads ≥100,000 • MAPD ≤0.5 • Mean AQ20 Read Length (bp) ≥64	If any of these specifications fail, the operator should rerun the sample, or repurify the sample using increased input material, if possible. **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 *(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. • Mapped fusion reads ≥6,000 • RNA Expression Controls detected ≥5	Rerun the sample alone, or with fewer samples. If the RNA sample library still fails this specification, repurify using more tissue input, if possible. **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] • SNV allelic frequency ≥2.5% • Deletion/Insertion allelic frequency ≥2% • 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 repurify 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] • Fusion Read Count ≥21, or • 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 222.	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 DNA only
	Sample Concentration RNA	Yes	Yes	No	No	Failed sample RNA 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 DNA only
	Mean AQ20 Read Length	No	No	Yes	Yes	Failed sample DNA only
	Mapped Reads	No	No	Yes	Yes	Failed sample DNA only
Sample QC - RNA	Mapped Fusion Reads	Yes	Yes	No	No	Failed sample RNA only
	RNA Expression Controls Detected	Yes	Yes	No	No	Failed sample RNA 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.
NOCALLxPredictedSSE	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.
NOCALLxPositiveSSE	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.
NOCALLxNegativeSSE	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.

Table 11 Variant call filters and No Call repeat strategies *(continued)*

Observation (Example FR tag)	Interpretation[1]	Repeat strategy
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.
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 *PREDICTIONRef SHIFTx*	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.

Table 11 Variant call filters and No Call repeat strategies *(continued)*

Observation (Example FR tag)	Interpretation[1]	Repeat strategy
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.
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_NONZERO_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.

Table 11 Variant call filters and No Call repeat strategies *(continued)*

Observation (Example FR tag)	Interpretation[1]	Repeat strategy
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.



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 study I—*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					
		Positive	Negative	Invalid[1]	No Calls	TNP[2]	Total
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 13. 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 Oncomine™ Dx Express Test 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 study II—*ERBB2*/*HER2* activating mutation biomarker

The accuracy of the Oncomine™ Dx Express Test for detecting *ERBB2*/*HER2* tyrosine kinase domain (TKD) activating mutations (SNVs and 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 92 tissue samples were obtained from subjects enrolled in the HYRNUO® (sevabertinib) clinical trial (SOHO-01; NCT05099172). All subjects were determined to be *ERBB2*/*HER2* activating mutation-positive based on the clinical trial assays (CTAs) used for enrollment. Additionally, 150 tissue samples were procured from commercial vendors and screened using NGS-based assays representative of the technologies commonly used for patient enrollment in the SOHO-01 trial to identify *ERBB2*/*HER2* activating mutation-negative cases. Of these biomarker-negative samples, 130 were confirmed as negative and 20 were excluded due to QC/extraction failures.

A total of 222 samples including 92 *ERBB2*/*HER2* activating mutation-positive samples and 130 *ERBB2*/*HER2* activating mutation-negative samples were initially submitted for testing with the Oncomine™ Dx Express Test. Of the 92 biomarker-positive samples, 89 were tested using the Oncomine™ Dx Express Test. All 130 biomarker-negative samples were tested with the Oncomine™ Dx Express Test. Among the 219 samples tested using the Oncomine™ Dx Express Test, 89 were confirmed as *ERBB2*/*HER2* activating mutation-positive and 130 were identified as negative. The 89 *ERBB2*/*HER2* activating mutation-positive samples (including both TKD activating and non-TKD activating variants) tested with the Oncomine™ Dx Express Test were submitted for testing with the orthogonal method; however, 25 samples could not be tested, and only 64 could be evaluated. Of these, 58 were confirmed as positive, 3 were identified as negative, and 3 produced invalid results by the orthogonal method. The 130 *ERBB2*/*HER2* activating mutation-negative samples tested with the Oncomine™ Dx Express Test were submitted for testing with the orthogonal method. Of these, 125 were confirmed as negative and 5 produced invalid results by the orthogonal method.

The analytical concordance between the results from the Oncomine™ Dx Express Test and the orthogonal method for detecting *ERBB2*/*HER2* activating mutations, based on a total of 222 samples (92 *ERBB2*/*HER2* activating mutation-positive samples and 130 *ERBB2*/*HER2* activating mutation-

negative samples) initially submitted for testing with the Oncomine™ Dx Express Test, is summarized in Table 14.

Table 14 Analytical concordance between the Oncomine™ Dx Express Test (ODxET) and orthogonal method for *ERBB2/HER2* activating mutations

		Orthogonal method				
		Positive	Negative	TNP[1]	Invalid[2]	Total
ODxET	Positive	58	3	25	3	89
	Negative	0	125	0	5	130
	TNP[1]	0	0	3	0	3
	Total	58	128	28	8	222

[1] TNP: Test Not Performed due to DNA concentration fail, tumor content fail, insufficient material, or DNA.
[2] Invalid were considered unknown or unevaluable.

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 15. The point estimates were 100.0% (58/58) for PPA, 97.7% (125/128) for NPA, and 98.4% (183/186) for OPA.

Table 15 Agreement between the Oncomine™ Dx Express Test (ODxET) and orthogonal method for *ERBB2/HER2* activating mutation detection

		Including or excluding ODxET unknowns		
Parameter	Agreed (N)	Total (N)	Percent agreement	95% CI
PPA	58	58	100.0%	(93.8%, 100.0%)
NPA	125	128	97.7%	(93.3%, 99.2%)
OPA	183	186	98.4%	(95.4%, 99.4%)

In the SOHO-01 trial, the study population was enriched for cases positive for *ERBB2/HER2* activating mutations. 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. At a prevalence of 1.8%, the adjusted point estimates for PPA and NPA were 100.0% (95% CI: 93.8%, 100.0%) and 99.1% (95% CI: 95.4%, 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 a prevalence of 1.8% for *ERBB2/HER2* activating mutations in NSCLC. The PPV was 43.9% (95% CI: 25.4%, 100.0%) and the NPV rounded to 100.0% (95% CI: 97.0%, 100.0%).

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 prescreened 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 prescreening 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 16.

Table 16 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 16 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 17.

Table 17 Accuracy results for Oncomine™ Dx Express Test (ODxET) tumor profiling variants by variant type and clinical significance—Accuracy Study I

Variant type	FDA Biomarker category[1]	Total unique variants/genes	ODxET + Comparator +[2]	ODxET + Comparator –[2]	ODxET – Comparator +[2]	ODxET – Comparator –[2]	PPA (n/N) [95% CI][3]	NPA (n/N) [95% CI][3]
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 17 Accuracy results for Oncomine Dx Express Test (ODxET) 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 +[2]	ODxET + Comparator –[2]	ODxET – Comparator +[2]	ODxET – Comparator –[2]	PPA (n/N) [95% CI][3]	NPA (n/N) [95% CI][3]
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[4]	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/A	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 17 Accuracy results for Oncomine Dx Express Test (ODxET) 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 +[2]	ODxET + Comparator –[2]	ODxET – Comparator +[2]	ODxET – Comparator –[2]	PPA (n/N) [95% CI][3]	NPA (n/N) [95% CI][3]
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] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[3] 95% 2-sided confidence interval calculated via the Wilson Score method.

[4] 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 18.

Table 18 Accuracy results for SNVs, insertions, and deletions in clinically and analytically meaningful bins—Accuracy Study I

Variant type	Bin	Total unique variants	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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[3]	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 18 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 +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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 18 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 +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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 18 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 +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[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 CNVs and RNA variants (including fusions and splice variants), excluding unknown results, is presented at the gene level in Table 19.

Table 19 Accuracy results for CNV and RNA variants at the gene level—Accuracy Study I

Variant type	Gene name	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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[3]	100.0% (9310/9310) [100.0%,100.0%]

Table 19 Accuracy results for CNV and RNA variants at the gene level—Accuracy Study I *(continued)*

Variant type	Gene name	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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%]

Table 19 Accuracy results for CNV and RNA variants at the gene level—Accuracy Study I *(continued)*

Variant type	Gene name	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
Splice variants/RNA exon variants	*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] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[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.

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 20. 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 20 Accuracy results for all variant types stratified by LoD level—Accuracy Study I

Variant type	LoD level	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N [95% CI][2]	NPA (n/N) [95% CI][2]	PPV (n/N) [95% CI][2]
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[3]	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%]
	≥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%]

Table 20 Accuracy results for all variant types stratified by LoD level—Accuracy Study I *(continued)*

Variant type	LoD level	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N [95% CI][2]	NPA (n/N) [95% CI][2]	PPV (n/N) [95% CI][2]
CNVs	<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%]
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%]

Table 20 Accuracy results for all variant types stratified by LoD level—Accuracy Study I *(continued)*

Variant type	LoD level	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N [95% CI][2]	NPA (n/N) [95% CI][2]	PPV (n/N) [95% CI][2]
RNA splice variants	≥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] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[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.

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 21 shows the 3 × 3 contingency table accounting for all 74 breast ERBB2

CNV samples tested by both the Oncomine™ Dx Express Test and the orthogonal method, along with the PPA and NPA estimates.

Table 21 Concordance between the Oncomine™ Dx Express Test (ODxET) 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	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 22 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 22 Concordance between Oncomine™ Dx Express Test (ODxET) and orthogonal method for detection of Level 2 *ALK* fusions in NSCLC—Accuracy study II

		Orthogonal method			
		Positive	Negative	Unknown[1]	Total
ODxET	Positive	26	1	13	40
	Negative	0	41	10	51
	Unknown	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%]			

Table 22 Concordance between Oncomine Dx Express Test (ODxET) and orthogonal method for detection of Level 2 ALK fusions in NSCLC—Accuracy study II *(continued)*

		Orthogonal method			
		Positive	Negative	Unknown[1]	Total
Excluding unknown	NPA (n/N) [95% CI][2]	97.6% (41/42) [87.7%, 99.6%]			
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 23 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 23 Concordance between Oncomine™ Dx Express Test (ODxET) 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	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.

A summary 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 24. 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 24 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 +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA {n/N) [95% CI][2]	NPA (n/N) [95% CI][2]	PPV (n/N) [95% CI][2]
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[3]	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%]

Table 24 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 +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA {n/N) [95% CI][2]	NPA (n/N) [95% CI][2]	PPV (n/N) [95% CI][2]
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%]
	≥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] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[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.

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 25. All *EGFR* variants were tested at levels above 1X LoD.

Table 25 Accuracy results for *EGFR* exon 19 deletions, L858R, and T790M tumor profiling variants—Accuracy study III

Variant type	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
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[3]	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%]
*EGFR*_non-homopolymers	4	0	—	—	100.0% (4/4) [51.0%, 100.0%]	N/E
*EGFR*_homopolymers	19	1	—	—	100.0% (19/19) [83.2%, 100.0%]	N/E

Table 25 Accuracy results for EGFR exon 19 deletions, L858R, and T790M tumor profiling variants—Accuracy study III *(continued)*

Variant type	ODxET + Comparator +[1]	ODxET + Comparator –[1]	ODxET – Comparator +[1]	ODxET – Comparator –[1]	PPA (n/N) [95% CI][2]	NPA (n/N) [95% CI][2]
*EGFR*_non-SSRs	19	1	—	—	100.0% (19/19) [83.2%, 100.0%]	N/E
*EGFR*_SSRs	4	0	—	—	100.0% (4/4) [51.0%, 100.0%]	N/E
*EGFR*_GC [30%–40%]	0	0	—	—	N/E	N/E
*EGFR*_GC [40%–50%]	11	0	—	—	100.0% (11/11) [74.1%, 100.0%]	N/E
*EGFR*_GC [50%–60%]	12	1	—	—	100.0% (12/12) [75.8%, 100.0%]	N/E
*EGFR*_GC [60%–75%]	0	0	—	—	N/E	N/E

[1] "+" in this context indicates variant-positive using the test; "–" indicates variant-negative using the test.

[2] 95% 2-sided confidence interval calculated via the Wilson Score method.

[3] 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 26). 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 26).

Table 26 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 1: *ERBB2*/*HER2* activating SNVs (43)	0% (0/[43 × 192])	0% (0/192)	0% (0/[43 × 192])	0% (0/192)
Level 1: *ERBB2*/*HER2* exon 20 insertion (44)	0% (0/[44 × 192])	0% (0/192)	0% (0/[44 × 192])	0% (0/192)
Level 2: Panel-wide SNVs (193)	0% (0/[193 × 192])	0% (0/192)	0% (0/[193 × 192])	0% (0/192)
Level 2: Panel-wide INDELs (410)	0% (0/[410 × 192])	0% (0/192)	0% (0/[410 × 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)

Table 26 False positive rates per position and per sample before and after masking of Level 3 SNVs with VAFs <5% *(continued)*

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 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 studies

Limit of Detection (LoD) study I

- Limit of Detection (LoD) for CDx variants—*EGFR* exon 20 insertion 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.

Limit of Detection (LoD) study II—*ERBB2/HER2* variants

The LoD of the Oncomine™ Dx Express Test for *ERBB2*/*HER2* 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 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 *ERBB2*/*HER2* exon 20 insertion variants ranged from 2.98% to 4.07% VAF (mean = 3.71%).

The LoD of the Oncomine™ Dx Express Test for *ERBB2*/*HER2* SNVs was determined using 4 patient-derived samples containing 5 SNVs (R678Q, T733I, D769Y, V842I, and R896H). DNA from these samples was blended with DNA extracted from a WT NSCLC sample to create 6 VAF levels per variant 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 *ERBB2*/*HER2* SNVs ranged from 3.60% to 6.42% VAF (mean = 4.46%).

Nucleic acid input studies

Nucleic acid input study I

- DNA and RNA input for CDx variants—*EGFR* exon 20 insertion 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 4 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 α=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.

Nucleic acid input study II—*ERBB2*/*HER2* 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 *ERBB2*/*HER2* 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 3 FFPE NSCLC patient samples containing *ERBB2*/*HER2* exon 20 insertions of 3, 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, except for the *ERBB2*/*HER2* exon 20 insertion of 12 bp, which showed the 7.5 ng input level generated lower VAFs than the 10 ng standard input by Tukey-Kramer pairwise comparison. The positive call rate for all DNA reference locations was 100% across all input levels. The lowest negative call rate 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 *ERBB2*/*HER2* exon 20 insertion variants.

To verify the performance of the Oncomine™ Dx Express Test at the product requirement of 10 ng DNA input and to establish the acceptable range of DNA input for accurate detection of *ERBB2*/*HER2* SNVs, 5 DNA input levels were evaluated (5 ng, 7.5 ng, 10 ng (standard input), 12.5 ng, and 15 ng). The study used DNA extracted from 2 *ERBB2*/*HER2* SNV-positive FFPE clinical samples, 1 tyrosine kinase domain variant (G776V) from lung and 1 non-tyrosine kinase domain variant (R896H) from non-lung, each blended with WT FFPE NSCLC DNA to achieve VAFs at 1–1.5X 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 positive call rate for all DNA reference locations was 100% across all input levels. These results support the performance of the Oncomine™ Dx Express Test at the recommended DNA input of 10 ng and confirm its robustness across a range of DNA input levels for the accurate detection of *ERBB2*/*HER2* SNVs.

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 quantification 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 the

minimum 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%.

Interfering substances studies

Interfering substances study I

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 27 shows the interferent evaluated.

Table 27 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 µmol/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.

Interfering substances study II—*ERBB2* variants

This study evaluated the impact of 5 potentially interfering substances on the ability of the Oncomine™ Dx Express Test to detect *ERBB2* single nucleotide variants (SNVs) in formalin-fixed, paraffin-embedded (FFPE) clinical samples on the Genexus™ Dx Integrated Sequencer.

A total of 2 FFPE clinical samples were used in the study, including 1 *ERBB2* SNV-positive sample and 1 SNV-negative (WT) sample (Table 28). The *ERBB2* SNV-positive sample was blended with the WT sample to generate a sample blend with variant frequency at 1–1.5X LoD. Six replicates were tested per condition in the study.

Table 28 Samples used in the study

Sample ID	Tissue	Sample type	Gene	Variant ID	Amino acid change	Variant AF
AD5573	Breast	Resection	*ERBB2*	COSM14060	p.L755S	10.7%
BN3103	Lung	Resection	WT	N/A	N/A	N/A

FFPE samples were processed through the entire assay workflow. The samples were spiked with added amounts of the listed substances at the processing step indicated in Table 29.

Table 29 Interfering substances and amounts

Condition	Interferent	Step in workflow	Amount of interferent
1	Paraffin	At the deparaffinization step, add 4X paraffin content to AutoLys tube	4X of normally expected levels
2	Xylene	Add 0.3 mL xylene into 1 mL ethanol during the first ethanol wash	6X residual expected (v/v) xylene in the ethanol bath
3	Wash buffer	Add 10% excess of Wash 2 Buffer used for nucleic acid dilution	5 µL wash buffer + 45 µL elution solution (equivalent to 10% Wash 2 Buffer carried over into eluate)
4	Proteinase K	Add 10X Proteinase K into the sample solution after the Proteinase K digestion and before transferring to purification plate 1	2.5 µL (>10X of expected residual Proteinase K after the heat-kill step)
5	Hemoglobin	Add hemoglobin to the Proteinase K digestion mixture before Proteinase K digestion step	4 mg/mL
6	Standard operating procedure (SOP) control	Process tissue sections without increased addition of potentially interfering substances	N/A

For each interferent, the positive concordance (No Calls included or excluded) was computed as the proportion of positive variant calls without interferent which were also positive in the presence of the interferent. For each interferent, the overall concordance (No Calls included or excluded) was computed as the proportion of all variant locations which yielded equivalent calls with and without interferent.

For all variants tested, the results showed that for each interferent, the positive concordance (No Calls excluded) was 100%. The overall concordance (No Calls excluded) for all variants was 100%. The study results show that the interfering substances listed in Table 29 do not affect the performance of the assay at the levels tested.

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, misprimed or 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 types of reproducibility studies were conducted to evaluate the precision and reproducibility of the Oncomine™ Dx Express Test: sample processing reproducibility studies and external panel reproducibility studies. Sample processing reproducibility studies began with the nucleic acid extraction step to assess the reproducibility and repeatability of the entire sample processing workflow. Panel reproducibility studies 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 studies, see "Assay panel reproducibility studies" on page 74.

Sample processing reproducibility study I—*EGFR* exon 20 insertions

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 30.

Table 30 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—*ERBB2*/*HER2* exon 20 insertions and SNVs

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 *ERBB2*/*HER2* exon 20 insertion variants in NSCLC FFPE tissue samples. Three NSCLC FFPE samples harboring *ERBB2*/*HER2* exon 20 insertions of 3, 6, and 12 bp in length, along with 1 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 *ERBB2*/*HER2* exon 20 insertion variants, with no instances of No Calls observed, and the negative call rate for the WT sample was also 100% (Table 31).

Table 31 Sample reproducibility for *ERBB2*/*HER2* 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]
ERBB2 exon 20 insertion (3 bp)	COSM12552	2.17X	12	12	0	0	100.0% (75.8%, 100.0%)	0.0% (0.0%, 24.2%)
ERBB2 exon 20 insertion (6 bp)	COSM303939	6.11X	12	12	0	0	100.0% (75.8%, 100.0%)	0.0% (0.0%, 24.2%)
ERBB2 exon 20 insertion (12 bp)	COSM20959	1.92X	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.

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 *ERBB2*/*HER2* SNVs in NSCLC FFPE tissue samples. Two FFPE samples harboring *ERBB2*/*HER2* SNVs (L755S and R678Q), along with 1 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 (4 replicates per sample per run) on separate days, resulting in 24 replicates per sample (4 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 *ERBB2*/*HER2* SNVs, with no instances of No Calls observed, and the negative call rate for the WT sample was also 100% (Table 32).

Table 32 Sample reproducibility for *ERBB2*/*HER2* SNVs

Variant	Variant ID	Total calls	Positive calls	Negative calls	No Calls	Positive call rate (95% CI)[1]	Negative call rate (95% CI)[1]
ERBB2 SNV (L755S)	COSM14060	24	24	0	0	100.0% (86.2%, 100.0%)	0.0% (0.0%, 13.8%)
ERBB2 SNV (R678Q)	COSM436498	24	24	0	0	100.0% (86.2%, 100.0%)	0.0% (0.0%, 13.8%)
WT	N/A	2088	0	2088	0	0.0% (0.0%, 0.2%)	100.0% (99.8%, 100.0%)

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

Sample processing reproducibility study III—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 33, 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 33 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 33 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 33 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 33 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 34 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 34 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 34 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 34 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 35 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 35 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 35 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 35 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 36 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 36 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 studies, see “Sample processing reproducibility studies” on page 61.

Assay panel reproducibility study I—*EGFR* exon 20 insertions

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 37 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 37 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)	COSM12378	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 37 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 38).

Table 38 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)	COSM12378	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 38 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	504	501	100.0% (98.3%, 99.9%)	100.0% (99.3%, 100.0%)
WT02	WT	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.

The PPA and NPA across all three test sites were 100% for all variants at both LoD levels tested.

Assay panel reproducibility study II—*ERBB2*/*HER2* exon 20 insertions and SNVs

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 *ERBB2*/*HER2* exon 20 insertion variants in NSCLC FFPE tissue samples. The study included 3 NSCLC FFPE samples harboring *ERBB2*/*HER2* exon 20 insertions of 3, 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 *ERBB2*/*HER2* 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 3 bp and 9 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 39 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 *ERBB2*/*HER2* 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 39 Panel reproducibility for *ERBB2*/*HER2* 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]
ERBB2 exon 20 insertion (3 bp)	COSM12553	1.38X	4.12%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
		2.66X	7.92%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
ERBB2 exon 20 insertion (9 bp)	COSM12556	1.31X	5.14%	72	72	0	0	100.0% (94.9, 100.0%)	0.0% (0.0%, 5.1%)
		2.53X	8.25%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
ERBB2 exon 20 insertion (12 bp)	COSM20959	1.00X	5.22%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
		1.41X	7.40%	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 *ERBB2*/*HER2* 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 40).

Table 40 Repeatability results—*ERBB2*/*HER2* 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]
ERBB2 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%)
ERBB2 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%)
ERBB2 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%)
WT01	N/A	N/A	1008	0	1005	3	504	501	100.0% (99.3%, 100.0%)	100.0% (98.3%, 99.9%)
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.

The PPA and NPA across all three test sites were 100% for all variants at both LoD levels tested, excluding No Calls.

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 *ERBB2*/*HER2* SNVs in NSCLC FFPE tissue samples. The study included 2 FFPE samples harboring *ERBB2*/*HER2* SNVs (G776V and R896H), 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 Oncomine™ Dx Express Test reagents and consumables. Each test site included 2 operators and 2 instruments. DNA blends containing *ERBB2*/*HER2* SNVs were prepared to represent two variant

levels: 1–1.5X LoD and 2–3X LoD. 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 41 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 *ERBB2*/*HER2* SNVs, with no instances of No Calls observed. The negative call rate, evaluated using 2 WT NSCLC samples (WT1 and WT2) and calculated for relevant hotspots, was ≥99.3%, excluding No Calls.

Table 41 Panel reproducibility for *ERBB2*/*HER2* SNVs

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]
ERBB2 SNV (G776V)	COSM18609	1.7X	5.4%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
		3.0X	9.8%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
ERBB2 SNV (R896H)	COSM119971	1.2X	4.6%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
		2.6X	9.5%	72	72	0	0	100.0% (94.9%, 100.0%)	0.0% (0.0%, 5.1%)
WT1	N/A	N/A	N/A	144	1	142	1	0.7% (0.1%, 3.9%)	99.3% (96.1%, 99.9%)
WT2	N/A	N/A	N/A	144	0	144	0	0.0% (0.0%, 2.6%)	100.0% (97.4%, 100.0%)

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

Repeatability for the detection of *ERBB2*/*HER2* SNVs was evaluated across within-run, between system, between-operator, between- site, between-lot, and total variability. When excluding No Calls from the reproducibility study data, the within-run repeatability was ≥97.2% (Table 42).

Table 42 Repeatability results—*ERBB2*/*HER2* SNVs

Sample	Variant	Variant ID	LoD level	Total pairs	Concordant pairs		Within-run repeatability	
					No Calls excluded	No Calls included	No Calls excluded (95% CI)[1]	No Calls included (95% CI)[1]
D01	*ERBB2* SNV (G776V)	COSM18609	1.7X	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)
D04	*ERBB2* SNV (G776V)	COSM18609	3.0X	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)
D02	*ERBB2* SNV (R896H)	COSM119971	1.2X	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)
D05	*ERBB2* SNV (R896H)	COSM119971	2.6X	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)
D03	WT1	COSM18609	N/A	36	36	36	100.0% (90.4%, 100.0%)	100.0% (97.2%, 100.0%)
D03	WT1	COSM119971	N/A	36	35	34	97.2% (85.8%, 99.5%)	94.4% (90.4%, 100.0%)
D06	WT2	COSM18609	N/A	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)
D06	WT2	COSM119971	N/A	36	36	36	100.0% (90.4%, 100.0%)	100.0% (90.4%, 100.0%)

[1] 95% 2-sided confidence interval calculated via the Wilson Score method.

Assay panel reproducibility study III—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 43. The PPA ranged from 87.3% (for *BRAF* V600E in colorectal cancer) to 100%, with no calls excluded from the analysis.

Table 43 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%)

Table 43 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	
4	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%)	98.86% (153177/154944) (98.81%, 98.91%)
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%)	

Table 43 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	
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%)
10	Level 3	AR (Amplifi-cation)	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 (Amplifi-cation)	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 (Amplifi-cation)	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 (Amplifi-cation)	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 (Amplifi-cation)	CNV	Melanoma	1–1.5X	6.64	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	99.09% (154253/155 664) (99.05%, 99.14%)
14	Level 3	KRAS (Amplifi-cation)	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% (152560/155 016) (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%)	

Table 43 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	
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%)
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%)	

Table 43 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	
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%)
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%)

Table 43 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	
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%)
30	Level 2	CCDC6-RET.C1R12.COSF1271.1	Fusion	Thyroid	1–1.5X	11.47	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	99.18% (70336/70920) (99.11%, 99.24%)
31	Level 2	FGFR3-TACC3.F17T11.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% (70336/70920) (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% (70406/70920) (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% (70342/70920) (99.12%, 99.25%)
34	Level 2	BCR-NTRK2.B1N17	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% (70326/70920) (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% (70345/70920) (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% (70344/70920) (99.12%, 99.25%)

Table 43 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	
37	Level 2	SQSTM1-NTRK3.S5N14	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% (70342/70920) (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% (70344/70920) (99.12%, 99.25%)
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%)

Table 43 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	1.11X	5.9	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	99.68% (154594/155088) (99.65%, 99.71%)
					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%)

Table 43 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	
44	Level 2	EGFR p.L858R	SNV	NSCLC	1.95X	10.64	98.6% (71/72) (92.5%, 99.8%)	100.0% (71/71) (94.9%, 100%)	98.44% (152591/155 016) (98.37%, 98.50%)
	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% (154193/155 016) (99.43%, 99.50%)
					2.15X	10.17	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	98.44% (152591/155 016) (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% (154643/155 088) (99.69%, 99.74%)
					2.76X	9.93	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	99.58% (154432/155 088) (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% (154423/155 016) (99.59%, 99.65%)
					1.84X	8.03	100.0% (72/72) (94.9%, 100%)	100.0% (72/72) (94.9%, 100%)	99.39% (154065/155 016) (99.35%, 99.42%)

Table 43 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	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%)
					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%)

Table 43 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	
48	Level 2	ERBB2 p.G778_P780dup (9 bp)	Insertion	NSCLC	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%)
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.G776delinsVC (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%)

Table 43 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	
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%)
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%)

Table 43 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	
56	Level 2	EML4-ALK .E20A20 .COSF409.2	Fusion	NSCLC	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%)
					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 44 and Table 45, 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 44 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%)
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 45 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%)
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. 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 (for example, 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, and 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-laboratory 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 46), 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 47), 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 46 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
EGFR L858R	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%)

Table 46 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
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%)
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%)

Table 46 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
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%)
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%)
		2.60X	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 47 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%)
		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.M13M15.1	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.M13M15.1	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%)

Table 47 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]
WT01	N/A	N/A	504	0	504	0	252	252	100.0% (98.5%, 100.0%)
WT02	N/A	N/A	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 48. 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 48 Designated hold time test conditions

Step	Control (SOP)	Test condition
Lysate stability	Immediate use	• 24 hours at 4°C • 72 hours at 4°C • 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 of 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), 6 months + 2 weeks (T1), 12 months + 2 weeks (T2), and 18 months + 2 weeks (T3) 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 Purification System and Genexus™ Dx Integrated Sequencer in the integrated purification and sequencing configuration. DNA and RNA quantification was performed to confirm that all samples met the minimum input requirement of 0.5 ng/µL.

All sample replicates achieved a 99.6% pass rate for DNA and 100% pass rate for RNA, with DNA and RNA concentrations meeting the Oncomine™ Dx Express Test minimum input requirement of 0.5 ng/µL at baseline (T0), 6 months + 2 weeks (T1), 12 months + 2 weeks (T2), and 18 months + 2 weeks (T3) time points. For FFPE resection samples, all DNA variants of interest except 1 block sample harboring *EGFR* exon 20 insertions demonstrated <30% drift in AF relative to baseline measurements at all time points. For FFPE resection samples, all RNA variants of interest demonstrated <40% drift in log fusion read counts relative to baseline measurements at all time points. For CNB and FNA cell block samples, concordance analysis confirmed 100% detection of all DNA and RNA variants of interest at T0, T1, and T3 time points. For T2, 27/28 variants were detected, but the missing variant was recovered in T3. These results support the stability of FFPE blocks, curls, and slides for up to 18 months under the specified storage conditions, except for up to 12 months for *EGFR* exon 20 insertion resection samples.

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 Genexus™ Dx FFPE DNA/RNA Purification Combo 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 variant allele frequencies (AFs) of 1–2X limit of detection (LoD) for DNA variants and 1–3X LoD for RNA variants. Extracted nucleic acids were aliquoted into Eppendorf™ 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), 6 months + 1 week (T1), and 12 months + 1 week (T2) 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), 6 months (T3), 12 months (T4), and 13 months (T5) time points. To evaluate freeze/thaw stability, aliquots from tubes were subjected to 3 freeze/thaw cycles before testing at each time point.

The impact of storage conditions and freeze/thaw cycles on sequencing performance was assessed by measuring drift in AF 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 AF 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 12 months when stored in plate formats and up to 11 months when stored in Eppendorf™ LoBind™ tubes, at the recommended temperature.

Reagent stability study

- **Oncomine™ Dx Express Test and Genexus™ Dx Integrated Sequencer and Genexus™ Dx Purification System 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 6 time points: baseline (T0), 4 months (T1), 5 months (T2), 9 months (T3), 12 months (T4), and 14 months (T5). 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 13 months when stored as recommended.

The stability of Genexus™ Dx FFPE DNA/RNA Purification Combo Kit reagents was also evaluated in a real-time stability study. The study evaluated 3 reagent lots at 4 time points: baseline (T0), 7 months (T1), 13 months (T2), and 19 months (T3). Lot 3 stability results at time point T3 are in progress. All reagent lots met predefined acceptance criteria for DNA and RNA yield in both purification PQ samples and clinical FFPE tissue samples at all tested time points. Variants of interest were consistently detected at expected mutation rates across all lots and time points, demonstrating maintained performance. The data support a 12-month shelf life for Genexus™ Dx Purification System reagents 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 studies—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 49.

Table 49 *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 50, 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 50.

Table 50 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, N/E[3])	9.8 months (5.6, N/E)
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
[3] N/E: Not evaluable.

ERBB2/HER2 activating mutations clinical study—clinical accuracy

To evaluate the ability of the Oncomine™ Dx Express Test to identify the *ERBB2/HER2* tyrosine kinase domain (TKD) activating mutations (SNVs and exon 20 insertions) in FFPE non-small cell lung cancer (NSCLC) tumor specimens, 210 samples (80 *ERBB2/HER2* TKD activating mutation-positive subjects (clinical trial assay (CTA)-positive) from participants enrolled in clinical trial SOHO-01 (NCT05099172) and 130 commercially-sourced biomarker-negative samples) were analyzed by comparing results with the Oncomine™ Dx Express Test to results obtained with the enrolling CTAs used in the trial.

Among the 80 *ERBB2/HER2* TKD activating mutation-positive subjects, all 80 were confirmed positive with the Oncomine™ Dx Express Test.

Of the 130 commercially-sourced samples, all 130 were confirmed as *ERBB2/HER2* TKD activating mutation-negative by NGS-based assays. Of these, 129 were confirmed negative and 1 was positive with the Oncomine™ Dx Express Test.

The positive percent agreement (PPA) was defined as the proportion of *ERBB2/HER2* TKD activating mutation-positive specimens as determined by the CTAs that were also *ERBB2/HER2* TKD activating mutation-positive as determined by the Oncomine™ Dx Express Test, and the negative percent agreement (NPA) was defined as the proportion of *ERBB2/HER2* TKD activating mutation-negative specimens as determined by the NGS-based assays (which are representative of the CTAs) that were

also *ERBB2*/*HER2* activating mutation-negative as determined by the Oncomine™ Dx Express Test. The PPA, NPA, and overall percent agreement (OPA) are shown in Table 51.

Table 51 *ERBB2*/*HER2* activating mutations—concordance with CTAs

Agreement measure	Percent agreement	Wilson Score 95% CI
Positive Percent Agreement (PPA)	100.0% (80/80)	(95.4%, 100.0%)
Negative Percent Agreement (NPA)	99.2% (129/130)	(95.8%, 99.9%)
Overall Percent Agreement (OPA)	99.5% (209/210)	(97.4%, 99.9%)

The positive predictive value (PPV) and negative predictive value (NPV) were calculated and adjusted based on the *ERBB2*/*HER2* TKD activating mutation-positive prevalence of 1.8% to account for potential bias due to enrichment. The adjusted PPV was 70.4% (95% CI: 25.3%, 94.4%) and the NPV was 100.0% (95% CI: not estimable).

ERBB2/*HER2* activating mutations clinical study—clinical effectiveness

The efficacy of HYRNUO® (sevabertinib) for treating patients with non-small cell lung cancer (NSCLC) who tested positive for an *ERBB2*/*HER2* tyrosine kinase domain (TKD) activating mutation (SNVs and exon 20 insertions) was evaluated in the SOHO-01 (NCT05099172) clinical trial. The primary efficacy population (Group D) included 70 patients with advanced non-squamous NSCLC with activating mutations in the TKD who had received prior systemic therapy but were naive to therapy targeting activating *ERBB2*/*HER2* mutations.

The clinical effectiveness of the Oncomine™ Dx Express Test in the SOHO-01 clinical trial was evaluated by measuring the objective response rate (ORR; complete response (CR) or partial response (PR)) for patients with NSCLC that tested positive for *ERBB2*/*HER2* TKD activating mutations. The objective response rate (ORR; based on RECIST version 1.1, as assessed by blinded independent central review) was calculated for patients selected for treatment with HYRNUO®.

The ORR in the clinical trial assay (CTA)-positive population was 71.4% (50/70; 95%CI: 59.4%, 81.6%), with 2.9% (2/70) of patients having a CR and 68.6% (48/70) having a PR. Among these, for FFPE samples, 46 patients were both CTA-positive and tested positive for *ERBB2*/*HER2* TKD activating mutations using the Oncomine™ Dx Express Test (ODxET+ | CTA+). In this subgroup (N=46), the ORR was 80.4% (37/46; 95%CI: 66.1%, 90.6%), with all the responses being PRs. The ORRs for the Oncomine™ Dx Express Test *ERBB2*/*HER2* TKD activating mutation-positive population align with those observed in the CTA *ERBB2*/*HER2* TKD activating mutation-positive population (Table 52).

The median duration of response (DoR) among the 71.4% (50/70) responders from the CTA-positive population was 9.2 months (95% CI: 6.3, 15.0), with 54.0% (27/50) of patients having a DoR ≥6 months. The median DoR among the 80.4% (37/46) responders from the ODxET + | CTA + population with the FFPE samples was 10.9 months (95% CI: 6.3, 15.9), with 59.5% (22/37) of patients having a DoR ≥6 months. The median DoR was based on Kaplan-Meier estimate using confirmed responses.

Efficacy was also evaluated in 52 patients with advanced non-squamous NSCLC with activating *ERBB2*/*HER2* mutations in the TKD who had received prior systemic therapy and had documented progression with *ERBB2*/*HER2*-targeted antibody-drug conjugates (Group E). The ORR in the clinical trial assay (CTA)-positive population was 38.5% (20/52; 95%CI: 25.3%, 53.0%), with 5.8% (3/52) of patients having a CR and 32.7% (17/52) having a PR. Among these, for FFPE samples, 34 patients were both CTA-positive and tested positive for *ERBB2*/*HER2* activating mutations using the Oncomine™ Dx Express Test (ODxET + | CTA +). In this subgroup (N=34), the ORR was 41.2% (14/34; 95%CI: 24.6%, 59.3%), with 2.9% (1/34) of patients having a CR and 38.2% (13/34) having a PR. The ORRs for the Oncomine™ Dx Express Test *ERBB2*/*HER2* TKD activating mutation-positive population align with those observed in the CTA *ERBB2*/*HER2* TKD activating mutation-positive population (Table 52)

Table 52 Efficacy results of the SOHO-01 study—FFPE samples

Efficacy parameter	Group D		Group E	
	HYRNUO® (N=70, CTA +)	ODxET + | CTA + (N=46)	HYRNUO® (N=52, CTA +)	ODxET + | CTA + (N=34)
Objective Response Rate (ORR), % (95% CI)[1]	71.4 (59.4, 81.6)	80.4 (66.1, 90.6)	38.5 (25.3, 53.0)	41.2 (24.6, 59.3)

[1] 95% CI for ORR was calculated based on Clopper-Pearson exact CI method. CI: confidence interval

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
*AR*AF	7	p.S214T	c.640T>A	COSM1558277
*AR*AF	7	p.S214A	c.640T>G	COSM1742787
*AR*AF	7	p.S214C	c.641C>G	COSM5044705
*AR*AF	7	p.S214Y	c.641C>A	COSM5416443
*AR*AF	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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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 **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_2257delGAATTAAGAGAAGCAACATCTCinsATCT	COSM133191
EGFR	19	p.E746_P753delinsLS	c.2236_2257delGAATTAAGAGAAGCAACATCTCinsCTCT	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_2251delAATTAAGAGAAGCAAinsCACCAT	COSM6924852
EGFR	19	p.E746_T751delinsVP	c.2237_2251delAATTAAGAGAAGCAAinsTCC	COSM6968288
EGFR	19	p.E746_T751delinsVA	c.2237_2251delAATTAAGAGAAGCAAinsTGG	COSM53205
EGFR	19	p.E746_T751delinsVP	c.2237_2251delAATTAAGAGAAGCAAinsTTC	COSM18421
EGFR	19	p.E746_T751delinsVS	c.2237_2251delAATTAAGAGAAGCAAinsTTT	COSM6980200
EGFR	19	p.E746_T751delinsV	c.2237_2252delAATTAAGAGAAGCAACinsT	COSM12386
EGFR	19	p.E746_T751delinsV	c.2237_2253delAATTAAGAGAAGCAACAinsTC	COSM133193
EGFR	19	p.E746_T751delinsVP	c.2237_2253delAATTAAGAGAAGCAACAinsTTCCT	COSM52935
EGFR	19	p.E746_T751delinsVA	c.2237_2253delAATTAAGAGAAGCAACAinsTTGCT	COSM12416
EGFR	19	p.E746_S752delinsA	c.2237_2254delAATTAAGAGAAGCAACAT	COSM12367
EGFR	19	p.E746_S752delinsV	c.2237_2255delAATTAAGAGAAGCAACATCinsT	COSM12384
EGFR	19	p.E746_S752delinsV	c.2237_2256delAATTAAGAGAAGCAACATCTinsTC	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 **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

(continued)

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 **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

(continued)

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.A763_Y764insFQEA	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.V769_D770insGVV	c.2308_2309insGGGTTGTGG	OMINDEL1187
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 **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.D770_N771delinsAGH	c.2309_2311delACAinsCTGGCC	MATNV09

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.D770_N771delinsAGG	c.2309_2312delACAAinsCTGGTGG	COSM12737
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.N771delinsKPH	c.2312_2313insACCCCA	OMINDEL1188
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.N771_P772insHN	c.2314_2315insACAACC	OMINDEL1084
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.P772_H773insTRNP	c.2316_2317insACCAGGAACCCC	OMINDEL1116
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.P772_H773insPHP	c.2317_2318insCCCACCCCC	OMINDEL1136
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 **Note:** The nucleotide change of COSM12380 overlaps that of MAN123, so a positive COSM12380 sample also results in a positive call for MAN123.

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.H773_V774insPHPH	c.2319_2320insCCCCACCCCCAC	p.H773_V774insPHPH_2
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
EGFR	20	p.G796A	c.2387G>C	COSM27568
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
ERBB2	8	p.S310Y	c.929_930delCCinsAT	COSM6906440
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.A775_G776insYLMA	c.2324_2325insATACCTGATGGC	SPRNV02

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
ERBB2	20	p.Y772_A775dup	c.2324_2325insATACGTGATGGC	COSM20959
ERBB2	20	p.A775_G776insYVMS	c.2324_2325insATACGTGATGTC	OMINDEL824
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_2326delGinsCTGT	SPRNV03
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.A775_G776insVVMA	c.2326_2327insTTGTGATGGCTG	SPRNV01
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
ERBB2	20	p.G776R	c.2326G>C	OM3421
ERBB2	20	p.G776C	c.2326G>T	COSM303938
ERBB2	20	p.G776delinsVV	c.2327_2327delGinsTTGT	COSM6977433
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
ERBB2	21	p.L869R	c.2606T>G	COSM249793
ERBB2	22	p.R896C	c.2686C>T	COSM14066
ERBB2	22	p.R896H	c.2687G>A	COSM119971
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
ESR1	9	p.L536Q	c.1607_1608delTCinsAG	COSM4766050
ESR1	9	p.L536R	c.1607T>G	COSM4774826
ESR1	9	p.Y537N	c.1609T>A	COSM1074635
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
FGFR2	12	p.N549H	c.1645A>C	COSM250083
FGFR2	12	p.N549S	c.1646A>G	COSM3665553
FGFR2	12	p.N549K	c.1647T>G	COSM36902
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
FGFR3	9	p.Y373C	c.1118A>G	COSM718
FGFR3	9	p.G380R	c.1138G>A	COSM24842
FGFR3	9	p.G380E	c.1139G>A	COSM51545
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
FLT3	20	p.D835N	c.2503G>A	COSM789
FLT3	20	p.D835del	c.2503_2505delGAT	COSM854
FLT3	20	p.D835K	c.2503_2505delGATinsAAG	OM3570
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
GNAS	9	p.Q227R	c.680A>G	COSM27896
GNAS	9	p.Q227H	c.681G>T	COSM27900
GNAS	9	p.Q227H	c.681G>C	COSM4168960
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
IDH1	4	p.R132V	c.394_395delCGinsGT	COSM28751
IDH1	4	p.R132C	c.394C>T	COSM28747
IDH1	4	p.R132G	c.394C>G	COSM28749
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
KEAP1	3	p.?	c.640-1G>T	OM3725
KEAP1	3	p.?	c.640-1G>C	OM3726
KEAP1	3	p.?	c.640-2A>C	OM3727
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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 **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 **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 **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 **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	p.?	c.1648-3_1671delCAGAAACCCATGTATGAAGTACAGTGG	OMINDEL901 **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 **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 **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 **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 **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 **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 **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 **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 **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 **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 **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 **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 **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.

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
KIT	11	p.M552_E561delinsK	c.1655_1681delTGTATGAAGTACAGTGGAAGGTTGTTG	COSM96880 **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 **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 **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 **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

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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 **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 **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 **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 **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 **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

(continued)

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 **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 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
KIT	11	p.W557_V560del	c.1669_1680delTGGAAGGTTGTT	COSM1223 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
KIT	11	p.K558_V560delinsL	c.1672_1680delAAGGTTGTTinsTTG	COSM6940807 **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

(continued)

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 **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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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 **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 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.2942-15_2946delTCTCTCTGTTTTAAGATCTG	COSM6948341 **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 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.2942-20_2942-3delTTCTTTCTCTCTGTTTTA	OMINDEL676 **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 **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 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.2942-14_2944delCTCTCTGTTTTAAGATC	OMINDEL681 **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

(continued)

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 **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 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.3075_3082+9delinsGGTATATTTT	OMINDEL687 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.3081_3082+4delAGGTAT	OMINDEL670 **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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
MET	14	p.?	c.3082_3082+20delGGTATATTTCAGTTTATTGTT	OMINDEL715 **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 **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

(continued)

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

(continued)

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

(continued)

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

(continued)

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 **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 **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

(continued)

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 **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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
RET	11	p.D627_L633delinsAH	c.1880_1899delinsCTCAC	OMINDEL1170 **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 **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

(continued)

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

(continued)

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

(continued)

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.D53GfsTer110	c.157_158insG	COSM391251
STK11	1	p.L67P	c.200T>C	OM3700
STK11	1	p.?	c.289_290+1delinsGAT	COSM6922318
STK11	1	p.?	c.289_290+2delAAGT	OMINDEL1000
STK11	1	p.?	c.290+1_290+2delGT	OMINDEL1001
STK11	1	p.?	c.290+1G>A	COSM6350392
STK11	1	p.?	c.290+1G>C	OM3701
STK11	1	p.?	c.290+1G>T	OM3702
STK11	1	p.?	c.290+2delT	OMINDEL1002
STK11	1	p.?	c.290+2T>A	COSM6925006
STK11	1	p.?	c.290+2T>G	COSM25857
STK11	2	p.?	c.291-1_291delinsTT	OM3705
STK11	2	p.?	c.291-1delG	OMINDEL1004
STK11	2	p.?	c.291-1G>A	OM3703
STK11	2	p.?	c.291-1G>C	OM3704
STK11	2	p.?	c.291-1G>T	COSM6083779

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
STK11	2	p.?	c.291-2A>G	COSM5575722
STK11	2	p.?	c.291-2A>T	COSM49010
STK11	3	p.?	c.375-1C>G	COSM4074282
STK11	3	p.?	c.375-1C>T	COSM6977173
STK11	3	p.?	c.375-2_386delACGTATATGGTGAT	OMINDEL1006
STK11	3	p.?	c.375-2A>C	OM3706
STK11	3	p.?	c.375-2A>G	OM3707
STK11	3	p.?	c.375-2A>T	COSM6967966
STK11	3	p.?	c.375-6_375-2delCCCCA	OMINDEL1005
STK11	4	p.?	c.465-1_465delinsTT	COSM6928792
STK11	4	p.?	c.465-1delG	OMINDEL1010
STK11	4	p.?	c.465-1G>A	COSM25855
STK11	4	p.?	c.465-1G>C	OM3709
STK11	4	p.?	c.465-1G>T	COSM21570
STK11	4	p.?	c.465-2_465-1delAG	OMINDEL1009
STK11	4	p.?	c.465-2_465-1delinsTC	OM3708
STK11	4	p.?	c.465-2A>C	COSM6933102
STK11	4	p.?	c.465-2A>G	COSM21387
STK11	4	p.?	c.465-2A>T	COSM1523962
STK11	4	p.?	c.465-4_472delGCAGGTACTTCT	OMINDEL1007
STK11	4	p.?	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
STK11	4	p.D194E	c.582C>A	OM3710
STK11	4	p.D194E	c.582C>G	OM3711

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
STK11	5	p.?	c.598-1_598delinsTT	OM3713
STK11	5	p.?	c.598-1_612delGGCACTGCACCCGTTC	OMINDEL1014
STK11	5	p.?	c.598-1delG	OMINDEL1013
STK11	5	p.?	c.598-1G>A	COSM3724562
STK11	5	p.?	c.598-1G>T	COSM51521
STK11	5	p.?	c.598-2_600delAGGCA	OMINDEL1012
STK11	5	p.?	c.598-2A>C	OM3712
STK11	5	p.?	c.598-2A>G	COSM6923168
STK11	5	p.?	c.598-2A>T	COSM25858
STK11	5	p.A205T	c.613G>A	COSM20953
STK11	5	p.S240W	c.719C>G	OM3714
STK11	5	p.?	c.727_734+9delGTCACCCTGTAAGTGCC	OMINDEL1015
STK11	5	p.?	c.731_734+9delCCCTGTAAGTGCC	OMINDEL1016
STK11	5	p.?	c.734+1G>A	COSM6916684
STK11	5	p.?	c.734+1G>C	COSM6970016
STK11	5	p.?	c.734+1G>T	COSM51523
STK11	5	p.?	c.734+2_734+10delTAAGTGCCC	OMINDEL1017
STK11	5	p.?	c.734+2T>A	OM3715
STK11	5	p.?	c.734+2T>G	COSM6961577
STK11	6	p.?	c.735-1G>A	COSM5732040
STK11	6	p.?	c.735-1G>T	COSM51522
STK11	6	p.?	c.735-2A>G	OM3716
STK11	6	p.?	c.735-2A>T	COSM25856
STK11	6	p.?	c.850_862+4delGACCTGCTGAAAGGTGG	OMINDEL1018
STK11	6	p.?	c.862+1G>A	COSM6929076
STK11	6	p.?	c.862+1G>C	OM3717
STK11	6	p.?	c.862+1G>T	COSM6149637

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
STK11	6	p.?	c.862+2T>A	COSM5576087
STK11	6	p.?	c.862+2T>G	OM3718
STK11	6	p.?	c.862+3delG	COSM8554973
STK11	7	p.?	c.863-1G>A	COSM6916725
STK11	7	p.?	c.863-1G>T	COSM6925330
STK11	7	p.?	c.863-2A>C	COSM6912582
STK11	7	p.?	c.863-2A>T	COSM401786
STK11	7	p.L290P	c.869T>C	OM3719
STK11	7	p.R304W	c.910C>T	COSM29468
STK11	7	p.?	c.918_920+11delCAGGTGAGCGGCCC	OMINDEL1019
STK11	7	p.?	c.920+1G>A	OM3720
STK11	7	p.?	c.920+1G>C	COSM4412472
STK11	7	p.?	c.920+1G>T	COSM6923458
STK11	7	p.?	c.920+2T>A	OM3721
STK11	8	p.?	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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

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Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

Gene	Exon	Amino acid change	Nucleotide change	Variant ID
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
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

(continued)

***ALK* fusion isoforms (N=213)**		
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
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

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***ALK* fusion isoforms (N=213)**		
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

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***BRAF* fusion isoforms (N=187)**		
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
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

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***BRAF* fusion isoforms (N=187)**		
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
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
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
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
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
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

(continued)

***NTRK1* fusion isoforms (N=89)**		
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
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
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

(continued)

***RET* fusion isoforms (N=89)**		
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

(continued)

***ROS1* fusion isoforms (N=57)**		
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 6.8 Reference User Guide* (MAN1001776).

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.A27A28
		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.N4N5	NTRK2.I.N13N14
	NTRK2.I.N6N7	NTRK2.I.N17N18
	NTRK2.I.N8N9	NTRK2.I.N19N20
		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 53) are prospectively masked in reporting.

Table 53 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

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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 MAN1001775

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 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

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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: MAN1001775 A (English)

Revision	Date	Description
A	3 August 2026	New IVD user guide for the Oncomine™ Dx Express Test 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 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.

Prerequisites

Category	Prerequisites
Instrument	• Genexus™ Dx Integrated Sequencer. • Reagents and supplies for operating the Genexus™ Dx Integrated Sequencer.
Software	• Genexus™ Dx Software 6.8.2.1 or later.

(continued)

Category	Prerequisites
Functional knowledge and understanding - all users	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to administrator credentials, SSH private keys and the associated SSH key passwords. • Main functions of the Genexus™ Dx Integrated Sequencer. *(Optional)* Main functions the Genexus™ Purification System. • Main features of the Genexus™ Dx Software.
Functional knowledge and understanding - system administrators	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to administrator credentials, SSH private keys and the associated SSH key passwords. • Use of security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

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 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. MAN1001774).

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. MAN1001774).

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. MAN1001774).

Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software version 6.8.2.1 or later. The operating system of the sequencer is Ubuntu™ 18.04 LTS. The operating system has an Ubuntu™ Pro license that includes expanded security maintenance coverage.

The Genexus™ Dx Integrated Sequencer and Genexus™ Dx Purification System have system firewalls enabled by default. Only ports required for the operation of the instruments and software are open.

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. MAN1001776).

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. Items 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) for FFPE samples.

Contents	Carrier color	Amount	Storage
Oncomine™ Dx Express Test Panel (Part No. A50871)			
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 is ordered separately.

Control	Cap color	Part No.	Sets provided	Amount/set	Storage
Oncomine™ Dx Express Test FFPE DNA and RNA Control (Cat. No. A52167)					
Oncomine™ Dx Express Test FFPE DNA Control	Brown	A50834	1	4 × 30 µL	–30°C to –10°C
Oncomine™ Dx Express Test FFPE RNA Control	White	A50835	1	4 × 30 µL	–90°C to –60°C
Oncomine™ Dx Express Test NTC Control	Purple	A50836	2	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. Items 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. A54106) are ordered as a set that contains two chips and two couplers, sufficient for up to eight 1-lane or two 4-lane 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 216.

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 216.

Component	Amount	Storage
Genexus™ Dx Filter	2 filters	15°C to 30°C

Genexus™ Dx Integrated Sequencer

Components	Cat. No.
Genexus™ Dx Integrated Sequencer	A53256
Genexus™ Dx Integrated Sequencer Install Kit	A52169

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 Thermo Scientific™ M10 Microplate Swinging Bucket Rotor (or equivalent), and Sealed Bucket; Capacity: 4 Standard or 2 Midi-Deepwell plates (Set of 2) (or equivalent)	75007200 75005706 75005721
Handheld Barcode Scanner[2] (Zebra Technologies Corp. DS4608)	4488442
Genexus™ Primer Pool Tube Holder	A40004840
Vortex mixer with a rubber platform	**MLS**
Uninterruptible Power Supply (UPS)[3]	**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
MicroAmp™ Adhesive Film Applicator (recommended)	4333183 or **MLS**
2-, 20-, 200-, and 1,000-µL pipettors and appropriate aerosol-resistant filtered pipette tips	**MLS**
Eppendorf™ DNA LoBind™ 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
Gloves, powder-free nitrile	**MLS**
Ice buckets and ice	—

(continued)

Item	Source
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] The Handheld Barcode Scanner can be used to scan extraction and control kit package barcodes during run planning.

[3] To protect against power outages and line voltage fluctuations, we require that you use an at least 2.0-kVA dual-conversion uninterruptible power supply (UPS) with a power rating of 1800W or greater per instrument. Additional battery pack capacity may be required, depending on how much hold up time is required. Power failures and other events that abruptly terminate the function of the instrument and computer can corrupt data and possibly damage the system.

Additional materials

The following Genexus™ Dx Purification System reagents, supplies, and equipment are used in the Sample to Result run workflow for the Oncomine™ Dx Express Test. The Sample to Result workflow is optional and not a required workflow for FFPE samples.

The items are ordered separately.

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. The sample purification kits are required for integrated Sample to Result runs only.

Genexus™ Dx FFPE DNA/RNA Purification Combo Kit

The Ion Torrent™ Genexus™ Dx FFPE DNA/RNA Purification Combo 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	

(continued)

Component	Amount	Storage
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	
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	
Genexus™ Dx AutoLys Tubes	2 × 24 each	
Genexus™ Dx AutoLys Caps	2 × 24 each	

[1] Store the Genexus™ Dx Quantitation Plate in the dark to help prevent photobleaching of the preloaded reagents.
[2] Can be stored at 15°C to 30°C on receipt.

Genexus™ Dx Purification System

Component	Cat. No.
Genexus™ Dx Purification System instrument	A53257

Also shipped with the Genexus™ Dx Purification System is an accessory kit for using Genexus™ Dx AutoLys Tubes. The kit is reorderable through Thermo Fisher Scientific customer service, if needed.

Component	Cat. No.
Genexus™ Dx AutoLys Accessories Kit • Genexus™ Dx AutoLys Tube Rack (1 set of 5 racks provided); Part No. A58869 • Genexus™ Dx AutoLys Pliers (1 provided); Part No. A58870 • Genexus™ Dx AutoLys Locking Lid (2 provided); Part No. A58871	A58873

Additional materials and equipment for the Genexus™ Dx Purification System 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.

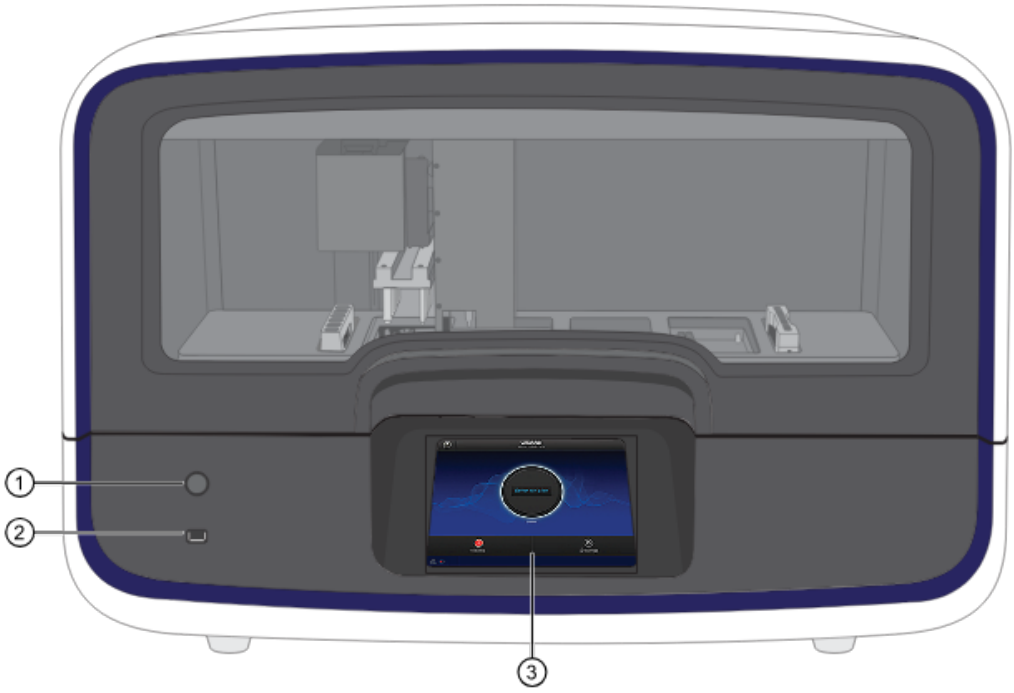
Item	Source
Equipment and consumables for AutoLys FFPE sample extraction[1]	
Economy Standard Incubator (2 incubators needed for 60°C and 90°C temperatures), or equivalent	S50441A **fisherscientific.com**
PCR cabinet or laminar flow hood (recommended)	**MLS**
Genexus™ Dx AutoLys Tube Lifter[2] (optional)	A58872
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**
Fisherbrand™ High Precision Straight Medium Point General Purpose Tweezers/Forceps, or equivalent, for handling FFPE curls	12-000-121
Other equipment	
Uninterruptible Power Supply (UPS)[3]	**MLS**

[1] For use with the Genexus™ FFPE DNA and RNA Purification kit.

[2] Orderable through Thermo Fisher Scientific customer service.

[3] To protect against power outages and line voltage fluctuations, we require that you use an at least 1.5-kVA dual-conversion uninterruptible power supply (UPS) with a power rating of 800W or greater per instrument. Additional battery pack capacity may be required, depending on how much hold up time is required. Power failures and other events that abruptly terminate the function of the instrument and computer can corrupt data and possibly damage the system.

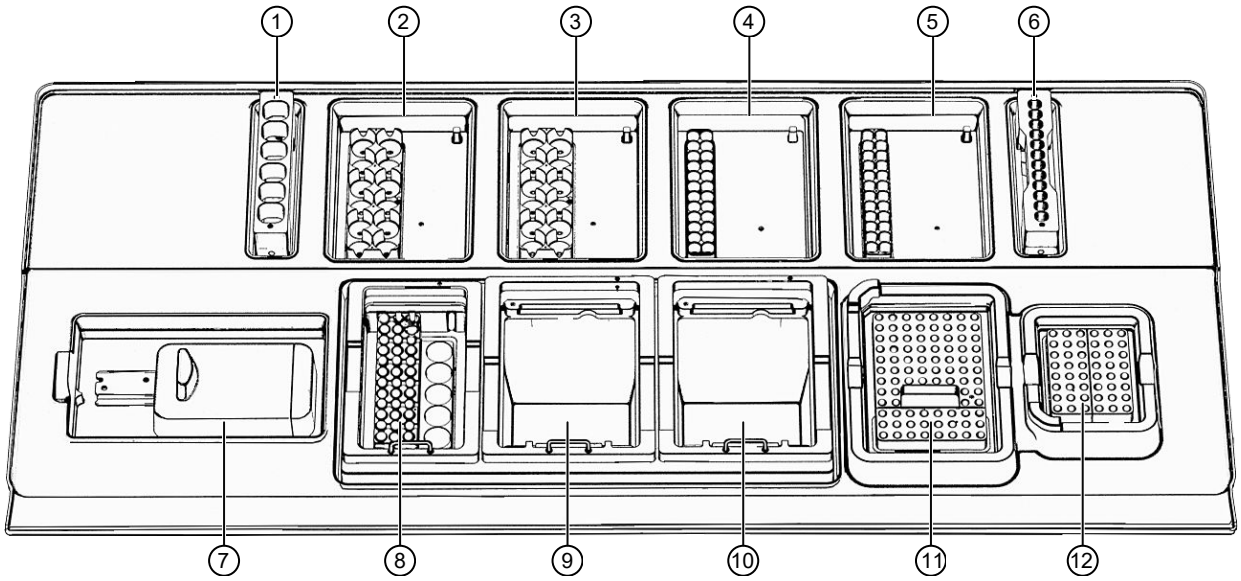
Genexus™ Dx Purification System features



External features of the Genexus™ Dx Purification System.

① Power button
② USB port
③ Touchscreen

Genexus™ Dx Purification System deck stations

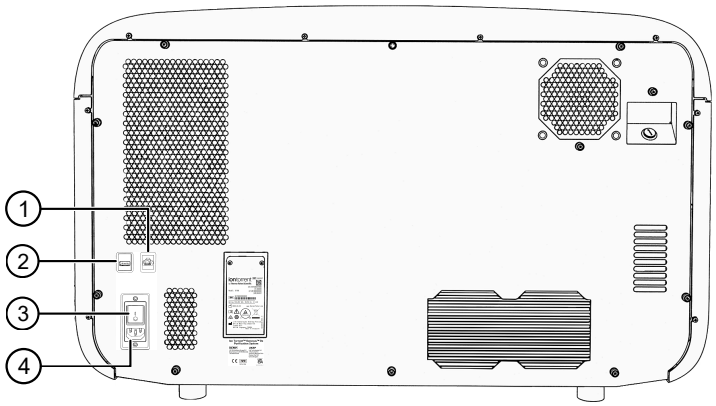


Interior deck stations of the Genexus™ Dx Purification System.

1. 6-well tip comb station
2. 24 deepwell plate station 1
3. 24 deepwell plate station 2
4. 96 deepwell plate station1
5. 96 deepwell plate station 2
6. 12-well tip comb station
7. Quantitation module
8. Quantitation tray
9. Tip box station 1
10. Tip box station 2
11. 96-well output plate station
12. 48-well archive plate station

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.



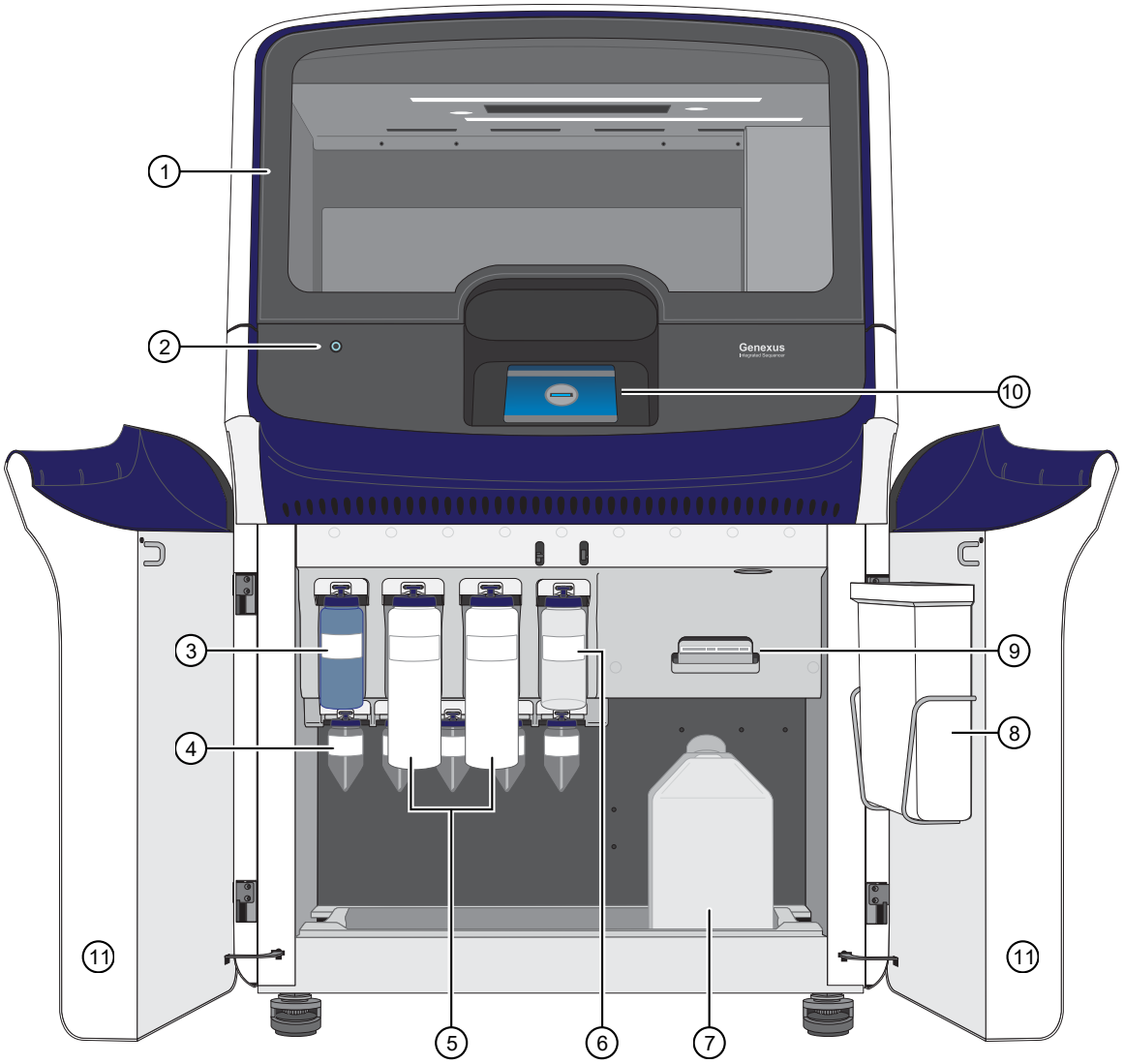
① Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the instrument and a local area network.

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

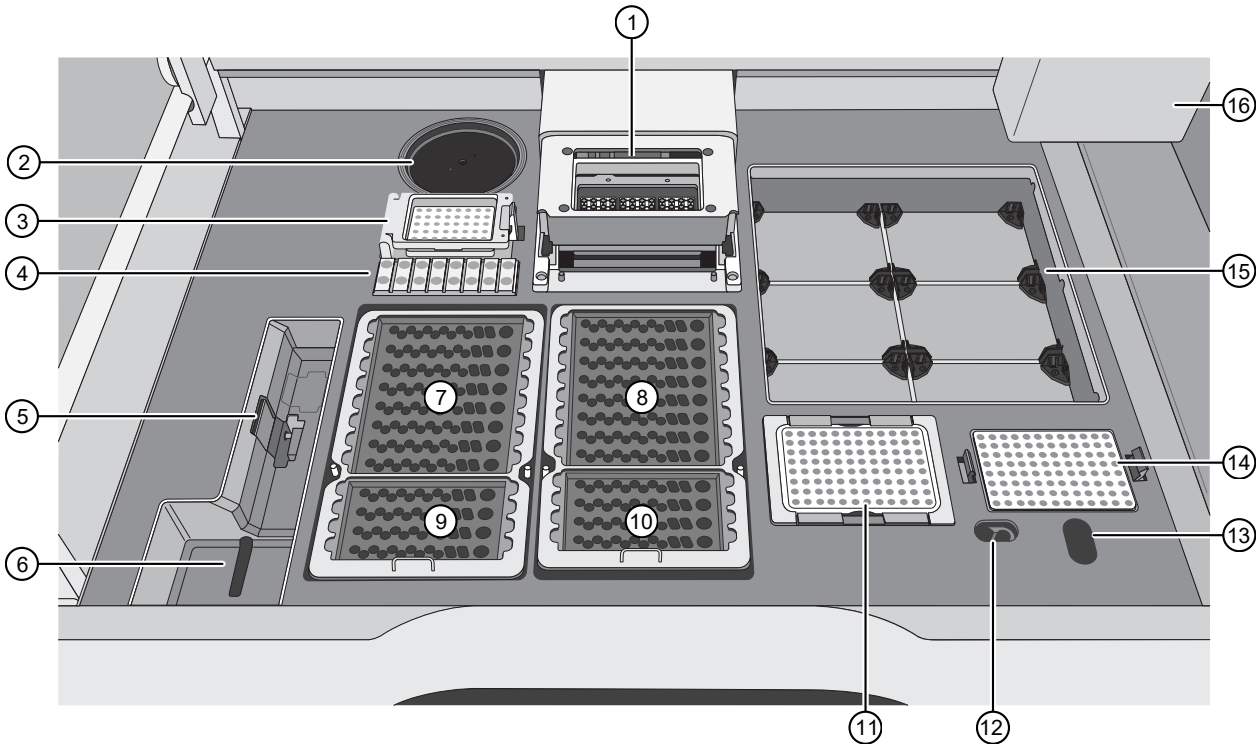


Major features and components of the exterior and sequencing reagents bay of the Genexus™ Dx Integrated Sequencer

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

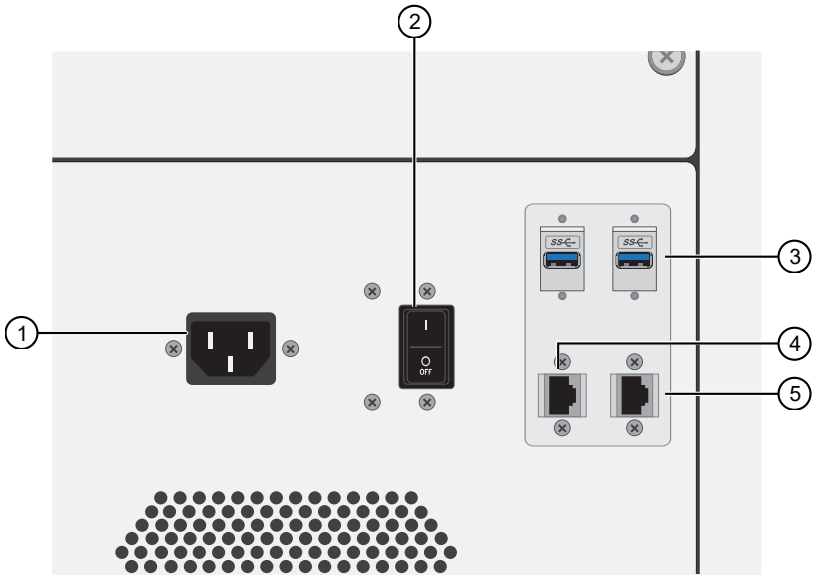


Interior Genexus™ Dx Integrated Sequencer deck components and stations

1. PCR amplification station
2. Microcentrifuge (not used)
3. Genexus™ Dx Barcodes 1–32 HD station
4. 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.

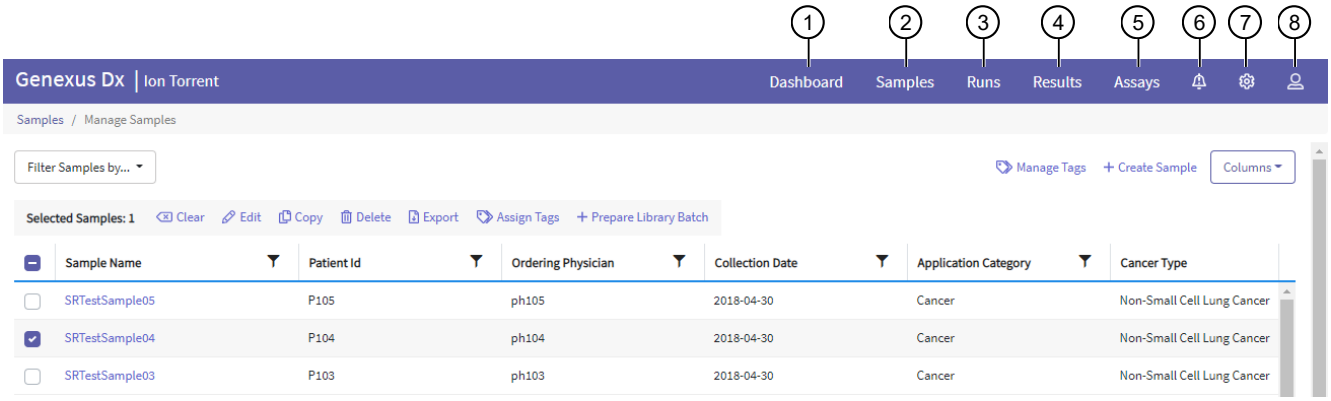


① Power port—100–240VAC port that provides power to the instrument.
② On/off switch—Power switch, where the states are on (|) or off (O).
③ USB ports—Connects a USB device to the instrument.
④ Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and a local area network.
⑤ Ethernet port—An RJ45 port that provides Ethernet (Gigabit) communication between the sequencer and an accessory instrument 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. MAN1001776), 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 38)

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 48)

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 73)

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 97)

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 103)

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 120)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



Review results in Genexus™ Dx Software (page 122)

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 38)

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 58)

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 99)

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 103)

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 120)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



Review results in Genexus™ Dx Software (page 122)

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 28
- Sample storage and stability 29
- Procedural guidelines 29
- Power the Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer on or off 35
- Getting started in Genexus™ Dx Software 35
- Network and password security requirements 36
- Installation and verification run 37

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 Genexus™ Dx AutoLys Tubes” on page 76 or “Prepare FFPE slide sections with Genexus™ Dx AutoLys Tubes” on page 78.

Sample storage and stability

For resection samples, FFPE blocks and slides can be stored at room temperature (15–30°C) for 18 months.

- For fine-needle aspirates (FNAs), FFPE blocks and slides can be stored at room temperature for up to 18 months.
- For core needle biopsies (CNBs), blocks and slides can be stored at room temperature for up to 18 months.
- FFPE curls can be stored at 2–8°C for up to 18 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 12 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 <40%, 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, and *ERBB2* SNV and 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 13 and "Additional materials" on page 16.
- 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 thawed 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 Genexus™ Dx Strip 2-HD and tube 5 of Genexus™ Dx Strip 3-GX5™ 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 105.

- 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 105. Alternatively, use the strip volume guide images provided on page 227 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.

  Note: Reinspect Genexus™ Dx Strip 2-HD before loading and dissolve if needed—reagent in tube 3 can reprecipitate on ice.

- 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 at least 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. **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, the sequencer performs a post-chip clean, and you are asked to remove the partially used chip. 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, for example up to 28 days. 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. MAN1001776), or in the software help system.

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 facility system administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions. The Genexus™ Dx Integrated Sequencer has a firewall enabled by default. Only ports required for the operation of the instrument and software are open.

CAUTION! Data can be lost or corrupted if you operate a sequencer that does not have sufficient security controls in place.

If external or network drives are connected to Genexus™ Dx Integrated Sequencer for archiving or backups, it is the sole responsibility of 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 Genexus™ Dx Software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN1001776).

Data backup and data storage

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.

Note: A paid data migration service is available if needed. Contact your Thermo Fisher Scientific representative for more information.

For more information about data storage compatibility, contact your local support team.

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.

Antivirus software

For recommendations for antivirus software that is compatible with Genexus™ Dx Software 6.8.2.1, see the current version of the Genexus™ Dx Software release notes.

Installation and verification run

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.

3 Enter samples and prepare library batches in Genexus™ Dx Software

- Enter or import samples 38
- Prepare or import a library batch 42
- No-template and positive controls 47

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.

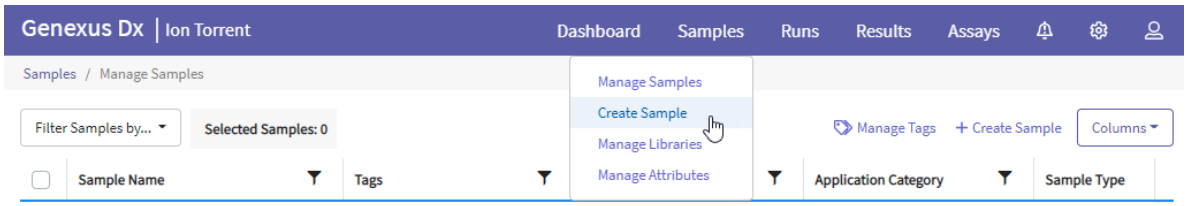
Before entering samples or preparing a library batch, you must sign in to Genexus™ Dx Software in IVD mode.

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 39 or “Import samples” on page 40.

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. MAN1001776), 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 40.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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. MAN1001776), 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 41. b. After editing the file, proceed to the second option.
Upload the sample data from an existing file.	a. Click **Browse**. b. Navigate to the file, then click **Open**. 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.

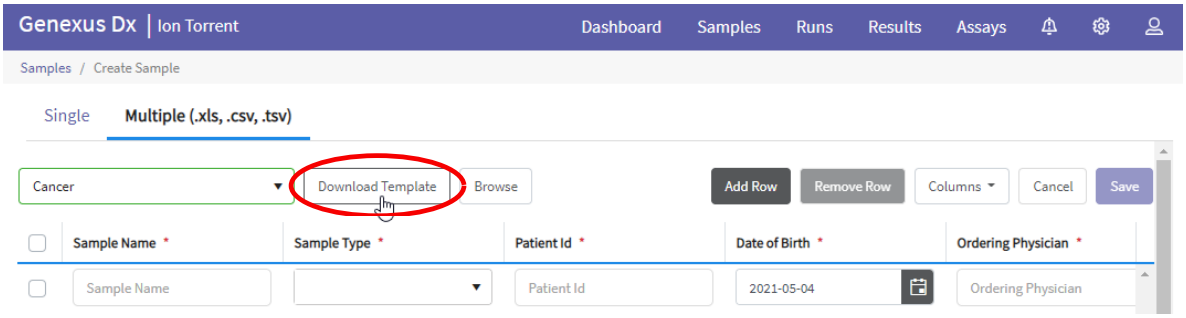
Template files contain two tabs. The **Instruction** tab in the spreadsheet lists and indicates required and optional attributes, which are the column headings in the **Specimen Format** tab. Use the **Specimen Format** tab to enter samples.

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 38.

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 50.

Note:

- Each library in a library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

Prepare a library batch

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 50.

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**.

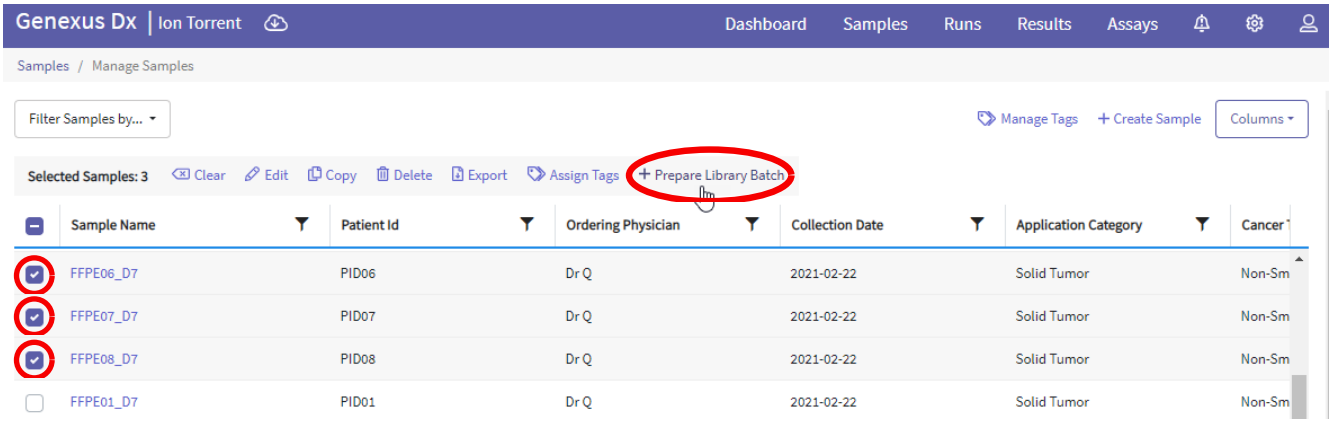
2. In the **Manage Samples** screen, in the **Filter Samples by** dropdown list, apply the **To Be Prepared** filter to limit the displayed samples to those samples that have not been placed in a library batch.

   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.

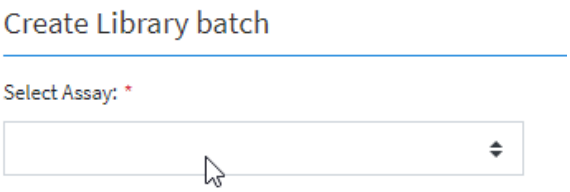
   For more information about how to filter the list of samples, see the software help system, or the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN1001776).

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.



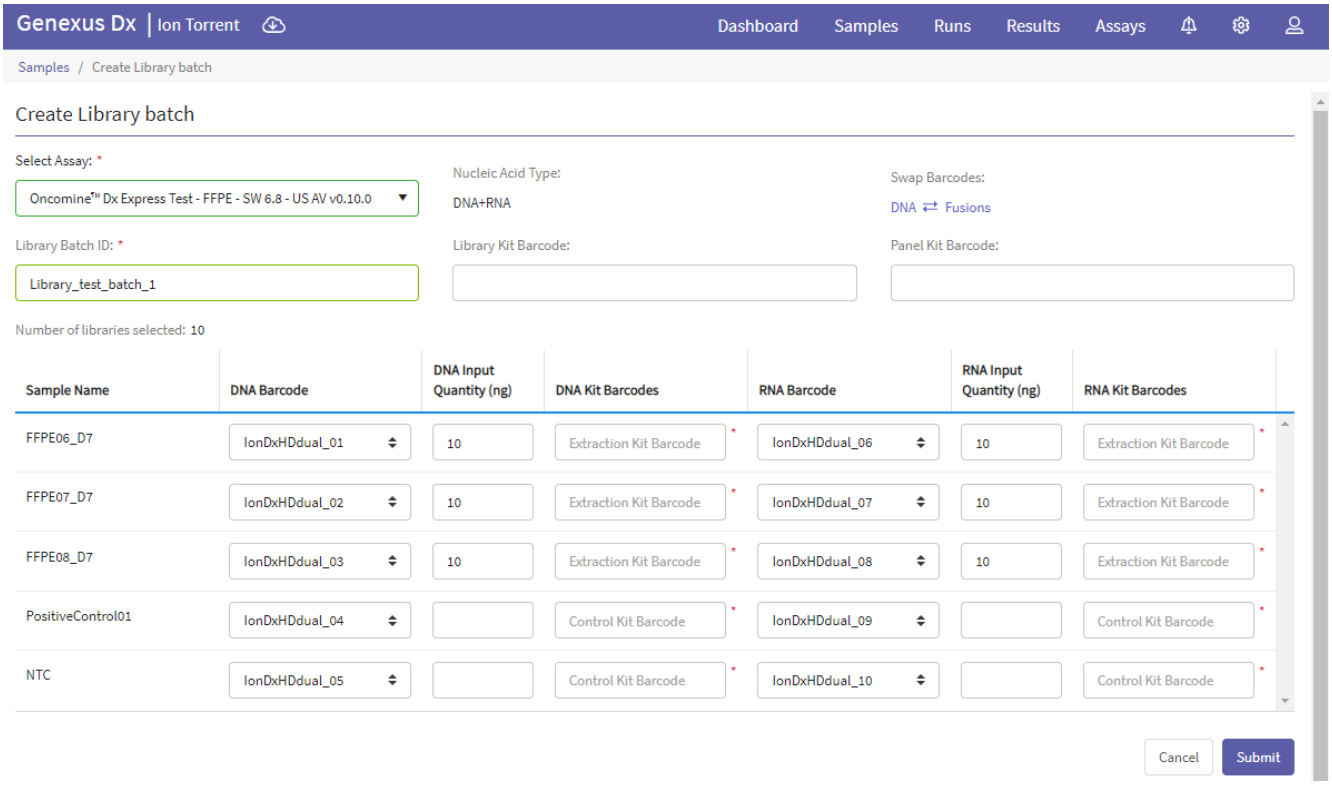
The assay determines parameter selections for the run, including any required controls and post-run data analysis settings. If you select an assay that includes one library prep type, the library preparation type automatically fills with the nucleic acid type specified by the assay you selected, for example DNA+RNA.

Note: If the cancer type of a sample in the library batch, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type. You can edit the cancer type of a sample, if needed. In the **Samples** ▸ **Manage Samples** screen, place the pointer over the row of the sample of interest, then click **Edit**. In the **Edit Sample** screen, change the cancer type.

---

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.

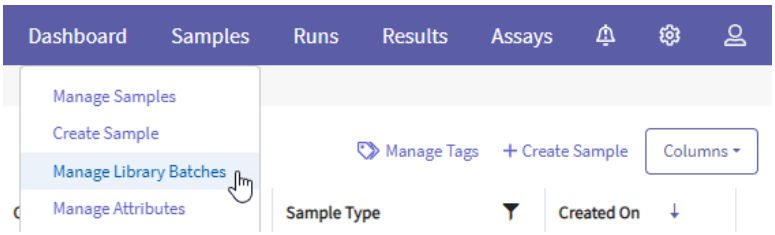
10. Click **Submit** to save and submit your selections and entries.

    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.

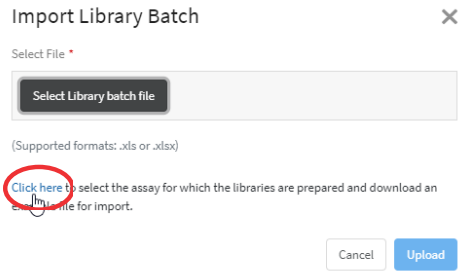
1. In the menu bar, click **Samples** ▸ **Manage Library Batches**.



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 182.

No-template and positive controls

A no-template control (NTC) and positive controls are automatically added to the run plan in Genexus™ Dx Software and their positions on the sample plate are determined by the number of samples selected. In a Library to Result run, the library batch includes the NTC and positive controls. Therefore, the operator does not need to add additional controls to the run plan or sample plate.

4 Plan a run in Genexus™ Dx Software

- Before you plan a run 49
- Mode switch with a partially used chip installed 49
- Integrated nucleic acid isolation in Sample to Result runs 49
- Plan a Sample to Result run 50
- Plan a Nucleic Acid to Result run 58
- Plan a Library to Result run 64
- Plan a Genexus™ Dx FFPE DNA and RNA Purification run—standalone configuration 69

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 223.

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 38.
- 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 42.

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.

Note: A mode switch cannot proceed if a run is loaded on the instrument, even if the run is complete.

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.

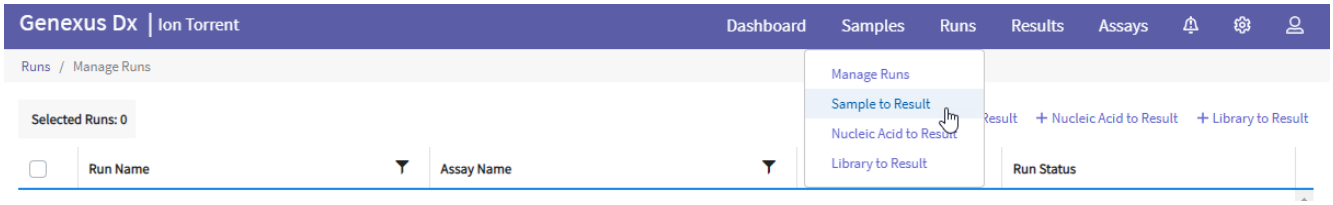
Ensure that the following prerequisites are complete before you plan a **Sample to Result** run.

- 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 39, or “Import samples” on page 40.
- 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.

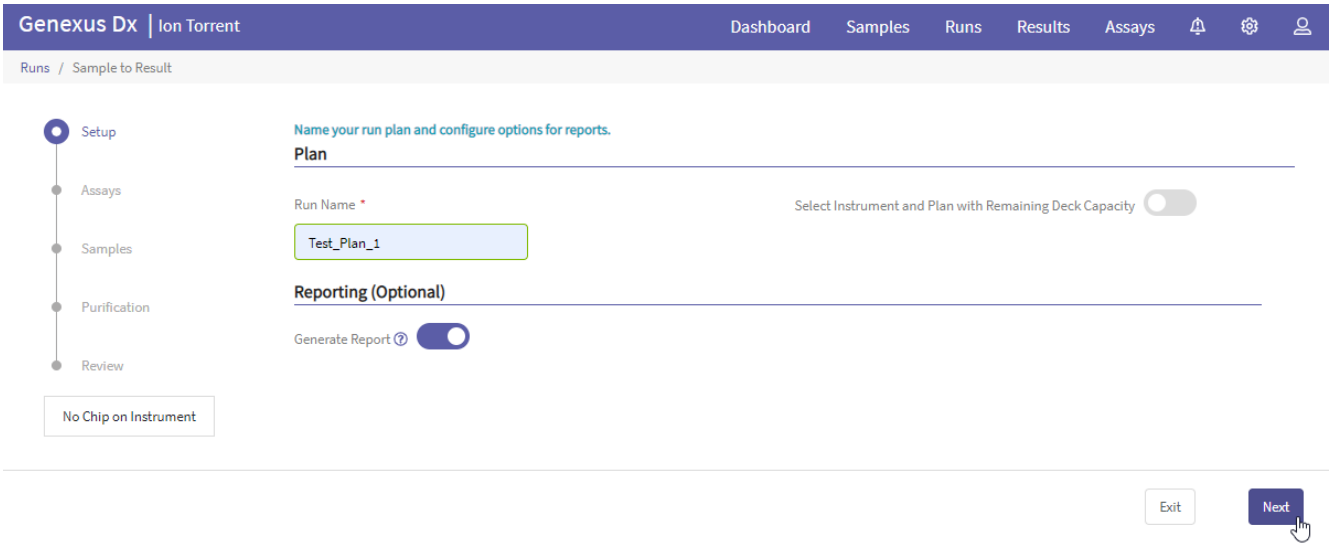


2. In the **Setup** step, enter or make the following selections.
   a. In the **Plan** section, enter a unique name.

      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**.



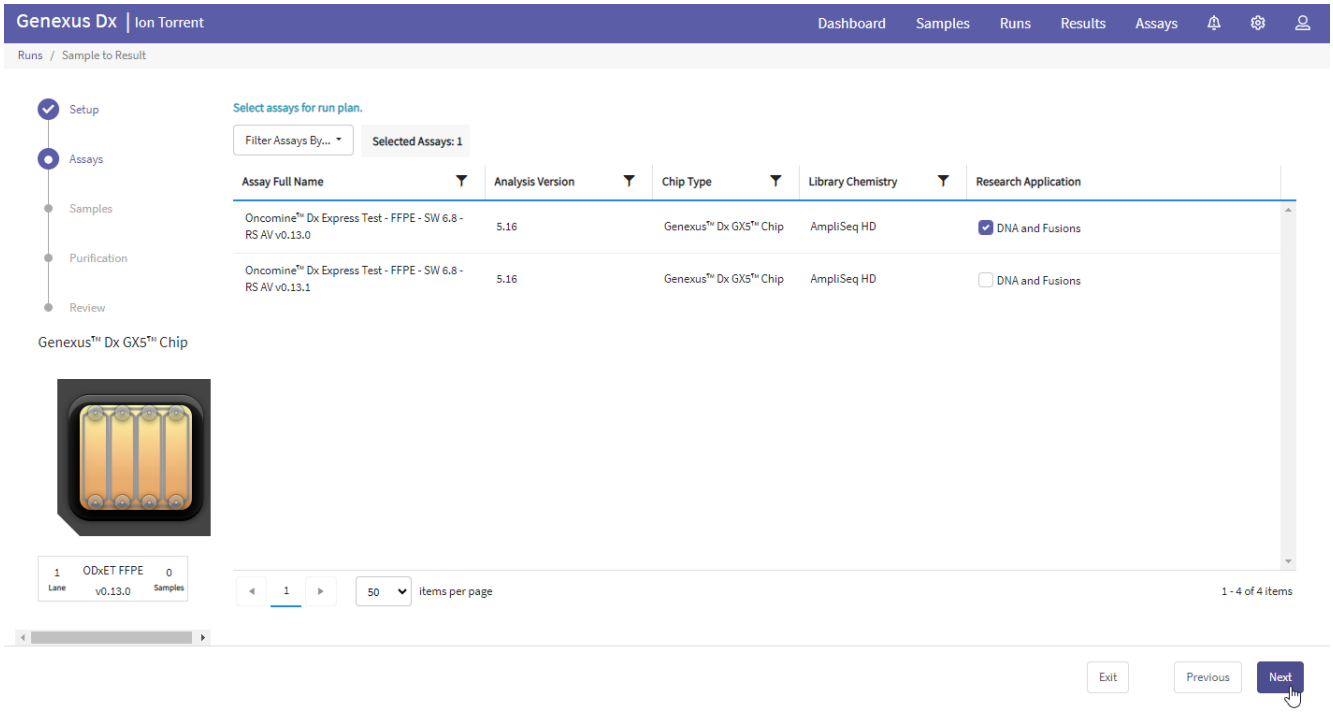
3. In the **Assays** step, select the assay that you want to include in the run.

   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.

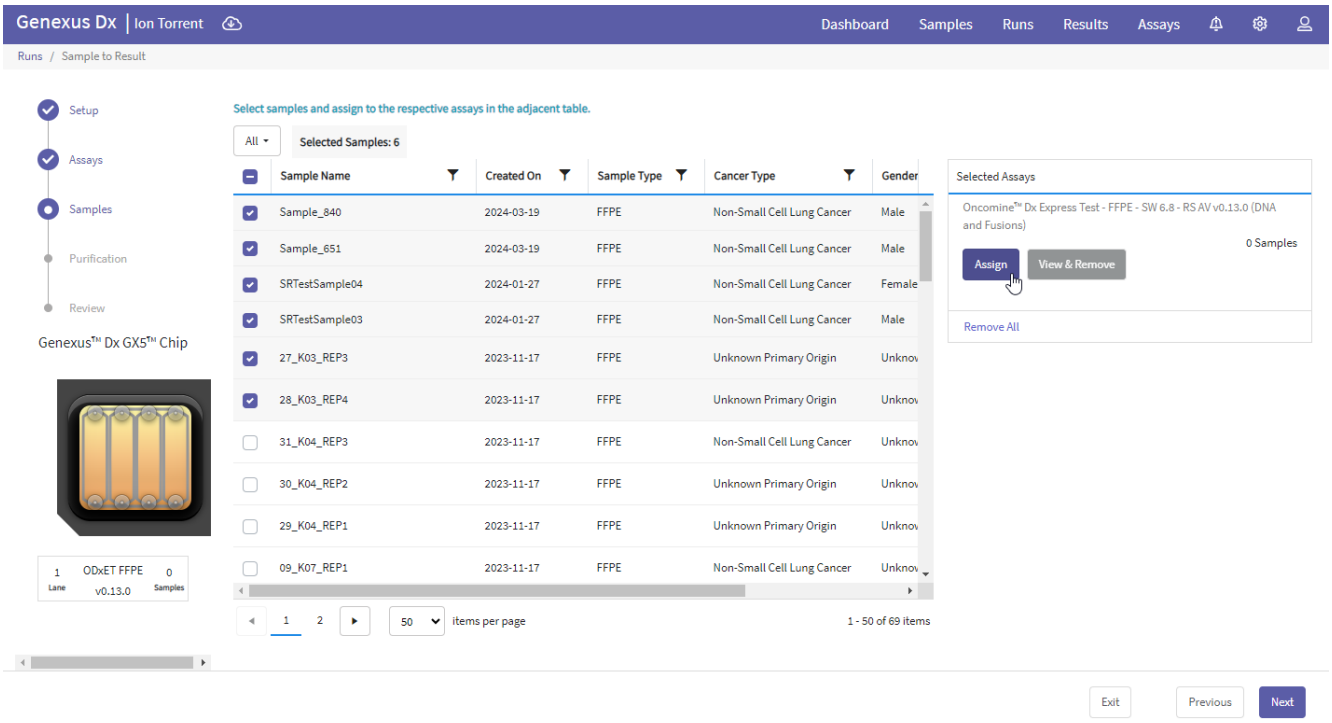
   a. Use the **(Filter)** tools in table column headers to find assays of interest, if desired.

   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: For FFPE DNA+RNA assays, 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. We recommend that you select 12 samples in a run for the most efficient use of purification consumables. You can include up to 14 samples in an FFPE DNA+RNA sample sequencing run, but doing so would require a second purification run and manual transfer of 2 samples of the second run to 4 wells of the first sample plate.

The **Chip View** updates to show the lanes used in the run. Lane usage is calculated based on the number of samples and positive 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. Red denotes a chip lane in the run containing assigned samples that exceed lane capacity.

Error messages can appear that may require you to change the samples you include in the run.

- 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 will 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.

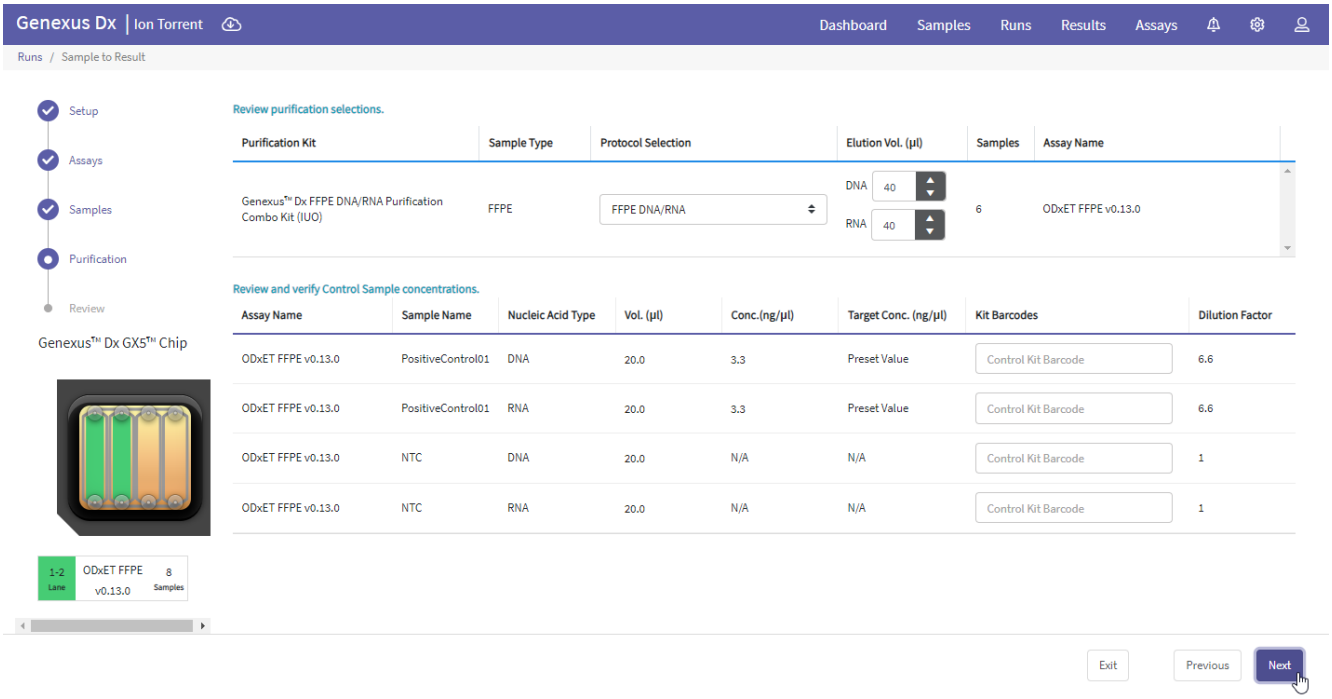
- If the cancer type of a sample, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type.

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	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.

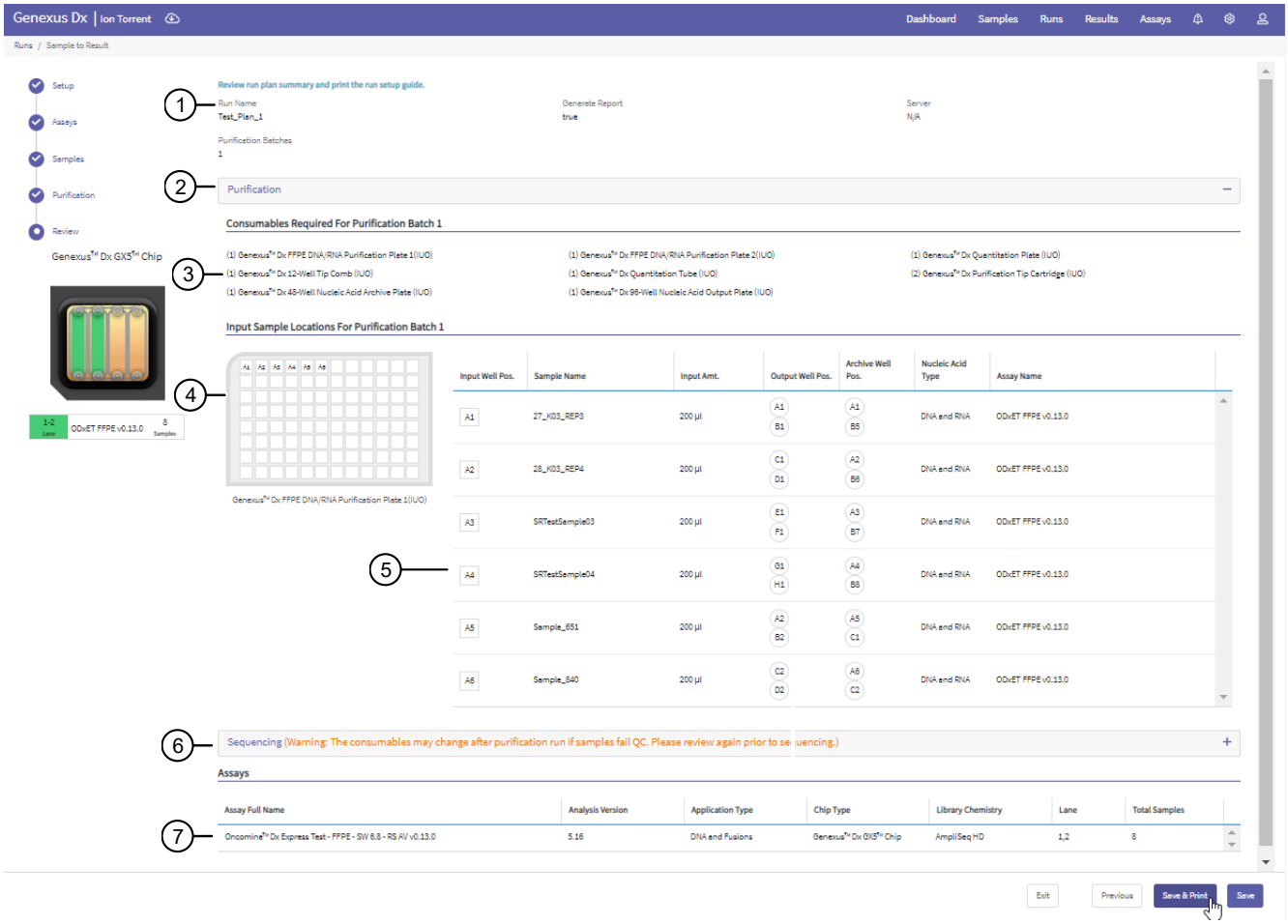
   If needed, you can enter the barcodes manually.

c. Click **Next**.

6. In the **Review** step, review the run setup guide, then click **Save**.

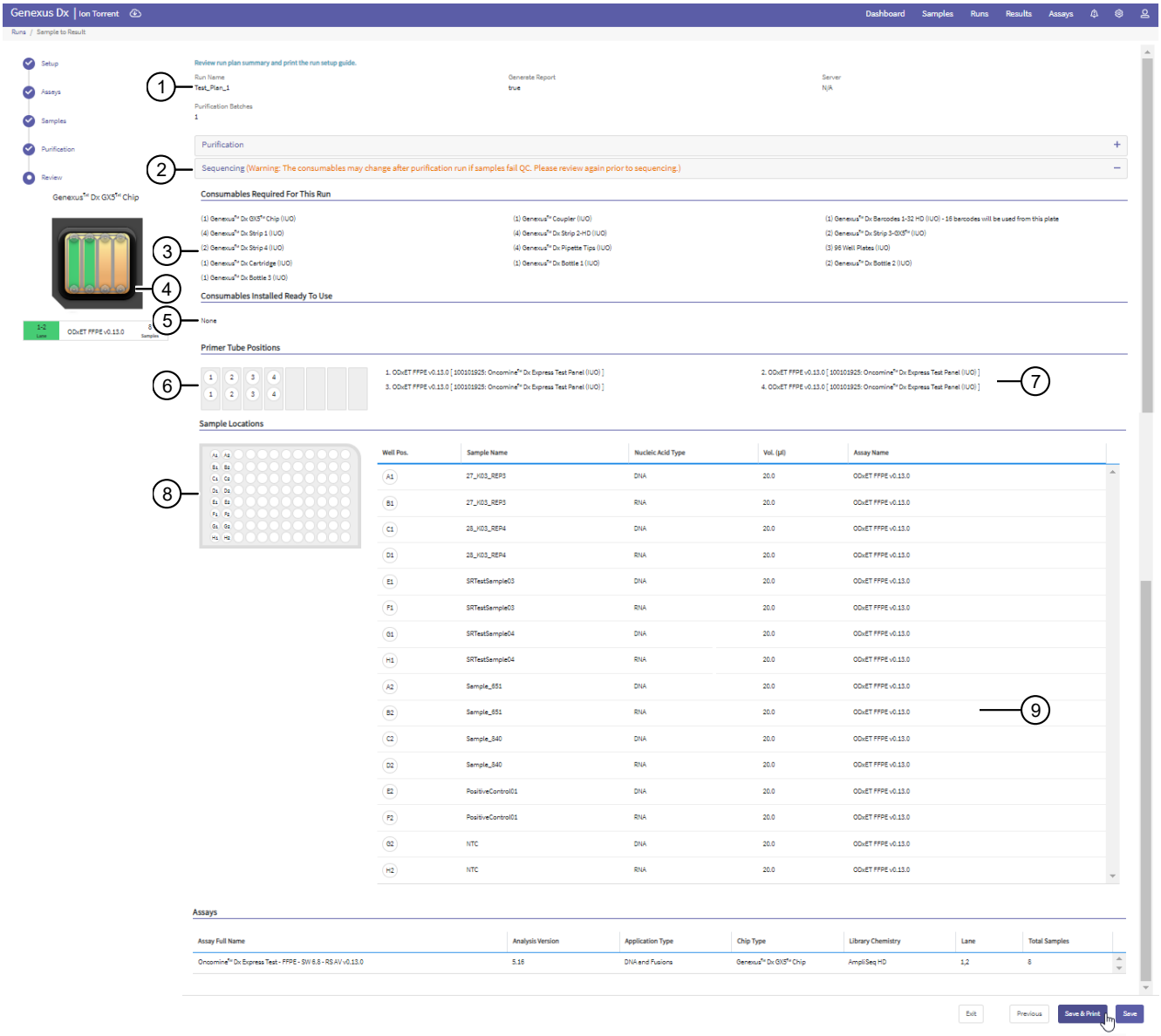
   The run plan summary lists the following details:

   - 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

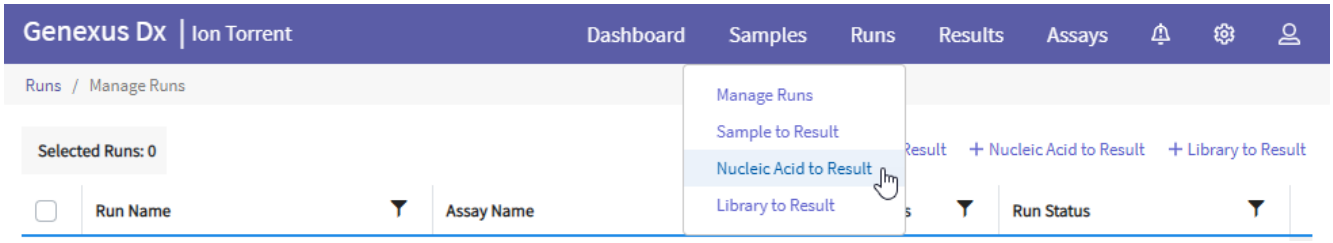
In Genexus™ Dx Software 6.8 and later, a run that starts with nucleic acid samples is called a **Nucleic Acid to Result** run.

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.

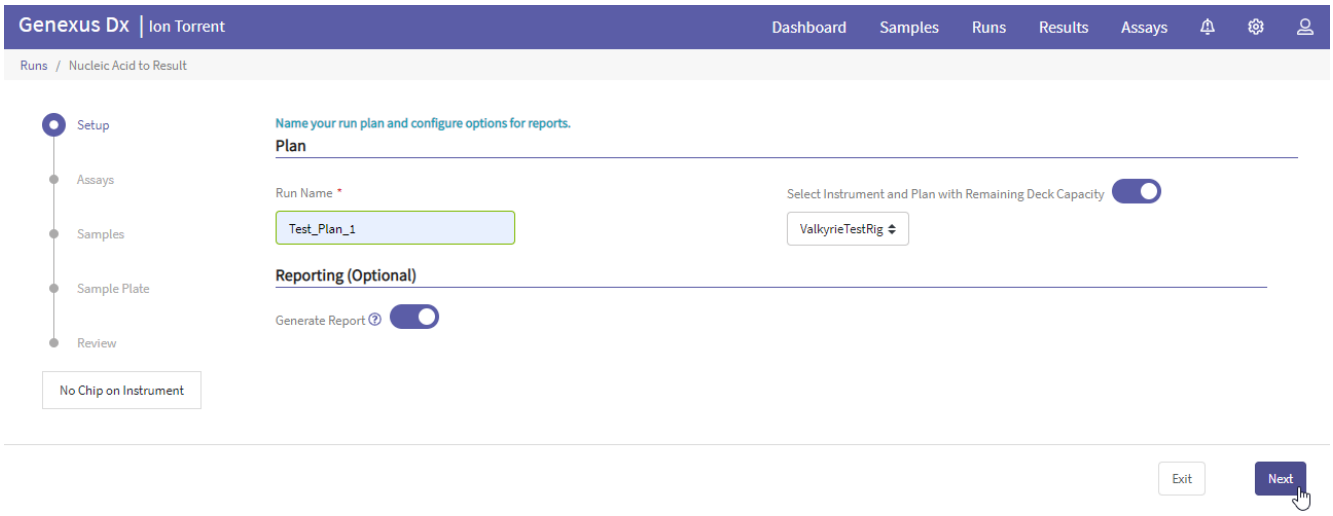
- Enter sample information into Genexus™ Dx Software. For more information, see “Create a new sample” on page 39, or “Import samples” on page 40.
- Quantify sample concentration.
- Connect the barcode scanner to the computer on which you use Genexus™ Dx Software to plan runs and verify that the barcode 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.

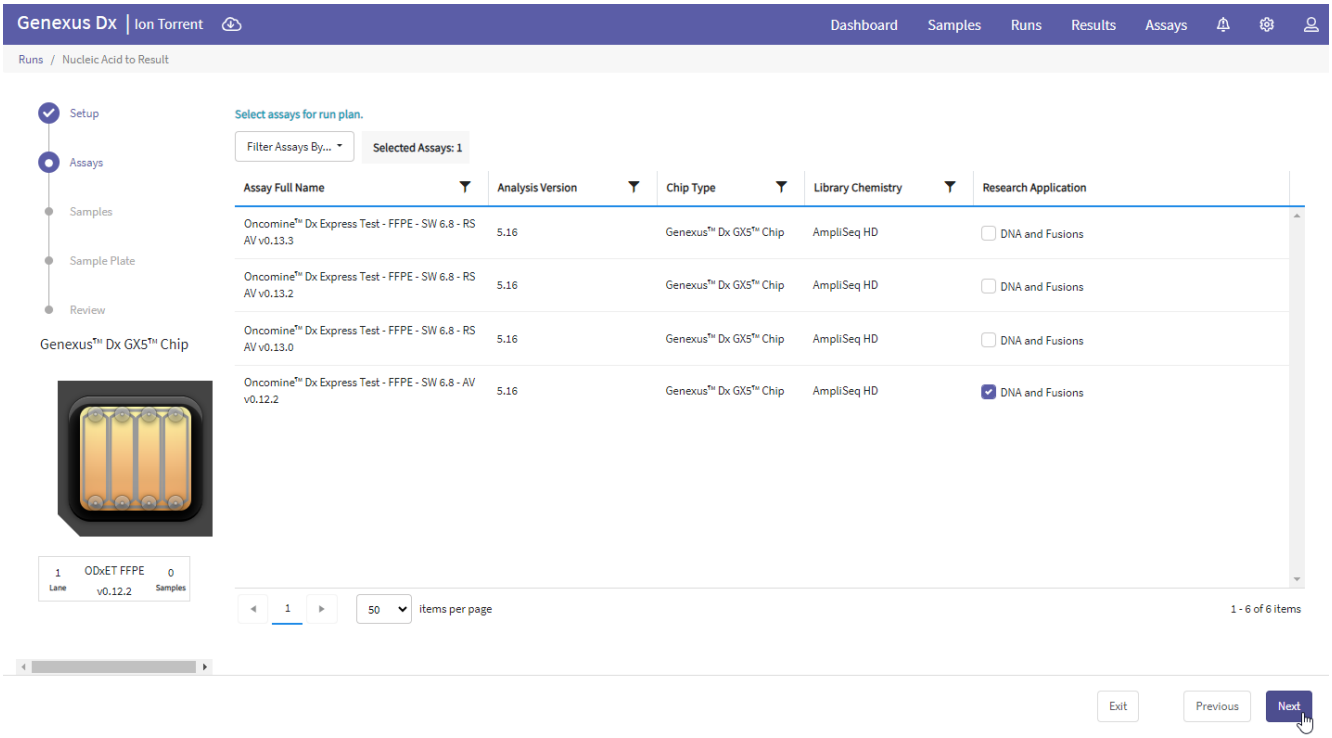
Note: Leave **Generate Report** enabled to generate a variant report using the default report template. To create a report template, click **Assays** ▸ **Manage Presets**, then in the **Report Templates** tab, click + **Add New**.

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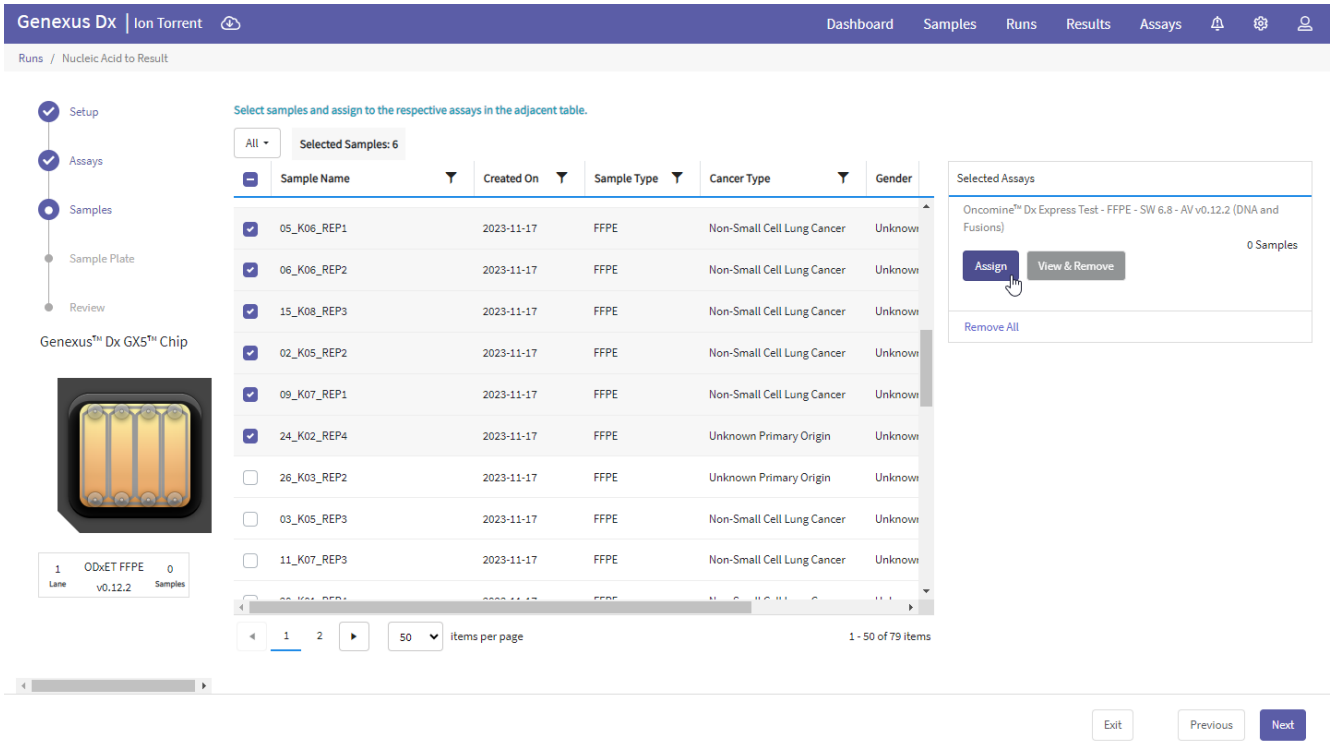
c. Click **Next**.

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 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. Red denotes a chip lane in the run containing assigned samples that exceed lane capacity.

Error messages can appear that may require you to change the samples you include in the run.

- 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 will 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.

- If the cancer type of a sample, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type.

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.

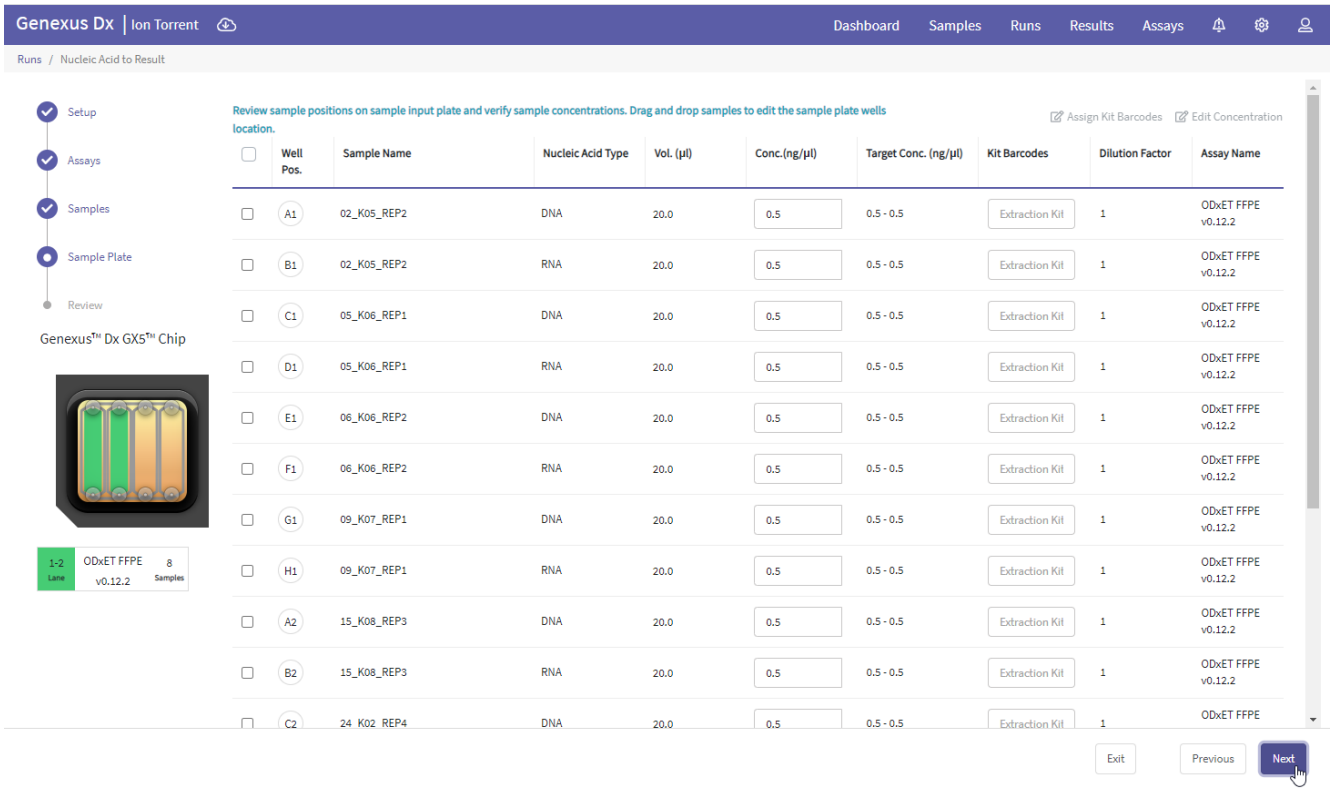
   Appropriate controls are automatically included in the sample plate list:
   - Oncomine™ Dx Express Test FFPE DNA Control
   - Oncomine™ Dx Express Test FFPE RNA Control
   - Oncomine™ Dx Express Test NTC Control

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.

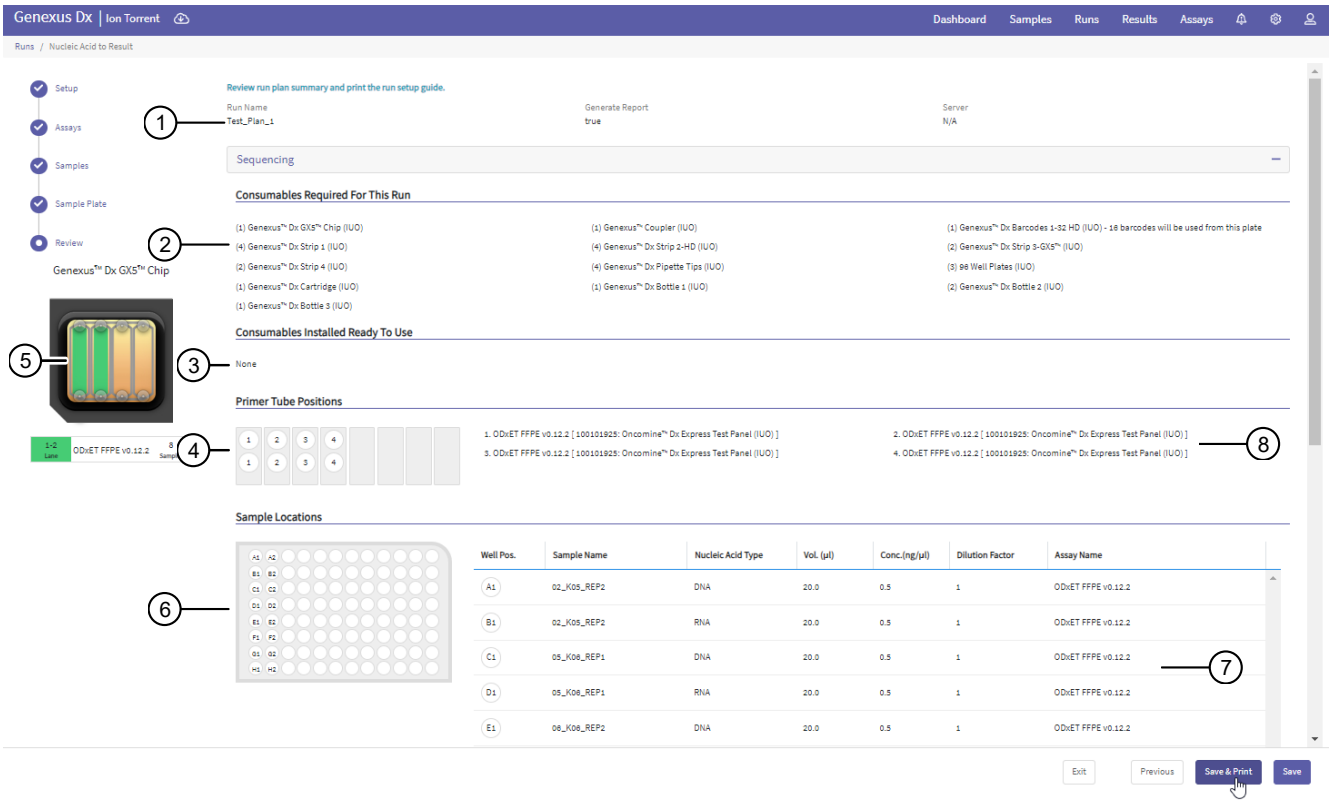
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c. Ensure that sample plate information is correct, then click **Next**.

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.

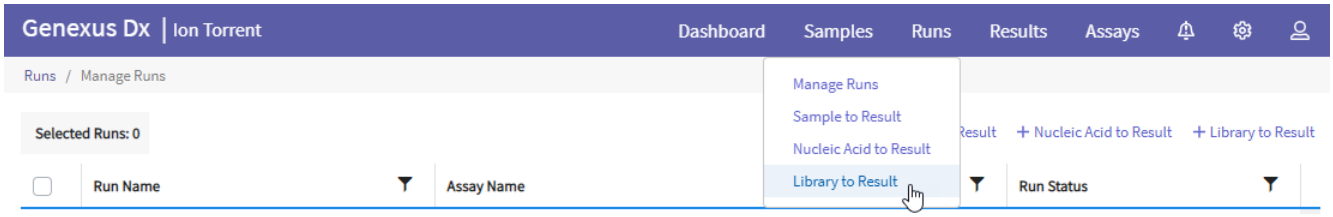
IMPORTANT! Before planning a Library to Result run using pooled libraries recovered from a previous run, confirm that the run failed due to a templating or sequencing problem, and that a sufficient volume of pooled sample library is available in well E4 of the PCR amplification plate. For more information, see "Recover pooled sample libraries and plan a Library to Result run" on page 223.

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 42.

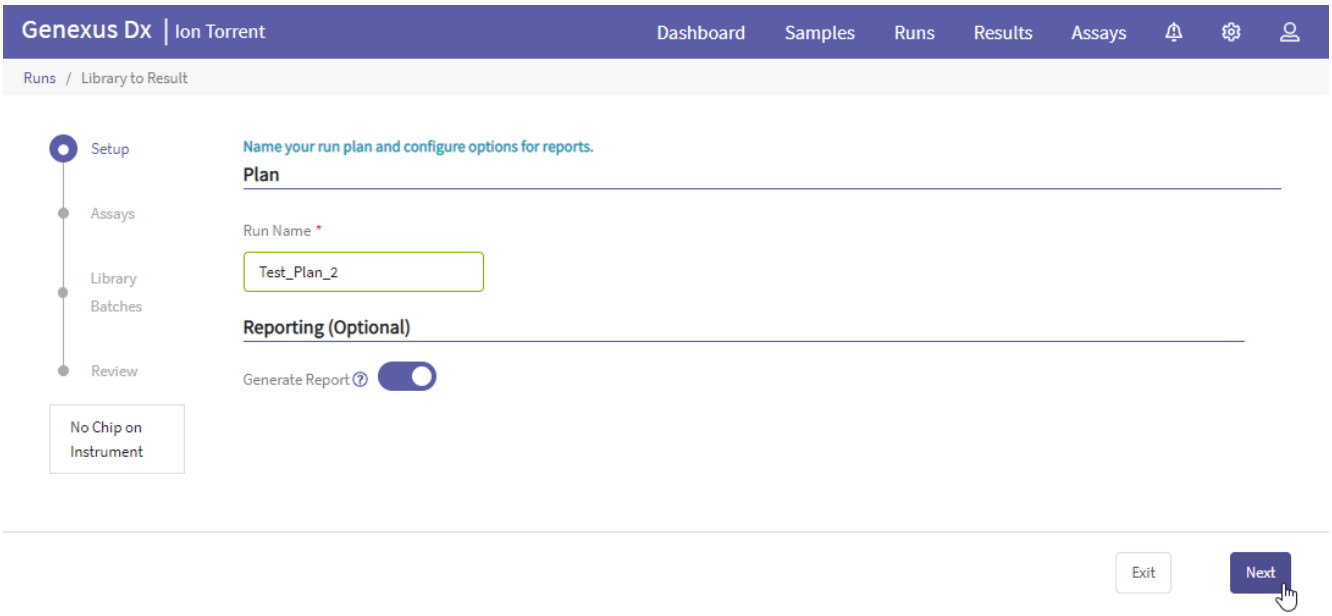
Genexus™ Dx Software guides you through the four steps of planning a Library to Result run: **Setup**, **Assays**, **Library Batches**, and **Review**. You can view progress through the steps in the upper left corner of the **Runs / Library to Result** screen.

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.



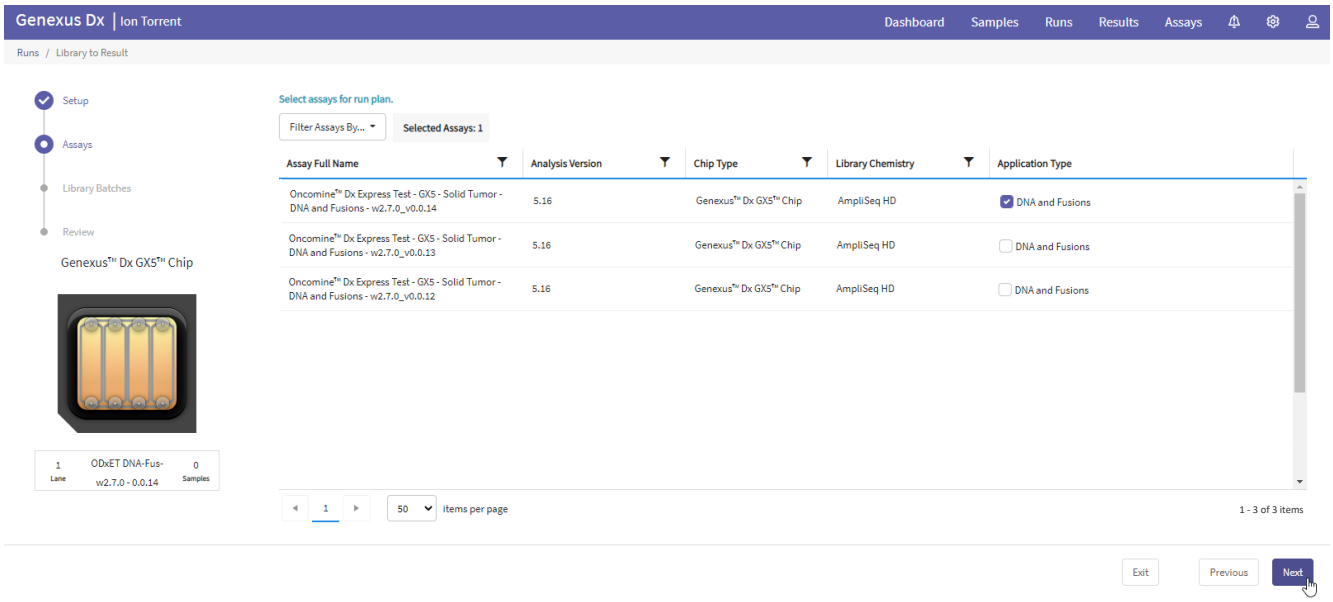
   b. *(Optional)* In the **Reporting (Optional)** section, select **Generate Report**, if needed.

      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.

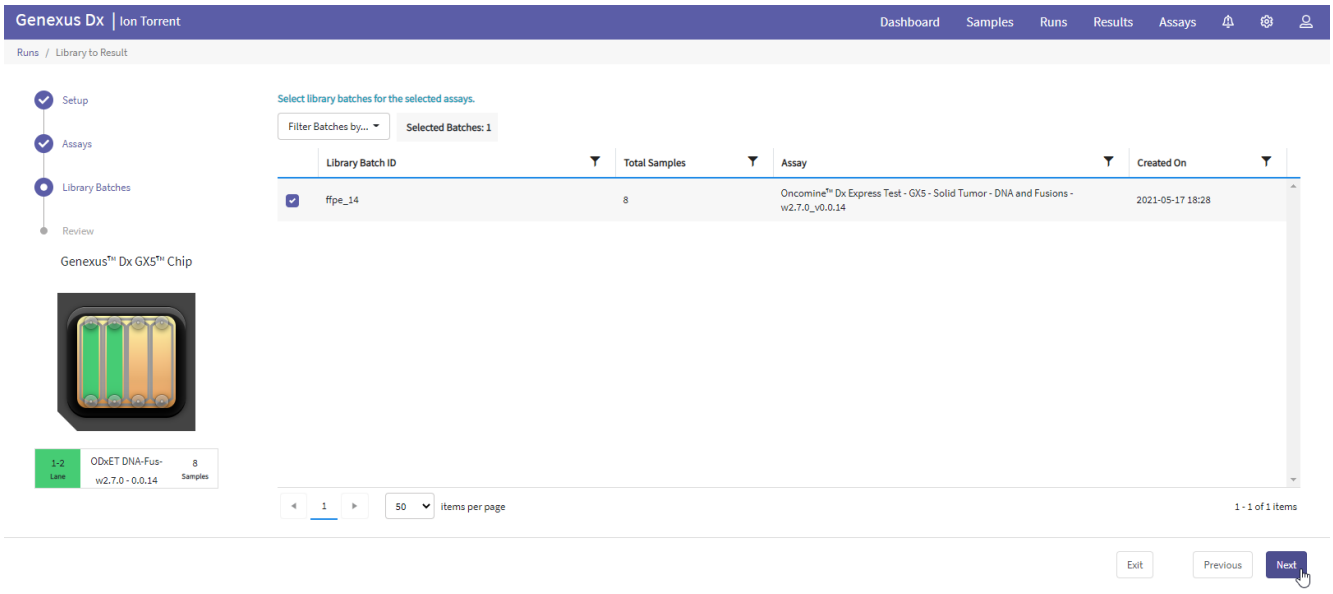
3. In the **Assays** step, select the assay that you want to use in the run, then click **Next**.

   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.



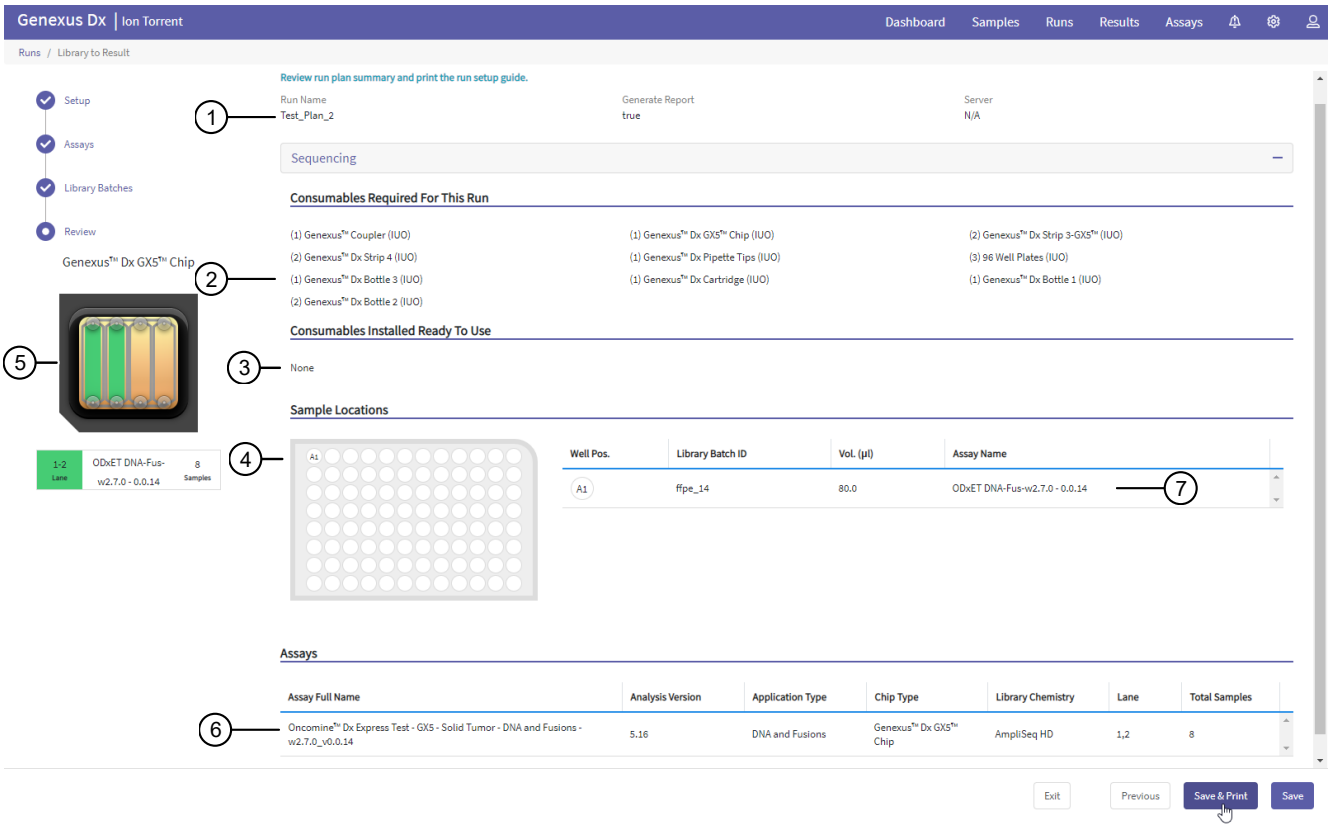
Green lane color indicates lane usage and sample assignment within lane capacity. Red lane color indicates sample assignment that exceeds lane capacity.

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 will 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

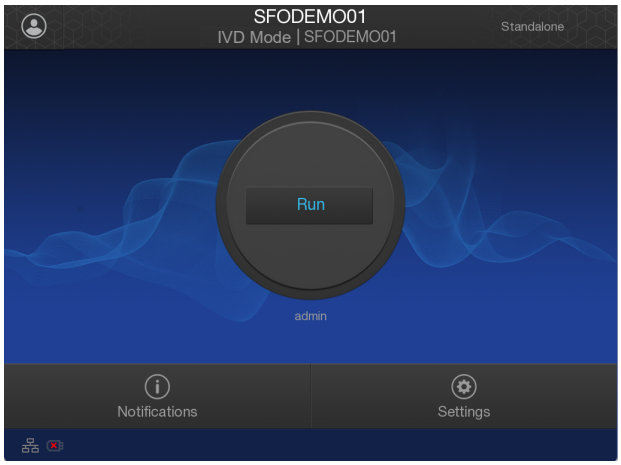
If you are purifying DNA and RNA from FFPE samples on the Genexus™ Dx Purification System in a standalone configuration, create a plan for the run using the touchscreen of the purification instrument. In the standalone instrument configuration, all purified sample DNA and RNA is loaded in an archive plate.

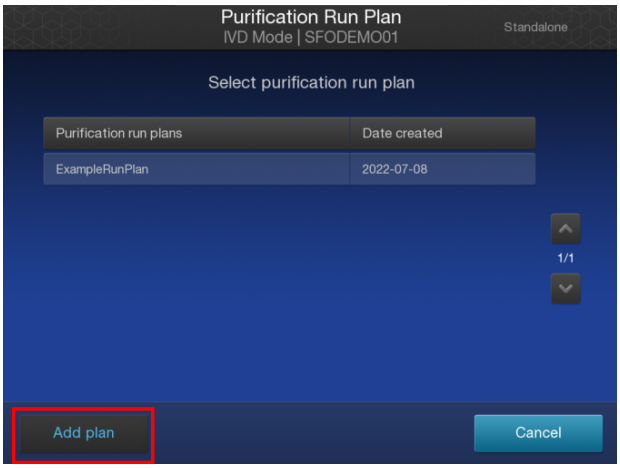
Note: We recommend that you plan the run before preparing the samples and loading into the Genexus™ Dx FFPE DNA and RNA Purification Plate 1. However, experienced users can save time by planning the purification run during the protease digestion step of sample preparation.

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1. Enter your username and password to sign in to the instrument.

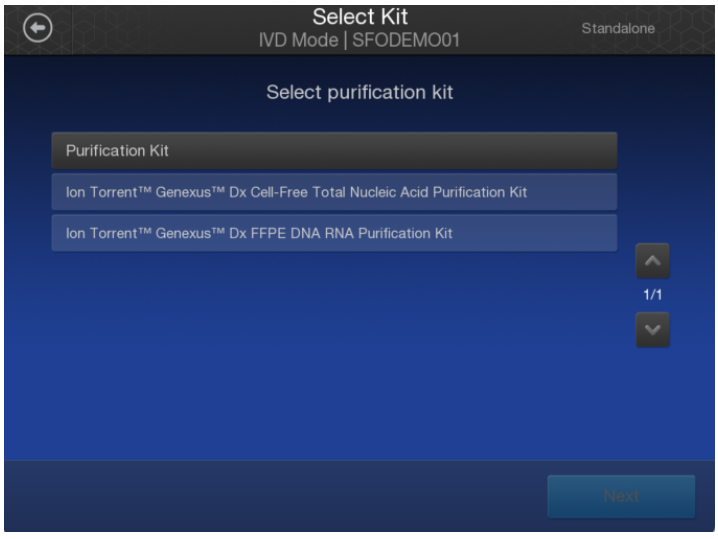
2. Tap **Run**, select **Purification Only**, then tap **Add plan**.



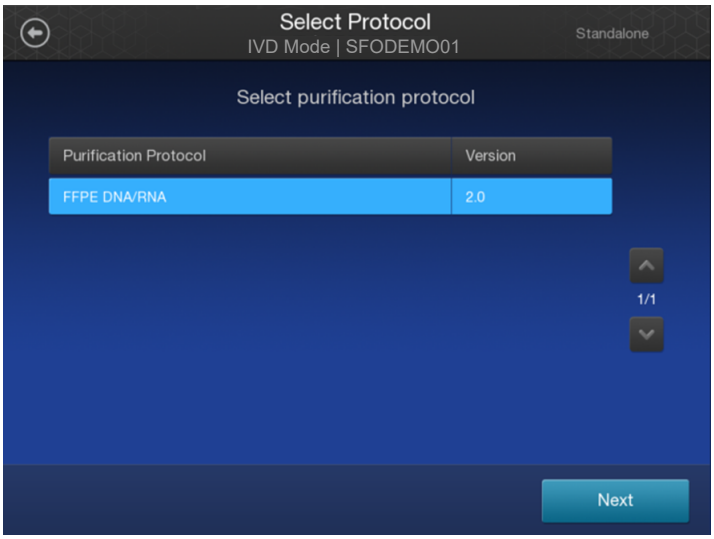


3. Tap in the entry box, enter a unique name for the run plan, then tap **Done** ▸ **Next**.

4. Select the Ion Torrent™ Genexus™ Dx FFPE DNA and RNA Purification Kit, then tap **Next**.



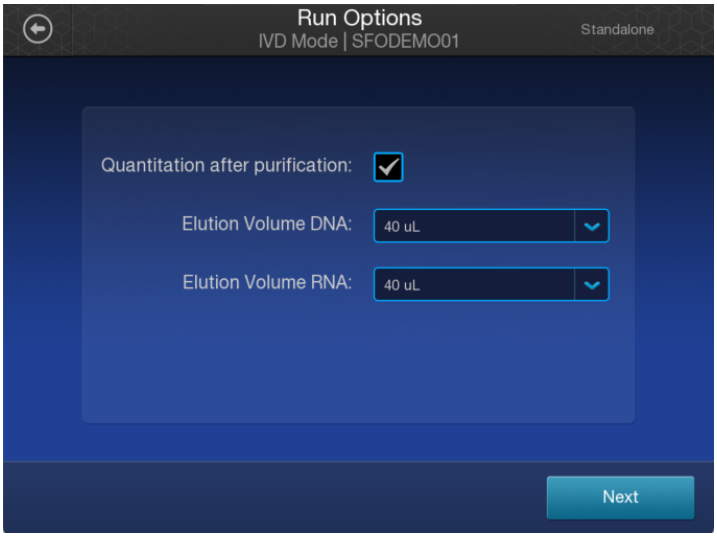
5. Select the **FFPE DNA/RNA** purification protocol, then tap **Next**.



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).

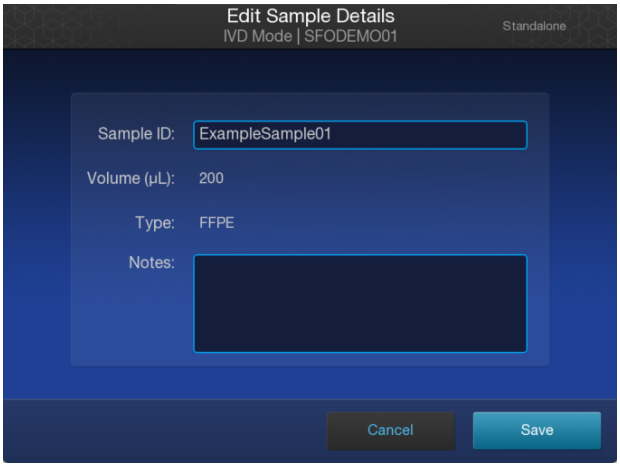
b. Tap on a sample ID to select the sample.

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 213.

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a. In the **Manage Samples** screen, select the samples for which sample information is being imported, then tap **Import**.

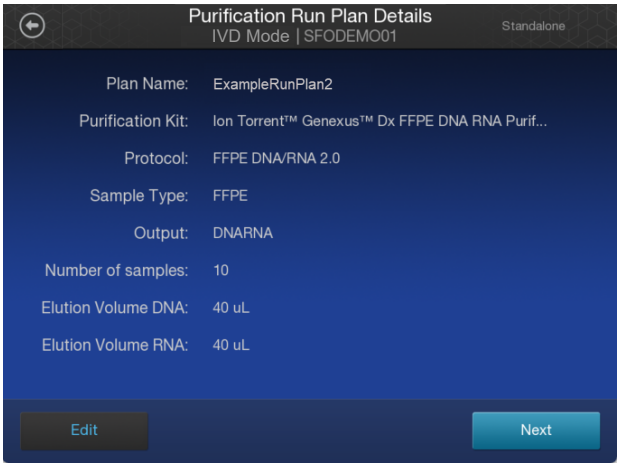
b. The **Sample Import** screen opens. Tap **Import** to proceed.

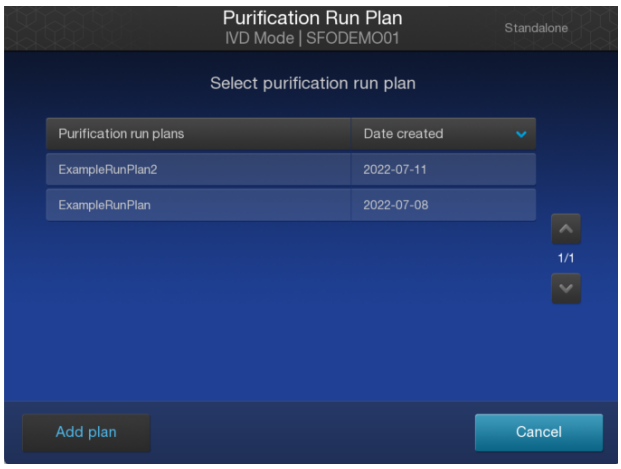
c. Insert the USB drive containing the sample import CSV file into the USB port on the front of the purification instrument. In the **Sample Import** screen, select the USB drive, then navigate to and select the sample import file.

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 214.

5 Start a purification run for FFPE samples

- Prepare FFPE samples 73
- Load the instrument and start the run 80
- Unload and UV clean the instrument 90
- View and export quantitation results 93

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 50). 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 69). 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.

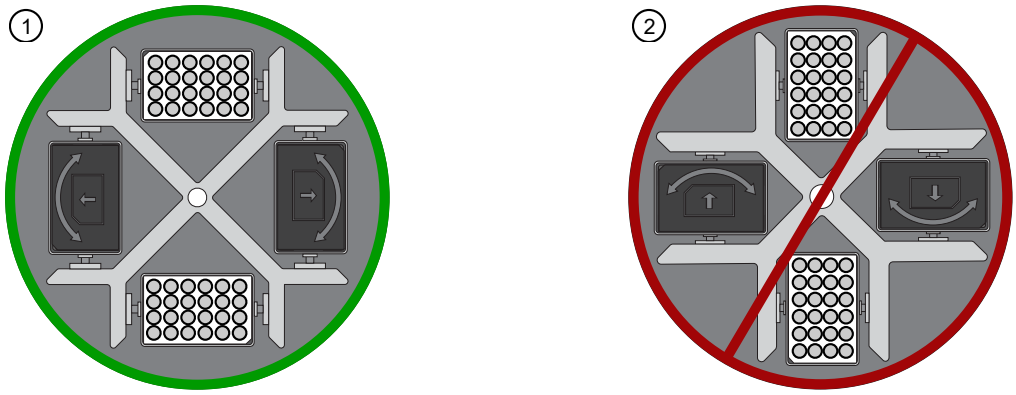
Prepare FFPE samples

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 –65°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 Tube Rack in "landscape" orientation.



① Landscape orientation—recommended

② Portrait orientation—not recommended

Note: Place AutoLys 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
- Genexus™ Dx AutoLys Tube Lifter or Genexus™ Dx AutoLys Pliers
- Genexus™ Dx AutoLys Tubes and Genexus™ Dx AutoLys Caps
- Genexus™ Dx AutoLys Tube Rack
- Genexus™ Dx AutoLys Locking Lid
- 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 samples.

Component	Volume per reaction
Genexus™ Dx FFPE Protease Buffer	(`n` + 1) × 225 µL
Genexus™ Dx Proteinase K (red cap)	(`n` + 1) × 10 µL
Total volume	**(`n` + 1) × 235 µL**

3. Vortex for ~5 seconds to mix, then briefly centrifuge to collect the contents.

Prepare FFPE curls with Genexus™ Dx AutoLys Tubes

Digest with protease

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).

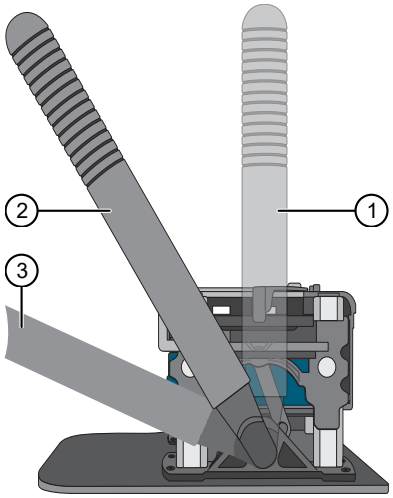
1. Label an AutoLys Tube for each FFPE tissue sample.
2. Transfer each FFPE section curl from its storage tube to a separate labeled AutoLys Tube.
   a. Invert a labeled AutoLys Tube over an uncapped storage tube containing the curl, then carefully invert the 2 tubes together so that the storage tube is on top.
   b. Tap or flick the storage tube so that the curl falls into the AutoLys Tube.

   If the curl adheres to the storage tube, gently grasp an edge of the curl with forceps, then transfer it to the AutoLys 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 Tubes, place the tubes in an AutoLys 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.

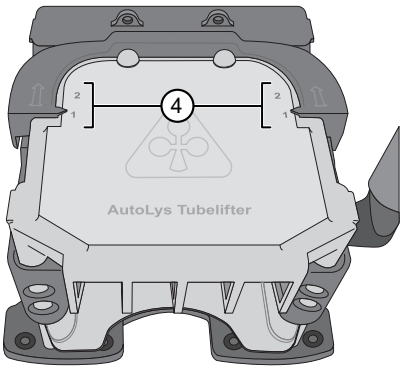
   Ensure that the samples are submerged in the Protease Digestion Master Mix. If tissue adheres to the side of the tube, use a sterile pipette tip to push the sample into the solution.
5. Securely recap each tube to limit evaporation, then incubate the capped tubes at 60°C for ≥60 minutes in an AutoLys 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. Securely retighten each tube cap to limit evaporation, then 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 81.
   - Equilibrate the Quantitation Plate to room temperature during the incubation.
7. Allow samples to cool to room temperature for 5–10 minutes before proceeding to lift the tubes.
8. Lift the tubes. The following steps describe use of the AutoLys Tube Lifter to process up to 24 samples simultaneously. Alternatively, AutoLys Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys Tube Lifter lever is in Position A (straight up) and the slider is in Position 1.
   b. Slide the 24-well AutoLys Tube Rack containing the lysed samples into the AutoLys Tube Lifter.
   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 Tube Lifter see the *AutoLys M TubeLifter User Guide* (Pub. No. MAN0017676).



① AutoLys Tube Lifter lever Position A
② AutoLys Tube Lifter lever Position B
③ AutoLys Tube Lifter lever Position C



④ AutoLys Tube Lifter slider positions 1 & 2

9. Slide the AutoLys 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 Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge. See “Procedural guidelines” on page 73.
- Ensure that AutoLys Tube caps are securely held by the AutoLys Locking Lid before you start the centrifugation.

10. Using the AutoLys Tube Lifter or AutoLys Pliers, separate the filter from the outer tube.
    a. Adjust the position of the AutoLys Tube Lifter slider to Position 2.
    b. Remove the AutoLys Locking Lid, then slide the rack into the AutoLys Tube Lifter.
    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 82.

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 Genexus™ Dx AutoLys Tubes

Collect the tissue

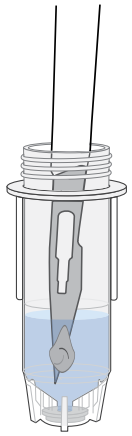
1. Label an AutoLys Tube for each FFPE tissue sample.

   Label each tube (cap and side) with its Sample ID using a marker that is resistant to 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 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 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.

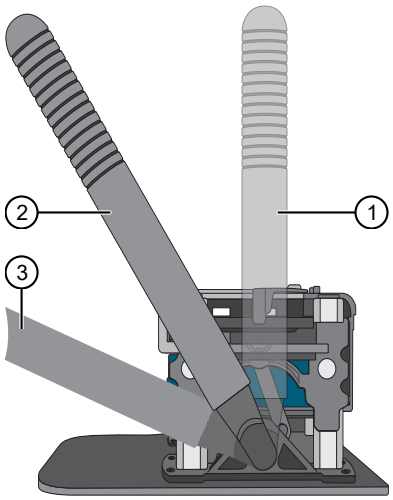
   Note: Ensure that the incubator loaded with sample tubes has sufficient space to allow the rapid transfer of heat within the incubator—do not overcrowd with tubes.

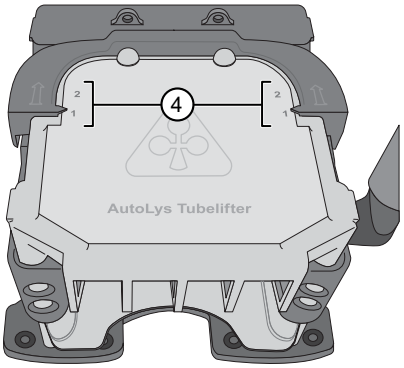
2. Securely retighten each tube cap to limit evaporation, then incubate at 90°C for 60 minutes.

   - Prepare the reagents and consumables that are needed by the instrument during the incubation. See “Prepare the consumables” on page 81.
   - Equilibrate the Genexus™ Dx Quantitation Plate to room temperature during the incubation.

3. Allow samples to cool to room temperature for at least 10 minutes before proceeding to lift the tubes.

4. Lift the tubes. The following steps describe use of the AutoLys Tube Lifter to process up to 24 samples simultaneously. Alternatively, AutoLys Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys Tube Lifter lever is in Position A (straight up) and the slider is in Position 1.
   b. Slide the 24-well AutoLys Tube Rack that contains the lysed samples into the AutoLys Tube Lifter.
   c. Press the lever down from Position A to Position B, then remove the rack from the lifter.





① AutoLys Tube Lifter lever Position A
② AutoLys Tube Lifter lever Position B
③ AutoLys Tube Lifter lever Position C

④ AutoLys Tube Lifter slider positions 1 & 2

5. Slide the AutoLys 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 Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge (see “Procedural guidelines” on page 73).
- Ensure that AutoLys Tube caps are securely held by the AutoLys Locking Lid before you start the centrifugation.

6. Using the AutoLys Tube Lifter or AutoLys Pliers, separate the filter from the outer tube.
   a. Adjust the position of the AutoLys Tube Lifter slider to Position 2.
   b. Remove the AutoLys Locking Lid, then slide the rack into the AutoLys Tube Lifter.
   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 82.

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 supplied in the kit for ~5 seconds, then briefly centrifuge to collect the contents.
2. Pulse-vortex the Genexus™ Dx DNase supplied in the kit for ~1 second, then briefly centrifuge to collect the contents.

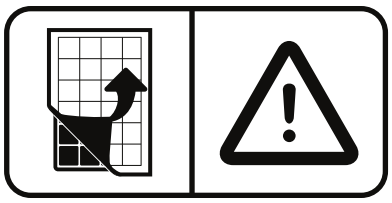
   Note: Genexus™ Dx DNase is not a shear-sensitive enzyme, but to avoid physical denaturation, perform a pulse-vortex to ensure homogeneity of the enzyme mix since it is stored at room temperature.
3. In a 1.5-mL low-retention microcentrifuge tube, prepare a 1X DNase Digestion Master Mix as indicated, where `n` is the number of 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**

4. With a pipette set to 100 µL, pipet the solution up and down 5 times to mix, then briefly centrifuge to collect the contents.

5. Centrifuge the Genexus™ Dx FFPE DNA and RNA Purification Plate 2 at 1,000 × *g* for 30 seconds to collect the contents.

6. Carefully remove the plate seal without disturbing the contents.



7. 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.

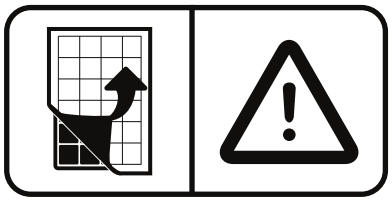
Note: You can add the samples to the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 and load the plate as the final steps of loading the instrument. This can be done to help ensure that other components are successfully loaded and accepted by the instrument before adding samples of possibly limited 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.

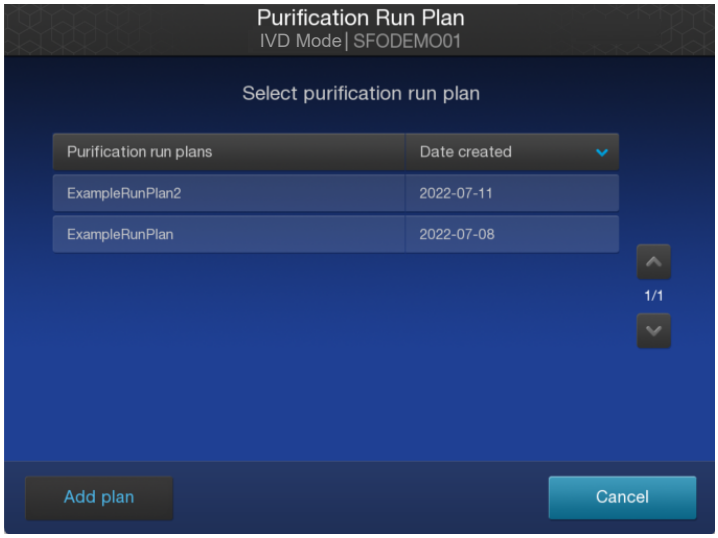
   A precipitate can form, but this does not interfere with the DNA binding. Proceed directly to the next step.

   Reagent consumables cannot be reused.

   IMPORTANT! Load the plate in the instrument (see page 85) 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.

3. *(Optional)* Import sample information.

   The import overwrites the existing **Sample ID** and **Notes** information for each sample. That is, if the run plan has 6 samples, the sample import file must include information for at least 6 samples.

   To import sample information, prepare a CSV sample import file and save it to a USB drive. See “Create a template for importing samples in standalone configuration purification run plans” on page 213.

   a. In the **Sample Assignment** screen, tap **Manage Samples**.

   b. In the **Manage Samples** screen, tap **Import**.

   c. In the **Sample Import** screen, tap **Import** to proceed.

   d. Insert the USB drive that contains 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.

   e. *(Optional)* Tap **Details** to view the CSV file that lists the sample names to be imported.

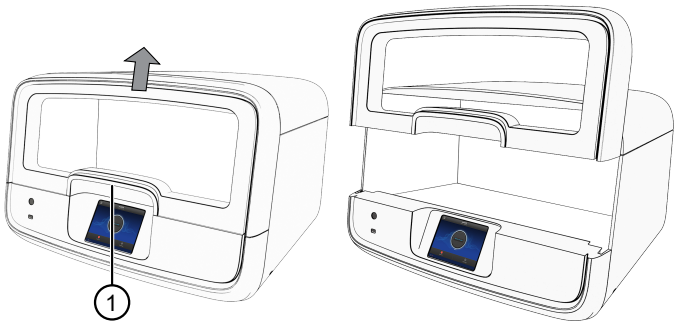
   f. Tap **Import**, then in the **Import Successful** screen, tap **OK**.

      The imported sample information is shown in the **Manage Samples** screen. If needed, select a sample, then tap **Edit** to modify the **Sample ID** or **Notes**.

4. Tap **Next**.

   The instrument performs a 2-minute UV cleaning, then unlocks the door.

5. Lift the instrument door to the stop.



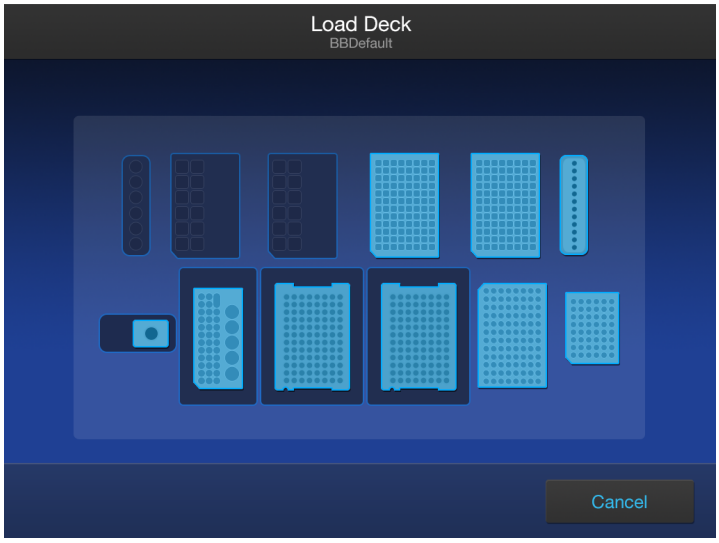
① Hold here, then lift.

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.

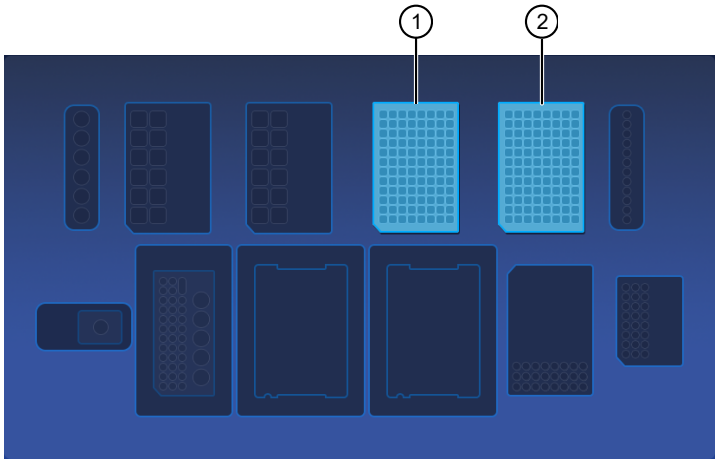


Screen display for an instrument fully loaded for an FFPE sample purification run (integrated configuration).

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 82.

2. Load the Genexus™ Dx FFPE DNA and RNA Purification Plate 2 (RNA plate) prepared in step 7 of "Add 1X DNase Digestion Master Mix to the Genexus™ Dx FFPE DNA and RNA Purification Plate 2" on page 81.



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

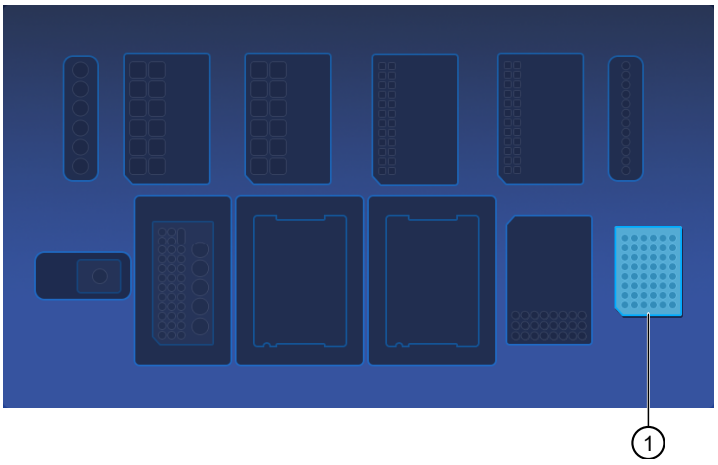
1. Unwrap, then load a new Genexus™ Dx 12-Well Tip Comb.

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.

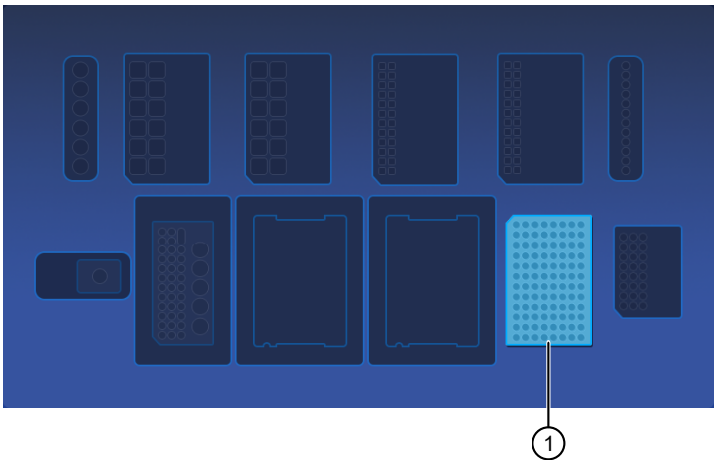


① Genexus™ Dx 48-Well Nucleic Acid Archive Plate position

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.

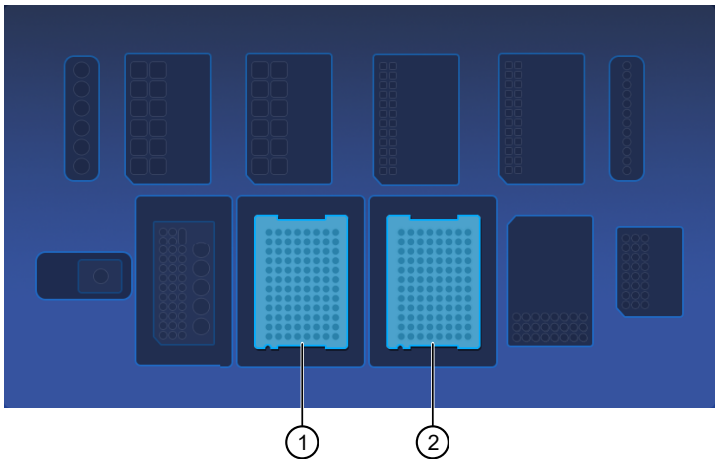
3. *(Integrated run configuration only)* Load a new 96-Well Nucleic Acid Output Plate (MicroAmp™ EnduraPlate™ Optical 96-Well Blue Reaction Plates with Barcode) into the output plate position.

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

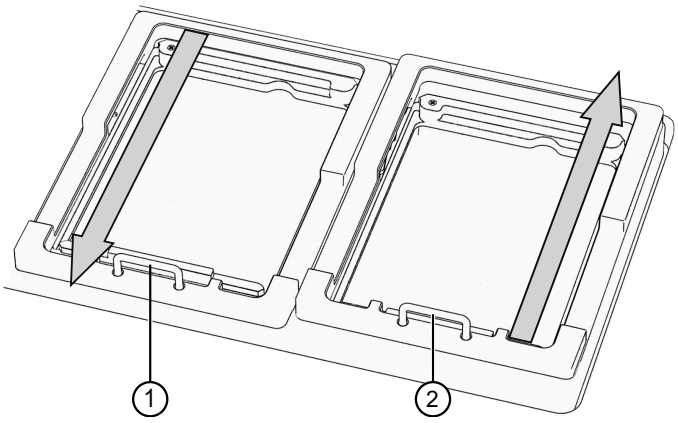
4. Unwrap two new Genexus™ Dx Purification Tip Cartridges and remove the cover to expose the pipette tips, then load them in positions 1 and 2.



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.

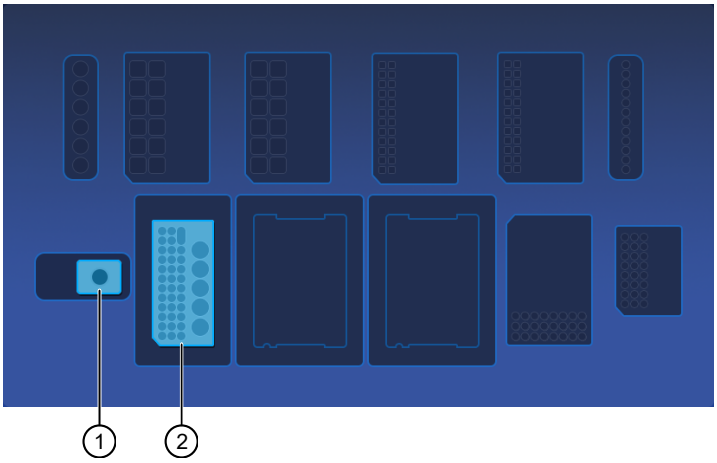


① Unlocked (forward)
② Locked (back)

Load the quantitation reagents and consumables

Note: Protect the Genexus™ Dx Quantitation Plate from direct bright light to prevent photobleaching of the preloaded reagents.

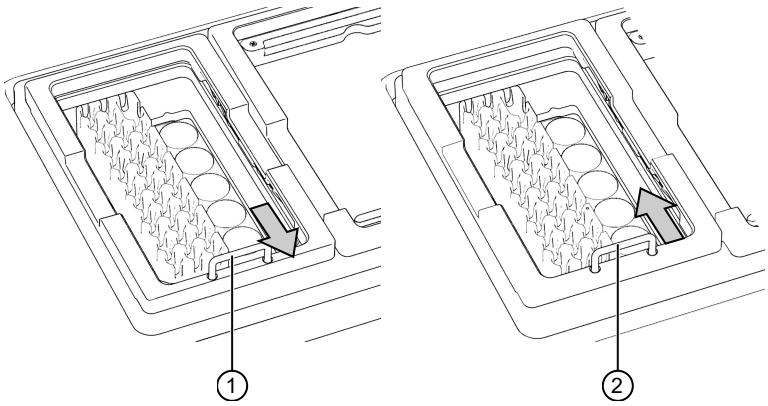
1. Centrifuge the Genexus™ Dx Quantitation Plate at 1,000 × *g* for 30 seconds to collect the contents.
2. Load the Quantitation Plate in position 2.



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.

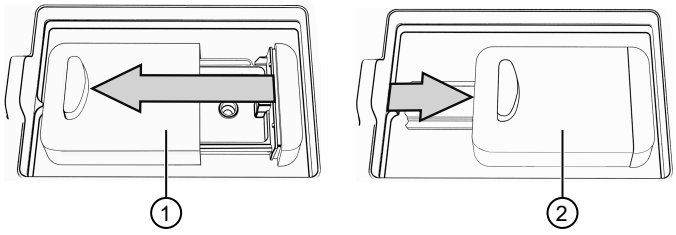


① Unlocked (forward)

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.

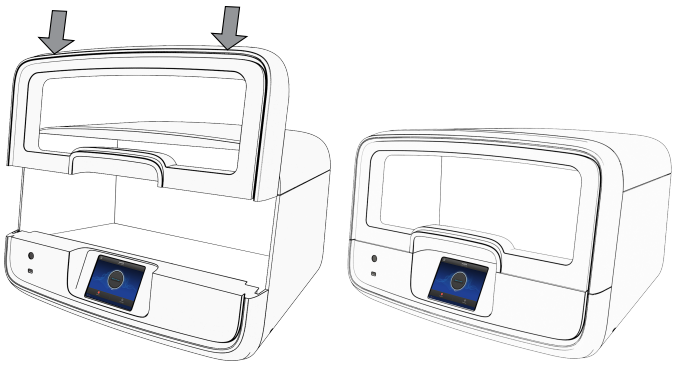
Confirm the following.

- 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 93.

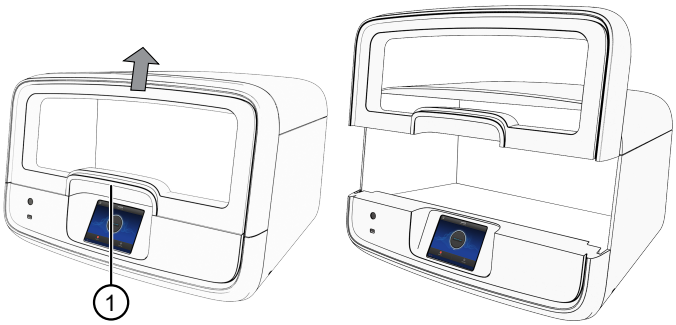
Unload and UV clean the instrument

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.

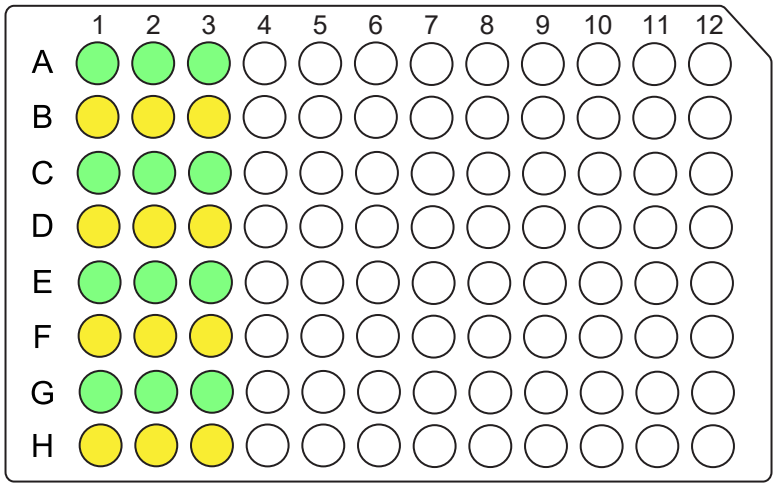


2. Lift the instrument door to access the instrument deck.



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 93.



In the 96-Well Nucleic Acid Output Plate, DNA and RNA for each sample are loaded pair-wise starting at wells A1 and B1

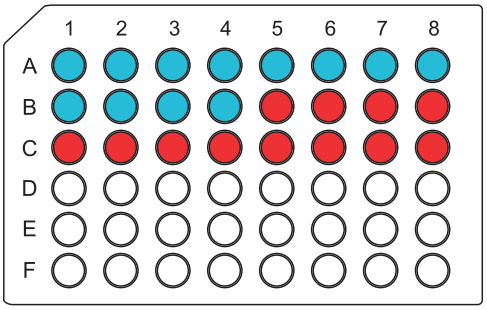
- DNA for 12 samples
- RNA for 12 samples

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 3 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 3 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 3 months after purification with 3 freeze-thaw cycles. 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.

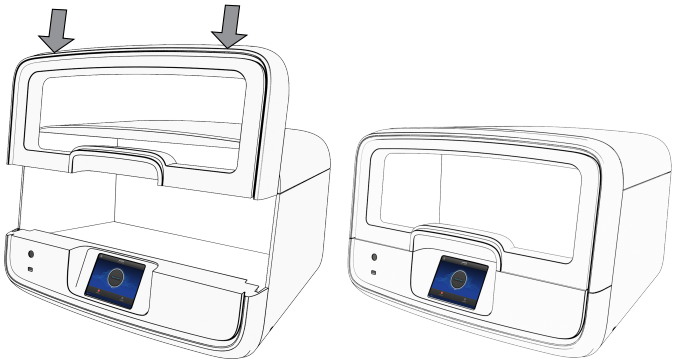
9. Unlock, then remove and dispose of the Genexus™ Dx Quantitation Plate.
   a. Dispose of the liquid waste by tipping the deep-well plate on one corner and pouring the waste into an appropriate liquid biohazardous waste container.

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**.

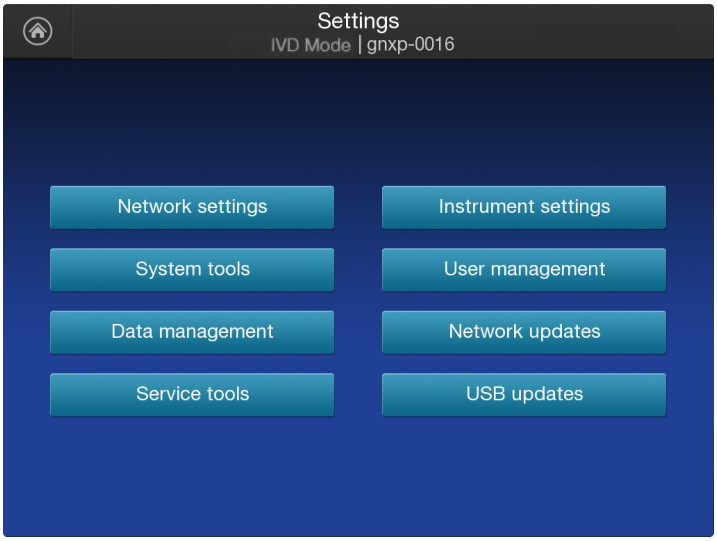


The **Saved Experiment Reports** screen opens. See step 4.

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 97
- Dilute the samples, if needed—Nucleic Acid to Result run 99
- Load the sample plate—Nucleic Acid to Result run 101

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.

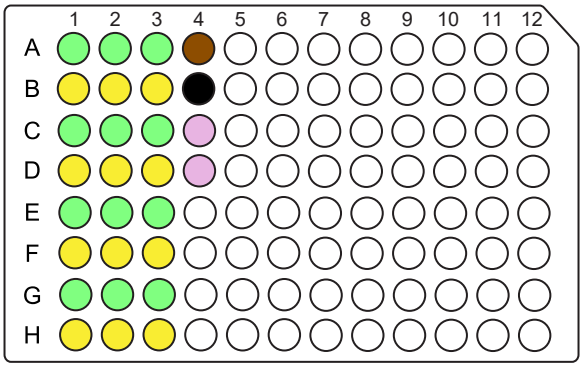
1. Add controls to the sample plate with the volume and in the well positions that are specified in the run setup guide.

   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)
   - Use a PCR hood (if available) when adding controls to the plate to reduce the risk of contamination.

   IMPORTANT! Before adding the NTC, replace your gloves with a new pair of gloves.

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

IMPORTANT! 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 99.

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)
   - Use a PCR hood (if available) when adding controls to the plate to reduce the risk of contamination.

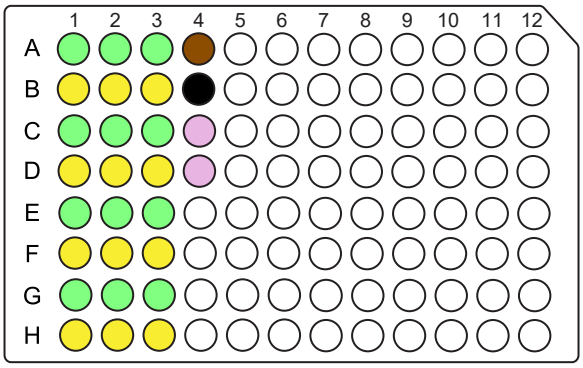
   IMPORTANT! Before adding the NTC, replace your gloves with a new pair of gloves.

   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

IMPORTANT! 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 (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. This helps 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 103
- Before you begin 104
- Load the sequencer and start a run 106
- Clear the instrument deck and perform a UV Clean 115
- Options for an expired sequencer initialization 118

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 30 and “Guidelines for preventing contamination” on page 31.

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 at least 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. If you put Genexus™ Dx Strip 2-HD back on ice, precipitate can reform. Precipitate needs to go back into solution before you load the strip.

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 and tube 5 of Genexus™ Dx Strip 3-GX5™ 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.

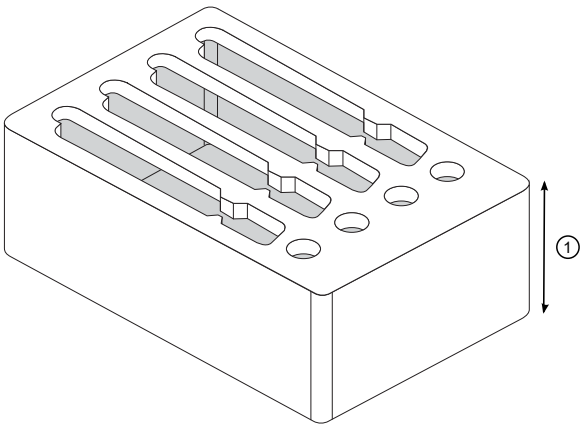
Proceed to “Centrifuge the library and templating reagent strips”.

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 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.

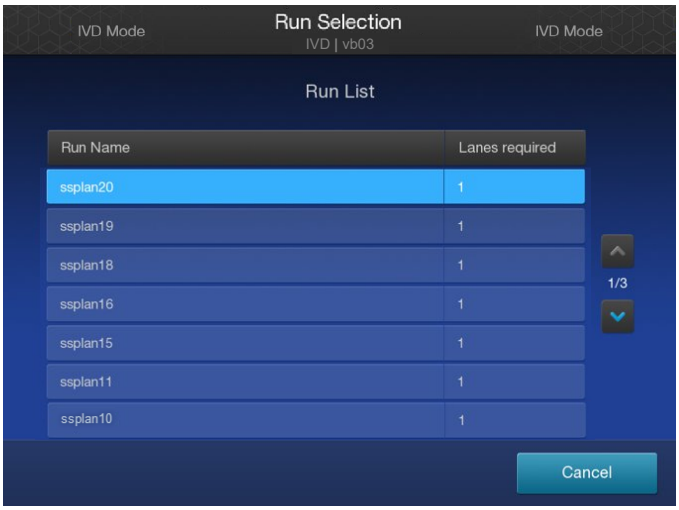
   Note: Alternatively, verify well reagent volumes by comparing fill levels with the Genexus™ Dx strip figures provided on page 227.
5. Repeat step 1 through step 4 for the remaining library and templating strips to be loaded for a sequencing run, if needed.
6. After centrifugation, return Genexus™ Dx 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 227.

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.

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3. In the **Review Run** screen, confirm the run and assay selections, then tap **Next**.



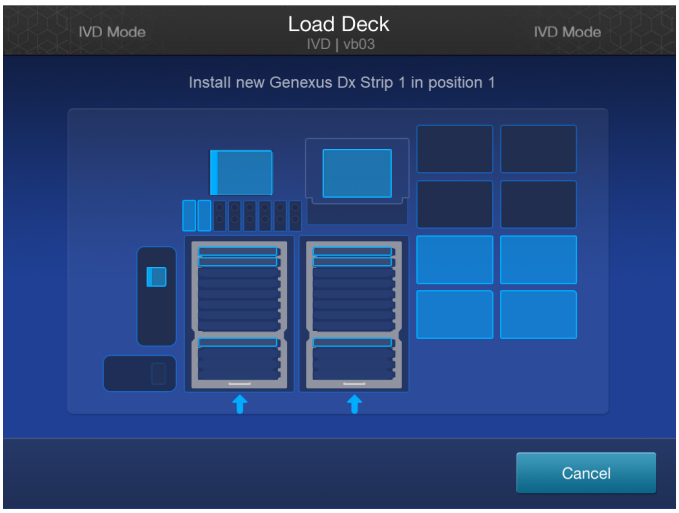
The deck door opens automatically.

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.

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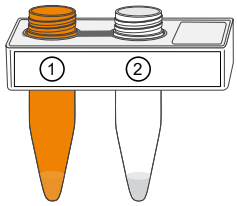
4. In the **Load Deck** screen, the sequencer instructs you step by step to load each required consumable and sample plate in a highlighted position on the deck. The sequencer detects the loading of each consumable in real time and advances to the next component automatically.



IMPORTANT!

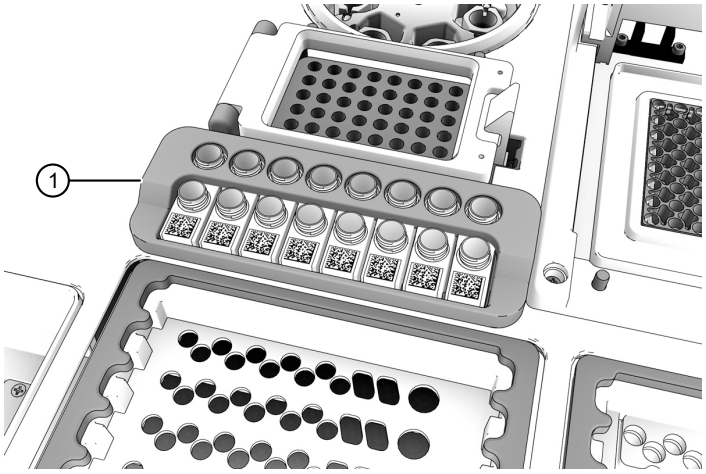
- Remove the primer pool tube caps before installing the tube carrier on the deck.
- Load the correct type of barcode plate, library strip, or template strip for the type of run you are setting up. The sequencer displays a warning if you have installed consumables that are incompatible with the run you 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. For Nucleic Acid to Report and Sample to Report sequencing runs that require loading of primer pool tubes, place the Genexus™ Primer Pool Tube Holder on top of the Oncomine™ Dx Express Test Panel tubes, lining up the holes in the tube holder to the tops of the primer tubes.



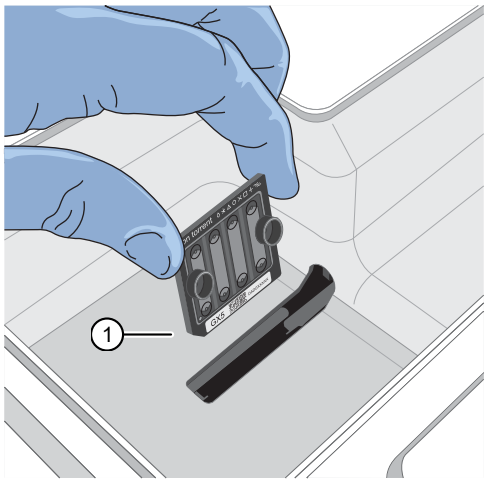
① Genexus™ Primer Pool Tube Holder

6. 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.

7. 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.





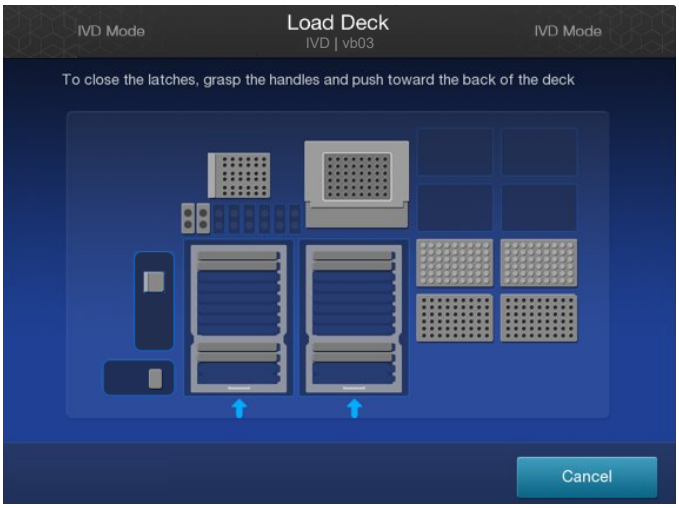
① Notched corner of chip

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.

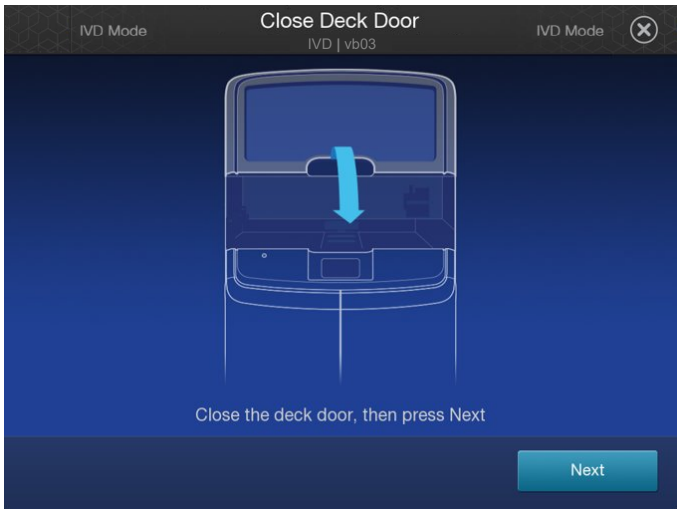
8. When the deck consumables and sample plate have been loaded, lock the library and templating strips in place by sliding the latches toward the rear of the deck.

Note: Load the sample plate last.



If a chip is detected and the strip latches are closed, the **Close Deck Door** screen appears.

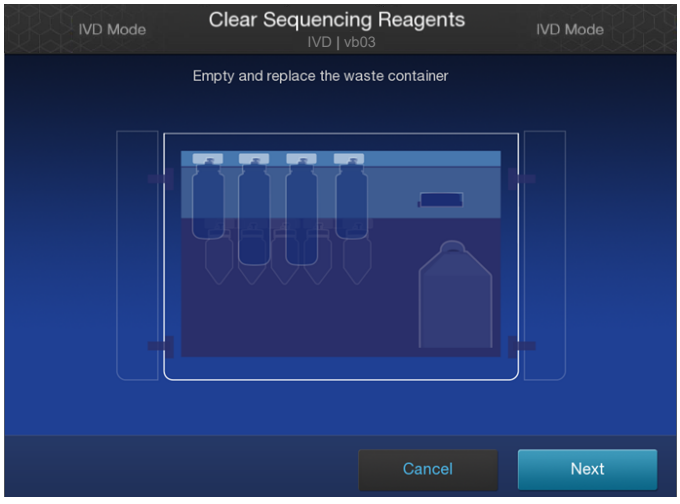
9. Close the deck door, then tap **Next**.



- 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 10.
- 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.

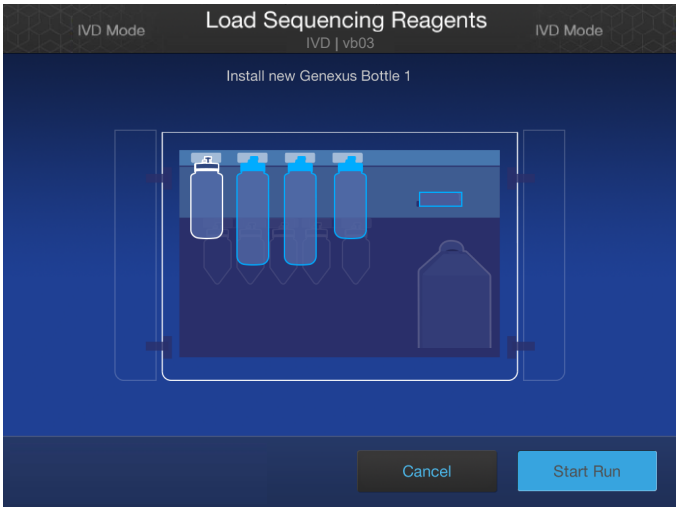
10. Follow on-screen instructions to empty the waste in the Waste carboy, remove waste pipette tips, remove the used Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2, Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge, then tap **Next**.



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.

11. Install a new Genexus™ Dx Bottle 1, Genexus™ Dx Bottle 2 (two required), Genexus™ Dx Bottle 3, and Genexus™ Dx Cartridge.

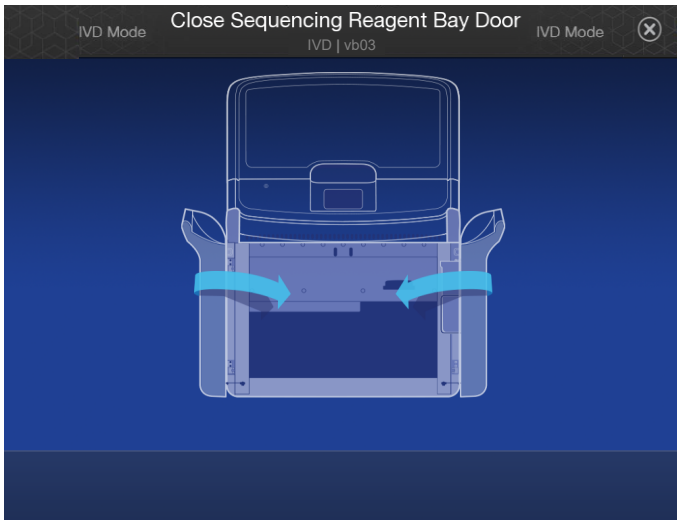


IMPORTANT!

- 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 on the sequencer should be used within 14 days of installation for optimal run performance.

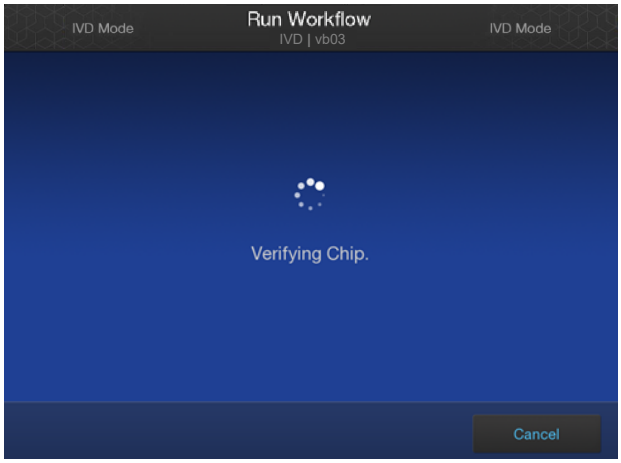
After reagents have been installed, the **Close Sequencing Reagent Bay Door** screen appears.

12. Close the sequencing reagents bay doors.

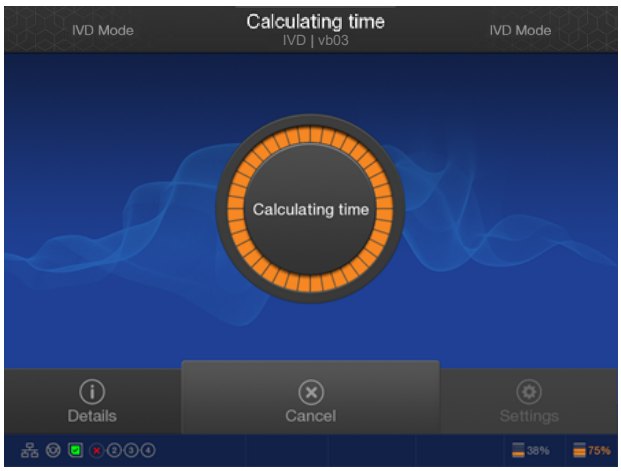


After the doors are closed, the sequencer automatically starts the run.

At the beginning of the run, the instrument verifies the chip, checks for leaks, then calculates run time.







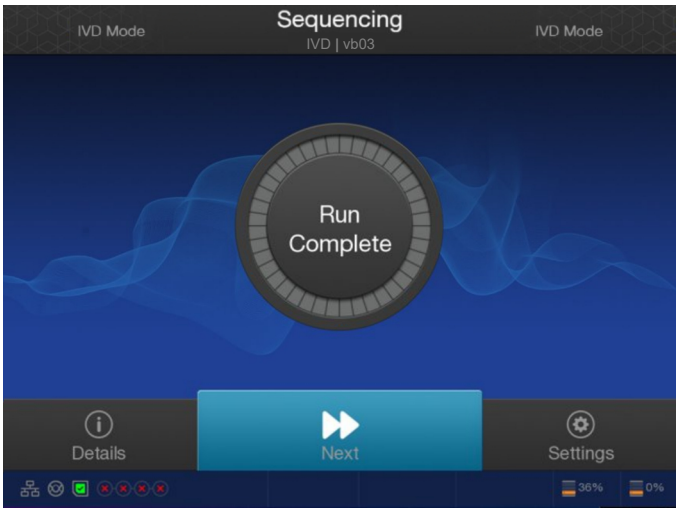
A sequencing run encompasses the following stages:

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.

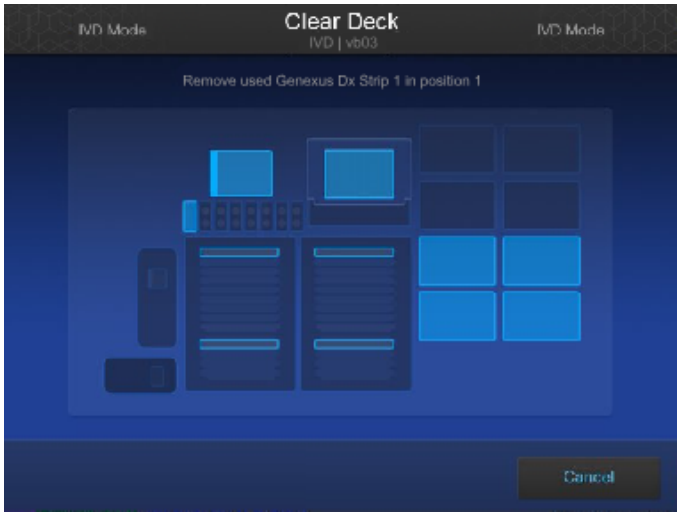
Clear the instrument deck and perform a UV Clean

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 and the sequencer if libraries are recovered for reuse. Store the sealed library plate at 4–8°C for up to 1 month in a post-amplification area.

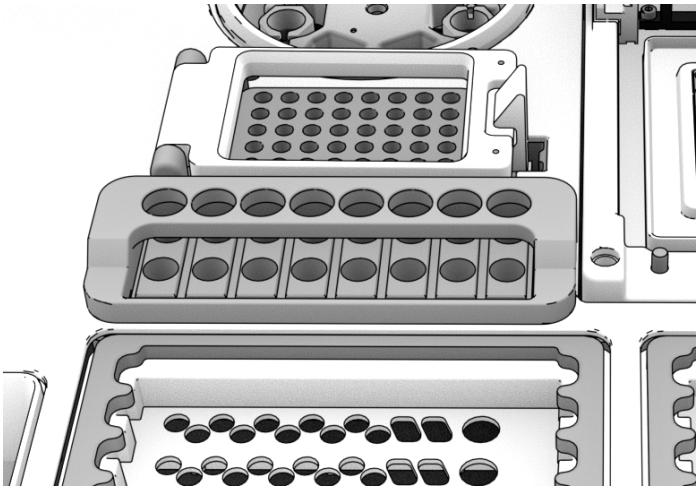


3. If panel primer tubes were used, remove the Genexus™ Primer Pool Tube Holder and discard the primer pool carriers from the deck.

   Note: If the Genexus™ Primer Pool Tube Holder shows reagent droplet accumulation from normal operation, it should be cleaned before proceeding. For more information, see “Clean the Genexus™ Primer Pool Tube Holder” on page 215.

4. Place the Genexus™ Primer Pool Tube Holder back on top of the Primer Pool Tube Station on the deck and store it there until next use.

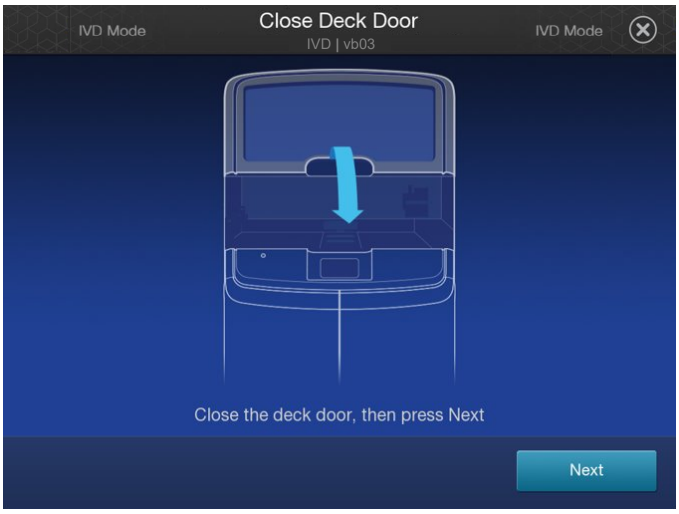
   IMPORTANT! Leave the Genexus™ Primer Pool Tube Holder in this location for the **UV Clean** step and to help reduce contamination.



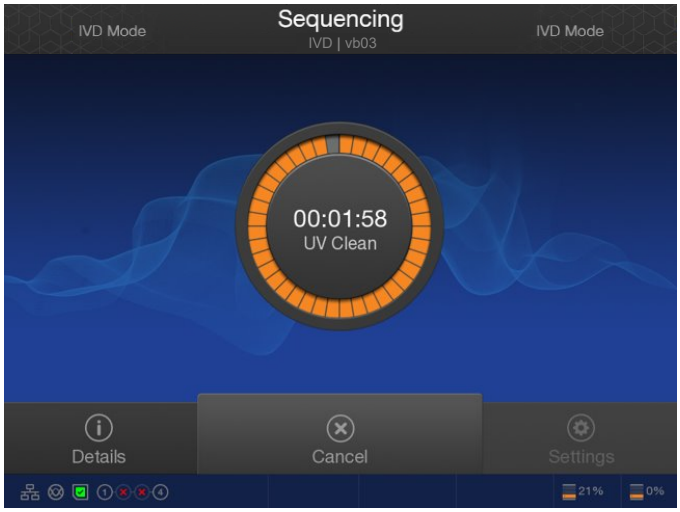
5. Inspect the Genexus™ Dx Filter in the liquid waste disposal port and verify that no standing liquid or debris is present.

   - 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 216.
   - If standing liquid is present, contact Technical Support and report a possible filter or deck liquid waste line clog.

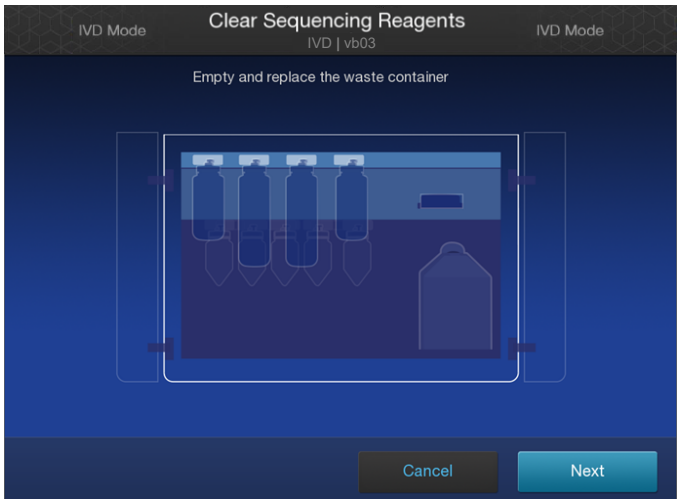
6. When finished, close the deck door, then tap **Next**.



A two-minute **UV Clean** starts.



7. 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 be required 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 216.

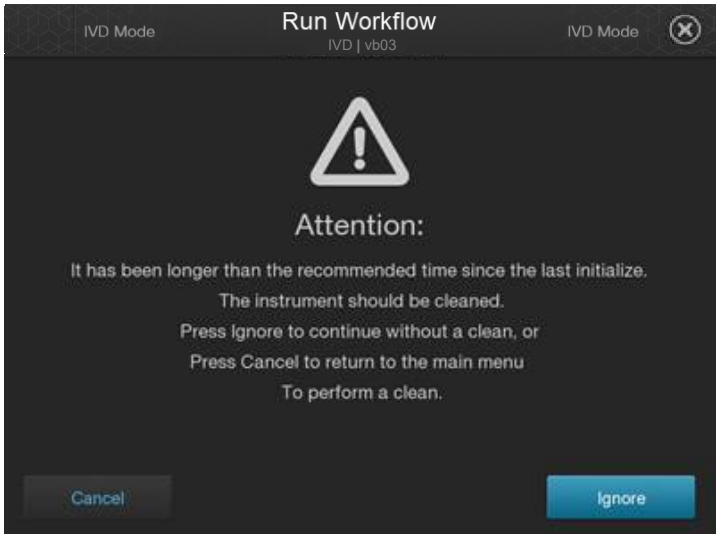
IMPORTANT! Follow all applicable local, state/provincial, and/or national regulations when recycling or disposing of Genexus™ Integrated Sequencer consumables and liquid waste.

CAUTION! The Genexus™ Dx Bottle 1 (small waste bottle) contains small amounts of formamide. Dispose of this waste appropriately.

8. 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

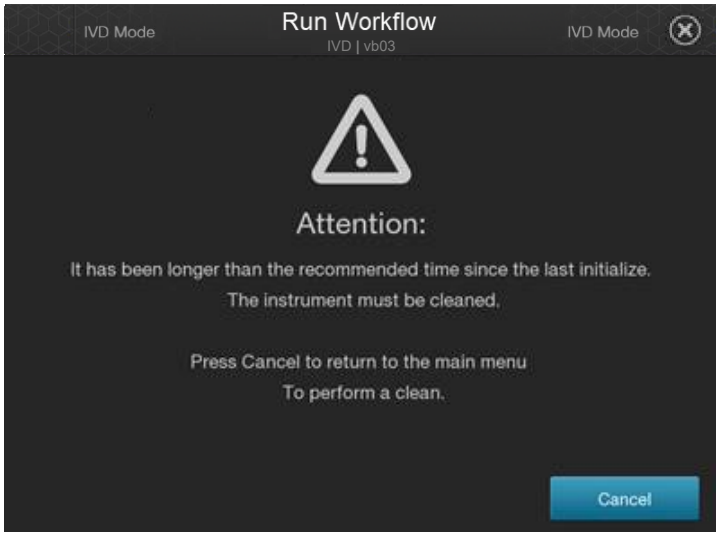
A sequencer initialization is defined by the installation of a new chip, chip coupler, sequencing bottles, and reagent cartridge on the sequencer before a run. Reagents are stable on the sequencer for 14 days, after which you can experience reduced performance. After 14 days, the following on-instrument 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:



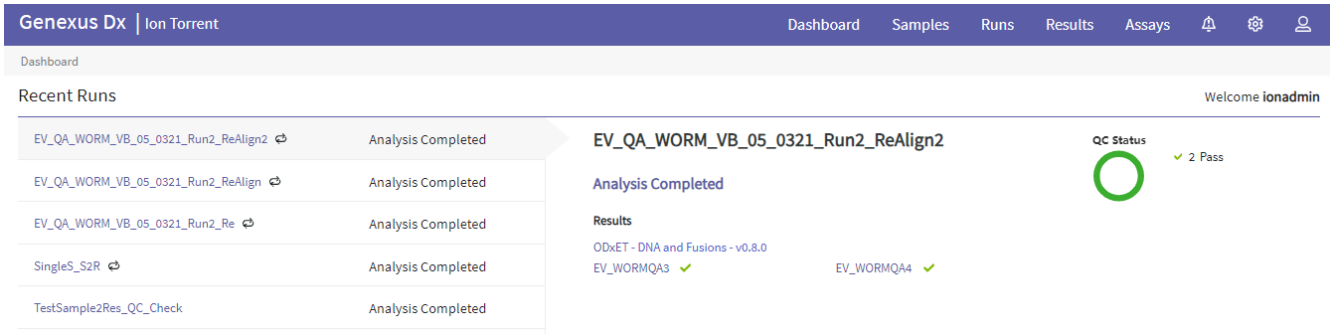
Tap **Cancel** to return to the main menu to perform an instrument clean.

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.

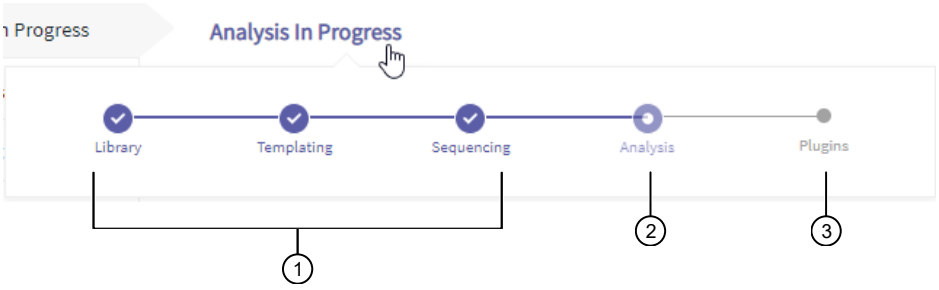
Runs that are reanalyzed are listed with ⟲ **(Reanalysis)** after the run name. For information on reanalyzing runs, see the *Oncomine™ Dx Express Test Part III: Genexus™ Dx Software 6.8 Reference User Guide* (Pub. No. MAN1001776), or the software help system.



1. In the menu bar, click **Dashboard**.
2. In the **Dashboard** screen, in the **Recent Runs** section, view information for recent runs.

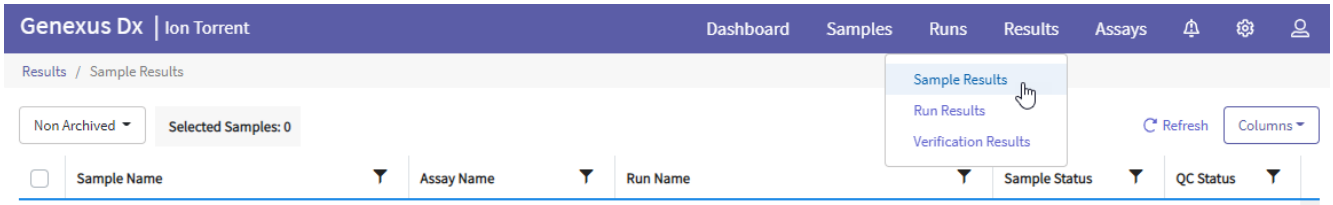
View option	Description
View detailed run result information for completed runs.	In the left side of the screen, click the run name of interest. The **Run Summary** tab opens. For more information, see “The run summary” on page 150. In the menu bar, click **Dashboard** to return to the **Dashboard**.
View the quality control information.	In the left side of the screen, click the row of the run of interest. 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 144.
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. 2. In the run details pane in the right side of the screen, click the assay name of interest. The **Assay Metrics** tab opens. For more information, see “Assay metrics and the run report” on page 153. 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. 2. In the run details pane in the right side of the screen, click the sample name of interest. The **Key Findings** tab opens. For more information, see "Key Findings" on page 126. 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. 2. In the run details pane in the right side of the screen, place the pointer over the status for the run. 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.  ① Completed steps are shown with a check mark. In this example, **Library** preparation, **Templating**, and **Sequencing** are complete. ② Steps that are in progress are shown with a flashing circle. In this example, **Analysis** is in progress. 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 122
- Review sample results 124
- Review run results 148
- Clinical Test Report and Clinical Laboratory Report 157
- Reanalyze a run or sample 166



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.

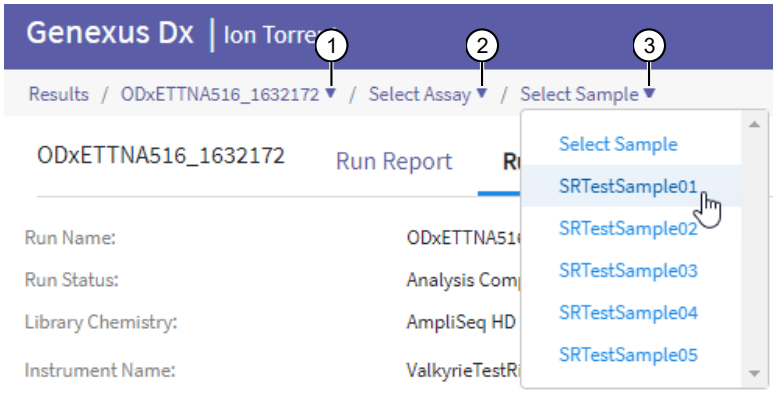
Selection	Description
Click **Results** ▸ **Sample Results**	Select this option to review completed sample results and reports.
Click **Results** ▸ **Run Results**	Select this option to review completed run results and reports by assay.
Click **Results** ▸ **Verification Results**	Select this option 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.

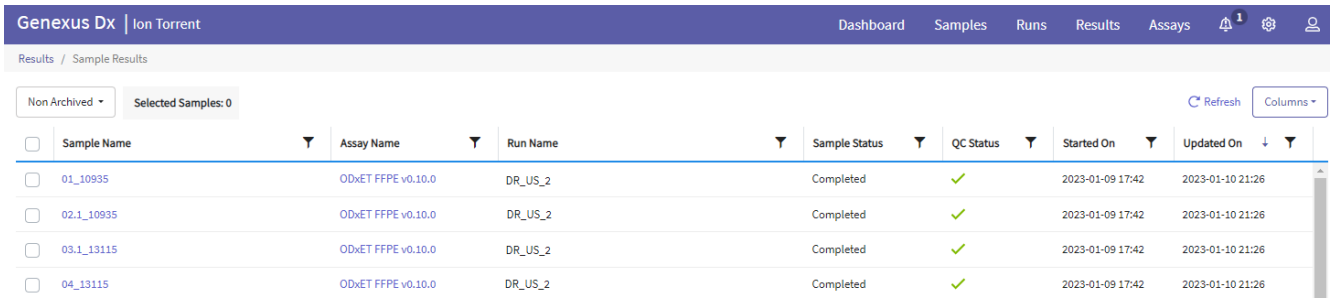


Callout	Dropdown menu	Description
1	**Run name**	The run name is listed. Multiple runs are listed only if the run has been reanalyzed.
2	**Select Assay**	You can click the assay name of interest to view assay metrics for the run. You can search for or toggle between different assays used in the run. If only one assay is associated with the run, only one assay name is listed. 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 153.
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. 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 153.

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. • ✔ (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.
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.

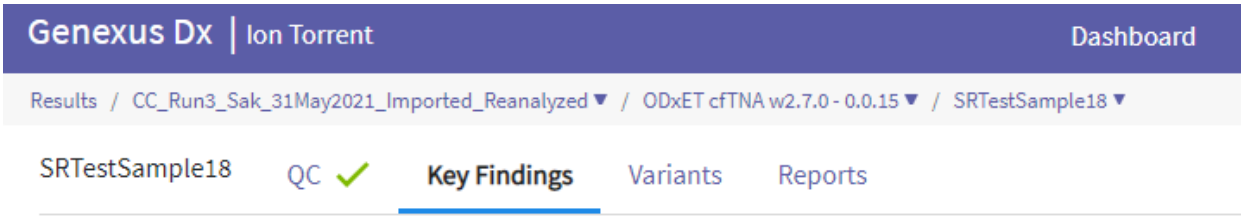
Option	Description
Actions	Available action links for a sample are shown when you place the pointer over the row of a sample. • **Report**—Download the report (available only for samples with a sample status of completed). • **Assign Tags**—Add or remove the tags assigned to a sample result. • **Audit**—View the audit trail for the run. • **Notes**—View, add, or delete notes for sample result. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. • **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. **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. MAN1001776), 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.
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

Biomarker level[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 161.

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 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

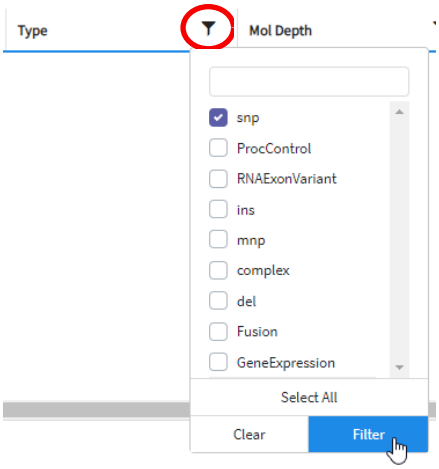
Variant type	Description
SNVs/Indels	• **SNVs**: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included. • **Indels**: insertion and deletion variants. For more information, see “View SNV/Indel results” on page 128.
Fusions	Translocations of genetic material and splicing variants. For more information, see “View fusion results” on page 130.
CNVs	Copy number variations (CNVs) are variations of the number of copies of a given gene. For more information, see “View CNV results” on page 135.

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 128.

Column	Description
Variant ID	The name of the hotspot as defined in the Browser Extensible Data (BED) file. 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. • **snp** (single nucleotide polymorphism) • **mnp** (multi-nucleotide polymorphism) • **ins** (insertion) • **del** (deletion) • **complex**
Test Result	Indicates the presence or absence of an SNV/Indel variant. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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). 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 128.

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. • **POSITIVE**—indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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. **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. Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline) where Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples) • 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample. • 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample. Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline) • 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene. • 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.

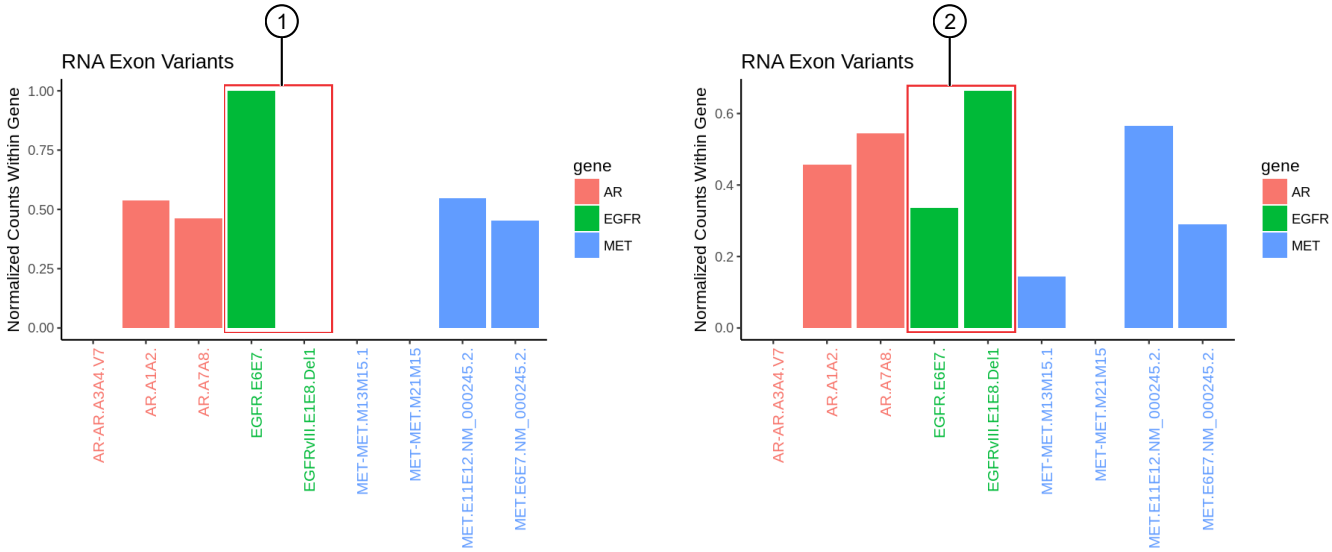
View RNA Exon Variants

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 "View RNA Exon Tile Fusion Imbalance" on page 133.

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 130.
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 list.

View RNA Exon Tile Fusion Imbalance

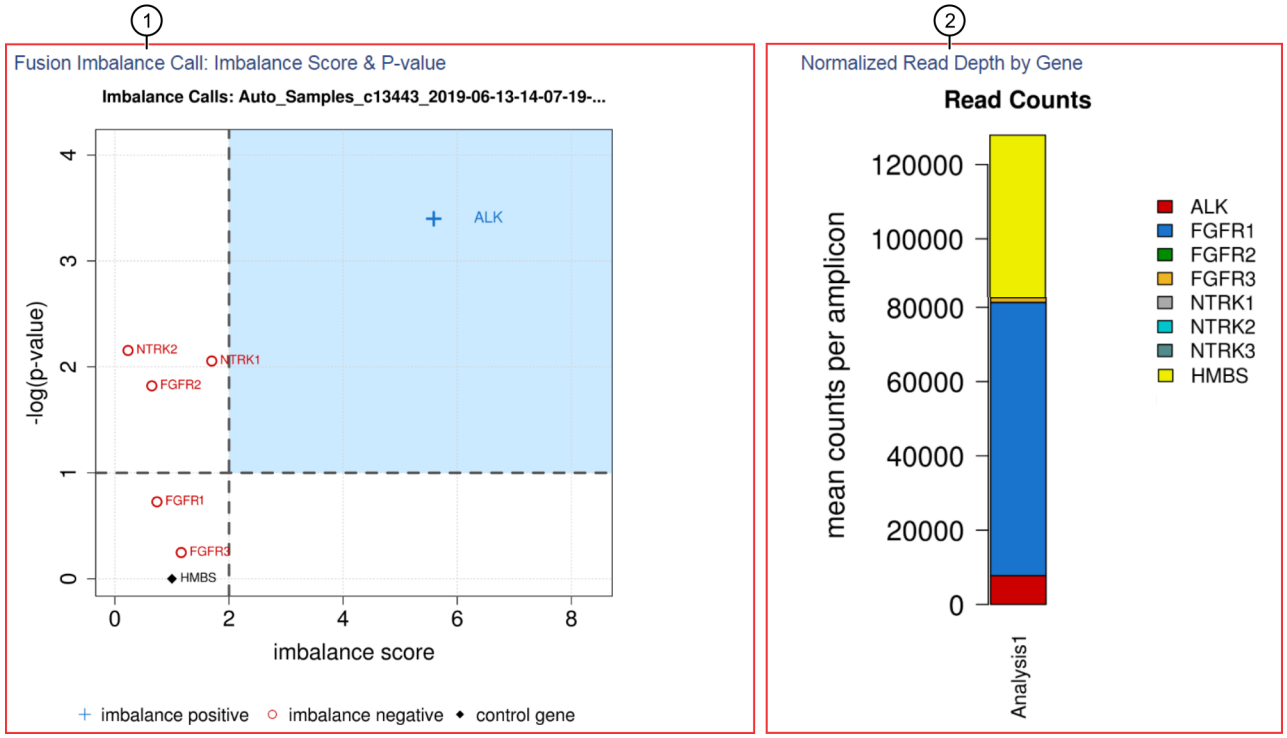
The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

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*.

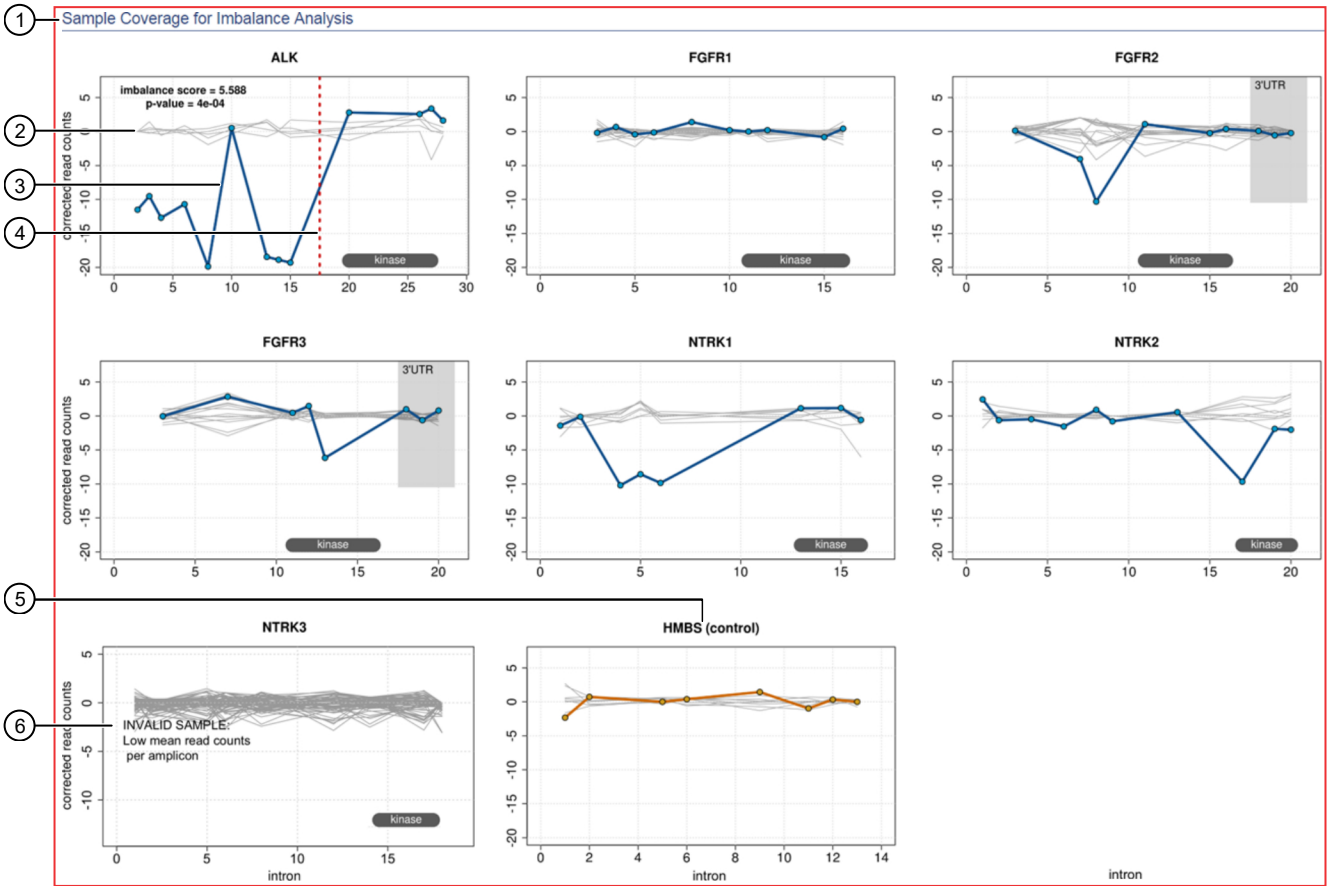
1. In the menu bar, click **Results** ▸ **Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see "Fusions table" on page 130.
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 list.

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 128.

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. 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. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds. • **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. • **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 138.

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**. This option is available only when the forward strand is shown.
View the forward stand	Click **Forward**. This option is available only when the reverse strand is shown.
View the reference sequence translation	Click **Three-Frame Translate**. This option is available only when the amino acid sequence is not shown.
Hide the reference sequence translation	Click **Close Translation**. 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.

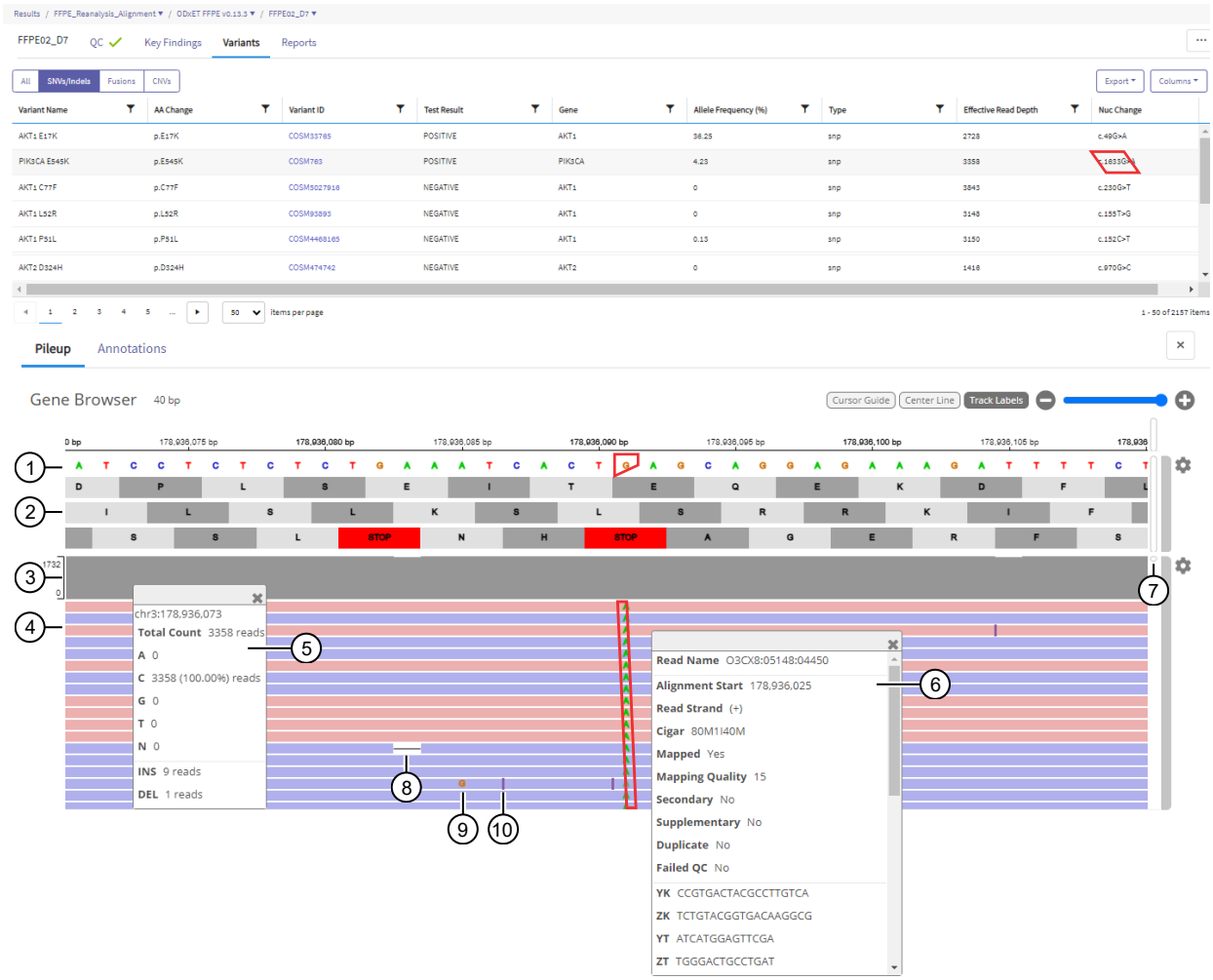
1. In the menu bar, click **Results** ▸ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

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**. 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**. 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 138.
Show all the bases in the BAM tracks	Click **Show all bases**. 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**. b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks. 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 137. 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 138.



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 138.

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.

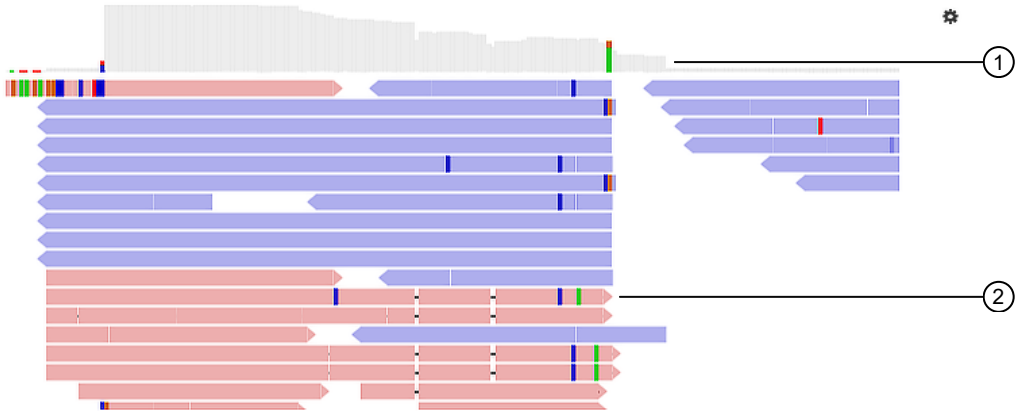
The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.



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.



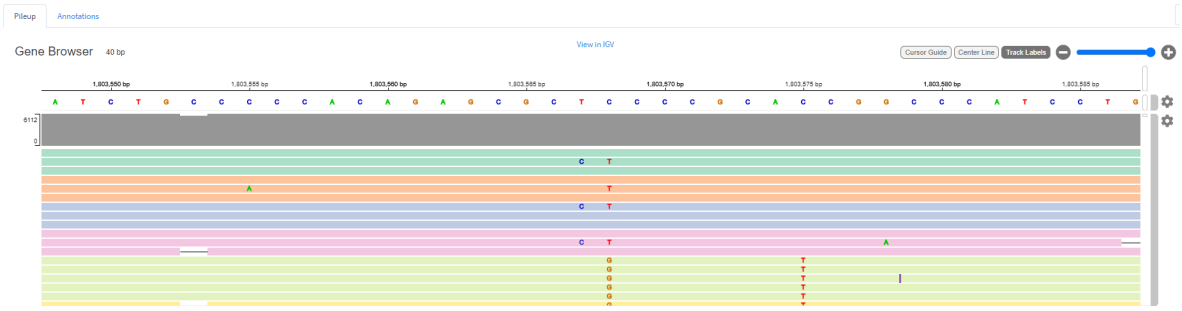
① Read depth
② Read location and direction of variants

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 138.
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.

Identification of potential positive control–derived variants in patient samples

Positive control (PC) and no-template control (NTC) are added to the sample plate manually by the operator before loading the sample plate to the Genexus™ Dx Integrated Sequencer. Although rare, it is possible that the positive control may contaminate the test samples or NTC during the workflow and specifically during the plate set up. These events are uncommon and typically arise from deviations from the user guide recommendations and best laboratory practices for preventing contamination.

During assay validation, the risk of carry-over or cross-contamination from positive controls was evaluated and determined to be low. However, as with any highly sensitive molecular workflow diagnostic assay, users should remain attentive to unexpected results that can indicate positive control contamination in patient samples.

The following observations can help identify potential positive control contamination in patient sample results:

- detection of multiple actionable variants that are rarely observed together, or
- detection of two or more of the positive-control variants in otherwise negative patient samples.

When reviewing data, operators should look for the variant patterns described above and consider factors such as the proximity of the sample to the positive control on the plate, the order in which samples and positive controls were plated, and whether other samples in the run show similar results. If positive control contamination is suspected, we recommend that you repeat the affected sample or, when feasible, the entire run.

Note: The occurrence of positive control–associated variants in sample results is rare and does not impact overall assay reliability when standard laboratory practices are followed. To minimize the risk of aerosolization and transfer of positive control material to patient samples, use a unidirectional workflow, perform sample and control plating in a PCR hood, and use dedicated pipettes for positive control handling. For additional guidance on contamination-prevention best practices, see “Guidelines for preventing contamination” on page 31.

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 144.

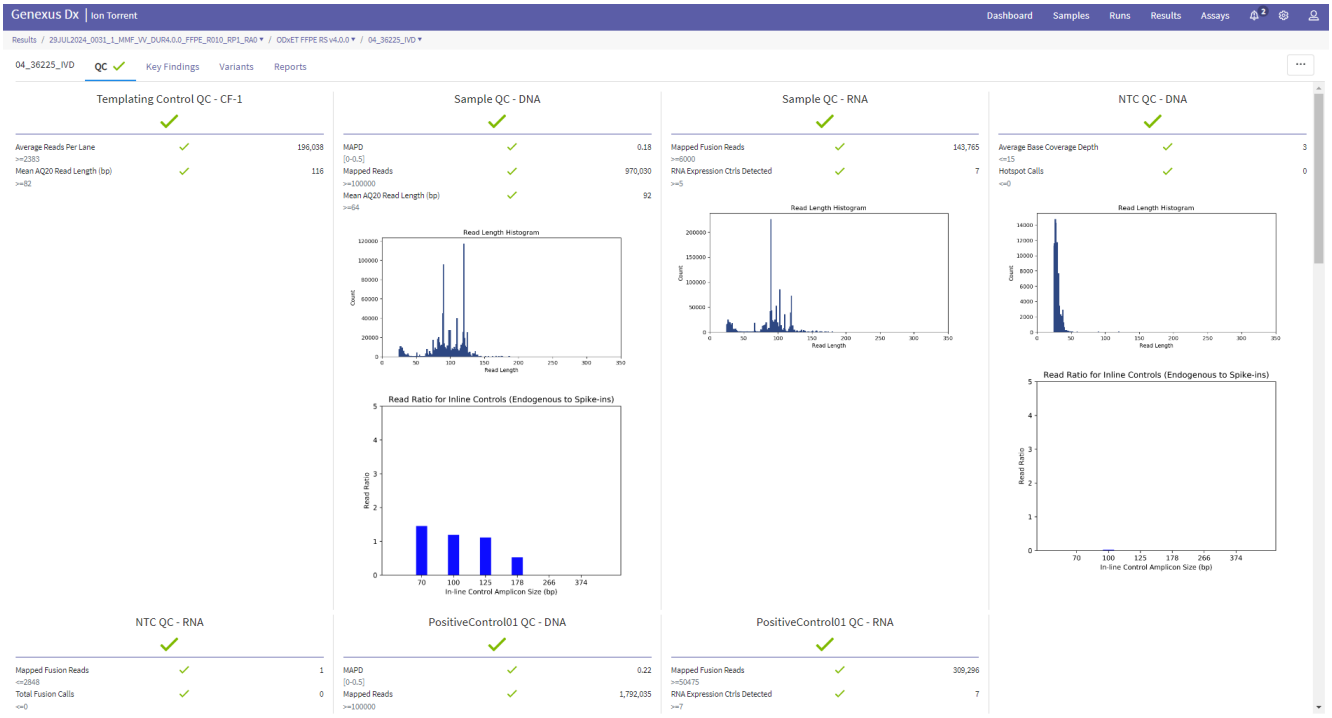
QC results

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.
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.

(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**.
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.
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.

(continued)

Metric	Description
<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**. 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. MAN1001776).
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.
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. MAN1001776).
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. • **Delete**—Delete the run. • **Audit**—View the audit trail for the run. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. • **Assign PCR Plate**—Enter a unique identifier for the 96-well plate used for library preparation and templating.

(continued)

Option	Description
Actions	These actions are available only for Sample to Result runs. • **View Plan**—View detailed run plan information. • **Assign Kit Barcodes**— • **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**. • **Clear**—Deselect the selected run or runs. • **Retain Data**—Click to keep run data regardless of disk cleanup settings. To undo, select **Release Data**. • **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. • **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**. 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. MAN1001776).
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**. 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. 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. 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. The status can be **N/A**, **Planned**, **Review**, **Started**, **Aborted**, or **Completed**.
Archive Position	The sample positions in the archive plate. 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. • ✔ (Checkmark) – indicates that the sample is selected for sequencing. • ⊘ – (No icon) indicates that the sample is not selected for sequencing. • **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.

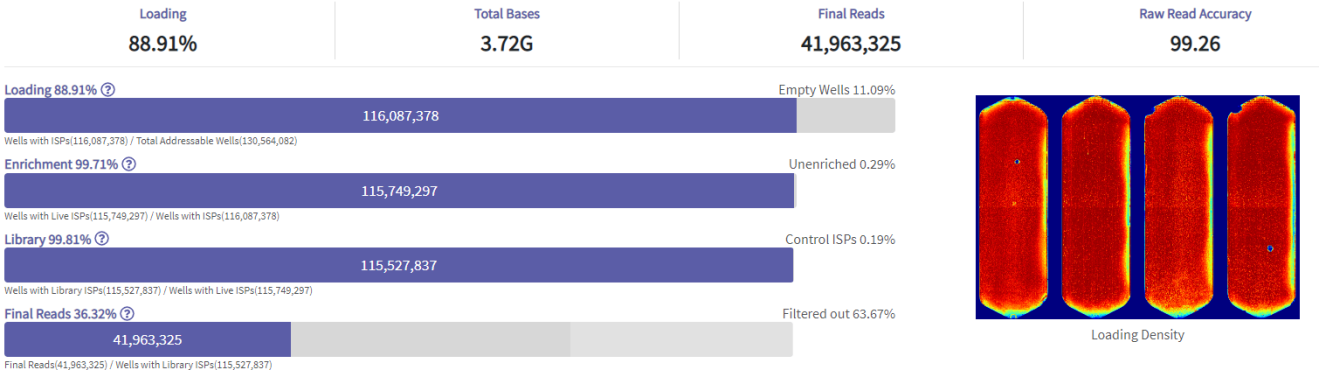
For runs with only one assay, the run report and assay metrics show the same information for assay metrics. In addition to the metrics for the assay, the run report shows information about the run and the instruments used in the run.

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.

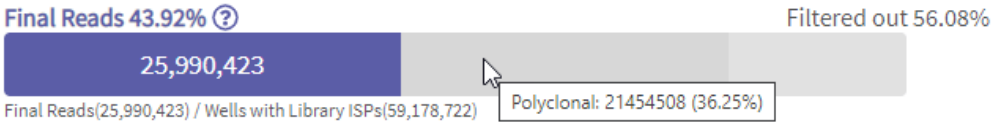
The **Run Report** is assay-specific and cannot be viewed within the sample results screens. To view the **Run Report**, ensure that **Select Sample** is selected in the **Select Sample** dropdown menu.

Assay metrics and the run report

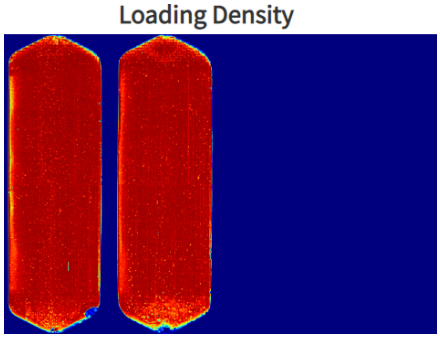
The run report provides detailed information, such as chip metrics for the run, well, and Ion Sphere™ Particles (ISPs) statistics. For runs with multiple assays, metrics are provided for each assay in the run. Sequencing metrics are shown at the top of the screen, followed by sample-specific metrics 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. To view polyclonal metrics, mouse over the first low quality portion (gray) of the **Final Reads** bar plot. 

Run Report *(continued)*

Metric	Description
Low Quality	Loaded wells with a low or unrecognizable signal. To view polyclonal metrics, mouse over the second low quality portion (gray) of the **Final Reads** bar plot. 
Adapter Dimer	Loaded wells with a library template of an insert size less than 8 bases. 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. 
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. 

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 153. 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 163.

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: • **Level 1: Companion Diagnostics (CDx) Biomarkers** • **Level 2: Cancer Mutations with Evidence of Clinical Significance** • **Level 3: Cancer Mutations with Potential Clinical Significance** If a detected variant is listed in the **Companion Diagnostics (CDx) Biomarkers** category, 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 161.
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 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 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. 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: • **Level 1: Companion Diagnostics (CDx) Biomarkers** • **Level 2: Cancer Mutations with Evidence of Clinical Significance** • **Level 3: Cancer Mutations with Potential Clinical Significance** If a detected variant is listed in the **Companion Diagnostics (CDx) Biomarkers** category, 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 161.
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 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 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. • **NTC QC – RNA**—A summary of the RNA no-template control quality control metrics. • **Sample QC – RNA**—A summary of RNA sample quality control metrics. • **Sample QC – DNA**—A summary of DNA sample quality control metrics. • **Templating Control QC – CF-1**—A summary of the CF-1 quality control metrics. • **Positive Control01 QC – DNA**—A summary of the DNA positive control quality control metrics. • **Positive Control01 QC – RNA**—A summary of the RNA positive control quality control metrics. • **Purification QC—A summary of QC metrics for the purification run.** This section is locked and cannot be customized.
Sequencing Run Details	Contains the following subsections: • **Assay Name**—The assay used in the run • **Panel**—The panel used in the run • **Started On**—The date and time that the run was started • **Completed On**—The date and time when the run was completed • **Operator**—The user who sent the run to the instrument • **Instrument**—The name of the instrument • **Serial Number**—The serial number of the instrument This section is locked and cannot be customized.
Reagents	Details about run consumables, including part numbers, expiration dates, and lot numbers. 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. 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 163.

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.

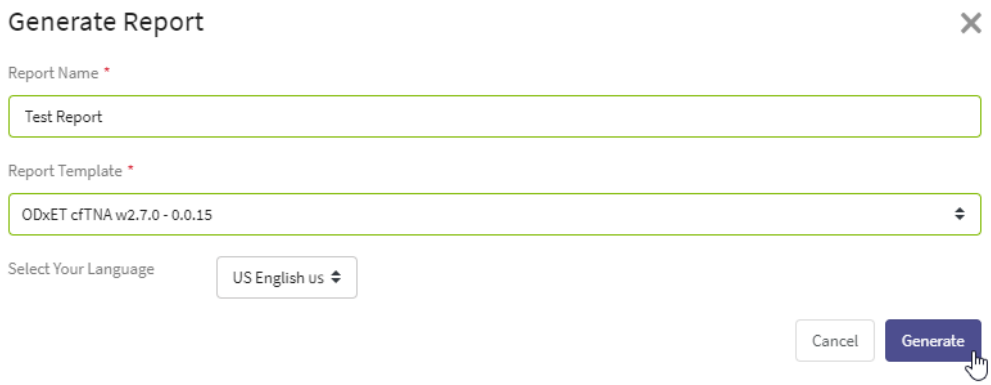
You can generate multiple reports for a sample result, if each report is named uniquely, and is generated in a different language, or uses a different report template.

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.

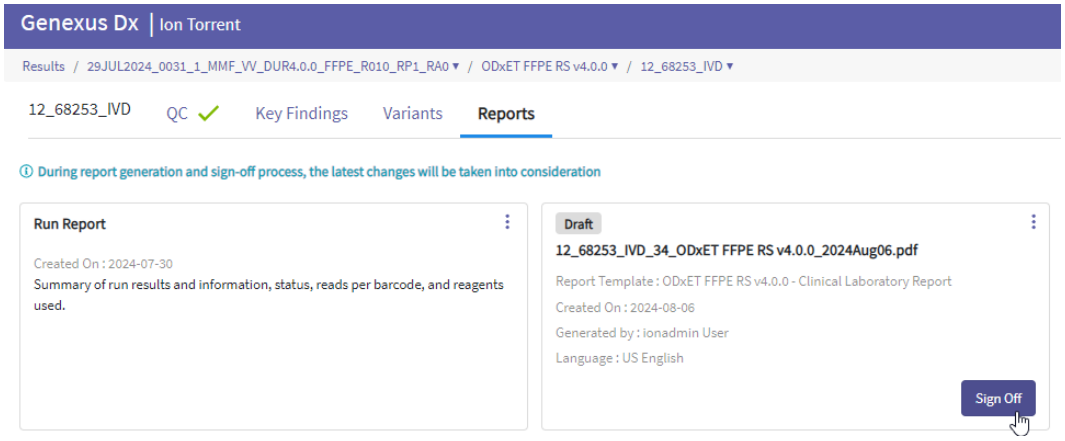
Multiple signatures can be added to the variant report that is used for sign-offs. Account permissions for electronic signatures are associated with a signature type. A lock-level signature type locks the report. If your signature permissions are associated with a lock-level signature type and you sign 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 157. 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. MAN1001776).

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

- Customer Support Archive (CSA) 167
- Genexus™ Dx Purification System 168
- Troubleshoot Genexus™ Dx Integrated Sequencer performance with CF-1 and inline controls 172
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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. 2. Export the Genexus™ Dx Purification System CSA file from the Run Report. 3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting. 4. Restart the run beginning from “Prepare samples”. If the run fails again, contact Technical Support to troubleshoot the problem.
	Power failure occurred during sample purification	**IMPORTANT!** If the power failure occurred during the purification process the samples and reagents cannot be recovered or reused. 1. Record the error message displayed on the instrument touchscreen. 2. Export the Genexus™ Dx Purification System CSA file from the Run Report. 3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting. 4. Restart the run beginning from “Prepare samples”.

Observation	Possible cause	Recommended action
Quantification failure during purification run	Instrument detected an error, or user aborted the run.	1. Record the error message displayed on the instrument touchscreen. 2. Export the Genexus™ Dx Purification System CSA file from the Run Report. 3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting. 4. Manually quantify samples present in the 48-well archive plate (reference the archive plate map for sample order) using the Qubit™ Fluorometer, or equivalent fluorometric method.
Unexpected sample concentration (negative or greater than normal concentration values for multiple samples)	• Erroneous standard measurements were made. • Pipetting error occurred during liquid transfers. • Tip blockage occurred during liquid transfer. • Bubble in Genexus™ Dx 48-Well Nucleic Acid Archive Plate sample well.	Manually quantify samples present in the 48-well archive plate (reference the archive plate map for FFPE sample order) 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 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.

Observation	Possible cause	Recommended action
Lower than expected yield *(continued)*	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 Tube was low.	Ensure that AutoLys Tube caps are securely closed before incubation.
		Allow AutoLys Tube to cool to room temperature before centrifugation.
	AutoLys Tube collapsed during centrifugation.	Ensure that AutoLys Tube caps are securely held by the AutoLys 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 sample input amount. See “Recommended input amount” on page 75.
		Increase the sample input if needed as follows: • FFPE tumor resections—up to 14 × 5-µm curls/slides, or equivalent amount • 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 the protocol for FFPE sample preparation with the AutoLys Tube closely.

Observation	Possible cause	Recommended action
Unexpected sample concentration (negative or greater than normal concentration values for multiple samples) **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 sample order). 2. Manually quantify samples present in the 48-well archive plate (reference archive plate maps for FFPE sample order) 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 the protocol for FFPE sample preparation with the AutoLys Tube 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 173.

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 Read ratio ~1–3	Low 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** **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 **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 199. After cleaning, start a new run. **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 **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 172.	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 **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 172.	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 104.
	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 **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 172.	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 104.

Observation	Possible cause	Recommended action
The number of sample reads is low, and CF-1 metrics fail QC **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 172. *(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 199. 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. **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 118.

<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

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.

1. Follow the procedure described in “Recover the pooled sample libraries” on page 225 to recover the pooled sample libraries from well E4 of the PCR amplification plate after the run.
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.

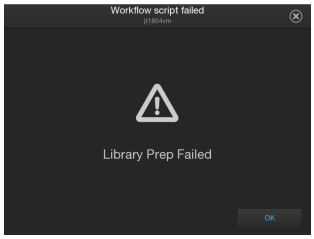
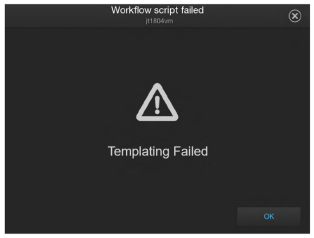
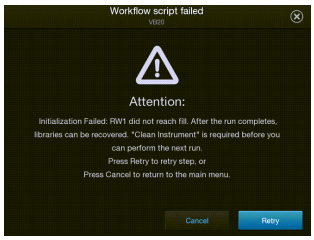
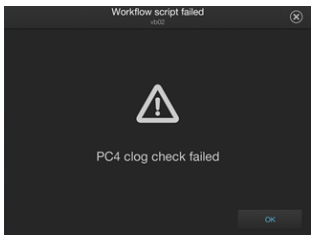
<table><tr><th>No. of lanes in run</th><th>Volume of pooled library</th><th>Volume of water or Low TE</th><th>Total volume in well A1</th></tr><tr><td>1</td><td>17.5 µL</td><td>32.5 µL</td><td>50 µL</td></tr><tr><td>2</td><td>28 µL</td><td>52 µL</td><td>80 µL</td></tr><tr><td>3</td><td>38.5 µL</td><td>71.5 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>
3. Pipet the contents of well A1 up and down 4–5 times to mix.
4. Plan and perform a Library to Result run with the diluted library.

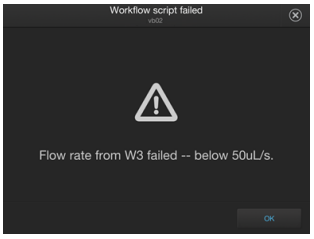
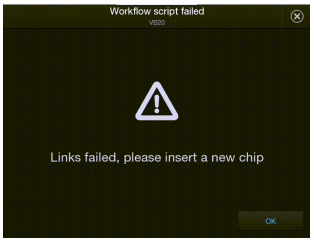
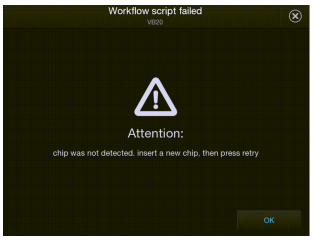
For more information, see “Recover pooled sample libraries and plan a Library to Result run” on page 223.</td></tr>
<tr><td>Some sample QC categories show N/A, and the Sample Status in the Sample Results screen shows "AnnotatorActor: FAILED"

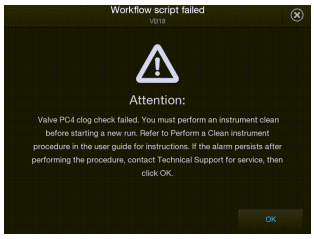
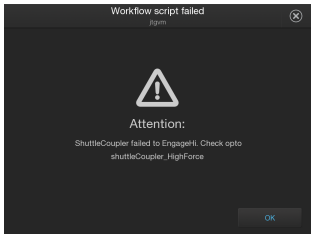
Details: Reanalysis does not rescue the sample.</td><td>A rare error occurred in the variant caller and caused the variant caller and downstream annotator actor to fail.</td><td>Repeat sequencing with the same nucleic acid sample (Nucleic Acid to Result run).

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 223.</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>
<tr><td>Panel primer tubes lifted from the Genexus™ Dx Primer Pool Tube station</td><td>The tubes became loose.</td><td>Use the Genexus™ Primer Pool Tube Holder to secure the tubes in place. For information about using the Genexus™ Primer Pool Tube Holder, see “Load the sequencer and start a run” on page 106.</td></tr>
</table>

Genexus™ Dx Integrated Sequencer error and warning messages

Observation	Possible cause	Recommended action
Error message: **Library Prep Failed** 	• Hardware error during pipette movement. • The sequencing chip failed to engage at pipette position.	Contact Technical Support for recommended action.
Error message: **Templating Failed** 	• The sequencer failed xy homing, *or* • 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. 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 223. **Note:** Diluting leftover pooled library more than 2-fold can result in low reads.
Error message: **Initialization Failed: XXX did not reach fill** 	Genexus™ Bottle 2 flow was restricted, or a flow sensor malfunctioned. OR During a post-chip clean, the Genexus™ Bottle 3 was empty, or Genexus™ Bottle 3 flow was restricted. **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. **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 223.
Error message: **XXX clog check failed** 	Clog check following Clean Instrument failed. Incomplete fluid flush or debris has caused a clog. **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** 	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 199. 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 199. You may or may not need to replace the chip during deck set up.
Error message: **Links failed, please insert a new chip** 	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** 	Chip connection was lost and sequencing failed.	1. Insert a new chip, then press **Retry**. 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. 3. Force an instrument clean by navigating to **Settings** ▸ **Clean instrument**. 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** 	The sequencing clamp hardware did not operate properly.	Instrument service is required to proceed. **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 223.
Error message: **Valve PC4 clog check failed** 	A clog was detected during the post-chip clean.	1. Perform an instrument clean by navigating to **Settings ▸ Clean instrument**. 2. After cleaning completes, begin a new run. 3. If the error message appears again, contact Technical Support for service.
Error message: **ShuttleCoupler failed to EngageHi** 	Chip shuttle hardware did not operate properly.	Instrument service is required to proceed. **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 223.
Warning message: **Pressure Leak Detected in Vac: vac pressure drop leakrate** **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** **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.

Observation	Possible cause	Recommended action
Warning message: **Pressure Leak Detected in Vac: vac pressure baseline** **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.
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: **Problem detected with coupler in Leak laneX** **Press Ok to open doors and fix the leak.**	Genexus™ Coupler leak was detected.	Remove the Genexus™ Coupler, inspect for issues, then reinstall on the deck. Close the deck door and continue with the run. If the coupler leak message appears again, replace the Genexus™ Coupler, then continue with the run. If the failure persists, contact Technical Support.
Warning message: **Problem detected with coupler in Clog laneX** **Press Ok to open doors and fix the leak.**	A clog in the Genexus™ Coupler or chip was detected.	Remove the Genexus™ Coupler, inspect for issues, then reinstall on the deck. Close the deck door and continue with the run. If the coupler clog message appears again, replace the Genexus™ Coupler and the Genexus™Chip with a new set. Close the door and continue with the run. If the failure persists, contact Technical Support.

Observation	Possible cause	Recommended action
Warning message: **Problem detected with bottles: W21 No or low Flow Press Ok to open doors and fix the leak.**	Unable to flow liquid from the bottle in the second position.	Remove the bottle and check for pinching at the nozzle. Reinstall the bottle securely, then continue with the run. If the flow message appears again, replace the bottle with a new one, then continue with the run. If the failure persists, contact Technical Support.
Warning message: **Problem detected with bottles: W22 No or low Flow Press Ok to open doors and fix the leak.**	Unable to flow liquid from the bottle in the third position.	Remove the bottle and check for pinching at the nozzle. Reinstall the bottle securely, then continue with the run. If the flow message appears again, replace the bottle with a new one, then continue with the run. If the failure persists, contact Technical Support.
Warning message: **Problem detected with bottles: W3 No or low Flow Press Ok to open doors and fix the leak.**	Unable to flow liquid from Genexus™ Dx Bottle 3.	Remove Genexus™ Dx Bottle 3 and check for pinching at the nozzle. Reinstall the bottle securely, then continue with the run. If the flow message appears again, replace with a new Genexus™ Dx Bottle 3, then continue with the run. If the failure persists, contact Technical Support.
Warning Message: **Detected a misplaced tip in tipRackX_tip#**	Tips used during Checking for Leaks step were not ditched properly back to the rack.	Ensure that the tip(s) listed are seated properly in the tip rack. 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.
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
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 209.
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 184
- Genexus™ Dx Integrated Sequencer 195

Genexus™ Dx Purification System

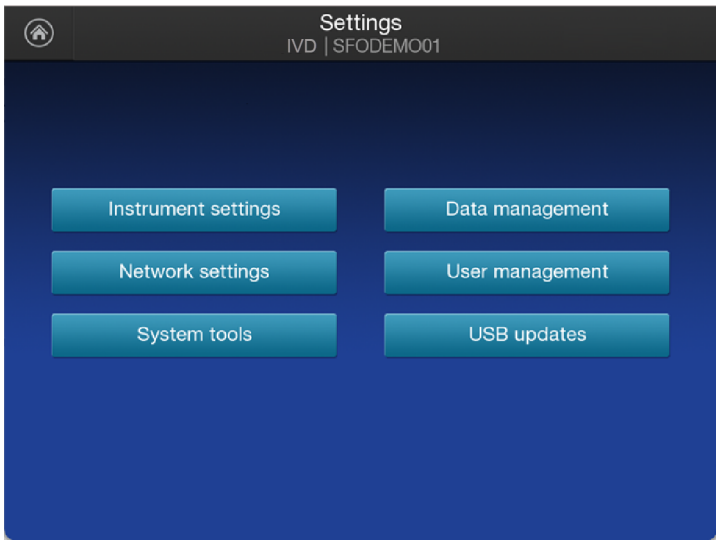
Touchscreen icons



Number	Icon	Description
1		User profile
2		USB – available
		USB – not available
3		Network connectivity – connected
		Network connectivity – not connected

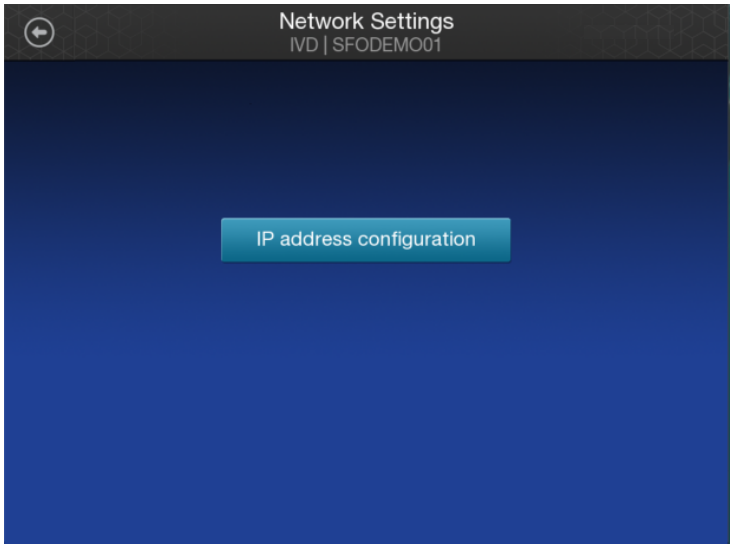
Settings

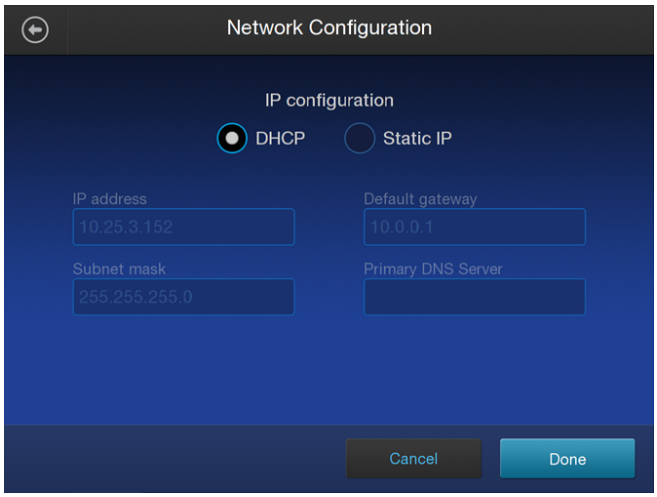
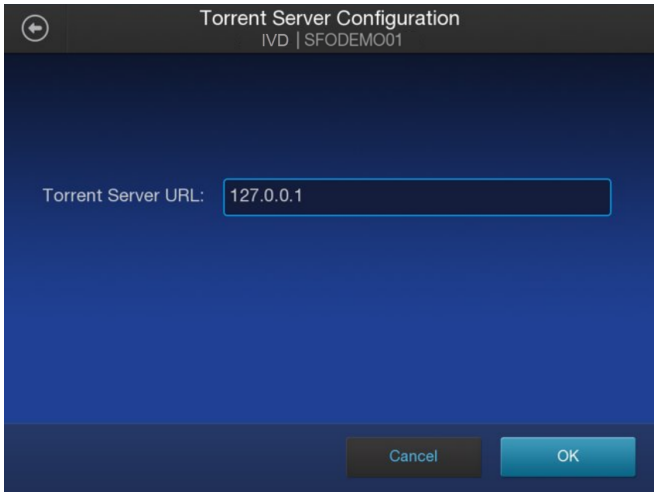
Use the **Settings** menu to view and change instrument settings, and to manage data.



Network Settings

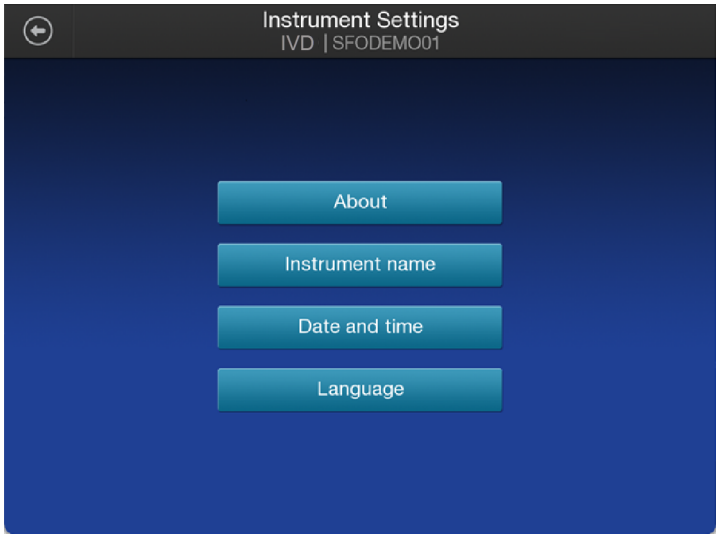
The **Network Settings** menu enables you to configure IP address and Ion Torrent™ Server settings, and update the server.

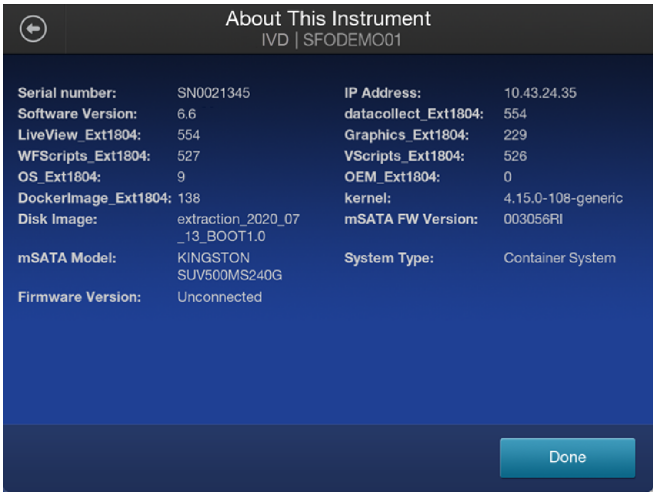


Item	Description	When/How to use
IP Address Configuration	Enables users to set or change the IP configuration (DHCP or Static IP). 	1. Select either the **DHCP** or **Static IP** option. 2. Touch the screen in the field you want to edit to activate the field. A virtual keyboard appears. 3. Enter the new information, then press **Done**.
Ion Torrent™ Server Configuration	Enables users to change the Ion Torrent™ Server IP address and user information.  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. **Note:** Do not change address unless directed to do so by a Thermo Fisher Scientific field service engineer.

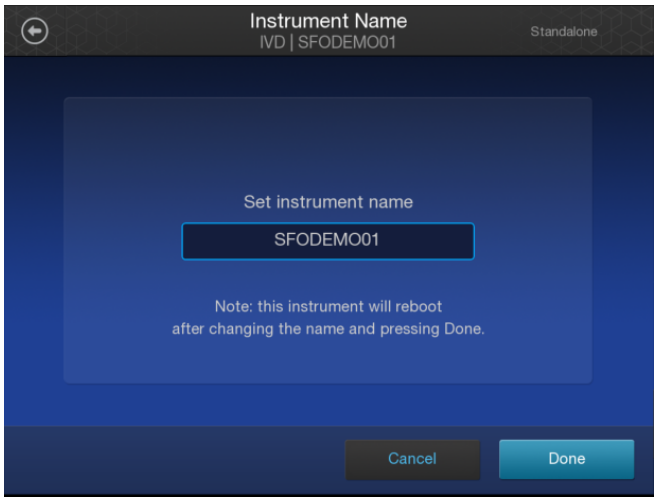
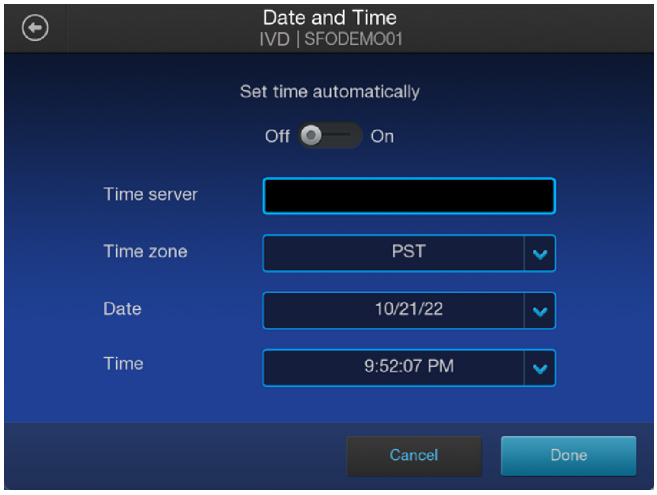
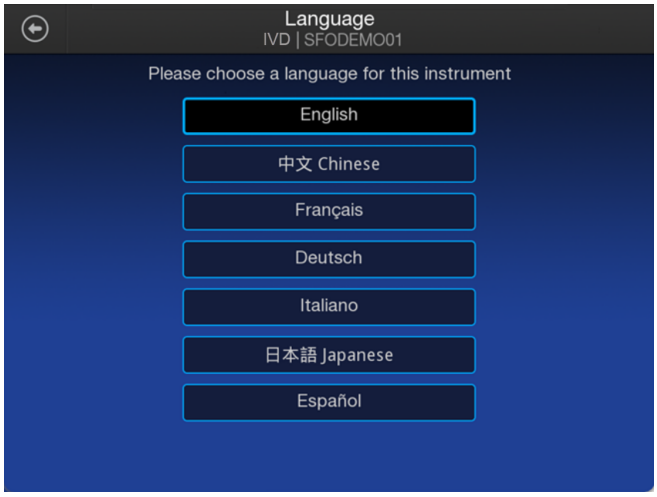
Instrument settings

Use the **Instrument Settings** menu to obtain information about the instrument, to set the instrument name, date and time, and language.



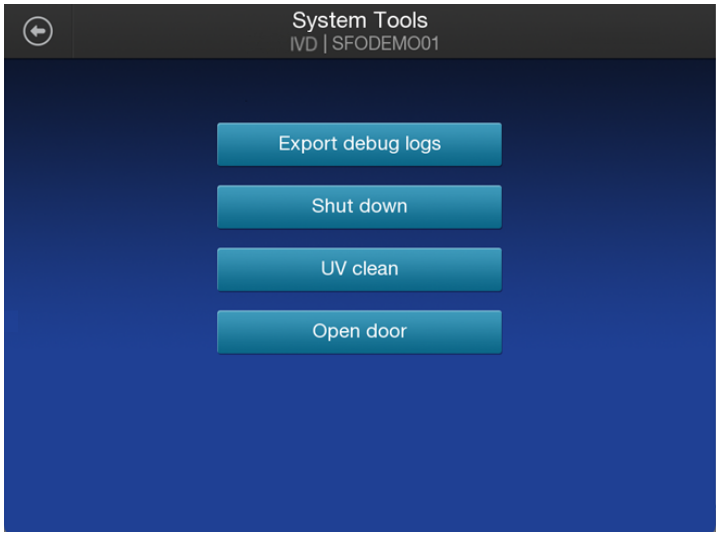
Item	Description	When and how to use
About	Provides instrument details. 	Use to view instrument reference information and performance qualification status.

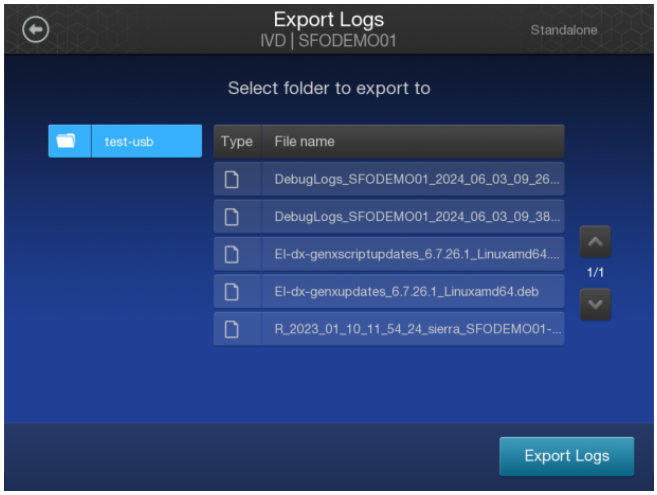
(continued)

Item	Description	When and how to use
Instrument Name		Use to change the instrument name. During installation the **Instrument Name** is set by the Thermo Fisher Scientific service representative. Renaming the instrument invalidaties the performance qualification (PQ). Only change the **Instrument Name** after consulting with your Thermo Fisher Scientific service representative.
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. **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. 	If changing the language. Tap to select a language, then tap **OK**. The instrument will reboot in the selected language.

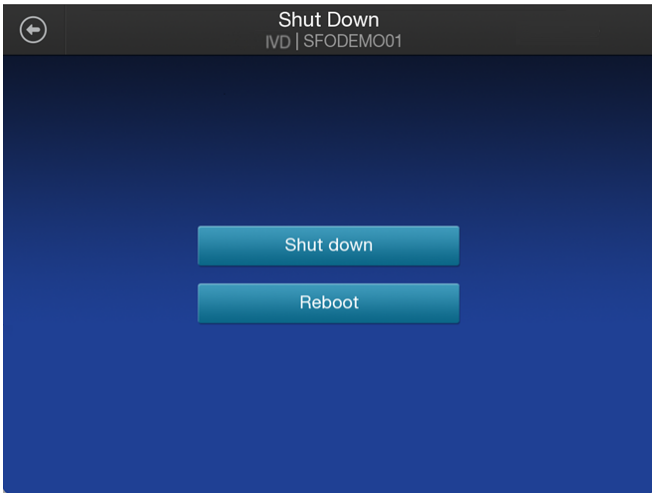
System Tools

Use the **System Tools** menu to upload instrument diagnostics, manage data, and shut down or reboot the instrument.



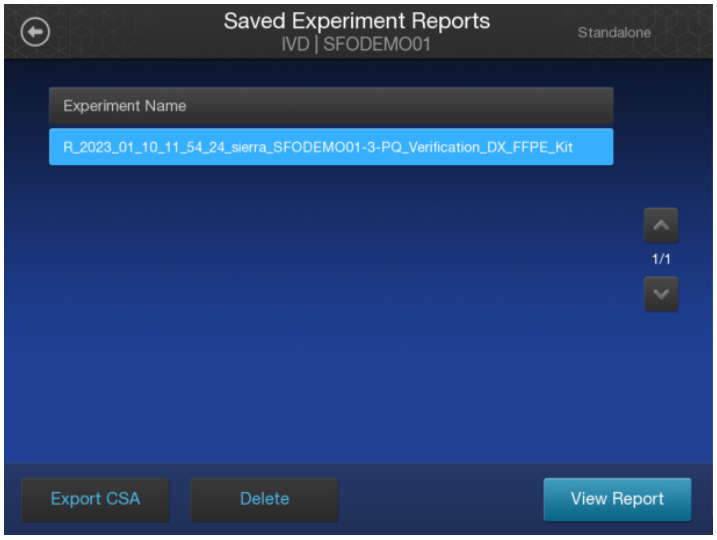
Item	Description	When and how to use
Export debug logs	Provides instrument error configuration information. 	Use for troubleshooting if directed to do so by Technical Support. 1. Tap ⚙ **(Settings) ▸ System Tools ▸ Export debug logs**. 2. In the **Export Logs** screen, tap the folder to export to. 3. From the file list, select a file to export. 4. Insert a USB drive into the USB port on the front of the instrument, then tap **Export Logs**. 5. When prompted, tap **Continue**. 6. When the export is complete, tap **Done**.

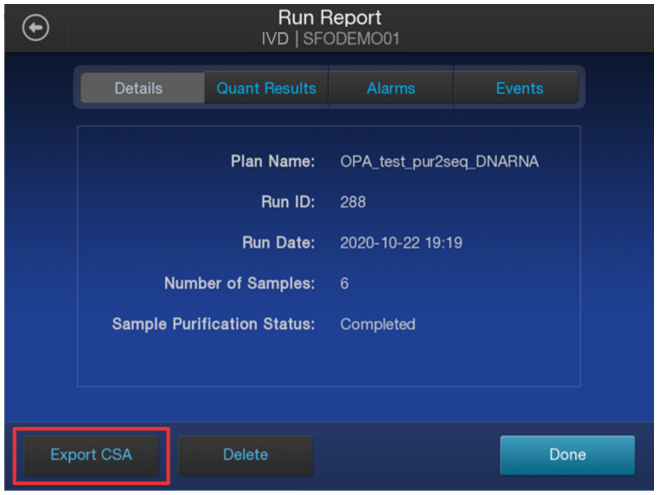
(continued)

Item	Description	When and how to use
Shut down	Provides access to **Shut down** and **Reboot** commands. 	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. 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 35.
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 210.
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).	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.



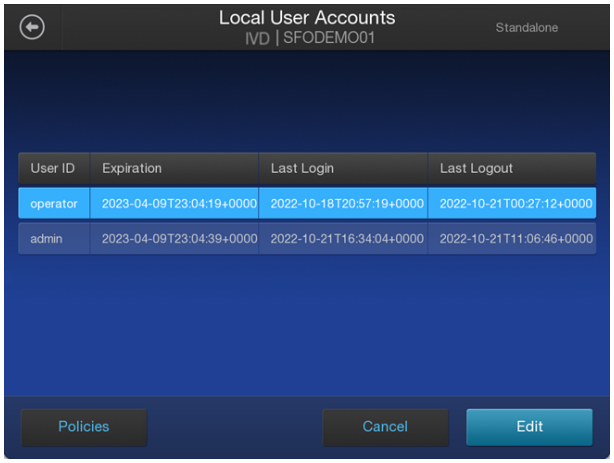
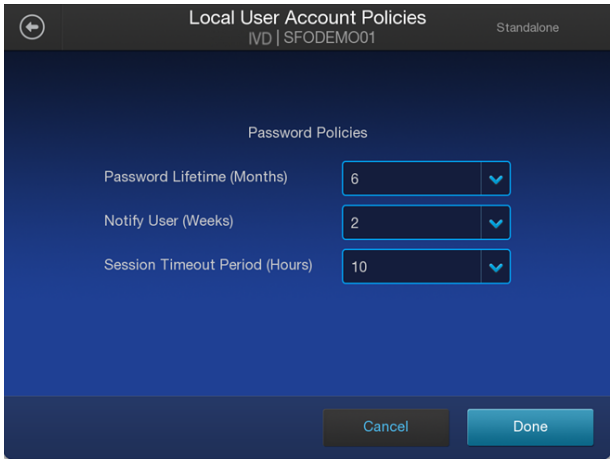
Item	Description	When and how to use
View Report	Purification runs that include sample quantitation produce sample concentration results that can be accessed after the run is complete.	To view reports, see "View and export quantitation results" on page 93.
Export CSA	Provides instrument error configuration information. 	Use for troubleshooting if directed to do so by Technical Support. 1. Tap ⚙ **(Settings)** ▸ **Data management**, then tap in the row of the **Experiment Name** of interest. 2. Tap **Export CSA** 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**. 4. When prompted, tap **Continue**. 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. **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 93.	1. Tap ⚙ **(Settings)** ▸ **Data management**, then tap in the row of the **Experiment Name** of interest. 2. Tap **Delete**. 3. When prompted, tap **Delete** again.

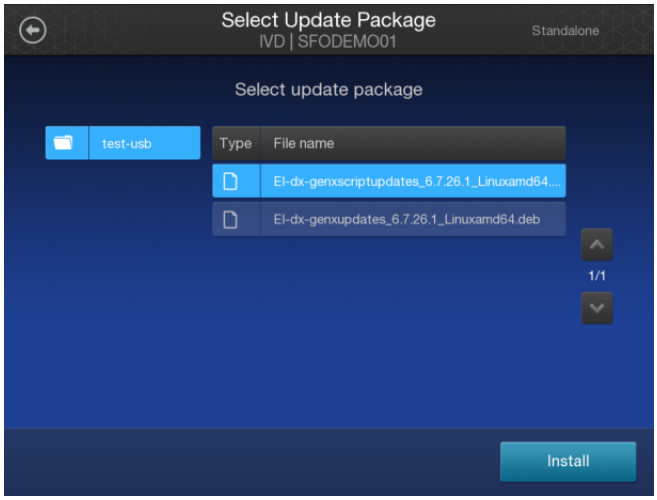
User management

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**. 2. Tap **Edit**, then enter then new password, then tap **Done**. 3. Tap **Done**, then when prompted, tap **Close**.
Policies		1. To update password policies, tap ⚙ **(Settings)** ▸ **User management**, then tap **Policies**. 2. Change the following settings as needed, then tap **Done**. • **Password Lifetime**—Set the number of months until a user password expires. • **Notify User**—Set the number of weeks before a user account password expires that the user receives a notification to reset their password. • **Session Timeout Period**—Set the number of hours before a session times out and a user must login to their user account again

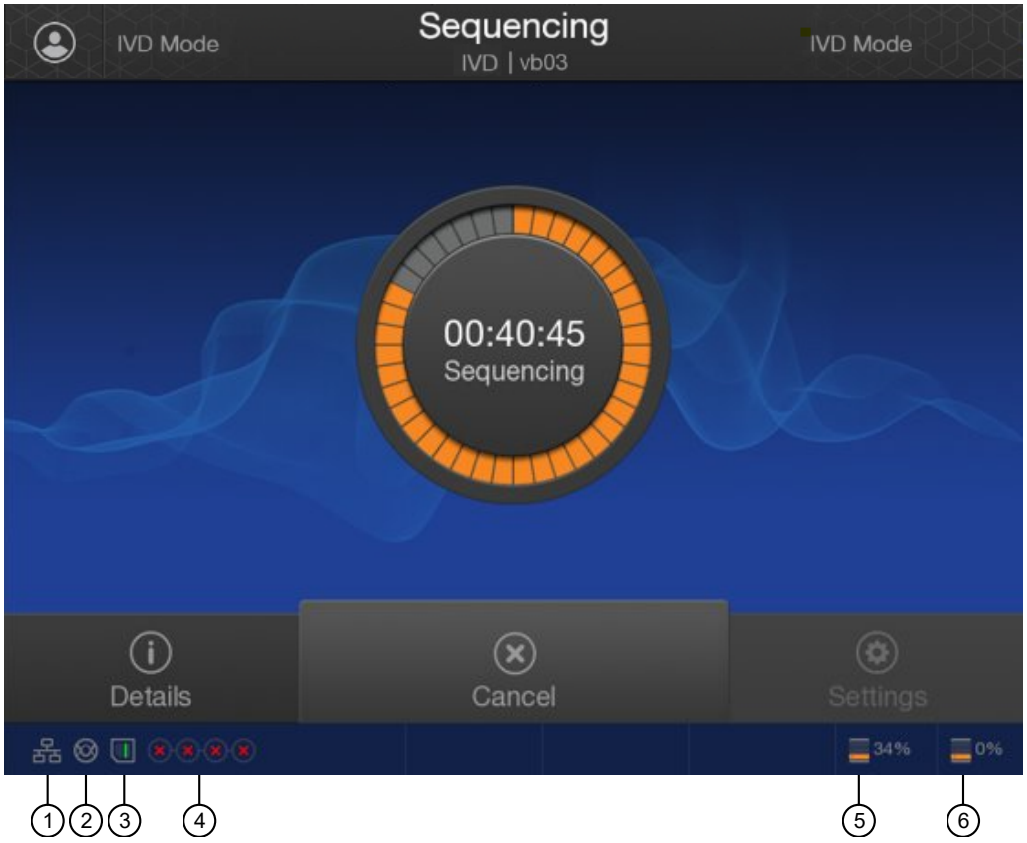
USB updates

Administrators use the **USB update** screen to perform software updates.

Item	Description	When and how to use
USB updates	Used to perform software updates. 	Administrators perform software updates as needed. 1. Insert the USB drive with the software upgrade package into the USB port on the front of the instrument. 2. Tap (Settings) ▸ **USB Updates**. 3. Navigate to the file destination on the USB drive, then tap **Install**. 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

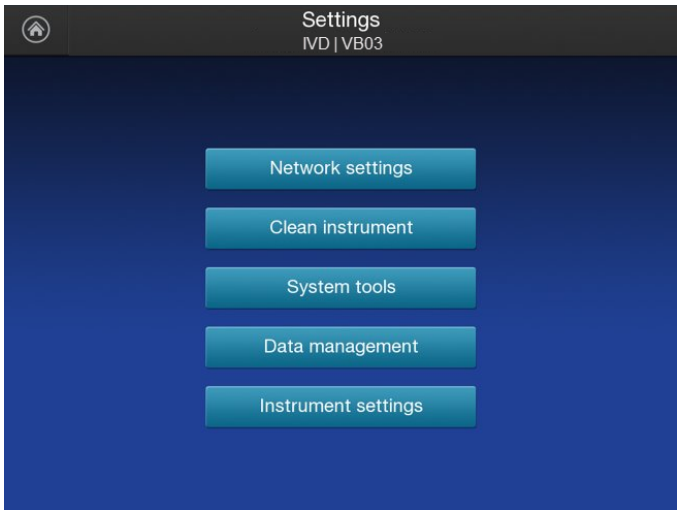
(continued)

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. 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. 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

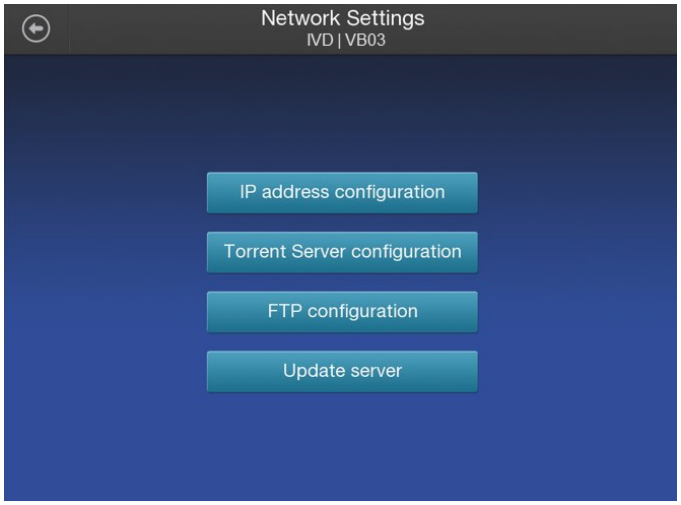
Use the **Settings** menu to view and/or change instrument settings, manage data and network configurations, and clean the instrument.

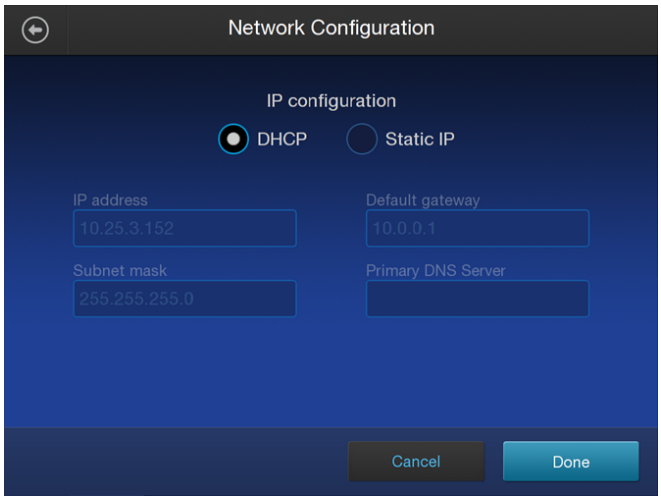


Note: The **System tools** option is for use only by trained service personnel.

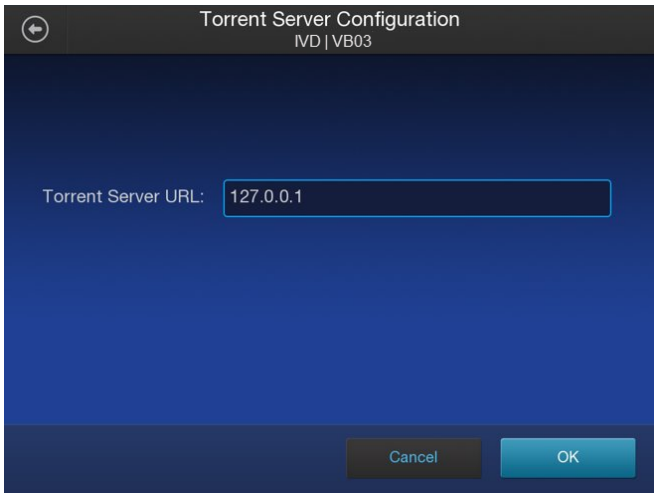
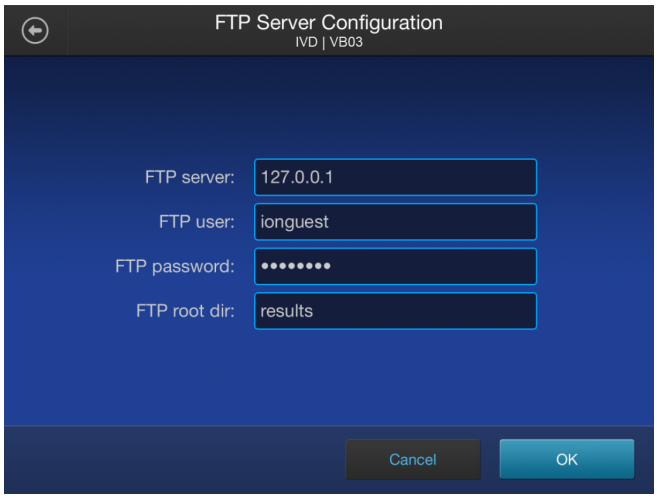
Network Settings

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). 	1. Select either the **DHCP** or the **Static IP** option. 2. Touch the screen in the field you want to edit to activate the field. A virtual keyboard appears. 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.  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. **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.  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. **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 221.

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 115.

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 115.

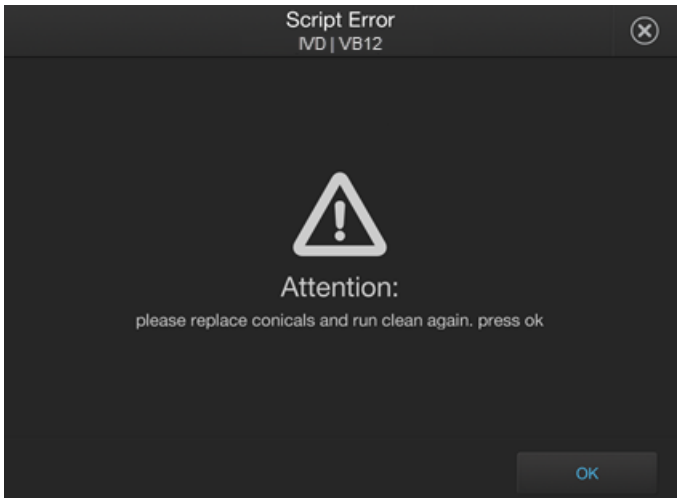
   Note: If a sequencing run completes with 1–3 unused lanes remaining on a chip, and the operator had selected **Do Force Clean** during run setup, the sequencer proceeds directly to the **Clean instrument** procedure after the run. The sequencing reagents bay doors remain closed and no prompts appear 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.

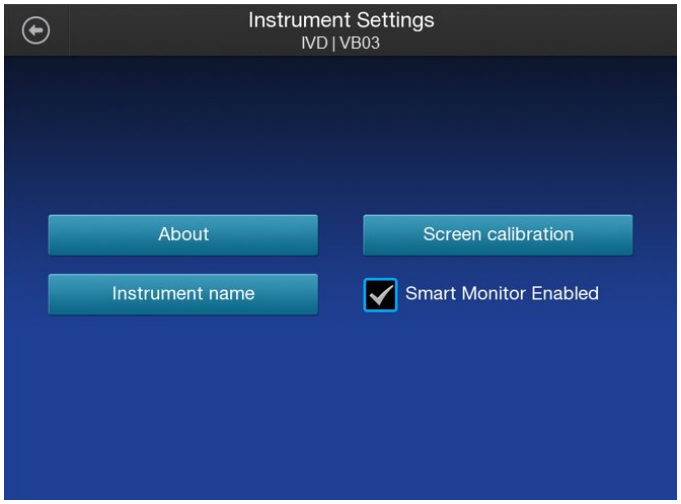


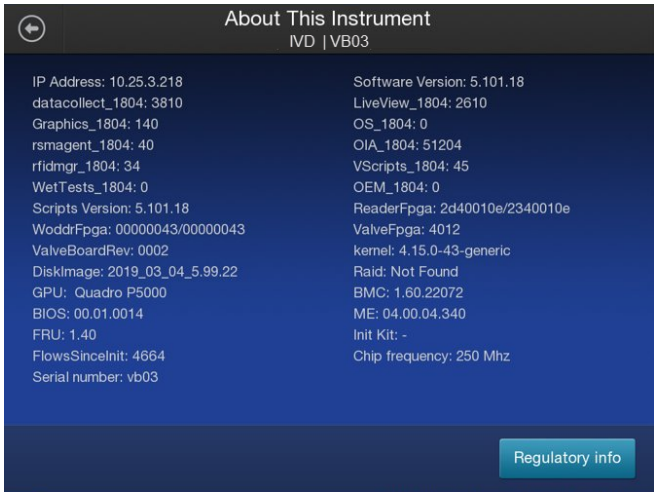
If you see this alert, follow these steps to replace the 5 Genexus™ Dx Conical Bottles.

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 216.

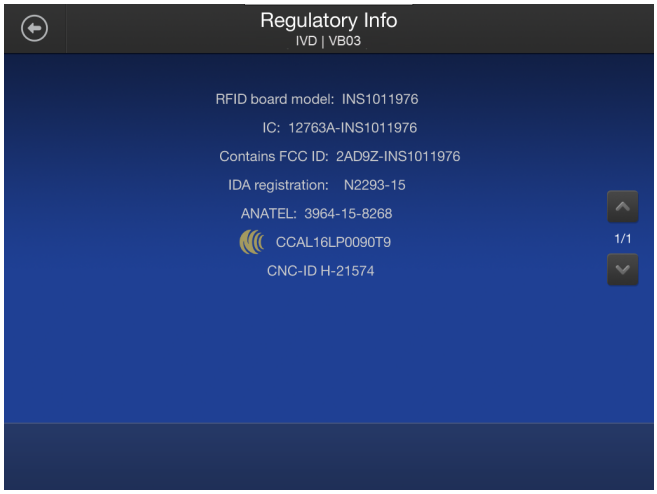
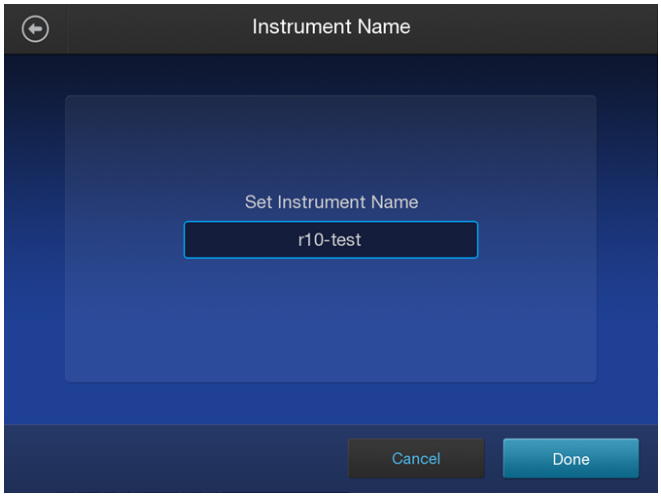
Instrument settings

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. 	Use the screen to view instrument reference information or access regulatory information. Click **Regulatory info** to view the following screen.

(continued)

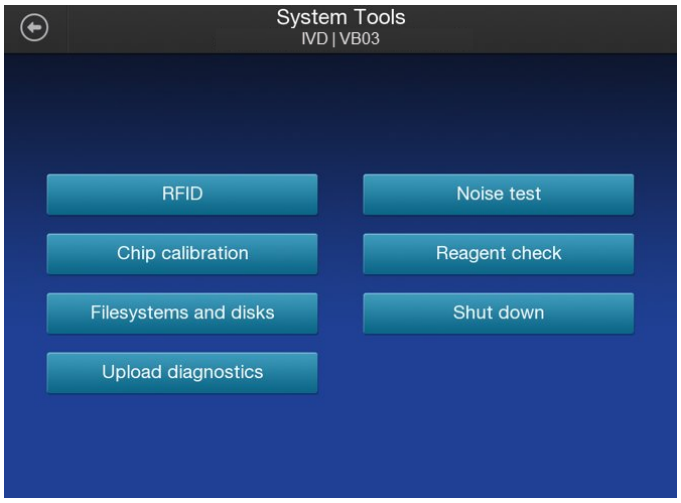
Button	Description	When/How to use
Regulatory info	Lists instrument-specific regulatory information. 	To view instrument RFID regulatory information. See “Radio compliance standards” on page 239 for compliance information.
Instrument name		To change the instrument name.

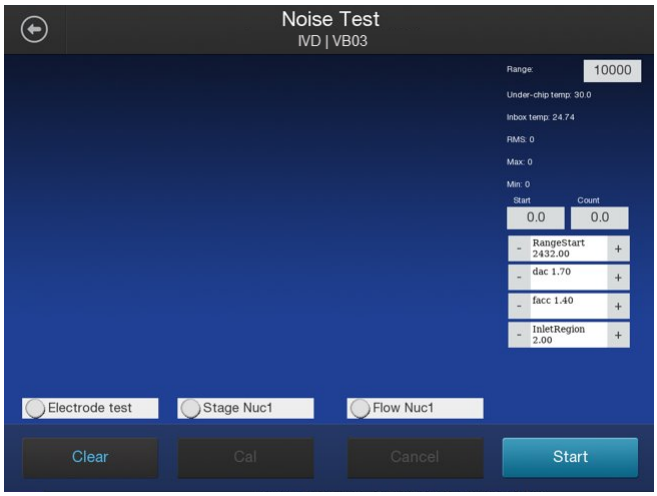
(continued)

Button	Description	When/How to use
Screen calibration		For troubleshooting if directed to do so by Technical Support. 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.

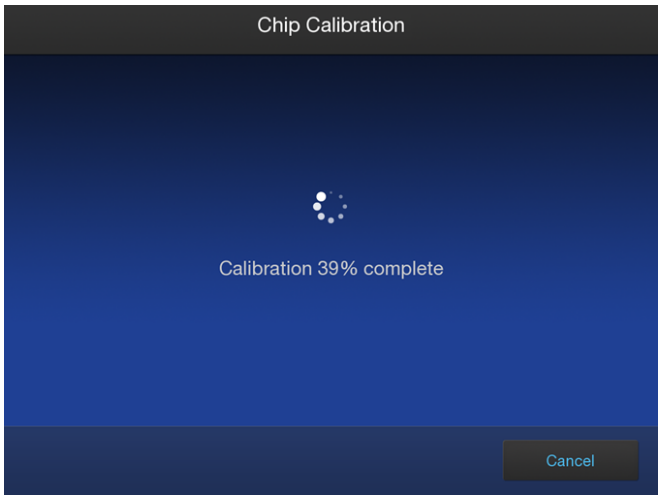
System Tools

The **System Tools** menu enables you to upload instrument diagnostics, manage data, and shut down or reboot the instrument.

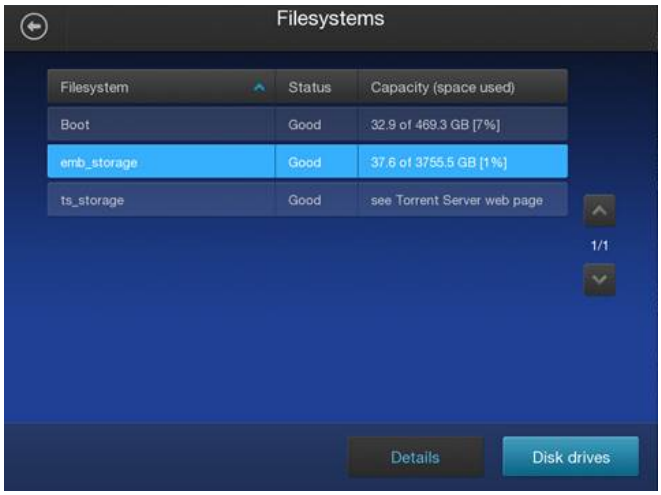
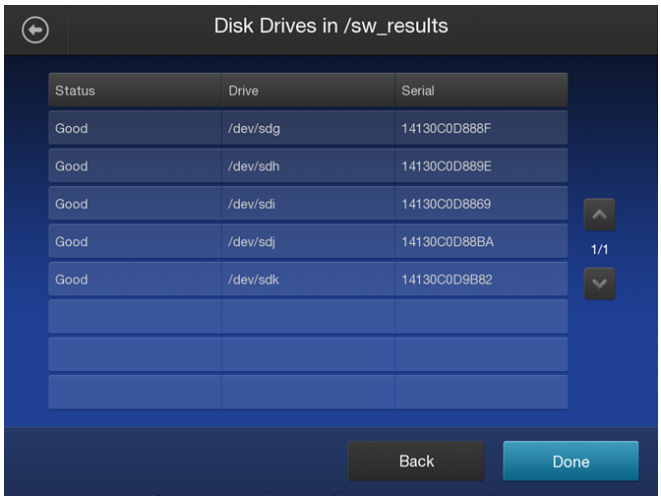
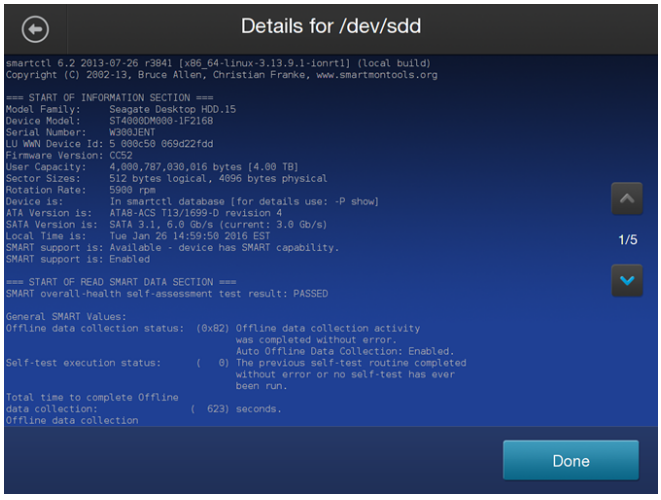


Tool	Description	When/How to use
RFID	Lists the product, product expiration date, and remaining uses of the product. 	To determine if reagents that were left on an instrument during a period of instrument shutdown have expired. 1. Tap the screen within the row of the product to view an **RFID Tag Detail** page. 2. Tap **Done** to return. **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 118.
Noise test	Provides real-time measurement of electrical noise readings on the chip. 	For troubleshooting if directed to do so by Technical Support.

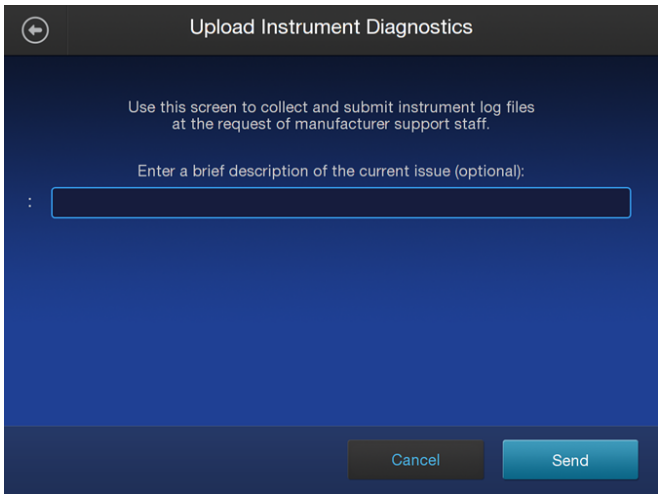
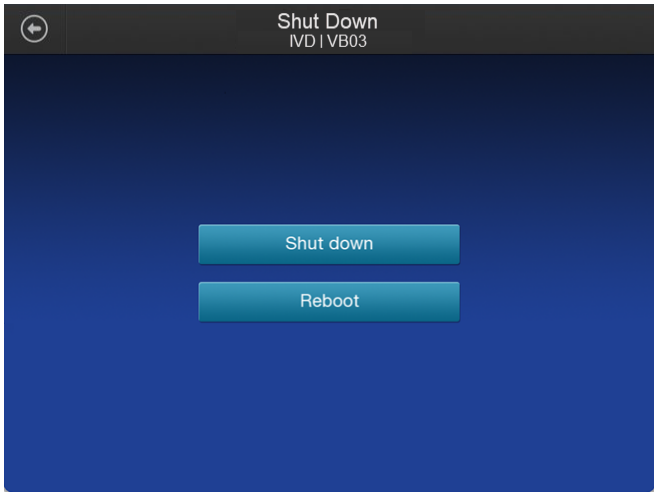
(continued)

Tool	Description	When/How to use
Chip calibration	Checks the status of a chip. **IMPORTANT!** Do not perform a chip calibration unless instructed to do so by a field service engineer. 	1. Insert a new chip into the chip installation slot. 2. In the System Tools menu, tap **Chip Calibration** to start the chip calibration. 3. In the dropdown lists, select the **Image** to display and the type of **Calibration** to perform. 4. After a chip calibration, perform a **Clean instrument** before removing the chip. **Note:** Tap **Cancel** to quit the chip calibration.

(continued)

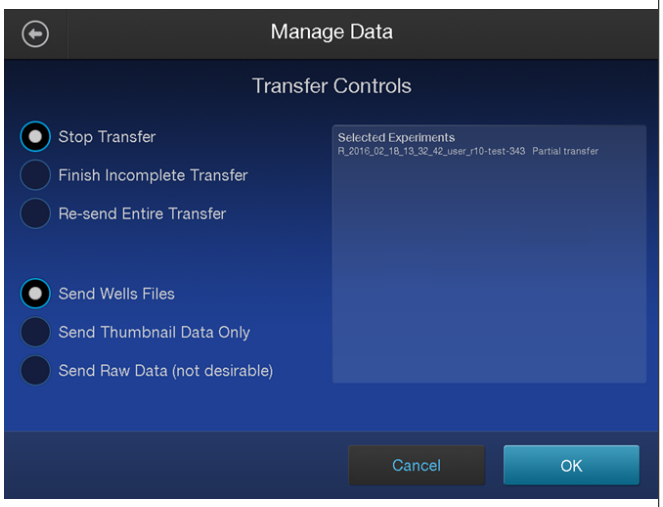
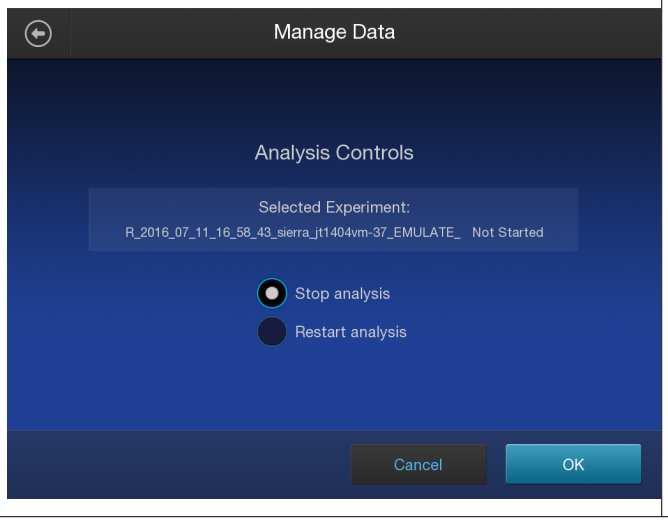
Tool	Description	When/How to use
Filesystems and disks	Provides real-time drive activity status and disk space usage.   	Use if directed to do so as part of a troubleshooting procedure: 1. In the System Tools menu, tap **Filesystems and disks**. The **Filesystems** screen opens. 2. Tap the screen within the row of the **Filesystem** that you want to view, then tap **Details**. The **Disk Drives in <Filesystem>** screen opens. 3. Tap the screen within the row of the **Drive** to view details of the disk drive. The **Details for <Drive>** screen opens. 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. 	Upload an instrument diagnostics file after an aborted run, or if otherwise directed to do so by Technical Support. **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. 	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. **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 35.

Data Management

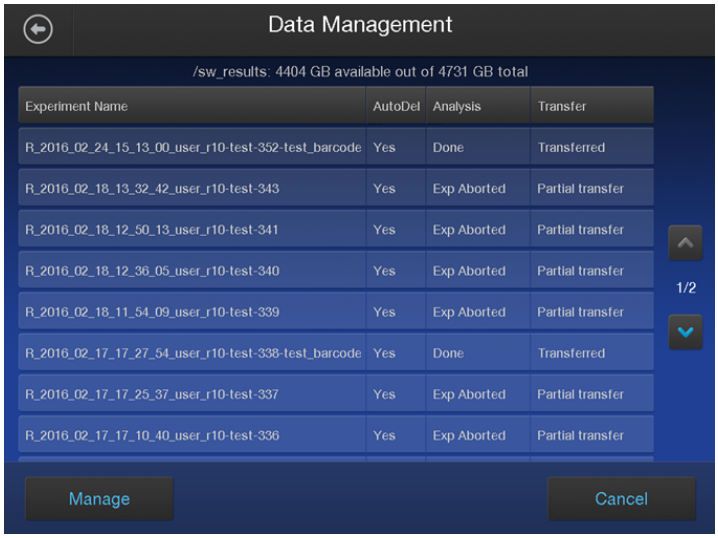
The **Data Management** function allows you to delete run data manually, or transfer data in the event of a failure of automatic transfer. Under normal conditions, run data are automatically transferred to the analysis server, then deleted from the instrument hard drive.

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 209.
Transfer Controls	Transfer run data from the instrument hard drive to the Ion Torrent™ Server. 	1. Tap the screen in the **Experiment Name** row of the individual experiment to be transferred. 2. Tap **Transfer**. 3. Select the transfer action to be performed. 4. Select the file transfer option. 5. Tap **OK**.
Analysis Controls	Stop the analysis of the data of individual runs. 	1. Tap the screen in the **Experiment Name** row of the individual experiment to be analyzed, then tap **Analysis**. 2. Select **Stop Analysis**, then tap **OK**. **Note: Restart analysis** is currently not supported.

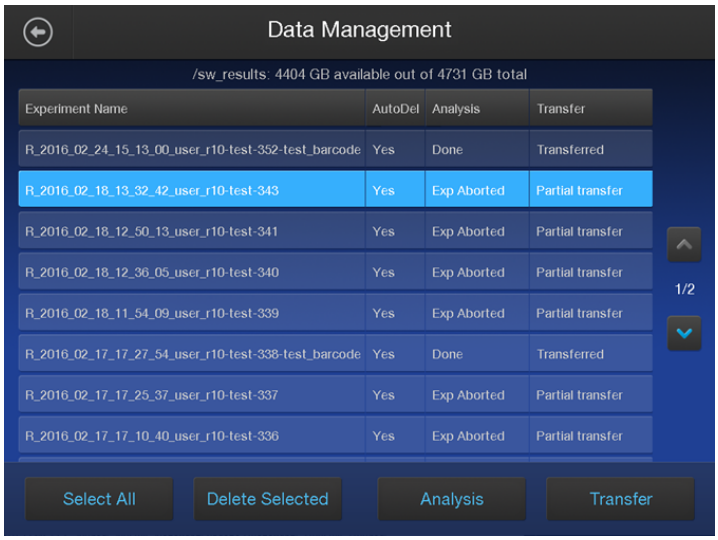
Manually delete run data

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**.

Supplemental information

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- Create a template for importing samples in standalone configuration purification run plans . . . 213
- Delete a run plan in the Genexus™ Dx Purification System touchscreen (standalone configuration) 214
- Maintain the Genexus™ Dx Integrated Sequencer 215
- Instrument planned maintenance 222
- Sequencer decommissioning and data sanitization 222
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- Genexus™ Dx reagent strip and panel tube fill volume guide 227

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 enable 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.

```
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
```

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.

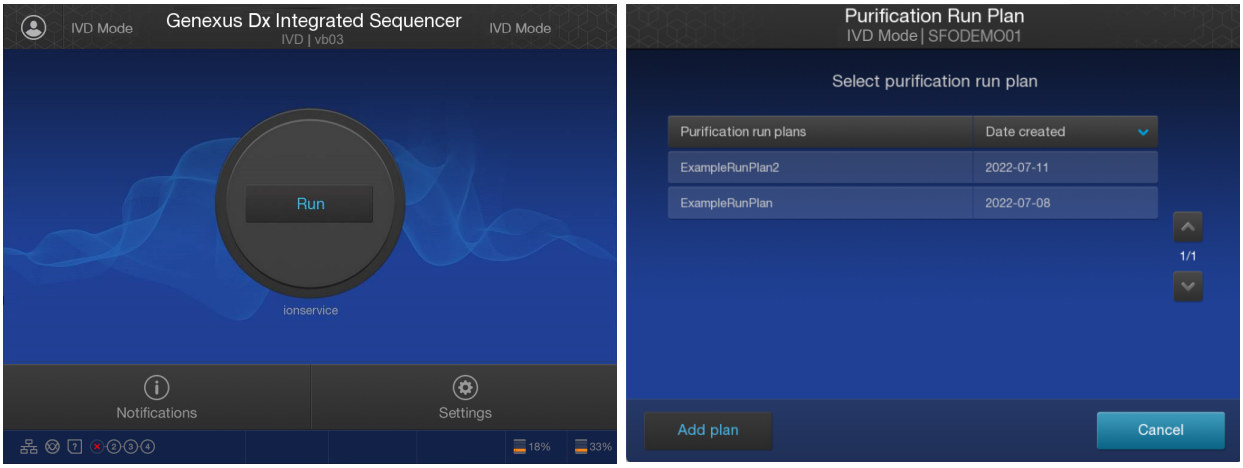
2. Save the file using a CSV file format to a USB drive.

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.

Clean the Genexus™ Primer Pool Tube Holder

Use this procedure to clean the Genexus™ Primer Pool Tube Holder.

Note: Dispose of all waste in appropriate liquid or solid waste containers.

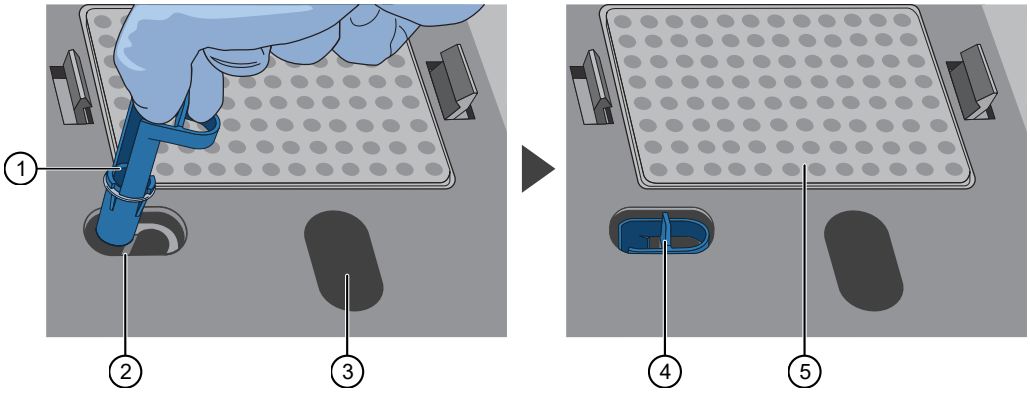
1. Inspect the Genexus™ Primer Pool Tube Holder for signs of reagent accumulation.
2. Using lint-free wipes, wipe the Genexus™ Primer Pool Tube Holder with 70% isopropanol.
3. Place the Genexus™ Primer Pool Tube Holder back on top of the Primer Pool Tube Station on the deck and allow to air-dry.
4. When the Genexus™ Primer Pool Tube Holder is dry, proceed with the **UV Clean**.

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 115.

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

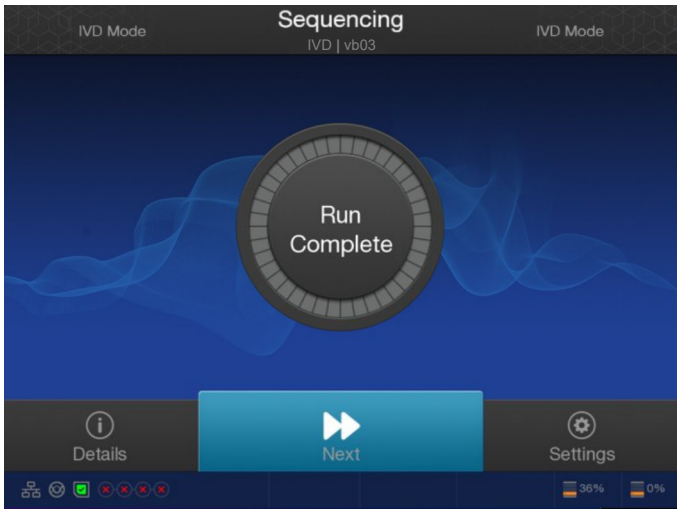
Replace the Genexus™ Dx Conical Bottles

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.

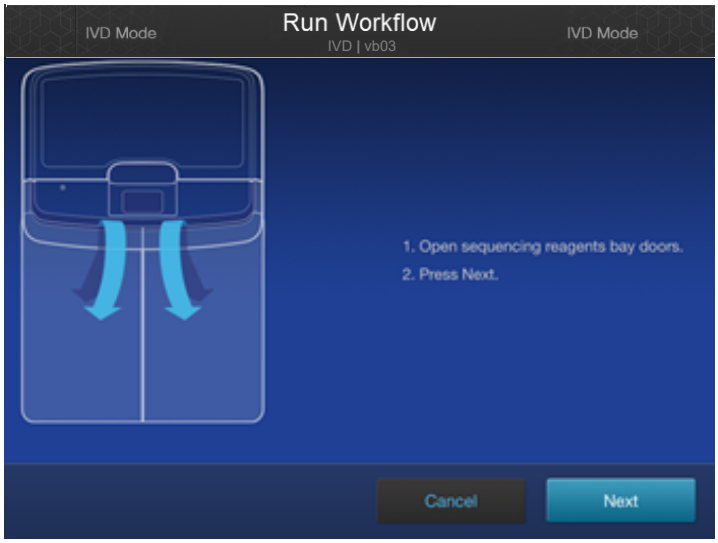
Follow these steps to replace the Genexus™ Dx Conical Bottles.

1. After a run, in the **Run Complete** screen, tap **Next**.



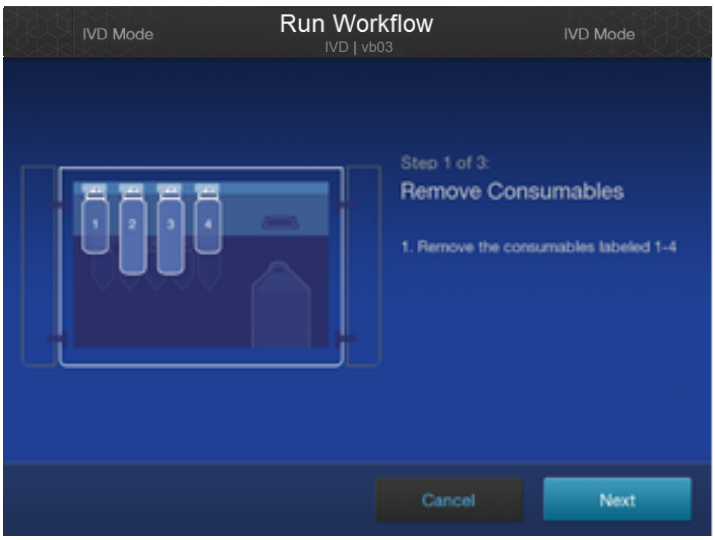
If the Conical Flow Rate test failed, a screen appears directing you to open the sequencing reagents bay doors.

2. Open the sequencing reagents bay doors, then tap **Next**.

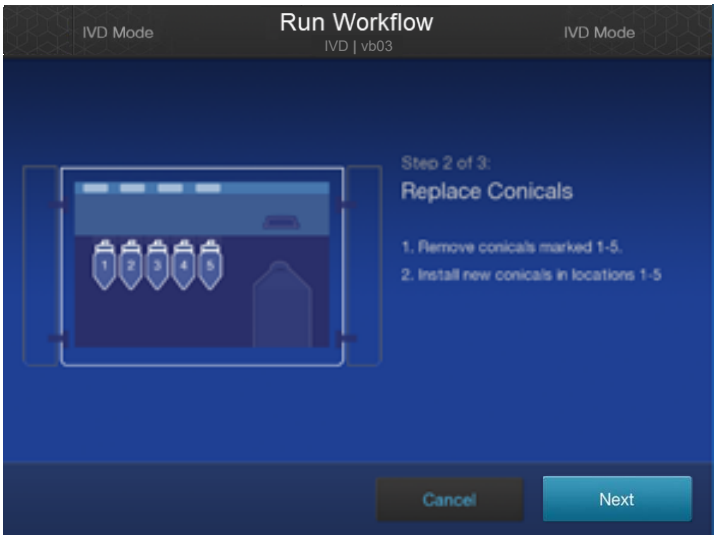


The **Remove Consumables** screen appears.

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 35.
  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 199, 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

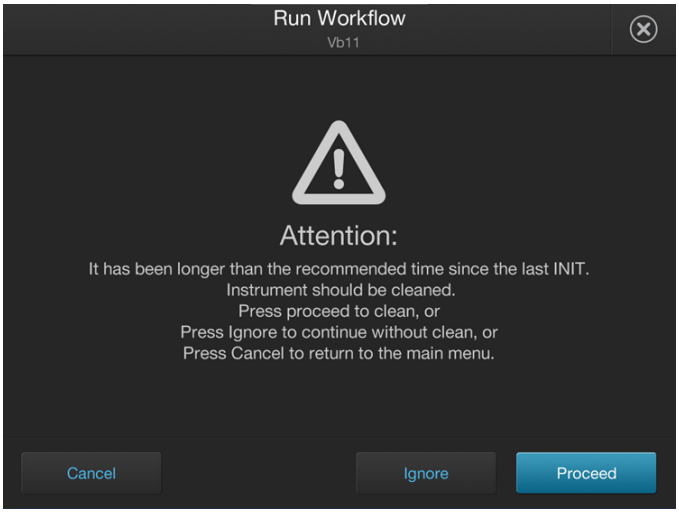
After a long-term shutdown (>28 days), follow these steps to power on the sequencer, as described in "Power on" on page 35.

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 199. 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 118.



   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.

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.

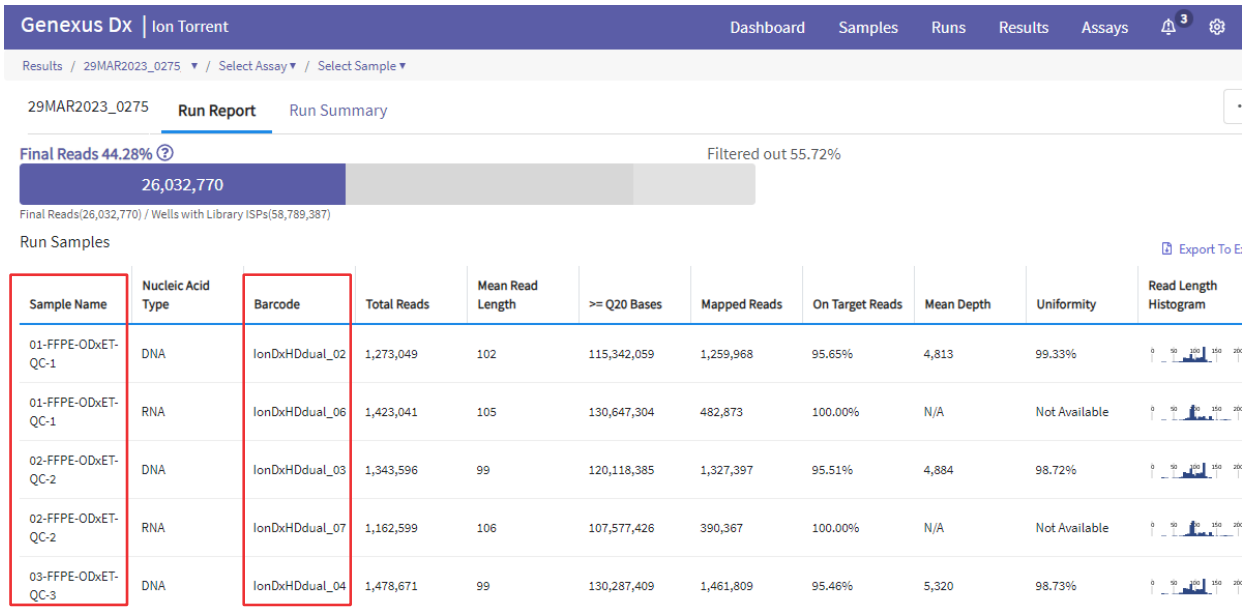
Recover pooled sample libraries and plan a Library to Result run

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 43.

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 64.

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**.

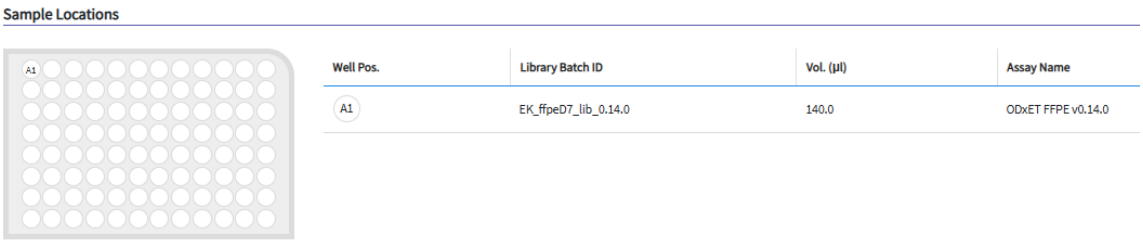
2. In the **Setup** step, enter a name for the run, configure the reporting options, then click **Next**.

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

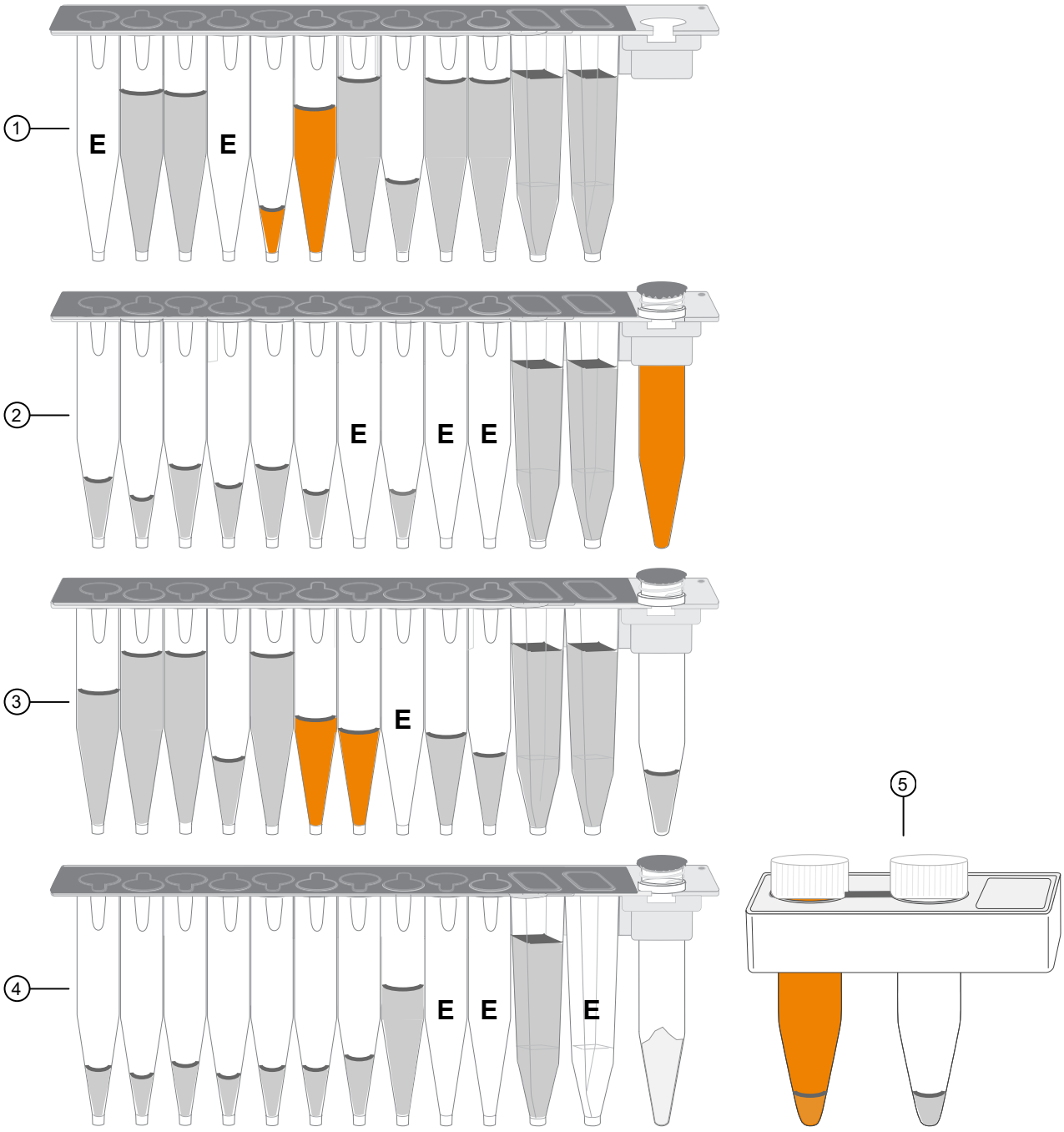
The Ion Torrent™ Genexus™ Dx Install Kit (Cat. No. A52172) is provided to first-time owners of a Genexus™ Dx Purification System and Genexus™ Dx Integrated Sequencer. The kit contains the following reagents, supplies, and controls that are used during the installation, training, and operation of the 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 • Genexus™ Dx PQ Panel-HD • Genexus™ Dx DNA PQ Control • 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	4 sets of 5 bottles	
Adhesive PCR Plate Foils	AB0626	1 box (100 foils)	

Genexus™ Dx reagent strip and panel tube fill volume guide

Use the figure below to check for correct 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 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. MAN1001774).

E

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.

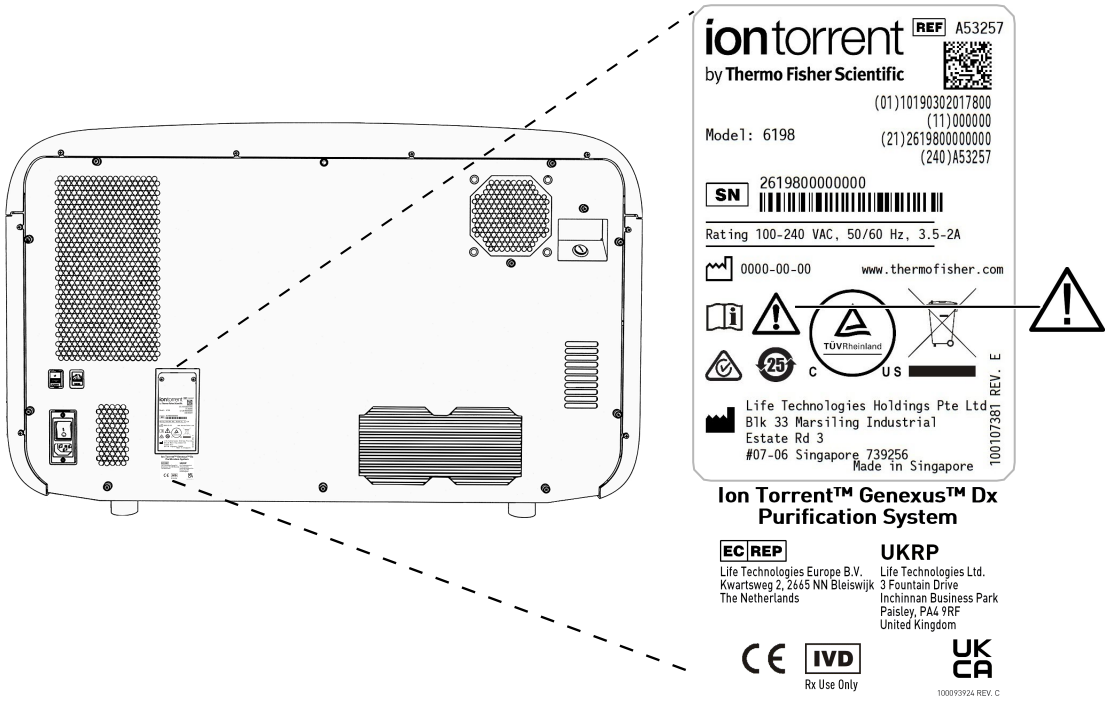
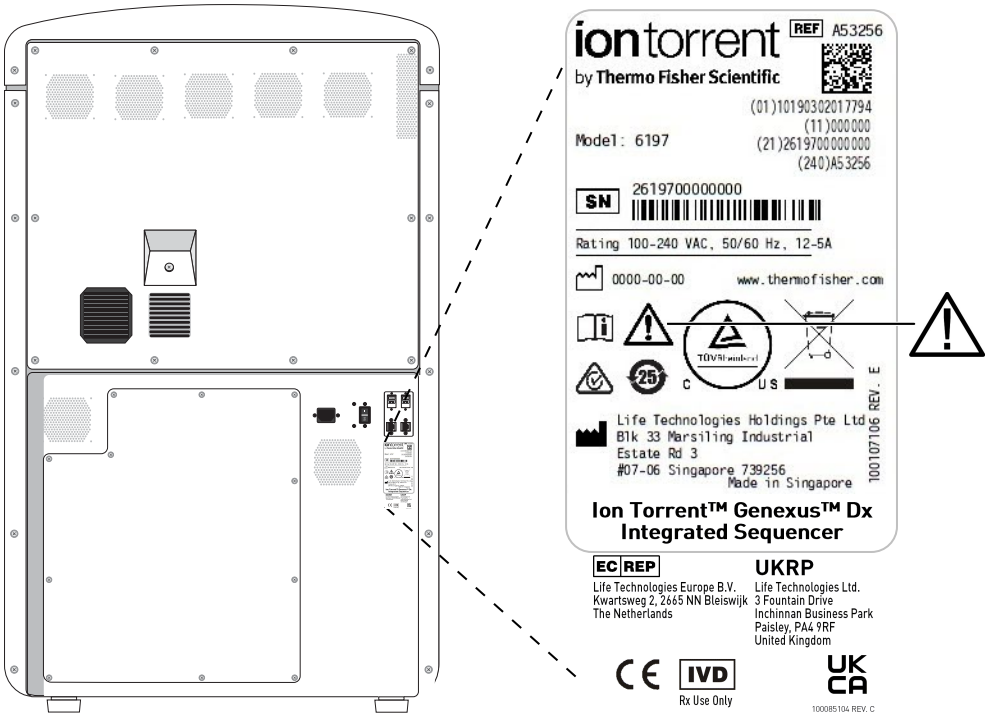
(continued)

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
 Purification instrument rear panel
 Sequencer rear panel

Control and connection symbols

Symbol	Description
	On (Power)
	Off (Power)
	Protective conductor terminal (main ground)
	Alternating current

Conformity symbols

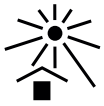
Conformity mark	Description
	Indicates conformity with safety requirements for Canada and U.S.A.
	Indicates conformity with China RoHS requirements.
	Indicates conformity with European Union requirements.
	Indicates conformity with Australian standards for electromagnetic compatibility.
	INDICATES CONFORMITY WITH THE WEEE DIRECTIVE 2012/19/EU. **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 EN 61010-1 UL 61010-1 CAN/CSA C22.2 No. 61010-1 GB 4793.1 (China)	*Safety requirements for electrical equipment for measurement, control, and laboratory use—Part 1: General requirements*
IEC 61010-2-010 EN 61010-2-010 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 EN 61010-2-020 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 EN 61010-2-081 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 EN 61010-2-101 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 IEC 61326-1 GB/T 18268.1 (China)	*Electrical Equipment for Measurement, Control and Laboratory Use. EMC Requirements—Part 1: General Requirements*
EN 61326-2-6 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* 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. 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 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): This device complies with Part 15 of the FCC Rules. Operation is subject to the following conditions: 1. This device may not cause harmful interference, and 2. This device must accept any interference received, Including interference that may cause undesired operation. Changes and modifications not expressly approved byThermo Fisher Scientific can void your authority to operate this equipment under Federal Communications Commissions rules.
RFID	Canada: This device complies with Industry Canada license-exempt RSS standard(s). Operation is subject to the following two conditions: (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): 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 : (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] Industrial and Information Department Radio, BRR[1]	China: Notice on issuing the technical requirements for micropower (short range) radio equipment.
GB/T 12572-2008	China: *Universal requirements and measurement methods of parameters for radio transmitting equipment*
ETSI EN 300 330-1 (2015-03)	China: *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: *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 MAN1001776

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: MAN1001776 A (English)

Revision	Date	Description
A	3 August 2026	New IVD user guide for the Oncomine™ Dx Express Test 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 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.

Prerequisites

Category	Prerequisites
Instrument	• Genexus™ Dx Integrated Sequencer. • Reagents and supplies for operating the Genexus™ Dx Integrated Sequencer.
Software	• Genexus™ Dx Software 6.8.2.1 or later.
Functional knowledge and understanding - all users	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to administrator credentials, SSH private keys and the associated SSH key passwords. • Main functions of the Genexus™ Dx Integrated Sequencer. *(Optional)* Main functions the Genexus™ Purification System. • Main features of the Genexus™ Dx Software.

(continued)

Category	Prerequisites
Functional knowledge and understanding - system administrators	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to administrator credentials, SSH private keys and the associated SSH key passwords. • Use of security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

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. MAN1001774).

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. MAN1001774).

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 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.
- 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 6.8.2.1 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. The operating system has an Ubuntu™ Pro license that includes expanded security maintenance coverage.

It is recommended that Genexus™ Dx Software be used only on computers that are maintained by facility system administrators and IT and security personnel, and include security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

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 facility system administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions. The Genexus™ Dx Integrated Sequencer has a firewall enabled by default. Only ports required for the operation of the instrument and software are open.

CAUTION! Data can be lost or corrupted if you operate a sequencer that does not have sufficient security controls in place.

If external or network drives are connected to Genexus™ Dx Integrated Sequencer for archiving or backups, it is the sole responsibility of 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 194.

Data backup and data storage

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.

Note: A paid data migration service is available if needed. Contact your Thermo Fisher Scientific representative for more information.

For more information about data storage compatibility, contact your local support team.

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.

Antivirus software

For recommendations for antivirus software that is compatible with Genexus™ Dx Software 6.8.2.1, see the current version of the Genexus™ Dx Software release notes.

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.

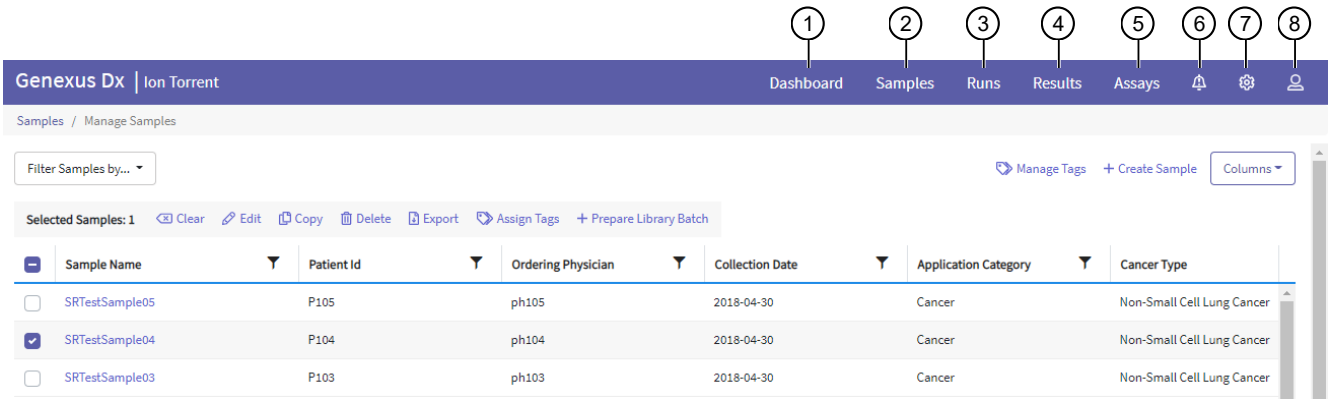
Sequencer decommissioning and data sanitization

Contact a Thermo Fisher Scientific Field Service Engineer for any request to decommission a Genexus™ Dx Integrated Sequencer. Decommissioning and data sanitization must be carried out by a Field Service Engineer.

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 • Generate, view, and sign variant reports • Send notifications • Download results files • View and edit notes for the sample result • View the audit trail for sample results • View notifications
Operator	Report functions, plus: • Add, import, and export sample files • Edit sample files • Prepare library batches • Plan, save, audit, and delete runs • Monitor runs • View results and reports • View the audit trail for samples, library batches, runs, and assays • Reanalyze runs

(continued)

Users at this level	Can perform these actions
Manager	Operator functions, plus: • Create, edit, and obsolete sample attributes • Import assays • Create report templates • Edit and delete sample files • Add or edit Thermo Fisher accounts • Restart a stalled or failed run • Access services information
Administrator	Operator and manager functions, plus: • View, export, and print audit records • Configure network settings • View and manage software updates • Configure backup settings and restore runs • Manage sequencer and software log files • 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 18.

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.

Note: A mode switch cannot proceed if a run is loaded on the instrument, even if the run is complete.

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**. 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**. 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**. b. In the **Edit Security questions and answers** dialog box, select a security question from the dropdown list, then enter an answer. c. Enter the password for the account that is associated with the profile. 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 233.

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. 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 22
Nucleic Acid to Result Library to Result	“Nucleic Acid to Result or Library to Result workflow” on page 23

Sample to Result workflow

Workflow for a Sample to Result purification and sequencing run

View and manage assays (page 144)

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 24)

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 54)

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 (see the *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports 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 *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports 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 68)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.

Review results in Genexus™ Dx Software (page 72)

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 144)

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 24)

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 57) **or Plan a Library to Result run** (page 61)

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 *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports 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 *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports 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 68)

Monitor the run in the Genexus™ Dx Software Dashboard in real time.



Review results in Genexus™ Dx Software (page 72)

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 29 and “System-installed sample attributes” on page 223.

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 33 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.

Show or hide columns in the samples table

The samples table is organized into columns of sample attributes. You can determine which columns are shown or hidden in the samples table with column preferences. The changes you make to the columns 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 show.
   - Deselect the checkbox next to each attribute you would like to hide in 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 25.

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 27.

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.

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. • In the search field, enter 3 or more characters for the sample attribute. • Select a checkbox next to one or more sample attributes. • Click **Select All** to select all sample attributes. • Click **Clear** to close the filter dialog box.
Limit the list of samples based on a creation date, or a range of dates.	Click **(Filter)** in the **Created On** column heading, and select **Is equal to**, **Is before or equal to**, or **is after or equal to** to set the criteria for the date or date range. Select a date in the calendar. *(Optional)* Select **And**, or **Or** to include a second set of criteria and further narrow the search. For more details, see "Filter the list of samples by the date of sample creation" on page 27.

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 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 28 and "Search for samples" on page 25.

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)**. b. In the first dropdown list, select **Is equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**.
List samples created within a date range	a. In the **Created On** column header, click **(Filter)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select **And**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date that the samples of interest were created. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select **Or**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date that the samples of interest were created. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is before or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**.
List only samples that were created after a date that you indicate	a. In the **Created On** column header, click **(Filter)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the date of interest. 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 29 and “Import samples” on page 30.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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 user-defined sample attribute (manager/administrator)” on page 34.
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 30.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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 223. 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 33. Upload the edited file using the **Browse** button. **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**. b. Navigate to the file, then click **Open**. 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 76.

For more information about sample attributes, see “System-installed sample attributes” on page 223.

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 39.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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**.

(continued)

Sample attribute	Description
%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. **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 30.

Add a user-defined 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 user-defined attributes and add or change values of those attributes.

1. To create an 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.

User-defined 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 29, “Create multiple samples” on page 29, “Import samples” on page 30, “Edit a sample” on page 39, and “System-installed sample attributes” on page 223.

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 39 and “Review sample edit history” on page 41.

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 102 and “Assign sample tags to multiple sample results” on page 103.

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 29 and "Import samples" on page 30.

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 37.

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**. b. Enter the note, then click **Save**. 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 120.

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 223 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 120.

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 29.
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 41.

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 24.

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 57.

Note:

- Each library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

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 57.

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.

   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 “Find samples” on page 25.

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**.

   Note: If the cancer type of a sample in the library batch, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type. You can edit the cancer type of a sample, if needed. For more information, see “Edit a sample” on page 39.

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)

(continued)

Template item	Description
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 45.

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 47.

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 45.

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 48.

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 45.

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. 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**. 2. Modify the information in the **Edit Library batch** screen, then click **Submit**. Alternatively, in the library batch details pane, click the library batch ID to perform this action. 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**. The **Audit Records** screen opens with time-stamped audit records that include details and changes for each action performed on the library batch. 2. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions. 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**. The **Add Notes** dialog box opens and notes that are associated with the library batch are listed by user, date, and time. 2. In the **Add Notes** dialog box, click + **Add Note**. 3. Enter the note, then click **Save**. 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 53.

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 24.
- 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 42.

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 51.

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 53.

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.

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 51.
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. • Select **Planned** to list all **Nucleic Acid to Result**, **Library to Result**, and **BAM to Result** planned runs. • 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 *Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide* (Pub. No. MAN1001775).

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 29, or “Import samples” on page 30.
- 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 positive 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. Red denotes a chip lane in the run containing assigned samples that exceed lane capacity.

      Error messages can appear that may require you to change the samples you include in the run.

      - 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 will 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.

      - If the cancer type of a sample, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type.

   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 53.

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 64.

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 53.

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 105.

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 29, or “Import samples” on page 30.
- 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 144.
- 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. MAN1001775).

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 positive 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. Red denotes a chip lane in the run containing assigned samples that exceed lane capacity.

      Error messages can appear that may require you to change the samples you include in the run.

      - 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 will 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.

      - If the cancer type of a sample, selected when the sample was created in the software, is not included in the assay definition file of the selected assay, an error message appears. Ensure that you select a cancer type at sample creation that is supported by the assay that you intend to use with the sample. If multiple assays are installed in the software, the cancer types and subtypes to select from at sample creation can exceed the number of cancer types supported by an assay. It is possible to select an unsupported cancer type.

   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.

   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. MAN1001775).

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 42.

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, 8 samples were included in a library batch instead of 7. The run is allowed if the lane capacity is exceeded, but you will 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 63.
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 66, "Add a note to a run" on page 66, "Review and download a run history" on page 65.

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.
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 106.

(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 106.

[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 53.

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 63.

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: • **Create** • **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 54, “Plan a Nucleic Acid to Result run” on page 57 and “Plan a Library to Result run” on page 61.

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.

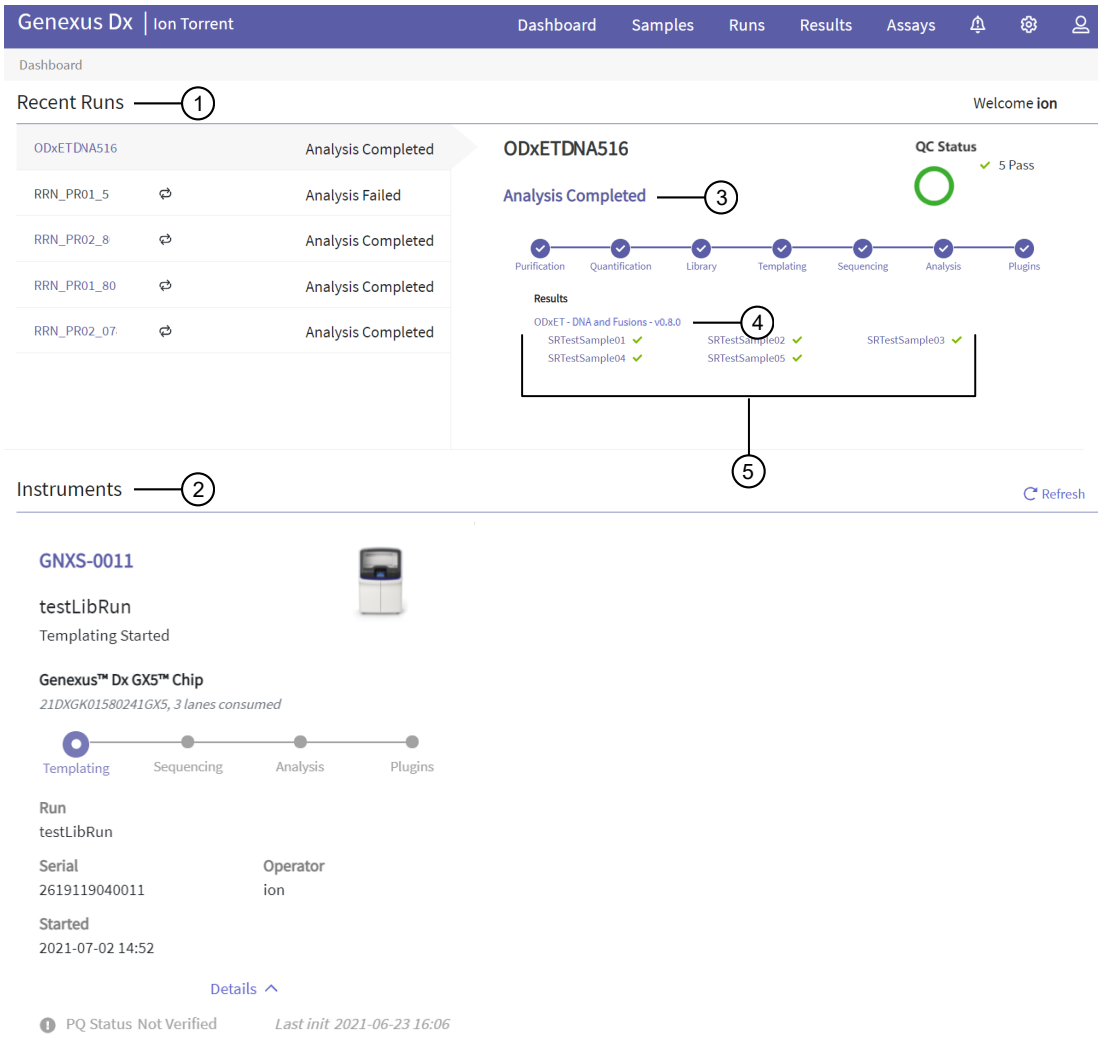
1. In the menu bar, click **Runs** ▸ **Manage Runs**.
2. In the **Manage Runs** screen, place the pointer over the row of the run plan of interest, then click **Notes**.
3. In the **Add Notes** pane, in the row of the note you want to delete, click **(Delete)**.

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 120.

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. The **Run Summary** tab opens. For more information, see “The run summary” on page 106. 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. 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 95.
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. b. In the run details pane in the right side of the screen, click the assay name of interest. The **Assay Metrics** tab opens. For more information, see “Assay metrics and the run report” on page 110. 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. b. In the run details pane in the right side of the screen, click the sample name of interest. The **Key Findings** tab opens. For more information, see “Key Findings” on page 76. 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. b. In the run details pane in the right side of the screen, place the pointer over the status for the run. 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 70.

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**. **(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 and any Genexus™ Dx Purification System that is integrated 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.

   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.

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. 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.

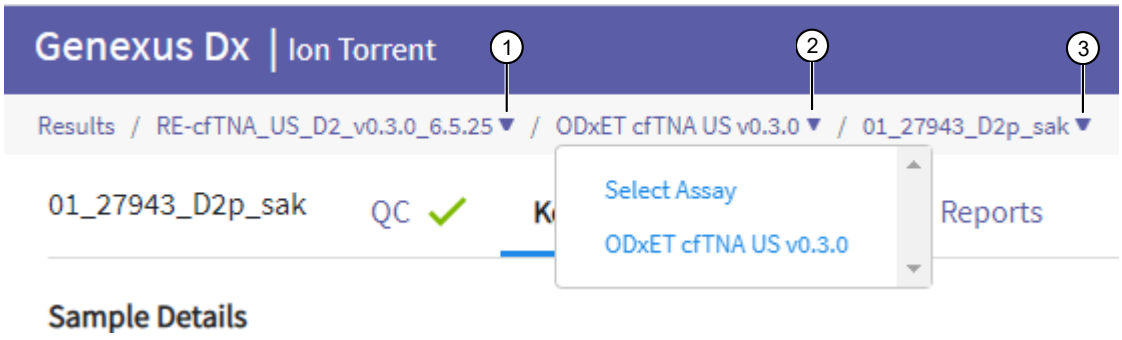
Selection	Description
Click **Results** ▸ **Sample Results**	Select this option to review completed sample results and reports.
Click **Results** ▸ **Run Results**	Select this option to review completed run results and reports by assay.
Click **Results** ▸ **Verification Results**	Select this option 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 120.
2	**Select Assay**	You can click the assay name of interest to view assay metrics for the run. 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. 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 110.
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. 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 110.

Review sample results

You can review completed sample results and reports in Genexus™ Dx Software.

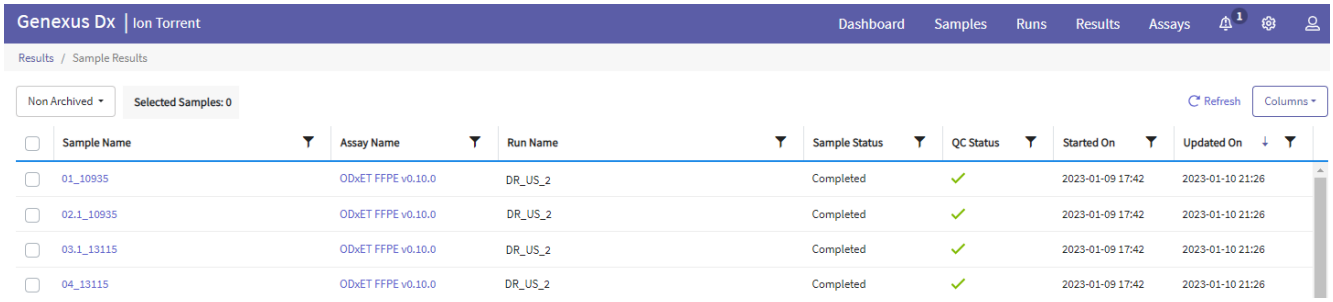
See “Find sample results” on page 98 for information about how to find sample results of interest.

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 72.

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 110.

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 "Show or hide columns in the samples table" on page 25.

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. • ✔ (Passed) indicates that the sample passed all QC metrics. • ✘ (Failed) indicates that the sample failed a QC metric. • — (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 75, "View the QC results" on page 94, "View variant results" on page 77, 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. • **Report**—Download the report (available only for samples with a sample status of completed). For more information, see "Clinical Test Report and Clinical Laboratory Report" on page 127. • **Assign Tags**—Add or remove the tags assigned to a sample result. For more information, see "Sample tags" on page 34. • **Audit**—View the audit trail for the run. For more information, see "Review and download a run history" on page 65. • **Notes**—View, add, or delete notes for sample result. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see "Download a Customer Support Archive" on page 227. • **Reanalyze**—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. For more information, see "Reanalyze a sample" on page 121.
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 76.

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.
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

Biomarker level[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 132.

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 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

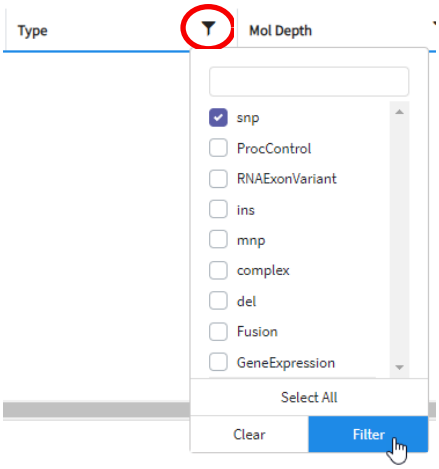
Variant type	Description
SNVs/Indels	• **SNVs**: missense and nonsense single-nucleotide variants. Multi-nucleotide variants are also included. • **Indels**: insertion and deletion variants. For more information, see “View SNV/Indel results” on page 79.
Fusions	Translocations of genetic material and splicing variants. For more information, see “View fusion results” on page 81.
CNVs	Copy number variations (CNVs) are variations of the number of copies of a given gene. For more information, see “View CNV results” on page 86.

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 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 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 78.

Column	Description
Variant ID	The name of the hotspot as defined in the Browser Extensible Data (BED) file. Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 88.
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. • **snp** (single nucleotide polymorphism) • **mnp** (multi-nucleotide polymorphism) • **ins** (insertion) • **del** (deletion) • **complex**
Test Result	Indicates the presence or absence of an SNV/Indel variant. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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). 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 82.
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 78.

Column	Description
Variant ID	The name of the fusion target as defined in the BED file. Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 88.
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. • **POSITIVE**—indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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. **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. Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline) where Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples) • 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample. • 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample. Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline) • 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene. • 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

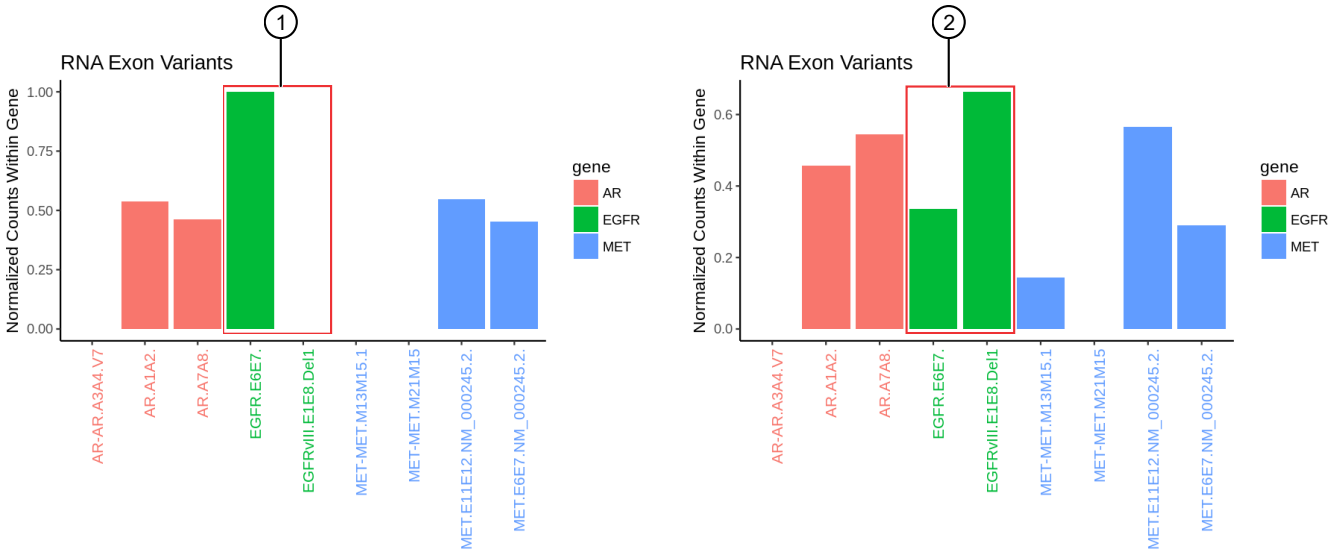
The **RNA Exon Variant** data view displays a bar graph summary of intragenic exon rearrangements or fusions. The displayed RNA exon variants are defined in the BED file that is associated with an assay. The **RNA Exon Variant** data view is available for all RNA and fusion assays.

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 82.

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 list.

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*.

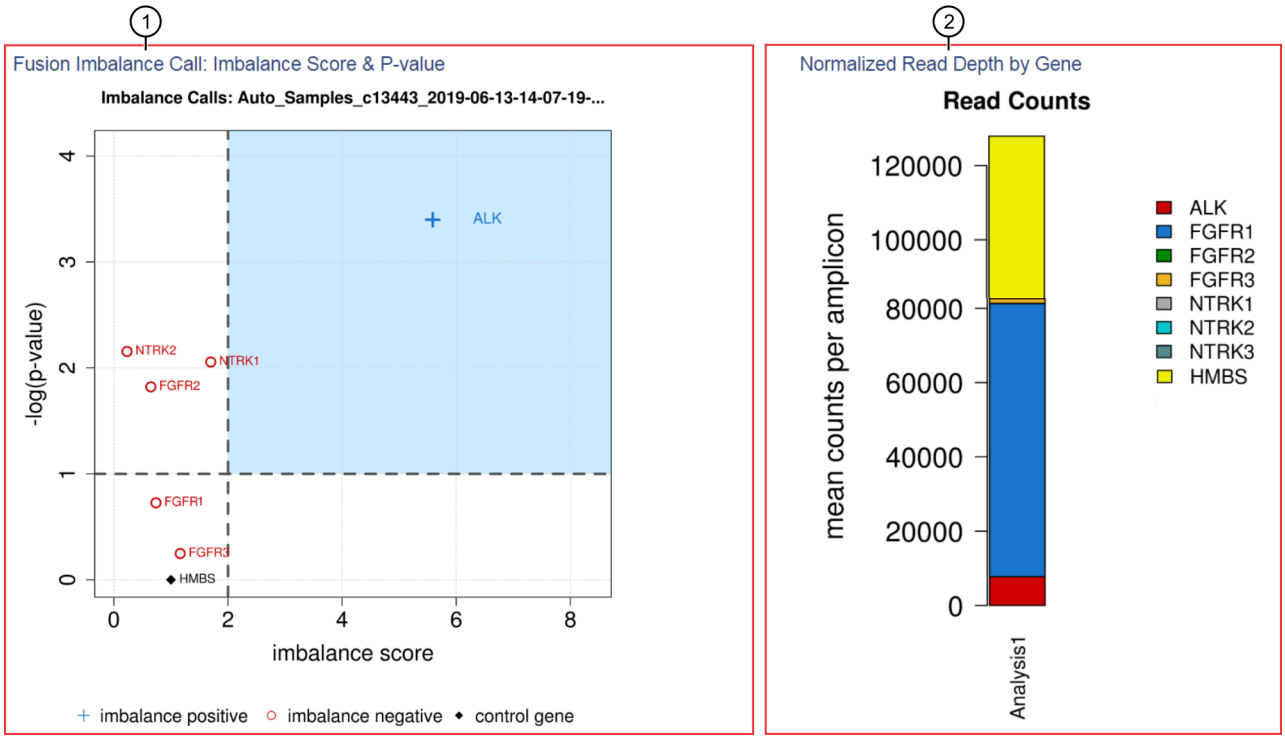
1. In the menu bar, click **Results** ▸ **Sample Results**.

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 82.

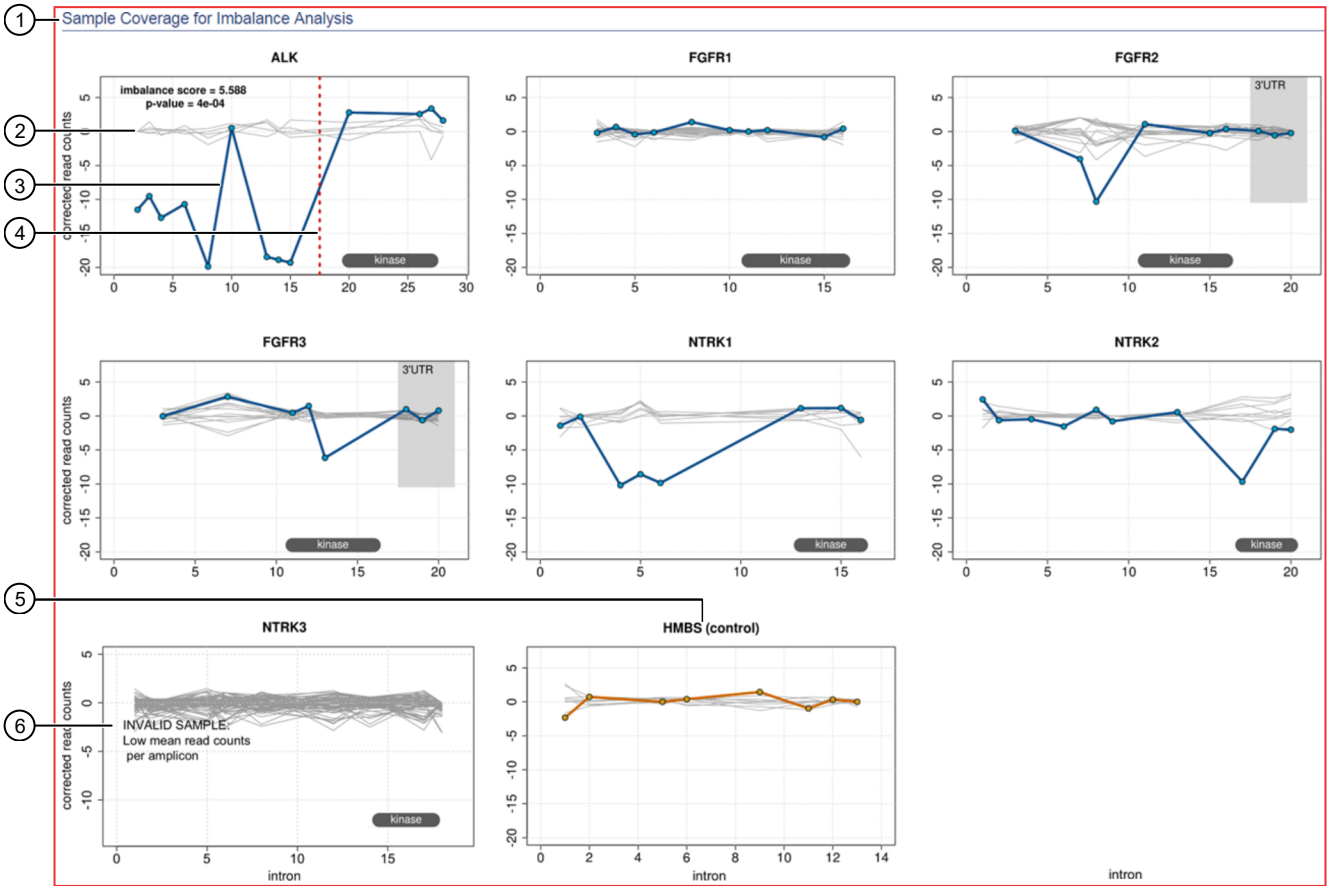
4. In the top right corner of the screen, click **Visualization** ▸ **RNA Exon Tile Fusion Imbalance**, then review the **RNA Exon Tile Fusion Imbalance** plots.

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 list.

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 87.

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 78.

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 223.

Column	Description
Variant ID	The identifier of the CNV variant. Click the link to view more annotation information. For more information, see “View more annotations and annotation sources” on page 88.
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. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds. • **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. • **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 88.
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 89.

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**. This option is available only when the forward strand is shown.
View the forward stand	Click **Forward**. This option is available only when the reverse strand is shown.
View the reference sequence translation	Click **Three-Frame Translate**. This option is available only when the amino acid sequence is not shown.
Hide the reference sequence translation	Click **Close Translation**. 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.

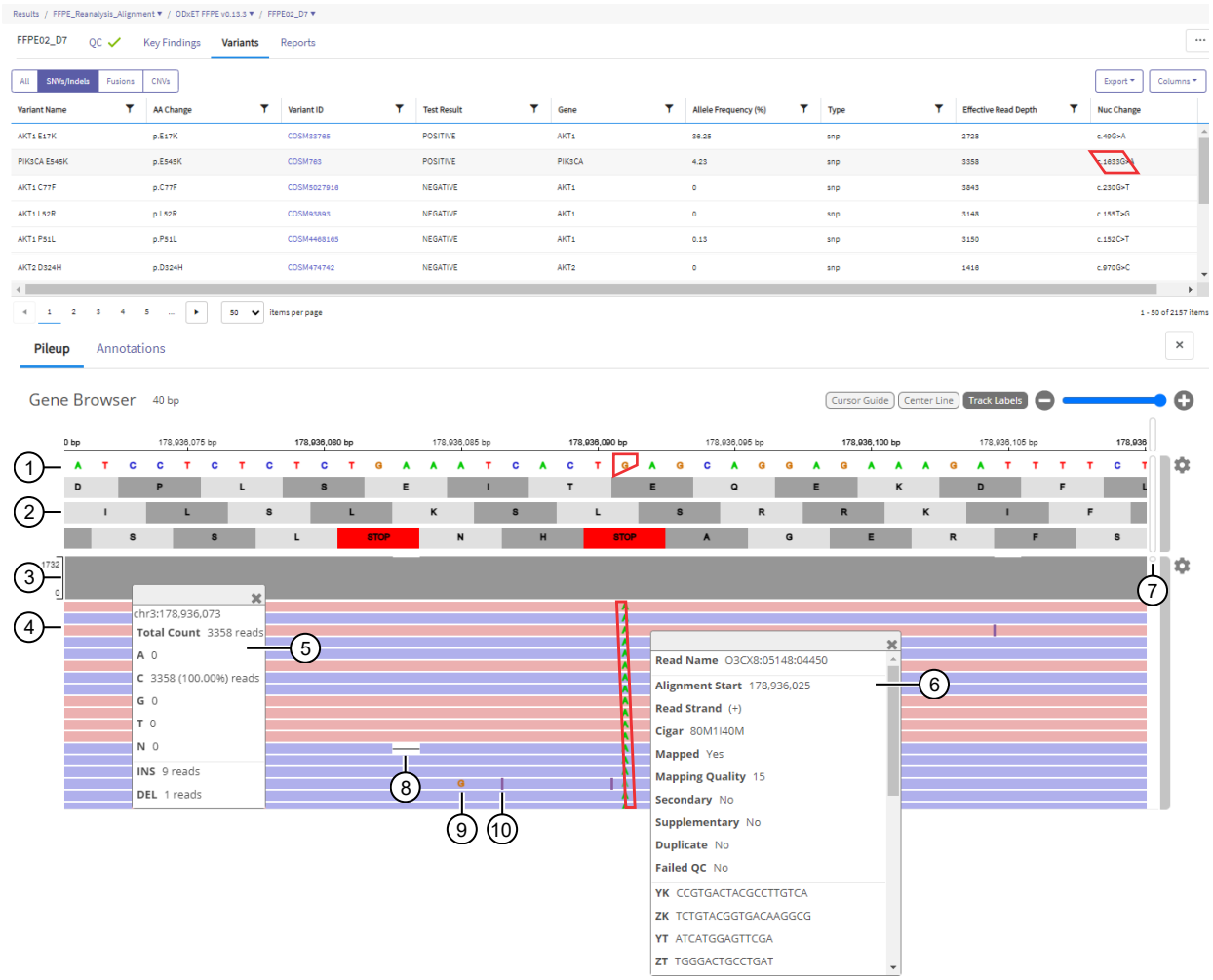
1. In the menu bar, click **Results** ▸ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

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**. 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**. 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 89.
Show all the bases in the BAM tracks	Click **Show all bases**. 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**. b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks. 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 89. 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 89.



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 90.

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**.

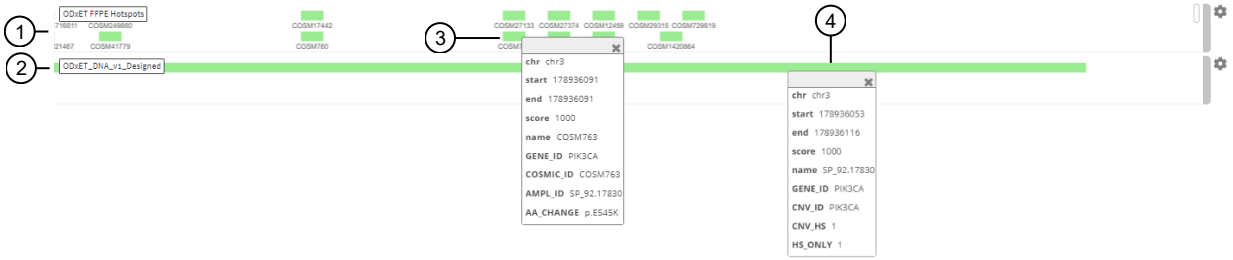
⑦ Use the slider to view more tracks.

⑧ Deletion.

⑨ Alternate base in the BAM read coverage track.

⑩ Insertion.

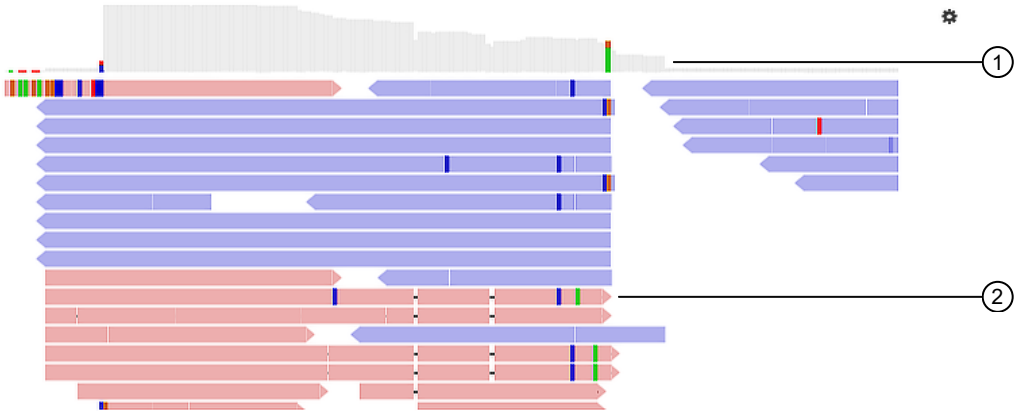
The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.



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.



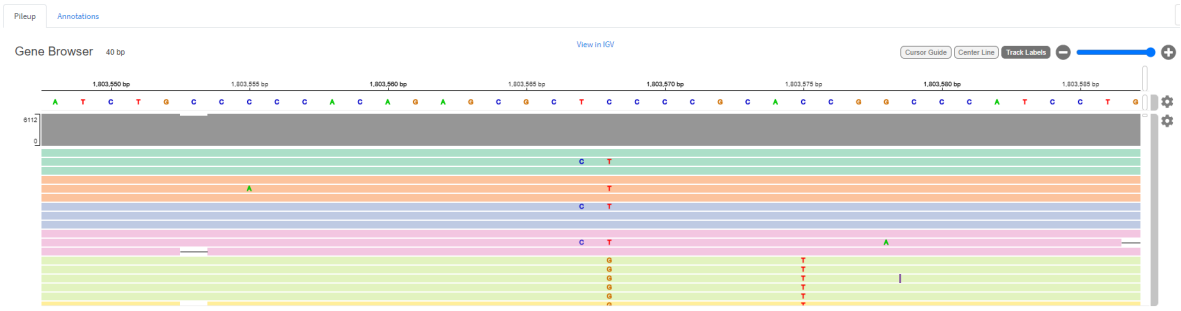
① Read depth
② Read location and direction of variants

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 89.
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 95.

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.
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.
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.
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**.

(continued)

Metric	Description
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.
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.

(continued)

Metric	Description
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.

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 98.

For information to filter sample results by a common trait, such as QC status, see “Filter the list of sample results” on page 99.

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 98.
Show or hide sample results that are archived	In the dropdown list in the upper left corner, select an archive status. • **Archived** • **Non Archived** 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. • **Completed** • **Running** • **Failed** • **Terminated** • **Stalled** • **Aborted** • **Pending** 2. Click **Filter**. 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. • **Failed** • **Not Calculated** • **Passed** 2. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the earliest date of the range. 4. In the Boolean operator dropdown list, select **And**. 5. In the second dropdown list, select **Is before or equal to**. 6. In the calendar that appears, select the latest date of the range. 7. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the earliest date for the sample results of interest. 4. In the Boolean operator dropdown list, select **Or**. 5. In the second dropdown list, select **Is before or equal to**. 6. In the calendar that appears, select the latest date for the sample results of interest. 7. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is before or equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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 102.

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 34.

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 35.

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 34.

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 35.
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 113 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 106. Runs that are reanalyzed are listed with **(Reanalysis)** after the run name. For more information, see “Reanalyze a run” on page 120.
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**). 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 53.
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 117.
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 105.
   - 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 109.

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. • **Delete**—Delete the run. • **Audit**—View the audit trail for the run. For more information, see "Review and download a run history" on page 65. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see "Download a Customer Support Archive" on page 227. • **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 117.
Actions[2]	• **View Plan**—View detailed run plan information. For more information, see "Run plan details" on page 63. • **Assign Kit Barcodes**—Assign purification kit barcodes used in the run. • **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**. For more information, see "Reanalyze a run" on page 120.

(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**. For more information, see "Customer Support Archive (CSA)" on page 227.
View the history of variant classifications.	In the upper right corner of the screen, click ••• **(More Options)** ▸ **Variant Audit**. 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 information *(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 Samples table *(continued)*

Item	Description
Conc. (ng/µl)	The concentration of the sample measured by the purification instrument. 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. 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. The status can be **N/A**, **Planned**, **Review**, **Started**, **Aborted**, or **Completed**.
Archive Position	The sample positions in the archive plate. 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. • ✔ (Checkmark) – indicates that the sample is selected for sequencing. • ⊘ – (No icon) indicates that the sample is not selected for sequencing. • **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 110.

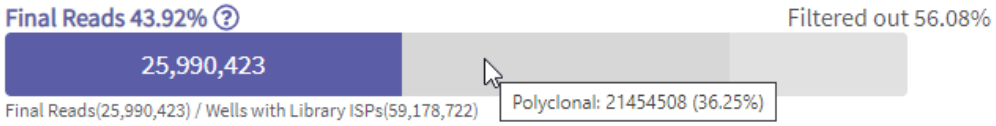
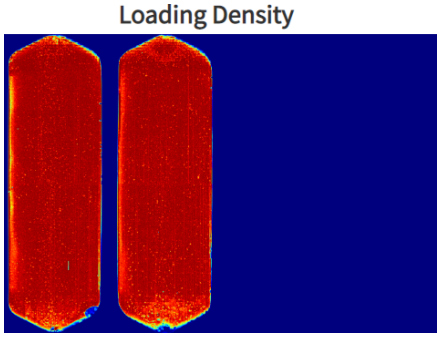
Assay metrics and the run report

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 *(continued)*

Metric	Description
Polyclonal	Wells with a live ISP that carries clones from two or more templates. To view polyclonal metrics, mouse over the first low quality portion (gray) of the **Final Reads** bar plot. 
Low Quality	Loaded wells with a low or unrecognizable signal. To view polyclonal metrics, mouse over the second low quality portion (gray) of the **Final Reads** bar plot. 
Adapter Dimer	Loaded wells with a library template of an insert size less than 8 bases. 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. 
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. 

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 110.

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 113.

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 115.

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 117.

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 113.
Show or hide run results that are archived.	a. In the dropdown list in the upper left corner, select an archive status. • **Archived** • **Non Archived** 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. • **Analysis Completed** • **Analysis In Progress** • **Analysis Failed** • **Analysis Aborted** • **Analysis Stalled** • **User Aborted** • **Run Aborted** • **Purification Completed** • **Purification Run Aborted** • **Purification Failed** • **Purification Review Required** • **Purification In Progress** • **Selected on Purification Instrument** • **Backup In Progress** • **Executing Plugins** • **Library Preparation Aborted** • **Library Preparation Completed** • **Library Preparation Started** • **Selected on Instrument** • **Sequencing Completed** • **Sequencing Started** • **Sequencing Aborted** • **Templating Aborted** • **Templating Completed** • **Templating Started** • **Kit Barcodes Required** b. Click **Filter**. 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)**. b. In the first dropdown list, select **Is equal to**.

Option	Description
	c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date of the range. d. In the Boolean operator dropdown list, select **And**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date of the range. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date for the sample results of interest. d. In the Boolean operator dropdown list, select **Or**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date for the sample results of interest. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is before or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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 181.

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 117.

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 117.

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 181.

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 113.

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. 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 119.

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. • When a run fails during the basecalling step or earlier. • When the run has not already been successfully reanalyzed. 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. 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. 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 170.

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 120.

Reanalysis at the alignment step uses BAM files.

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 124.

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).

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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 127.

(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. 2. Click ••• **(More Options)** ▸ **Download Files**.

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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. 2. Click the run or assay of interest from the results navigation bar. 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 122.

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 122.

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 122.

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 170.

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 143)

  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 110.

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 136.

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 134.

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 138.
Tumor Mutation Profiling Results: Summary	A summary of results for variants detected in the sample classified in 3 biomarker levels: • **Level 1: Companion Diagnostics (CDx) Biomarkers** • **Level 2: Cancer Mutations with Evidence of Clinical Significance** • **Level 3: Cancer Mutations with Potential Clinical Significance** If a detected variant is listed in the **Companion Diagnostics (CDx) Biomarkers** category, 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 132.
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 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 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. 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 138.
Tumor Mutation Profiling Results: Summary	A summary of results for variants detected in the sample classified in 3 biomarker levels: • **Level 1: Companion Diagnostics (CDx) Biomarkers** • **Level 2: Cancer Mutations with Evidence of Clinical Significance** • **Level 3: Cancer Mutations with Potential Clinical Significance** If a detected variant is listed in the **Companion Diagnostics (CDx) Biomarkers** category, 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 132.

(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 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 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. • **NTC QC – RNA**—A summary of the RNA no-template control quality control metrics. • **Sample QC – RNA**—A summary of RNA sample quality control metrics. • **Sample QC – DNA**—A summary of DNA sample quality control metrics. • **Templating Control QC – CF-1**—A summary of the CF-1 quality control metrics. • **Positive Control01 QC – DNA**—A summary of the DNA positive control quality control metrics. • **Positive Control01 QC – RNA**—A summary of the RNA positive control quality control metrics. • **Purification QC—A summary of QC metrics for the purification run.** This section is locked and cannot be customized.

(continued)

Section	Description
Sequencing Run Details	Contains the following subsections: • **Assay Name**—The assay used in the run • **Panel**—The panel used in the run • **Started On**—The date and time that the run was started • **Completed On**—The date and time when the run was completed • **Operator**—The user who sent the run to the instrument • **Instrument**—The name of the instrument • **Serial Number**—The serial number of the instrument This section is locked and cannot be customized.
Reagents	Details about run consumables, including part numbers, expiration dates, and lot numbers. 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. 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. 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 134.

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 138.
   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 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 127.

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 133.

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 166.

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 18.

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 222.

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 137.
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. • **Locked** • **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 129.

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 134.

- 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 129.

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**. 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)**. Repeat for each section to include in the report template. c. Click **Apply**.
Remove a section.	In the box for the section, click ✕ **(Remove)**.
Remove multiple sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click the section name of interest, then click « **(Remove)**. Repeat for each section to remove from the report template. c. Click **Apply**.
Add all sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, click **Select All**. c. Click **Apply**.
Remove all sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click **Select All**. 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 134.
- **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 142.

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**. b. Enter the note, then click **Save**. 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 110. 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 143.

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 110. 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 145.

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 146.

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***.

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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. • 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**. 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. • 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**. The filtered list of assays is shown in the **Manage Assays** screen. • 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. • 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**. The filtered list of assays is shown in the **Manage Assays** screen. • 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. 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: • **Locked**
Reference	Genome reference that is available in the annotation set. • **hg19** • **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 152.

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. 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** • **CF-1 Control** • **Purification QC** • **NTC QC – DNA** • **Run QC** • **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: • **Create** **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:

- View system information and settings.

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. b. In the app, click ＋. 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. b. In the Connect website, click **InstrumentConnect** in the left sidebar. c. Click **Link instrument** in the upper right corner of the screen. d. Enter the QR code text shown in the **Link Instrument** screen. 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 166.

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 166.

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 (administrator)

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 166.

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**. • 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. • 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.

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.

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 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 134.

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. 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 157 and “Edit user details (administrator)” on page 158.

(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. 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 158.
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**. 2. In the **Audit Trail - Electronic Signature** screen: 3. • Review the audit trail details for the most recent change. • Click a time stamp of interest to view the details for other changes that have been made to the selected signature. • Click **Download** to export the record of the single change to a PDF file. • 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 165.

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 184.

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 open the **Software Updates** screen.

- The currently installed software versions for each software module, assay definition files, plugin, configuration package or security patch update that is installed are shown when the **Installed Software** link is selected.
- New software updates that are available to be downloaded and installed are shown when the **Software Updates** link is selected.

Security patch updates and security configuration updates for Genexus™ Software do not include any software features. These update packages are identified by fourth digit in the software version number. For example, Genexus™ Software `x.x.x.1`.

After the system update is installed, entries are logged in the installation history and audit history in Genexus™ Dx Software.

For users who have set up a Thermo Fisher™ Connect Platform account, **(Notifications)** appear 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 167
Download and install software with a USB drive	"Format a USB drive for a software update" on page 168 "Download and install software with a USB drive (administrator)" on page 169

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 do the following.

- Configure a Thermo Fisher Connect account. For instructions, see "Configure a Thermo Fisher™ Connect Platform account with a QR code (manager/administrator)" on page 155.
- Ensure that the Genexus™ Dx Integrated Sequencer has the latest software version, Genexus™ Dx Software 6.8.2.1.
- Connect the Genexus™ Dx Integrated Sequencer to the internet.

When software updates are downloaded to a computer from the Thermo Fisher App Store on Connect, the checksum information for the downloaded software updates are verified by Genexus™ Dx Software before the software package is installed.

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 155.
- Format a USB drive for the software update. For more information, see “Format a USB drive for a software update” on page 168.

When software updates are downloaded to a USB drive from the Thermo Fisher App Store on Connect, checksum information is available in the App Store screen. Use this information to confirm that the correct software update package is downloaded to the USB drive.

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. Verify that the checksum of the downloaded software package is the same as the checksum listed on the App Store.
8. Copy the downloaded software package (ZIP file) to the prepared USB drive.
9. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the Genexus™ Integrated Sequencer.
10. In the menu bar of Genexus™ Dx Software, click **(Settings)** ▸ **Software Updates**, then click **Software Updates** in the upper right corner.
11. Select **USB**.
12. 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 179.

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. 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. 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. 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.
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. 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. 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. 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 helps ensure 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 185.

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 • VCF • 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 170.

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 181.

[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 122.

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 178.

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. 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. 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. For more information, see “Add or change the backup directory (administrator)” on page 175.
Change the email notification settings	Select when administrator-level users receive email notifications. For more information, see “Change email notification settings (administrator)” on page 184.
Change the backup settings	Backup .wells files and intermediate files. For more information, see “Change the backup settings (administrator)” on page 179.
Change the disk cleanup settings	Set the schedule to permanently delete analysis results files. Set the conditions to permanently delete other files. For more information, see “Set a schedule to permanently delete files (administrator)” on page 181.
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. This setting overrides all other backup settings.

(continued)

Option	Description
Start backup	Click **Start Now** to start: • the backup of system data and analysis results to the selected backup directory. • if enabled, the backup of sequencing output and intermediate files to the selected backup directory. • data clean up based on cleanup settings. Backup can take a considerable amount of time when the backup is set up for first time on the system.

3. Click **Save**.

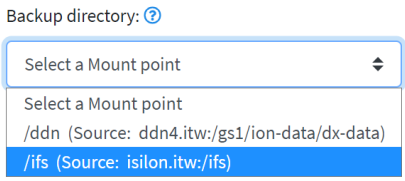
Add or change the backup directory (administrator)

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**. b. In the **Add Mount** dialog box: • Enter the name of the mount directory. For example, ***ddn***. **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. You can type a new name to create a new directory. • Enter the storage source. For example, ***ddn.itw:/gs1/ion-data/dx-data*** c. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is restarted. 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. You must add a mount point before it can be selected in the dropdown. 
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**. b. In the **Remove Mount** dialog box, select the mounted drive you want to remove. c. Click **Remove**. 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.

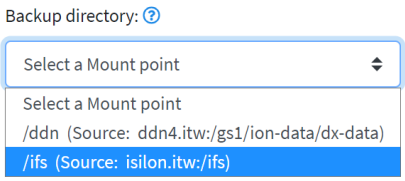
Add or change the cloud backup directory (administrator)

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**. 2. In the **Add Mount** dialog box: • Enter the name of the **Mount Point directory**. For example, ***ddn***. You can type a new name to create a new directory. • Enter the **Cloud Provider**. For example, ***AWS-S3***. 3. Enter the **Configuration** information. • **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. **Note:** The following information must be entered correctly to help ensure that the mount works correctly. Information should be provided by an administrator for the network or Amazon™ Web Services (AWS). • **Bucket Name**—The name of the AWS S3 bucket. • **Access Key Id**—A key identifier that, similar to a username, provides programmatic and CLI access to AWS resource, such as the S3 bucket • **Secret Access Key**—An access key that, similar to a password, provides programmatic and CLI access to AWS resource, such as the S3 bucket. • **Region**—The AWS region in which for the S3 bucket resides. For example, ***us-east-1***, ***us-west-1***. 4. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is rebooted. When the option is unselected, you must mount a directory after a reboot. 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** dropdown list. You must add a mount point before for the backup directory can be selected. 
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**. 2. In the **Remove Mount** dialog box, select the mounted drive you want to remove. 3. Click **Remove**. 4. Click **Save**. 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 175.

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 170.

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 173.

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. 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. 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 117.

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. 1. In **Delete Analysis Results**, click **Yes**. 2. In **months after completion**, set the schedule to delete the files: • 3 months • 6 months • 9 months • 12 months • 24 months – default setting • 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. The system automatically deletes .wells and intermediate files when the disk space falls below the selected threshold. 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 help 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 161.

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 analyses 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 184.

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 166.

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 170 and “Add or change the backup directory (administrator)” on page 175. 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 181. 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 185.
- For information about how to restore or import multiple run results, see “Restore or import multiple archived run results (administrator)” on page 185.

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. **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.

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 30.
- 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 152.

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 34.

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**. 2. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 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**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to restore, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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**. 2. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 3. In the **Select backup folder to list runs** dialog box, navigate to the folder for a different system, then click **Select**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to import, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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. 1. In the type of runs dropdown list, select **All runs**. 2. In the dropdown list, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 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**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to restore, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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. For more information, see “Start a stopped service (manager/administrator)” on page 192.
Active Jobs	• Shows a list of runs in progress that are active and queued on the sequencer. • The message “No records available” indicates that no jobs are currently active. • Allows manager- and administrator-level users to stop a job. For more information, see “Terminate an active job (manager/administrator)” on page 192.
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 192.

(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.



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 227.

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 For more information, see “Get report templates” on page 206.	Returns report templates.	`https://<host>/genexus/api/lims/v2/report-templates`
Get a list of assays For more information, see “Get assay list” on page 197.	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 For more information, see “Get samples” on page 198.	Returns a list of the samples in the system.	`https://<host>/genexus/api/lims/v2/samples`
Get a list of library batches For more information, see “Get library batches” on page 199.	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 For more information, see “Get run plans” on page 202.	Returns a list of available run plans in the system.	`https://<host>/genexus/api/lims/v2/plans`
Get results For more information, see “Get results” on page 207.	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 For more information, see “Get signed off samples” on page 212.	Returns a list of the samples that are designated with ***(Signed Off)***.	`https://<host>/genexus/api/lims/v2/signedOffSamples`
Download files for completed runs For more information, see “Download files for completed runs” on page 213.	Returns a compressed file in ZIP format that contains the requested files for the sample.	`https://<host>/genexus/api/lims/v2/download`
Create samples For more information, see “Create samples” on page 213.	Create samples.	`https://<host>/genexus/api/lims/v2/samples`
Create a library batch For more information, see “Create a library batch” on page 215.	Create a library batch from existing samples.	`https://<host>/genexus/api/lims/v2/library-batches`
Create a Sample to Result (purification) run For more information, see “Plan a Sample to Result run” on page 54.	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 For more information, see “Create a Nucleic Acid to Result run” on page 216.	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 For more information, see “Create a library run” on page 217.	Create a library run from one or more library batches and an assay.	`https://<host>/genexus/api/lims/v2/plan-by-library`

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 Accept:application/json
Response	Content-Type:application/json

Response fields

Code	Status
200	Successful response
400	Bad request Error response: `{ "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". 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` String. *Optional*. Assay name. • `status` String, *Optional*. Valid values are "draft", "obsolete", or "locked".

Example response	{
	"total": 1,
	"objects": [
	{
	"id": 18,
	"assayName": "Generic_DNA_and_Fusions_EU_v0.3.0",
	"assayShortName": "Generic DNA-Fus EU v0.3.0",
	"assayDisplayedName": "Generic - DNA and Fusions - EU v0.3.0",
	"application": "DNA+RNA",
	"createdBy": "System Installed",
	"createdDate": "2021-07-14T00:05:12.000+0000",
	"isReanalysisAssay": false,
	"genomeReference": "hg19",
	"targetRegions": "Ronin_w2.7.0_v0.3.0_dnafusion_FFPE_DNA_v1.20210604.designed.bed",
	"hotspots": "Ronin_w2.7.0_dnafusion_FFPE_DNA.v1.20210608.hotspots_0.3.0.bed",
	"fusionReference": "Ronin_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.fasta",
	"fusionTargetRegions":
	"Ronin_w2.7.0_dnafusion_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.bed",
	"chipType": "Genexus™ Dx GX5™ Chip",
	"panelKit": "Generic Assay Panel (IVD)",
	"controlKit": "",
	"extractionKit": "",
	"libraryKit": "Genexus™ Dx Library Strips 1 and 2-HD (IVD)",
	"templatingKit": "Genexus™ Dx Templating Strips 3-GX5™ and 4 (IVD)",
	"barcodeKit": "Genexus™ Dx Barcodes 1-32 HD (IVD)",
	"sequencingKit": "Genexus™ Dx Sequencing Kit (IVD)",
	"flows": 500,
	"templatingSize": 200,
	"status": "Locked",
	"tumorCellularity": "1",
	"reanalysisAssay": false
	}
	]
	}

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 -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/samples'`

(continued)

samples	
Request query parameters	• `sampleId` String. *Optional*. 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` String. *Required*. Library batch ID.

(continued)

library-batches	

Example response	{
	  "total": 1,
	  "objects": [
	  {
	  "libraries": [
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_05",
	  "libraryName": "NTC_DNAST_LibBatch_DNA_RNA_3",
	  "inputQuantity": 87.0,
	  "sampleId": "NTC",
	  "tumorCellularity": null,
	  "controlKitBarcode": "01101903020165751724060110123456724 0A51339",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_06",
	  "libraryName": "NTC_RNAST_LibBatch_DNA_RNA_3",
	  "inputQuantity": 55.3,
	  "sampleId": "NTC",
	  "tumorCellularity": null,
	  "controlKitBarcode": "01101903020165751724060110123456724 0A51339",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_03",
	  "libraryName": "PositiveControl01_DNAST_LibBatch_DNA_RNA_1",
	  "inputQuantity": 87.0,
	  "sampleId": "PositiveControl01",
	  "tumorCellularity": null,
	  "controlKitBarcode": "01101903020165441725093010260745 5240A51337",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_04",
	  "libraryName": "PositiveControl01_RNAST_LibBatch_DNA_RNA_1",
	  "inputQuantity": 55.3,
	  "sampleId": "PositiveControl01",
	  "tumorCellularity": null,
	  "controlKitBarcode": "01101903020165441725093010260745 5240A51338",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_01",
	  "libraryName": "LIMS_Sample_55_DNAST_LibBatch_DNA_RNA_2",
	  "inputQuantity": 57.0,
	  "sampleId": "LIMS_Sample_55",
	  "tumorCellularity": null,
	  "controlKitBarcode": null,
	  "extractionKitBarcode": "91100090559101234567172110 19"
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_02",
	  "libraryName": "LIMS_Sample_55_RNAST_LibBatch_DNA_RNA_2",
	  "inputQuantity": 51.3,
	  "sampleId": "LIMS_Sample_55",
	  "tumorCellularity": null,
	  "controlKitBarcode": null,
	  "extractionKitBarcode": "91100090560101234567172112 31"
	  }
	  ],
	
	  "id": 13,
	  "assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",
	  "libraryBatchId": "ST_LibBatch_DNA_RNA",
	  "libraryKitBarcode": "",
	  "panelKitBarcode": "",
	  "libraryPrepType": "DNA+RNA",
	  "ampliSeqHd": false
	  }
	  ]
	}

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` String. *Optional*. Name of the run plan. • `viewType` String. *Optional*. Valid values are "all" or "toBeProcessed".
Example response	{ "objects": [{ "id": 1, "planName": "EK_Generic_FFPE_EU_Ronin_D5_0.3.0_6.5.26", "planShortId": "EQ55D", "type": "sample", "createdBy": "System Installed", "updatedOn": "2021-07-17T20:33:48.000+0000", "noOfSamples": 8, "noOfLibraries": 16, "samples": [{ "id": 8, "sampleId": "NTC", "assayName": "Generic - DNA and Fusions - EU v0.3.0", "dnaBarcodes": ["IonDxHDdual_28"], "rnaBarcodes": ["IonDxHDdual_32"], "libraryBatchId": "1", "libraryPrepType": "DNA+RNA", "pauseForReview": false }, { "id": 5, "sampleId": "SRTestSample64", "assayName": "Generic - DNA and Fusions - EU v0.3.0", "dnaBarcodes": ["IonDxHDdual_25"], "rnaBarcodes": ["IonDxHDdual_29"], "libraryBatchId": "1", "libraryPrepType": "DNA+RNA", "pauseForReview": false },

(continued)

plans	

Example response, continued	{
	"id": 6,
	"sampleId": "SRTestSample65",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_26"
	],
	"rnaBarcodes": [
	"IonDxHDdual_30"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 2,
	"sampleId": "SRTestSample61",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_18"
	],
	"rnaBarcodes": [
	"IonDxHDdual_22"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 7,
	"sampleId": "SRTestSample66",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_27"
	],
	"rnaBarcodes": [
	"IonDxHDdual_31"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	}, {
	"id": 3,
	"sampleId": "SRTestSample62",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_19"
	],
	"rnaBarcodes": [
	"IonDxHDdual_23"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 4,
	"sampleId": "SRTestSample63",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"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	{
	"id": 50,
	"sampleId": "SRTestSample61",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_18"
	],
	"rnaBarcodes": [
	"IonDxHDdual_22"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 53,
	"sampleId": "SRTestSample64",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_25"
	],
	"rnaBarcodes": [
	"IonDxHDdual_29"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 54,
	"sampleId": "SRTestSample65",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_26"
	],
	"rnaBarcodes": [
	"IonDxHDdual_30"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 51,
	"sampleId": "SRTestSample62",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_19"
	],
	"rnaBarcodes": [
	"IonDxHDdual_23"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 56,
	"sampleId": "NTC",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_28"
	],
	"rnaBarcodes": [
	"IonDxHDdual_32"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	}
	],
	"runAssays": [
	"Generic Assay - DNA and Fusions - EU v0.3.0"
	]
	}
	],
	"total": 2
	}

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 -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/report-templates'`
Request query parameters	`assayName` String. *Optional*. Get the report template for the given assay. Use the **Assay Name**. Do not use the **Assay Full Name**.

Example response	{
	"mode": "dx",
	"reportingTemplate": [
	{
	"name": "LAB_ODxET_DNA_QC_6.8_v1.0.0",
	"id": 22,
	"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - DNA QC v1.0.0",
	"assayId": 41,
	"description": "ODxET DNA QC v1.0.0 - Lab Report",
	"locale": "en"
	},
	{
	"name": "ev_template",
	"id": 21,
	"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",
	"assayId": 40,
	"description": "EV_Template",
	"locale": "en"
	}
	],
	"total": 2
	}

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` String. *Optional*. Results for the selected result id. Example: ***123*** • `assay_id` String. *Optional*. Results for the assay id. Example: ***123*** • `plan_id` String. *Optional*. Results for selected run plan id. Example: ***123*** • `sampleId` String. *Optional*. Results for the selected sample name. Example: ***MySample*** • `planName` String. *Optional*. Results for selected run plan name. Example: ***MyPlan*** • `analysisComplete` String. *Optional*. Results for completed runs. Results included for failed and successfully completed runs. Example: ***true*** • `updatedBefore` String. *Optional*. Returns results updated before the selected date. Use date format: `yyyy-MM-dd HH:MM`. Example: ***2019-01-15 10:30*** • `updatedAfter` String. *Optional*. Returns results updated after the selected date. Use date format: `yyyy-MM-dd` Example: ***2019-01-15*** • `orderBy` String. *Optional*. 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": []
                }
            ],
            "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 -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/signedOffSamples'`
Request query parameters	• `start_date` String. *Optional*. Provide the start date as yyyy-mm-dd. • `end_date` String, *Optional*. Provide the end date as yyyy-mm-dd. • `signedOff` String, *Required* A **Boolean** data type. Valid values are "true" or a "false". 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 -X GET --header 'username:<user name>' --header 'password: <user password>' 'https://<host>/genexus/api/lims/v2/download'`
Request query parameters	• `file_list` String. *Required*. Provide the file name in comma separated format. • `path` String, *Required*. 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 212.

Example request	1. Perform a checksum function to query for the list of files for a sample that has been sequenced and analyzed.
	`https://BASE_URL/download?file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q`
	2. Parse the checksum to get the file name, then request that file.
	`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`
	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>' --header 'Content-Type: application/json'`

(continued)

samples	
Example URL	`curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/samples'`
Attribute definitions	• `Patient Id`. String. *Required*. A unique identifier for the patient. • `Date of Birth`. String. *Required*. The date of birth for the patient. • `Ordering Physician`. String. *Required*. The name of the physician who created the order for the sample. • `sampleId`. String. *Required*. Sample name. • `sampleType`. String. *Required*. Examples of valid values are "DNA" or "RNA". • `collectionDate`. String. *Required*. The date that the sample was collected. • `gender`. String. *Required*. The biological sex of the sample: Female, Male, or Unknown. • `diseaseCategory`. String. *Required*. The sample application category. • `cancerStage`. String. *Required*. Cancer stage. • `origin`. String. *Required*. Cancer type. • `notes`. String. *Required*. An open-entry field for more sample information. • `necrosis`. String. *Optional*. The percentage of cellular necrosis in the sample. • `tumorCellularity`. String. *Optional*. The percentage of tumor cells over normal cells in the sample. • `condition`. String. *Optional*. The condition of the sample.

Example request	{
	"objects":[
	{
	"sampleId":"Sample_NSCLC_99",
	"diseaseCategory":"Cancer",
	"sampleType":"DNA & RNA",
	"origin":"Non-Small Cell Lung Cancer",
	"cancerStage":"Unknown",
	"necrosis":100,
	"tumorCellularity":31,
	"gender":"Unknown",
	"collectionDate":"2018-10-02",
	"condition":"good",
	"tags":["NSCLC"],
	"notes":"LIMS API created Cancer sample and existing Tag NSCLC",
	"otherAttributes":
	{
	"Patient Id":"MRND12121",
	"Ordering Physician":"Dr Fred",
	"Date of Birth": "2000-01-01"
	}
	}
	]
	}

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>' --header 'Content-Type: application/json'`
Example URL	`curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/plan-by-purification'`

Example request	<pre>{
	"reportTemplateName": "",
	"samples":[
	{
	"assayName": "Oncomine™ Dx Express Test - DNA and Fusions - US v0.3.0",
	"sampleId":"Test_Sample4",
	"purificationProtocol": "FFPE DNA/RNA",
	"elutionVolumeDNA": 40,
	"elutionVolumeRNA": 40,
	"pauseForReview": false
	}
	],
	"notes":"Sample to Result run created by LIMS API",
	"planName":"PurificationPlan_DNF_US_1Sample_01"
	}</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>' --header 'Content-Type: application/json'`

(continued)

plan-by-samples	
Example URL	`curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' 'https://<host>/genexus/api/lims/v2/plan-by-samples'`
Example request	`{ "reportTemplateName": "", "samples":[{ "assayName": "Generic Assay - DNA and Fusions - EU v0.3.0", "sampleId":"Test_Sample1" }, { "assayName": "{{DNA_RNA_Assay}}", "sampleId":"Test_Sample2" }, { "assayName": "{{DNA_RNA_Assay}}", "sampleId":"Test_Sample3" }, { "assayName": "{{DNA_RNA_Assay}}", "sampleId":"Test_Sample4" }, { "assayName": "{{DNA_RNA_Assay}}", "sampleId":"Test_Sample5" }, { "assayName": "{{DNA_RNA_Assay}}", "sampleId":"Test_Sample6" }], "notes":"Nucleic Acid Run created by LIMS API", "planName":"NucleicAcidRun_DNF_6Samples_Run" }`

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>' --header 'Content-Type: application/json'`

(continued)

plan-by-library	
Example URL	`curl -k -X POST --header 'username:<user name>' --header 'password: <user password>' --header 'Content-Type: application/json' --data '<input payload>' '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

Variant calling criteria

In addition to run and clinical sample validity criteria, 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 *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 *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 *PTEN* deletion
RNA exon variants	Specific RNA exon variants, identified by variant ID.	*MET* exon 14 skipping *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 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.

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. This plugin generates QC metrics. For more information, see “QC results” on page 95.
molecularCoverageAnalysis	This plugin generates QC metrics. For more information, see “QC results” on page 95.

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. 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. **IMPORTANT!** To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample. **Note:** • Samples that have been used in a run cannot be deleted. • 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. Click **Calendar** to select the date in the correct format.
Gender[1]	The biological sex of the sample: **Female**, **Male**, or **Unknown**. **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**. **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. 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. 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. **IMPORTANT!** • If this value is not set, **% Cellularity** is assumed to be 100% in calculations that use this attribute. • **% Cellularity** is a required attribute for CNV analyses. Do not leave the field blank. • 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 87.
% 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**



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. 2. Plan a new run for the sample or sample library with the correct assay. 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. MAN1001774).

Documentation and support

Customer and technical support

Visit **thermofisher.com/support** for the latest service and support information.

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- 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	MAN1001240
Genexus™ Dx Purification System User Guide	MAN1000137
Oncomine™ Dx Express Test Part I: Test Description and Performance Characteristics User Guide	MAN1001774
Oncomine™ Dx Express Test Part II: Sample Purification, Sequencing, and Results Reports User Guide	MAN1001775

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**.

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**.

Glossary

Genexus™ Dx Software definitions

adapter dimer (ISPs)

Primer and adapter sequences that anneal during library preparation or templating to form a short fragment that can be sequenced. These adapter dimer ISPs generate unusable reads that the software filters out of the final sequencing data. High levels of adapter dimer ISPs can reduce the number of valid reads.

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 targeted sequencing 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 variants. Specifying a hotspots file to use in a run enables the Torrent Variant Caller (TVC) module to identify variants that may be present in sample DNA. A hotspots file instructs the Torrent Variant Caller (TVC) 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

The Median of the Absolute values of all Pairwise Differences (MAPD) score is reported on runs that detect CNVs.

MAPD is one metric that is used to determine 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 of each tile for a 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 an estimate of copy number variability in each sequencing run that is 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.

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 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 filters.

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 that contain ISPs have a delayed pH change due to the presence of an ISP that slow 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 Purification System

USER GUIDE

Catalog Number A53580

Publication Number MAN1000137

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 Purification System

Life Technologies Corporation |
7335 Executive Way |
Frederick, Maryland 21704 USA

Products manufactured at this site:

- Genexus™ Dx FFPE DNA/RNA Purification Kit

For descriptions of symbols on product labels or product documents, go to thermofisher.com/symbols-definition.

Revision history: B MAN1000137 (English)

Revision	Date	Description
B	13 January 2026	• Updated requirements for use of an uninterruptible power supply. • Added software compatibility, firewall, and data backup information.
A	8 May 2025	Genexus™ Dx Purification System user guide for regulatory body 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 Purification System in standalone configuration (not connected to the Genexus™ Dx Integrated Sequencer). This guide provides detailed information about the product, accessory materials, preparation for use, main protocols, troubleshooting, and instrument maintenance. In addition, the guide provides basic instructions for sample entry and run planning through the instrument touchscreen.

Prerequisites

Category	Prerequisites
Instrument	• Genexus™ Dx Purification System • Reagents and supplies for operating the Genexus™ Dx Purification System
Functional knowledge and understanding	Main functions of the Genexus™ Dx Purification System
Software	Genexus™ Dx Software 6.8.2 or later

1 Product information

Product description

The Ion Torrent™ Genexus™ Dx Purification System automates purification and quantification of DNA and RNA from formalin-fixed paraffin-embedded (FFPE) tissue samples.

The Ion Torrent™ Genexus™ Dx Purification System is intended for use with the Genexus™ Dx FFPE DNA/RNA Purification Combo Kit.

Intended use

Ion Torrent™ Genexus™ Dx Purification System

The Genexus™ Dx Purification System is a sample preparation instrument which automates nucleic acid extraction, purification, and quantification of DNA and RNA from FFPE tissue lysates and cfTNA from plasma for use in next-generation sequencing applications. The system uses MagMAX™ magnetic bead-based technology to perform nucleic acid extraction. Purified nucleic acid is quantified using the onboard Qubit™ Fluorometer which uses fluorescence to measure nucleic acid.

Ion Torrent™ Genexus™ Dx FFPE DNA/RNA Purification Combo Kit

The Genexus™ Dx FFPE DNA/RNA Purification Combo Kit provides components for automated nucleic acid isolation, and quantification from FFPE tissue. The kit uses MagMAX™ magnetic bead-based technology for isolation and purification of genomic DNA and cellular RNA from formalin-fixed paraffin-embedded (FFPE) tissue. The isolated DNA and RNA are quantified using the onboard Qubit™ Fluorometer and fluorescence-based quantification reagents provided in the kit.

The product is intended for professional users, such as technicians who are trained in molecular biological techniques. The Genexus™ Dx FFPE DNA/RNA Purification Combo Kit is for in vitro diagnostic use.

Theory of operation

Overview

Formalin-fixed paraffin-embedded (FFPE) tissue lysates are treated with protease, then loaded into the Genexus™ Dx Purification System along with consumables needed for automated purification and quantification of DNA and RNA. On completion of the run, purified and quantified nucleic acid samples are automatically transferred into a 48-well nucleic acid archive plate.

Sample preparation

Protease-treated FFPE lysates are 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 into a 48-well nucleic acid archive plate.

Indications for use

Same as intended use.

Genexus™ Dx Purification System

The Genexus™ Dx Purification System (Cat. No. A53580) is an orderable item that includes the instrument shown in the following table.

Instrument	Cat. No.
Genexus™ Dx Purification System	A53257

The Genexus™ Dx Install Kit (Cat. No. A52172, not available for separate purchase) is provided to first-time owners of a Genexus™ Dx Purification System and is shipped with the instrument. The kit contains the following supplies that are required by the field service engineer for installation and qualification of the instrument.

Component	Amount	Storage
Genexus™ Dx Purification System Install Kit (Part. No. A48549)	1	15°C to 30°C
Genexus™ Purification Integrated Workflow PQ Controls (Part. No. A49291)	8 AutoLys Tubes, each containing 1 control curl	2°C to 8°C
Genexus™ Dx FFPE DNA and RNA Purification Kit (Part. No. A56024)	1 (48 reactions)	15°C to 30°C
Genexus™ Dx Nucleic Acid Quantitation Kit (Part. No. A56025)	1 (48 reactions)	2°C to 8°C
Genexus™ Dx Purification Supplies 2 (Part. No. A56026)	1 (48 reactions)	15°C to 30°C

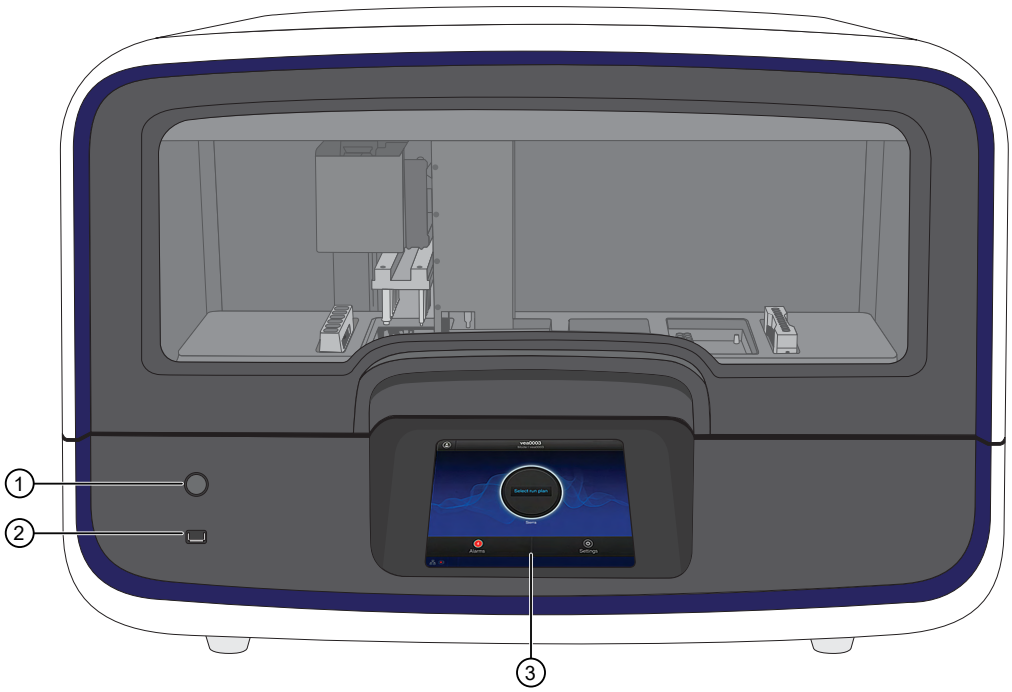
Also shipped with the system is the Genexus™ Dx AutoLys Accessories Kit for using AutoLys Tubes.

Components	Cat. No.
Genexus™ Dx AutoLys Accessories Kit • Ion Torrent™ Genexus™ Dx AutoLys Tube Rack; Part No. A58869 • Ion Torrent™ Genexus™ Dx AutoLys Pliers; Part No. A58870 • Ion Torrent™ Genexus™ Dx AutoLys Locking Lid; Part No. A58871	A58873

Warnings and limitations

- Use of this product must be limited to personnel trained in the use of the Genexus™ Dx Purification System.
- The Genexus™ Dx Purification System has only been validated for use with FFPE tumor specimens.
- Nucleic acids extracted, purified and quantified with the Genexus™ Dx Purification System have been validated for use with the Genexus™ Dx Integrated Sequencer.
- Both the DNA and RNA from a single sample extraction must meet the concentration requirements specified in the downstream analysis method.
- The effects of potential variations in FFPE specimen fixation have not been evaluated.
- 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.

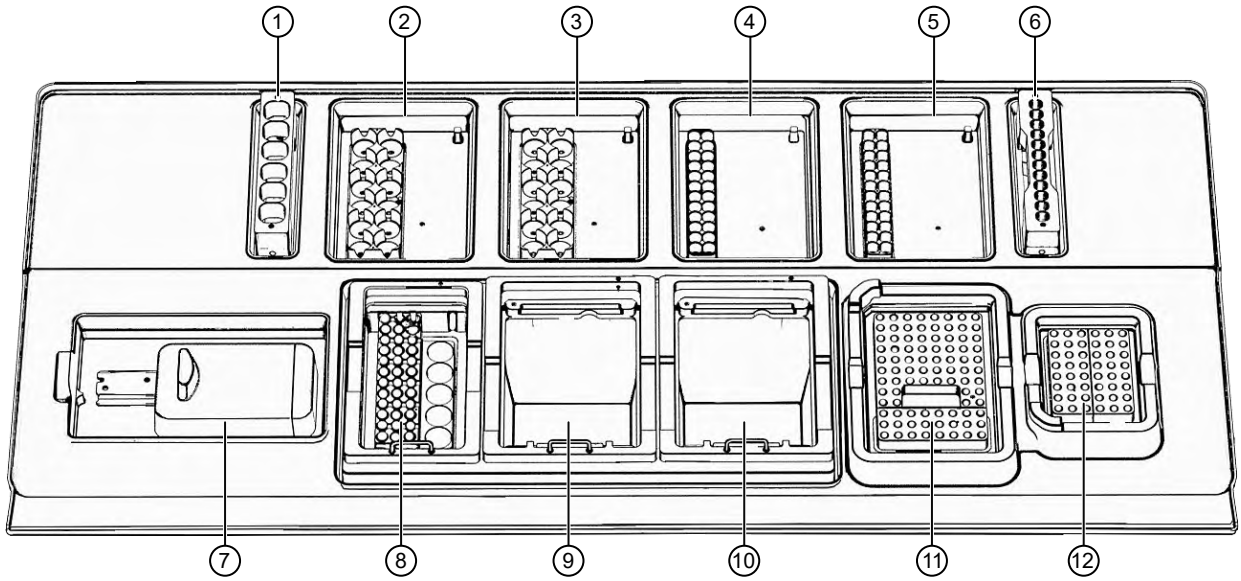
Genexus™ Dx Purification System features



External features of the Genexus™ Dx Purification System.

① Power button
② USB port
③ Touchscreen

Genexus™ Dx Purification System deck stations

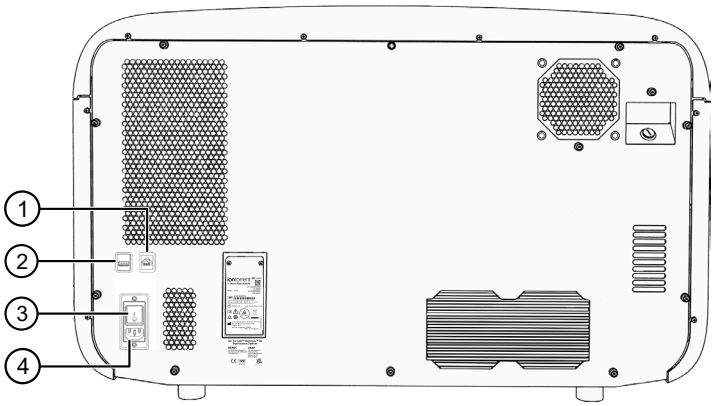


Interior deck stations of the Genexus™ Dx Purification System.

1. 6-well tip comb station
2. 24 deepwell plate station 1
3. 24 deepwell plate station 2
4. 96 deepwell plate station1
5. 96 deepwell plate station 2
6. 12-well tip comb station
7. Quantitation module
8. Quantitation tray
9. Tip box station 1
10. Tip box station 2
11. 96-well output plate station
12. 48-well archive plate station

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.

2 Reagents, supplies, and required materials

- Genexus™ Dx FFPE DNA/RNA Purification Combo Kit 13
- Required materials not supplied for use with the Genexus™ Dx Purification System 14

Genexus™ Dx Purification System reagents and supplies can be ordered as kits but most 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 FFPE DNA/RNA Purification Combo Kit

The Ion Torrent™ Genexus™ Dx FFPE DNA/RNA Purification Combo 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	

(continued)

Component	Amount	Storage
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	
Genexus™ Dx AutoLys Tubes	2 × 24 each	
Genexus™ Dx AutoLys Caps	2 × 24 each	

[1] Store the Genexus™ Dx Quantitation Plate in the dark to help prevent photobleaching of the preloaded reagents.
[2] Can be stored at 15°C to 30°C on receipt.

Required materials not supplied for use with the Genexus™ Dx Purification System

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.

Item	Source
General laboratory equipment and supplies	
Bench top microcentrifuge [1]	**MLS**
1,000 µL multichannel pipette	**MLS**
20-, 200-, and 1,000-µL pipettes and appropriate filtered tips[2]	**MLS**
Aerosol-resistant pipette tips	**MLS**
Microcentrifuge tubes, 1.5-mL or 1.7-mL (low retention for nucleic acids)	**MLS**
1.5-mL Eppendorf™ LoBind™ tube (for long term storage of archived samples)	13-698-791 (Eppendorf™ 022431021) or **MLS**
Vortex mixer with a rubber platform	**MLS**
Gloves, powder-free nitrile	**MLS**
Isopropanol, 100% and 70% solution	**MLS**
Wipes, disposable lint-free	**MLS**
Uninterruptible Power Supply (UPS) [3]	**MLS**

(continued)

Item	Source
For use with Genexus™ Dx FFPE DNA/RNA Purification Combo Kit	
Sorvall™ ST 8 Small Benchtop Centrifuge (or equivalent)[4]	75007200 (USA) 15135233 (Europe) **eu.fishersci.com**
Thermo Scientific™ M10 Microplate Swinging Bucket Rotor (or equivalent)[5]	75005706 (USA) 15105283 (Europe) **eu.fishersci.com**
Sealed Bucket; Capacity: 4 Standard or 2 Midi-Deepwell plates (Set of 2) (or equivalent)	75005721 (USA) 15241026 (Europe) **eu.fishersci.com**
Economy Standard Incubator (2, 60°C and 90°C) (or equivalent)	S50441A (USA) **fisherscientific.com** 15652086 (Europe) **eu.fishersci.com**
Equipment and consumables for AutoLys Tube FFPE sample extraction[6]	
Genexus™ Dx AutoLys Tube Lifter (optional)	A58872
Integra™ Miltex™ Sterile Standard Scalpel; Mfr. No. 4411, flat blade for scraping FFPE slides	12-460-452 or **MLS**
Integra™ Miltex™ Sterile Standard Scalpel; Mfr. No. 4410, rounded blade for tissue macrodissection, if needed	12-460-451 or **MLS**
Fisherbrand™ High Precision Straight Medium Point General Purpose Tweezers/Forceps, or equivalent, for handling FFPE curls	12-000-121
Tubes, plates, and other consumables	
Reagent reservoir, 50–100 mL, for use with multichannel pipettor	**MLS**
DNA*Zap*™ PCR DNA Degradation Solutions	AM9890
RNA*later*™ Stabilization Solution	AM7020
RNase*Zap*™ RNase Decontamination Solution	AM9780
RNase *AWAY*™ Surface Decontaminant	7002PK
Nuclease-Free Water (not DEPC-Treated)	AM9930 or **MLS**

(continued)

Item	Source
TE Buffer, 1X Solution pH 8.0, Low EDTA	J75793.AP or **MLS**
Ethanol, 100%	**MLS**

[1] Must fit standard 0.2- and 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 recommend use of positive displacement pipettes and RNase free tips for use when isolating RNA.

[3] To protect against power outages and line voltage fluctuations, we require that you use a double-conversion 1.5-kVA (or higher) uninterruptible power supply with a power rating of 800W or higher. Additional battery pack capacity may be required depending on how much holdup time is required. Power failures and other events that abruptly terminate the function of the instrument can possibly damage the system. These recommendations are for 1 single system.

[4] Centrifuge must achieve a centrifugal force of 2000 × *g*, have a swinging bucket rotor and accommodate deepwell plates and AutoLys Tube Rack (minimum depth 6.5 cm).

[5] Swinging bucket rotor must carry deepwell plates in the landscape orientation (see page 32).

[6] For use with the Genexus™ Dx FFPE DNA/RNA Purification Combo Kit.

3 Before you begin

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 curl samples with AutoLys Tubes” on page 34 or “Prepare FFPE slide samples with AutoLys Tubes” on page 36.

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 6 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 5 months.

Guidelines for FFPE samples

- 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, macrodissection may be required to enrich the tumor content if the tumor content is below the acceptable level recommended by the downstream application or assay. Following tumor enrichment, proceed with the extraction protocol.
- 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.
- Necrotic samples: Highly necrotic samples do not appear to interfere with extraction. However, we recommend that you macrodissect to remove highly necrotic areas or select alternate samples if possible.
- 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.
- We recommend that you extract sufficient nucleic acid from samples to enable repeat analyses, if possible.

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.

Equipment and instrument cleaning

- We recommend situating the instrument in a dedicated sample preparation (pre-PCR) laboratory space to decrease the possibility of amplicon contamination.
- If you are preparing samples and sample plates in a hood (recommended), illuminate the hood with UV light for 15 minutes before use.
- Before preparing for a purification run, decontaminate surfaces of the hood or bench where samples or sample plates are handled, and other equipment such as vortexers, microcentrifuges, and pipettors. 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 bench and equipment surfaces with wipes moistened with 70% isopropanol or 70% ethanol, or with prepared alcohol wipes.

- Before and after a run, sequentially wipe the deck surface of the Genexus™ Dx Purification System with lint-free wipes moistened with the two DNA*Zap*™ decontamination solutions followed by wipes moistened with nuclease-free water. Follow with wiping of deck surfaces with 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.

  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.

Guidelines for Genexus™ Dx Purification System operation

- Follow guidance that is provided by the software in the instrument touchscreen when you create a run plan to determine which consumables must be loaded for each run.
- Follow guidance that is provided by the software in the instrument touchscreen 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 Genexus™ Dx Purification System 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.
- Ensure that the 48-Well Nucleic Acid Archive Plate is seated properly. If the plate is curved or bent, gently bend the plate in the direction opposite to the curvature to straighten the plate before loading.
- If the instrument determines there is a problem (for example, stuck pipet tip or obstructed movement of the gantry arm) the run is aborted and you must restart the run with new consumables.

Power the Genexus™ Dx Purification System On or Off

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.

- Power the instrument on.

  If the touchscreen is unresponsive, check the power switch on the back of the instrument to ensure that the switch is in the on (|) position. If the power switch is in the off (O) position, proceed with substep 1a. If the power switch is already in the on position, proceed to substep 1b.

  a. Turn the power switch on the back of the instrument to the on (|) position.

  b. Press the power button on the front of the instrument.
  The button illuminates.

c. In the **Sign In** screen, enter the username and password created at installation.
When the instrument home screen appears, the instrument is ready for use.

- Power off the instrument.

Note: It is not necessary to power off the instrument overnight or over the weekend. If the instrument will not be used for more than 3 days, power off the instrument as follows:

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.

Reboot the instrument

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.

1. In the **Home** screen, tap **Settings** ▸ **System Tools** ▸ **Shut down**.
2. Select **Reboot**.
A confirmation message appears. Select **Yes** to reboot the instrument.

Software compatibility and requirements

The procedures in this guide are designed for use with Genexus™ Dx Software version 6.8.2 or later. The operating system of the Genexus™ Dx Purification System is Ubuntu™ 18.04 LTS. The operating system has an Ubuntu™ Pro license that includes expanded security maintenance coverage.

Network configuration and security

The network configuration and security settings of your laboratory or facility (such as firewalls, anti-virus software, network passwords) are the sole responsibility of your facility administrator, IT, and security personnel. This product does not provide any network or security configuration files, utilities, or instructions. The Genexus™ Dx Purification System has a firewall enabled by default. Only ports required for the operation of the instrument and software are open.

If external or network drives are connected to the software, it is the responsibility of your IT personnel to ensure that such drives are configured and secured correctly to prevent data corruption or loss. It is the responsibility of your facility administrator, IT, and security personnel to prevent the use of any unsecured ports (such as USB, Ethernet) and ensure that the system security is maintained.

Password security

Thermo Fisher Scientific strongly recommends that you maintain unique passwords for all the operator and admin accounts in use on this instrument. All passwords should be reset on first sign in to the instrument.

Type a new password, confirm the password, then click Change. Passwords are case-sensitive and must the following formatting critera.

- Contain between 8 and 30 characters
- Only alphanumeric characters (numbers 0 to 9 and letters A to Z), no spaces or special characters
- At least one lower case and one upper case alphabetic character (a–z, A–Z)
- At least one numeric character (0–9)

Change passwords according to the password policy for your organization. For information on how to manage existing passwords, see “User management” on page 73.

Data backup and data storage

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.

Note: A paid data migration service is available if needed. Contact your Thermo Fisher Scientific representative for more information.

For more information about data storage compatibility, contact your local support team.

Antivirus software

For recommendations for antivirus software that is compatible with Genexus™ Dx Software 6.8.2, see the current version of the Genexus™ Dx Software release notes.

Switch user accounts

The Genexus™ Dx Purification System has three user accounts: Operator, Administrator, and Service. Additional accounts cannot be created. To switch between user accounts:

1. Sign out from the user account in use. Tap **(User account profile)**, then tap **Sign out**.
2. Enter the credentials for the desired user account (for example, Operator + password), then tap **Sign in**.

User access levels on the Genexus™ Dx Purification System

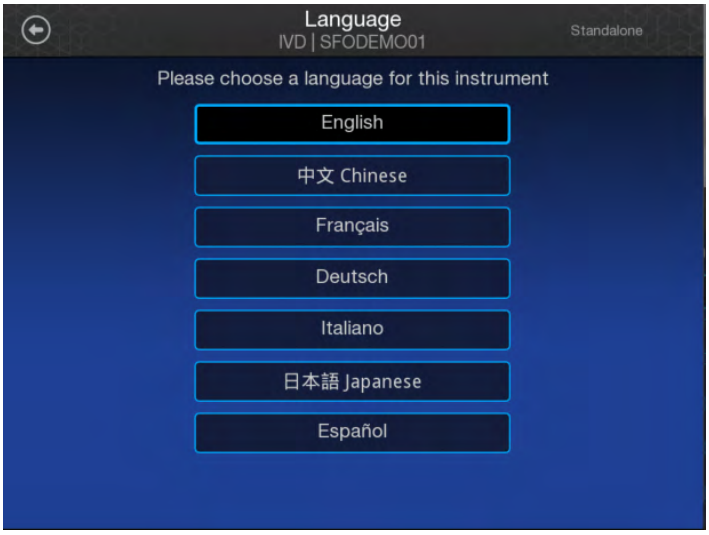
User level	Allowed actions
Operator	• Plan, save, and delete runs • Perform purification runs • View results • Access system tools
Administrator	Operator functions, plus: • Manage instrument settings • Manage network settings • Manage software updates
Service	Access service tools

Switch the software language

You can switch the language of the software user interface on the Genexus™ Dx Purification System:

- English
- Français
- Deutsch
- Italiano
- Español
- Chinese
- Japanese

1. In the Home screen, tap **Settings** ▸ **Instrument settings** ▸ **Language**.
2. In the **Language** screen, select the desired language, then tap **OK**.

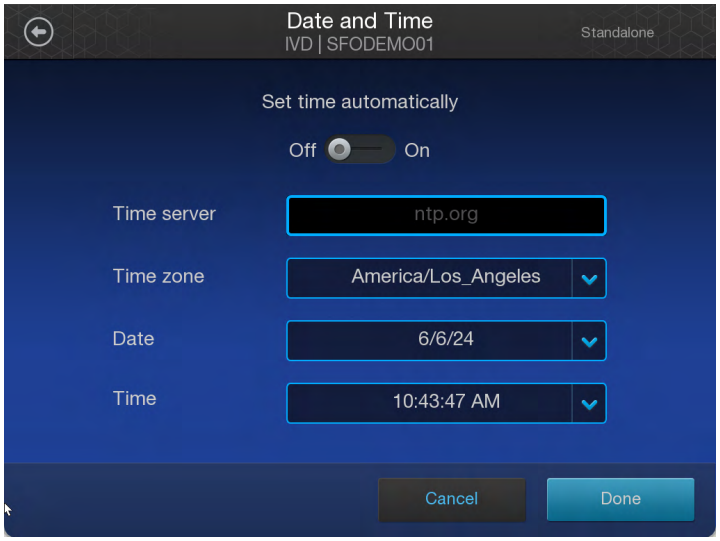


The software will restart in the selected language.

3. After the instrument reboot is complete. Enter your **User name** and **Password**, then tap **Sign in**.

Set the date and time

1. In the **Home** screen, tap **Settings** ▸ **Instrument Settings** ▸ **Date and time**.
2. Next to the **Date** field, tap ⌄, then select the current date.
3. Next to the **Time** field, tap ⌄, then enter the current time.
4. Tap **Done**.



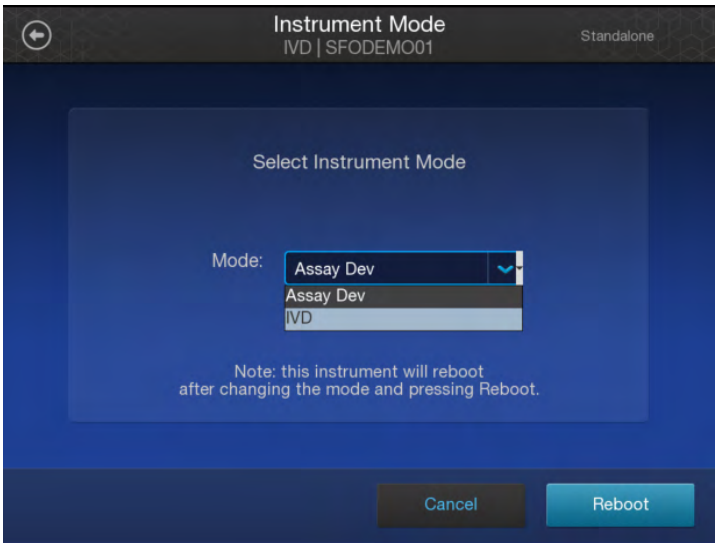
IMPORTANT! Do not leave the **Time server** field blank. This will cause the instrument to stop responding and require a reboot to restore functionality.

Switch the software mode

You can switch between two separate software modes controlling the Genexus™ Dx Purification System:

- Assay Development mode
- IVD mode

1. In the Home screen, tap **Settings** ▸ **System tools** ▸ **Instrument mode**.
2. In the **Instrument Mode** screen, tap ⌄, then select the mode that you want to use.



3. Tap **Reboot**, then tap **Reboot** again to confirm.
4. After the instrument reboot is complete. Enter your **User name** and **Password**, then tap **Sign in**.

View instrument PQ status

Before using the Genexus™ Dx Purification System in IVD mode, the instrument must first pass perfomance qualification (PQ). Performance qualification is performed by a service representative during installation. The PQ status and the time remaining before it must be renewed is available in the **About This Instrument** screen.

Note: Performance qualification of the Genexus™ Dx Purification System is required to use the instrument in IVD mode, but not required to use the instrument in Assay Development mode.

1. In the **Home** screen, tap **Settings** ▸ **Instrument Settings** ▸ **About**.

   Note: You need to be signed in to the Administrator user account to view the PQ status.

   The **About This Instrument** screen displays the **PQ State** (PQ status of the instrument). This screen also displays the expiration date of the current PQ, and the number of days until the PQ expires.

2. To update the PQ status, contact your local service representative.

Precautions

Avoid nucleic acid contamination

IMPORTANT! DNA fragments from sample processing are a common source of contamination. Do not introduce amplified DNA into the work area where the instrument is located.

- Use good laboratory practices to minimize cross-contamination of products and reagents.
- When designing the laboratory layout, dedicate separate areas for purification and sequencing activities. Dedicate laboratory supplies and/or equipment to the appropriate area.

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.

Avoid instrument vibration

IMPORTANT! The Genexus™ Dx Purification System must be installed on a bench that is free from vibrations. The bench should not be in contact with equipment that can cause vibrations to the bench, such as freezers, pumps, large benchtop centrifuges, and other similar equipment. An air table is not required, and securing the instrument to the bench is not required.

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).

4 Genexus™ Dx FFPE DNA and RNA Purification protocol

- Workflow 27
- Plan a Genexus™ Dx FFPE DNA and RNA Purification run 28
- Prepare samples 32
- Load the instrument and start the run 39
- Unload purified DNA and RNA 49

Workflow

Genexus™ Dx FFPE DNA and RNA Purification workflow

5 min

Plan a purification run (page 28)

Add a new purification run plan or copy-edit an existing purification run plan that best represents your experiment. Purification run plans include instrument settings that are used in sample purification.



3 hr

Prepare samples from FFPE curls (page 34)

OR

Prepare samples from FFPE slides (page 36)

Samples are deparaffinized and digested with protease in preparation for isolation of DNA and RNA.



5 min

Load the Genexus™ Dx Purification System (page 39)

The purification run plan is selected and the run initiated. The instrument performs a pre-run UV clean, then reagents and consumables are loaded on to the instrument.



2 hr

Start the run (page 48)

After the sample plate and all reagents and consumables have been loaded the instrument door is closed and the run started.

Note: Sample quantification adds ~2 hours to the run time.



5 min

Unload the purified nucleic acids (page 49)

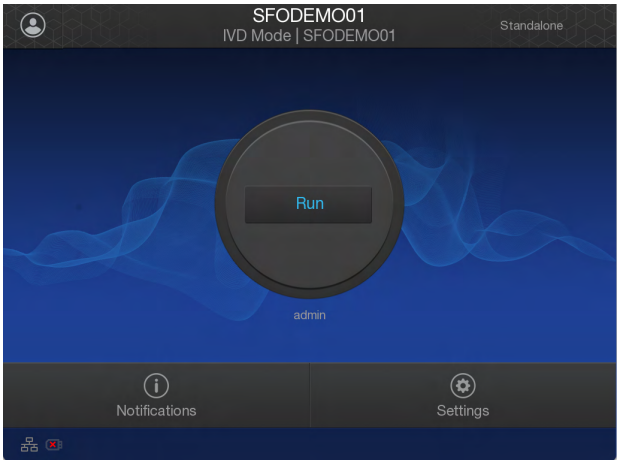
Remove and seal the 48-Well Nucleic Acid Archive Plate or proceed immediately to sequencing of the purified sample. Used reagents and consumables are removed from the instrument and a UV clean is performed.

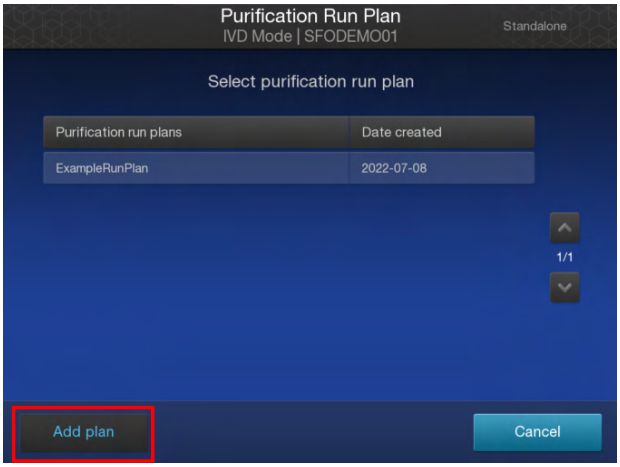


Plan a Genexus™ Dx FFPE DNA and RNA Purification run

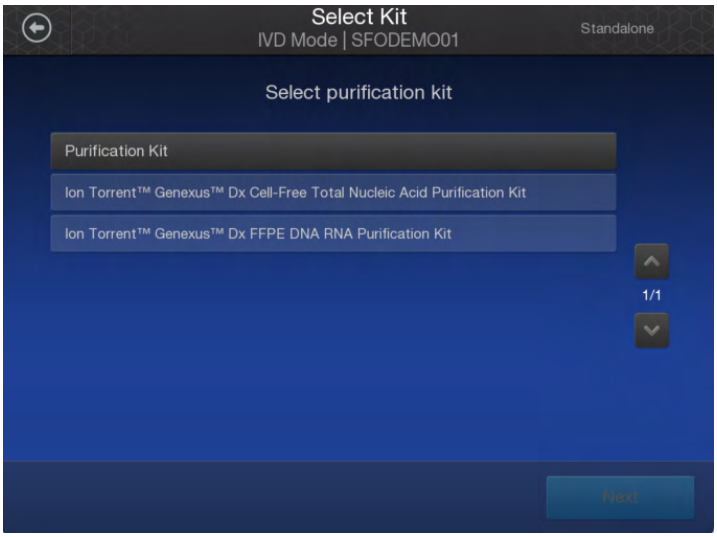
Users plan a purification run through the instrument touchscreen. Plan the run before you prepare samples and load the samples into the Genexus™ Dx FFPE DNA and RNA Purification Plate 1.

1. Enter your username and password to sign in to the instrument.
2. Tap **Run**, then tap **Add plan**.

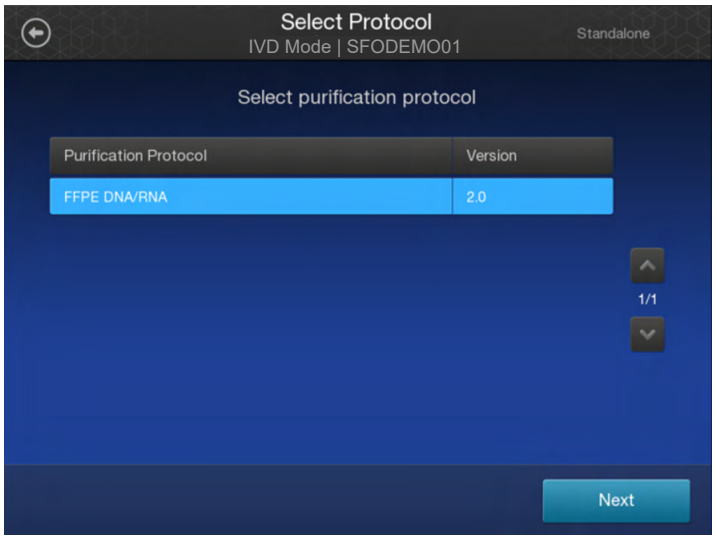




3. Tap in the entry box, enter a unique name for the run plan, then tap **Done** ▸ **Next**.
4. Select the Ion Torrent™ Genexus™ Dx FFPE DNA and RNA Purification Kit, then tap **Next**.



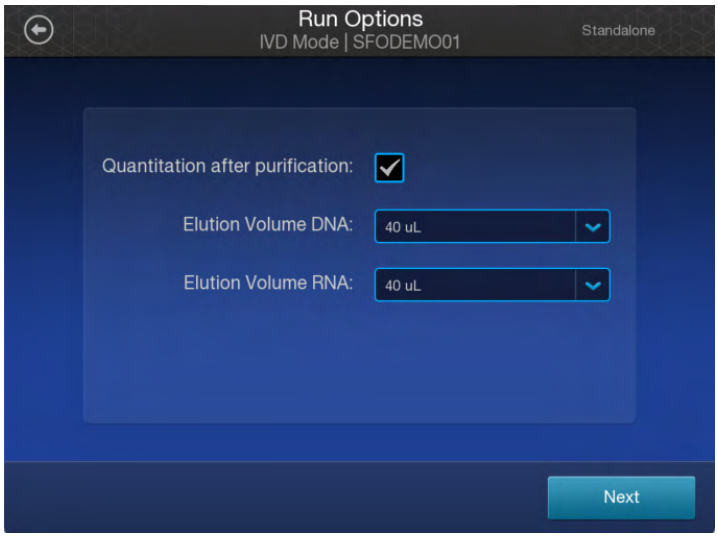
5. Select the **FFPE DNA/RNA** purification protocol, then tap **Next**.



6. Enable or disable **Quantitation after Purification**.

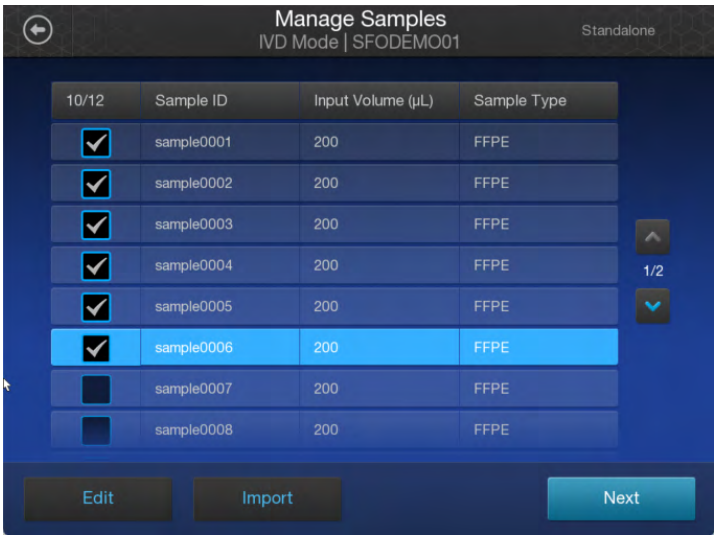
Note:

- The Quantitation Plate is required even if **Quantitation after Purification** is disabled.
- Disabling **Quantitation after Purification** may reduce the purification run time by up to 2.5 hours.
- Quantitation requires up to 5 µL of the eluted sample. If the expected sample yield is limiting, manual sample quantitation using less volume may be preferred to preserve sample.

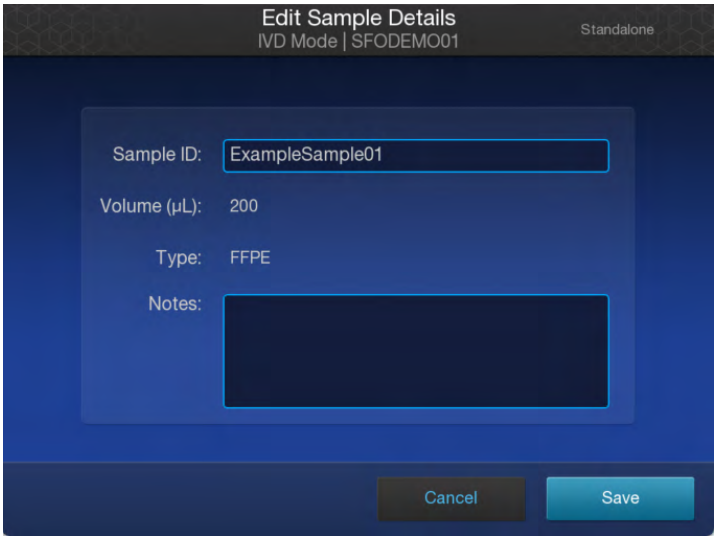


7. Accept the default elution volume. If needed, select the desired elution volume from the dropdown list, then tap **Next**.

8. *(Optional)* Change the number of samples and the sample details.
   a. In the **Manage Samples** screen, deselect extra samples from the end of the range (for example, if you are only running ten 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 sample information.

   The import overwrites the existing **Sample ID** and **Notes** information for each sample. That is, if the run plan has 6 samples, the sample import file must include information for at least 6 samples.

To import sample information, prepare a CSV sample import file and save it to a USB drive. See “Create a template for importing samples in standalone configuration purification run plans” on page 59.

a. In the **Manage Samples** screen, tap **Import**.

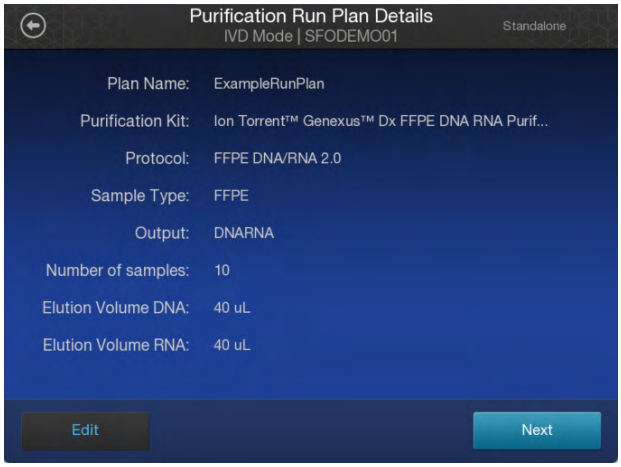
b. In the **Sample Import** screen, tap **Import** to proceed.

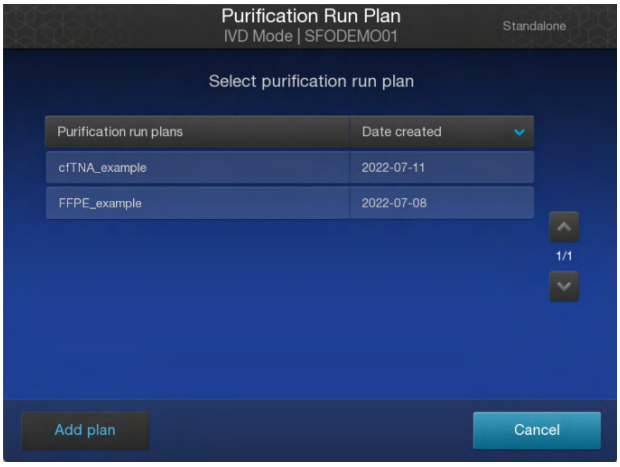
c. Insert the USB drive that contains the sample import CSV file into the USB port on the front of the purification instrument. In the **Sample Import** screen, select the USB drive, then navigate to and select the sample import file.

d. *(Optional)* Tap **Details** to view the CSV file that lists 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. If needed, select a sample, then tap **Edit** to modify the **Sample ID** or **Notes**.

10. Review the **Purification Run Plan Details**. Tap **Edit** to change any of your selections, otherwise tap **Next**.





The new purification run plan appears in the list of available **Purification run plans**.

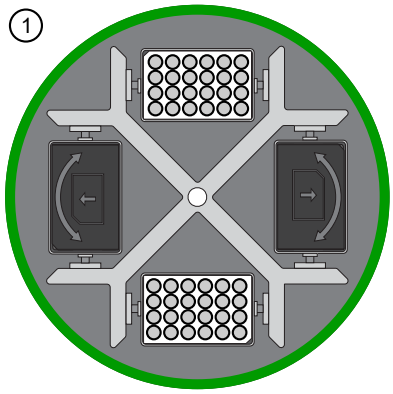
To delete an existing run plan, see “Delete a run plan” on page 59.

Prepare samples

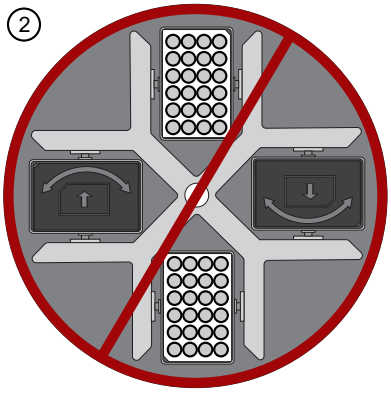
Procedural guidelines

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 –65°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 Tube Rack in "landscape" orientation.



① Landscape orientation—recommended



② Portrait orientation—not recommended

Note: Place AutoLys 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 48-Well Nucleic Acid Archive Plate with purified nucleic acids from the instrument within 1 hour of run completion and 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

- We recommend the use of incubators when using AutoLys Tubes.
- Preheat incubators to 60°C and 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 FFPE DNA and RNA Purification Plate 1
  - Genexus™ Dx Proteinase K (red cap)
  - Genexus™ Dx FFPE Protease Buffer
- Genexus™ Dx Purification Supplies 2 (Part. No. A51081)
  - AutoLys Tubes and Caps
- AutoLys Tube Lifter or AutoLys Pliers
- AutoLys Tube Rack
- Plate centrifuge
- Incubators (see “Required materials not supplied for use with the Genexus™ Dx Purification System” on page 14 for a list of recommended incubators)

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 samples.

Component	Volume per reaction
Genexus™ Dx FFPE Protease Buffer	(n + 1) × 225 µL
Genexus™ Dx Proteinase K (red cap)	(n + 1) × 10 µL
Total volume	**(n + 1) × 235 µL**

3. Vortex for ~5 seconds to mix, then briefly centrifuge to collect the contents.

Prepare FFPE curl samples with AutoLys Tubes

Digest with protease

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 Tube for each FFPE tissue sample.

2. Transfer each FFPE section curl from its storage tube to a separate labeled AutoLys Tube.
   a. Invert a labeled AutoLys 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 Tube.

   If the curl adheres to the storage tube, gently grasp an edge of the curl with forceps, then transfer it to the AutoLys 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 Tubes, place the tubes in an AutoLys 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 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. Securely retighten each tube cap to limit evaporation, then 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 39.
   - Equilibrate the Quantitation Plate to room temperature during the incubation.

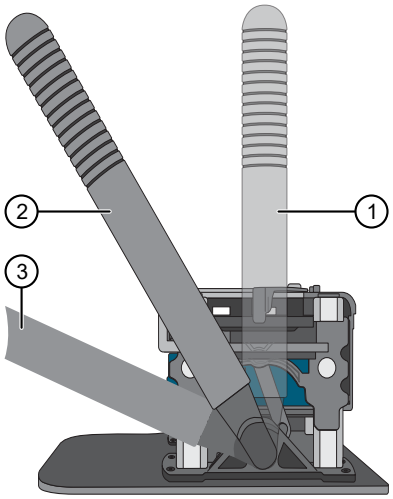
7. Allow samples to cool to room temperature for 5–10 minutes before proceeding to lift the tubes.

8. Lift the tubes. The following steps describe use of the AutoLys Tube Lifter to process up to 24 samples simultaneously. Alternatively, AutoLys Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys Tube Lifter lever is in Position A (straight up) and the slider is in Position 1.

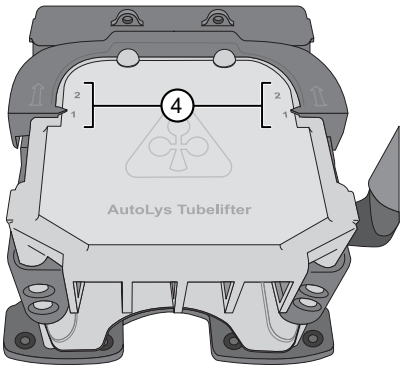
   b. Slide the 24-well AutoLys Tube Rack containing the lysed samples into the AutoLys Tube Lifter.

   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 Tube Lifter see the *AutoLys M TubeLifter User Guide* (Pub. No. MAN0017676).



① AutoLys Tube Lifter lever Position A
② AutoLys Tube Lifter lever Position B
③ AutoLys Tube Lifter lever Position C



④ AutoLys Tube Lifter slider positions 1 & 2

9. Slide the AutoLys 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 Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge. See page 32.
   - Ensure that AutoLys Tube caps are securely held by the AutoLys Locking Lid before you start the centrifugation.

10. Using the AutoLys Tube Lifter or AutoLys Pliers, separate the filter from the outer tube.
    a. Adjust the position of the AutoLys Tube Lifter slider to Position 2.
    b. Remove the AutoLys Locking Lid, then slide the rack into the AutoLys Tube Lifter.
    c. Press the lever down from Position B to Position C.

    Keep the samples on ice or at 4°C.

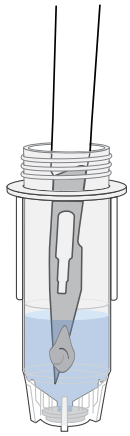
Proceed to "Load the instrument and start the run" on page 39.

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 samples with AutoLys Tubes

Collect the tissue

1. Label an AutoLys Tube for each FFPE tissue sample.
   Label each tube (cap and side) with its Sample ID using a marker that is resistant to 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 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 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.

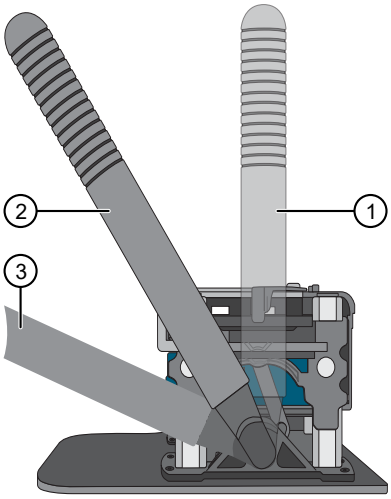
2. Securely retighten each tube cap to limit evaporation, then incubate at 90°C for 60 minutes.
   - Prepare the reagents and consumables that are needed by the instrument during the incubation. See “Prepare the consumables” on page 39.
   - Equilibrate the Genexus™ Dx Quantitation Plate to room temperature during the incubation.

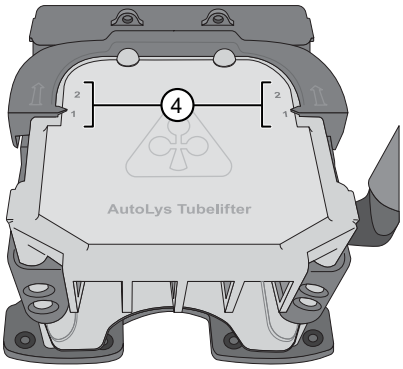
3. Allow samples to cool to room temperature for at least 10 minutes before proceeding to lift the tubes.

4. Lift the tubes. The following steps describe use of the AutoLys Tube Lifter to process up to 24 samples simultaneously. Alternatively, AutoLys Pliers can be used to process tubes individually.
   a. Ensure that the AutoLys Tube Lifter lever is in Position A (straight up) and the slider is in Position 1.

   b. Slide the 24-well AutoLys Tube Rack that contains the lysed samples into the AutoLys Tube Lifter.

c. Press the lever down from Position A to Position B, then remove the rack from the lifter.





① AutoLys Tube Lifter lever Position A
② AutoLys Tube Lifter lever Position B
③ AutoLys Tube Lifter lever Position C

④ AutoLys Tube Lifter slider positions 1 & 2

5. Slide the AutoLys 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 Locking Lid points away from the center of rotation (landscape orientation) when placed in the centrifuge (see page 32).
- Ensure that AutoLys Tube caps are securely held by the AutoLys Locking Lid before you start the centrifugation.

6. Using the AutoLys Tube Lifter or AutoLys Pliers, separate the filter from the outer tube.
   a. Adjust the position of the AutoLys Tube Lifter slider to Position 2.
   b. Remove the AutoLys Locking Lid, then slide the rack into the AutoLys Tube Lifter.
   c. Press the lever down from Position B to Position C.

Keep the samples on ice or at 4°C.

Proceed to “Load the instrument and start the run” on page 39.

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)
  - 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
  - 48-Well Nucleic Acid Archive Plate
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal
- 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
  - 48-Well Nucleic Acid Archive Plate
  - Genexus™ Dx 48-Well Nucleic Acid Archive Plate Seal
- 12-Well Tip Comb

Equilibrate the Quantitation Plate

IMPORTANT! Allow at least 30 minutes for the Genexus™ Dx Quantitation Plate to equilibrate to room temperature.

The Genexus™ Dx Quantitation Plate is required even if your run plan does not include sample quantitation.

The Genexus™ Dx Quantitation Plate can be equilibrated to room temperature during the protease digestion to save time.

Centrifuge the Genexus™ Dx Quantitation Plate at 1,000 × *g* for 30 seconds to collect the contents.

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.

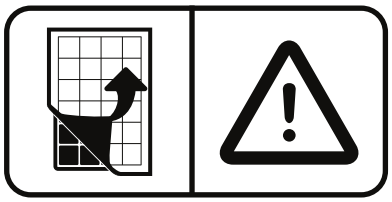
2. In a 1.5-mL low-retention microcentrifuge tube, prepare a 1X DNase Digestion Master Mix as indicated, where n is the number of samples.

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.

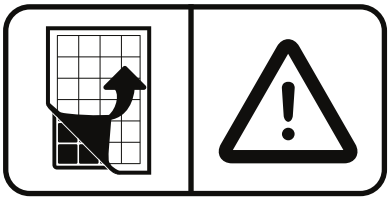
Note: You can add the samples to the Genexus™ Dx FFPE DNA and RNA Purification Plate 1 and load the plate as the final steps of loading the instrument. This can be done to help ensure that other components are successfully loaded and accepted by the instrument before adding samples of possibly limited 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.

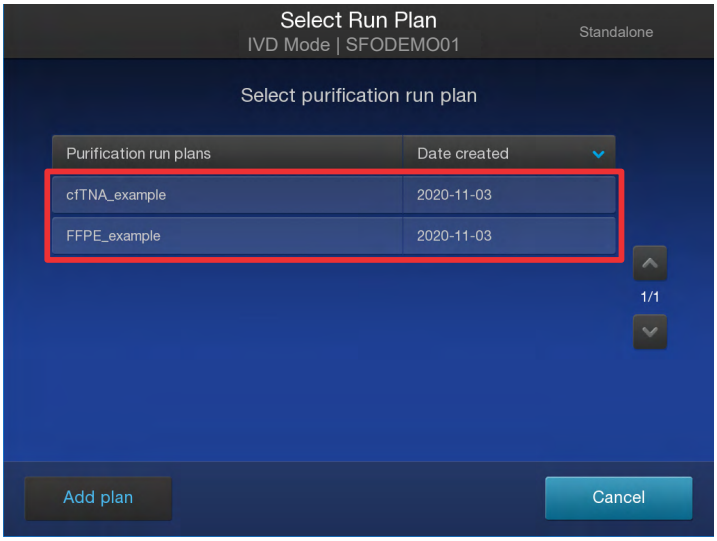
   A precipitate can form, but this does not interfere with the DNA binding. Proceed directly to the next step.

   Reagent consumables cannot be reused.

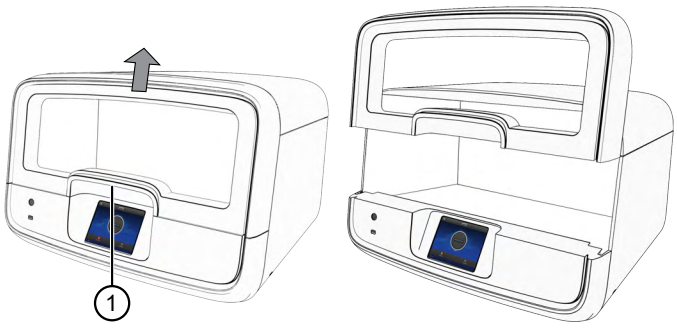
   IMPORTANT! Load the plate in the instrument (see page 44) within 15 minutes of adding samples to the plate.

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. Lift the instrument door to the stop.



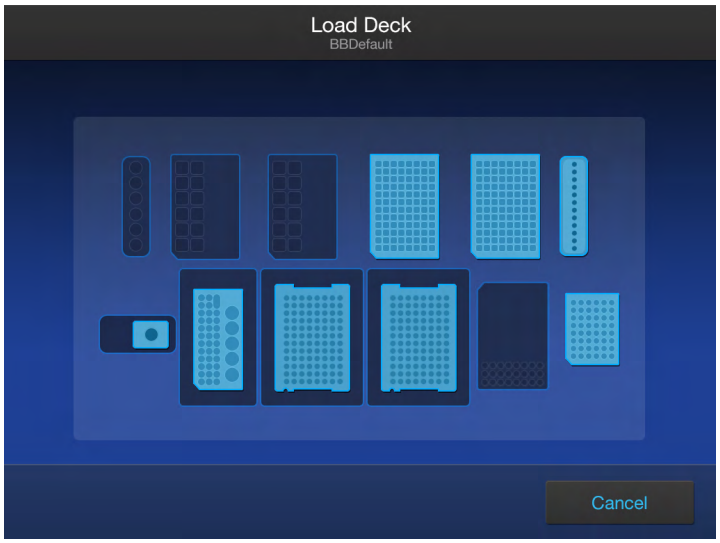
① Hold here, then lift.

Load the Genexus™ Dx Purification System

IMPORTANT!

- Do NOT load any consumables onto the instrument until after the instrument has performed the prerun UV cleaning.
- Ensure that all components are clean and dry before loading them onto the instrument.
- 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.

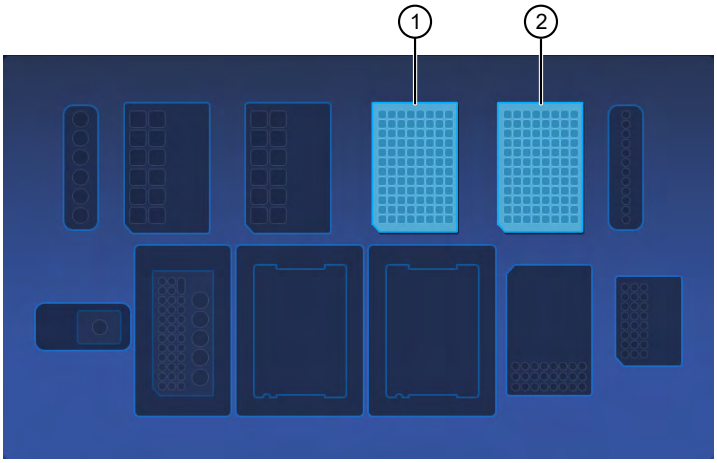


Screen display for an instrument fully loaded for an FFPE sample purification run.

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 41.

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 40.



① Genexus™ Dx FFPE DNA and RNA Purification Plate 1 position
② Genexus™ Dx FFPE DNA and RNA Purification Plate 2 position

Load the 12-Well Tip Comb, Genexus™ Dx Purification Tip Cartridges, and 48-Well Nucleic Acid Archive Plate

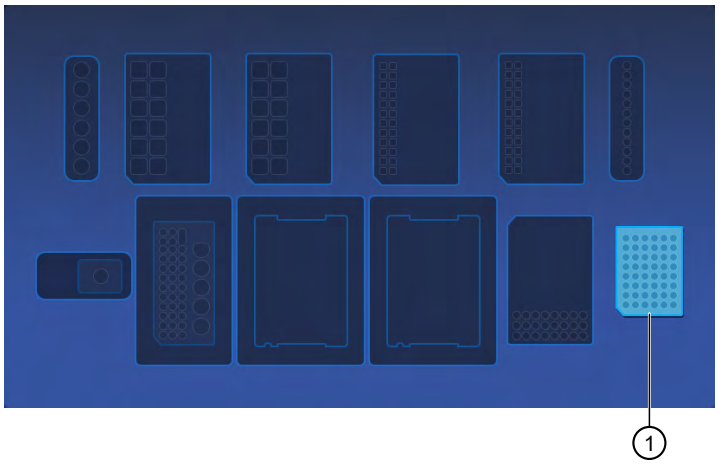
1. Unwrap, then load a new 12-Well Tip Comb.

   Ensure that the 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 12-Well Tip Comb position

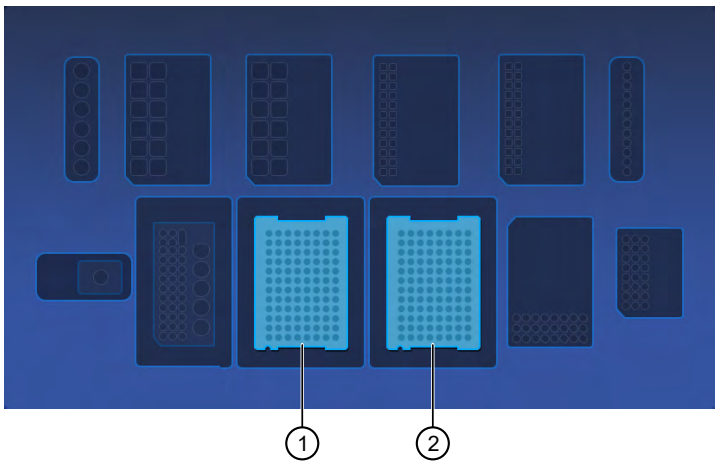
2. Unwrap, then load a new 48-Well Nucleic Acid Archive Plate.



① 48-Well Nucleic Acid Archive Plate position

Note: Ensure that the 48-Well Nucleic Acid Archive Plate is seated properly. If the plate is curved or bent, gently bend the plate in the direction opposite to the curvature to straighten the plate before loading.

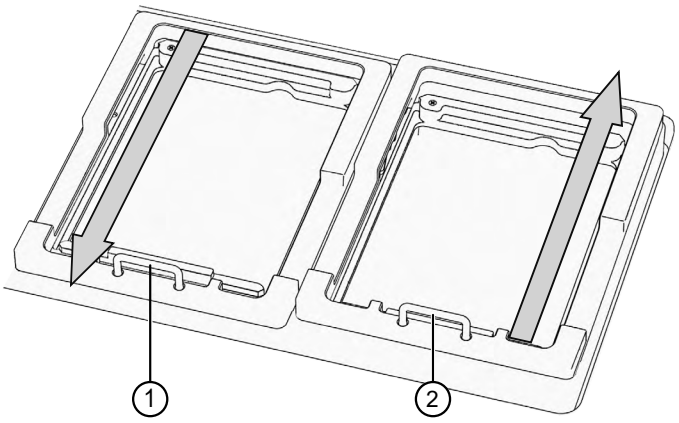
3. Unwrap two new Genexus™ Dx Purification Tip Cartridges, remove the cover to expose the pipette tips, then load the cartridges in positions 1 and 2.



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.

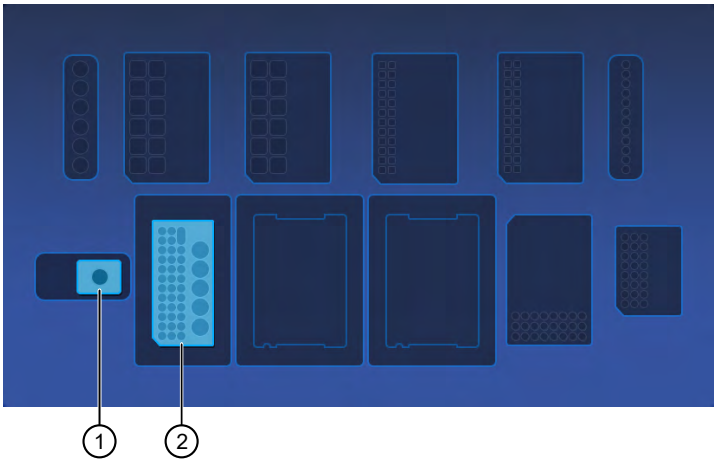


① Unlocked (forward) ② Locked (back)

Load the quantitation reagents and consumables

Note: Protect the Genexus™ Dx Quantitation Plate from direct bright light to prevent photobleaching of the preloaded reagents.

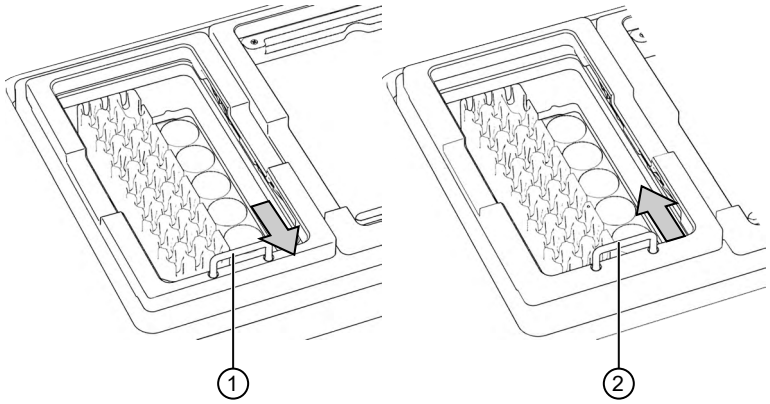
1. Centrifuge the Genexus™ Dx Quantitation Plate at 1,000 × *g* for 30 seconds to collect the contents.
2. Load the Genexus™ Dx Quantitation Plate in position 2.



① Genexus™ Dx Quantitation Tube position
② Genexus™ Dx Quantitation Plate position

a. Pull the locking mechanism handle forward, then place the Genexus™ Dx Quantitation Plate in the open position.

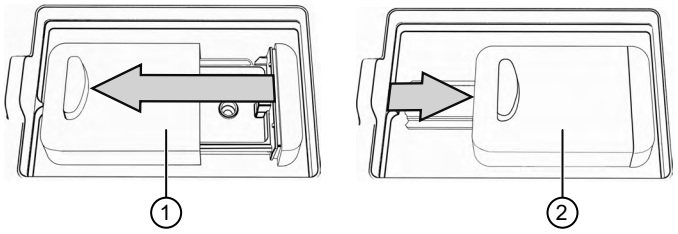
b. Push the locking mechanism handle back to lock the plate in place.



① Unlocked (forward) ② Locked (back)

3. Slide and hold the quantitation module cover to the left, then insert the Quantitation Tube. **Touching only the top of the 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.

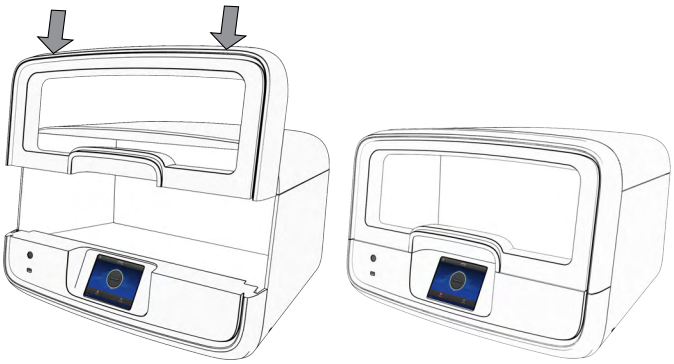
- Foil seals are removed from the purification plates. Do not remove foil seal from the Genexus™ Dx Quantitation Plate.
- Each component is at the correct location and in the correct orientation. Press down on all plates and cartridges to ensure that they are firmly seated in place.
- The 12-Well Tip 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 52.

Unload purified DNA and RNA

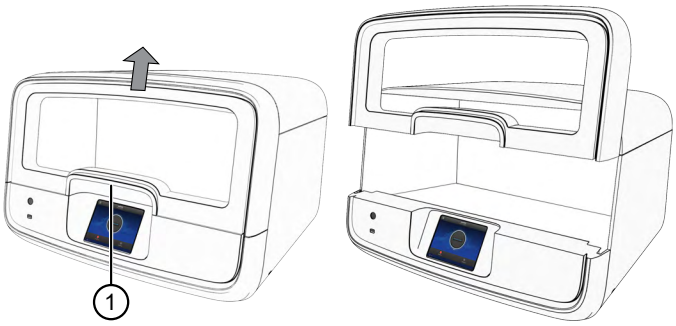
IMPORTANT! Do not allow purified nucleic acids to sit on the instrument. Store as directed or use in downstream applications within 60 minutes.

1. In the touchscreen, tap **Unload**.



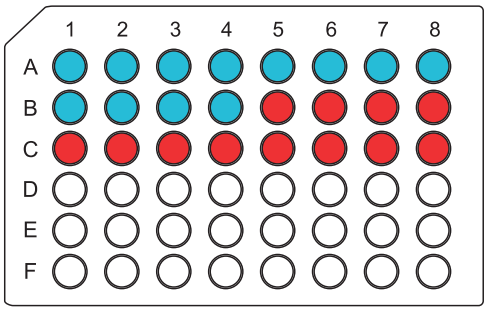
The door unlocks.

2. Lift the instrument door to access the instrument deck.



① Hold here, then lift.

3. Remove the 48-Well Nucleic Acid Archive Plate that contains the purified sample DNA and RNA.



- DNA is in wells A1–B4. DNA samples always start in well A1.
- RNA is in wells B5–C8. RNA samples always start in well B5 regardless of how many samples are purified.

4. For short-term storage, seal the plate with a 48-Well Nucleic Acid Archive Plate Seal. Store the plate at –20°C for up to 3 months. For long-term storage (>3 months), transfer samples to labeled low-retention tubes, then store the DNA samples at –30°C to –10°C and the RNA samples at –90°C to –65°C for up to 12 months.

   If the archive plate is thawed during short-term storage, transfer the DNA and RNA into labeled low-retention tubes. Do not reseal the archive plate with the used plate seal.

Dispose of used consumables and UV clean the instrument

Unload purified nucleic acid samples before disposal of used consumables.

IMPORTANT! Do not allow purified nucleic acids to sit on the instrument. Store as directed or use in sequencing reactions within 60 minutes.

1. 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 1a through substep 1c to discard the Genexus™ Dx FFPE DNA and RNA Purification Plate 2.
2. Unlock, then remove and dispose of the Purification Tip Cartridges in an appropriate waste container.

3. Unlock, then remove and dispose of the 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.

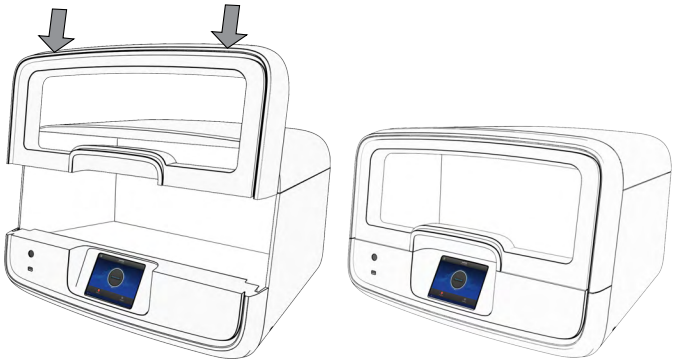


WARNING! No data are currently available that address the mutagenicity or toxicity of the Qubit™ RNA BR Reagent (Component A). This reagent is known to bind nucleic acid and is provided as a solution in DMSO. Treat the Qubit™ RNA BR Reagent with the same safety precautions as all other potential mutagens and dispose of the dye in accordance with local regulations.

   b. Dispose of the deep-well plate in an appropriate waste container.

4. Open the quantitation module cover, remove and discard the Quantitation Tube, then allow the module cover to close.

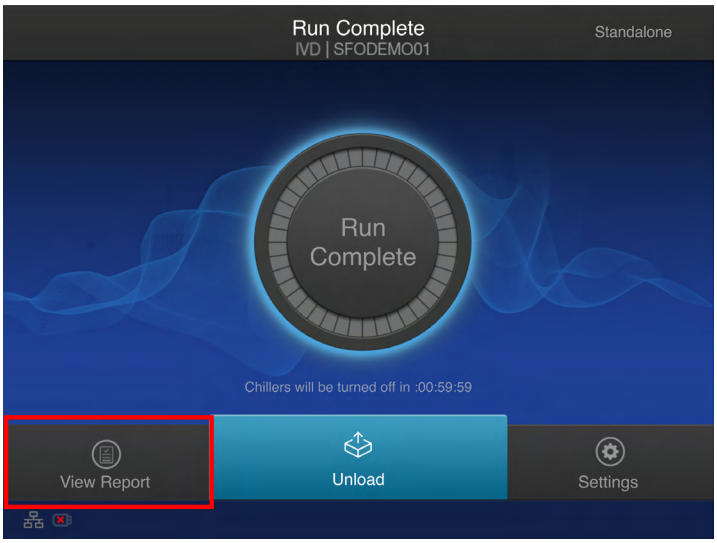
5. Close and lock the instrument door by pressing down on both top corners, then tap **Start UV Clean**.



The time remaining in the UV cleaning is displayed. When complete, the instrument is ready to start a new purification run.

View and export quantitation results

1. In the **Run Complete** screen, tap **View report**.

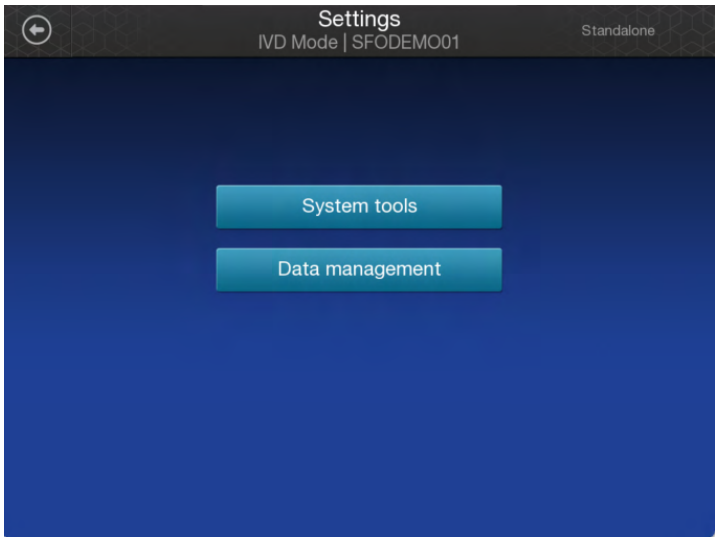


   The **Saved Experiment Reports** screen opens. See step 4.

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. Results displayed in the **Concentration** column are in ng/µL.



6. Insert a USB drive into the USB port on the front of the instrument, then tap **Export CSV**. Navigate to the file destination, then tap **Save**.

Troubleshooting

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. 2. Export the Genexus™ Dx Purification System CSA file from the Run Report. 3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting. 4. Restart the run beginning from “Prepare samples”. If the run fails again, contact Technical Support to troubleshoot the problem.
	Power failure occurred during sample purification	**IMPORTANT!** If the power failure occurred during the purification process the samples and reagents cannot be recovered or reused. 1. Record the error message displayed on the instrument touchscreen. 2. Export the Genexus™ Dx Purification System CSA file from the Run Report. 3. Remove the consumables from the deck, then clean the instrument. If possible, retain the consumables for troubleshooting. 4. Restart the run beginning from “Prepare samples”.
Unexpected sample concentration (negative or greater than normal concentration values for multiple samples)	• Erroneous standard measurements were made. • Pipetting error occurred during liquid transfers. • Tip blockage occurred during liquid transfer. • Bubble in 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.
Lower than expected DNA yield	Samples left on instrument for an extended period of time after run completion.	Unload the 48-Well Nucleic Acid Archive Plate as soon as possible after the purification run is complete. The onboard plate chiller turns off after 60 minutes.

Observation	Possible cause	Recommended action
Low sample volume in some 48-Well Nucleic Acid Archive Plate wells	Pipetting error, elution buffer not centered in the well.	Reference archive plate maps and check the elution row in the 48-Well Nucleic Acid Archive Plate. Recover any eluate left in the wells.
Magnetic beads in 48-Well Nucleic Acid Archive Plate	Carry over of magnetic beads.	Carry over of magnetic beads can lead to incorrect quantitation values, but may not affect downstream applications such as PCR. Quantify manually using the Qubit™ Fluorometer or an equivalent fluorometric method, if needed.
		If excessive amount of beads are present, remove the beads by centrifugation or magnetic stand.

Genexus™ Dx FFPE DNA and RNA Purification

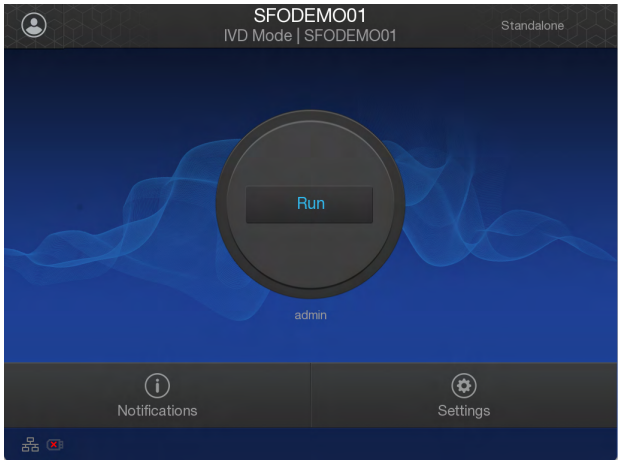
Observation	Possible cause	Recommended action
Lower than expected yield	Protease digestion incubation temperatures were below 60°C and 90°C.	Use a calibrated thermometer to verify incubation oven temperature. Reset oven temperature, if needed.
		Ensure that incubators have reached target temperature 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 Tube was low.	Ensure that AutoLys Tube caps are securely closed before incubation.
		Allow AutoLys Tube to cool to room temperature before centrifugation.
	AutoLys Tube collapsed during centrifugation.	Ensure that AutoLys Tube caps are securely held by the AutoLys 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 sample input amount. See “Recommended input amount” on page 33.
		Increase the sample input if needed as follows: • FFPE tumor resections—up to 14 × 5-µm curls/slides, or equivalent amount • 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 the protocol for FFPE sample preparation with the AutoLys Tube closely.

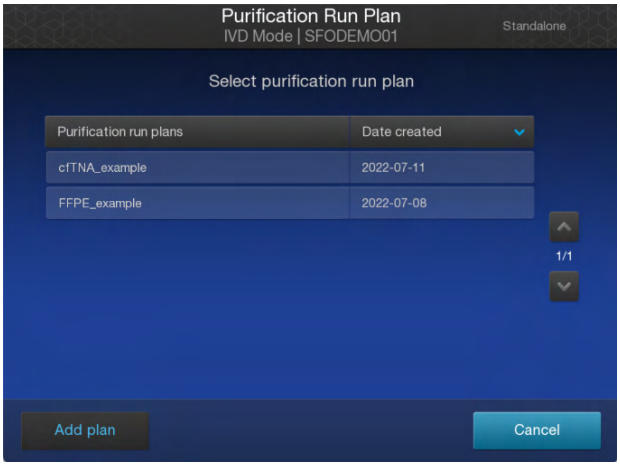
Observation	Possible cause	Recommended action
Cloudy samples in some 48-Well Nucleic Acid Archive Plate wells	Deparaffinization was incomplete.	Follow the protocol for FFPE sample preparation with the AutoLys Tube closely. This situation may not affect downstream applications.

A Supplemental information

Delete a run plan

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.

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.

```
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
```

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.

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.

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.

B Touchscreen reference

Note: This section provides information on the actions that can be taken by users signed in with the Administrator profile. Users signed with the Operator profile have access to a subset of these features. For information about user profiles, see “User access levels on the Genexus™ Dx Purification System” on page 22.

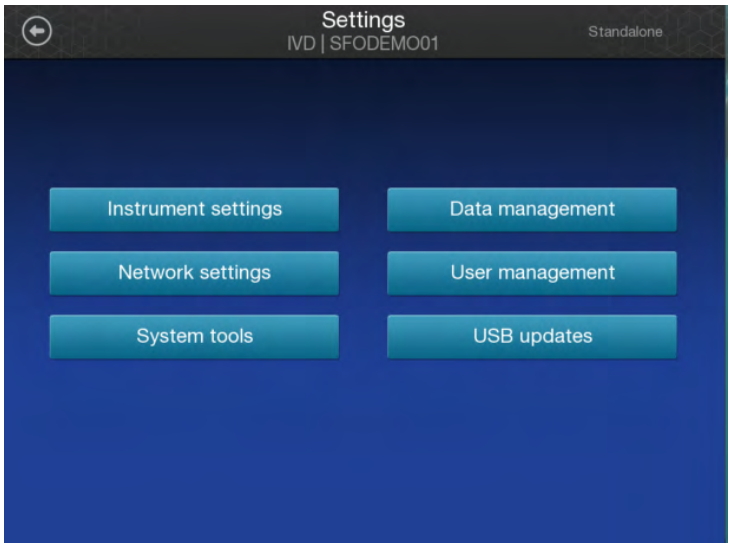
Touchscreen icons



Number	Icon	Description
1		User profile
2		USB – available
		USB – not available
3		Network connectivity – connected
		Network connectivity – not connected

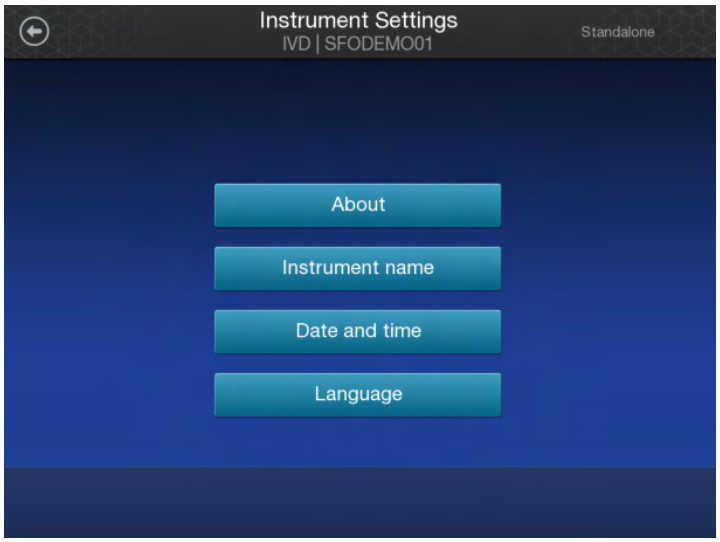
Settings

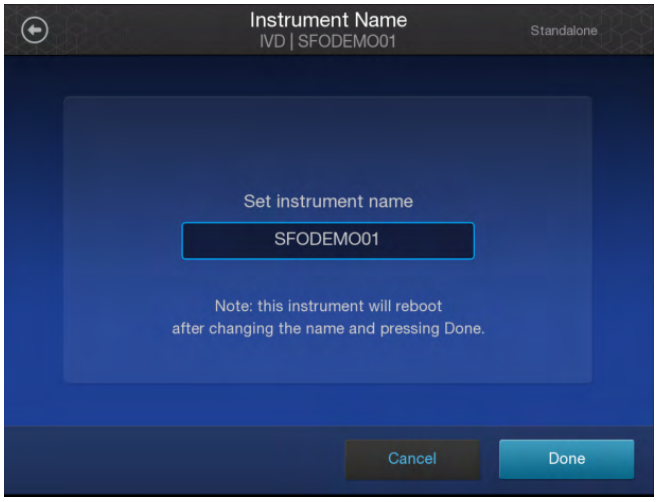
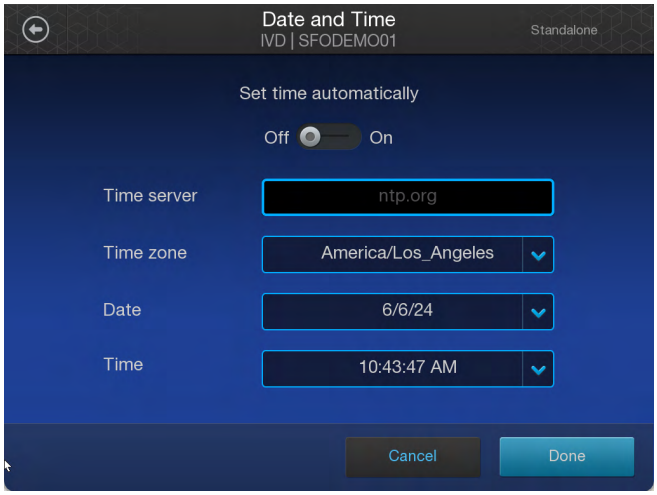
Use the **Settings** menu to view and change instrument settings, and to manage data.



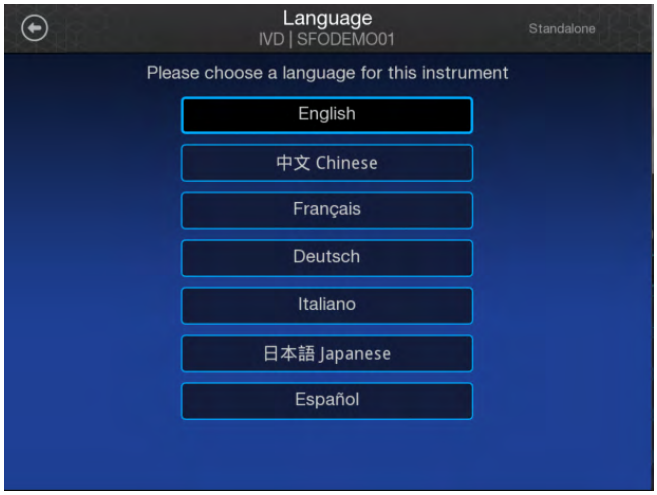
Instrument settings

Use the **Instrument Settings** menu to obtain information about the instrument, to set the instrument name, date and time, and language.



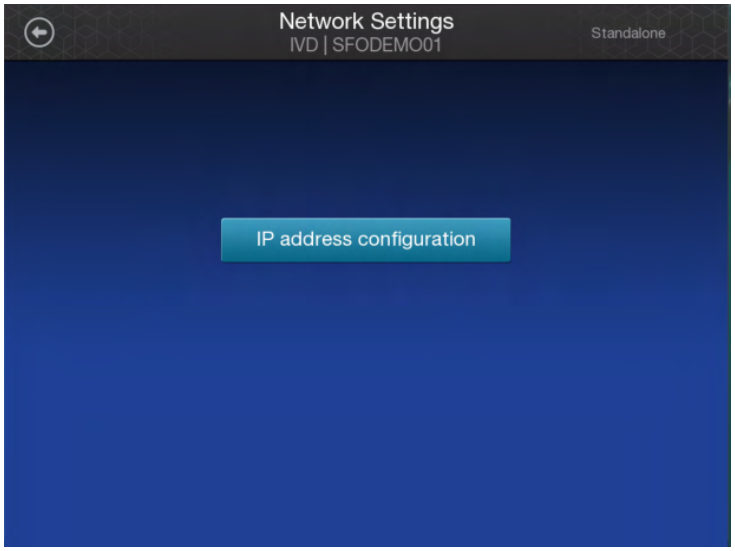
Item	Description	When and how to use
About	Provides instrument details. 	Use to view instrument reference information and performance qualification status.
Instrument Name		Use to change the instrument name. During installation the **Instrument Name** is set by the Thermo Fisher Scientific service representative. Renaming the instrument invalidaties the performance qualification (PQ). Only change the **Instrument Name** after consulting with your Thermo Fisher Scientific service representative.
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. **IMPORTANT!** Do not leave the **Time server** field blank. This will cause the instrument to stop responding and require a reboot to restore functionality.

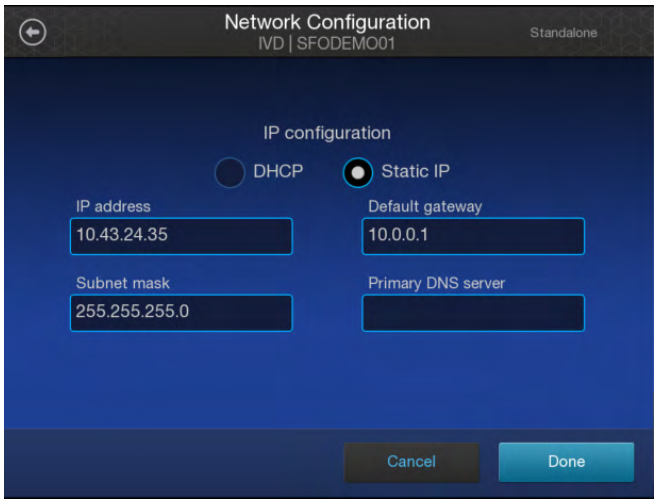
(continued)

Item	Description	When and how to use
Language	Lists available user interface languages. 	If changing the language. Tap to select a language, then tap **OK**. The instrument will reboot in the selected language.

Network Settings

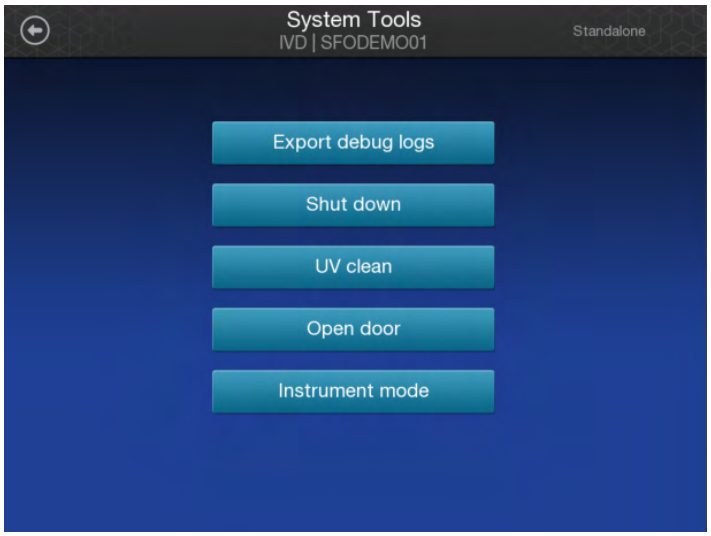
Use the **Network Settings** menu to configure the network IP address.

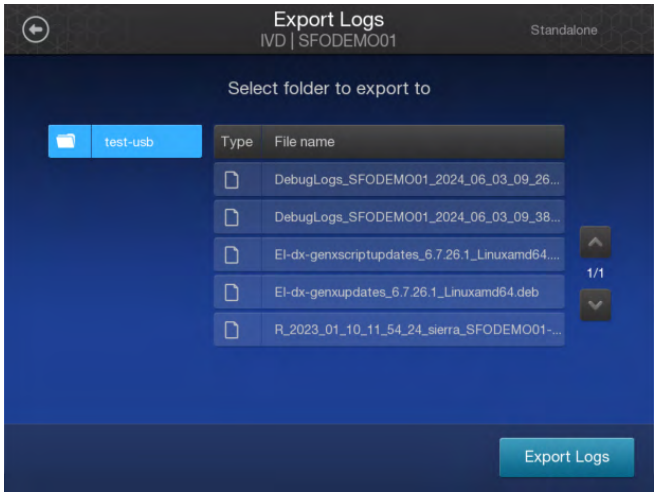
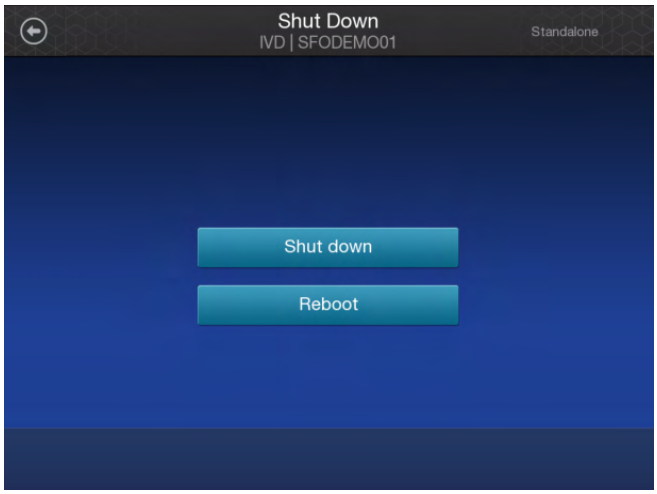


Item	Description	When and how to use
IP Address Configuration	Use this screen to set or change the IP configuration (DHCP or Static IP). 	1. Select either the **DHCP** or **Static IP** option. 2. Touch the screen in the field that you want to edit to activate the field. A virtual keyboard appears. 3. Enter the new information, then press **Done**.

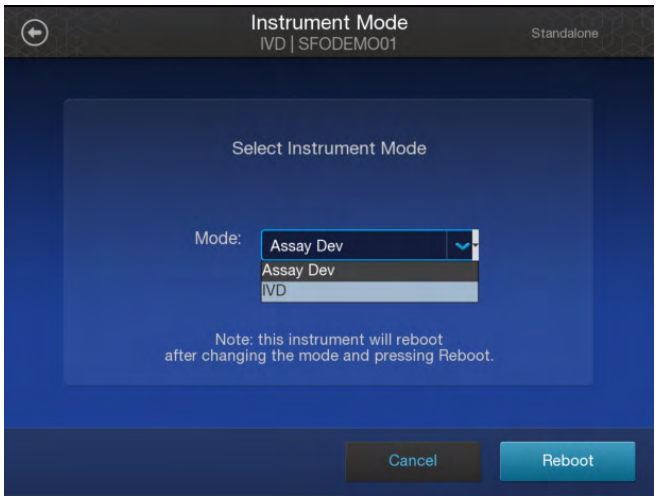
System Tools

Use the **System Tools** menu to upload instrument diagnostics, manage data, and shut down or reboot the instrument.



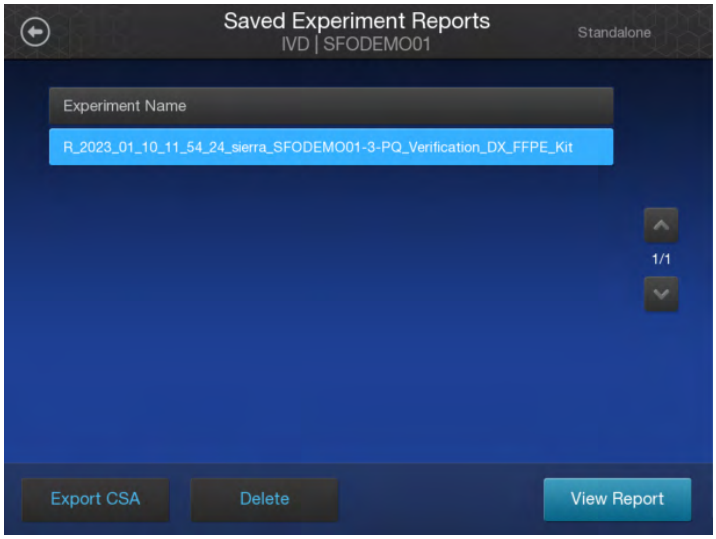
Item	Description	When and how to use
Export debug logs	Provides instrument error configuration information. 	Use for troubleshooting if directed to do so by Technical Support. 1. Tap ⚙ **(Settings) ▸ System Tools ▸ Export debug logs**. 2. In the **Export Logs** screen, tap the folder to export to. 3. From the file list, select a file to export. 4. Insert a USB drive into the USB port on the front of the instrument, then tap **Export Logs**. 5. When prompted, tap **Continue**. 6. When the export is complete, tap **Done**.
Shut down	Provides access to **Shut down** and **Reboot** commands. 	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. 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 the Genexus™ Dx Purification System On or Off” on page 19.
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 61.
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.

(continued)

Item	Description	When and how to use
Instrument mode	Sets the mode of the instrument to either **IVD** or **Assay Dev** (Assay Develoment). 	If changing the mode, select the mode from the dropdown list, then click **Reboot**.

Data management

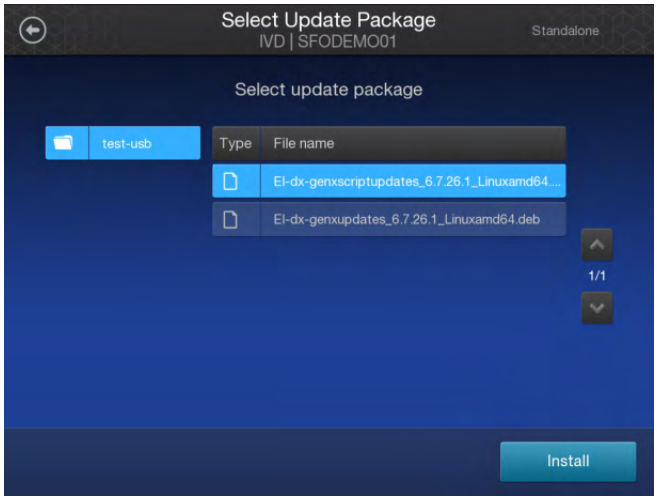
Use the **Data management** screen view run purification report information, export CSA information, or delete purification run reports.



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 52.
Export CSA	Provides instrument error configuration information. 	Use for troubleshooting if directed to do so by Technical Support. 1. Tap ⚙ **(Settings)** ▸ **Data management**, then tap in the row of the **Experiment Name** of interest. 2. Tap **Export CSA** 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**. 4. When prompted, tap **Continue**. 5. When the export is complete, tap **Done**.
Delete	Delete purification run reports that are no longer needed. **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 52.	1. Tap ⚙ **(Settings)** ▸ **Data management**, then tap in the row of the **Experiment Name** of interest. 2. Tap **Delete**. 3. When prompted, tap **Delete** again.

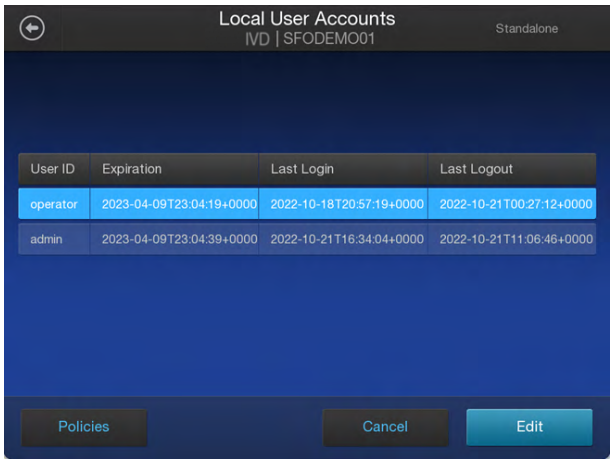
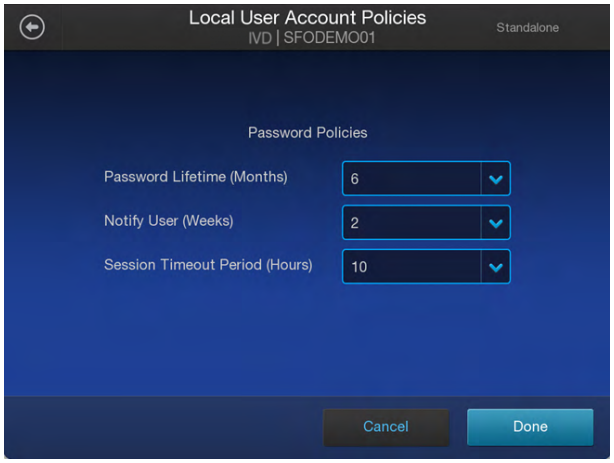
USB updates

Administrators use the **USB update** screen to perform software updates.

Item	Description	When and how to use
USB updates	Used to perform software updates. 	Administrators perform software updates as needed. 1. Insert the USB drive with the software upgrade package into the USB port on the front of the instrument. 2. Tap (Settings) ▸ **USB Updates**. 3. Navigate to the file destination on the USB drive, then tap **Install**. 4. Follow the prompts on the screen to complete the install.

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**. 2. Tap **Edit**, then enter then new password, then tap **Done**. 3. Tap **Done**, then when prompted, tap **Close**.
Policies		1. To update password policies, tap ⚙ **(Settings)** ▸ **User management**, then tap **Policies**. 2. Change the following settings as needed, then tap **Done**. • **Password Lifetime**—Set the number of months until a user password expires. • **Notify User**—Set the number of weeks before a user account password expires that the user receives a notification to reset their password. • **Session Timeout Period**—Set the number of hours before a session times out and a user must login to their user account again

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.

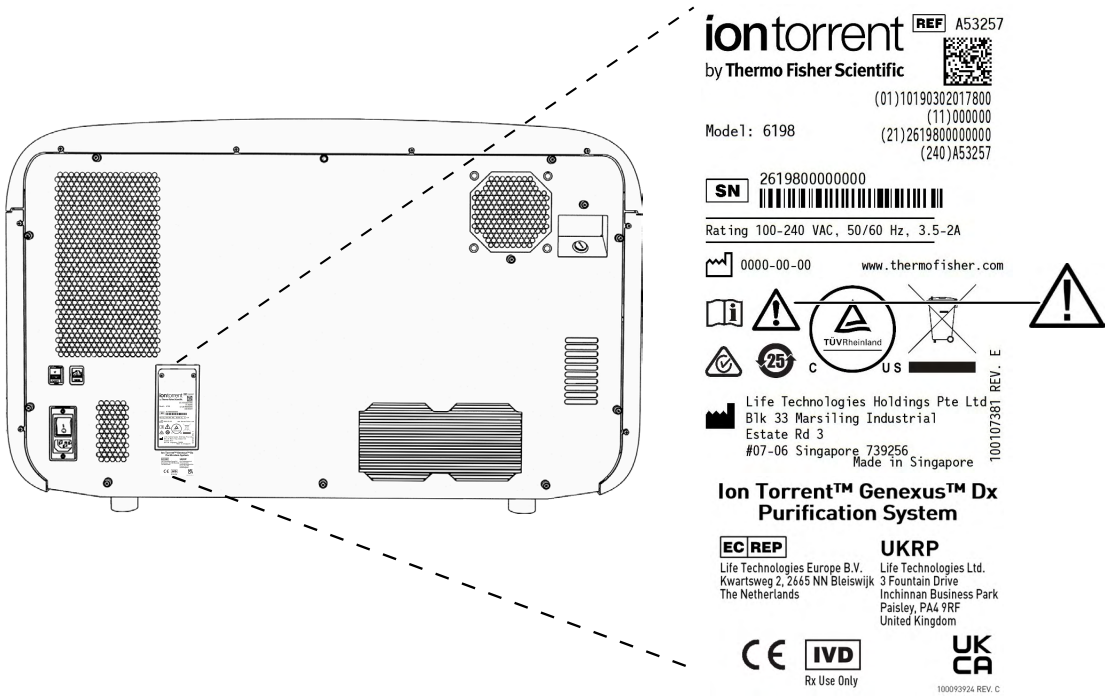
Standard safety symbols

Symbol and description
CAUTION! Risk of danger. Consult the manual for further safety information.
CAUTION! Risk of electrical shock.
CAUTION! Hot surface.
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
 Rear panel

Control and connection symbols

Symbol	Description
	On (Power)
	Off (Power)
	Protective conductor terminal (main ground)

(continued)

Symbol	Description
~	Alternating current

Conformity symbols

Conformity mark	Description
	Indicates conformity with safety requirements for Canada and U.S.A.
	Indicates conformity with China RoHS requirements.
CE	Indicates conformity with European Union requirements.
	Indicates conformity with Australian standards for electromagnetic compatibility.
	INDICATES CONFORMITY WITH THE WEEE DIRECTIVE 2012/19/EU. **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.

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 lifting or moving the instrument or accessories:

- Depending on the weight, moving or lifting may require 2 or more persons.
- If you decide to lift or move the instrument after it is installed, do not attempt to do so without the assistance of others, the use of appropriate moving equipment, and proper lifting techniques.
- Ensure that the path from where the object is to where it is being moved is clear of obstructions.
- Participants should coordinate lift and move intentions with each other before lifting and carrying.



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 that 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. Position 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 EN 61010-1 UL 61010-1 CAN/CSA C22.2 No. 61010-1	*Safety requirements for electrical equipment for measurement, control, and laboratory use – Part 1: General requirements*
IEC 61010-2-010 EN 61010-2-010	*Safety requirements for electrical equipment for measurement, control and laboratory use – Part 2-010: Particular requirements for laboratory equipment for the heating of materials*

(continued)

Reference	Description
IEC 61010-2-081 EN 61010-2-081	*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 EN 61010-2-101	*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 IEC 61326-1	*Electrical Equipment for Measurement, Control and Laboratory Use – EMC Requirements – Part 1: General Requirements*
EN 61326-2-6 IEC 61326-2-6	*Electrical equipment for measurement, control and laboratory use. EMC requirements. Particular requirements. In vitro diagnostic (IVD) medical equipment*
FCC Part 18 (47 CFR)	U.S. Standard *Industrial, Scientific, and Medical Equipment*
AS/NZS CISPR 11	*Limits and Methods of Measurement of Electromagnetic Disturbance Characteristics of Industrial, Scientific, and Medical (ISM) Radiofrequency Equipment*
ICES-001, Issue 4	*Industrial, Scientific and Medical (ISM) Radio Frequency Generators*
FCC Part 15 Subpart B (47 CFR)	*U.S. Standard Radio Frequency Devices* This equipment was tested and found to comply with the limits for a Class A digital device, pursuant to part 15 of the FCC Rules. These limits are designed to provide reasonable protection against harmful interference when the equipment is operated in a commercial environment. This equipment generates, uses, and can radiate radio frequency energy and, if not installed and used in accordance with the instruction manual, may cause harmful interference to radio communications. Operation of this equipment in a residential area is likely to cause harmful interference, in which case the user is required to correct the interference at his own expense. The Genexus™ Purification System has no user serviceable parts. Contact your Thermo Fisher Scientific field service engineer for instrument service or repair with approved parts.
IEC 60601-1-2	Medical electrical equipment—Part 1-2: General requirements for basic safety and essential performance—Collateral Standard: Electromagnetic disturbances – Requirements and tests

IEC 60601-1-2 immunity tests

Immunity test	IEC 60601-1-2 test level	Compliance level	Electromagnetic environment - guidance
Electrostatic discharge (ESD) IEC 61000-4-2	+/- 8 kV contact +/- 15 kV air	+/-8 kV contact +/-15 kV air	Use flooring made of wood, concrete or ceramic tile. If floors are covered with synthetic material, maintain the relative humidity at least 30%.
Electrical fast transient/burst IEC 61000-4-4	+/- 2 kV 100 Hz repetition frequency	+/- 2 kV 100 Hz repetition frequency	Use mains power quality of a typical commercial or hospital environment.
Surge IEC 61000-4-5	+/- 1 kV line to line	+/- 1 kV line to line	Use mains power quality of a typical commercial or hospital environment.
	+/- 2 kV line to earth	+/- 2 kV line to earth	
Power frequency (50/60 Hz) magnetic field IEC 61000-4-8	30 A/m	30 A/m	Maintain power frequency magnetic fields at levels characteristic of a typical location in a typical commercial or hospital environment.
Conducted RF IEC 61000-4-6	3 Vrms 150 kHz to 80 MHz 6 Vrms in ISM bands between 150 kHz and 80 MHz 80% AM at 1 kHz	3 Vrms 150 kHz to 80 MHz 6 Vrms in ISM bands between 150 kHz and 80 MHz 80% AM at 1 kHz	Field strengths from fixed RF transmitters, as determined by an electromagnetic site survey, should be less than the compliance level in each frequency range. Interference may occur in the vicinity of equipment marked with the following symbol:
Radiated RF IEC 61000-4-3	3V/m 80 MHz to 2.5 GHz 80 MHz - 2.7 GHz 80% AM at 1 kHz	3V/m 80 MHz to 2.5 GHz 80 MHz - 2.7 GHz 80% AM at 1 kHz	

Note:

- UT is the AC mains voltage before application of the test level.
- At 80 MHz and 800 MHz, the higher frequency range applies.
- These guidelines may not apply in all situations. Electromagnetic propagation is affected by absorption and reflection from structures, objects, and people.

Field strengths from fixed transmitters, such as base stations for radio (cellular/cordless) telephones and land mobile radios, amateur radio, AM and FM radio broadcast and TV broadcast cannot be predicted theoretically with accuracy. Over the frequency range of 150 kHz to 80 MHz, maintain field strengths less than 3 V/m.

Emissions test	Compliance	Electromagnetic environment - guidance
RF emissions CISPR 11	Group 1 Class A	The EMISSIONS characteristics of this equipment make it suitable for use in industrial areas and hospitals (CISPR 11 class A). If it is used in a residential environment (for which CISPR 11 class B is normally required), this equipment might not offer adequate protection to radio-frequency communication services. The user might need to take mitigation measures, such as relocating or reorienting the equipment.
Harmonics emissions IEC 61000-3-2	Class A	
Voltage fluctuations/ flicker emissions IEC 61000-3-3	Complies	

Environmental design standards

Reference	Description
Directive 2012/19/EU	European Union “WEEE Directive”—Waste electrical and electronic equipment
RoHS Directive 2011/65/EU & Commission Delegated Directive (EU) 2015/863	European Union “RoHS Directive”—Restriction of hazardous substances in electrical and electronic equipment
SJ/T 11364-2014	“China RoHS” Standard—Marking for the Restricted Use of Hazardous Substances in Electronic and Electrical Products For instrument specific certificates, visit our customer resource page at **www.thermofisher.com/us/en/home/technical-resources/rohs-certificates.html**.

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.

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

Documentation and support

Related documentation

Document	Publication number
Genexus™ Dx Purification System User Guide for use in Assay Development mode	MAN1000104
Genexus™ Dx Purification System Site Preparation Guide	MAN1001599
Genexus™ Dx Integrated Sequencer User Guide	MAN1000073
Genexus™ Dx Software 6.8 User Guide	MAN1000074

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**.

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**.

ion torrent

Genexus™ Dx Software 6.8

USER GUIDE

Publication Number MAN1000074

Revision C

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 C (English)

Revision	Date	Description
C	23 November 2025	• Updated with software cybersecurity features, requirements, and guidelines. • Minor updates made in run planning topics.
B	24 July 2025	• Updated "Sign in to a new user account" on page 18 and "Reset a user password (administrator)" on page 155 to correct password requirements in Genexus™ Dx Software 6.8.2. • Added "Sequencer decommissioning and data sanitization" on page 13.
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 6.8.2.1 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. • Reagents and supplies for operating the Genexus™ Dx Integrated Sequencer.
Software	• Genexus™ Dx Software 6.8.2.1 or later.
Functional knowledge and understanding - all users	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to adminstrator credentials, SSH private keys and the associated SSH key passwords. • Main functions of the Genexus™ Dx Integrated Sequencer. • Main features of the Genexus™ Dx Software.
Functional knowledge and understanding - system administrators	• Proficiency in the use of Linux operating system. Knowledge of cybersecurity measures and best practices for credentials security, including but not limited to adminstrator credentials, SSH private keys and the associated SSH key passwords. • Use of security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

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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 6.8.2.1 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. The operating system has an Ubuntu™ Pro license that includes expanded security maintenance coverage.

It is recommended that Genexus™ Dx Software be used only on computers that are maintained by facility system administrators and IT and security personnel, and include security software, including but not limited to firewalls, antivirus software, encryption-based passwords systems.

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 facility system administrators and IT and security personnel. Genexus™ Dx Software does not provide any network or security configuration files, utilities, or instructions. The Genexus™ Dx Integrated Sequencer has a firewall enabled by default. Only ports required for the operation of the instrument and software are open.



CAUTION! Data can be lost or corrupted if you operate a sequencer that does not have sufficient security controls in place.

If external or network drives are connected to Genexus™ Dx Integrated Sequencer for archiving or backups, it is the sole responsibility of 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 191.

Data backup and data storage

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.

Note: A paid data migration service is available if needed. Contact your Thermo Fisher Scientific representative for more information.

For more information about data storage compatibility, contact your local support team.

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.

Antivirus software

For recommendations for antivirus software that is compatible with Genexus™ Dx Software 6.8.2.1, see the current version of the Genexus™ Dx Software release notes.

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.

Sequencer decommissioning and data sanitization

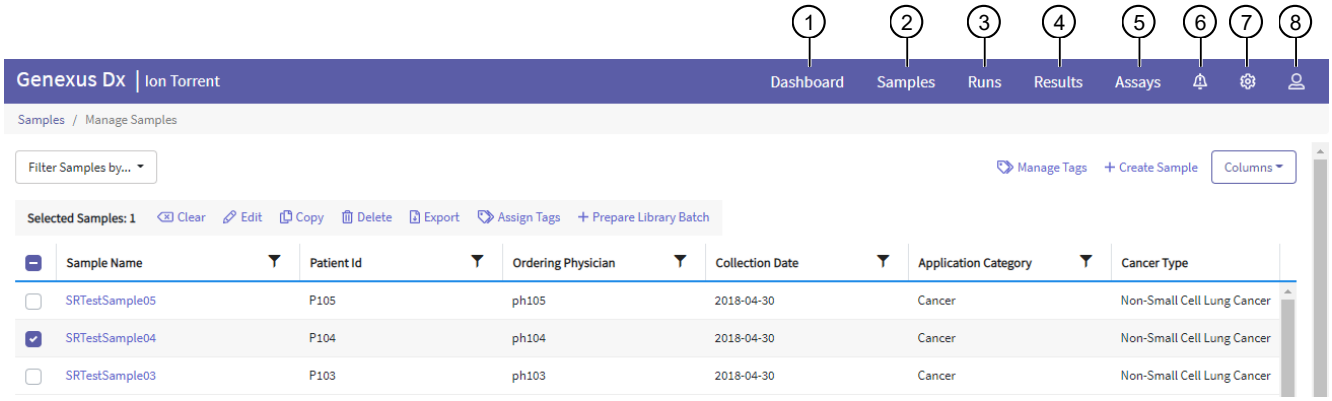
Contact a Thermo Fisher Scientific Field Service Engineer for any request to decommission a Genexus™ Dx Integrated Sequencer. Decomissioning and data sanitization must be carried out by a Field Service Engineer.

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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 • Generate, view, and sign variant reports • Send notifications • Download results files • View and edit notes for the sample result • View the audit trail for sample results • View notifications
Operator	Report functions, plus: • Add, import, and export sample files • Edit sample files • Prepare library batches • Plan, save, audit, and delete runs • Monitor runs • View results and reports • View the audit trail for samples, library batches, runs, and assays • Reanalyze runs

(continued)

Users at this level	Can perform these actions
Manager	Operator functions, plus: • Create, edit, and obsolete sample attributes • Import assays • Create report templates • Edit and delete sample files • Add or edit Thermo Fisher accounts • Restart a stalled or failed run • Access services information
Administrator	Operator and manager functions, plus: • View, export, and print audit records • Configure network settings • View and manage software updates • Configure backup settings and restore runs • Manage sequencer and software log files • 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 18.

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**. 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**. 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**. b. In the **Edit Security questions and answers** dialog box, select a security question from the dropdown list, then enter an answer. c. Enter the password for the account that is associated with the profile. 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 228.

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. 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.

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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 Library to Result	“Nucleic Acid to Result or Library to Result workflow” on page 22

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 24)

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 53)

OR

Plan a Library to Result run (page 57)

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 66)

Monitor the run in Genexus™ Dx Software in real time.



Review data and results (page 70)

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 29 and “System-installed sample attributes” on page 219.

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 33 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 25.

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 27.

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***.

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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 28 and "Search for samples" on page 25.

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)**. b. In the first dropdown list, select **Is equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**.
List samples created within a date range	a. In the **Created On** column header, click **(Filter)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select **And**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date that the samples of interest were created. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date that the samples of interest were created. d. In the Boolean operator dropdown list, select **Or**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date that the samples of interest were created. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is before or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**.
List only samples that were created after a date that you indicate	a. In the **Created On** column header, click **(Filter)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the date of interest. 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 29 and “Import samples” on page 30.

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 34.
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 30.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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.

Note: Genexus™ Dx Software encrypts data to protect the information that you enter for a sample.

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 219. 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 33. Upload the edited file using the **Browse** button. **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**. b. Navigate to the file, then click **Open**. 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 219.

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 39.

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 over normal cells in the sample. Percent tumor content.

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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. **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 30.

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 29, “Create multiple samples” on page 29, “Import samples” on page 30, “Edit a sample” on page 39, and “System-installed sample attributes” on page 219.

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 39 and “Review sample edit history” on page 41.

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 29 and "Import samples" on page 30.

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 37.

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**. b. Enter the note, then click **Save**. 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 219 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 29.
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 41.

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 24.

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 53.

Note:

- Each library batch must have a unique library batch ID.
- Fields identified with a red asterisk (*) are required.

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 53.

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.

   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 “Find samples” on page 25.

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, 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 224.

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 45.

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 47.

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 45.

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 48.

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 45.

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. 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**. 2. Modify the information in the **Edit Library batch** screen, then click **Submit**. Alternatively, in the library batch details pane, click the library batch ID to perform this action. 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**. The **Audit Records** screen opens with time-stamped audit records that include details and changes for each action performed on the library batch. 2. Click the time stamp and action listed in the left pane of the screen to view audit details for other actions. 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**. The **Add Notes** dialog box opens and notes that are associated with the library batch are listed by user, date, and time. 2. In the **Add Notes** dialog box, click + **Add Note**. 3. Enter the note, then click **Save**. 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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- Plan a Nucleic Acid to Result run 53
- Plan a Library to Result run 57
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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 24.
- 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 42.

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 51.

For information to filter run plans by a common trait, such as run status, see “Filter the list of run plans” on page 53.

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.

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 51.
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. • 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 29, or “Import samples” on page 30.
- 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 *Genexus™ Dx Integrated Sequencer User Guide* (Pub. No. MAN1000073).

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 one or more assays 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.

      After selecting an assay and the application type for the assay, the list is filtered to show compatible assays that can be selected and run at the same time.

   c. If more assays are included in the run, repeat substep 3b for each extra assay.

   d. 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 positive 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. Red denotes a chip lane in the run containing assigned samples that exceed 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 will 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 you selected more than one research application or assay, repeat substep 4a for each research application for each assay in the run plan.

   c. 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.

   d. 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.

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 *Genexus™ Dx Integrated Sequencer User Guide* (Pub. No. MAN1000073).

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 42.

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 one or more assays 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.

      After you select 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.

      Note: To create a new assay, see Chapter 8, "View and manage assays (manager/ administrator)".

   c. If more assays are included in the run, repeat substep 3b for each extra assay.

   d. 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, 8 samples were included in a library batch instead of 7. The run is allowed if the lane capacity is exceeded, but you will 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.

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 59.

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 60.

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***.

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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 53, or “Plan a Library to Result run” on page 57.

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. 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. b. Select **Ion Torrent™ GX5™ Chip** then click **Filter**.
List assays according to library chemistry	a. Click **(Filter)** in the **Library Chemistry** column. b. Select the library chemistry: • **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. 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 53 or “Plan a Library to Result run” on page 57.

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 63, "Add a note to a run" on page 64, "Review and download a run history" on page 62.

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.

(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 103.
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 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: • **Create** • **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 53 and “Plan a Library to Result run” on page 57.

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)**.

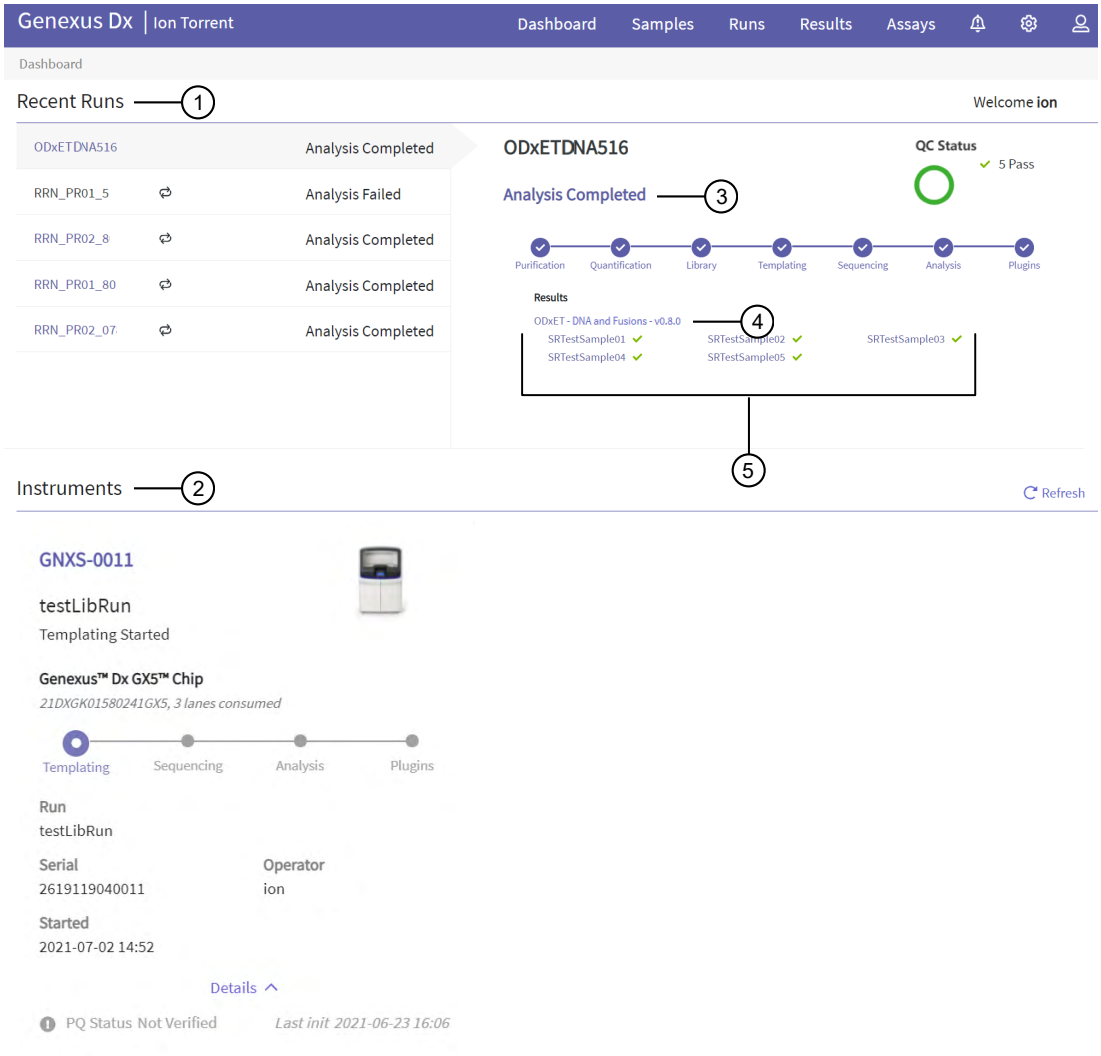
The note is no longer listed in the **Add Notes** dialog box and the note is removed from the software.

Click **Close** to return to the **Manage Samples** screen.

5 Genexus™ Dx Software Dashboard

- View run progress on the instrument 67
- View the status of an instrument 69
- Information about connected instruments 69

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. The **Run Summary** tab opens. For more information, see “The run summary” on page 103. 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. 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. b. In the run details pane in the right side of the screen, click the assay name of interest. The **Assay Metrics** tab opens. For more information, see “Assay metrics and the run report” on page 106. 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. b. In the run details pane in the right side of the screen, click the sample name of interest. The **Key Findings** tab opens. For more information, see “Key Findings” on page 74. In the menu bar, click **Dashboard** to return to the **Dashboard**.

Option	Description
View the progress of the run.	a. In the left side of the screen, click the row of the run of interest. b. In the run details pane in the right side of the screen, place the pointer over the status for the run. 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** 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**. **(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

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. 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

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- Reanalyze a sample 118
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- Data files and flow 123

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.

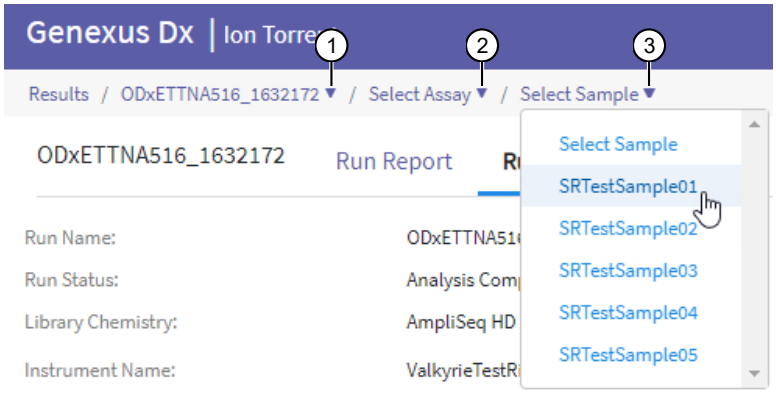
Selection	Description
Click **Results** ▸ **Sample Results**	Select this option to review completed sample results and reports.
Click **Results** ▸ **Run Results**	Select this option to review completed run results and reports by assay.
Click **Results** ▸ **Verification Results**	Select this option 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. 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. 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. 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.

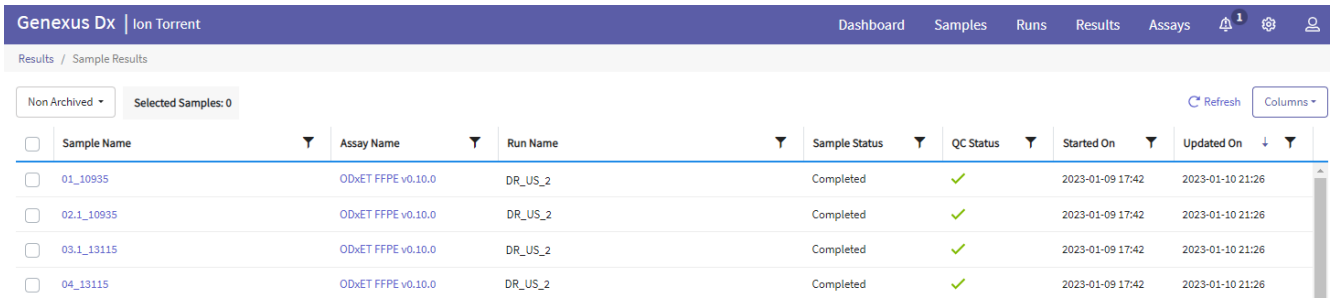
See “Find sample results” on page 96 for information about how to find sample results of interest.

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 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 25.

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. • ✔ (Passed) indicates that the sample passed all QC metrics. • ✘ (Failed) indicates that the sample failed a QC metric. • — (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 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. • **Report**—Download the report (available only for samples with a sample status of completed). For more information, see "Variant report" on page 124. • **Assign Tags**—Add or remove the tags assigned to a sample result. For more information, see "Sample tags" on page 34. • **Audit**—View the audit trail for the run. For more information, see "Review and download a run history" on page 62. • **Notes**—View, add, or delete notes for sample result. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see "Download a Customer Support Archive" on page 223. • **Reanalyze**—Reanalyze a sample from the alignment step when a sample or reanalysis successfully completes the basecalling step. 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 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	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 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. • **Indels**: insertion and deletion variants. For more information, see “View SNV/INDEL results” on page 77.
Fusions	Translocations of genetic material and splicing variants. 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. For more information, see “View CNV results” on page 84.

Search and filter the list of variants

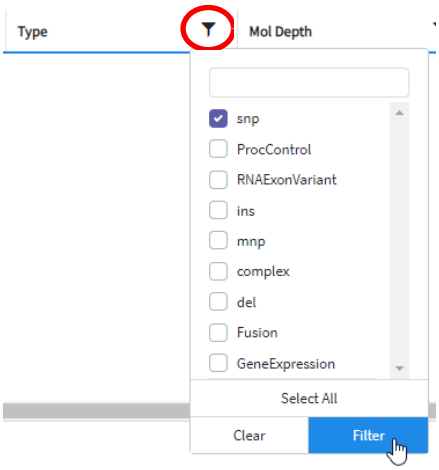
You can search and filter to immediately narrow the list of results that are shown in the variant tables. You can apply filters to columns of information that appear in the screen. The filters, available at the top of each column, immediately narrow the list of information in any columns to which filters 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 77.
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 75.

Column	Description
Variant ID	The name of the hotspot as defined in the Browser Extensible Data (BED) file. 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.

(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. • **snp** (single nucleotide polymorphism) • **mnp** (multi-nucleotide polymorphism) • **ins** (insertion) • **del** (deletion) • **complex**
Test Result	Indicates the presence or absence of an SNV/Indel variant. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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). 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 79.
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 75.

Column	Description
Variant ID	The name of the fusion target as defined in the BED file. 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. 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. • **POSITIVE**—indicates a high confidence call that passes all filter thresholds at a given variant position. • **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. • **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. **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. Imbalance score = Observed imbalance (in samples) / Expected imbalance (in the baseline) where Observed imbalance = 3' of breakpoint (in samples) / 5' of breakpoint (in samples) • 3' of breakpoint is the sum of the normalized reads of amplicons 3' of a predicted breakpoint for the gene in the sample. • 5' of breakpoint is the total normalized reads of the amplicons for the gene in the sample. Expected imbalance = 3' of breakpoint (in the baseline) / 5' of breakpoint (in the baseline) • 3' of breakpoint is the sum of normalized baseline values of amplicons 3' of the breakpoint for the gene. • 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.

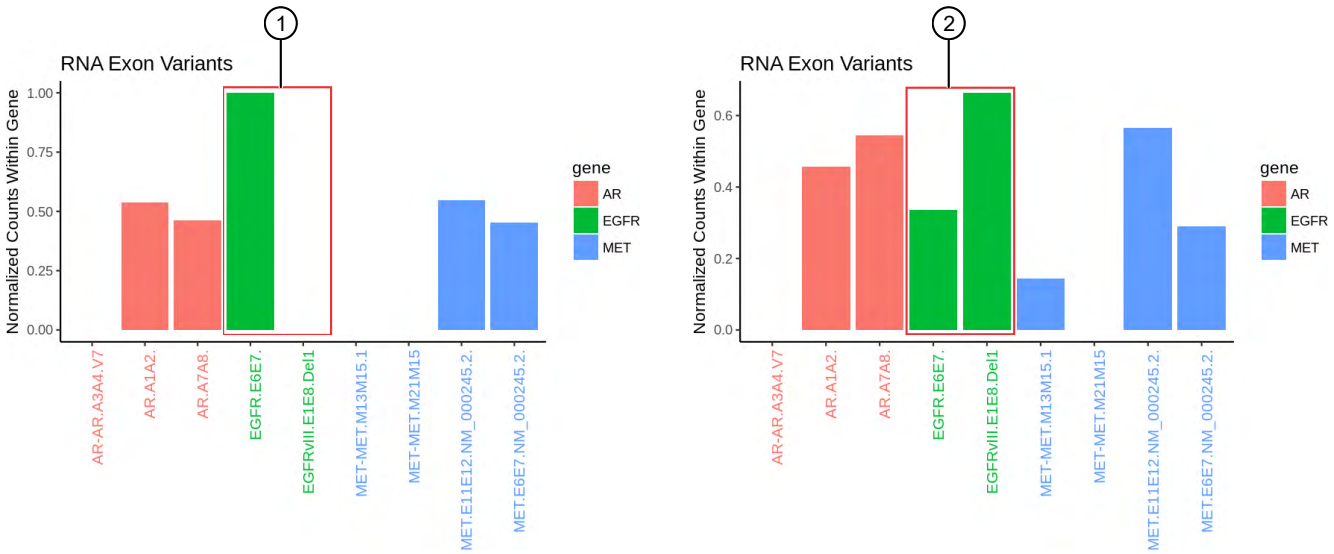
[1] Column appears only in analyses that use the exon tiling fusion detection method.

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 79.
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

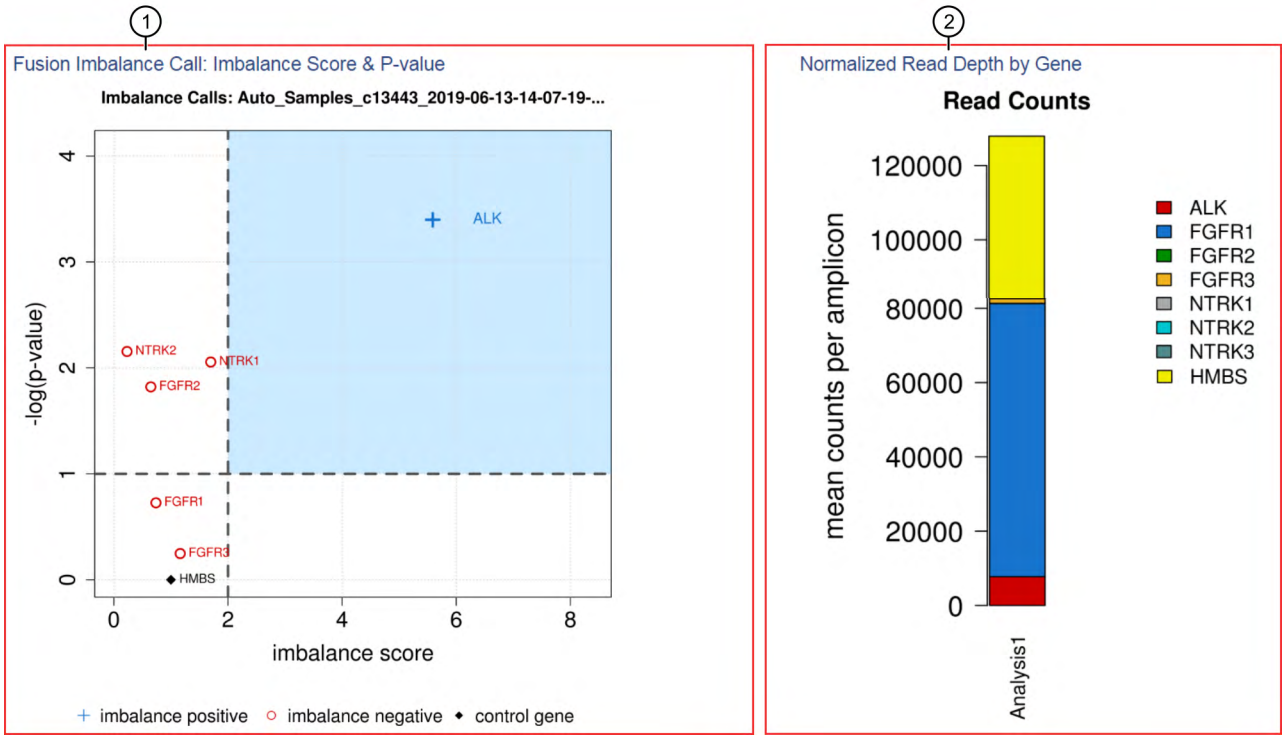
The **RNA Exon Tile Fusion Imbalance** data view provides a visual representation of the RNA fusion imbalance analyses.

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.

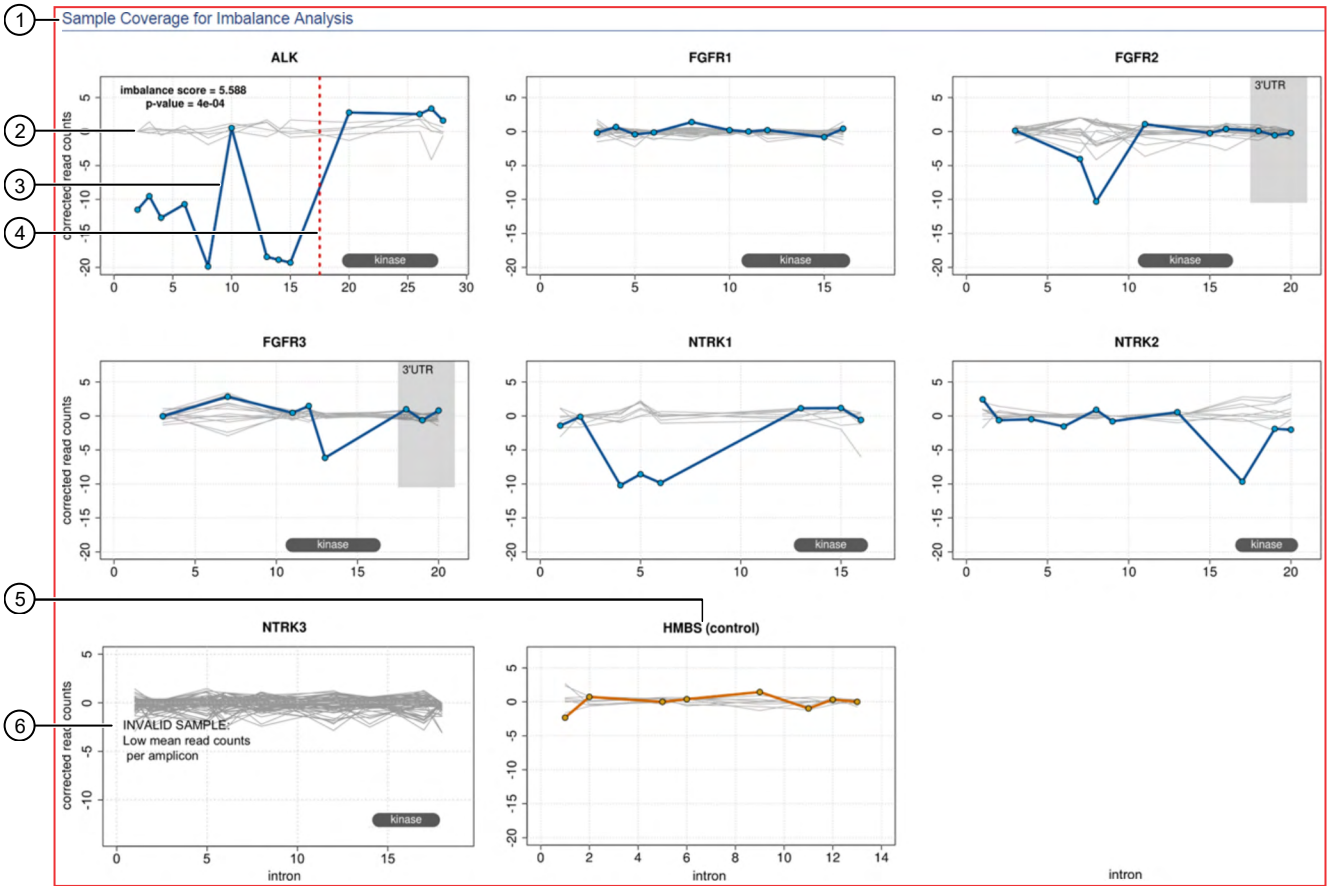
1. In the menu bar, click **Results** ▸ **Sample Results**.
2. In the **Sample name** column, click the name of the sample of interest.
3. Click the **Variants** tab, then click **Fusions**.
   The **Fusions** table opens to display fusions results. For more information, see “Fusions table” on page 79.
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 75.

IMPORTANT! *(FFPE samples only)* If the **%Cellularity** value for a sample is set to <100, then the magnitude of copy number gain or loss can be decreased. For more information, see “System-installed sample attributes” on page 219.

Column	Description
Variant ID	The identifier of the CNV variant. 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. • **POSITIVE**—Indicates a high confidence call that passes all filter thresholds. • **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. • **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.

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**. This option is available only when the forward strand is shown.
View the forward stand	Click **Forward**. This option is available only when the reverse strand is shown.
View the reference sequence translation	Click **Three-Frame Translate**. This option is available only when the amino acid sequence is not shown.
Hide the reference sequence translation	Click **Close Translation**. 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.

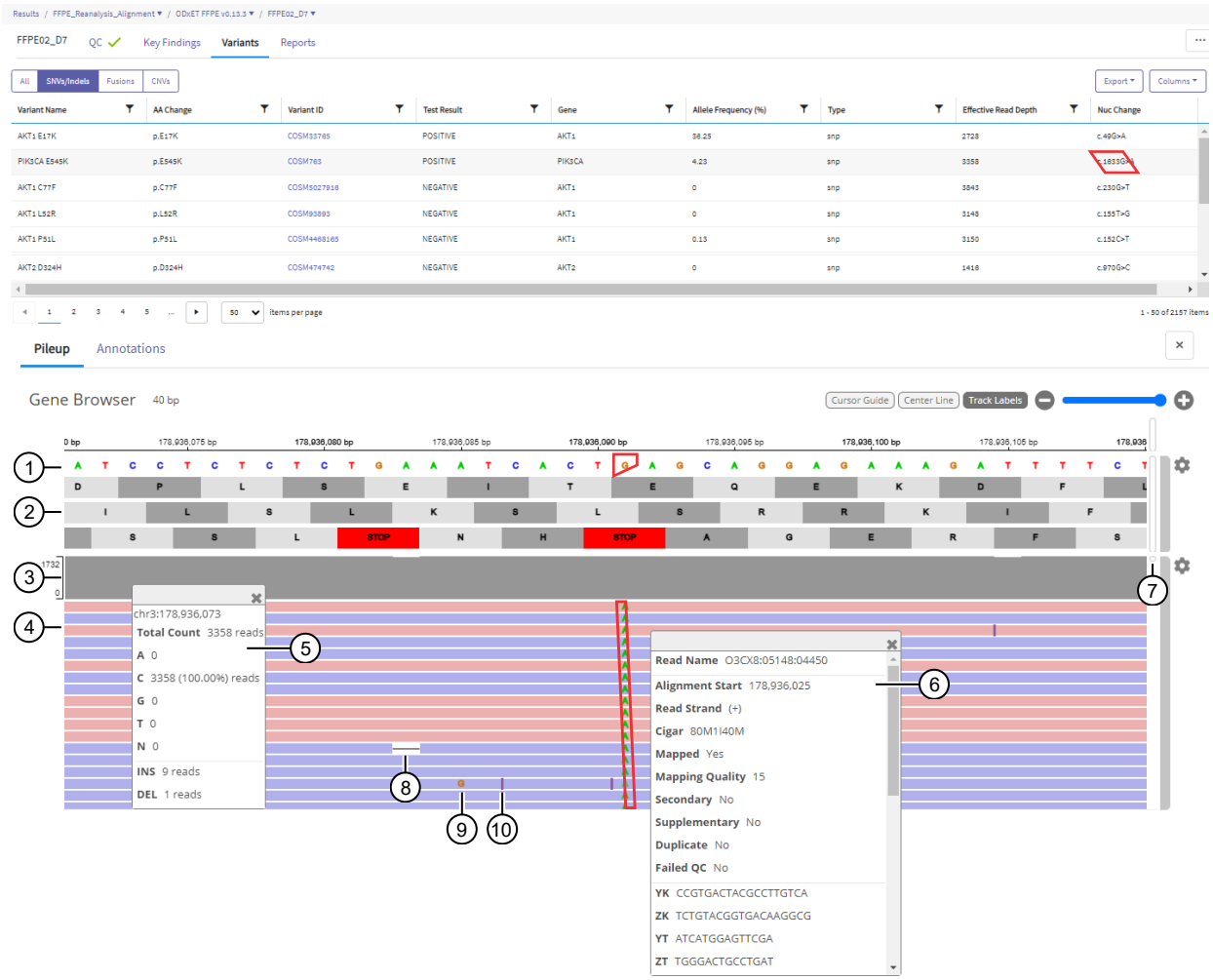
1. In the menu bar, click **Results** ▸ **Sample Results**.
2. Click a sample name to open the **Results** screen.
3. Click the **Variants** tab.
4. In the variants table, in the **Variant ID** column, click the identifier of the variant of interest.

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**. 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**. 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**. 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**. b. In the **Set visibility window** dialog box, enter the minimum number of bases needed to show the BAM tracks. 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**.

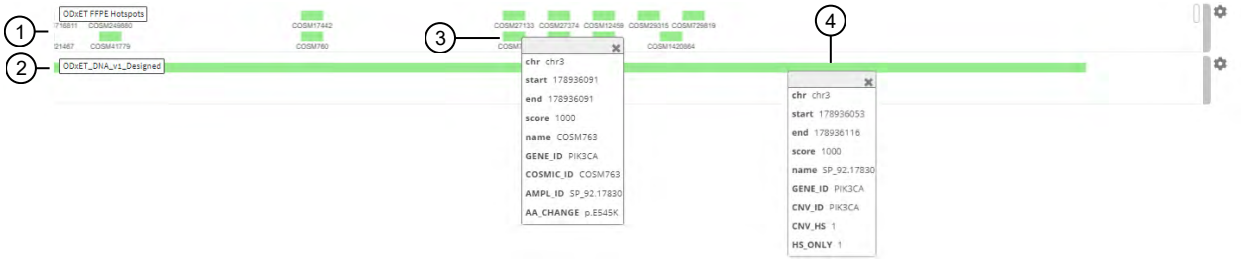
⑦ Use the slider to view more tracks.

⑧ Deletion.

⑨ Alternate base in the BAM read coverage track.

⑩ Insertion.

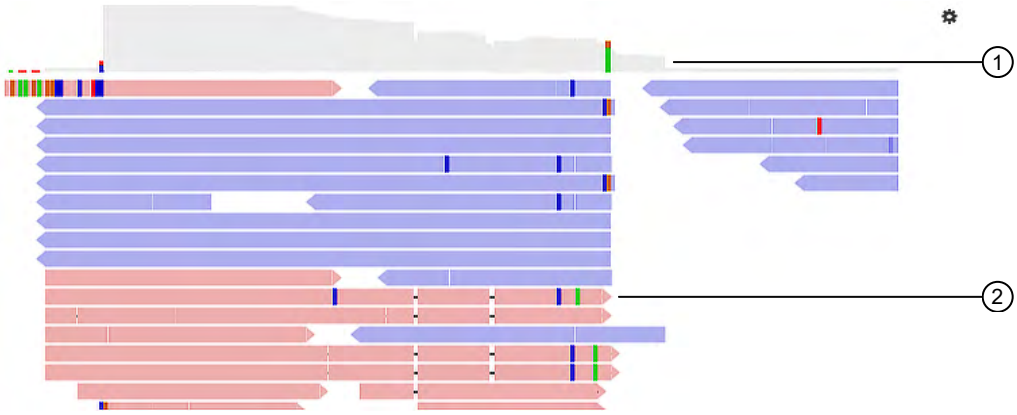
The following figure is an example of the reference tracks shown beneath the BAM tracks in the **Gene Browser**.



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.



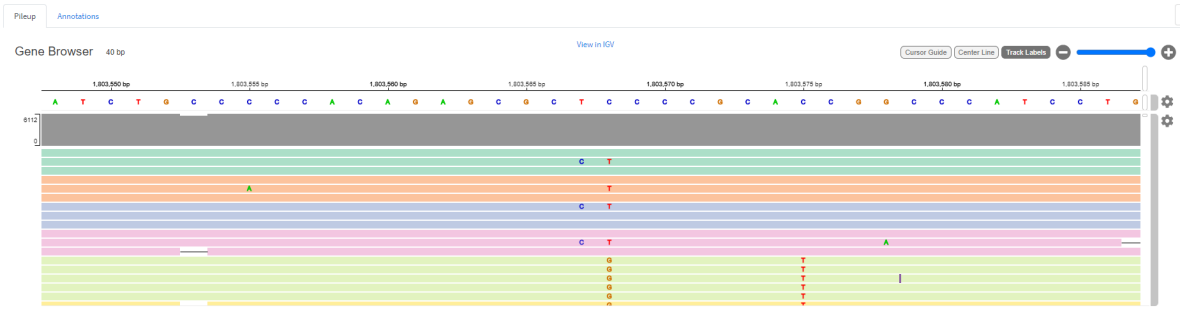
① Read depth
② Read location and direction of variants

View the variants from an 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 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.

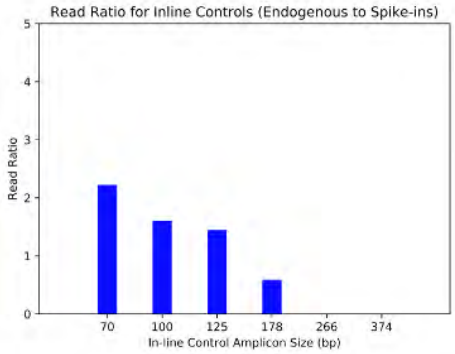
If a sample fails a single test metric, the sample fails that QC test. A sample must meet all QC parameter thresholds of a particular QC test in order to pass. The QC status is divided into the following categories.

- ✔ (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
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.
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 0.4 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.

(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.  **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).

(continued)

Metric	Description
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).

[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. • **Archived** • **Non Archived** 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. • **Completed** • **Running** • **Failed** • **Terminated** • **Stalled** • **Aborted** • **Pending** 2. Click **Filter**. 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. • **Failed** • **Passed** • **Not Calculated** 2. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the earliest date of the range. 4. In the Boolean operator dropdown list, select **And**. 5. In the second dropdown list, select **Is before or equal to**. 6. In the calendar that appears, select the latest date of the range. 7. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the earliest date for the sample results of interest. 4. In the Boolean operator dropdown list, select **Or**. 5. In the second dropdown list, select **Is before or equal to**. 6. In the calendar that appears, select the latest date for the sample results of interest. 7. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is before or equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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)**. 2. In the first dropdown list, select **Is after or equal to**. 3. In the calendar that appears, select the date of interest. 4. Click **Filter**. 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 34.

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 35.

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 34.

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 35.
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. 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. • **Delete**—Delete the run. • **Audit**—View the audit trail for the run. For more information, see "Review and download a run history" on page 62. • **CSA**—Download customer support archive (CSA) log files for the run to help with troubleshooting. For more information, see "Download a Customer Support Archive" on page 223. • **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**. 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**. For more information, see "Customer Support Archive (CSA)" on page 223.
View the history of variant classifications.	In the upper right corner of the screen, click ••• **(More Options)** ▸ **Variant Audit**. 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.

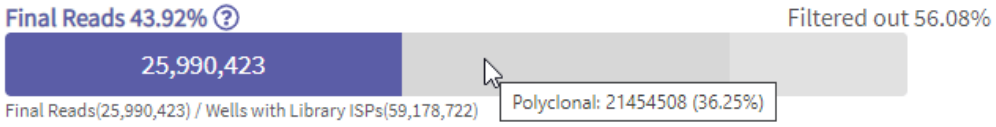
Assay metrics and the run report

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.

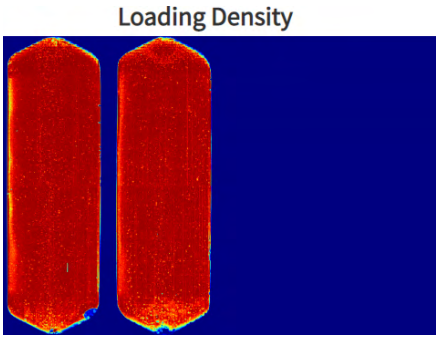
Run Report

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. To view polyclonal metrics, mouse over the first low quality portion (gray) of the **Final Reads** bar plot. 
Low Quality	Loaded wells with a low or unrecognizable signal. To view polyclonal metrics, mouse over the second low quality portion (gray) of the **Final Reads** bar plot. 
Adapter Dimer	Loaded wells with a library template of an insert size less than 8 bases. 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. 

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. 

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). 

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***.

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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 110.
Show or hide run results that are archived	a. In the dropdown list in the upper left corner, select an archive status. b. • **Archived** • **Non Archived** 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. • **Analysis Completed** • **Analysis In Progress** • **Analysis Failed** • **Analysis Aborted** • **Analysis Stalled** • **User Aborted** • **Run Aborted** • **Backup In Progress** • **Executing Plugins** • **Library Preparation Aborted** • **Library Preparation Completed** • **Library Preparation Started** • **Selected on Instrument** • **Sequencing Completed** • **Sequencing Started** • **Sequencing Aborted** • **Templating Aborted** • **Templating Completed** • **Templating Started** • **Kit Barcodes Required** b. Click **Filter**. 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)**. b. In the first dropdown list, select **Is equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date of the range. d. In the Boolean operator dropdown list, select **And**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date of the range. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the earliest date for the sample results of interest. d. In the Boolean operator dropdown list, select **Or**. e. In the second dropdown list, select **Is before or equal to**. f. In the calendar that appears, select the latest date for the sample results of interest. g. Click **Filter**. 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)**. b. In the first dropdown list, select **Is before or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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)**. b. In the first dropdown list, select **Is after or equal to**. c. In the calendar that appears, select the date of interest. d. Click **Filter**. 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. 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. • When a run fails during the basecalling step or earlier. • When the run has not already been successfully reanalyzed. 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. 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. 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 *Genexus™ Dx Integrated Sequencer 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 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.

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 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. 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. 2. Click the run or assay of interest from the results navigation bar. 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 • **NTC QC – DNA**—A summary of the DNA no template control quality control metrics • **NTC QC – RNA**—A summary of the RNA no template control quality control metrics • **Positive Control01 QC – DNA**—A summary of the DNA positive control quality control metrics • **Positive Control01 QC – RNA**—A summary of the RNA positive control quality control metrics • **Sample QC – RNA**—A summary of RNA sample quality control metrics • **Sample QC – DNA**—A summary of DNA sample quality control metrics • **Templating Control QC – CF-1**—A summary of the CF-1 quality control metrics This section is locked and cannot be customized.
Sequencing Run Details	Contains the following subsections: • **Assay Name**—The assay used in the run • **Panel**—The panel used in the run • **Started On**—The date and time that the run was started • **Completed On**—The date and time when the run was completed • **Operator**—The user who sent the run to the instrument • **Instrument**—The name of the instrument • **Serial Number**—The serial number of the instrument This section is locked and cannot be customized.
Reagents	Details about run consumables, including part numbers, expiration dates, and lot numbers. 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. 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 18.

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. • **Locked** • **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**. 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)**. Repeat for each section to include in the report template. c. Click **Apply**.
Remove a section.	In the box for the section, click ✕ **(Remove)**.
Remove multiple sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click the section name of interest, then click « **(Remove)**. Repeat for each section to remove from the report template. c. Click **Apply**.
Add all sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Available Sections** pane on the left, click **Select All**. c. Click **Apply**.
Remove all sections.	a. Click **Bulk Add**. b. In the **Bulk Add** dialog box, in the **Configured Sections** pane on the right, click **Select All**. 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**. b. Enter the note, then click **Save**. 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. • 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**. 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. • 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**. The filtered list of assays is shown in the **Manage Assays** screen. • 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. • 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**. The filtered list of assays is shown in the **Manage Assays** screen. • 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. 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: • **Locked**
Reference	Genome reference that is available in the annotation set. • **hg19** • **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**. • **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. • **Version**–The revision number of the assay.
Panel	In addition to other components, the panel section lists the reference files used in an assay. 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** • **CF-1 Control** • **NTC QC – DNA** • **Run QC** • **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: • **Create** **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. b. In the app, click +. 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. b. In the Connect website, click **InstrumentConnect** in the left sidebar. c. Click **Link instrument** in the upper right corner of the screen. d. Enter the QR code text shown in the **Link Instrument** screen. 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**. • 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. • 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.

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.

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. 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. 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**. 2. In the **Audit Trail - Electronic Signature** screen: 3. • Review the audit trail details for the most recent change. • Click a time stamp of interest to view the details for other changes that have been made to the selected signature. • Click **Download** to export the record of the single change to a PDF file. • 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.
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 open the **Software Updates** screen.

- The currently installed software versions for each software module, assay definition files, plugin, configuration package or security patch update that is installed are shown when the **Installed Software** link is selected.
- New software updates that are available to be downloaded and installed are shown when the **Software Updates** link is selected.

Security patch updates and security configuration updates for Genexus™ Software do not include any software features. These update packages are identified by fourth digit in the software version number. For example, Genexus™ Software `x.x.x.1`.

After the system update is installed, entries are logged in the installation history and audit history in Genexus™ Dx Software.

For users who have set up a Thermo Fisher™ Connect Platform account, **(Notifications)** appear 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 "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 do the following.

- 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 6.8.2.1.
- Connect the Genexus™ Dx Integrated Sequencer to the internet.

When software updates are downloaded to a computer from the Thermo Fisher App Store on Connect, the checksum information for the downloaded software updates are verified by Genexus™ Dx Software before the software package is installed.

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.

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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.

When software updates are downloaded to a USB drive from the Thermo Fisher App Store on Connect, checksum information is available in the App Store screen. Use this information to confirm that the correct software update package is downloaded to the USB drive.

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. Verify that the checksum of the downloaded software package is the same as the checksum listed on the App Store.
8. Copy the downloaded software package (ZIP file) to the prepared USB drive.
9. After completion, unmount the USB drive, then insert the drive into one of the USB ports at the back of the Genexus™ Integrated Sequencer.
10. In the menu bar of Genexus™ Dx Software, click **(Settings)** ▸ **Software Updates**, then click **Software Updates** in the upper right corner.
11. Select **USB**.
12. 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. 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. 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. 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. 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. 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. 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 • VCF • 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. 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. 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. 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. For more information, see “Change email notification settings (administrator)” on page 180.
Change the backup settings	Backup .wells files and intermediate files. 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. Set the conditions to permanently delete other files. 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. This setting overrides all other backup settings.

(continued)

Option	Description
Start backup	Click **Start Now** to start: • the backup of system data and analysis results to the selected backup directory. • if enabled, the backup of sequencing output and intermediate files to the selected backup directory. • data clean up based on cleanup settings. Backup can take a considerable amount of time when the backup is set up for first time on the system.

3. Click **Save**.

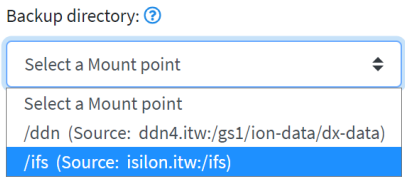
Add or change the backup directory (administrator)

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**. b. In the **Add Mount** dialog box: • Enter the name of the mount directory. For example, ***ddn***. **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. You can type a new name to create a new directory. • Enter the storage source. For example, ***ddn.itw:/gs1/ion-data/dx-data*** c. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is restarted. 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. You must add a mount point before it can be selected in the dropdown. 
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**. b. In the **Remove Mount** dialog box, select the mounted drive you want to remove. c. Click **Remove**. 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.

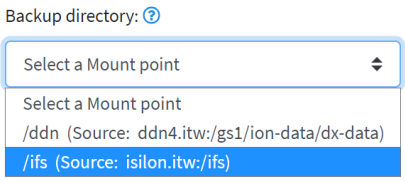
Add or change the cloud backup directory (administrator)

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**. 2. In the **Add Mount** dialog box: • Enter the name of the **Mount Point directory**. For example, ***ddn***. You can type a new name to create a new directory. • Enter the **Cloud Provider**. For example, ***AWS-S3***. 3. Enter the **Configuration** information. • **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. **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). • **Bucket Name**—The name of the AWS S3 bucket. • **Access Key Id**—A key identifier that, similar to a username, provides programmatic and CLI access to AWS resource, such as the S3 bucket • **Secret Access Key**—An access key that, similar to a password, provides programmatic and CLI access to AWS resource, such as the S3 bucket. • **Region**—The AWS region in which for the S3 bucket resides. For example, ***us-east-1***, ***us-west-1***. 4. *(Optional)* Select **Automount** to allow the system to mount backup directories automatically when the system is rebooted. When the option is unselected, you must mount a directory after a reboot. 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. You must add a mount point before for the backup directory can be selected. 
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**. 2. In the **Remove Mount** dialog box, select the mounted drive you want to remove. 3. Click **Remove**. 4. Click **Save**. 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.

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 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. 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. 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. 1. In **Delete Analysis Results**, click **Yes**. 2. In **months after completion**, set the schedule to delete the files: • 3 months • 6 months • 9 months • 12 months • 24 months – default setting • 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. The system automatically deletes .wells and intermediate files when the disk space falls below the selected threshold. 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 30.
- 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 34.

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**. 2. In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 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**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to restore, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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**. 2. In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 3. In the **Select backup folder to list runs** dialog box, navigate to the folder for a different system, then click **Select**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to import, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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. 1. In the type of runs dropdown menu, select **All runs**. 2. In the dropdown menu, select the mounted drive that contains the runs that you want to restore, then click **Browse**. 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**. 4. In the **Restore Runs** tab, select the checkbox in the row for each run that you want to restore, then click **Restore Runs**. Use the **Search By** dropdown list and the search field to search for samples by a run name or by sample tags. 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. 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. 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. • The message "No records available" indicates that no jobs are currently active. • Allows manager- and administrator-level users to stop a job. 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**.

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 223.

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 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 For more information, see “Get assay list” on page 194.	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 For more information, see “Get samples” on page 195.	Returns a list of the samples in the system.	`https://<host>/genexus/api/lims/v2/samples`
Get a list of library batches 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 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 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 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 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 For more information, see “Create samples” on page 210.	Create samples.	`https://<host>/genexus/api/lims/v2/samples`
Create a library batch 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 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 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`

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` `Accept:`application/json
Response	`Content-Type:`application/json

Response fields

Code	Status
200	Successful response
400	Bad request Error response: `{ "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". 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` String. *Optional*. Assay name. • `status` String, *Optional*. Valid values are "draft", "obsolete", or "locked".

Example response	{
	"total": 1,
	"objects": [
	{
	"id": 18,
	"assayName": "Generic_DNA_and_Fusions_EU_v0.3.0",
	"assayShortName": "Generic DNA-Fus EU v0.3.0",
	"assayDisplayedName": "Generic - DNA and Fusions - EU v0.3.0",
	"application": "DNA+RNA",
	"createdBy": "System Installed",
	"createdDate": "2021-07-14T00:05:12.000+0000",
	"isReanalysisAssay": false,
	"genomeReference": "hg19",
	"targetRegions": "Ronin_w2.7.0_v0.3.0_dnafusion_FFPE_DNA_v1.20210604.designed.bed",
	"hotspots": "Ronin_w2.7.0_dnafusion_FFPE_DNA.v1.20210608.hotspots_0.3.0.bed",
	"fusionReference": "Ronin_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.fasta",
	"fusionTargetRegions":
	"Ronin_w2.7.0_dnafusion_FFPE_RNA_fusions_imbalance_v3.6_042621_Reference_0.3.0.bed",
	"chipType": "Genexus™ Dx GX5™ Chip",
	"panelKit": "Generic Assay Panel (IVD)",
	"controlKit": "",
	"extractionKit": "",
	"libraryKit": "Genexus™ Dx Library Strips 1 and 2-HD (IVD)",
	"templatingKit": "Genexus™ Dx Templating Strips 3-GX5™ and 4 (IVD)",
	"barcodeKit": "Genexus™ Dx Barcodes 1-32 HD (IVD)",
	"sequencingKit": "Genexus™ Dx Sequencing Kit (IVD)",
	"flows": 500,
	"templatingSize": 200,
	"status": "Locked",
	"tumorCellularity": "1",
	"reanalysisAssay": false
	}
	]
	}

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` String. *Optional*. 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` String. *Required*. Library batch ID.

(continued)

library-batches	

Example response	{
	  "total": 1,
	  "objects": [
	  {
	  "libraries": [
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_05",
	  "libraryName": "NTC_DNAST_LibBatch_DNA_RNA_3",
	  "inputQuantity": 87.0,
	  "sampleId": "NTC",
	  "tumorCellularity": null,
	  "controlKitBarcode": "011019030201657517240601101234567240A51339",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_06",
	  "libraryName": "NTC_RNAST_LibBatch_DNA_RNA_3",
	  "inputQuantity": 55.3,
	  "sampleId": "NTC",
	  "tumorCellularity": null,
	  "controlKitBarcode": "011019030201657517240601101234567240A51339",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_03",
	  "libraryName": "PositiveControl01_DNAST_LibBatch_DNA_RNA_1",
	  "inputQuantity": 87.0,
	  "sampleId": "PositiveControl01",
	  "tumorCellularity": null,
	  "controlKitBarcode": "011019030201654417250930102607455240A51337",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_04",
	  "libraryName": "PositiveControl01_RNAST_LibBatch_DNA_RNA_1",
	  "inputQuantity": 55.3,
	  "sampleId": "PositiveControl01",
	  "tumorCellularity": null,
	  "controlKitBarcode": "011019030201654417250930102607455240A51338",
	  "extractionKitBarcode": null
	  },
	  {
	  "libraryType": "DNA",
	  "barcodeID": "IonDxHDdual_01",
	  "libraryName": "LIMS_Sample_55_DNAST_LibBatch_DNA_RNA_2",
	  "inputQuantity": 57.0,
	  "sampleId": "LIMS_Sample_55",
	  "tumorCellularity": null,
	  "controlKitBarcode": null,
	  "extractionKitBarcode": "91100090559101234567172110 19"
	  },
	  {
	  "libraryType": "RNA",
	  "barcodeID": "IonDxHDdual_02",
	  "libraryName": "LIMS_Sample_55_RNAST_LibBatch_DNA_RNA_2",
	  "inputQuantity": 51.3,
	  "sampleId": "LIMS_Sample_55",
	  "tumorCellularity": null,
	  "controlKitBarcode": null,
	  "extractionKitBarcode": "91100090560101234567172112 31"
	  }
	  ],
	
	  "id": 13,
	  "assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",
	  "libraryBatchId": "ST_LibBatch_DNA_RNA",
	  "libraryKitBarcode": "",
	  "panelKitBarcode": "",
	  "libraryPrepType": "DNA+RNA",
	  "ampliSeqHd": false
	  }
	  ]
	}

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` String. *Optional*. Name of the run plan. • `viewType` String. *Optional*. Valid values are "all" or "toBeProcessed".
Example response	{ "objects": [{ "id": 1, "planName": "EK_Generic_FFPE_EU_Ronin_D5_0.3.0_6.5.26", "planShortId": "EQ55D", "type": "sample", "createdBy": "System Installed", "updatedOn": "2021-07-17T20:33:48.000+0000", "noOfSamples": 8, "noOfLibraries": 16, "samples": [{ "id": 8, "sampleId": "NTC", "assayName": "Generic - DNA and Fusions - EU v0.3.0", "dnaBarcodes": ["IonDxHDdual_28"], "rnaBarcodes": ["IonDxHDdual_32"], "libraryBatchId": "1", "libraryPrepType": "DNA+RNA", "pauseForReview": false }, { "id": 5, "sampleId": "SRTestSample64", "assayName": "Generic - DNA and Fusions - EU v0.3.0", "dnaBarcodes": ["IonDxHDdual_25"], "rnaBarcodes": ["IonDxHDdual_29"], "libraryBatchId": "1", "libraryPrepType": "DNA+RNA", "pauseForReview": false },

(continued)

plans	

Example response, continued	{
	"id": 6,
	"sampleId": "SRTestSample65",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_26"
	],
	"rnaBarcodes": [
	"IonDxHDdual_30"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 2,
	"sampleId": "SRTestSample61",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_18"
	],
	"rnaBarcodes": [
	"IonDxHDdual_22"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 7,
	"sampleId": "SRTestSample66",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_27"
	],
	"rnaBarcodes": [
	"IonDxHDdual_31"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	}, {
	"id": 3,
	"sampleId": "SRTestSample62",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_19"
	],
	"rnaBarcodes": [
	"IonDxHDdual_23"
	],
	"libraryBatchId": "1",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 4,
	"sampleId": "SRTestSample63",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"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	{
	"id": 50,
	"sampleId": "SRTestSample61",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_18"
	],
	"rnaBarcodes": [
	"IonDxHDdual_22"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 53,
	"sampleId": "SRTestSample64",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_25"
	],
	"rnaBarcodes": [
	"IonDxHDdual_29"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 54,
	"sampleId": "SRTestSample65",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_26"
	],
	"rnaBarcodes": [
	"IonDxHDdual_30"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 51,
	"sampleId": "SRTestSample62",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_19"
	],
	"rnaBarcodes": [
	"IonDxHDdual_23"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	},
	{
	"id": 56,
	"sampleId": "NTC",
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"dnaBarcodes": [
	"IonDxHDdual_28"
	],
	"rnaBarcodes": [
	"IonDxHDdual_32"
	],
	"libraryBatchId": "9",
	"libraryPrepType": "DNA+RNA",
	"pauseForReview": false
	}
	],
	"runAssays": [
	"Generic Assay - DNA and Fusions - EU v0.3.0"
	]
	}
	],
	"total": 2
	}

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` String. *Optional*. Get the report template for the given assay. Use the **Assay Name**. Do not use the **Assay Full Name**.

Example response	{
	"mode": "dx",
	"reportingTemplate": [
	{
	"name": "LAB_ODxET_DNA_QC_6.8_v1.0.0",
	"id": 22,
	"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - DNA QC v1.0.0",
	"assayId": 41,
	"description": "ODxET DNA QC v1.0.0 - Lab Report",
	"locale": "en"
	},
	{
	"name": "ev_template",
	"id": 21,
	"assayName": "Oncomine™ Dx Express Test - FFPE - SW 6.8 - RS AV v0.13.0",
	"assayId": 40,
	"description": "EV_Template",
	"locale": "en"
	}
	],
	"total": 2
	}

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` String. *Optional*. Results for the selected result id. Example: ***123*** • `assay_id` String. *Optional*. Results for the assay id. Example: ***123*** • `plan_id` String. *Optional*. Results for selected run plan id. Example: ***123*** • `sampleId` String. *Optional*. Results for the selected sample name. Example: ***MySample*** • `planName` String. *Optional*. Results for selected run plan name. Example: ***MyPlan*** • `analysisComplete` String. *Optional*. Results for completed runs. Results included for failed and successfully completed runs. Example: ***true*** • `updatedBefore` String. *Optional*. Returns results updated before the selected date. Use date format: `yyyy-MM-dd HH:MM`. Example: ***2019-01-15 10:30*** • `updatedAfter` String. *Optional*. Returns results updated after the selected date. Use date format: `yyyy-MM-dd` Example: ***2019-01-15*** • `orderBy` String. *Optional*. 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` String. *Optional*. Provide the start date as yyyy-mm-dd. • `end_date` String, *Optional*. Provide the end date as yyyy-mm-dd. • `signedOff` String, *Required* A **Boolean** data type. Valid values are "true" or a "false". 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` String. *Required*. Provide the file name in comma separated format. • `path` String, *Required*. 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.
	`https://BASE_URL/download?file_list=CHECKSUM&path=AssayDev_Sample_C1_qualification_ampliseq_Genexus_pq_template516_1.3.2_3MI8Q_3MI8Q`
	2. Parse the checksum to get the file name, then request that file.
	`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`
	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. • `Date of Birth`. String. *Required*. The date of birth for the patient. • `Ordering Physician`. String. *Required*. The name of the physician who created the order for the sample. • `sampleId`. String. *Required*. Sample name. • `sampleType`. String. *Required*. Examples of valid values are "DNA" or "RNA". • `collectionDate`. String. *Required*. The date that the sample was collected. • `gender`. String. *Required*. The biological sex of the sample: Female, Male, or Unknown. • `diseaseCategory`. String. *Required*. The sample application category. • `cancerStage`. String. *Required*. Cancer stage. • `origin`. String. *Required*. Cancer type. • `notes`. String. *Required*. An open-entry field for more sample information. • `necrosis`. String. *Optional*. The percentage of cellular necrosis in the sample. • `tumorCellularity`. String. *Optional*. The percentage of tumor cells over normal cells in the sample. • `condition`. String. *Optional*. The condition of the sample.

Example request	{
	"objects":[
	{
	"sampleId":"Sample_NSCLC_99",
	"diseaseCategory":"Cancer",
	"sampleType":"DNA & RNA",
	"origin":"Non-Small Cell Lung Cancer",
	"cancerStage":"Unknown",
	"necrosis":100,
	"tumorCellularity":31,
	"gender":"Unknown",
	"collectionDate":"2018-10-02",
	"condition":"good",
	"tags":["NSCLC"],
	"notes":"LIMS API created Cancer sample and existing Tag NSCLC",
	"otherAttributes":
	{
	"Patient Id":"MRND12121",
	"Ordering Physician":"Dr Fred",
	"Date of Birth": "2000-01-01"
	}
	}
	]
	}

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>{
	"reportTemplateName": "",
	"samples":[
	{
	"assayName": "Generic Assay - DNA and Fusions - EU v0.3.0",
	"sampleId":"Test_Sample1"
	},
	{
	"assayName": "{{DNA_RNA_Assay}}",
	"sampleId":"Test_Sample2"
	},
	{
	"assayName": "{{DNA_RNA_Assay}}",
	"sampleId":"Test_Sample3"
	},
	{
	"assayName": "{{DNA_RNA_Assay}}",
	"sampleId":"Test_Sample4"
	},
	{
	"assayName": "{{DNA_RNA_Assay}}",
	"sampleId":"Test_Sample5"
	},
	{
	"assayName": "{{DNA_RNA_Assay}}",
	"sampleId":"Test_Sample6"
	}
	],
	"notes":"Nucleic Acid Run created by LIMS API",
	"planName":"NucleicAcidRun_DNF_6Samples_Run"
	}</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 *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 *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 *PTEN* deletion
RNA exon variants	Specific RNA exon variants, identified by variant ID.	*MET* exon 14 skipping *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 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.

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. This plugin generates QC metrics. For more information, see “QC results” on page 93.
molecularCoverageAnalysis	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. 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. **IMPORTANT!** To prevent erroneous sample selection during run planning, ensure that you assign a unique and distinguishable sample name for each sample. **Note:** • Samples that have been used in a run cannot be deleted. • 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. Click **Calendar** to select the date in the correct format.
Gender[1]	The biological sex of the sample: **Female**, **Male**, or **Unknown**. **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**. **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. 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. 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. **IMPORTANT!** • If this value is not set, **% Cellularity** is assumed to be 100% in calculations that use this attribute. • **% Cellularity** is a required attribute for CNV analyses. Do not leave the field blank. • 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**

Troubleshooting

- Download Instrument Diagnostics 222
- Customer Support Archive (CSA) 223
- Troubleshoot Genexus™ Dx Software 223

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 *Genexus™ Dx Integrated Sequencer 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

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

- Obtaining Certificates of Analysis 227
- Obtaining Certificates of Conformance 227
- Serious incidents 227
- Cybersecurity incidents 227

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.

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**.

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.

Sample		Patient	
Sample Name:	**FFPE01**	Patient Id:	**PID01**
Cancer Type:	**Non-Small Cell Lung Cancer**	Gender:	**Male**
Cancer Stage:	**Stage III**	Date of Birth:	**30 MAR 1975**
Sample Type:	**FFPE**	Collection Date:	**22 FEB 2021**
%Cellularity:	**100**	Ordering Physician:	**Dr.Q**
%Necrosis:		Created On:	**06 NOV 2025 14:24**

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
EGFR	EGFR exon 20 insertion	p.?	c.2284-6_2284-5ins TCCAGGAAGCCT	POSITIVE	COSM26720	ins
KRAS	KRAS G12C	p.G12C	c.34_36delGGTi nsTGC	POSITIVE	COSM513	mnp

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
SQSTM1-NTRK3 fusion	SQSTM1(5) - NTRK3(14)	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
AKT1	AKT1 E17K	p.E17K	c.49G>A	POSITIVE	COSM33765	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
AR-V7	AR(3) - AR(4)	RNAExonVariant	POSITIVE

ⓘ 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 therapies 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)
NSCLC	ERBB2	ERBB2/HER2 activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions)	HYRNUO® (sevabertinib)

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 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 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 >70% necrotic tissue may yield unreliable sequencing results for the detection of *EGFR* exon 20 insertion and *ERBB2* 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).

30. For *MET* Exon 14 skipping results for cancer types other than non-small cell lung cancer, confirmatory testing using a clinically validated assay is recommended.

31. For NSCLC, the Oncomine™ Dx Express Test assay definition file includes prevalent but not all rare or newly identified *EGFR* exon 20 insertions, *ERBB2/HER2* exon 20 insertions, and *ERBB2/HER2* SNVs in exons 18-21 within the tyrosine kinase domain. The Oncomine™ Dx Express Test may miss rare or newly identified:

- *EGFR* exon 20 insertions carried by a subset of patients who may derive benefit from sunvozertinib
- *ERBB2/HER2* activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions) carried by a subset of patients who may derive benefit from sevabertinib

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 C77F	p.C77F	COSM5027916	NEGATIVE	SNV
AKT1 L52R	p.L52R	COSM93893	NEGATIVE	SNV
AKT1 P51L	p.P51L	COSM4468165	NEGATIVE	SNV
AKT1 P51S	p.P51S	COSM5991558	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AKT1 P70_C77dup	p.P70_C77dup	COSM6568358	NEGATIVE	ins
AKT1 Q79K	p.Q79K	COSM159008	NEGATIVE	SNV
AKT1 R76_C77insWPNTFIIR	p.R76_C77insWPNTFIIR	COSM5045024	NEGATIVE	ins
AKT2 D324G	p.D324G	COSM474741	NEGATIVE	SNV
AKT2 D324H	p.D324H	COSM474742	NEGATIVE	SNV
AKT2 E17K	p.E17K	COSM159009	NEGATIVE	SNV
AKT3 E17K	p.E17K	COSM224779	NEGATIVE	SNV
AKT3 L51R	p.L51R	COSM309035	NEGATIVE	SNV
AKT3 Q78K	p.Q78K	COSM242892	NEGATIVE	SNV
ALK A1200V	p.A1200V	COSM317003	NEGATIVE	SNV
ALK A1280V	p.A1280V	COSM6914238	NEGATIVE	SNV
ALK C1156F	p.C1156F	BT144	NEGATIVE	SNV
ALK C1156T	p.C1156T	OM3350	NEGATIVE	mnp
ALK C1156Y	p.C1156Y	COSM99136	NEGATIVE	SNV
ALK D1203A	p.D1203A	OM3313	NEGATIVE	SNV
ALK D1203N	p.D1203N	COSM6191344	NEGATIVE	SNV
ALK E1129V	p.E1129V	BT117	NEGATIVE	SNV
ALK E1154Q	p.E1154Q	OM3324	NEGATIVE	SNV
ALK E1210K	p.E1210K	OM3161	NEGATIVE	SNV
ALK F1174C	p.F1174C	COSM28059	NEGATIVE	SNV
ALK F1174I	p.F1174I	COSM28491	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28055	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28061	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK F1174L	p.F1174L	COSM28057	NEGATIVE	SNV
ALK F1174S	p.F1174S	COSM53063	NEGATIVE	SNV
ALK F1174V	p.F1174V	COSM28054	NEGATIVE	SNV
ALK F1245C	p.F1245C	COSM28500	NEGATIVE	SNV
ALK F1245I	p.F1245I	COSM28492	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28062	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28493	NEGATIVE	SNV
ALK F1245V	p.F1245V	COSM28499	NEGATIVE	SNV
ALK G1123D	p.G1123D	OM3262	NEGATIVE	SNV
ALK G1123S	p.G1123S	COSM5045108	NEGATIVE	SNV
ALK G1128A	p.G1128A	COSM98475	NEGATIVE	SNV
ALK G1202R	p.G1202R	COSM144250	NEGATIVE	SNV
ALK G1202del	p.G1202del	OMINDEL698	NEGATIVE	del
ALK G1269A	p.G1269A	COSM1169707	NEGATIVE	SNV
ALK I1171H	p.I1171H	OM3163	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM1169448	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM28498	NEGATIVE	SNV
ALK I1171S	p.I1171S	COSM4440758	NEGATIVE	SNV
ALK I1171T	p.I1171T	COSM4381100	NEGATIVE	SNV
ALK I1268V	p.I1268V	BT149	NEGATIVE	SNV
ALK L1152P	p.L1152P	COSM1407659	NEGATIVE	SNV
ALK L1152R	p.L1152R	COSM97185	NEGATIVE	SNV
ALK L1152V	p.L1152V	OM3513	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK L1196M	p.L1196M	COSM99137	NEGATIVE	SNV
ALK L1196Q	p.L1196Q	COSM1169447	NEGATIVE	SNV
ALK L1198F	p.L1198F	COSM97028	NEGATIVE	SNV
ALK L1198P	p.L1198P	OM3162	NEGATIVE	SNV
ALK L1198V	p.L1198V	OM3314	NEGATIVE	SNV
ALK R1192P	p.R1192P	OM3259	NEGATIVE	SNV
ALK R1192Q	p.R1192Q	OM3372	NEGATIVE	SNV
ALK R1209Q	p.R1209Q	COSM327056	NEGATIVE	SNV
ALK R1231Q	p.R1231Q	COSM5967491	NEGATIVE	SNV
ALK R1231W	p.R1231W	BT148	NEGATIVE	SNV
ALK R1275L	p.R1275L	COSM28060	NEGATIVE	SNV
ALK R1275Q	p.R1275Q	COSM28056	NEGATIVE	SNV
ALK S1157F	p.S1157F	BT145	NEGATIVE	SNV
ALK S1206C	p.S1206C	BT147	NEGATIVE	SNV
ALK S1206F	p.S1206F	COSM6909200	NEGATIVE	SNV
ALK S1206Y	p.S1206Y	COSM144251	NEGATIVE	SNV
ALK T1151K	p.T1151K	OM3261	NEGATIVE	SNV
ALK T1151M	p.T1151M	COSM98478	NEGATIVE	SNV
ALK T1151R	p.T1151R	OM3260	NEGATIVE	SNV
ALK T1151dup	p.T1151dup	COSM144252	NEGATIVE	ins
ALK V1180L	p.V1180L	COSM4381101	NEGATIVE	SNV
ALK Y1278S	p.Y1278S	COSM28058	NEGATIVE	SNV
AR D880E	p.D880E	COSM5945750	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AR D891H	p.D891H	COSM5945767	NEGATIVE	SNV
AR E894K	p.E894K	COSM5945748	NEGATIVE	SNV
AR E898G	p.E898G	OM3322	NEGATIVE	SNV
AR F814V	p.F814V	COSM150793	NEGATIVE	SNV
AR F877L	p.F877L	COSM5945752	NEGATIVE	SNV
AR F877L	p.F877L	COSM5946164	NEGATIVE	SNV
AR H875Q	p.H875Q	COSM5945766	NEGATIVE	SNV
AR H875Y	p.H875Y	COSM238555	NEGATIVE	SNV
AR L702H	p.L702H	COSM238553	NEGATIVE	SNV
AR L882I	p.L882I	COSM5945751	NEGATIVE	SNV
AR M896T	p.M896T	OM3321	NEGATIVE	SNV
AR M896V	p.M896V	COSM5945749	NEGATIVE	SNV
AR S889G	p.S889G	COSM5945753	NEGATIVE	SNV
AR T878A	p.T878A	COSM236693	NEGATIVE	SNV
AR T878S	p.T878S	COSM5570418	NEGATIVE	SNV
AR V716M	p.V716M	COSM3094417	NEGATIVE	SNV
AR V731M	p.V731M	COSM1468984	NEGATIVE	SNV
AR W742C	p.W742C	COSM5572266	NEGATIVE	SNV
AR W742C	p.W742C	COSM5944171	NEGATIVE	SNV
AR W742L	p.W742L	COSM5944172	NEGATIVE	SNV
ARAF S214A	p.S214A	COSM1742787	NEGATIVE	SNV
ARAF S214C	p.S214C	COSM5044705	NEGATIVE	SNV
ARAF S214F	p.S214F	COSM612884	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ARAF S214T	p.S214T	COSM1558277	NEGATIVE	SNV
ARAF S214Y	p.S214Y	COSM5416443	NEGATIVE	SNV
BRAF A598T	p.A598T	COSM28505	NEGATIVE	SNV
BRAF A598V	p.A598V	COSM21549	NEGATIVE	SNV
BRAF D594A	p.D594A	COSM1583010	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM1121	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM253330	NEGATIVE	SNV
BRAF D594G	p.D594G	COSM467	NEGATIVE	SNV
BRAF D594H	p.D594H	COSM144576	NEGATIVE	SNV
BRAF D594N	p.D594N	COSM27639	NEGATIVE	SNV
BRAF D594V	p.D594V	COSM466	NEGATIVE	SNV
BRAF D594Y	p.D594Y	COSM6928839	NEGATIVE	SNV
BRAF D594_T599dup	p.D594_T599dup	COSM26504	NEGATIVE	ins
BRAF E586K	p.E586K	COSM463	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM468	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM53198	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM21612	NEGATIVE	SNV
BRAF F595S	p.F595S	COSM1123	NEGATIVE	SNV
BRAF G464E	p.G464E	COSM449	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1111	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1448615	NEGATIVE	SNV
BRAF G464V	p.G464V	COSM450	NEGATIVE	SNV
BRAF G466A	p.G466A	COSM452	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF G466E	p.G466E	COSM453	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM1112	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM253328	NEGATIVE	SNV
BRAF G466V	p.G466V	COSM451	NEGATIVE	SNV
BRAF G469A	p.G469A	COSM460	NEGATIVE	SNV
BRAF G469E	p.G469E	COSM461	NEGATIVE	SNV
BRAF G469L	p.G469L	COSM6056848	NEGATIVE	mnp
BRAF G469R	p.G469R	COSM455	NEGATIVE	SNV
BRAF G469R	p.G469R	COSM457	NEGATIVE	SNV
BRAF G469S	p.G469S	COSM1113	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM27912	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM458	NEGATIVE	mnp
BRAF G469V	p.G469V	COSM459	NEGATIVE	SNV
BRAF G596C	p.G596C	COSM6936824	NEGATIVE	SNV
BRAF G596D	p.G596D	COSM26506	NEGATIVE	SNV
BRAF G596R	p.G596R	COSM469	NEGATIVE	SNV
BRAF G596V	p.G596V	COSM9145962	NEGATIVE	SNV
BRAF I463S	p.I463S	COSM448	NEGATIVE	SNV
BRAF K601E	p.K601E	COSM478	NEGATIVE	SNV
BRAF K601I	p.K601I	COSM26491	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM1132	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM6265	NEGATIVE	SNV
BRAF K601R	p.K601R	COSM1568586	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF K601T	p.K601T	COSM3878760	NEGATIVE	SNV
BRAF L597P	p.L597P	COSM1448593	NEGATIVE	SNV
BRAF L597Q	p.L597Q	COSM1125	NEGATIVE	SNV
BRAF L597R	p.L597R	COSM471	NEGATIVE	SNV
BRAF L597S	p.L597S	COSM1126	NEGATIVE	mnp
BRAF L597V	p.L597V	COSM470	NEGATIVE	SNV
BRAF N581I	p.N581I	COSM26460	NEGATIVE	SNV
BRAF N581S	p.N581S	COSM462	NEGATIVE	SNV
BRAF N581T	p.N581T	COSM1699452	NEGATIVE	SNV
BRAF R462I	p.R462I	COSM447	NEGATIVE	SNV
BRAF T599I	p.T599I	COSM472	NEGATIVE	SNV
BRAF T599R	p.T599R	COSM4172020	NEGATIVE	SNV
BRAF T599_V600insEAT	p.T599_V600insEAT	COSM4166148	NEGATIVE	ins
BRAF T599_V600insETT	p.T599_V600insETT	COSM4172018	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM144982	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM30730	NEGATIVE	ins
BRAF V600A	p.V600A	COSM18443	NEGATIVE	SNV
BRAF V600D	p.V600D	COSM308550	NEGATIVE	mnp
BRAF V600D	p.V600D	COSM477	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM475	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM476	NEGATIVE	SNV
BRAF V600G	p.V600G	COSM6137	NEGATIVE	SNV
BRAF V600K	p.V600K	COSM473	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF V600L	p.V600L	COSM219798	NEGATIVE	SNV
BRAF V600L	p.V600L	COSM33808	NEGATIVE	SNV
BRAF V600M	p.V600M	COSM1130	NEGATIVE	SNV
BRAF V600Q	p.V600Q	COSM249889	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM1583011	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM474	NEGATIVE	mnp
BRAF V600_K601delinsE	p.V600_K601delinsE	COSM1133	NEGATIVE	del
BRAF Y472C	p.Y472C	COSM1133046	NEGATIVE	SNV
BRAF [G593=;D594N]	p.[G593=;D594N]	COSM211600	NEGATIVE	mnp
BRAF [T599=;V600R]	p.[T599=;V600R]	COSM1127	NEGATIVE	mnp
BRAF [T599I;V600K]	p.[T599I;V600K]	COSM1735763	NEGATIVE	mnp
BRAF [T599R;V600G]	p.[T599R;V600G]	OM3547	NEGATIVE	mnp
CDK4 K22M	p.K22M	COSM3463915	NEGATIVE	SNV
CDK4 K22Q	p.K22Q	OM3153	NEGATIVE	SNV
CDK4 K22R	p.K22R	COSM232013	NEGATIVE	SNV
CDK4 R24C	p.R24C	COSM1677139	NEGATIVE	SNV
CDK4 R24H	p.R24H	COSM1989836	NEGATIVE	SNV
CDK4 R24L	p.R24L	COSM363684	NEGATIVE	SNV
CDK4 R24S	p.R24S	COSM3463914	NEGATIVE	SNV
CHEK2 K373E	p.K373E	COSM42871	NEGATIVE	SNV
CHEK2 M381V	p.M381V	COSM1559601	NEGATIVE	SNV
CHEK2 R523H	p.R523H	COSM1559596	NEGATIVE	SNV
CHEK2 R523P	p.R523P	COSM1559595	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CHEK2 [S372=;K373E]	p.[S372=;K373E]	COSM1294063	NEGATIVE	mnp
CTNNB1 D32A	p.D32A	COSM5690	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM22939	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM5766	NEGATIVE	SNV
CTNNB1 D32G	p.D32G	COSM5681	NEGATIVE	SNV
CTNNB1 D32H	p.D32H	COSM5668	NEGATIVE	SNV
CTNNB1 D32N	p.D32N	COSM5672	NEGATIVE	SNV
CTNNB1 D32V	p.D32V	COSM5691	NEGATIVE	SNV
CTNNB1 D32Y	p.D32Y	COSM5661	NEGATIVE	SNV
CTNNB1 G34A	p.G34A	COSM5988492	NEGATIVE	SNV
CTNNB1 G34E	p.G34E	COSM5671	NEGATIVE	SNV
CTNNB1 G34I	p.G34I	COSM1666841	NEGATIVE	mnp
CTNNB1 G34R	p.G34R	COSM5684	NEGATIVE	SNV
CTNNB1 G34R	p.G34R	COSM5686	NEGATIVE	SNV
CTNNB1 G34V	p.G34V	COSM5670	NEGATIVE	SNV
CTNNB1 H36P	p.H36P	COSM5678	NEGATIVE	SNV
CTNNB1 H36R	p.H36R	COSM27378	NEGATIVE	SNV
CTNNB1 H36Y	p.H36Y	COSM5703	NEGATIVE	SNV
CTNNB1 I35K	p.I35K	COSM28727	NEGATIVE	mnp
CTNNB1 I35M	p.I35M	COSM28711	NEGATIVE	SNV
CTNNB1 I35N	p.I35N	COSM5695	NEGATIVE	SNV
CTNNB1 I35S	p.I35S	COSM5674	NEGATIVE	SNV
CTNNB1 I35T	p.I35T	COSM13168	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 I35V	p.I35V	COSM1566774	NEGATIVE	SNV
CTNNB1 K335I	p.K335I	COSM17797	NEGATIVE	SNV
CTNNB1 K335Q	p.K335Q	COSM4421324	NEGATIVE	SNV
CTNNB1 K335R	p.K335R	COSM5945941	NEGATIVE	SNV
CTNNB1 K335T	p.K335T	COSM1725761	NEGATIVE	SNV
CTNNB1 L46_S47del	p.L46_S47del	COSM6129	NEGATIVE	del
CTNNB1 L46_S47del	p.L46_S47del	COSM6132	NEGATIVE	del
CTNNB1 P44A	p.P44A	COSM17661	NEGATIVE	SNV
CTNNB1 P44L	p.P44L	COSM5761	NEGATIVE	SNV
CTNNB1 P44S	p.P44S	COSM5704	NEGATIVE	SNV
CTNNB1 S33A	p.S33A	COSM5683	NEGATIVE	SNV
CTNNB1 S33C	p.S33C	COSM5677	NEGATIVE	SNV
CTNNB1 S33F	p.S33F	COSM5669	NEGATIVE	SNV
CTNNB1 S33L	p.S33L	COSM6098	NEGATIVE	mnp
CTNNB1 S33N	p.S33N	COSM6099	NEGATIVE	mnp
CTNNB1 S33P	p.S33P	COSM5682	NEGATIVE	SNV
CTNNB1 S33T	p.S33T	COSM27311	NEGATIVE	SNV
CTNNB1 S33Y	p.S33Y	COSM5673	NEGATIVE	SNV
CTNNB1 S37A	p.S37A	COSM5675	NEGATIVE	SNV
CTNNB1 S37C	p.S37C	COSM5679	NEGATIVE	SNV
CTNNB1 S37F	p.S37F	COSM5662	NEGATIVE	SNV
CTNNB1 S37P	p.S37P	COSM5687	NEGATIVE	SNV
CTNNB1 S37T	p.S37T	COSM5729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 S37Y	p.S37Y	COSM5666	NEGATIVE	SNV
CTNNB1 S45A	p.S45A	COSM5685	NEGATIVE	SNV
CTNNB1 S45C	p.S45C	COSM5689	NEGATIVE	SNV
CTNNB1 S45F	p.S45F	COSM5667	NEGATIVE	SNV
CTNNB1 S45P	p.S45P	COSM5663	NEGATIVE	SNV
CTNNB1 S45T	p.S45T	COSM5719	NEGATIVE	SNV
CTNNB1 S45Y	p.S45Y	COSM5692	NEGATIVE	SNV
CTNNB1 S45_G48del	p.S45_G48del	COSM5992	NEGATIVE	del
CTNNB1 S45_S47del	p.S45_S47del	COSM1617582	NEGATIVE	del
CTNNB1 S45_S47delinsC	p.S45_S47delinsC	COSM28708	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6128	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6131	NEGATIVE	del
CTNNB1 T41A	p.T41A	COSM5664	NEGATIVE	SNV
CTNNB1 T41I	p.T41I	COSM5676	NEGATIVE	SNV
CTNNB1 T41N	p.T41N	COSM5730	NEGATIVE	SNV
CTNNB1 T41P	p.T41P	COSM5688	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5716	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5701	NEGATIVE	SNV
CTNNB1 [S45E;L46=]	p.[S45E;L46=]	COSM86011	NEGATIVE	mnp
EGFR A289D	p.A289D	COSM21685	NEGATIVE	SNV
EGFR A289T	p.A289T	COSM21686	NEGATIVE	SNV
EGFR A289V	p.A289V	COSM21687	NEGATIVE	SNV
EGFR A750P	p.A750P	COSM6219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR C775Y	p.C775Y	COSM22954	NEGATIVE	SNV
EGFR C797G	p.C797G	COSM6493936	NEGATIVE	SNV
EGFR C797N	p.C797N	OM3543	NEGATIVE	mnp
EGFR C797S	p.C797S	COSM6493937	NEGATIVE	SNV
EGFR C797S	p.C797S	COSM5945664	NEGATIVE	SNV
EGFR E709A	p.E709A	COSM13427	NEGATIVE	SNV
EGFR E709G	p.E709G	COSM13009	NEGATIVE	SNV
EGFR E709H	p.E709H	COSM12428	NEGATIVE	mnp
EGFR E709K	p.E709K	COSM12988	NEGATIVE	SNV
EGFR E709Q	p.E709Q	COSM116882	NEGATIVE	SNV
EGFR E709V	p.E709V	COSM12371	NEGATIVE	SNV
EGFR E709_T710delinsA	p.E709_T710delinsA	COSM85796	NEGATIVE	del
EGFR E709_T710delinsD	p.E709_T710delinsD	COSM51525	NEGATIVE	del
EGFR E709_T710delinsG	p.E709_T710delinsG	COSM48981	NEGATIVE	complex
EGFR E746G	p.E746G	COSM6849765	NEGATIVE	SNV
EGFR G465E	p.G465E	BT129	NEGATIVE	SNV
EGFR G465R	p.G465R	COSM601581	NEGATIVE	SNV
EGFR G598A	p.G598A	COSM3412196	NEGATIVE	SNV
EGFR G598V	p.G598V	COSM21690	NEGATIVE	SNV
EGFR G719A	p.G719A	COSM6239	NEGATIVE	SNV
EGFR G719C	p.G719C	COSM6253	NEGATIVE	SNV
EGFR G719D	p.G719D	COSM18425	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR G719S	p.G719S	COSM6252	NEGATIVE	SNV
EGFR G721D	p.G721D	COSM22993	NEGATIVE	SNV
EGFR G721R	p.G721R	OM3325	NEGATIVE	SNV
EGFR G721S	p.G721S	COSM22992	NEGATIVE	SNV
EGFR G724S	p.G724S	COSM13979	NEGATIVE	SNV
EGFR G796A	p.G796A	COSM27568	NEGATIVE	SNV
EGFR G796C	p.G796C	OM3318	NEGATIVE	SNV
EGFR G796R	p.G796R	COSM6493935	NEGATIVE	SNV
EGFR G796S	p.G796S	COSM20891	NEGATIVE	SNV
EGFR H835L	p.H835L	COSM6227	NEGATIVE	SNV
EGFR I491M	p.I491M	BT135	NEGATIVE	SNV
EGFR I744T	p.I744T	COSM87245	NEGATIVE	SNV
EGFR K467T	p.K467T	BT132	NEGATIVE	SNV
EGFR K757R	p.K757R	COSM133588	NEGATIVE	SNV
EGFR L718M	p.L718M	COSM1716255	NEGATIVE	SNV
EGFR L718Q	p.L718Q	COSM6503269	NEGATIVE	SNV
EGFR L718V	p.L718V	OM3316	NEGATIVE	SNV
EGFR L747P	p.L747P	COSM24267	NEGATIVE	mnp
EGFR L747S	p.L747S	COSM26704	NEGATIVE	SNV
EGFR L747V	p.L747V	OM3537	NEGATIVE	SNV
EGFR L792F	p.L792F	COSM6493933	NEGATIVE	SNV
EGFR L792H	p.L792H	COSM6493934	NEGATIVE	SNV
EGFR L792P	p.L792P	COSM13190	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR L792R	p.L792R	OM3267	NEGATIVE	SNV
EGFR L792V	p.L792V	OM3265	NEGATIVE	SNV
EGFR L792Y	p.L792Y	OM3266	NEGATIVE	mnp
EGFR L798I	p.L798I	OM3268	NEGATIVE	SNV
EGFR L833F	p.L833F	COSM219794	NEGATIVE	SNV
EGFR L833V	p.L833V	COSM13424	NEGATIVE	SNV
EGFR L833W	p.L833W	COSM133204	NEGATIVE	SNV
EGFR L858K	p.L858K	COSM24268	NEGATIVE	mnp
EGFR L858M	p.L858M	COSM12366	NEGATIVE	SNV
EGFR L858Q	p.L858Q	COSM29578	NEGATIVE	SNV
EGFR L858R	p.L858R	COSM13553	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM12429	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM133630	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM6224	NEGATIVE	SNV
EGFR L861Q	p.L861Q	COSM6213	NEGATIVE	SNV
EGFR L861R	p.L861R	COSM12374	NEGATIVE	SNV
EGFR L861V	p.L861V	COSM24848	NEGATIVE	SNV
EGFR N771T	p.N771T	OM3174	NEGATIVE	SNV
EGFR P848L	p.P848L	COSM22943	NEGATIVE	SNV
EGFR R108G	p.R108G	COSM1451536	NEGATIVE	SNV
EGFR R108K	p.R108K	COSM21683	NEGATIVE	SNV
EGFR R451C	p.R451C	COSM3266583	NEGATIVE	SNV
EGFR R836C	p.R836C	COSM28604	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR S464L	p.S464L	BT126	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236671	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236670	NEGATIVE	SNV
EGFR S720I	p.S720I	OM3317	NEGATIVE	mnp
EGFR S720P	p.S720P	COSM88729	NEGATIVE	SNV
EGFR S768G	p.S768G	COSM1451595	NEGATIVE	SNV
EGFR S768I	p.S768I	COSM6241	NEGATIVE	SNV
EGFR S768N	p.S768N	COSM12989	NEGATIVE	SNV
EGFR S768T	p.S768T	COSM291998	NEGATIVE	SNV
EGFR T790A	p.T790A	COSM1272071	NEGATIVE	SNV
EGFR T790M	p.T790M	COSM6240	NEGATIVE	SNV
EGFR T854A	p.T854A	COSM28537	NEGATIVE	SNV
EGFR T854I	p.T854I	COSM87246	NEGATIVE	SNV
EGFR T854S	p.T854S	COSM1235378	NEGATIVE	SNV
EGFR V292L	p.V292L	COSM43067	NEGATIVE	SNV
EGFR V769L	p.V769L	COSM6242	NEGATIVE	SNV
EGFR V769M	p.V769M	COSM13425	NEGATIVE	SNV
EGFR [H773L;V774M]	p.[H773L;V774M]	COSM6978402	NEGATIVE	mnp
EGFR [K745=;E746I;L747P]	p.[K745=;E746I;L747P]	OM3355	NEGATIVE	mnp
EGFR [L718=;G719C]	p.[L718=;G719C]	COSM18441	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM6984779	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM85750	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	OMINDEL1181	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR [V769G;D770I]	p.[V769G;D770I]	OM3540	NEGATIVE	mnp
EGFR exon 19 deletion	p.I744_E749delinsLKR	COSM133186	NEGATIVE	complex
EGFR exon 19 deletion	p.I744_A750delinsVK	COSM85798	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750del	COSM221565	NEGATIVE	del
EGFR exon 19 deletion	p.K745_E749del	COSM26038	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM3734668	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750delinsT	COSM1190791	NEGATIVE	del
EGFR exon 19 deletion	p.E746del	COSM18420	NEGATIVE	del
EGFR exon 19 deletion	p.E746_E749del	COSM28517	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751del	COSM24869	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.E746_T751del	COSM12728	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752del	COSM133189	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsA	COSM13549	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM13550	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM13551	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM13552	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_E749del	COSM6218	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM6225	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsFPS	COSM6506513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsA	COSM12367	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsRP	COSM12413	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsA	COSM12678	NEGATIVE	del
EGFR exon 19 deletion	p.E746_P753delinsLS	COSM13200	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133188	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133190	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsIS	COSM133191	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsQP	COSM13557	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsQ	COSM22999	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_K754del	COSM24970	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsS	COSM26513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM26680	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsL	COSM51502	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM51526	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_A750delinsP	COSM6947327	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM6966471	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsM	OMINDEL716	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751del	COSM12369	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752delinsV	COSM12384	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM12386	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM12416	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM133193	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsAP	COSM144207	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM18421	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.L747_P753del	COSM255211	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsVP	COSM28623	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsVQ	COSM51524	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM52935	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM53205	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsD	COSM6220	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_S752delinsV	COSM674057	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_T751delinsVS	COSM6980200	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750del	COSM9179903	NEGATIVE	del
EGFR exon 19 deletion	p.L747_T751delinsQ	COSM12419	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQH	COSM12421	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12422	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsDP	COSM18428	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM22944	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQ	COSM26441	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.E746_A750delinsD	COSM6974307	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM1235296	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_P753delinsS	COSM12370	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12382	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM12383	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM12387	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_S752delinsQH	COSM133196	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_A750delinsP	COSM4170220	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsS	COSM4170221	NEGATIVE	del
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM51501	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsN	COSM51503	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsS	COSM6210	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A755delinsAN	COSM85891	NEGATIVE	complex
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_E758delinsP	COSM26440	NEGATIVE	complex

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.T751_E758del	COSM133207	NEGATIVE	del
EGFR exon 19 deletion	p.A750_I759delinsGS	COSM5023005	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM1667024	NEGATIVE	complex
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.S752_I759del	COSM13556	NEGATIVE	del
EGFR exon 19 deletion	p.T751_I759delinsS	COSM1667027	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsREA	COSM22956	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM24270	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM96856	NEGATIVE	complex
EGFR exon 19 deletion	p.S752_I759del	COSM6256	NEGATIVE	del
EGFR exon 19 deletion	p.P753_I759del	COSM24269	NEGATIVE	del
EGFR exon 19 deletion	p.K754_I759del	COSM5271797	NEGATIVE	del
EGFR exon 19 insertion	p.I740_K745dup	COSM12423	NEGATIVE	ins
EGFR exon 19 insertion	p.I740_K745dup	COSM51504	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insVPVAIK	COSM26444	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insTPVAIK	COSM255152	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.?	OMINDEL1050	NEGATIVE	ins
EGFR exon 20 insertion	p.?	COSM26720	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_D770dup	COSM6962256	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_V765insHH	OMINDEL1051	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM12376	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_S768insTLA	COSM12425	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM20884	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVASV	OMINDEL1800	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.S768_P772dup	OMINDEL1053	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSSV	OMINDEL1054	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insTSV	OMINDEL1055	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.S768_V769insLDS	OMINDEL1802	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insRSV	OMINDEL952	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.S768_N771dup	S768_N771dup	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.V769_D770insCV	COSM12379	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRVDNP	OMINDEL1056	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insANV	COSM1651741	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	COSM18430	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_N771dup	COSM20885	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_P772dup	OMINDEL1057	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_V769delinsISW	OMINDEL1058	NEGATIVE	complex
EGFR exon 20 insertion	p.S768delinsRRGH	OMINDEL1059	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTV	OMINDEL1060	NEGATIVE	ins
EGFR exon 20 insertion	p.V769dup	COSM6506514	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	OMINDEL1187	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	OMINDEL1803	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTM	OMINDEL1061	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1062	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1063	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1064	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1065	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1066	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM6845099	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM85795	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGG	MATNV07	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_D770insGE	OMINDEL1069	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGQ	OMINDEL1070	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1071	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.V769_D770insERG	COSM1651742	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM1651745	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM22955	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insMASVD	COSM28638	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	MATNV03	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771insGTD	OMINDEL1074	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1075	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insP	OMINDEL1076	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	OMINDEL1804	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771dup	OMINDEL1805	NEGATIVE	ins
EGFR exon 20 insertion	p.D770dup	OMINDEL1806	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insY	COSM1238030	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	COSM12381	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGY	COSM12427	NEGATIVE	ins
EGFR exon 20 insertion	p.N771dup	COSM13003	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGTH	COSM6983510	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGNPH	D770_N771insGNPH	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_1	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_2	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.D770delins
ASVH

OMINDEL108
0

NEGATIVE

ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771insH	OMINDEL1081	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.N771_P772insHN	OMINDEL1084	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.D770_N771insH	OMINDEL1807	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	OMINDEL1808	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1809	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsAGG	COSM12737	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insG	COSM13004	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM13558	NEGATIVE	complex
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_N771insT	COSM5023008	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insGNPH	COSM51544	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	COSM6977296	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGV	D770delinsGV	NEGATIVE	complex

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.D770_N771delinsAGH	MATNV09	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insPNP	OMINDEL1087	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1088	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsGGT	OMINDEL1090	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsEF	OMINDEL1091	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsEG	OMINDEL1092	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
EGFR exon 20 insertion	p.D770_N771insP	OMINDEL1096	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1097	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insANP	OMINDEL1098	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_H773insNPNP	OMINDEL1101	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1102	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1103	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1104	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1105	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1106	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1107	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1108	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1109	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insYPNP	OMINDEL1110	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772delinsASVDNR	OMINDEL1801	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsAS	OMINDEL1810	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insVDSVDNP	OMINDEL953	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	COSM12378	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGT	COSM1238029	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insSVE	COSM1651743	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insAPW	COSM20886	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_N771insSVE	D770_N771insSVE	NEGATIVE	ins
EGFR exon 20	p.D770_N771insG	MATNV05	NEGATIVE	ins

insertion

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771delinsERG	MATNV06	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1111	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1112	NEGATIVE	ins
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.P772_H773insTRNP	OMINDEL1116	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_N771insF	OMINDEL1812	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRNP	OMINDEL1813	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	COSM12380	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insH	COSM1238031	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSGH	COSM1651744	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insRH	COSM166390	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsGF	COSM18431	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsTH	COSM22946	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSH	COSM24434	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insTHP	COSM48923	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsVH	COSM5023007	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGY	COSM53189	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSTH	COSM6920147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insHH	COSM6931207	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsPH	MATNV04	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsHH	OMINDEL1118	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGD	OMINDEL1119	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1120	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1121	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insHPH	OMINDEL1122	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1123	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1124	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1125	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1126	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1127	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1128	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1129	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1130	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSPH	OMINDEL1131	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1132	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1133	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1134	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1135	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.N771delinsKPH	OMINDEL1188	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsCH	OMINDEL1814	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSQRGH	OMINDEL1815	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsHHR	OMINDEL1816	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGC	OMINDEL904	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL954	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insPHPH	p.H773_V774insPHPH_2	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsSVDNR	COSM13554	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsKL	COSM6438147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insV	COSM6922328	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insT	OMINDEL1138	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1139	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSVDS	OMINDEL1140	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1141	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1142	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insG	OMINDEL1143	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1144	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1145	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1146	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1183	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPP	OMINDEL955	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_C775dup	OMINDEL956	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_V774dup	COSM6845098	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1147	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_P772insT	OMINDEL1148	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1149	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1150	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.H773_V774insTH	OMINDEL1151	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insT	OMINDEL1152	NEGATIVE	ins
EGFR exon 20 insertion	p.H773dup	COSM12377	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774dup	COSM12389	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insNH	COSM5023006	NEGATIVE	ins
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_H773insL	OMINDEL1155	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insR	OMINDEL1156	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTP	COSM12388	NEGATIVE	complex
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.H773_V774insPHPH	H773_V774insPHPH	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.H773_V774insGH	OMINDEL1157	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1158	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1159	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	OMINDEL1184	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1811	NEGATIVE	complex
EGFR exon 20 insertion	p.H773delinsPNPY	COSM1735761	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsRY	MATNV08	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	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.H773delinsPNPY	OMINDEL1165	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1166	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1167	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insY	OMINDEL1168	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insTQPP	COSM3727813	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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
ERBB2 C311R	p.C311R	COSM35763	NEGATIVE	SNV
ERBB2 D769H	p.D769H	COSM13170	NEGATIVE	SNV
ERBB2 D769N	p.D769N	COSM1302747	NEGATIVE	SNV
ERBB2 D769Y	p.D769Y	COSM1251412	NEGATIVE	SNV
ERBB2 E321G	p.E321G	COSM35764	NEGATIVE	SNV
ERBB2 G309A	p.G309A	COSM436496	NEGATIVE	SNV
ERBB2 G309E	p.G309E	COSM94224	NEGATIVE	SNV
ERBB2 G660D	p.G660D	COSM4681497	NEGATIVE	SNV
ERBB2 G776A	p.G776A	OBENV03	NEGATIVE	SNV
ERBB2 G776C	p.G776C	COSM303938	NEGATIVE	SNV
ERBB2 G776L	p.G776L	COSM144255	NEGATIVE	mnp
ERBB2 G776R	p.G776R	OM3421	NEGATIVE	SNV
ERBB2 G776S	p.G776S	COSM685	NEGATIVE	SNV
ERBB2 G776V	p.G776V	COSM18609	NEGATIVE	SNV
ERBB2 I767M	p.I767M	COSM51317	NEGATIVE	SNV
ERBB2 L755A	p.L755A	COSM6906940	NEGATIVE	mnp
ERBB2 L755F	p.L755F	OBENV01	NEGATIVE	SNV
ERBB2 L755F	p.L755F	OBENV02	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 L755M	p.L755M	COSM1205571	NEGATIVE	SNV
ERBB2 L755S	p.L755S	COSM14060	NEGATIVE	SNV
ERBB2 L755W	p.L755W	COSM436499	NEGATIVE	SNV
ERBB2 L755_T759del	p.L755_T759del	COSM7345568	NEGATIVE	del
ERBB2 L755_T759del	p.L755_T759del	COSM5029269	NEGATIVE	del
ERBB2 L869R	p.L869R	COSM249793	NEGATIVE	SNV
ERBB2 R678Q	p.R678Q	COSM436498	NEGATIVE	SNV
ERBB2 R868W	p.R868W	COSM289684	NEGATIVE	SNV
ERBB2 R896C	p.R896C	COSM14066	NEGATIVE	SNV
ERBB2 R896H	p.R896H	COSM119971	NEGATIVE	SNV
ERBB2 S310F	p.S310F	COSM48358	NEGATIVE	SNV
ERBB2 S310P	p.S310P	COSM1563863	NEGATIVE	SNV
ERBB2 S310Y	p.S310Y	COSM6906440	NEGATIVE	mnp
ERBB2 S310Y	p.S310Y	COSM94225	NEGATIVE	SNV
ERBB2 T733I	p.T733I	COSM14059	NEGATIVE	SNV
ERBB2 T798I	p.T798I	OM3210	NEGATIVE	SNV
ERBB2 V659E	p.V659E	COSM3724566	NEGATIVE	mnp
ERBB2 V659E	p.V659E	COSM6503262	NEGATIVE	mnp
ERBB2 V777A	p.V777A	COSM35496	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM14062	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM436500	NEGATIVE	SNV
ERBB2 V777M	p.V777M	COSM14064	NEGATIVE	SNV
ERBB2 V842E	p.V842E	COSM6005745	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 V842I	p.V842I	COSM14065	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.Y772_V773insMMAY	OMINDEL820	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.A775_G776insVVMA	SPRNV01	NEGATIVE	ins
ERBB2 exon 20 insertion	p.M774dup	OMINDEL823	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insYLMA	SPRNV02	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_G776insV	MAN307	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insVAG	OMINDEL817	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insC	OMINDEL825	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.A775_G776insV	MAN300	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN308	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN309	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12552	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12553	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsLC	COSM12554	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM19875	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM20895	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsVC	COSM5235349	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsAVGC	COSM6907033	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM85995	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_V777delinsSVA	G776_V777delinsSVA_3	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_S779insCVG	OMINDEL1179	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsIC	OMINDEL826	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	SPRNV03	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776_V777insL	COSM6438151	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_S779dup	COSM681	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVV	COSM6977433	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.V777dup	COSM9869865	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_G778delinsVVGD	OMINDEL1189	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_P780dup	COSM12555	NEGATIVE	ins
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	COSM303948	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_P780dup	COSM6865893	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPG	COSM7449770	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insCPG	OMINDEL1180	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_G778insC	OMINDEL827	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPS	COSM5802314	NEGATIVE	ins
ERBB3 D297H	p.D297H	COSM1935392	NEGATIVE	SNV
ERBB3 D297N	p.D297N	COSM941489	NEGATIVE	SNV
ERBB3 D297V	p.D297V	COSM941490	NEGATIVE	SNV
ERBB3 D297Y	p.D297Y	COSM160822	NEGATIVE	SNV
ERBB3 E332K	p.E332K	COSM254677	NEGATIVE	SNV
ERBB3 E332Q	p.E332Q	COSM4838033	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM5413554	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB3 G284R	p.G284R	COSM941488	NEGATIVE	SNV
ERBB3 M60K	p.M60K	COSM254678	NEGATIVE	SNV
ERBB3 M60L	p.M60L	COSM1606366	NEGATIVE	SNV
ERBB3 M60R	p.M60R	COSM941484	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM122890	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM1299636	NEGATIVE	SNV
ERBB3 R103G	p.R103G	COSM6907312	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM160824	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM191840	NEGATIVE	SNV
ERBB3 V104M	p.V104M	COSM172423	NEGATIVE	SNV
ERBB4 R711C	p.R711C	COSM160827	NEGATIVE	SNV
ERBB4 R711L	p.R711L	OM3150	NEGATIVE	SNV
ESR1 D538G	p.D538G	COSM94250	NEGATIVE	SNV
ESR1 E380Q	p.E380Q	COSM3829320	NEGATIVE	SNV
ESR1 K303R	p.K303R	COSM4745827	NEGATIVE	SNV
ESR1 L536Q	p.L536Q	COSM4766050	NEGATIVE	mnp
ESR1 L536R	p.L536R	COSM4774826	NEGATIVE	SNV
ESR1 P535H	p.P535H	BT24	NEGATIVE	SNV
ESR1 P535T	p.P535T	COSM5666097	NEGATIVE	SNV
ESR1 R555C	p.R555C	BT32	NEGATIVE	SNV
ESR1 S463P	p.S463P	COSM4771561	NEGATIVE	SNV
ESR1 V392I	p.V392I	COSM1545070	NEGATIVE	SNV
ESR1 Y537C	p.Y537C	COSM1074637	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ESR1 Y537N	p.Y537N	COSM1074635	NEGATIVE	SNV
ESR1 Y537S	p.Y537S	COSM1074639	NEGATIVE	SNV
FGFR1 K687E	p.K687E	COSM35673	NEGATIVE	SNV
FGFR1 K687M	p.K687M	COSM4771556	NEGATIVE	SNV
FGFR1 K687N	p.K687N	COSM6578115	NEGATIVE	SNV
FGFR1 V592M	p.V592M	COSM6840954	NEGATIVE	SNV
FGFR2 A314D	p.A314D	COSM49171	NEGATIVE	SNV
FGFR2 A315T	p.A315T	COSM30777	NEGATIVE	SNV
FGFR2 A648T	p.A648T	COSM29836	NEGATIVE	SNV
FGFR2 C382G	p.C382G	COSM6197913	NEGATIVE	SNV
FGFR2 C382R	p.C382R	COSM36906	NEGATIVE	SNV
FGFR2 C382Y	p.C382Y	COSM915493	NEGATIVE	SNV
FGFR2 E565A	p.E565A	OM3292	NEGATIVE	SNV
FGFR2 E565G	p.E565G	OM3563	NEGATIVE	SNV
FGFR2 G613V	p.G613V	COSM94318	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM1474415	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM48382	NEGATIVE	SNV
FGFR2 I547V	p.I547V	COSM36913	NEGATIVE	SNV
FGFR2 K310R	p.K310R	COSM36901	NEGATIVE	SNV
FGFR2 K641R	p.K641R	COSM5010388	NEGATIVE	SNV
FGFR2 K659E	p.K659E	COSM36909	NEGATIVE	SNV
FGFR2 K659M	p.K659M	COSM49175	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM49173	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR2 K659N	p.K659N	COSM683054	NEGATIVE	SNV
FGFR2 L617M	p.L617M	OM3562	NEGATIVE	SNV
FGFR2 L617V	p.L617V	OM3291	NEGATIVE	SNV
FGFR2 M391R	p.M391R	COSM36907	NEGATIVE	SNV
FGFR2 N549D	p.N549D	COSM4604460	NEGATIVE	SNV
FGFR2 N549H	p.N549H	COSM250083	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36902	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36912	NEGATIVE	SNV
FGFR2 N549S	p.N549S	COSM3665553	NEGATIVE	SNV
FGFR2 N549T	p.N549T	COSM6952624	NEGATIVE	SNV
FGFR2 P253L	p.P253L	COSM537801	NEGATIVE	SNV
FGFR2 P253R	p.P253R	COSM49170	NEGATIVE	SNV
FGFR2 P253S	p.P253S	COSM1346291	NEGATIVE	SNV
FGFR2 S252W	p.S252W	COSM36903	NEGATIVE	SNV
FGFR2 V395D	p.V395D	COSM250081	NEGATIVE	mnp
FGFR2 V564F	p.V564F	OM3293	NEGATIVE	SNV
FGFR2 V564I	p.V564I	OM3247	NEGATIVE	SNV
FGFR2 V564L	p.V564L	OM3564	NEGATIVE	SNV
FGFR2 V679I	p.V679I	COSM4822956	NEGATIVE	SNV
FGFR2 Y375C	p.Y375C	COSM36904	NEGATIVE	SNV
FGFR2 Y375H	p.Y375H	COSM1560916	NEGATIVE	SNV
FGFR3 A391E	p.A391E	COSM721	NEGATIVE	SNV
FGFR3 A391T	p.A391T	OM3151	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 A391V	p.A391V	COSM1737909	NEGATIVE	SNV
FGFR3 D641G	p.D641G	COSM1428721	NEGATIVE	SNV
FGFR3 D641N	p.D641N	COSM29438	NEGATIVE	SNV
FGFR3 G370C	p.G370C	COSM716	NEGATIVE	SNV
FGFR3 G370D	p.G370D	COSM5966955	NEGATIVE	SNV
FGFR3 G380E	p.G380E	COSM51545	NEGATIVE	SNV
FGFR3 G380R	p.G380R	COSM24842	NEGATIVE	SNV
FGFR3 K650E	p.K650E	COSM719	NEGATIVE	SNV
FGFR3 K650M	p.K650M	COSM720	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM1428730	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM3993568	NEGATIVE	SNV
FGFR3 K650Q	p.K650Q	COSM726	NEGATIVE	SNV
FGFR3 K650R	p.K650R	OM3224	NEGATIVE	SNV
FGFR3 K650T	p.K650T	COSM731	NEGATIVE	SNV
FGFR3 K715M	p.K715M	COSM732992	NEGATIVE	SNV
FGFR3 R248C	p.R248C	COSM714	NEGATIVE	SNV
FGFR3 R248H	p.R248H	COSM1757310	NEGATIVE	SNV
FGFR3 R399C	p.R399C	COSM296687	NEGATIVE	SNV
FGFR3 S249C	p.S249C	COSM715	NEGATIVE	SNV
FGFR3 S249T	p.S249T	COSM29431	NEGATIVE	SNV
FGFR3 S371C	p.S371C	COSM17461	NEGATIVE	SNV
FGFR3 V553M	p.V553M	OM3223	NEGATIVE	SNV
FGFR3 V555M	p.V555M	COSM303914	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 V677I	p.V677I	COSM178247	NEGATIVE	SNV
FGFR3 Y373C	p.Y373C	COSM718	NEGATIVE	SNV
FGFR4 V550E	p.V550E	COSM9215998	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM1066172	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM3738141	NEGATIVE	SNV
FGFR4 V550M	p.V550M	COSM6163	NEGATIVE	SNV
FLT3 D835A	p.D835A	COSM27650	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM787	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM788	NEGATIVE	SNV
FLT3 D835F	p.D835F	COSM19844	NEGATIVE	mnp
FLT3 D835G	p.D835G	COSM1366188	NEGATIVE	SNV
FLT3 D835H	p.D835H	COSM785	NEGATIVE	SNV
FLT3 D835K	p.D835K	OM3570	NEGATIVE	mnp
FLT3 D835K	p.D835K	OM3571	NEGATIVE	mnp
FLT3 D835L	p.D835L	OM3572	NEGATIVE	mnp
FLT3 D835N	p.D835N	COSM789	NEGATIVE	SNV
FLT3 D835V	p.D835V	COSM784	NEGATIVE	SNV
FLT3 D835Y	p.D835Y	COSM783	NEGATIVE	SNV
FLT3 D835_I836del	p.D835_I836del	COSM5879385	NEGATIVE	del
FLT3 D835_I836delinsV	p.D835_I836delinsV	COSM27651	NEGATIVE	del
FLT3 D835del	p.D835del	COSM854	NEGATIVE	del
FLT3 D839E	p.D839E	COSM5945287	NEGATIVE	SNV
FLT3 D839G	p.D839G	COSM1166729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FLT3 D839H	p.D839H	COSM3399304	NEGATIVE	SNV
FLT3 D839N	p.D839N	COSM6005768	NEGATIVE	SNV
FLT3 I836D	p.I836D	OM3569	NEGATIVE	mnp
FLT3 I836L	p.I836L	COSM19649	NEGATIVE	SNV
FLT3 I836S	p.I836S	COSM19843	NEGATIVE	SNV
FLT3 I836T	p.I836T	COSM5879386	NEGATIVE	SNV
FLT3 I836del	p.I836del	COSM19836	NEGATIVE	del
GNAS Q227E	p.Q227E	COSM4962873	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM27900	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM4168960	NEGATIVE	SNV
GNAS Q227K	p.Q227K	COSM28618	NEGATIVE	SNV
GNAS Q227L	p.Q227L	COSM27888	NEGATIVE	SNV
GNAS Q227R	p.Q227R	COSM27896	NEGATIVE	SNV
GNAS R201C	p.R201C	COSM27887	NEGATIVE	SNV
GNAS R201G	p.R201G	COSM1757313	NEGATIVE	SNV
GNAS R201H	p.R201H	COSM27895	NEGATIVE	SNV
GNAS R201L	p.R201L	COSM99221	NEGATIVE	SNV
GNAS R201S	p.R201S	COSM27899	NEGATIVE	SNV
HRAS G12A	p.G12A	COSM485	NEGATIVE	SNV
HRAS G12C	p.G12C	COSM481	NEGATIVE	SNV
HRAS G12D	p.G12D	COSM484	NEGATIVE	SNV
HRAS G12N	p.G12N	COSM4517473	NEGATIVE	mnp
HRAS G12R	p.G12R	COSM482	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
HRAS G12S	p.G12S	COSM480	NEGATIVE	SNV
HRAS G12V	p.G12V	COSM483	NEGATIVE	SNV
HRAS G13C	p.G13C	COSM488	NEGATIVE	SNV
HRAS G13D	p.G13D	COSM490	NEGATIVE	SNV
HRAS G13R	p.G13R	COSM486	NEGATIVE	SNV
HRAS G13S	p.G13S	COSM487	NEGATIVE	SNV
HRAS G13V	p.G13V	COSM489	NEGATIVE	SNV
HRAS Q61E	p.Q61E	COSM497	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM502	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM503	NEGATIVE	SNV
HRAS Q61K	p.Q61K	COSM496	NEGATIVE	SNV
HRAS Q61L	p.Q61L	COSM52978	NEGATIVE	mnp
HRAS Q61L	p.Q61L	COSM498	NEGATIVE	SNV
HRAS Q61P	p.Q61P	COSM500	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM33695	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM501	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM499	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM52979	NEGATIVE	mnp
IDH1 G70D	p.G70D	COSM96531	NEGATIVE	SNV
IDH1 R132C	p.R132C	COSM28747	NEGATIVE	SNV
IDH1 R132G	p.R132G	COSM28749	NEGATIVE	SNV
IDH1 R132H	p.R132H	COSM86993	NEGATIVE	mnp
IDH1 R132H	p.R132H	COSM28746	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
IDH1 R132L	p.R132L	COSM28750	NEGATIVE	SNV
IDH1 R132Q	p.R132Q	OM3221	NEGATIVE	mnp
IDH1 R132Q	p.R132Q	OM3222	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM1169216	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM28748	NEGATIVE	SNV
IDH1 R132V	p.R132V	COSM28751	NEGATIVE	mnp
IDH1 S280F	p.S280F	OM3315	NEGATIVE	SNV
IDH2 I319M	p.I319M	OM3319	NEGATIVE	SNV
IDH2 Q316E	p.Q316E	OM3320	NEGATIVE	SNV
IDH2 R140G	p.R140G	COSM1737874	NEGATIVE	SNV
IDH2 R140L	p.R140L	COSM41875	NEGATIVE	SNV
IDH2 R140Q	p.R140Q	COSM41590	NEGATIVE	SNV
IDH2 R140W	p.R140W	COSM41877	NEGATIVE	SNV
IDH2 R172G	p.R172G	COSM33731	NEGATIVE	SNV
IDH2 R172K	p.R172K	COSM33733	NEGATIVE	SNV
IDH2 R172M	p.R172M	COSM33732	NEGATIVE	SNV
IDH2 R172N	p.R172N	COSM256046	NEGATIVE	mnp
IDH2 R172S	p.R172S	COSM133672	NEGATIVE	SNV
IDH2 R172S	p.R172S	COSM34090	NEGATIVE	SNV
IDH2 R172T	p.R172T	COSM1636997	NEGATIVE	SNV
IDH2 R172V	p.R172V	OM3327	NEGATIVE	mnp
IDH2 R172W	p.R172W	COSM34039	NEGATIVE	SNV
KEAP1 D422N	p.D422N	COSM710198	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 G333C	p.G333C	COSM1193323	NEGATIVE	SNV
KEAP1 M409*	p.M409*	COSM6916353	NEGATIVE	del
KEAP1 R320L	p.R320L	COSM2812614	NEGATIVE	SNV
KEAP1 R320P	p.R320P	COSM346494	NEGATIVE	SNV
KEAP1 R320Q	p.R320Q	COSM710195	NEGATIVE	SNV
KEAP1 R320W	p.R320W	COSM1524062	NEGATIVE	SNV
KEAP1 R415G	p.R415G	COSM6196677	NEGATIVE	SNV
KEAP1 R470C	p.R470C	COSM564847	NEGATIVE	SNV
KEAP1 R470G	p.R470G	OM3724	NEGATIVE	SNV
KEAP1 R470H	p.R470H	COSM1524064	NEGATIVE	SNV
KEAP1 R470S	p.R470S	COSM564846	NEGATIVE	SNV
KEAP1 R554Q	p.R554Q	COSM1524066	NEGATIVE	SNV
KEAP1 S243C	p.S243C	COSM710191	NEGATIVE	SNV
KEAP1 V155F	p.V155F	COSM710187	NEGATIVE	SNV
KEAP1 V167F	p.V167F	COSM710188	NEGATIVE	SNV
KEAP1 Y584*	p.Y584*	COSM6201646	NEGATIVE	ins
KEAP1 c.1531+1G>T	p.?	COSM6956065	NEGATIVE	SNV
KEAP1 c.1531+2T>G	p.?	OM3723	NEGATIVE	SNV
KEAP1 c.1532-1G>T	p.?	COSM6969960	NEGATIVE	SNV
KEAP1 c.1532-2A>G	p.?	COSM381459	NEGATIVE	SNV
KEAP1 c.1708+1G>T	p.?	COSM7738757	NEGATIVE	SNV
KEAP1 c.1708+2T>A	p.?	COSM6149754	NEGATIVE	SNV
KEAP1 c.1708_1708+1delinsT	p.?	COSM6927565	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
T				
KEAP1 c.1709-1G>T	p.?	COSM7724088	NEGATIVE	SNV
KEAP1 c.1709-2A>G	p.?	COSM7088100	NEGATIVE	SNV
KEAP1 c.640-1G>A	p.?	COSM6972470	NEGATIVE	SNV
KEAP1 c.640-1G>C	p.?	OM3726	NEGATIVE	SNV
KEAP1 c.640-1G>T	p.?	OM3725	NEGATIVE	SNV
KEAP1 c.640-2A>C	p.?	OM3727	NEGATIVE	SNV
KIT A502_Y503dup	p.A502_Y503dup	COSM1326	NEGATIVE	ins
KIT A502_Y503insFA	p.A502_Y503insFA	COSM53306	NEGATIVE	ins
KIT A829P	p.A829P	COSM13172	NEGATIVE	SNV
KIT A829V	p.A829V	COSM133757	NEGATIVE	SNV
KIT D419_L421delinsF	p.D419_L421delinsF	COSM1152	NEGATIVE	complex
KIT D419_L421delinsH	p.D419_L421delinsH	COSM1578133	NEGATIVE	complex
KIT D419_R420del	p.D419_R420del	COSM1578132	NEGATIVE	del
KIT D419_R420insFF	p.D419_R420insFF	COSM36334	NEGATIVE	ins
KIT D419_R420insFL	p.D419_R420insFL	COSM36332	NEGATIVE	ins
KIT D419del	p.D419del	COSM29014	NEGATIVE	del
KIT D419del	p.D419del	COSM1600400	NEGATIVE	complex
KIT D816A	p.D816A	COSM24675	NEGATIVE	SNV
KIT D816E	p.D816E	COSM19285	NEGATIVE	SNV
KIT D816E	p.D816E	COSM5749965	NEGATIVE	SNV
KIT D816F	p.D816F	COSM1312	NEGATIVE	mnp
KIT D816G	p.D816G	COSM12711	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT D816H	p.D816H	COSM1311	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1600404	NEGATIVE	mnp
KIT D816I	p.D816I	COSM21979	NEGATIVE	mnp
KIT D816N	p.D816N	COSM1313	NEGATIVE	SNV
KIT D816V	p.D816V	COSM1314	NEGATIVE	SNV
KIT D816Y	p.D816Y	COSM1310	NEGATIVE	SNV
KIT D820A	p.D820A	COSM133670	NEGATIVE	SNV
KIT D820E	p.D820E	COSM12709	NEGATIVE	SNV
KIT D820E	p.D820E	COSM19280	NEGATIVE	SNV
KIT D820G	p.D820G	COSM1316	NEGATIVE	SNV
KIT D820H	p.D820H	COSM22379	NEGATIVE	SNV
KIT D820N	p.D820N	COSM17947	NEGATIVE	SNV
KIT D820V	p.D820V	COSM1317	NEGATIVE	SNV
KIT D820Y	p.D820Y	COSM12710	NEGATIVE	SNV
KIT F522C	p.F522C	OM3237	NEGATIVE	SNV
KIT H697Y	p.H697Y	COSM36053	NEGATIVE	SNV
KIT I653T	p.I653T	COSM19326	NEGATIVE	SNV
KIT K550I	p.K550I	COSM1165	NEGATIVE	SNV
KIT K550N	p.K550N	COSM1167	NEGATIVE	SNV
KIT K550R	p.K550R	COSM1166	NEGATIVE	SNV
KIT K558N	p.K558N	OM3238	NEGATIVE	SNV
KIT K558N	p.K558N	COSM19216	NEGATIVE	SNV
KIT K642E	p.K642E	COSM1304	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT K642N	p.K642N	COSM4125155	NEGATIVE	SNV
KIT K642Q	p.K642Q	COSM96871	NEGATIVE	SNV
KIT L576F	p.L576F	COSM33966	NEGATIVE	SNV
KIT L576P	p.L576P	COSM1290	NEGATIVE	SNV
KIT L576R	p.L576R	COSM5945006	NEGATIVE	SNV
KIT N505I	p.N505I	OM3236	NEGATIVE	SNV
KIT N655K	p.N655K	COSM235682	NEGATIVE	SNV
KIT N655K	p.N655K	COSM4413464	NEGATIVE	SNV
KIT N655S	p.N655S	OM3302	NEGATIVE	SNV
KIT N655T	p.N655T	OM3303	NEGATIVE	SNV
KIT N680K	p.N680K	COSM5352270	NEGATIVE	SNV
KIT N822H	p.N822H	COSM1318	NEGATIVE	SNV
KIT N822I	p.N822I	COSM3604523	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1321	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1322	NEGATIVE	SNV
KIT N822S	p.N822S	COSM33975	NEGATIVE	SNV
KIT N822Y	p.N822Y	COSM19109	NEGATIVE	SNV
KIT P838L	p.P838L	OM3240	NEGATIVE	SNV
KIT R796A	p.R796A	COSM1272091	NEGATIVE	mnp
KIT R796G	p.R796G	COSM51381	NEGATIVE	SNV
KIT R796K	p.R796K	COSM1600411	NEGATIVE	SNV
KIT R796P	p.R796P	COSM1272092	NEGATIVE	mnp
KIT S501_A502dup	p.S501_A502dup	COSM5702501	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT S501_A502dup	p.S501_A502dup	COSM219786	NEGATIVE	ins
KIT T417_D419delinsI	p.T417_D419delinsI	COSM1148	NEGATIVE	complex
KIT T417_D419delinsRG	p.T417_D419delinsRG	COSM1149	NEGATIVE	complex
KIT T670A	p.T670A	OM3304	NEGATIVE	SNV
KIT T670E	p.T670E	COSM19293	NEGATIVE	mnp
KIT T670I	p.T670I	COSM12708	NEGATIVE	SNV
KIT V530I	p.V530I	COSM1155	NEGATIVE	SNV
KIT V559A	p.V559A	COSM1255	NEGATIVE	SNV
KIT V559C	p.V559C	OM3239	NEGATIVE	mnp
KIT V559D	p.V559D	COSM1252	NEGATIVE	SNV
KIT V559G	p.V559G	COSM1253	NEGATIVE	SNV
KIT V559I	p.V559I	COSM1251	NEGATIVE	SNV
KIT V559K	p.V559K	COSM96869	NEGATIVE	mnp
KIT V559P	p.V559P	COSM1272086	NEGATIVE	mnp
KIT V560A	p.V560A	COSM36302	NEGATIVE	SNV
KIT V560D	p.V560D	COSM1257	NEGATIVE	SNV
KIT V560E	p.V560E	COSM1333	NEGATIVE	mnp
KIT V560G	p.V560G	COSM1260	NEGATIVE	SNV
KIT V654A	p.V654A	COSM12706	NEGATIVE	SNV
KIT V654L	p.V654L	COSM5941904	NEGATIVE	SNV
KIT V825A	p.V825A	COSM1323	NEGATIVE	SNV
KIT V825I	p.V825I	COSM19110	NEGATIVE	SNV
KIT W557C	p.W557C	COSM19108	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT W557C	p.W557C	COSM133700	NEGATIVE	SNV
KIT W557G	p.W557G	COSM1221	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1216	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1219	NEGATIVE	SNV
KIT W557S	p.W557S	COSM1227	NEGATIVE	SNV
KIT Y553C	p.Y553C	COSM4413463	NEGATIVE	SNV
KIT Y553N	p.Y553N	COSM133763	NEGATIVE	SNV
KIT Y570H	p.Y570H	COSM1430152	NEGATIVE	SNV
KIT Y578C	p.Y578C	COSM6503446	NEGATIVE	SNV
KIT Y823C	p.Y823C	COSM18682	NEGATIVE	SNV
KIT Y823D	p.Y823D	COSM18681	NEGATIVE	SNV
KIT Y823H	p.Y823H	COSM1651649	NEGATIVE	SNV
KIT Y823N	p.Y823N	COSM22378	NEGATIVE	SNV
KIT [K558S;V559S]	p.[K558S;V559S]	COSM28637	NEGATIVE	mnp
KIT [W557C;K558P]	p.[W557C;K558P]	COSM21978	NEGATIVE	mnp
KIT [W557C;K558T]	p.[W557C;K558T]	COSM28992	NEGATIVE	mnp
KIT [W557F;K558P]	p.[W557F;K558P]	COSM1224	NEGATIVE	mnp
KIT [W557S;K558S]	p.[W557S;K558S]	COSM96948	NEGATIVE	mnp
KIT exon 11 deletion	p.?	OMINDEL901	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1163	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1158	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1160	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1162	NEGATIVE	del

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.P551_E554del	COSM1172	NEGATIVE	del
KIT exon 11 deletion	p.K550_V555delinsQ	COSM6938892	NEGATIVE	complex
KIT exon 11 deletion	p.K550_Q556delinsII	COSM1161	NEGATIVE	complex
KIT exon 11 deletion	p.P551_V555del	COSM1169	NEGATIVE	del
KIT exon 11 deletion	p.K550_W557delinsIL	COSM6936468	NEGATIVE	complex
KIT exon 11 deletion	p.K550_E554delinsI	COSM6953101	NEGATIVE	complex
KIT exon 11 deletion	p.K550_P551delinsI	COSM6971267	NEGATIVE	complex
KIT exon 11 deletion	p.K550_W557delinsIR	COSM6977972	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Q556del	COSM1170	NEGATIVE	del
KIT exon 11 deletion	p.P551del	COSM1171	NEGATIVE	del
KIT exon 11 deletion	p.P551_V555delinsL	COSM1174	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.P551_V555delinsTL	COSM30550	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Y553delinsH	COSM6934157	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1177	NEGATIVE	del
KIT exon 11 deletion	p.M552_V555del	COSM6957862	NEGATIVE	del
KIT exon 11 deletion	p.M552_Y553del	COSM96881	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsL	COSM1178	NEGATIVE	complex
KIT exon 11 deletion	p.M552_Y553del	COSM1179	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1180	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554del	COSM1181	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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_V555del	COSM18827	NEGATIVE	del
KIT exon 11 deletion	p.M552_K558del	OMINDEL902	NEGATIVE	complex
KIT exon 11 deletion	p.M552_V555delinsI	COSM1185	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557delinsR	COSM133636	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.M552_Y553delinsN	COSM96863	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsK	COSM96868	NEGATIVE	del
KIT exon 11 deletion	p.M552_E561delinsK	COSM96880	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_K558del	COSM27909	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554delinsI	COSM6944981	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559del	COSM1191	NEGATIVE	del
KIT exon 11 deletion	p.Y553_E554del	COSM6923694	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559delinsE	COSM96946	NEGATIVE	complex
KIT exon 11 deletion	p.E554_N564del	COSM96947	NEGATIVE	del
KIT exon 11 deletion	p.E554_V559del	OMINDEL701	NEGATIVE	del
KIT exon 11 deletion	p.E554_K558del	COSM1192	NEGATIVE	del
KIT exon 11 deletion	p.V555_E562del	COSM1194	NEGATIVE	del
KIT exon 11 deletion	p.E554_E562del	COSM36310	NEGATIVE	del

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.V555_G565del	COSM37027	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1198	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559del	COSM1199	NEGATIVE	del
KIT exon 11 deletion	p.V555_Q556del	COSM1201	NEGATIVE	del
KIT exon 11 deletion	p.V555_V560del	COSM1202	NEGATIVE	del
KIT exon 11 deletion	p.V555_N566delinsD	COSM96870	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1204	NEGATIVE	del
KIT exon 11 deletion	p.Q556_E561del	COSM1206	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558del	COSM23418	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.W557_K558del	COSM6986076	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsF	COSM1207	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558del	COSM1217	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561del	COSM1332	NEGATIVE	del
KIT exon 11 deletion	p.Q556_W557delinsR	COSM133642	NEGATIVE	del
KIT exon 11 deletion	p.Q556_I563delinsL	COSM1651755	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsTTF	COSM27904	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E562del	COSM36312	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsH	COSM1213	NEGATIVE	del
KIT exon 11 deletion	p.W557del	COSM1220	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559delinsH	COSM133641	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_E561delinsHH	COSM1208	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V559delinsHT	COSM1214	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559del	COSM1218	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560del	COSM1223	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561delinsPS	COSM6922994	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E561delinsQ	COSM6954755	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsI	COSM1222	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsF	COSM1226	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560delinsF	COSM1230	NEGATIVE	del
KIT exon 11 deletion	p.W557_K558delinsE	COSM24748	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558delinsQ	COSM6946777	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V560delinsC	COSM1232	NEGATIVE	del
KIT exon 11 deletion	p.W557_V559delinsC	COSM1233	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.K558_V560del	COSM1234	NEGATIVE	del
KIT exon 11 deletion	p.K558_V559del	COSM1235	NEGATIVE	del
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.W557_K558delinsC	COSM96887	NEGATIVE	complex
KIT exon 11 deletion	p.K558_G565delinsR	COSM1240	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K558_V560delinsI	COSM1241	NEGATIVE	del
KIT exon 11 deletion	p.V559_E561del	COSM1250	NEGATIVE	del
KIT exon 11 deletion	p.K558_Y570delinsN	COSM28991	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsL	COSM6940807	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V560delinsM	COSM1242	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V559delinsN	COSM1244	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsN	COSM27069	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM6971633	NEGATIVE	del
KIT exon 11 deletion	p.V560del	COSM1247	NEGATIVE	del
KIT exon 11 deletion	p.V559_V560del	COSM1248	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM1249	NEGATIVE	del
KIT exon 11 deletion	p.V559_I571del	COSM36301	NEGATIVE	del
KIT exon 11 deletion	p.V560_I563del	COSM18897	NEGATIVE	del
KIT exon 11 deletion	p.V559_N566delinsD	COSM6977976	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1262	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1263	NEGATIVE	del
KIT exon 11 deletion	p.E562_V569delinsD	COSM1267	NEGATIVE	del
KIT exon 11 deletion	p.I563_D572del	COSM1268	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.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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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_Q575del	COSM1281	NEGATIVE	del
KIT exon 11 deletion	p.V569_D572del	COSM1282	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 deletion	p.I571_Y578del	OMINDEL1186	NEGATIVE	del
KIT exon 11 deletion	p.I571_L576del	COSM133754	NEGATIVE	del
KIT exon 11 deletion	p.Q575del	COSM96875	NEGATIVE	del
KIT exon 11 deletion	p.Q575_L576del	COSM6987753	NEGATIVE	del
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.Q575_P577delinsT	COSM96865	NEGATIVE	complex
KIT exon 11 deletion	p.L576del	COSM1289	NEGATIVE	del
KIT exon 11 deletion	p.P577_Y578del	COSM1291	NEGATIVE	del
KIT exon 11 deletion	p.P577del	COSM1684822	NEGATIVE	del
KIT exon 11 deletion	p.P577_D579del	COSM98379	NEGATIVE	del
KIT exon 11 deletion	p.D579del	COSM1294	NEGATIVE	del
KIT exon 11 deletion	p.H580del	COSM1295	NEGATIVE	del
KIT exon 11 insertion	p.K558delinsNP	COSM21976	NEGATIVE	ins
KIT exon 11 insertion	p.K558delinsNP	COSM1245	NEGATIVE	complex
KIT exon 11 insertion	p.I571_D579dup	COSM23417	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 insertion	p.D572_D579dup	COSM308544	NEGATIVE	ins
KIT exon 11 insertion	p.P573_H580dup	COSM34126	NEGATIVE	ins
KIT exon 11 insertion	p.P573_D579dup	COSM18668	NEGATIVE	ins
KIT exon 11 insertion	p.T574_D579dup	OMINDEL702	NEGATIVE	ins
KIT exon 11 insertion	p.Q575_L576dup	COSM1292	NEGATIVE	ins
KIT exon 11 insertion	p.Y578_P585dup	COSM41632	NEGATIVE	ins
KIT exon 11 insertion	p.P577_H580dup	COSM1296	NEGATIVE	ins
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
KRAS A146G	p.A146G	COSM1360828	NEGATIVE	SNV
KRAS A146P	p.A146P	COSM19905	NEGATIVE	SNV
KRAS A146S	p.A146S	COSM5967494	NEGATIVE	SNV
KRAS A146T	p.A146T	COSM19404	NEGATIVE	SNV
KRAS A146V	p.A146V	COSM19900	NEGATIVE	SNV
KRAS A59E	p.A59E	COSM547	NEGATIVE	SNV
KRAS A59G	p.A59G	COSM28518	NEGATIVE	SNV
KRAS A59S	p.A59S	COSM1235389	NEGATIVE	SNV
KRAS A59T	p.A59T	COSM546	NEGATIVE	SNV
KRAS G12A	p.G12A	COSM522	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G12C	p.G12C	COSM516	NEGATIVE	SNV
KRAS G12D	p.G12D	COSM14209	NEGATIVE	mnp
KRAS G12D	p.G12D	COSM521	NEGATIVE	SNV
KRAS G12E	p.G12E	COSM30566	NEGATIVE	mnp
KRAS G12E	p.G12E	COSM519	NEGATIVE	mnp
KRAS G12F	p.G12F	COSM512	NEGATIVE	mnp
KRAS G12I	p.G12I	COSM34144	NEGATIVE	mnp
KRAS G12L	p.G12L	COSM514	NEGATIVE	mnp
KRAS G12N	p.G12N	COSM13643	NEGATIVE	mnp
KRAS G12R	p.G12R	COSM518	NEGATIVE	SNV
KRAS G12S	p.G12S	COSM517	NEGATIVE	SNV
KRAS G12V	p.G12V	COSM515	NEGATIVE	mnp
KRAS G12V	p.G12V	COSM520	NEGATIVE	SNV
KRAS G12W	p.G12W	COSM36281	NEGATIVE	mnp
KRAS G12Y	p.G12Y	COSM25081	NEGATIVE	mnp
KRAS G13A	p.G13A	COSM533	NEGATIVE	SNV
KRAS G13C	p.G13C	COSM527	NEGATIVE	SNV
KRAS G13D	p.G13D	COSM531	NEGATIVE	mnp
KRAS G13D	p.G13D	COSM532	NEGATIVE	SNV
KRAS G13E	p.G13E	COSM30567	NEGATIVE	mnp
KRAS G13E	p.G13E	COSM87280	NEGATIVE	mnp
KRAS G13I	p.G13I	COSM525	NEGATIVE	mnp
KRAS G13N	p.G13N	COSM53283	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G13R	p.G13R	COSM526	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM529	NEGATIVE	SNV
KRAS G13S	p.G13S	COSM528	NEGATIVE	SNV
KRAS G13V	p.G13V	COSM12721	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM530	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM534	NEGATIVE	SNV
KRAS G15D	p.G15D	COSM539	NEGATIVE	SNV
KRAS G15S	p.G15S	COSM538	NEGATIVE	SNV
KRAS K117E	p.K117E	COSM6854421	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM19940	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM28519	NEGATIVE	SNV
KRAS K117R	p.K117R	COSM4696721	NEGATIVE	SNV
KRAS Q61E	p.Q61E	COSM550	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM554	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM555	NEGATIVE	SNV
KRAS Q61K	p.Q61K	COSM549	NEGATIVE	SNV
KRAS Q61L	p.Q61L	COSM553	NEGATIVE	SNV
KRAS Q61P	p.Q61P	COSM551	NEGATIVE	SNV
KRAS Q61R	p.Q61R	COSM1168052	NEGATIVE	mnp
KRAS Q61R	p.Q61R	COSM552	NEGATIVE	SNV
KRAS [G12=;G13C]	p.[G12=;G13C]	COSM87281	NEGATIVE	mnp
KRAS [G13D;V14I]	p.[G13D;V14I]	COSM30568	NEGATIVE	mnp
KRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM87298	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 C121S	p.C121S	COSM555601	NEGATIVE	SNV
MAP2K1 C121S	p.C121S	COSM1315829	NEGATIVE	SNV
MAP2K1 E203K	p.E203K	COSM232755	NEGATIVE	SNV
MAP2K1 E203V	p.E203V	COSM3386991	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3249	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	COSM1235480	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3250	NEGATIVE	SNV
MAP2K1 F53C	p.F53C	COSM1562837	NEGATIVE	SNV
MAP2K1 F53I	p.F53I	COSM3503329	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM555604	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM1725008	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM2257208	NEGATIVE	SNV
MAP2K1 F53S	p.F53S	ClinVar13350	NEGATIVE	SNV
MAP2K1 F53V	p.F53V	COSM5077832	NEGATIVE	SNV
MAP2K1 F53Y	p.F53Y	COSM5588244	NEGATIVE	SNV
MAP2K1 G128D	p.G128D	COSM1374186	NEGATIVE	SNV
MAP2K1 G128V	p.G128V	COSM1517232	NEGATIVE	SNV
MAP2K1 K57E	p.K57E	COSM5369532	NEGATIVE	SNV
MAP2K1 K57M	p.K57M	MAN124	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM1235478	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM5520914	NEGATIVE	SNV
MAP2K1 K57Q	p.K57Q	OM3155	NEGATIVE	SNV
MAP2K1 K57T	p.K57T	COSM4756761	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 L115P	p.L115P	OM3248	NEGATIVE	SNV
MAP2K1 L215P	p.L215P	OM3252	NEGATIVE	SNV
MAP2K1 P124L	p.P124L	COSM1315861	NEGATIVE	SNV
MAP2K1 P124Q	p.P124Q	COSM1167912	NEGATIVE	SNV
MAP2K1 P124S	p.P124S	COSM235614	NEGATIVE	SNV
MAP2K1 Q56P	p.Q56P	COSM1235481	NEGATIVE	SNV
MAP2K1 V211D	p.V211D	OM3251	NEGATIVE	SNV
MAP2K1 V60E	p.V60E	COSM2257209	NEGATIVE	SNV
MAP2K1 V60M	p.V60M	OM3576	NEGATIVE	SNV
MAP2K2 C125S	p.C125S	COSM5855814	NEGATIVE	SNV
MAP2K2 N126D	p.N126D	COSM1731742	NEGATIVE	SNV
MAP2K2 V35M	p.V35M	COSM5855836	NEGATIVE	SNV
MET H1112L	p.H1112L	COSM698	NEGATIVE	SNV
MET H1112R	p.H1112R	COSM703	NEGATIVE	SNV
MET H1112Y	p.H1112Y	COSM696	NEGATIVE	SNV
MET Y1021F	p.Y1021F	COSM339515	NEGATIVE	SNV
MET Y1021S	p.Y1021S	COSM7450124	NEGATIVE	SNV
MET c.2942-20_2942-19delinsAGAG	p.?	OMINDEL804	NEGATIVE	complex
MET c.2942-21_2942-20delinsGC	p.?	COSM8631602	NEGATIVE	mnp
MET c.2942-29_2942-28delinsGG	p.?	OM3305	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM29635	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.?	OMINDEL674	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM49015	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6980941	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL709	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6056832	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL675	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL676	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL710	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.?	COSM6057022	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL640	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL697	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6947399	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL642	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL677	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL679	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL712	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6948341	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL680	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL681	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.?	COSM6976260	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL684	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL706	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL685	NEGATIVE	ins
MET exon 14 skipping (DNA)	p.?	OMINDEL808	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OM3177	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3178	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL809	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.A1023IfsTer19	OMINDEL810	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6057025	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL686	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6937267	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6977526	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL654	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL655	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL811	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL656	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL658	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL688	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL687	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL689	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL662	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.?	COSM6965225	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.?	OMINDEL812	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL813	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.?	OMINDEL814	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.?	OMINDEL694	NEGATIVE	del
MET exon 14 skipping (DNA)	p.D1028IfsTer19	COSM24687	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.D1028Y	COSM6917314	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.D1028N	COSM6924587	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL695	NEGATIVE	complex
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.?	COSM6928344	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6940825	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6983140	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL815	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6108460	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6947995	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3185	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3217	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM744044	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3186	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3188	NEGATIVE	SNV
NRAS A146T	p.A146T	COSM27174	NEGATIVE	SNV
NRAS A146V	p.A146V	COSM4170228	NEGATIVE	SNV
NRAS A59D	p.A59D	COSM253327	NEGATIVE	SNV
NRAS A59T	p.A59T	COSM578	NEGATIVE	SNV
NRAS G12A	p.G12A	COSM565	NEGATIVE	SNV
NRAS G12C	p.G12C	COSM562	NEGATIVE	SNV
NRAS G12D	p.G12D	COSM564	NEGATIVE	SNV
NRAS G12E	p.G12E	COSM144577	NEGATIVE	mnp
NRAS G12N	p.G12N	COSM12723	NEGATIVE	mnp
NRAS G12P	p.G12P	COSM559	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS G12R	p.G12R	COSM561	NEGATIVE	SNV
NRAS G12S	p.G12S	COSM563	NEGATIVE	SNV
NRAS G12V	p.G12V	COSM566	NEGATIVE	SNV
NRAS G12Y	p.G12Y	COSM560	NEGATIVE	mnp
NRAS G13A	p.G13A	COSM575	NEGATIVE	SNV
NRAS G13C	p.G13C	COSM570	NEGATIVE	SNV
NRAS G13D	p.G13D	COSM573	NEGATIVE	SNV
NRAS G13N	p.G13N	COSM24668	NEGATIVE	mnp
NRAS G13R	p.G13R	COSM569	NEGATIVE	SNV
NRAS G13S	p.G13S	COSM571	NEGATIVE	SNV
NRAS G13V	p.G13V	COSM572	NEGATIVE	mnp
NRAS G13V	p.G13V	COSM574	NEGATIVE	SNV
NRAS G13Y	p.G13Y	COSM568	NEGATIVE	mnp
NRAS K117N	p.K117N	MAN13	NEGATIVE	SNV
NRAS Q61E	p.Q61E	COSM581	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM585	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM586	NEGATIVE	SNV
NRAS Q61K	p.Q61K	COSM53223	NEGATIVE	mnp
NRAS Q61K	p.Q61K	COSM580	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM30646	NEGATIVE	mnp
NRAS Q61L	p.Q61L	COSM583	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM12725	NEGATIVE	mnp
NRAS Q61P	p.Q61P	COSM582	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS Q61R	p.Q61R	COSM33693	NEGATIVE	mnp
NRAS Q61R	p.Q61R	COSM584	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM579	NEGATIVE	mnp
NRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM12730	NEGATIVE	mnp
NRAS [Q61H;E62K]	p.[Q61H;E62K]	COSM26494	NEGATIVE	mnp
NTRK1 F589C	p.F589C	OM3255	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT106	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT107	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT108	NEGATIVE	SNV
NTRK1 F589Y	p.F589Y	BT105	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT109	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT110	NEGATIVE	SNV
NTRK1 G667A	p.G667A	OM3301	NEGATIVE	SNV
NTRK1 G667C	p.G667C	BT111	NEGATIVE	SNV
NTRK1 G667S	p.G667S	BT112	NEGATIVE	SNV
NTRK1 V573M	p.V573M	BT104	NEGATIVE	SNV
NTRK2 F633C	p.F633C	OM3278	NEGATIVE	SNV
NTRK2 F633I	p.F633I	OM3276	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3277	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3308	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3309	NEGATIVE	SNV
NTRK2 F633Y	p.F633Y	OM3307	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3279	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NTRK2 G639R	p.G639R	OM3280	NEGATIVE	SNV
NTRK2 G709A	p.G709A	OM3310	NEGATIVE	SNV
NTRK2 Q612E	p.Q612E	OM3272	NEGATIVE	SNV
NTRK2 Q612P	p.Q612P	OM3273	NEGATIVE	SNV
NTRK2 R646K	p.R646K	OM3281	NEGATIVE	SNV
NTRK2 V617D	p.V617D	OM3306	NEGATIVE	SNV
NTRK2 V617G	p.V617G	OM3275	NEGATIVE	SNV
NTRK2 V617M	p.V617M	OM3274	NEGATIVE	mnp
NTRK3 F617L	p.F617L	OM3297	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3298	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3299	NEGATIVE	SNV
NTRK3 F617Y	p.F617Y	OM3311	NEGATIVE	SNV
NTRK3 G623R	p.G623R	COSM6951362	NEGATIVE	SNV
NTRK3 G623R	p.G623R	OM3253	NEGATIVE	SNV
NTRK3 G696A	p.G696A	OM3296	NEGATIVE	SNV
NTRK3 G696C	p.G696C	OM3294	NEGATIVE	mnp
NTRK3 G696C	p.G696C	OM3295	NEGATIVE	mnp
NTRK3 G696E	p.G696E	COSM5850004	NEGATIVE	SNV
NTRK3 V601F	p.V601F	OM3312	NEGATIVE	SNV
NTRK3 V601M	p.V601M	OM3300	NEGATIVE	mnp
PDGFRA A530_I543dup	p.A530_I543dup	COSM7002381	NEGATIVE	ins
PDGFRA D568N	p.D568N	OM3225	NEGATIVE	SNV
PDGFRA D842F	p.D842F	COSM29007	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA D842H	p.D842H	COSM734174	NEGATIVE	SNV
PDGFRA D842I	p.D842I	COSM12398	NEGATIVE	mnp
PDGFRA D842V	p.D842V	COSM736	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12396	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12397	NEGATIVE	mnp
PDGFRA D846Y	p.D846Y	COSM12399	NEGATIVE	SNV
PDGFRA G853D	p.G853D	OM3235	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3228	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3229	NEGATIVE	SNV
PDGFRA H845Y	p.H845Y	COSM96893	NEGATIVE	SNV
PDGFRA N659H	p.N659H	COSM51516	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22414	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22415	NEGATIVE	SNV
PDGFRA N659R	p.N659R	OM3230	NEGATIVE	mnp
PDGFRA N659S	p.N659S	COSM6960333	NEGATIVE	SNV
PDGFRA N659Y	p.N659Y	COSM22416	NEGATIVE	SNV
PDGFRA N848K	p.N848K	COSM28052	NEGATIVE	SNV
PDGFRA P577S	p.P577S	COSM6438084	NEGATIVE	SNV
PDGFRA Q579R	p.Q579R	OM3226	NEGATIVE	SNV
PDGFRA R841K	p.R841K	OM3233	NEGATIVE	SNV
PDGFRA S566_E571delinsR	p.S566_E571delinsR	COSM12418	NEGATIVE	del
PDGFRA V536E	p.V536E	COSM43059	NEGATIVE	SNV
PDGFRA V561A	p.V561A	COSM1430077	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA V561D	p.V561D	COSM739	NEGATIVE	SNV
PDGFRA W559_R560del	p.W559_R560del	OMINDEL700	NEGATIVE	del
PDGFRA Y555C	p.Y555C	COSM133645	NEGATIVE	SNV
PDGFRA Y849C	p.Y849C	COSM94952	NEGATIVE	SNV
PDGFRA Y849S	p.Y849S	OM3234	NEGATIVE	SNV
PDGFRA exon 18 deletion	p.D842_M844del	COSM96895	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_M844del	COSM12401	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842del	COSM12406	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_D846delinsN	COSM12411	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_I843delinsV	COSM405285	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_H845delinsY	COSM96896	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsRI	OMINDEL900	NEGATIVE	complex
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.I843del	COSM4969655	NEGATIVE	del
PDGFRA exon 18 deletion	p.I843_D846del	COSM12400	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_D846delinsE	COSM12408	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA exon 18 deletion	p.D842_S847delinsEA	COSM12415	NEGATIVE	complex
PDGFRA exon 18 deletion	p.I843_S847delinsT	COSM12407	NEGATIVE	del
PIK3CA A1046E	p.A1046E	COSM27156	NEGATIVE	SNV
PIK3CA A1046T	p.A1046T	COSM27273	NEGATIVE	SNV
PIK3CA A1046V	p.A1046V	COSM6146	NEGATIVE	SNV
PIK3CA C420R	p.C420R	COSM757	NEGATIVE	SNV
PIK3CA D1017H	p.D1017H	COSM27377	NEGATIVE	SNV
PIK3CA D1045G	p.D1045G	COSM1421003	NEGATIVE	SNV
PIK3CA D1045N	p.D1045N	COSM25086	NEGATIVE	SNV
PIK3CA D1045V	p.D1045V	COSM729813	NEGATIVE	SNV
PIK3CA D538G	p.D538G	COSM29123	NEGATIVE	SNV
PIK3CA D538N	p.D538N	COSM21467	NEGATIVE	SNV
PIK3CA D538Y	p.D538Y	COSM1716811	NEGATIVE	SNV
PIK3CA E109_I112delinsD	p.E109_I112delinsD	COSM445995	NEGATIVE	del
PIK3CA E110del	p.E110del	COSM24710	NEGATIVE	del
PIK3CA E39G	p.E39G	COSM1041451	NEGATIVE	SNV
PIK3CA E39K	p.E39K	COSM30625	NEGATIVE	SNV
PIK3CA E39V	p.E39V	COSM1420731	NEGATIVE	SNV
PIK3CA E418K	p.E418K	COSM12582	NEGATIVE	SNV
PIK3CA E542A	p.E542A	COSM1041494	NEGATIVE	SNV
PIK3CA E542G	p.E542G	COSM761	NEGATIVE	SNV
PIK3CA E542K	p.E542K	COSM760	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA E542Q	p.E542Q	COSM17442	NEGATIVE	SNV
PIK3CA E542R	p.E542R	COSM1659152	NEGATIVE	mnp
PIK3CA E542V	p.E542V	COSM762	NEGATIVE	SNV
PIK3CA E545A	p.E545A	COSM12458	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM27374	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM765	NEGATIVE	SNV
PIK3CA E545G	p.E545G	COSM764	NEGATIVE	SNV
PIK3CA E545K	p.E545K	COSM763	NEGATIVE	SNV
PIK3CA E545Q	p.E545Q	COSM27133	NEGATIVE	SNV
PIK3CA E545V	p.E545V	COSM27155	NEGATIVE	SNV
PIK3CA E547D	p.E547D	COSM729819	NEGATIVE	SNV
PIK3CA E547G	p.E547G	COSM1420864	NEGATIVE	SNV
PIK3CA E547K	p.E547K	COSM29315	NEGATIVE	SNV
PIK3CA E81A	p.E81A	COSM5347052	NEGATIVE	SNV
PIK3CA E81K	p.E81K	COSM27502	NEGATIVE	SNV
PIK3CA G1049A	p.G1049A	COSM27158	NEGATIVE	SNV
PIK3CA G1049C	p.G1049C	COSM1168056	NEGATIVE	SNV
PIK3CA G1049D	p.G1049D	COSM308548	NEGATIVE	SNV
PIK3CA G1049R	p.G1049R	COSM12597	NEGATIVE	SNV
PIK3CA G1049S	p.G1049S	COSM777	NEGATIVE	SNV
PIK3CA G106A	p.G106A	COSM166339	NEGATIVE	SNV
PIK3CA G106R	p.G106R	COSM86046	NEGATIVE	SNV
PIK3CA G106V	p.G106V	COSM748	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA G118D	p.G118D	COSM751	NEGATIVE	SNV
PIK3CA H1047L	p.H1047L	COSM776	NEGATIVE	SNV
PIK3CA H1047N	p.H1047N	COSM5029128	NEGATIVE	SNV
PIK3CA H1047P	p.H1047P	COSM249874	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM1041525	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM24714	NEGATIVE	SNV
PIK3CA H1047R	p.H1047R	COSM775	NEGATIVE	SNV
PIK3CA H1047Y	p.H1047Y	COSM774	NEGATIVE	SNV
PIK3CA H1048D	p.H1048D	COSM249881	NEGATIVE	SNV
PIK3CA H1048L	p.H1048L	COSM3734761	NEGATIVE	SNV
PIK3CA H1048N	p.H1048N	COSM1421010	NEGATIVE	SNV
PIK3CA H1048Q	p.H1048Q	COSM27157	NEGATIVE	SNV
PIK3CA H1048R	p.H1048R	COSM36289	NEGATIVE	SNV
PIK3CA H1048Y	p.H1048Y	COSM249875	NEGATIVE	SNV
PIK3CA K111E	p.K111E	COSM13570	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM12580	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM27505	NEGATIVE	SNV
PIK3CA K111R	p.K111R	COSM14052	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM29313	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM773	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM94984	NEGATIVE	SNV
PIK3CA M1043L	p.M1043L	COSM5731063	NEGATIVE	SNV
PIK3CA M1043T	p.M1043T	COSM12463	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA M1043V	p.M1043V	COSM12591	NEGATIVE	SNV
PIK3CA N1044D	p.N1044D	COSM27134	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM12592	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM27504	NEGATIVE	SNV
PIK3CA N1044S	p.N1044S	COSM22541	NEGATIVE	SNV
PIK3CA N1044Y	p.N1044Y	COSM36288	NEGATIVE	SNV
PIK3CA N345D	p.N345D	COSM1420788	NEGATIVE	SNV
PIK3CA N345H	p.N345H	COSM3846786	NEGATIVE	SNV
PIK3CA N345I	p.N345I	COSM94978	NEGATIVE	SNV
PIK3CA N345K	p.N345K	COSM754	NEGATIVE	SNV
PIK3CA N345S	p.N345S	COSM13476	NEGATIVE	SNV
PIK3CA N345T	p.N345T	COSM446002	NEGATIVE	SNV
PIK3CA P104L	p.P104L	COSM163485	NEGATIVE	SNV
PIK3CA P104R	p.P104R	COSM747	NEGATIVE	SNV
PIK3CA P104T	p.P104T	COSM1666660	NEGATIVE	SNV
PIK3CA P539R	p.P539R	COSM759	NEGATIVE	SNV
PIK3CA P539S	p.P539S	COSM249880	NEGATIVE	SNV
PIK3CA P539T	p.P539T	COSM41779	NEGATIVE	SNV
PIK3CA Q546E	p.Q546E	COSM6147	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM24712	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM5881064	NEGATIVE	SNV
PIK3CA Q546K	p.Q546K	COSM766	NEGATIVE	SNV
PIK3CA Q546L	p.Q546L	COSM25041	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Q546P	p.Q546P	COSM767	NEGATIVE	SNV
PIK3CA Q546R	p.Q546R	COSM12459	NEGATIVE	SNV
PIK3CA R108C	p.R108C	COSM6438095	NEGATIVE	SNV
PIK3CA R108H	p.R108H	COSM27497	NEGATIVE	SNV
PIK3CA R108L	p.R108L	COSM729828	NEGATIVE	SNV
PIK3CA R108P	p.R108P	COSM749	NEGATIVE	SNV
PIK3CA R38C	p.R38C	COSM744	NEGATIVE	SNV
PIK3CA R38G	p.R38G	COSM40945	NEGATIVE	SNV
PIK3CA R38H	p.R38H	COSM745	NEGATIVE	SNV
PIK3CA R38L	p.R38L	COSM729833	NEGATIVE	SNV
PIK3CA R38S	p.R38S	COSM87310	NEGATIVE	SNV
PIK3CA T1025A	p.T1025A	COSM771	NEGATIVE	SNV
PIK3CA T1025I	p.T1025I	COSM36285	NEGATIVE	SNV
PIK3CA T1025N	p.T1025N	COSM772	NEGATIVE	SNV
PIK3CA T1025P	p.T1025P	COSM6023925	NEGATIVE	SNV
PIK3CA T1025S	p.T1025S	COSM12590	NEGATIVE	SNV
PIK3CA V344A	p.V344A	COSM86951	NEGATIVE	SNV
PIK3CA V344E	p.V344E	COSM6746438	NEGATIVE	SNV
PIK3CA V344G	p.V344G	COSM22540	NEGATIVE	SNV
PIK3CA V344M	p.V344M	COSM253279	NEGATIVE	SNV
PIK3CA Y1021C	p.Y1021C	COSM12461	NEGATIVE	SNV
PIK3CA Y1021F	p.Y1021F	COSM328025	NEGATIVE	SNV
PIK3CA Y1021H	p.Y1021H	COSM17444	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Y1021N	p.Y1021N	COSM6143	NEGATIVE	SNV
PTEN A126D	p.A126D	COSM28884	NEGATIVE	SNV
PTEN A126G	p.A126G	COSM4391544	NEGATIVE	SNV
PTEN A126P	p.A126P	COSM5211	NEGATIVE	SNV
PTEN A126S	p.A126S	COSM28889	NEGATIVE	SNV
PTEN A126T	p.A126T	COSM5051	NEGATIVE	SNV
PTEN A126V	p.A126V	COSM5041	NEGATIVE	SNV
PTEN C136F	p.C136F	COSM5228	NEGATIVE	SNV
PTEN C136R	p.C136R	COSM5096	NEGATIVE	SNV
PTEN C136Y	p.C136Y	COSM12734	NEGATIVE	SNV
PTEN C71Y	p.C71Y	COSM5102	NEGATIVE	SNV
PTEN D24E	p.D24E	COSM5198	NEGATIVE	SNV
PTEN D24G	p.D24G	COSM5107	NEGATIVE	SNV
PTEN D24H	p.D24H	COSM1603823	NEGATIVE	SNV
PTEN D24N	p.D24N	COSM5079	NEGATIVE	SNV
PTEN D24V	p.D24V	COSM4016448	NEGATIVE	SNV
PTEN D24Y	p.D24Y	COSM5237	NEGATIVE	SNV
PTEN F341V	p.F341V	COSM5255	NEGATIVE	SNV
PTEN G127E	p.G127E	COSM5143	NEGATIVE	SNV
PTEN G129E	p.G129E	COSM28917	NEGATIVE	SNV
PTEN G129R	p.G129R	COSM246853	NEGATIVE	SNV
PTEN G129V	p.G129V	COSM5276	NEGATIVE	SNV
PTEN L181P	p.L181P	MCH17	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN L345Q	p.L345Q	COSM5213	NEGATIVE	SNV
PTEN N48K	p.N48K	COSM4392678	NEGATIVE	SNV
PTEN R130*	p.R130*	COSM5152	NEGATIVE	SNV
PTEN R130G	p.R130G	COSM5219	NEGATIVE	SNV
PTEN R130L	p.R130L	COSM5216	NEGATIVE	SNV
PTEN R130P	p.R130P	COSM5277	NEGATIVE	SNV
PTEN R130Q	p.R130Q	COSM5033	NEGATIVE	SNV
PTEN R173C	p.R173C	COSM5089	NEGATIVE	SNV
PTEN R173H	p.R173H	COSM5039	NEGATIVE	SNV
PTEN R173P	p.R173P	COSM12735	NEGATIVE	SNV
PTEN S170N	p.S170N	COSM5045	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH20	NEGATIVE	SNV
PTEN S170R	p.S170R	COSM6006454	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH21	NEGATIVE	SNV
PTEN T131I	p.T131I	COSM5104	NEGATIVE	SNV
PTEN V133I	p.V133I	COSM5044	NEGATIVE	SNV
PTEN V343E	p.V343E	MCH18	NEGATIVE	SNV
PTEN Y155C	p.Y155C	COSM5144	NEGATIVE	SNV
PTEN Y16C	p.Y16C	COSM5133	NEGATIVE	SNV
PTEN Y174C	p.Y174C	COSM5575601	NEGATIVE	SNV
PTEN Y174N	p.Y174N	COSM5221	NEGATIVE	SNV
RAF1 S257L	p.S257L	COSM181063	NEGATIVE	SNV
RAF1 S257P	p.S257P	ClinVar40600	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RAF1 S257W	p.S257W	COSM581519	NEGATIVE	SNV
RAF1 S259A	p.S259A	COSM20741	NEGATIVE	SNV
RAF1 S259C	p.S259C	COSM5719965	NEGATIVE	SNV
RAF1 S259F	p.S259F	COSM1536534	NEGATIVE	SNV
RAF1 S259P	p.S259P	COSM1418555	NEGATIVE	SNV
RAF1 S259T	p.S259T	ClinVar40601	NEGATIVE	SNV
RET A883F	p.A883F	COSM977	NEGATIVE	mnp
RET A883S	p.A883S	COSM133167	NEGATIVE	SNV
RET A883T	p.A883T	COSM100081	NEGATIVE	SNV
RET A883V	p.A883V	COSM4594153	NEGATIVE	SNV
RET A883Y	p.A883Y	OM3415	NEGATIVE	mnp
RET C618F	p.C618F	OM3401	NEGATIVE	SNV
RET C618G	p.C618G	COSM9214400	NEGATIVE	SNV
RET C618R	p.C618R	COSM29803	NEGATIVE	SNV
RET C618S	p.C618S	COSM87267	NEGATIVE	SNV
RET C618S	p.C618S	OM3402	NEGATIVE	SNV
RET C618T	p.C618T	OM3400	NEGATIVE	mnp
RET C618W	p.C618W	OM3403	NEGATIVE	SNV
RET C618Y	p.C618Y	COSM980	NEGATIVE	SNV
RET C620F	p.C620F	OM3405	NEGATIVE	SNV
RET C620G	p.C620G	OM3404	NEGATIVE	SNV
RET C620R	p.C620R	COSM29804	NEGATIVE	SNV
RET C620S	p.C620S	COSM5946160	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET C620S	p.C620S	COSM29805	NEGATIVE	SNV
RET C620W	p.C620W	COSM249815	NEGATIVE	SNV
RET C620Y	p.C620Y	COSM7403807	NEGATIVE	SNV
RET C630F	p.C630F	OM3406	NEGATIVE	SNV
RET C630G	p.C630G	COSM29806	NEGATIVE	SNV
RET C630R	p.C630R	COSM964	NEGATIVE	SNV
RET C630Y	p.C630Y	COSM9233214	NEGATIVE	SNV
RET C634F	p.C634F	COSM1237919	NEGATIVE	SNV
RET C634G	p.C634G	COSM1738369	NEGATIVE	SNV
RET C634L	p.C634L	OM3408	NEGATIVE	mnp
RET C634R	p.C634R	COSM966	NEGATIVE	SNV
RET C634S	p.C634S	COSM1237918	NEGATIVE	SNV
RET C634S	p.C634S	COSM1666664	NEGATIVE	SNV
RET C634W	p.C634W	COSM975	NEGATIVE	SNV
RET C634Y	p.C634Y	COSM974	NEGATIVE	SNV
RET C634_R635dup	p.C634_R635dup	OMINDEL1177	NEGATIVE	ins
RET C634_R635insHELC	p.C634_R635insHELC	OMINDEL1174	NEGATIVE	ins
RET C634_T636dup	p.C634_T636dup	COSM9213763	NEGATIVE	ins
RET D631Y	p.D631Y	COSM1738370	NEGATIVE	SNV
RET D631_C634del	p.D631_C634del	OMINDEL1172	NEGATIVE	del
RET D631_L633delinsE	p.D631_L633delinsE	COSM983	NEGATIVE	del
RET D631_L633delinsV	p.D631_L633delinsV	OMINDEL1175	NEGATIVE	del
RET D631del	p.D631del	OMINDEL1173	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET D898_E901del	p.D898_E901del	COSM962	NEGATIVE	del
RET E632K	p.E632K	COSM1702268	NEGATIVE	SNV
RET E632_C634delinsG	p.E632_C634delinsG	COSM973	NEGATIVE	del
RET E632_C634dup	p.E632_C634dup	OMINDEL1176	NEGATIVE	ins
RET E632_L633del	p.E632_L633del	COSM968	NEGATIVE	del
RET E632_L633delinsV	p.E632_L633delinsV	COSM982	NEGATIVE	del
RET E632_T636delinsSS	p.E632_T636delinsSS	COSM1048	NEGATIVE	complex
RET E768D	p.E768D	COSM21338	NEGATIVE	SNV
RET E768D	p.E768D	OM3413	NEGATIVE	SNV
RET E768G	p.E768G	COSM1347811	NEGATIVE	SNV
RET E768Q	p.E768Q	COSM1716312	NEGATIVE	SNV
RET E884K	p.E884K	COSM960	NEGATIVE	SNV
RET E884V	p.E884V	COSM1570338	NEGATIVE	SNV
RET G736A	p.G736A	OM3411	NEGATIVE	SNV
RET G810A	p.G810A	OM3290	NEGATIVE	SNV
RET G810C	p.G810C	OM3557	NEGATIVE	SNV
RET G810R	p.G810R	OM3289	NEGATIVE	SNV
RET G810S	p.G810S	OM3288	NEGATIVE	SNV
RET G810V	p.G810V	OM3558	NEGATIVE	SNV
RET G911D	p.G911D	COSM20888	NEGATIVE	SNV
RET I788N	p.I788N	OM3284	NEGATIVE	SNV
RET L730Q	p.L730Q	OM3409	NEGATIVE	SNV
RET L730R	p.L730R	OM3410	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET L760Q	p.L760Q	OM3412	NEGATIVE	SNV
RET L790F	p.L790F	rs75030001	NEGATIVE	SNV
RET L881V	p.L881V	OM3414	NEGATIVE	SNV
RET P628_L633del	p.P628_L633del	OMINDEL1171	NEGATIVE	del
RET R770Q	p.R770Q	COSM6006462	NEGATIVE	SNV
RET R886W	p.R886W	COSM6942691	NEGATIVE	SNV
RET R897*	p.R897*	COSM6983129	NEGATIVE	SNV
RET R912L	p.R912L	COSM188545	NEGATIVE	SNV
RET R912P	p.R912P	OM3419	NEGATIVE	SNV
RET R912Q	p.R912Q	COSM3437793	NEGATIVE	SNV
RET R912W	p.R912W	COSM3415038	NEGATIVE	SNV
RET S891A	p.S891A	rs75234356	NEGATIVE	SNV
RET S904C	p.S904C	OM3418	NEGATIVE	SNV
RET S904F	p.S904F	COSM6438204	NEGATIVE	SNV
RET V804G	p.V804G	OM3554	NEGATIVE	SNV
RET V804L	p.V804L	OM3285	NEGATIVE	SNV
RET V804L	p.V804L	OM3286	NEGATIVE	SNV
RET V804M	p.V804M	OM3195	NEGATIVE	SNV
RET V899_D903del	p.V899_D903del	COSM5991595	NEGATIVE	del
RET Y806C	p.Y806C	OM3287	NEGATIVE	SNV
RET Y806F	p.Y806F	OM3556	NEGATIVE	SNV
RET [D903E;S904P]	p.[D903E;S904P]	OM3416	NEGATIVE	mnp
RET [D903E;S904P]	p.[D903E;S904P]	OM3417	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET [E632D;L633V;C634R]	p.[E632D;L633V;C634R]	OM3407	NEGATIVE	mnp
RET [V882=;A883F]	p.[V882=;A883F]	COSM981	NEGATIVE	mnp
RET c.1880_1899delinsCTCAC	p.?	OMINDEL1170	NEGATIVE	complex
ROS1 D2033N	p.D2033N	COSM6924065	NEGATIVE	SNV
ROS1 G2032K	p.G2032K	OM3533	NEGATIVE	mnp
ROS1 G2032R	p.G2032R	MAN10	NEGATIVE	SNV
ROS1 G2032R	p.G2032R	MAN11	NEGATIVE	SNV
ROS1 L1951M	p.L1951M	COSM1072523	NEGATIVE	SNV
ROS1 L1951R	p.L1951R	OM3264	NEGATIVE	SNV
ROS1 L2026M	p.L2026M	BT151	NEGATIVE	SNV
ROS1 S1986F	p.S1986F	COSM6929870	NEGATIVE	SNV
ROS1 S1986Y	p.S1986Y	OM3170	NEGATIVE	SNV
STK11 A205T	p.A205T	COSM20953	NEGATIVE	SNV
STK11 D176N	p.D176N	COSM4827690	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3710	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3711	NEGATIVE	SNV
STK11 D194H	p.D194H	COSM5325765	NEGATIVE	SNV
STK11 D194N	p.D194N	COSM25847	NEGATIVE	SNV
STK11 D194V	p.D194V	COSM20957	NEGATIVE	SNV
STK11 D194Y	p.D194Y	COSM20944	NEGATIVE	SNV
STK11 D53Gfs*110	p.D53GfsTer110	COSM391251	NEGATIVE	ins
STK11 L290P	p.L290P	OM3719	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 L67P	p.L67P	OM3700	NEGATIVE	SNV
STK11 R304W	p.R304W	COSM29468	NEGATIVE	SNV
STK11 S240W	p.S240W	OM3714	NEGATIVE	SNV
STK11 c.1108+1G>A	p.?	OM3722	NEGATIVE	SNV
STK11 c.289_290+1delinsGAT	p.?	COSM6922318	NEGATIVE	mnp
STK11 c.289_290+2delAAGT	p.?	OMINDEL1000	NEGATIVE	del
STK11 c.290+1G>A	p.?	COSM6350392	NEGATIVE	SNV
STK11 c.290+1G>C	p.?	OM3701	NEGATIVE	SNV
STK11 c.290+1G>T	p.?	OM3702	NEGATIVE	SNV
STK11 c.290+1_290+2delGT	p.?	OMINDEL1001	NEGATIVE	del
STK11 c.290+2T>A	p.?	COSM6925006	NEGATIVE	SNV
STK11 c.290+2T>G	p.?	COSM25857	NEGATIVE	SNV
STK11 c.290+2delT	p.?	OMINDEL1002	NEGATIVE	del
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.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.375-1C>G	p.?	COSM4074282	NEGATIVE	SNV
STK11 c.375-1C>T	p.?	COSM6977173	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.375-2A>C	p.?	OM3706	NEGATIVE	SNV
STK11 c.375-2A>G	p.?	OM3707	NEGATIVE	SNV
STK11 c.375-2A>T	p.?	COSM6967966	NEGATIVE	SNV
STK11 c.375-2_386delACGTATATGGTGAT	p.?	OMINDEL1006	NEGATIVE	del
STK11 c.375-6_375-2delCCCCA	p.?	OMINDEL1005	NEGATIVE	del
STK11 c.465-1G>A	p.?	COSM25855	NEGATIVE	SNV
STK11 c.465-1G>C	p.?	OM3709	NEGATIVE	SNV
STK11 c.465-1G>T	p.?	COSM21570	NEGATIVE	SNV
STK11 c.465-1_465delinsTT	p.?	COSM6928792	NEGATIVE	mnp
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.465-2_465-1delAG	p.?	OMINDEL1009	NEGATIVE	del
STK11 c.465-2_465-1delinsTC	p.?	OM3708	NEGATIVE	mnp
STK11 c.465-4_472delGCAGGTACTTCT	p.?	OMINDEL1007	NEGATIVE	del
STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT	p.?	OMINDEL1008	NEGATIVE	del
STK11 c.598-1G>A	p.?	COSM3724562	NEGATIVE	SNV
STK11 c.598-1G>T	p.?	COSM51521	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.598-1_598delinsTT	p.?	OM3713	NEGATIVE	mnp
STK11 c.598-1_612delGGCACTGCACCCGTTC	p.?	OMINDEL1014	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.598-2_600delAGGCA	p.?	OMINDEL1012	NEGATIVE	del
STK11 c.727_734+9delGTCACCCTGTAAGTGCC	p.?	OMINDEL1015	NEGATIVE	del
STK11 c.731_734+9delCCCTGTAAGTGCC	p.?	OMINDEL1016	NEGATIVE	del
STK11 c.734+1G>A	p.?	COSM6916684	NEGATIVE	SNV
STK11 c.734+1G>C	p.?	COSM6970016	NEGATIVE	SNV
STK11 c.734+1G>T	p.?	COSM51523	NEGATIVE	SNV
STK11 c.734+2T>A	p.?	OM3715	NEGATIVE	SNV
STK11 c.734+2T>G	p.?	COSM6961577	NEGATIVE	SNV
STK11 c.734+2_734+10delTAAGTGCCC	p.?	OMINDEL1017	NEGATIVE	del
STK11 c.735-1G>A	p.?	COSM5732040	NEGATIVE	SNV
STK11 c.735-1G>T	p.?	COSM51522	NEGATIVE	SNV
STK11 c.735-2A>G	p.?	OM3716	NEGATIVE	SNV
STK11 c.735-2A>T	p.?	COSM25856	NEGATIVE	SNV
STK11	c.850_862+4delGACCTG			

p.?

OMINDEL1018

NEGATIVE

del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTGAAAGGTGG				
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+2T>A	p.?	COSM5576087	NEGATIVE	SNV
STK11 c.862+2T>G	p.?	OM3718	NEGATIVE	SNV
STK11 c.862+3delG	p.?	COSM8554973	NEGATIVE	del
STK11 c.863-1G>A	p.?	COSM6916725	NEGATIVE	SNV
STK11 c.863-1G>T	p.?	COSM6925330	NEGATIVE	SNV
STK11 c.863-2A>C	p.?	COSM6912582	NEGATIVE	SNV
STK11 c.863-2A>T	p.?	COSM401786	NEGATIVE	SNV
STK11 c.918_920+11delCAGGT GAGCGGCCC	p.?	OMINDEL1019	NEGATIVE	del
STK11 c.920+1G>A	p.?	OM3720	NEGATIVE	SNV
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
TP53 A138V	p.A138V	COSM43818	NEGATIVE	SNV
TP53 C135F	p.C135F	COSM10647	NEGATIVE	SNV
TP53 C135G	p.C135G	COSM44829	NEGATIVE	SNV
TP53 C135R	p.C135R	COSM10684	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44643	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44910	NEGATIVE	SNV
TP53 C135W	p.C135W	COSM44219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C135Y	p.C135Y	COSM10801	NEGATIVE	SNV
TP53 C141F	p.C141F	COSM44911	NEGATIVE	SNV
TP53 C141G	p.C141G	COSM45794	NEGATIVE	SNV
TP53 C141R	p.C141R	COSM43901	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM293910	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM44564	NEGATIVE	SNV
TP53 C141W	p.C141W	COSM44204	NEGATIVE	SNV
TP53 C141Y	p.C141Y	COSM43708	NEGATIVE	SNV
TP53 C176F	p.C176F	COSM10645	NEGATIVE	SNV
TP53 C176G	p.C176G	COSM44692	NEGATIVE	SNV
TP53 C176R	p.C176R	COSM44948	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44645	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44146	NEGATIVE	SNV
TP53 C176W	p.C176W	COSM11114	NEGATIVE	SNV
TP53 C176Y	p.C176Y	COSM10687	NEGATIVE	SNV
TP53 C238F	p.C238F	COSM43778	NEGATIVE	SNV
TP53 C238G	p.C238G	COSM46336	NEGATIVE	SNV
TP53 C238R	p.C238R	COSM44321	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM44653	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM43700	NEGATIVE	SNV
TP53 C238W	p.C238W	COSM44676	NEGATIVE	SNV
TP53 C238Y	p.C238Y	COSM11059	NEGATIVE	SNV
TP53 C242F	p.C242F	COSM10810	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C242G	p.C242G	COSM44135	NEGATIVE	SNV
TP53 C242R	p.C242R	COSM11738	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM11133	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM44935	NEGATIVE	SNV
TP53 C242W	p.C242W	COSM11356	NEGATIVE	SNV
TP53 C242Y	p.C242Y	COSM10646	NEGATIVE	SNV
TP53 C275F	p.C275F	COSM10701	NEGATIVE	SNV
TP53 C275G	p.C275G	COSM11501	NEGATIVE	SNV
TP53 C275R	p.C275R	COSM43902	NEGATIVE	SNV
TP53 C275S	p.C275S	COSM45413	NEGATIVE	SNV
TP53 C275W	p.C275W	COSM43823	NEGATIVE	SNV
TP53 C275Y	p.C275Y	COSM10893	NEGATIVE	SNV
TP53 C277F	p.C277F	COSM10749	NEGATIVE	SNV
TP53 D281A	p.D281A	COSM11665	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43837	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43906	NEGATIVE	SNV
TP53 D281G	p.D281G	COSM11232	NEGATIVE	SNV
TP53 D281H	p.D281H	COSM10943	NEGATIVE	SNV
TP53 D281N	p.D281N	COSM43596	NEGATIVE	SNV
TP53 D281V	p.D281V	COSM45729	NEGATIVE	SNV
TP53 D281Y	p.D281Y	COSM11516	NEGATIVE	SNV
TP53 E258A	p.E258A	COSM44719	NEGATIVE	SNV
TP53 E258D	p.E258D	COSM44962	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 E258D	p.E258D	COSM46410	NEGATIVE	SNV
TP53 E258G	p.E258G	COSM44168	NEGATIVE	SNV
TP53 E258K	p.E258K	COSM10988	NEGATIVE	SNV
TP53 E258Q	p.E258Q	COSM10751	NEGATIVE	SNV
TP53 E258V	p.E258V	COSM44450	NEGATIVE	SNV
TP53 E285A	p.E285A	COSM43614	NEGATIVE	SNV
TP53 E285G	p.E285G	COSM45649	NEGATIVE	SNV
TP53 E285K	p.E285K	COSM10722	NEGATIVE	SNV
TP53 E285Q	p.E285Q	COSM45138	NEGATIVE	SNV
TP53 E285V	p.E285V	COSM44227	NEGATIVE	SNV
TP53 E286A	p.E286A	COSM45892	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45439	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45492	NEGATIVE	SNV
TP53 E286G	p.E286G	COSM43565	NEGATIVE	SNV
TP53 E286K	p.E286K	COSM10726	NEGATIVE	SNV
TP53 E286Q	p.E286Q	COSM44250	NEGATIVE	SNV
TP53 E286V	p.E286V	COSM43936	NEGATIVE	SNV
TP53 G154V	p.G154V	COSM6815	NEGATIVE	SNV
TP53 G244A	p.G244A	COSM12013	NEGATIVE	SNV
TP53 G244C	p.G244C	COSM11524	NEGATIVE	SNV
TP53 G244D	p.G244D	COSM10883	NEGATIVE	SNV
TP53 G244R	p.G244R	COSM44221	NEGATIVE	SNV
TP53 G244S	p.G244S	COSM10941	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 G244V	p.G244V	COSM43652	NEGATIVE	SNV
TP53 G245A	p.G245A	COSM43965	NEGATIVE	SNV
TP53 G245C	p.G245C	COSM11081	NEGATIVE	SNV
TP53 G245D	p.G245D	COSM43606	NEGATIVE	SNV
TP53 G245R	p.G245R	COSM10957	NEGATIVE	SNV
TP53 G245S	p.G245S	COSM6932	NEGATIVE	SNV
TP53 G245V	p.G245V	COSM11196	NEGATIVE	SNV
TP53 G262V	p.G262V	COSM11198	NEGATIVE	SNV
TP53 G266A	p.G266A	COSM45488	NEGATIVE	SNV
TP53 G266E	p.G266E	COSM10867	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM10794	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM11205	NEGATIVE	SNV
TP53 G266V	p.G266V	COSM10958	NEGATIVE	SNV
TP53 G279E	p.G279E	COSM43714	NEGATIVE	SNV
TP53 H179D	p.H179D	COSM44776	NEGATIVE	SNV
TP53 H179L	p.H179L	COSM43635	NEGATIVE	SNV
TP53 H179N	p.H179N	COSM44151	NEGATIVE	SNV
TP53 H179P	p.H179P	COSM44218	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM11249	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM44214	NEGATIVE	SNV
TP53 H179R	p.H179R	COSM10889	NEGATIVE	SNV
TP53 H179Y	p.H179Y	COSM10768	NEGATIVE	SNV
TP53 H193D	p.H193D	COSM44002	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 H193L	p.H193L	COSM11066	NEGATIVE	SNV
TP53 H193N	p.H193N	COSM43935	NEGATIVE	SNV
TP53 H193P	p.H193P	COSM43833	NEGATIVE	SNV
TP53 H193R	p.H193R	COSM10742	NEGATIVE	SNV
TP53 H193Y	p.H193Y	COSM10672	NEGATIVE	SNV
TP53 H214D	p.H214D	COSM45115	NEGATIVE	SNV
TP53 H214L	p.H214L	COSM5632535	NEGATIVE	SNV
TP53 H214P	p.H214P	COSM1640841	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM44407	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM46272	NEGATIVE	SNV
TP53 H214R	p.H214R	COSM43687	NEGATIVE	SNV
TP53 H214Y	p.H214Y	COSM44112	NEGATIVE	SNV
TP53 I195F	p.I195F	COSM44633	NEGATIVE	SNV
TP53 I195L	p.I195L	COSM45773	NEGATIVE	SNV
TP53 I195M	p.I195M	COSM562637	NEGATIVE	SNV
TP53 I195N	p.I195N	COSM44877	NEGATIVE	SNV
TP53 I195S	p.I195S	COSM44539	NEGATIVE	SNV
TP53 I195T	p.I195T	COSM11089	NEGATIVE	SNV
TP53 K132E	p.K132E	COSM10813	NEGATIVE	SNV
TP53 K132M	p.K132M	COSM43592	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM10991	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM43963	NEGATIVE	SNV
TP53 K132Q	p.K132Q	COSM11224	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 K132R	p.K132R	COSM11582	NEGATIVE	SNV
TP53 K132T	p.K132T	COSM43912	NEGATIVE	SNV
TP53 L145P	p.L145P	COSM43899	NEGATIVE	SNV
TP53 L194F	p.L194F	COSM10995	NEGATIVE	SNV
TP53 L194H	p.L194H	COSM43623	NEGATIVE	SNV
TP53 L194I	p.L194I	COSM45330	NEGATIVE	SNV
TP53 L194P	p.L194P	COSM43827	NEGATIVE	SNV
TP53 L194R	p.L194R	COSM44571	NEGATIVE	SNV
TP53 L194V	p.L194V	COSM46117	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM10834	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM11063	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM44415	NEGATIVE	SNV
TP53 M237K	p.M237K	COSM43952	NEGATIVE	SNV
TP53 M237L	p.M237L	COSM44965	NEGATIVE	SNV
TP53 M237R	p.M237R	COSM45032	NEGATIVE	SNV
TP53 M237T	p.M237T	COSM45329	NEGATIVE	SNV
TP53 M237V	p.M237V	COSM44525	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM10757	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM44310	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM46136	NEGATIVE	SNV
TP53 M246K	p.M246K	COSM44103	NEGATIVE	SNV
TP53 M246L	p.M246L	COSM45992	NEGATIVE	SNV
TP53 M246R	p.M246R	COSM11376	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 M246T	p.M246T	COSM11355	NEGATIVE	SNV
TP53 M246V	p.M246V	COSM43555	NEGATIVE	SNV
TP53 N239D	p.N239D	COSM10777	NEGATIVE	SNV
TP53 N239I	p.N239I	COSM307333	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44510	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44647	NEGATIVE	SNV
TP53 N239S	p.N239S	COSM44094	NEGATIVE	SNV
TP53 N239T	p.N239T	COSM43801	NEGATIVE	SNV
TP53 N239Y	p.N239Y	COSM45870	NEGATIVE	SNV
TP53 P151A	p.P151A	COSM44944	NEGATIVE	SNV
TP53 P151H	p.P151H	COSM11476	NEGATIVE	SNV
TP53 P151L	p.P151L	COSM44288	NEGATIVE	SNV
TP53 P151R	p.P151R	COSM44003	NEGATIVE	SNV
TP53 P151S	p.P151S	COSM10905	NEGATIVE	SNV
TP53 P151T	p.P151T	COSM43911	NEGATIVE	SNV
TP53 P152A	p.P152A	COSM44788	NEGATIVE	SNV
TP53 P152L	p.P152L	COSM10790	NEGATIVE	SNV
TP53 P152Q	p.P152Q	COSM44613	NEGATIVE	SNV
TP53 P152R	p.P152R	COSM45505	NEGATIVE	SNV
TP53 P152S	p.P152S	COSM43582	NEGATIVE	SNV
TP53 P152T	p.P152T	COSM44561	NEGATIVE	SNV
TP53 P177_C182del	p.P177_C182del	COSM43570	NEGATIVE	del
TP53 P190L	p.P190L	COSM43657	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 P191del	p.P191del	COSM45140	NEGATIVE	del
TP53 P250L	p.P250L	COSM10771	NEGATIVE	SNV
TP53 P278A	p.P278A	COSM10814	NEGATIVE	SNV
TP53 P278H	p.P278H	COSM43755	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 Q192*	p.Q192*	COSM10733	NEGATIVE	SNV
TP53 R156P	p.R156P	COSM10760	NEGATIVE	SNV
TP53 R175C	p.R175C	COSM43680	NEGATIVE	SNV
TP53 R175G	p.R175G	COSM10870	NEGATIVE	SNV
TP53 R175H	p.R175H	COSM10648	NEGATIVE	SNV
TP53 R175L	p.R175L	COSM10718	NEGATIVE	SNV
TP53 R175P	p.R175P	COSM45416	NEGATIVE	SNV
TP53 R175S	p.R175S	COSM43931	NEGATIVE	SNV
TP53 R196*	p.R196*	COSM10705	NEGATIVE	SNV
TP53 R213*	p.R213*	COSM10654	NEGATIVE	SNV
TP53 R213G	p.R213G	COSM44102	NEGATIVE	SNV
TP53 R213L	p.R213L	COSM43650	NEGATIVE	SNV
TP53 R213P	p.R213P	COSM11860	NEGATIVE	SNV
TP53 R213Q	p.R213Q	COSM10735	NEGATIVE	SNV
TP53 R248G	p.R248G	COSM11564	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R248L	p.R248L	COSM6549	NEGATIVE	SNV
TP53 R248P	p.R248P	COSM11491	NEGATIVE	SNV
TP53 R248Q	p.R248Q	COSM10662	NEGATIVE	SNV
TP53 R248W	p.R248W	COSM10656	NEGATIVE	SNV
TP53 R249G	p.R249G	COSM10668	NEGATIVE	SNV
TP53 R249K	p.R249K	COSM44091	NEGATIVE	SNV
TP53 R249M	p.R249M	COSM43871	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10785	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10817	NEGATIVE	SNV
TP53 R249T	p.R249T	COSM43665	NEGATIVE	SNV
TP53 R249W	p.R249W	COSM43629	NEGATIVE	SNV
TP53 R273C	p.R273C	COSM10659	NEGATIVE	SNV
TP53 R273G	p.R273G	COSM43843	NEGATIVE	SNV
TP53 R273H	p.R273H	COSM10660	NEGATIVE	SNV
TP53 R273L	p.R273L	COSM10779	NEGATIVE	SNV
TP53 R273P	p.R273P	COSM43896	NEGATIVE	SNV
TP53 R273S	p.R273S	COSM43909	NEGATIVE	SNV
TP53 R280G	p.R280G	COSM11123	NEGATIVE	SNV
TP53 R280I	p.R280I	COSM11287	NEGATIVE	SNV
TP53 R280K	p.R280K	COSM10728	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44171	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44233	NEGATIVE	SNV
TP53 R280T	p.R280T	COSM10724	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R282G	p.R282G	COSM10992	NEGATIVE	SNV
TP53 R282L	p.R282L	COSM44470	NEGATIVE	SNV
TP53 R282P	p.R282P	COSM44306	NEGATIVE	SNV
TP53 R282Q	p.R282Q	COSM44338	NEGATIVE	SNV
TP53 R282W	p.R282W	COSM10704	NEGATIVE	SNV
TP53 R283P	p.R283P	COSM10743	NEGATIVE	SNV
TP53 S240G	p.S240G	COSM43973	NEGATIVE	SNV
TP53 S241A	p.S241A	COSM44224	NEGATIVE	SNV
TP53 S241C	p.S241C	COSM10709	NEGATIVE	SNV
TP53 S241F	p.S241F	COSM10812	NEGATIVE	SNV
TP53 S241P	p.S241P	COSM44578	NEGATIVE	SNV
TP53 S241T	p.S241T	COSM44067	NEGATIVE	SNV
TP53 S241Y	p.S241Y	COSM10935	NEGATIVE	SNV
TP53 V157F	p.V157F	COSM10670	NEGATIVE	SNV
TP53 V157L	p.V157L	COSM45120	NEGATIVE	SNV
TP53 V172F	p.V172F	COSM44240	NEGATIVE	SNV
TP53 V173A	p.V173A	COSM44327	NEGATIVE	SNV
TP53 V173E	p.V173E	COSM45731	NEGATIVE	SNV
TP53 V173G	p.V173G	COSM44383	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM43559	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM44057	NEGATIVE	SNV
TP53 V173M	p.V173M	COSM11084	NEGATIVE	SNV
TP53 V197M	p.V197M	COSM43779	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 V216A	p.V216A	COSM44567	NEGATIVE	SNV
TP53 V216E	p.V216E	COSM44274	NEGATIVE	SNV
TP53 V216G	p.V216G	COSM43681	NEGATIVE	SNV
TP53 V216L	p.V216L	COSM11210	NEGATIVE	SNV
TP53 V216M	p.V216M	COSM10667	NEGATIVE	SNV
TP53 V272A	p.V272A	COSM44294	NEGATIVE	SNV
TP53 V272E	p.V272E	COSM44580	NEGATIVE	SNV
TP53 V272G	p.V272G	COSM44870	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM10859	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM45898	NEGATIVE	SNV
TP53 V272M	p.V272M	COSM10891	NEGATIVE	SNV
TP53 V274F	p.V274F	COSM10769	NEGATIVE	SNV
TP53 V274L	p.V274L	COSM44443	NEGATIVE	SNV
TP53 Y220C	p.Y220C	COSM10758	NEGATIVE	SNV
TP53 Y220D	p.Y220D	COSM11847	NEGATIVE	SNV
TP53 Y220F	p.Y220F	COSM1564197	NEGATIVE	SNV
TP53 Y220H	p.Y220H	COSM44637	NEGATIVE	SNV
TP53 Y220N	p.Y220N	COSM44672	NEGATIVE	SNV
TP53 Y220S	p.Y220S	COSM43850	NEGATIVE	SNV
TP53 Y234C	p.Y234C	COSM10725	NEGATIVE	SNV
TP53 Y234D	p.Y234D	COSM43768	NEGATIVE	SNV
TP53 Y234F	p.Y234F	COSM44315	NEGATIVE	SNV
TP53 Y234H	p.Y234H	COSM11152	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 Y234N	p.Y234N	COSM43956	NEGATIVE	SNV
TP53 Y234S	p.Y234S	COSM43865	NEGATIVE	SNV
TP53 Y236C	p.Y236C	COSM10731	NEGATIVE	SNV
TP53 Y236D	p.Y236D	COSM43602	NEGATIVE	SNV
TP53 Y236F	p.Y236F	COSM3958809	NEGATIVE	SNV
TP53 Y236H	p.Y236H	COSM44326	NEGATIVE	SNV
TP53 Y236N	p.Y236N	COSM43826	NEGATIVE	SNV
TP53 Y236S	p.Y236S	COSM44693	NEGATIVE	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
A2M-ALK fusion	A2M(22) - ALK(19)	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
AGAP1-ALK fusion	AGAP1(6) - ALK(18)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATIC-ALK fusion	ATIC(7) - ALK(20)	Fusion	NEGATIVE
ATRNL1-ALK fusion	ATRNL1(19) - ALK(20)	Fusion	NEGATIVE
BCL11A-ALK fusion	BCL11A(4) - ALK(20)	Fusion	NEGATIVE
BEND5-ALK fusion	BEND5(3) - ALK(20)	Fusion	NEGATIVE
BIRC6-ALK fusion	BIRC6(10) - ALK(20)	Fusion	NEGATIVE
CAD-ALK fusion	CAD(35) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CAD-ALK fusion	CAD(36) - ALK(20)	Fusion	NEGATIVE
CAMKMT-ALK fusion	CAMKMT(3) - ALK(20)	Fusion	NEGATIVE
CARS-ALK fusion	CARS(17) - ALK(20)	Fusion	NEGATIVE
CCDC149-ALK fusion	CCDC149(10) - ALK(20)	Fusion	NEGATIVE
CCDC61-ALK fusion	CCDC61(13) - ALK(20)	Fusion	NEGATIVE
CCDC88A-ALK fusion	CCDC88A(12) - ALK(20)	Fusion	NEGATIVE
CEBPZ-ALK fusion	CEBPZ(2) - ALK(20)	Fusion	NEGATIVE
CENPF-ALK fusion	CENPF(11) - ALK(20)	Fusion	NEGATIVE
CLIP1-ALK fusion	CLIP1(13) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(10) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(14) - ALK(17)	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
COL25A1-ALK fusion	COL25A1(3) - ALK(20)	Fusion	NEGATIVE
CPSF7-ALK fusion	CPSF7(5) - ALK(20)	Fusion	NEGATIVE
CREB1-ALK fusion	CREB1(1) - ALK(9)	Fusion	NEGATIVE
CTNND1-ALK fusion	CTNND1(16) - ALK(20)	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
DDX6-ALK fusion	DDX6(2) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DES-ALK fusion	DES(7) - ALK(19)	Fusion	NEGATIVE
DMD-ALK fusion	DMD(55) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(8) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(6) - ALK(20)	Fusion	NEGATIVE
EIF2AK3-ALK fusion	EIF2AK3(2) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(17) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(2) - 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(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(3) - 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(19)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(18)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(17)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(16)	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

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(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(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(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(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

Variant Name	Genes (Exons)	Variant Type	Test Result
EML4-ALK fusion	EML4(22) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(4) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(5) - ALK(20)	Fusion	NEGATIVE
FBXO36-ALK fusion	FBXO36(2) - 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
GALNT14-ALK fusion	GALNT14(1) - ALK(19)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(13) - ALK(20)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(19) - 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
HNRNPA1-ALK fusion	HNRNPA1(2) - ALK(18)	Fusion	NEGATIVE
IGFBP5-ALK fusion	IGFBP5(1) - ALK(19)	Fusion	NEGATIVE
INTS9-ALK fusion	INTS9(8) - ALK(20)	Fusion	NEGATIVE
ITSN2-ALK fusion	ITSN2(14) - ALK(2)	Fusion	NEGATIVE
KANK2-ALK fusion	KANK2(4) - ALK(16)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KCNQ5-ALK fusion	KCNQ5(1) - ALK(10)	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
KLC1-ALK fusion	KLC1(9) - ALK(20)	Fusion	NEGATIVE
KTN1-ALK fusion	KTN1(43) - ALK(19)	Fusion	NEGATIVE
LMNA-ALK fusion	LMNA(2) - ALK(20)	Fusion	NEGATIVE
LMO7-ALK fusion	LMO7(15) - ALK(20)	Fusion	NEGATIVE
LRPPRC-ALK fusion	LRPPRC(23) - ALK(20)	Fusion	NEGATIVE
LRRFIP2-ALK fusion	LRRFIP2(26) - ALK(20)	Fusion	NEGATIVE
LTBP1-ALK fusion	LTBP1(19) - ALK(20)	Fusion	NEGATIVE
MALAT1-ALK fusion	MALAT1(1) - ALK(16)	Fusion	NEGATIVE
MANBA-ALK fusion	MANBA(14) - ALK(20)	Fusion	NEGATIVE
MAPRE3-ALK fusion	MAPRE3(7) - ALK(20)	Fusion	NEGATIVE
MCFD2-ALK fusion	MCFD2(1) - ALK(20)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(2) - ALK(7)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(1) - ALK(2)	Fusion	NEGATIVE
MLPH-ALK fusion	MLPH(11) - ALK(20)	Fusion	NEGATIVE
MPRIP-ALK fusion	MPRIP(21) - ALK(20)	Fusion	NEGATIVE
MROH2B-ALK fusion	MROH2B(21) - ALK(12)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(9) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MYH10-ALK fusion	MYH10(38) - 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
MYO5C-ALK fusion	MYO5C(30) - ALK(20)	Fusion	NEGATIVE
MYT1L-ALK fusion	MYT1L(14) - ALK(20)	Fusion	NEGATIVE
NCOA1-ALK fusion	NCOA1(21) - ALK(1)	Fusion	NEGATIVE
NPM1-ALK fusion	NPM1(4) - ALK(20)	Fusion	NEGATIVE
NUMA1-ALK fusion	NUMA1(19) - ALK(20)	Fusion	NEGATIVE
OTOF-ALK fusion	OTOF(2) - ALK(3)	Fusion	NEGATIVE
PABPC1-ALK fusion	PABPC1(9) - ALK(20)	Fusion	NEGATIVE
PCM1-ALK fusion	PCM1(23) - ALK(20)	Fusion	NEGATIVE
PDE3A-ALK fusion	PDE3A(10) - ALK(8)	Fusion	NEGATIVE
PDE8B-ALK fusion	PDE8B(11) - ALK(20)	Fusion	NEGATIVE
PHACTR1-ALK fusion	PHACTR1(4) - ALK(20)	Fusion	NEGATIVE
PLEKHA7-ALK fusion	PLEKHA7(26) - ALK(19)	Fusion	NEGATIVE
PNPT1-ALK fusion	PNPT1(5) - ALK(4)	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
PPM1B-ALK fusion	PPM1B(1) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
PPP1R21-ALK fusion	PPP1R21(7) - ALK(20)	Fusion	NEGATIVE
PPP1R21-ALK fusion	PPP1R21(17) - ALK(20)	Fusion	NEGATIVE
PPP4R3B-ALK fusion	PPP4R3B(10) - ALK(2)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(2) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(5) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(10) - ALK(20)	Fusion	NEGATIVE
PRKAR1B-ALK fusion	PRKAR1B(4) - ALK(20)	Fusion	NEGATIVE
PRKAR2A-ALK fusion	PRKAR2A(2) - ALK(20)	Fusion	NEGATIVE
PRKCE-ALK fusion	PRKCE(1) - ALK(20)	Fusion	NEGATIVE
RANBP2-ALK fusion	RANBP2(18) - ALK(20)	Fusion	NEGATIVE
RNF213-ALK fusion	RNF213(20) - ALK(20)	Fusion	NEGATIVE
RRBP1-ALK fusion	RRBP1(20) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(6) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(5) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(22) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(21) - ALK(20)	Fusion	NEGATIVE
SFT2D1-ALK fusion	SFT2D1(1) - ALK(6)	Fusion	NEGATIVE
SLC34A2-ALK fusion	SLC34A2(1) - ALK(20)	Fusion	NEGATIVE
SLC8A1-ALK fusion	SLC8A1(1) - ALK(20)	Fusion	NEGATIVE
SLMAP-ALK fusion	SLMAP(11) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SP100-ALK fusion	SP100(4) - ALK(20)	Fusion	NEGATIVE
SPDYA-ALK fusion	SPDYA(6) - ALK(20)	Fusion	NEGATIVE
SPECC1L-ALK fusion	SPECC1L(9) - 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
SQSTM1-ALK fusion	SQSTM1(5) - ALK(20)	Fusion	NEGATIVE
SQSTM1-ALK fusion	SQSTM1(5) - ALK(19)	Fusion	NEGATIVE
STK32B-ALK fusion	STK32B(11) - ALK(20)	Fusion	NEGATIVE
STRN-ALK fusion	STRN(3) - ALK(20)	Fusion	NEGATIVE
TERT-ALK fusion	TERT(11) - ALK(5)	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(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(5) - ALK(19)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(6) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(19)	Fusion	NEGATIVE
THBS1-ALK fusion	THBS1(4) - ALK(19)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(20)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(19)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(32) - ALK(13)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(28) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(20)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(16) - ALK(17)	Fusion	NEGATIVE
TPM1-ALK fusion	TPM1(8) - ALK(20)	Fusion	NEGATIVE
TPM3-ALK fusion	TPM3(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(15) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(4) - ALK(20)	Fusion	NEGATIVE
TRAF1-ALK fusion	TRAF1(6) - ALK(20)	Fusion	NEGATIVE
TRMT61B-ALK fusion	TRMT61B(1) - ALK(9)	Fusion	NEGATIVE
VCL-ALK fusion	VCL(16) - ALK(20)	Fusion	NEGATIVE
YPEL5-ALK fusion	YPEL5(1) - ALK(18)	Fusion	NEGATIVE
ZNF2-ALK fusion	ZNF2(1) - ALK(20)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(11)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(9)	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
AGK-BRAF fusion	AGK(2) - BRAF(8)	Fusion	NEGATIVE
AGTRAP-BRAF fusion	AGTRAP(5) - BRAF(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
AP3B1-BRAF fusion	AP3B1(22) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(3) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(4) - BRAF(11)	Fusion	NEGATIVE
ATG7-BRAF fusion	ATG7(18) - BRAF(9)	Fusion	NEGATIVE
BAIAP2L1-BRAF fusion	BAIAP2L1(12) - BRAF(9)	Fusion	NEGATIVE
BBS9-BRAF fusion	BBS9(19) - BRAF(4)	Fusion	NEGATIVE
BCAP29-BRAF fusion	BCAP29(7) - BRAF(11)	Fusion	NEGATIVE
BCL2L11-BRAF fusion	BCL2L11(3) - BRAF(10)	Fusion	NEGATIVE
BTF3L4-BRAF fusion	BTF3L4(3) - BRAF(11)	Fusion	NEGATIVE
C7orf73-BRAF fusion	C7orf73(2) - BRAF(9)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(2) - BRAF(8)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(1) - BRAF(9)	Fusion	NEGATIVE
CCDC91-BRAF fusion	CCDC91(11) - BRAF(9)	Fusion	NEGATIVE
CCNY-BRAF fusion	CCNY(1) - BRAF(10)	Fusion	NEGATIVE
CDC27-BRAF fusion	CDC27(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CDC42BPB-BRAF fusion	CDC42BPB(36) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(8)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(12) - BRAF(11)	Fusion	NEGATIVE
CEP89-BRAF fusion	CEP89(16) - BRAF(9)	Fusion	NEGATIVE
CLCN6-BRAF fusion	CLCN6(2) - BRAF(11)	Fusion	NEGATIVE
CLIP2-BRAF fusion	CLIP2(6) - BRAF(11)	Fusion	NEGATIVE
COBL-BRAF fusion	COBL(7) - BRAF(10)	Fusion	NEGATIVE
CUL1-BRAF fusion	CUL1(7) - BRAF(9)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(8) - BRAF(8)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(10) - BRAF(9)	Fusion	NEGATIVE
DGKI-BRAF fusion	DGKI(7) - BRAF(10)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(5) - BRAF(9)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(3) - BRAF(10)	Fusion	NEGATIVE
DOCK4-BRAF fusion	DOCK4(41) - BRAF(9)	Fusion	NEGATIVE
DSTYK-BRAF fusion	DSTYK(1) - BRAF(10)	Fusion	NEGATIVE
DYNC1I2-BRAF fusion	DYNC1I2(7) - BRAF(10)	Fusion	NEGATIVE
EML4-BRAF fusion	EML4(6) - BRAF(10)	Fusion	NEGATIVE
EPS15-BRAF fusion	EPS15(22) - BRAF(10)	Fusion	NEGATIVE
EPS15L1-BRAF fusion	EPS15L1(16) - BRAF(9)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(12) - BRAF(10)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(17) - BRAF(8)	Fusion	NEGATIVE
ESYT2-BRAF fusion	ESYT2(8) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FAM114A2-BRAF fusion	FAM114A2(9) - BRAF(11)	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
FAM73A-BRAF fusion	FAM73A(7) - BRAF(9)	Fusion	NEGATIVE
FCHSD1-BRAF fusion	FCHSD1(13) - BRAF(9)	Fusion	NEGATIVE
FXR1-BRAF fusion	FXR1(13) - BRAF(10)	Fusion	NEGATIVE
GATM-BRAF fusion	GATM(2) - BRAF(11)	Fusion	NEGATIVE
GHR-BRAF fusion	GHR(1) - BRAF(10)	Fusion	NEGATIVE
GIPC2-BRAF fusion	GIPC2(3) - BRAF(10)	Fusion	NEGATIVE
GIT2-BRAF fusion	GIT2(14) - BRAF(9)	Fusion	NEGATIVE
GNAI1-BRAF fusion	GNAI1(1) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(4) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(20) - BRAF(10)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(4) - BRAF(7)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(6) - BRAF(7)	Fusion	NEGATIVE
KANK1-BRAF fusion	KANK1(7) - BRAF(9)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(3) - BRAF(8)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(4) - BRAF(8)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(12) - BRAF(4)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(11) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(18) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(17) - BRAF(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(10)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(8)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(8)	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
KLHL7-BRAF fusion	KLHL7(4) - BRAF(10)	Fusion	NEGATIVE
KLHL7-BRAF fusion	KLHL7(5) - BRAF(9)	Fusion	NEGATIVE
LMO7-BRAF fusion	LMO7(7) - BRAF(9)	Fusion	NEGATIVE
LSM12-BRAF fusion	LSM12(3) - BRAF(9)	Fusion	NEGATIVE
LSM14A-BRAF fusion	LSM14A(9) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(5) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(60) - BRAF(9)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(17) - BRAF(10)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MBP-BRAF fusion	MBP(2) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(11)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(10)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(8)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(3) - BRAF(10)	Fusion	NEGATIVE
MYRIP-BRAF fusion	MYRIP(16) - BRAF(9)	Fusion	NEGATIVE
MZT1-BRAF fusion	MZT1(2) - BRAF(11)	Fusion	NEGATIVE
NFIC-BRAF fusion	NFIC(6) - BRAF(10)	Fusion	NEGATIVE
NRF1-BRAF fusion	NRF1(5) - BRAF(9)	Fusion	NEGATIVE
NUB1-BRAF fusion	NUB1(3) - BRAF(9)	Fusion	NEGATIVE
NUDCD3-BRAF fusion	NUDCD3(4) - BRAF(9)	Fusion	NEGATIVE
NUP214-BRAF fusion	NUP214(21) - BRAF(10)	Fusion	NEGATIVE
OSBPL1A-BRAF fusion	OSBPL1A(16) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(11) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(14) - BRAF(9)	Fusion	NEGATIVE
PACSIN2-BRAF fusion	PACSIN2(7) - BRAF(9)	Fusion	NEGATIVE
PAPSS1-BRAF fusion	PAPSS1(5) - BRAF(9)	Fusion	NEGATIVE
PARP12-BRAF fusion	PARP12(9) - BRAF(11)	Fusion	NEGATIVE
PCM1-BRAF fusion	PCM1(37) - BRAF(9)	Fusion	NEGATIVE
PHTF2-BRAF fusion	PHTF2(17) - BRAF(9)	Fusion	NEGATIVE
PICALM-BRAF fusion	PICALM(19) - BRAF(9)	Fusion	NEGATIVE
PJA2-BRAF fusion	PJA2(7) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PLIN3-BRAF fusion	PLIN3(1) - BRAF(9)	Fusion	NEGATIVE
PLXNA2-BRAF fusion	PLXNA2(9) - BRAF(2)	Fusion	NEGATIVE
POR-BRAF fusion	POR(3) - BRAF(11)	Fusion	NEGATIVE
PPFIBP2-BRAF fusion	PPFIBP2(3) - BRAF(11)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(10) - BRAF(9)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(9) - BRAF(9)	Fusion	NEGATIVE
PRKAR2B-BRAF fusion	PRKAR2B(1) - BRAF(10)	Fusion	NEGATIVE
RAD18-BRAF fusion	RAD18(7) - BRAF(10)	Fusion	NEGATIVE
RBM33-BRAF fusion	RBM33(3) - BRAF(11)	Fusion	NEGATIVE
RBMS3-BRAF fusion	RBMS3(11) - BRAF(11)	Fusion	NEGATIVE
RNF11-BRAF fusion	RNF11(1) - BRAF(11)	Fusion	NEGATIVE
RNF130-BRAF fusion	RNF130(3) - BRAF(9)	Fusion	NEGATIVE
RP2-BRAF fusion	RP2(3) - BRAF(10)	Fusion	NEGATIVE
RUNDC1-BRAF fusion	RUNDC1(1) - BRAF(9)	Fusion	NEGATIVE
SCRIB-BRAF fusion	SCRIB(27) - BRAF(11)	Fusion	NEGATIVE
SLC12A7-BRAF fusion	SLC12A7(17) - BRAF(11)	Fusion	NEGATIVE
SLC45A3-BRAF fusion	SLC45A3(1) - 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(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(9) - BRAF(8)	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(11)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(8)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(6) - BRAF(9)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(5) - BRAF(9)	Fusion	NEGATIVE
STRN3-BRAF fusion	STRN3(3) - BRAF(10)	Fusion	NEGATIVE
STXBP6-BRAF fusion	STXBP6(4) - BRAF(10)	Fusion	NEGATIVE
TANK-BRAF fusion	TANK(4) - BRAF(9)	Fusion	NEGATIVE
TAX1BP1-BRAF fusion	TAX1BP1(8) - BRAF(11)	Fusion	NEGATIVE
TMEM106B-BRAF fusion	TMEM106B(4) - BRAF(8)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(1) - BRAF(10)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(2) - BRAF(9)	Fusion	NEGATIVE
TMPRSS2-BRAF fusion	TMPRSS2(3) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TOM1L2-BRAF fusion	TOM1L2(11) - BRAF(9)	Fusion	NEGATIVE
TPR-BRAF fusion	TPR(14) - BRAF(7)	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
TRIM4-BRAF fusion	TRIM4(6) - BRAF(10)	Fusion	NEGATIVE
UBN2-BRAF fusion	UBN2(3) - BRAF(11)	Fusion	NEGATIVE
ZBTB8A-BRAF fusion	ZBTB8A(4) - BRAF(9)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(7) - BRAF(11)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(3) - BRAF(10)	Fusion	NEGATIVE
ZKSCAN5-BRAF fusion	ZKSCAN5(2) - BRAF(9)	Fusion	NEGATIVE
ZNF207-BRAF fusion	ZNF207(3) - BRAF(10)	Fusion	NEGATIVE
ZNF767P-BRAF fusion	ZNF767P(1) - BRAF(11)	Fusion	NEGATIVE
ZSCAN30-BRAF fusion	ZSCAN30(3) - BRAF(10)	Fusion	NEGATIVE
EGFRvIII	EGFR(1) - EGFR(8)	RNAExonVariant	NEGATIVE
ESR1-ARMT1 fusion	ESR1(3) - ARMT1(4)	Fusion	NEGATIVE
ESR1-ARMT1 fusion	ESR1(6) - ARMT1(2)	Fusion	NEGATIVE
ESR1-ARNT2 fusion	ESR1(7) - ARNT2(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-ATP2B2 fusion	ESR1(4) - ATP2B2(4)	Fusion	NEGATIVE
ESR1-BMPER fusion	ESR1(6) - BMPER(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(2)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(5)	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(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(10)	Fusion	NEGATIVE
ESR1-CDK13 fusion	ESR1(7) - CDK13(13)	Fusion	NEGATIVE
ESR1-COA5 fusion	ESR1(4) - COA5(2)	Fusion	NEGATIVE
ESR1-DAB2 fusion	ESR1(6) - DAB2(3)	Fusion	NEGATIVE
ESR1-GYG2 fusion	ESR1(6) - GYG2(2)	Fusion	NEGATIVE
ESR1-MKL1 fusion	ESR1(6) - MKL1(7)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(6) - MTHFD1L(21)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(7) - MTHFD1L(27)	Fusion	NEGATIVE
ESR1-NKAIN2 fusion	ESR1(6) - NKAIN2(2)	Fusion	NEGATIVE
ESR1-NOP2 fusion	ESR1(6) - NOP2(16)	Fusion	NEGATIVE
ESR1-PCDH11X fusion	ESR1(6) - PCDH11X(6)	Fusion	NEGATIVE
ESR1-PDE10A fusion	ESR1(7) - PDE10A(6)	Fusion	NEGATIVE
ESR1-PLEKHG1 fusion	ESR1(6) - PLEKHG1(12)	Fusion	NEGATIVE
ESR1-POLH fusion	ESR1(6) - POLH(2)	Fusion	NEGATIVE
ESR1-RMND1 fusion	ESR1(6) - RMND1(9)	Fusion	NEGATIVE
ESR1-SOX9 fusion	ESR1(6) - SOX9(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-SYNE1 fusion	ESR1(6) - SYNE1(59)	Fusion	NEGATIVE
ESR1-TFG fusion	ESR1(6) - TFG(4)	Fusion	NEGATIVE
ESR1-TNRC6B fusion	ESR1(6) - TNRC6B(14)	Fusion	NEGATIVE
ESR1-YAP1 fusion	ESR1(6) - YAP1(4)	Fusion	NEGATIVE
PLEKHG1-ESR1 fusion	PLEKHG1(3) - ESR1(2)	Fusion	NEGATIVE
TNS3-ESR1 fusion	TNS3(16) - ESR1(5)	Fusion	NEGATIVE
ACPP-FGFR1 fusion	ACPP(10) - FGFR1(4)	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
BCR-FGFR1 fusion	BCR(4) - FGFR1(10)	Fusion	NEGATIVE
CNTRL-FGFR1 fusion	CNTRL(40) - FGFR1(10)	Fusion	NEGATIVE
CPSF6-FGFR1 fusion	CPSF6(8) - FGFR1(10)	Fusion	NEGATIVE
CUX1-FGFR1 fusion	CUX1(11) - FGFR1(10)	Fusion	NEGATIVE
EGFR-FGFR1 fusion	EGFR(24) - FGFR1(10)	Fusion	NEGATIVE
ERLIN2-FGFR1 fusion	ERLIN2(8) - FGFR1(2)	Fusion	NEGATIVE
ERVK3-1-FGFR1 fusion	ERVK3-1(3) - FGFR1(10)	Fusion	NEGATIVE
FGFR1-ADAM32 fusion	FGFR1(17) - ADAM32(14)	Fusion	NEGATIVE
FGFR1-EVI5 fusion	FGFR1(17) - EVI5(12)	Fusion	NEGATIVE
FGFR1-HOOK3 fusion	FGFR1(17) - HOOK3(5)	Fusion	NEGATIVE
FGFR1-TACC1 fusion	FGFR1(17) - TACC1(7)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(5) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(6) - FGFR1(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR1OP-FGFR1 fusion	FGFR1OP(7) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP2-FGFR1 fusion	FGFR1OP2(4) - 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(4)	Fusion	NEGATIVE
FN1-FGFR1 fusion	FN1(22) - FGFR1(3)	Fusion	NEGATIVE
KAT6A-FGFR1 fusion	KAT6A(5) - FGFR1(2)	Fusion	NEGATIVE
LRRFIP1-FGFR1 fusion	LRRFIP1(8) - FGFR1(10)	Fusion	NEGATIVE
MYO18A-FGFR1 fusion	MYO18A(33) - FGFR1(10)	Fusion	NEGATIVE
RANBP2-FGFR1 fusion	RANBP2(20) - FGFR1(10)	Fusion	NEGATIVE
SNX29-FGFR1 fusion	SNX29(1) - FGFR1(4)	Fusion	NEGATIVE
SQSTM1-FGFR1 fusion	SQSTM1(6) - FGFR1(10)	Fusion	NEGATIVE
TPR-FGFR1 fusion	TPR(22) - FGFR1(10)	Fusion	NEGATIVE
TRIM24-FGFR1 fusion	TRIM24(11) - FGFR1(10)	Fusion	NEGATIVE
WHSC1L1-FGFR1 fusion	WHSC1L1(14) - FGFR1(5)	Fusion	NEGATIVE
ZMYM2-FGFR1 fusion	ZMYM2(17) - FGFR1(10)	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
CD44-FGFR2 fusion	CD44(1) - FGFR2(3)	Fusion	NEGATIVE
COL25A1-FGFR2 fusion	COL25A1(3) - FGFR2(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CTNNB1-FGFR2 fusion	CTNNB1(1) - FGFR2(10)	Fusion	NEGATIVE
FGFR2-ACSL5 fusion	FGFR2(17) - ACSL5(15)	Fusion	NEGATIVE
FGFR2-AFF3 fusion	FGFR2(17) - AFF3(7)	Fusion	NEGATIVE
FGFR2-AHCYL1 fusion	FGFR2(17) - AHCYL1(2)	Fusion	NEGATIVE
FGFR2-ANKRD24 fusion	FGFR2(17) - ANKRD24(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(2)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(10)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(16)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(18)	Fusion	NEGATIVE
FGFR2-CASP7 fusion	FGFR2(17) - CASP7(2)	Fusion	NEGATIVE
FGFR2-CBX5 fusion	FGFR2(17) - CBX5(2)	Fusion	NEGATIVE
FGFR2-CCAR2 fusion	FGFR2(17) - CCAR2(4)	Fusion	NEGATIVE
FGFR2-CCDC186 fusion	FGFR2(17) - CCDC186(4)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(2)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(1)	Fusion	NEGATIVE
FGFR2-CELF2 fusion	FGFR2(17) - CELF2(4)	Fusion	NEGATIVE
FGFR2-CIT fusion	FGFR2(17) - CIT(23)	Fusion	NEGATIVE
FGFR2-CLIP2 fusion	FGFR2(17) - CLIP2(10)	Fusion	NEGATIVE
FGFR2-COL14A1 fusion	FGFR2(17) - COL14A1(34)	Fusion	NEGATIVE
FGFR2-CREB5 fusion	FGFR2(17) - CREB5(8)	Fusion	NEGATIVE
FGFR2-DDX21 fusion	FGFR2(17) - DDX21(11)	Fusion	NEGATIVE
FGFR2-EIF4A2 fusion	FGFR2(17) - EIF4A2(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-FAM76A fusion	FGFR2(17) - FAM76A(2)	Fusion	NEGATIVE
FGFR2-GAB2 fusion	FGFR2(17) - GAB2(2)	Fusion	NEGATIVE
FGFR2-INA fusion	FGFR2(17) - INA(2)	Fusion	NEGATIVE
FGFR2-KCTD1 fusion	FGFR2(17) - KCTD1(2)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(3)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(5)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(6)	Fusion	NEGATIVE
FGFR2-KRT8 fusion	FGFR2(17) - KRT8(6)	Fusion	NEGATIVE
FGFR2-LAMC1 fusion	FGFR2(17) - LAMC1(27)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MGEA5 fusion	FGFR2(17) - MGEA5(12)	Fusion	NEGATIVE
FGFR2-NOL4 fusion	FGFR2(17) - NOL4(7)	Fusion	NEGATIVE
FGFR2-NRAP fusion	FGFR2(17) - NRAP(24)	Fusion	NEGATIVE
FGFR2-NRBF2 fusion	FGFR2(17) - NRBF2(4)	Fusion	NEGATIVE
FGFR2-OFD1 fusion	FGFR2(17) - OFD1(3)	Fusion	NEGATIVE
FGFR2-OGDH fusion	FGFR2(17) - OGDH(3)	Fusion	NEGATIVE
FGFR2-OPTN fusion	FGFR2(17) - OPTN(5)	Fusion	NEGATIVE
FGFR2-PASD1 fusion	FGFR2(17) - PASD1(10)	Fusion	NEGATIVE
FGFR2-PCM1 fusion	FGFR2(17) - PCM1(7)	Fusion	NEGATIVE
FGFR2-PHC1 fusion	FGFR2(17) - PHC1(2)	Fusion	NEGATIVE
FGFR2-PKD2L1 fusion	FGFR2(17) - PKD2L1(7)	Fusion	NEGATIVE
FGFR2-PPHLN1 fusion	FGFR2(17) - PPHLN1(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-RABGAP1L fusion	FGFR2(17) - RABGAP1L(20)	Fusion	NEGATIVE
FGFR2-RASAL2 fusion	FGFR2(17) - RASAL2(3)	Fusion	NEGATIVE
FGFR2-ROCK1 fusion	FGFR2(17) - ROCK1(2)	Fusion	NEGATIVE
FGFR2-SBNO2 fusion	FGFR2(17) - SBNO2(19)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(7)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(5)	Fusion	NEGATIVE
FGFR2-SMN1 fusion	FGFR2(17) - SMN1(2)	Fusion	NEGATIVE
FGFR2-STK26 fusion	FGFR2(17) - STK26(3)	Fusion	NEGATIVE
FGFR2-TACC2 fusion	FGFR2(17) - TACC2(11)	Fusion	NEGATIVE
FGFR2-TACC3 fusion	FGFR2(17) - TACC3(11)	Fusion	NEGATIVE
FGFR2-TBC1D1 fusion	FGFR2(17) - TBC1D1(9)	Fusion	NEGATIVE
FGFR2-TFEC fusion	FGFR2(17) - TFEC(2)	Fusion	NEGATIVE
FGFR2-TJP2 fusion	FGFR2(17) - TJP2(5)	Fusion	NEGATIVE
FGFR2-TXLNA fusion	FGFR2(17) - TXLNA(6)	Fusion	NEGATIVE
FGFR2-UBQLN1 fusion	FGFR2(17) - UBQLN1(8)	Fusion	NEGATIVE
FGFR2-UTRN fusion	FGFR2(17) - UTRN(51)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(2)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(7)	Fusion	NEGATIVE
FGFR2-WAC fusion	FGFR2(17) - WAC(5)	Fusion	NEGATIVE
FGFR2-WARS fusion	FGFR2(17) - WARS(3)	Fusion	NEGATIVE
FGFR2-ZMYM4 fusion	FGFR2(17) - ZMYM4(10)	Fusion	NEGATIVE
KLK2-FGFR2 fusion	KLK2(1) - FGFR2(5)	Fusion	NEGATIVE
PARK2-FGFR2 fusion	PARK2(9) - FGFR2(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDHX-FGFR2 fusion	PDHX(1) - FGFR2(7)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(2)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(1)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(8) - FGFR2(7)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(7) - FGFR2(7)	Fusion	NEGATIVE
ETV6-FGFR3 fusion	ETV6(5) - FGFR3(9)	Fusion	NEGATIVE
FGFR3-ADD1 fusion	FGFR3(17) - ADD1(3)	Fusion	NEGATIVE
FGFR3-AES fusion	FGFR3(17) - AES(2)	Fusion	NEGATIVE
FGFR3-AMBRA1 fusion	FGFR3(17) - AMBRA1(14)	Fusion	NEGATIVE
FGFR3-BAIAP2L1 fusion	FGFR3(17) - BAIAP2L1(2)	Fusion	NEGATIVE
FGFR3-BRAP fusion	FGFR3(17) - BRAP(5)	Fusion	NEGATIVE
FGFR3-CAMK2A fusion	FGFR3(18) - CAMK2A(14)	Fusion	NEGATIVE
FGFR3-ELAVL3 fusion	FGFR3(17) - ELAVL3(2)	Fusion	NEGATIVE
FGFR3-FBXO28 fusion	FGFR3(17) - FBXO28(4)	Fusion	NEGATIVE
FGFR3-JAKMIP1 fusion	FGFR3(17) - JAKMIP1(4)	Fusion	NEGATIVE
FGFR3-NBR1 fusion	FGFR3(17) - NBR1(7)	Fusion	NEGATIVE
FGFR3-RBBP4 fusion	FGFR3(17) - RBBP4(12)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(6)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(8)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(11)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(18) - TACC3(11)	Fusion	NEGATIVE
BAIAP2L1-MET fusion	BAIAP2L1(9) - MET(15)	Fusion	NEGATIVE
C8orf34-MET fusion	C8orf34(2) - 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
CLIP2-MET fusion	CLIP2(11) - MET(15)	Fusion	NEGATIVE
DCTN1-MET fusion	DCTN1(26) - MET(15)	Fusion	NEGATIVE
EPS15-MET fusion	EPS15(21) - MET(15)	Fusion	NEGATIVE
ETV6-MET fusion	ETV6(4) - MET(15)	Fusion	NEGATIVE
GTF2I-MET fusion	GTF2I(6) - MET(15)	Fusion	NEGATIVE
HLA-DRB1-MET fusion	HLA-DRB1(5) - MET(15)	Fusion	NEGATIVE
KIF5B-MET fusion	KIF5B(24) - MET(15)	Fusion	NEGATIVE
LRRFIP1-MET fusion	LRRFIP1(19) - MET(15)	Fusion	NEGATIVE
MET KDD	MET(21) - MET(15)	RNAExonVariant	NEGATIVE
MET exon 14 skipping (RNA)	MET(13) - MET(15)	RNAExonVariant	NEGATIVE
OXR1-MET fusion	OXR1(9) - MET(13)	Fusion	NEGATIVE
OXR1-MET fusion	OXR1(1) - MET(13)	Fusion	NEGATIVE
PPFIBP1-MET fusion	PPFIBP1(9) - MET(15)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
SCAF11-MET fusion	SCAF11(1) - MET(2)	Fusion	NEGATIVE
SLC34A2-MET fusion	SLC34A2(4) - MET(15)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(2)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(11)	Fusion	NEGATIVE
STARD3NL-MET fusion	STARD3NL(6) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(5) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(7) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(36) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(4) - MET(15)	Fusion	NEGATIVE
TRIM4-MET fusion	TRIM4(6) - MET(15)	Fusion	NEGATIVE
ZKSCAN1-MET fusion	ZKSCAN1(3) - MET(15)	Fusion	NEGATIVE
AKAP13-NRG1 fusion	AKAP13(5) - NRG1(2)	Fusion	NEGATIVE
ATP1B1-NRG1 fusion	ATP1B1(2) - 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
CDH1-NRG1 fusion	CDH1(2) - NRG1(2)	Fusion	NEGATIVE
CLU-NRG1 fusion	CLU(2) - NRG1(6)	Fusion	NEGATIVE
COX10-AS1-NRG1 fusion	COX10-AS1(1) - NRG1(2)	Fusion	NEGATIVE
HMBOX1-NRG1 fusion	HMBOX1(1) - NRG1(6)	Fusion	NEGATIVE
KIF13B-NRG1 fusion	KIF13B(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
NOTCH2-NRG1 fusion	NOTCH2(4) - NRG1(6)	Fusion	NEGATIVE
PARP8-NRG1 fusion	PARP8(1) - NRG1(2)	Fusion	NEGATIVE
PDE7A-NRG1 fusion	PDE7A(3) - NRG1(6)	Fusion	NEGATIVE
RAB3IL1-NRG1 fusion	RAB3IL1(9) - NRG1(6)	Fusion	NEGATIVE
RALGAPA1-NRG1 fusion	RALGAPA1(20) - NRG1(6)	Fusion	NEGATIVE
ROCK1-NRG1 fusion	ROCK1(2) - NRG1(2)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(4) - NRG1(6)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(2) - NRG1(6)	Fusion	NEGATIVE
SETD4-NRG1 fusion	SETD4(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(5) - NRG1(6)	Fusion	NEGATIVE
SMAD4-NRG1 fusion	SMAD4(1) - NRG1(6)	Fusion	NEGATIVE
TBL1XR1-NRG1 fusion	TBL1XR1(1) - NRG1(2)	Fusion	NEGATIVE
TENM4-NRG1 fusion	TENM4(12) - NRG1(2)	Fusion	NEGATIVE
THAP7-NRG1 fusion	THAP7(3) - NRG1(2)	Fusion	NEGATIVE
THBS1-NRG1 fusion	THBS1(6) - NRG1(6)	Fusion	NEGATIVE
TNC-NRG1 fusion	TNC(10) - NRG1(6)	Fusion	NEGATIVE
TSHZ2-NRG1 fusion	TSHZ2(1) - NRG1(6)	Fusion	NEGATIVE
VAMP2-NRG1 fusion	VAMP2(4) - NRG1(4)	Fusion	NEGATIVE
VTCN1-NRG1 fusion	VTCN1(2) - NRG1(2)	Fusion	NEGATIVE
WHSC1L1-NRG1 fusion	WHSC1L1(1) - NRG1(2)	Fusion	NEGATIVE
WRN-NRG1 fusion	WRN(30) - NRG1(2)	Fusion	NEGATIVE
ZMYM2-NRG1 fusion	ZMYM2(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
AFAP1-NTRK1 fusion	AFAP1(4) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF2-NTRK1 fusion	ARHGEF2(21) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF25-NTRK1 fusion	ARHGEF25(11) - NTRK1(8)	Fusion	NEGATIVE
BANP-NTRK1 fusion	BANP(4) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(12) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(13) - NTRK1(11)	Fusion	NEGATIVE
CD74-NTRK1 fusion	CD74(7) - NTRK1(10)	Fusion	NEGATIVE
CEL-NTRK1 fusion	CEL(7) - NTRK1(9)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(10)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(11)	Fusion	NEGATIVE
CTRC-NTRK1 fusion	CTRC(1) - NTRK1(9)	Fusion	NEGATIVE
EPHB2-NTRK1 fusion	EPHB2(3) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
GON4L-NTRK1 fusion	GON4L(21) - NTRK1(9)	Fusion	NEGATIVE
GRIPAP1-NTRK1 fusion	GRIPAP1(22) - NTRK1(11)	Fusion	NEGATIVE
GSN-NTRK1 fusion	GSN(7) - NTRK1(10)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(10)	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
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(11)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(10)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(5) - NTRK1(11)	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(11)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(12)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(13)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(11) - NTRK1(11)	Fusion	NEGATIVE
LRRC71-NTRK1 fusion	LRRC71(1) - NTRK1(9)	Fusion	NEGATIVE
MDM4-NTRK1 fusion	MDM4(3) - NTRK1(5)	Fusion	NEGATIVE
MPRIP-NTRK1 fusion	MPRIP(21) - NTRK1(12)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
NFASC-NTRK1 fusion	NFASC(20) - NTRK1(10)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(16) - DYNC2H1(85)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(17) - DYNC2H1(85)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PEAR1-NTRK1 fusion	PEAR1(2) - NTRK1(9)	Fusion	NEGATIVE
PEAR1-NTRK1 fusion	PEAR1(15) - NTRK1(10)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(21) - NTRK1(9)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(16) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(11)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(13)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(12) - NTRK1(13)	Fusion	NEGATIVE
RABGAP1L-NTRK1 fusion	RABGAP1L(14) - NTRK1(16)	Fusion	NEGATIVE
RFWD2-NTRK1 fusion	RFWD2(2) - NTRK1(3)	Fusion	NEGATIVE
RNF213-NTRK1 fusion	RNF213(15) - NTRK1(12)	Fusion	NEGATIVE
SCYL3-NTRK1 fusion	SCYL3(11) - NTRK1(12)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(2) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(12)	Fusion	NEGATIVE
SSBP2-NTRK1 fusion	SSBP2(12) - NTRK1(12)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
TP53-NTRK1 fusion	TP53(8) - NTRK1(9)	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(9)	Fusion	NEGATIVE
TPM3-NTRK1 fusion	TPM3(7) - 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(6) - NTRK1(11)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(9)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(10)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(12)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(13)	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
TRIM63-NTRK1 fusion	TRIM63(8) - NTRK1(9)	Fusion	NEGATIVE
VANGL2-NTRK1 fusion	VANGL2(1) - NTRK1(12)	Fusion	NEGATIVE
AFAP1-NTRK2 fusion	AFAP1(14) - NTRK2(12)	Fusion	NEGATIVE
AGBL4-NTRK2 fusion	AGBL4(6) - NTRK2(16)	Fusion	NEGATIVE
BCR-NTRK2 fusion	BCR(1) - NTRK2(17)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DAB2IP-NTRK2 fusion	DAB2IP(1) - NTRK2(17)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(4) - NTRK2(16)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(15)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(16)	Fusion	NEGATIVE
GKAP1-NTRK2 fusion	GKAP1(9) - NTRK2(13)	Fusion	NEGATIVE
GNAQ-NTRK2 fusion	GNAQ(2) - NTRK2(16)	Fusion	NEGATIVE
KCTD8-NTRK2 fusion	KCTD8(1) - NTRK2(13)	Fusion	NEGATIVE
NACC2-NTRK2 fusion	NACC2(4) - NTRK2(13)	Fusion	NEGATIVE
NAV1-NTRK2 fusion	NAV1(15) - NTRK2(11)	Fusion	NEGATIVE
NOS1AP-NTRK2 fusion	NOS1AP(9) - NTRK2(11)	Fusion	NEGATIVE
PAN3-NTRK2 fusion	PAN3(1) - NTRK2(17)	Fusion	NEGATIVE
QKI-NTRK2 fusion	QKI(6) - NTRK2(16)	Fusion	NEGATIVE
SLMAP-NTRK2 fusion	SLMAP(14) - NTRK2(16)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(13)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(17)	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
STRN3-NTRK2 fusion	STRN3(7) - NTRK2(16)	Fusion	NEGATIVE
TBC1D2-NTRK2 fusion	TBC1D2(6) - NTRK2(12)	Fusion	NEGATIVE
TLE4-NTRK2 fusion	TLE4(7) - NTRK2(15)	Fusion	NEGATIVE
TP63-NTRK2 fusion	TP63(8) - NTRK2(12)	Fusion	NEGATIVE
TRAF2-NTRK2 fusion	TRAF2(9) - NTRK2(15)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(15)	Fusion	NEGATIVE
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(16)	Fusion	NEGATIVE
VCAN-NTRK2 fusion	VCAN(6) - NTRK2(10)	Fusion	NEGATIVE
VCL-NTRK2 fusion	VCL(16) - NTRK2(12)	Fusion	NEGATIVE
WNK2-NTRK2 fusion	WNK2(24) - NTRK2(16)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(14) - NTRK3(14)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(16) - NTRK3(15)	Fusion	NEGATIVE
BTBD1-NTRK3 fusion	BTBD1(4) - NTRK3(14)	Fusion	NEGATIVE
COX5A-NTRK3 fusion	COX5A(1) - NTRK3(15)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(2) - NTRK3(14)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(6) - NTRK3(14)	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(15)	Fusion	NEGATIVE
ETV6-NTRK3 fusion	ETV6(5) - NTRK3(14)	Fusion	NEGATIVE
FAT1-NTRK3 fusion	FAT1(2) - NTRK3(7)	Fusion	NEGATIVE
FBXL20-NTRK3 fusion	FBXL20(1) - NTRK3(6)	Fusion	NEGATIVE
HNRNPA2B1-NTRK3 fusion	HNRNPA2B1(7) - NTRK3(14)	Fusion	NEGATIVE
KANK1-NTRK3 fusion	KANK1(3) - NTRK3(14)	Fusion	NEGATIVE
LYN-NTRK3 fusion	LYN(8) - NTRK3(14)	Fusion	NEGATIVE
MYH9-NTRK3 fusion	MYH9(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(13)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(11)	Fusion	NEGATIVE
NTRK3-HOMER1 fusion	NTRK3(17) - HOMER1(2)	Fusion	NEGATIVE
NTRK3-HOMER2 fusion	NTRK3(17) - HOMER2(2)	Fusion	NEGATIVE
NTRK3-LOXL2 fusion	NTRK3(3) - LOXL2(5)	Fusion	NEGATIVE
RBPMS-NTRK3 fusion	RBPMS(5) - 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
TPM4-NTRK3 fusion	TPM4(7) - NTRK3(12)	Fusion	NEGATIVE
UBE2R2-NTRK3 fusion	UBE2R2(3) - NTRK3(4)	Fusion	NEGATIVE
ZNF710-NTRK3 fusion	ZNF710(1) - NTRK3(14)	Fusion	NEGATIVE
AFF1-NUTM1 fusion	AFF1(17) - NUTM1(3)	Fusion	NEGATIVE
BCORL1-NUTM1 fusion	BCORL1(1) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CIC-NUTM1 fusion	CIC(17) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(2)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(20) - NUTM1(5)	Fusion	NEGATIVE
MXD1-NUTM1 fusion	MXD1(5) - NUTM1(2)	Fusion	NEGATIVE
WHSC1L1-NUTM1 fusion	WHSC1L1(7) - NUTM1(2)	Fusion	NEGATIVE
ZNF618-NUTM1 fusion	ZNF618(12) - NUTM1(2)	Fusion	NEGATIVE
ABI3BP-RET fusion	ABI3BP(34) - RET(12)	Fusion	NEGATIVE
ACBD5-RET fusion	ACBD5(11) - RET(12)	Fusion	NEGATIVE
ADD3-RET fusion	ADD3(1) - RET(12)	Fusion	NEGATIVE
AFAP1-RET fusion	AFAP1(3) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(6) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(2) - RET(12)	Fusion	NEGATIVE
AKAP13-RET fusion	AKAP13(35) - RET(12)	Fusion	NEGATIVE
ANK3-RET fusion	ANK3(1) - RET(12)	Fusion	NEGATIVE
BCR-RET fusion	BCR(4) - RET(12)	Fusion	NEGATIVE
CCDC186-RET fusion	CCDC186(7) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CCDC6-RET fusion	CCDC6(1) - RET(10)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(12)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CUX1-RET fusion	CUX1(10) - RET(12)	Fusion	NEGATIVE
DLG5-RET fusion	DLG5(13) - RET(12)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(11)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(12)	Fusion	NEGATIVE
EPHA5-RET fusion	EPHA5(2) - 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
FGFR1OP-RET fusion	FGFR1OP(11) - RET(11)	Fusion	NEGATIVE
FGFR1OP-RET fusion	FGFR1OP(12) - RET(12)	Fusion	NEGATIVE
FKBP15-RET fusion	FKBP15(25) - RET(12)	Fusion	NEGATIVE
FRMD4A-RET fusion	FRMD4A(12) - RET(12)	Fusion	NEGATIVE
GOLGA5-RET fusion	GOLGA5(7) - RET(12)	Fusion	NEGATIVE
GPHN-RET fusion	GPHN(9) - RET(12)	Fusion	NEGATIVE
HOOK3-RET fusion	HOOK3(11) - RET(12)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(10)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1468-RET fusion	KIAA1468(10) - RET(12)	Fusion	NEGATIVE
KIF13A-RET fusion	KIF13A(18) - 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(8)	Fusion	NEGATIVE
KIF5B-RET fusion	KIF5B(23) - RET(12)	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
KIF5B-RET fusion	KIF5B(15) - RET(12)	Fusion	NEGATIVE
KTN1-RET fusion	KTN1(29) - RET(12)	Fusion	NEGATIVE
MPRIP-RET fusion	MPRIP(19) - RET(12)	Fusion	NEGATIVE
MYH10-RET fusion	MYH10(33) - RET(12)	Fusion	NEGATIVE
MYH13-RET fusion	MYH13(35) - RET(12)	Fusion	NEGATIVE
MYO5A-RET fusion	MYO5A(23) - RET(11)	Fusion	NEGATIVE
MYO5C-RET fusion	MYO5C(25) - 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
OLFM4-RET fusion	OLFM4(4) - RET(10)	Fusion	NEGATIVE
PCM1-RET fusion	PCM1(29) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDCD10-RET fusion	PDCD10(4) - RET(11)	Fusion	NEGATIVE
PICALM-RET fusion	PICALM(19) - RET(12)	Fusion	NEGATIVE
PPFIBP2-RET fusion	PPFIBP2(9) - RET(12)	Fusion	NEGATIVE
PRKAR1A-RET fusion	PRKAR1A(7) - RET(12)	Fusion	NEGATIVE
PRKG1-RET fusion	PRKG1(3) - RET(12)	Fusion	NEGATIVE
RASAL2-RET fusion	RASAL2(15) - RET(12)	Fusion	NEGATIVE
RASSF4-RET fusion	RASSF4(3) - RET(12)	Fusion	NEGATIVE
RRBP1-RET fusion	RRBP1(9) - RET(12)	Fusion	NEGATIVE
RUFY1-RET fusion	RUFY1(17) - RET(12)	Fusion	NEGATIVE
RUFY2-RET fusion	RUFY2(9) - RET(12)	Fusion	NEGATIVE
SATB1-RET fusion	SATB1(7) - RET(12)	Fusion	NEGATIVE
SNRNP70-RET fusion	SNRNP70(7) - RET(12)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(11)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(12)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(5) - RET(11)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(6) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(11)	Fusion	NEGATIVE
TFG-RET fusion	TFG(4) - RET(11)	Fusion	NEGATIVE
TNIP1-RET fusion	TNIP1(11) - RET(12)	Fusion	NEGATIVE
TNIP2-RET fusion	TNIP2(5) - RET(12)	Fusion	NEGATIVE
TRIM24-RET fusion	TRIM24(9) - RET(12)	Fusion	NEGATIVE
TRIM27-RET fusion	TRIM27(3) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
WAC-RET fusion	WAC(3) - RET(12)	Fusion	NEGATIVE
CCDC30-ROS1 fusion	CCDC30(10) - ROS1(36)	Fusion	NEGATIVE
CCDC6-ROS1 fusion	CCDC6(5) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(7) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(33)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(32)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(4) - ROS1(33)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(12) - ROS1(35)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(8) - ROS1(36)	Fusion	NEGATIVE
CLIP1-ROS1 fusion	CLIP1(20) - ROS1(36)	Fusion	NEGATIVE
CLTC-ROS1 fusion	CLTC(31) - ROS1(35)	Fusion	NEGATIVE
ERC1-ROS1 fusion	ERC1(11) - ROS1(36)	Fusion	NEGATIVE
EYS-ROS1 fusion	EYS(12) - ROS1(9)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(35)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(34)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(33)	Fusion	NEGATIVE
GOLGB1-ROS1 fusion	GOLGB1(9) - ROS1(35)	Fusion	NEGATIVE
GOPC-ROS1 fusion	GOPC(8) - ROS1(35)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
GOPC-ROS1 fusion	GOPC(4) - ROS1(36)	Fusion	NEGATIVE
HLA-A-ROS1 fusion	HLA-A(7) - ROS1(34)	Fusion	NEGATIVE
LIMA1-ROS1 fusion	LIMA1(10) - ROS1(36)	Fusion	NEGATIVE
LRIG3-ROS1 fusion	LRIG3(16) - ROS1(35)	Fusion	NEGATIVE
MSN-ROS1 fusion	MSN(9) - ROS1(34)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(32) - ROS1(35)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(23) - ROS1(35)	Fusion	NEGATIVE
MYO5C-ROS1 fusion	MYO5C(30) - ROS1(35)	Fusion	NEGATIVE
NCOR2-ROS1 fusion	NCOR2(7) - ROS1(36)	Fusion	NEGATIVE
NFKB2-ROS1 fusion	NFKB2(14) - ROS1(36)	Fusion	NEGATIVE
PPFIBP1-ROS1 fusion	PPFIBP1(9) - ROS1(35)	Fusion	NEGATIVE
PWWP2A-ROS1 fusion	PWWP2A(1) - ROS1(36)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(5) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(32)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(32)	Fusion	NEGATIVE
SHTN1-ROS1 fusion	SHTN1(11) - ROS1(36)	Fusion	NEGATIVE
SLC16A10-ROS1 fusion	SLC16A10(3) - ROS1(3)	Fusion	NEGATIVE
SLC22A16-ROS1 fusion	SLC22A16(7) - ROS1(16)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(35)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(32)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(33)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(32)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - 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
TMEM106B-ROS1 fusion	TMEM106B(3) - ROS1(35)	Fusion	NEGATIVE
TPD52L1-ROS1 fusion	TPD52L1(3) - ROS1(33)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(7) - ROS1(35)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(3) - ROS1(36)	Fusion	NEGATIVE
TSPAN3-ROS1 fusion	TSPAN3(5) - ROS1(31)	Fusion	NEGATIVE
YWHAE-ROS1 fusion	YWHAE(4) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(3) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(2) - ROS1(36)	Fusion	NEGATIVE
EIF3E-RSPO2 fusion	EIF3E(1) - RSPO2(2)	Fusion	NEGATIVE
GRHL2-RSPO2 fusion	GRHL2(8) - 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

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
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

QC Metrics

NTC QC - DNA

Metric Name	Value	Reference Range	QC Status
Average Base Coverage Depth	3	<=15	Passed
Hotspot Calls	0	<=0	Passed

NTC QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	0	<=2848	Passed
Total Fusion Calls	0	<=0	Passed

Sample QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	94944	>=6000	Passed
RNA Expression Ctrls Detected	7	>=5	Passed

Sample QC - DNA

Metric Name	Value	Reference Range	QC Status
MAPD	0.18	[0-0.5]	Passed
Mapped Reads	862563	>=100000	Passed
Mean AQ20 Read Length (bp)	88	>=64	Passed

Templating Control QC - CF-1

Metric Name	Value	Reference Range	QC Status
Average Reads Per Lane	26283	>=2383	Passed
Mean AQ20 Read Length (bp)	108	>=82	Passed

PositiveControl01 QC - DNA

Metric Name	Value	Reference Range	QC Status
MAPD	0.25	[0-0.5]	Passed
Mapped Reads	1250723	>=100000	Passed
Mean AQ20 Read Length (bp)	93	>=76	Passed
COSM1072523	POSITIVE	POSITIVE	Passed
COSM1072523_AF	0.1172	>=0.0511	Passed
COSM12376	POSITIVE	POSITIVE	Passed
COSM12376_AF	0.1278	>=0.02	Passed
COSM12428	POSITIVE	POSITIVE	Passed
COSM12428_AF	0.1465	>=0.0529	Passed
COSM12558	POSITIVE	POSITIVE	Passed
COSM12558_AF	0.1273	>=0.02	Passed
COSM133167	POSITIVE	POSITIVE	Passed
COSM133167_AF	0.1299	>=0.025	Passed
COSM144252	POSITIVE	POSITIVE	Passed
COSM144252_AF	0.1483	>=0.02	Passed
COSM256046	POSITIVE	POSITIVE	Passed
COSM256046_AF	0.1281	>=0.0489	Passed
COSM28055	POSITIVE	POSITIVE	Passed
COSM28055_AF	0.1201	>=0.0423	Passed
COSM28747	POSITIVE	POSITIVE	Passed
COSM28747_AF	0.0973	>=0.0448	Passed
COSM33765	POSITIVE	POSITIVE	Passed

Metric Name	Value	Reference Range	QC Status
COSM33765_AF	0.1132	>=0.025	Passed
COSM41590	POSITIVE	POSITIVE	Passed
COSM41590_AF	0.1215	>=0.032	Passed
COSM476	POSITIVE	POSITIVE	Passed
COSM476_AF	0.1186	>=0.0394	Passed
COSM48358	POSITIVE	POSITIVE	Passed
COSM48358_AF	0.108	>=0.0539	Passed
COSM516	POSITIVE	POSITIVE	Passed
COSM516_AF	0.1053	>=0.025	Passed
COSM5219	POSITIVE	POSITIVE	Passed
COSM5219_AF	0.1412	>=0.0555	Passed
COSM6223	POSITIVE	POSITIVE	Passed
COSM6223_AF	0.1388	>=0.0348	Passed
COSM6224	POSITIVE	POSITIVE	Passed
COSM6224_AF	0.147	>=0.0471	Passed
COSM6240	POSITIVE	POSITIVE	Passed
COSM6240_AF	0.139	>=0.025	Passed
COSM683	POSITIVE	POSITIVE	Passed
COSM683_AF	0.1128	>=0.0297	Passed
COSM775	POSITIVE	POSITIVE	Passed
COSM775_AF	0.1155	>=0.0261	Passed
COSM962	POSITIVE	POSITIVE	Passed
COSM962_AF	0.1342	>=0.0309	Passed

Metric Name	Value	Reference Range	QC Status
OMINDEL1177	POSITIVE	POSITIVE	Passed
OMINDEL1177_AF	0.1305	>=0.0334	Passed
OMINDEL698	POSITIVE	POSITIVE	Passed
OMINDEL698_AF	0.1345	>=0.0456	Passed
ERBB2 CNV	POSITIVE	POSITIVE	Passed
ERBB2 Copy Number	9.6	>=5	Passed
MET CNV	POSITIVE	POSITIVE	Passed
MET Copy Number	9.9	>=7	Passed

PositiveControl01 QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	341314	>=50475	Passed
RNA Expression Ctrls Detected	7	>=7	Passed
ALK Targeted Isoform	POSITIVE	POSITIVE	Passed
AR Targeted Isoform	POSITIVE	POSITIVE	Passed
BRAF Targeted Isoform	POSITIVE	POSITIVE	Passed
EGFR Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR1 Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR2 Expression Imbalance	POSITIVE	POSITIVE	Passed
FGFR2 Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR3 Expression Imbalance	POSITIVE	POSITIVE	Passed
FGFR3 Targeted Isoform	POSITIVE	POSITIVE	Passed
MET Targeted Isoform	POSITIVE	POSITIVE	Passed
NRG1 Targeted Isoform	POSITIVE	POSITIVE	Passed

Metric Name	Value	Reference Range	QC Status
NTRK1 Expression Imbalance	POSITIVE	POSITIVE	Passed
NTRK1 Targeted Isoform	POSITIVE	POSITIVE	Passed
NTRK2 Targeted Isoform	POSITIVE	POSITIVE	Passed
NTRK3 Targeted Isoform	POSITIVE	POSITIVE	Passed
NUTM1 Targeted Isoform	POSITIVE	POSITIVE	Passed
RET Targeted Isoform	POSITIVE	POSITIVE	Passed
ROS1 Targeted Isoform	POSITIVE	POSITIVE	Passed
RSPO2 Targeted Isoform	POSITIVE	POSITIVE	Passed
RSPO3 Targeted Isoform	POSITIVE	POSITIVE	Passed

Sequencing Run Details

Assay Name: **Oncomine™ Dx Express Test - FFPE - RS**

Panel: **Oncomine™ Dx Express Test - FFPE Panel - RS v5.2.0**

Started On: **v5.2.0 15 FEB 2022 18:38**

Completed On: **06 NOV 2025 18:23**

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

Kit Barcodes

Sample Name	Nucleic Acid Type	Kit Type	Kit Information
FFPE01	DNA	Extraction	100101850
FFPE01	RNA	Extraction	100101851
FFPE02	DNA	Extraction	100101850
FFPE02	RNA	Extraction	100101851
FFPE03	DNA	Extraction	100101850
FFPE03	RNA	Extraction	100101851
FFPE04	DNA	Extraction	100101850
FFPE04	RNA	Extraction	100101851
FFPE05	DNA	Extraction	100101850
FFPE05	RNA	Extraction	100101851
FFPE06	DNA	Extraction	100101850
FFPE06	RNA	Extraction	100101851
FFPE07	DNA	Extraction	100101850
FFPE07	RNA	Extraction	100101851
FFPE08	DNA	Extraction	100101850
FFPE08	RNA	Extraction	100101851
FFPE09	DNA	Extraction	100101850

Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** | Date of Birth: 30 MAR 1975 | Date: 13 JAN 2026

Sample Name	Nucleic Acid Type	Kit Type	Kit Information
FFPE09	RNA	Extraction	100101851
FFPE10	DNA	Extraction	100101850
FFPE10	RNA	Extraction	100101851
FFPE11	DNA	Extraction	100101850
FFPE11	RNA	Extraction	100101851
FFPE12	DNA	Extraction	100101850
FFPE12	RNA	Extraction	100101851
PositiveControl01	DNA	Control	A50834(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
PositiveControl01	RNA	Control	A50835(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
NTC	DNA	Control	A50836(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
NTC	RNA	Control	A50836(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)

180 Oyster Point Blvd
South San Francisco, CA 94080

Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 13 JAN 2026

Sample		Patient	
Sample Name:	**FFPE01**	Patient Id:	**PID01**
Cancer Type:	**Non-Small Cell Lung Cancer**	Gender:	**Male**
Cancer Stage:	**Stage III**	Date of Birth:	**30 MAR 1975**
Sample Type:	**FFPE**	Collection Date:	**22 FEB 2021**
%Cellularity:	**100**	Ordering Physician:	**Dr.Q**
%Necrosis:		Created On:	**06 NOV 2025 14:24**

Companion Diagnostics (CDx) Biomarkers

SNVs/Indels

Gene	Variant Name	AA Change	Nuc Change	Test Result	Variant ID	Associated Therapy
ERBB2	ERBB2 L755P	p.L755P	c.2263_2264delTTinsCC	POSITIVE	COSM683	HYRNUO® (sevabertinib)
ERBB2	ERBB2 exon 20 insertion	p.G776_V777insL	c.2328_2329insCTT	POSITIVE	COSM6438151	HYRNUO® (sevabertinib)

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
EGFR	EGFR exon 20 insertion	p.?	c.2284-6_2284-5insTCCAGGAAGCCT	POSITIVE	COSM26720	ins
KRAS	KRAS G12C	p.G12C	c.34_36delGGTinsTGC	POSITIVE	COSM513	mnp

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
SQSTM1-NTRK3 fusion	SQSTM1(5) - NTRK3(14)	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
AKT1	AKT1 E17K	p.E17K	c.49G>A	POSITIVE	COSM33765	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
AR-V7	AR(3) - AR(4)	RNAExonVariant	POSITIVE

ⓘ 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 therapies 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)
NSCLC	ERBB2	ERBB2/HER2 activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions)	HYRNUO® (sevabertinib)

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 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 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 >70% necrotic tissue may yield unreliable sequencing results for the detection of *EGFR* exon 20 insertion and *ERBB2* 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).

30. For *MET* Exon 14 skipping results for cancer types other than non-small cell lung cancer, confirmatory testing using a clinically validated assay is recommended.

31. For NSCLC, the Oncomine™ Dx Express Test assay definition file includes prevalent but not all rare or newly identified *EGFR* exon 20 insertions, *ERBB2/HER2* exon 20 insertions, and *ERBB2/HER2* SNVs in exons 18-21 within the tyrosine kinase domain. The Oncomine™ Dx Express Test may miss rare or newly identified:

- *EGFR* exon 20 insertions carried by a subset of patients who may derive benefit from sunvozertinib
- *ERBB2/HER2* activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions) carried by a subset of patients who may derive benefit from sevabertinib

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 C77F	p.C77F	COSM5027916	NEGATIVE	SNV
AKT1 L52R	p.L52R	COSM93893	NEGATIVE	SNV
AKT1 P51L	p.P51L	COSM4468165	NEGATIVE	SNV
AKT1 P51S	p.P51S	COSM5991558	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AKT1 P70_C77dup	p.P70_C77dup	COSM6568358	NEGATIVE	ins
AKT1 Q79K	p.Q79K	COSM159008	NEGATIVE	SNV
AKT1 R76_C77insWPNTFIIR	p.R76_C77insWPNTFIIR	COSM5045024	NEGATIVE	ins
AKT2 D324G	p.D324G	COSM474741	NEGATIVE	SNV
AKT2 D324H	p.D324H	COSM474742	NEGATIVE	SNV
AKT2 E17K	p.E17K	COSM159009	NEGATIVE	SNV
AKT3 E17K	p.E17K	COSM224779	NEGATIVE	SNV
AKT3 L51R	p.L51R	COSM309035	NEGATIVE	SNV
AKT3 Q78K	p.Q78K	COSM242892	NEGATIVE	SNV
ALK A1200V	p.A1200V	COSM317003	NEGATIVE	SNV
ALK A1280V	p.A1280V	COSM6914238	NEGATIVE	SNV
ALK C1156F	p.C1156F	BT144	NEGATIVE	SNV
ALK C1156T	p.C1156T	OM3350	NEGATIVE	mnp
ALK C1156Y	p.C1156Y	COSM99136	NEGATIVE	SNV
ALK D1203A	p.D1203A	OM3313	NEGATIVE	SNV
ALK D1203N	p.D1203N	COSM6191344	NEGATIVE	SNV
ALK E1129V	p.E1129V	BT117	NEGATIVE	SNV
ALK E1154Q	p.E1154Q	OM3324	NEGATIVE	SNV
ALK E1210K	p.E1210K	OM3161	NEGATIVE	SNV
ALK F1174C	p.F1174C	COSM28059	NEGATIVE	SNV
ALK F1174I	p.F1174I	COSM28491	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28055	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28061	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK F1174L	p.F1174L	COSM28057	NEGATIVE	SNV
ALK F1174S	p.F1174S	COSM53063	NEGATIVE	SNV
ALK F1174V	p.F1174V	COSM28054	NEGATIVE	SNV
ALK F1245C	p.F1245C	COSM28500	NEGATIVE	SNV
ALK F1245I	p.F1245I	COSM28492	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28062	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28493	NEGATIVE	SNV
ALK F1245V	p.F1245V	COSM28499	NEGATIVE	SNV
ALK G1123D	p.G1123D	OM3262	NEGATIVE	SNV
ALK G1123S	p.G1123S	COSM5045108	NEGATIVE	SNV
ALK G1128A	p.G1128A	COSM98475	NEGATIVE	SNV
ALK G1202R	p.G1202R	COSM144250	NEGATIVE	SNV
ALK G1202del	p.G1202del	OMINDEL698	NEGATIVE	del
ALK G1269A	p.G1269A	COSM1169707	NEGATIVE	SNV
ALK I1171H	p.I1171H	OM3163	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM1169448	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM28498	NEGATIVE	SNV
ALK I1171S	p.I1171S	COSM4440758	NEGATIVE	SNV
ALK I1171T	p.I1171T	COSM4381100	NEGATIVE	SNV
ALK I1268V	p.I1268V	BT149	NEGATIVE	SNV
ALK L1152P	p.L1152P	COSM1407659	NEGATIVE	SNV
ALK L1152R	p.L1152R	COSM97185	NEGATIVE	SNV
ALK L1152V	p.L1152V	OM3513	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK L1196M	p.L1196M	COSM99137	NEGATIVE	SNV
ALK L1196Q	p.L1196Q	COSM1169447	NEGATIVE	SNV
ALK L1198F	p.L1198F	COSM97028	NEGATIVE	SNV
ALK L1198P	p.L1198P	OM3162	NEGATIVE	SNV
ALK L1198V	p.L1198V	OM3314	NEGATIVE	SNV
ALK R1192P	p.R1192P	OM3259	NEGATIVE	SNV
ALK R1192Q	p.R1192Q	OM3372	NEGATIVE	SNV
ALK R1209Q	p.R1209Q	COSM327056	NEGATIVE	SNV
ALK R1231Q	p.R1231Q	COSM5967491	NEGATIVE	SNV
ALK R1231W	p.R1231W	BT148	NEGATIVE	SNV
ALK R1275L	p.R1275L	COSM28060	NEGATIVE	SNV
ALK R1275Q	p.R1275Q	COSM28056	NEGATIVE	SNV
ALK S1157F	p.S1157F	BT145	NEGATIVE	SNV
ALK S1206C	p.S1206C	BT147	NEGATIVE	SNV
ALK S1206F	p.S1206F	COSM6909200	NEGATIVE	SNV
ALK S1206Y	p.S1206Y	COSM144251	NEGATIVE	SNV
ALK T1151K	p.T1151K	OM3261	NEGATIVE	SNV
ALK T1151M	p.T1151M	COSM98478	NEGATIVE	SNV
ALK T1151R	p.T1151R	OM3260	NEGATIVE	SNV
ALK T1151dup	p.T1151dup	COSM144252	NEGATIVE	ins
ALK V1180L	p.V1180L	COSM4381101	NEGATIVE	SNV
ALK Y1278S	p.Y1278S	COSM28058	NEGATIVE	SNV
AR D880E	p.D880E	COSM5945750	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AR D891H	p.D891H	COSM5945767	NEGATIVE	SNV
AR E894K	p.E894K	COSM5945748	NEGATIVE	SNV
AR E898G	p.E898G	OM3322	NEGATIVE	SNV
AR F814V	p.F814V	COSM150793	NEGATIVE	SNV
AR F877L	p.F877L	COSM5945752	NEGATIVE	SNV
AR F877L	p.F877L	COSM5946164	NEGATIVE	SNV
AR H875Q	p.H875Q	COSM5945766	NEGATIVE	SNV
AR H875Y	p.H875Y	COSM238555	NEGATIVE	SNV
AR L702H	p.L702H	COSM238553	NEGATIVE	SNV
AR L882I	p.L882I	COSM5945751	NEGATIVE	SNV
AR M896T	p.M896T	OM3321	NEGATIVE	SNV
AR M896V	p.M896V	COSM5945749	NEGATIVE	SNV
AR S889G	p.S889G	COSM5945753	NEGATIVE	SNV
AR T878A	p.T878A	COSM236693	NEGATIVE	SNV
AR T878S	p.T878S	COSM5570418	NEGATIVE	SNV
AR V716M	p.V716M	COSM3094417	NEGATIVE	SNV
AR V731M	p.V731M	COSM1468984	NEGATIVE	SNV
AR W742C	p.W742C	COSM5572266	NEGATIVE	SNV
AR W742C	p.W742C	COSM5944171	NEGATIVE	SNV
AR W742L	p.W742L	COSM5944172	NEGATIVE	SNV
ARAF S214A	p.S214A	COSM1742787	NEGATIVE	SNV
ARAF S214C	p.S214C	COSM5044705	NEGATIVE	SNV
ARAF S214F	p.S214F	COSM612884	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ARAF S214T	p.S214T	COSM1558277	NEGATIVE	SNV
ARAF S214Y	p.S214Y	COSM5416443	NEGATIVE	SNV
BRAF A598T	p.A598T	COSM28505	NEGATIVE	SNV
BRAF A598V	p.A598V	COSM21549	NEGATIVE	SNV
BRAF D594A	p.D594A	COSM1583010	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM1121	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM253330	NEGATIVE	SNV
BRAF D594G	p.D594G	COSM467	NEGATIVE	SNV
BRAF D594H	p.D594H	COSM144576	NEGATIVE	SNV
BRAF D594N	p.D594N	COSM27639	NEGATIVE	SNV
BRAF D594V	p.D594V	COSM466	NEGATIVE	SNV
BRAF D594Y	p.D594Y	COSM6928839	NEGATIVE	SNV
BRAF D594_T599dup	p.D594_T599dup	COSM26504	NEGATIVE	ins
BRAF E586K	p.E586K	COSM463	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM468	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM53198	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM21612	NEGATIVE	SNV
BRAF F595S	p.F595S	COSM1123	NEGATIVE	SNV
BRAF G464E	p.G464E	COSM449	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1111	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1448615	NEGATIVE	SNV
BRAF G464V	p.G464V	COSM450	NEGATIVE	SNV
BRAF G466A	p.G466A	COSM452	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF G466E	p.G466E	COSM453	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM1112	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM253328	NEGATIVE	SNV
BRAF G466V	p.G466V	COSM451	NEGATIVE	SNV
BRAF G469A	p.G469A	COSM460	NEGATIVE	SNV
BRAF G469E	p.G469E	COSM461	NEGATIVE	SNV
BRAF G469L	p.G469L	COSM6056848	NEGATIVE	mnp
BRAF G469R	p.G469R	COSM455	NEGATIVE	SNV
BRAF G469R	p.G469R	COSM457	NEGATIVE	SNV
BRAF G469S	p.G469S	COSM1113	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM27912	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM458	NEGATIVE	mnp
BRAF G469V	p.G469V	COSM459	NEGATIVE	SNV
BRAF G596C	p.G596C	COSM6936824	NEGATIVE	SNV
BRAF G596D	p.G596D	COSM26506	NEGATIVE	SNV
BRAF G596R	p.G596R	COSM469	NEGATIVE	SNV
BRAF G596V	p.G596V	COSM9145962	NEGATIVE	SNV
BRAF I463S	p.I463S	COSM448	NEGATIVE	SNV
BRAF K601E	p.K601E	COSM478	NEGATIVE	SNV
BRAF K601I	p.K601I	COSM26491	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM1132	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM6265	NEGATIVE	SNV
BRAF K601R	p.K601R	COSM1568586	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF K601T	p.K601T	COSM3878760	NEGATIVE	SNV
BRAF L597P	p.L597P	COSM1448593	NEGATIVE	SNV
BRAF L597Q	p.L597Q	COSM1125	NEGATIVE	SNV
BRAF L597R	p.L597R	COSM471	NEGATIVE	SNV
BRAF L597S	p.L597S	COSM1126	NEGATIVE	mnp
BRAF L597V	p.L597V	COSM470	NEGATIVE	SNV
BRAF N581I	p.N581I	COSM26460	NEGATIVE	SNV
BRAF N581S	p.N581S	COSM462	NEGATIVE	SNV
BRAF N581T	p.N581T	COSM1699452	NEGATIVE	SNV
BRAF R462I	p.R462I	COSM447	NEGATIVE	SNV
BRAF T599I	p.T599I	COSM472	NEGATIVE	SNV
BRAF T599R	p.T599R	COSM4172020	NEGATIVE	SNV
BRAF T599_V600insEAT	p.T599_V600insEAT	COSM4166148	NEGATIVE	ins
BRAF T599_V600insETT	p.T599_V600insETT	COSM4172018	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM144982	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM30730	NEGATIVE	ins
BRAF V600A	p.V600A	COSM18443	NEGATIVE	SNV
BRAF V600D	p.V600D	COSM308550	NEGATIVE	mnp
BRAF V600D	p.V600D	COSM477	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM475	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM476	NEGATIVE	SNV
BRAF V600G	p.V600G	COSM6137	NEGATIVE	SNV
BRAF V600K	p.V600K	COSM473	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF V600L	p.V600L	COSM219798	NEGATIVE	SNV
BRAF V600L	p.V600L	COSM33808	NEGATIVE	SNV
BRAF V600M	p.V600M	COSM1130	NEGATIVE	SNV
BRAF V600Q	p.V600Q	COSM249889	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM1583011	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM474	NEGATIVE	mnp
BRAF V600_K601delinsE	p.V600_K601delinsE	COSM1133	NEGATIVE	del
BRAF Y472C	p.Y472C	COSM1133046	NEGATIVE	SNV
BRAF [G593=;D594N]	p.[G593=;D594N]	COSM211600	NEGATIVE	mnp
BRAF [T599=;V600R]	p.[T599=;V600R]	COSM1127	NEGATIVE	mnp
BRAF [T599I;V600K]	p.[T599I;V600K]	COSM1735763	NEGATIVE	mnp
BRAF [T599R;V600G]	p.[T599R;V600G]	OM3547	NEGATIVE	mnp
CDK4 K22M	p.K22M	COSM3463915	NEGATIVE	SNV
CDK4 K22Q	p.K22Q	OM3153	NEGATIVE	SNV
CDK4 K22R	p.K22R	COSM232013	NEGATIVE	SNV
CDK4 R24C	p.R24C	COSM1677139	NEGATIVE	SNV
CDK4 R24H	p.R24H	COSM1989836	NEGATIVE	SNV
CDK4 R24L	p.R24L	COSM363684	NEGATIVE	SNV
CDK4 R24S	p.R24S	COSM3463914	NEGATIVE	SNV
CHEK2 K373E	p.K373E	COSM42871	NEGATIVE	SNV
CHEK2 M381V	p.M381V	COSM1559601	NEGATIVE	SNV
CHEK2 R523H	p.R523H	COSM1559596	NEGATIVE	SNV
CHEK2 R523P	p.R523P	COSM1559595	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CHEK2 [S372=;K373E]	p.[S372=;K373E]	COSM1294063	NEGATIVE	mnp
CTNNB1 D32A	p.D32A	COSM5690	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM22939	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM5766	NEGATIVE	SNV
CTNNB1 D32G	p.D32G	COSM5681	NEGATIVE	SNV
CTNNB1 D32H	p.D32H	COSM5668	NEGATIVE	SNV
CTNNB1 D32N	p.D32N	COSM5672	NEGATIVE	SNV
CTNNB1 D32V	p.D32V	COSM5691	NEGATIVE	SNV
CTNNB1 D32Y	p.D32Y	COSM5661	NEGATIVE	SNV
CTNNB1 G34A	p.G34A	COSM5988492	NEGATIVE	SNV
CTNNB1 G34E	p.G34E	COSM5671	NEGATIVE	SNV
CTNNB1 G34I	p.G34I	COSM1666841	NEGATIVE	mnp
CTNNB1 G34R	p.G34R	COSM5684	NEGATIVE	SNV
CTNNB1 G34R	p.G34R	COSM5686	NEGATIVE	SNV
CTNNB1 G34V	p.G34V	COSM5670	NEGATIVE	SNV
CTNNB1 H36P	p.H36P	COSM5678	NEGATIVE	SNV
CTNNB1 H36R	p.H36R	COSM27378	NEGATIVE	SNV
CTNNB1 H36Y	p.H36Y	COSM5703	NEGATIVE	SNV
CTNNB1 I35K	p.I35K	COSM28727	NEGATIVE	mnp
CTNNB1 I35M	p.I35M	COSM28711	NEGATIVE	SNV
CTNNB1 I35N	p.I35N	COSM5695	NEGATIVE	SNV
CTNNB1 I35S	p.I35S	COSM5674	NEGATIVE	SNV
CTNNB1 I35T	p.I35T	COSM13168	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 I35V	p.I35V	COSM1566774	NEGATIVE	SNV
CTNNB1 K335I	p.K335I	COSM17797	NEGATIVE	SNV
CTNNB1 K335Q	p.K335Q	COSM4421324	NEGATIVE	SNV
CTNNB1 K335R	p.K335R	COSM5945941	NEGATIVE	SNV
CTNNB1 K335T	p.K335T	COSM1725761	NEGATIVE	SNV
CTNNB1 L46_S47del	p.L46_S47del	COSM6129	NEGATIVE	del
CTNNB1 L46_S47del	p.L46_S47del	COSM6132	NEGATIVE	del
CTNNB1 P44A	p.P44A	COSM17661	NEGATIVE	SNV
CTNNB1 P44L	p.P44L	COSM5761	NEGATIVE	SNV
CTNNB1 P44S	p.P44S	COSM5704	NEGATIVE	SNV
CTNNB1 S33A	p.S33A	COSM5683	NEGATIVE	SNV
CTNNB1 S33C	p.S33C	COSM5677	NEGATIVE	SNV
CTNNB1 S33F	p.S33F	COSM5669	NEGATIVE	SNV
CTNNB1 S33L	p.S33L	COSM6098	NEGATIVE	mnp
CTNNB1 S33N	p.S33N	COSM6099	NEGATIVE	mnp
CTNNB1 S33P	p.S33P	COSM5682	NEGATIVE	SNV
CTNNB1 S33T	p.S33T	COSM27311	NEGATIVE	SNV
CTNNB1 S33Y	p.S33Y	COSM5673	NEGATIVE	SNV
CTNNB1 S37A	p.S37A	COSM5675	NEGATIVE	SNV
CTNNB1 S37C	p.S37C	COSM5679	NEGATIVE	SNV
CTNNB1 S37F	p.S37F	COSM5662	NEGATIVE	SNV
CTNNB1 S37P	p.S37P	COSM5687	NEGATIVE	SNV
CTNNB1 S37T	p.S37T	COSM5729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 S37Y	p.S37Y	COSM5666	NEGATIVE	SNV
CTNNB1 S45A	p.S45A	COSM5685	NEGATIVE	SNV
CTNNB1 S45C	p.S45C	COSM5689	NEGATIVE	SNV
CTNNB1 S45F	p.S45F	COSM5667	NEGATIVE	SNV
CTNNB1 S45P	p.S45P	COSM5663	NEGATIVE	SNV
CTNNB1 S45T	p.S45T	COSM5719	NEGATIVE	SNV
CTNNB1 S45Y	p.S45Y	COSM5692	NEGATIVE	SNV
CTNNB1 S45_G48del	p.S45_G48del	COSM5992	NEGATIVE	del
CTNNB1 S45_S47del	p.S45_S47del	COSM1617582	NEGATIVE	del
CTNNB1 S45_S47delinsC	p.S45_S47delinsC	COSM28708	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6128	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6131	NEGATIVE	del
CTNNB1 T41A	p.T41A	COSM5664	NEGATIVE	SNV
CTNNB1 T41I	p.T41I	COSM5676	NEGATIVE	SNV
CTNNB1 T41N	p.T41N	COSM5730	NEGATIVE	SNV
CTNNB1 T41P	p.T41P	COSM5688	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5716	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5701	NEGATIVE	SNV
CTNNB1 [S45E;L46=]	p.[S45E;L46=]	COSM86011	NEGATIVE	mnp
EGFR A289D	p.A289D	COSM21685	NEGATIVE	SNV
EGFR A289T	p.A289T	COSM21686	NEGATIVE	SNV
EGFR A289V	p.A289V	COSM21687	NEGATIVE	SNV
EGFR A750P	p.A750P	COSM6219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR C775Y	p.C775Y	COSM22954	NEGATIVE	SNV
EGFR C797G	p.C797G	COSM6493936	NEGATIVE	SNV
EGFR C797N	p.C797N	OM3543	NEGATIVE	mnp
EGFR C797S	p.C797S	COSM6493937	NEGATIVE	SNV
EGFR C797S	p.C797S	COSM5945664	NEGATIVE	SNV
EGFR E709A	p.E709A	COSM13427	NEGATIVE	SNV
EGFR E709G	p.E709G	COSM13009	NEGATIVE	SNV
EGFR E709H	p.E709H	COSM12428	NEGATIVE	mnp
EGFR E709K	p.E709K	COSM12988	NEGATIVE	SNV
EGFR E709Q	p.E709Q	COSM116882	NEGATIVE	SNV
EGFR E709V	p.E709V	COSM12371	NEGATIVE	SNV
EGFR E709_T710delinsA	p.E709_T710delinsA	COSM85796	NEGATIVE	del
EGFR E709_T710delinsD	p.E709_T710delinsD	COSM51525	NEGATIVE	del
EGFR E709_T710delinsG	p.E709_T710delinsG	COSM48981	NEGATIVE	complex
EGFR E746G	p.E746G	COSM6849765	NEGATIVE	SNV
EGFR G465E	p.G465E	BT129	NEGATIVE	SNV
EGFR G465R	p.G465R	COSM601581	NEGATIVE	SNV
EGFR G598A	p.G598A	COSM3412196	NEGATIVE	SNV
EGFR G598V	p.G598V	COSM21690	NEGATIVE	SNV
EGFR G719A	p.G719A	COSM6239	NEGATIVE	SNV
EGFR G719C	p.G719C	COSM6253	NEGATIVE	SNV
EGFR G719D	p.G719D	COSM18425	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR G719S	p.G719S	COSM6252	NEGATIVE	SNV
EGFR G721D	p.G721D	COSM22993	NEGATIVE	SNV
EGFR G721R	p.G721R	OM3325	NEGATIVE	SNV
EGFR G721S	p.G721S	COSM22992	NEGATIVE	SNV
EGFR G724S	p.G724S	COSM13979	NEGATIVE	SNV
EGFR G796A	p.G796A	COSM27568	NEGATIVE	SNV
EGFR G796C	p.G796C	OM3318	NEGATIVE	SNV
EGFR G796R	p.G796R	COSM6493935	NEGATIVE	SNV
EGFR G796S	p.G796S	COSM20891	NEGATIVE	SNV
EGFR H835L	p.H835L	COSM6227	NEGATIVE	SNV
EGFR I491M	p.I491M	BT135	NEGATIVE	SNV
EGFR I744T	p.I744T	COSM87245	NEGATIVE	SNV
EGFR K467T	p.K467T	BT132	NEGATIVE	SNV
EGFR K757R	p.K757R	COSM133588	NEGATIVE	SNV
EGFR L718M	p.L718M	COSM1716255	NEGATIVE	SNV
EGFR L718Q	p.L718Q	COSM6503269	NEGATIVE	SNV
EGFR L718V	p.L718V	OM3316	NEGATIVE	SNV
EGFR L747P	p.L747P	COSM24267	NEGATIVE	mnp
EGFR L747S	p.L747S	COSM26704	NEGATIVE	SNV
EGFR L747V	p.L747V	OM3537	NEGATIVE	SNV
EGFR L792F	p.L792F	COSM6493933	NEGATIVE	SNV
EGFR L792H	p.L792H	COSM6493934	NEGATIVE	SNV
EGFR L792P	p.L792P	COSM13190	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR L792R	p.L792R	OM3267	NEGATIVE	SNV
EGFR L792V	p.L792V	OM3265	NEGATIVE	SNV
EGFR L792Y	p.L792Y	OM3266	NEGATIVE	mnp
EGFR L798I	p.L798I	OM3268	NEGATIVE	SNV
EGFR L833F	p.L833F	COSM219794	NEGATIVE	SNV
EGFR L833V	p.L833V	COSM13424	NEGATIVE	SNV
EGFR L833W	p.L833W	COSM133204	NEGATIVE	SNV
EGFR L858K	p.L858K	COSM24268	NEGATIVE	mnp
EGFR L858M	p.L858M	COSM12366	NEGATIVE	SNV
EGFR L858Q	p.L858Q	COSM29578	NEGATIVE	SNV
EGFR L858R	p.L858R	COSM13553	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM12429	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM133630	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM6224	NEGATIVE	SNV
EGFR L861Q	p.L861Q	COSM6213	NEGATIVE	SNV
EGFR L861R	p.L861R	COSM12374	NEGATIVE	SNV
EGFR L861V	p.L861V	COSM24848	NEGATIVE	SNV
EGFR N771T	p.N771T	OM3174	NEGATIVE	SNV
EGFR P848L	p.P848L	COSM22943	NEGATIVE	SNV
EGFR R108G	p.R108G	COSM1451536	NEGATIVE	SNV
EGFR R108K	p.R108K	COSM21683	NEGATIVE	SNV
EGFR R451C	p.R451C	COSM3266583	NEGATIVE	SNV
EGFR R836C	p.R836C	COSM28604	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR S464L	p.S464L	BT126	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236671	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236670	NEGATIVE	SNV
EGFR S720I	p.S720I	OM3317	NEGATIVE	mnp
EGFR S720P	p.S720P	COSM88729	NEGATIVE	SNV
EGFR S768G	p.S768G	COSM1451595	NEGATIVE	SNV
EGFR S768I	p.S768I	COSM6241	NEGATIVE	SNV
EGFR S768N	p.S768N	COSM12989	NEGATIVE	SNV
EGFR S768T	p.S768T	COSM291998	NEGATIVE	SNV
EGFR T790A	p.T790A	COSM1272071	NEGATIVE	SNV
EGFR T790M	p.T790M	COSM6240	NEGATIVE	SNV
EGFR T854A	p.T854A	COSM28537	NEGATIVE	SNV
EGFR T854I	p.T854I	COSM87246	NEGATIVE	SNV
EGFR T854S	p.T854S	COSM1235378	NEGATIVE	SNV
EGFR V292L	p.V292L	COSM43067	NEGATIVE	SNV
EGFR V769L	p.V769L	COSM6242	NEGATIVE	SNV
EGFR V769M	p.V769M	COSM13425	NEGATIVE	SNV
EGFR [H773L;V774M]	p.[H773L;V774M]	COSM6978402	NEGATIVE	mnp
EGFR [K745=;E746I;L747P]	p.[K745=;E746I;L747P]	OM3355	NEGATIVE	mnp
EGFR [L718=;G719C]	p.[L718=;G719C]	COSM18441	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM6984779	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM85750	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	OMINDEL1181	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR [V769G;D770I]	p.[V769G;D770I]	OM3540	NEGATIVE	mnp
EGFR exon 19 deletion	p.I744_E749delinsLKR	COSM133186	NEGATIVE	complex
EGFR exon 19 deletion	p.I744_A750delinsVK	COSM85798	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750del	COSM221565	NEGATIVE	del
EGFR exon 19 deletion	p.K745_E749del	COSM26038	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM3734668	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750delinsT	COSM1190791	NEGATIVE	del
EGFR exon 19 deletion	p.E746del	COSM18420	NEGATIVE	del
EGFR exon 19 deletion	p.E746_E749del	COSM28517	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751del	COSM24869	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.E746_T751del	COSM12728	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752del	COSM133189	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsA	COSM13549	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM13550	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM13551	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM13552	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_E749del	COSM6218	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM6225	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsFPS	COSM6506513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsA	COSM12367	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsRP	COSM12413	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsA	COSM12678	NEGATIVE	del
EGFR exon 19 deletion	p.E746_P753delinsLS	COSM13200	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133188	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133190	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsIS	COSM133191	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsQP	COSM13557	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsQ	COSM22999	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_K754del	COSM24970	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsS	COSM26513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM26680	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsL	COSM51502	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM51526	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_A750delinsP	COSM6947327	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM6966471	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsM	OMINDEL716	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751del	COSM12369	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752delinsV	COSM12384	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM12386	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM12416	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM133193	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsAP	COSM144207	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM18421	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.L747_P753del	COSM255211	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsVP	COSM28623	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsVQ	COSM51524	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM52935	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM53205	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsD	COSM6220	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_S752delinsV	COSM674057	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_T751delinsVS	COSM6980200	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750del	COSM9179903	NEGATIVE	del
EGFR exon 19 deletion	p.L747_T751delinsQ	COSM12419	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQH	COSM12421	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12422	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsDP	COSM18428	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM22944	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQ	COSM26441	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.E746_A750delinsD	COSM6974307	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM1235296	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_P753delinsS	COSM12370	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12382	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM12383	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM12387	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_S752delinsQH	COSM133196	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_A750delinsP	COSM4170220	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsS	COSM4170221	NEGATIVE	del
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM51501	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsN	COSM51503	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsS	COSM6210	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A755delinsAN	COSM85891	NEGATIVE	complex
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_E758delinsP	COSM26440	NEGATIVE	complex

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.T751_E758del	COSM133207	NEGATIVE	del
EGFR exon 19 deletion	p.A750_I759delinsGS	COSM5023005	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM1667024	NEGATIVE	complex
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.S752_I759del	COSM13556	NEGATIVE	del
EGFR exon 19 deletion	p.T751_I759delinsS	COSM1667027	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsREA	COSM22956	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM24270	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM96856	NEGATIVE	complex
EGFR exon 19 deletion	p.S752_I759del	COSM6256	NEGATIVE	del
EGFR exon 19 deletion	p.P753_I759del	COSM24269	NEGATIVE	del
EGFR exon 19 deletion	p.K754_I759del	COSM5271797	NEGATIVE	del
EGFR exon 19 insertion	p.I740_K745dup	COSM12423	NEGATIVE	ins
EGFR exon 19 insertion	p.I740_K745dup	COSM51504	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insVPVAIK	COSM26444	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insTPVAIK	COSM255152	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.?	OMINDEL1050	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_D770dup	COSM6962256	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_V765insHH	OMINDEL1051	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM12376	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_S768insTLA	COSM12425	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM20884	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVASV	OMINDEL1800	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.S768_P772dup	OMINDEL1053	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSSV	OMINDEL1054	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insTSV	OMINDEL1055	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.S768_V769insLDS	OMINDEL1802	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insRSV	OMINDEL952	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_N771dup	S768_N771dup	NEGATIVE	ins

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.A767_S768insYVM	COSM1651740	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	COSM12379	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRVDNP	OMINDEL1056	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insANV	COSM1651741	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	COSM18430	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_N771dup	COSM20885	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_P772dup	OMINDEL1057	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_V769delinsISW	OMINDEL1058	NEGATIVE	complex
EGFR exon 20 insertion	p.S768delinsRRGH	OMINDEL1059	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTV	OMINDEL1060	NEGATIVE	ins
EGFR exon 20 insertion	p.V769dup	COSM6506514	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	OMINDEL1187	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	OMINDEL1803	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTM	OMINDEL1061	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1062	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1063	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1064	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1065	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1066	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM6845099	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM85795	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGG	MATNV07	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_D770insGE	OMINDEL1069	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGQ	OMINDEL1070	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1071	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.V769_D770insERG	COSM1651742	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM1651745	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM22955	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insMASVD	COSM28638	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	MATNV03	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGTD	OMINDEL1074	NEGATIVE	ins

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.V769_D770insP	OMINDEL1076	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	OMINDEL1804	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771dup	OMINDEL1805	NEGATIVE	ins
EGFR exon 20 insertion	p.D770dup	OMINDEL1806	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insY	COSM1238030	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	COSM12381	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGY	COSM12427	NEGATIVE	ins
EGFR exon 20 insertion	p.N771dup	COSM13003	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGTH	COSM6983510	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGNPH	D770_N771insGNPH	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_1	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_2	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.D770delinsASVH	OMINDEL1080	NEGATIVE	ins

EGFR exon 20 insertion	p.D770_N771insH	OMINDEL1081	NEGATIVE	ins	

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.N771_P772insHN	OMINDEL1084	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.D770_N771insH	OMINDEL1807	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	OMINDEL1808	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1809	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsAGG	COSM12737	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insG	COSM13004	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM13558	NEGATIVE	complex
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_N771insT	COSM5023008	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insGNPH	COSM51544	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	COSM6977296	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGV	D770delinsGV	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_P772dup	MAN123	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771delinsAGH	MATNV09	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insPNP	OMINDEL1087	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1088	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsGGT	OMINDEL1090	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsEF	OMINDEL1091	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsEG	OMINDEL1092	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
EGFR exon 20 insertion	p.D770_N771insP	OMINDEL1096	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1097	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insANP	OMINDEL1098	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_H773insNPNP	OMINDEL1101	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1102	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1103	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1104	NEGATIVE	ins

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.P772_H773insTPNP	OMINDEL1106	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1107	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1108	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1109	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insYPNP	OMINDEL1110	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772delinsASVDNR	OMINDEL1801	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsAS	OMINDEL1810	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insVDSVDNP	OMINDEL953	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	COSM12378	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGT	COSM1238029	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insSVE	COSM1651743	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insAPW	COSM20886	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_N771insSVE	D770_N771insSVE	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	MATNV05	NEGATIVE	ins
EGFR exon 20	p.D770_N771delinsERG	MATNV06	NEGATIVE	complex

insertion

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.N771_V774dup	OMINDEL1112	NEGATIVE	ins
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.P772_H773insTRNP	OMINDEL1116	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_N771insF	OMINDEL1812	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRNP	OMINDEL1813	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	COSM12380	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insH	COSM1238031	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSGH	COSM1651744	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insRH	COSM166390	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsGF	COSM18431	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsTH	COSM22946	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSH	COSM24434	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTHP	COSM48923	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsVH	COSM5023007	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGY	COSM53189	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSTH	COSM6920147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insHH	COSM6931207	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsPH	MATNV04	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsHH	OMINDEL1118	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGD	OMINDEL1119	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1120	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1121	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insHPH	OMINDEL1122	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1123	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1124	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1125	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1126	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1127	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1128	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1129	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1130	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsSPH	OMINDEL1131	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1132	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1133	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1134	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1135	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.N771delinsKPH	OMINDEL1188	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsCH	OMINDEL1814	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSQRGH	OMINDEL1815	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsHHR	OMINDEL1816	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGC	OMINDEL904	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL954	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insPHPH	p.H773_V774insPHPH_2	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsSVDNR	COSM13554	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKL	COSM6438147	NEGATIVE	ins

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.N771_P772insT	OMINDEL1138	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1139	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSVDS	OMINDEL1140	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1141	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1142	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insG	OMINDEL1143	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1144	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1145	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1146	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1183	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPP	OMINDEL955	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_C775dup	OMINDEL956	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_V774dup	COSM6845098	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.N771_P772insL	OMINDEL1149	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1150	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.H773_V774insTH	OMINDEL1151	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insT	OMINDEL1152	NEGATIVE	ins
EGFR exon 20 insertion	p.H773dup	COSM12377	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774dup	COSM12389	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insNH	COSM5023006	NEGATIVE	ins
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_H773insL	OMINDEL1155	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insR	OMINDEL1156	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTP	COSM12388	NEGATIVE	complex
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.H773_V774insPHPH	H773_V774insPHPH	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insGH	OMINDEL1157	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1158	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1159	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	OMINDEL1184	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1811	NEGATIVE	complex
EGFR exon 20 insertion	p.H773delinsPNPY	COSM1735761	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsRY	MATNV08	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	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.H773delinsPNPY	OMINDEL1165	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1166	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1167	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insY	OMINDEL1168	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insTQPP	COSM3727813	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insHV	OMINDEL1169	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.H773_V774insQ	COSM131552	NEGATIVE	ins
EGFR exon 20 insertion	p.V774_C775insPR	COSM4170223	NEGATIVE	ins
ERBB2 C311R	p.C311R	COSM35763	NEGATIVE	SNV
ERBB2 D769H	p.D769H	COSM13170	NEGATIVE	SNV
ERBB2 D769N	p.D769N	COSM1302747	NEGATIVE	SNV
ERBB2 D769Y	p.D769Y	COSM1251412	NEGATIVE	SNV
ERBB2 E321G	p.E321G	COSM35764	NEGATIVE	SNV
ERBB2 G309A	p.G309A	COSM436496	NEGATIVE	SNV
ERBB2 G309E	p.G309E	COSM94224	NEGATIVE	SNV
ERBB2 G660D	p.G660D	COSM4681497	NEGATIVE	SNV
ERBB2 G776A	p.G776A	OBENV03	NEGATIVE	SNV
ERBB2 G776C	p.G776C	COSM303938	NEGATIVE	SNV
ERBB2 G776L	p.G776L	COSM144255	NEGATIVE	mnp
ERBB2 G776R	p.G776R	OM3421	NEGATIVE	SNV
ERBB2 G776S	p.G776S	COSM685	NEGATIVE	SNV
ERBB2 G776V	p.G776V	COSM18609	NEGATIVE	SNV
ERBB2 I767M	p.I767M	COSM51317	NEGATIVE	SNV
ERBB2 L755A	p.L755A	COSM6906940	NEGATIVE	mnp
ERBB2 L755F	p.L755F	OBENV01	NEGATIVE	SNV
ERBB2 L755F	p.L755F	OBENV02	NEGATIVE	SNV
ERBB2 L755M	p.L755M	COSM1205571	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 L755S	p.L755S	COSM14060	NEGATIVE	SNV
ERBB2 L755W	p.L755W	COSM436499	NEGATIVE	SNV
ERBB2 L755_T759del	p.L755_T759del	COSM7345568	NEGATIVE	del
ERBB2 L755_T759del	p.L755_T759del	COSM5029269	NEGATIVE	del
ERBB2 L869R	p.L869R	COSM249793	NEGATIVE	SNV
ERBB2 R678Q	p.R678Q	COSM436498	NEGATIVE	SNV
ERBB2 R868W	p.R868W	COSM289684	NEGATIVE	SNV
ERBB2 R896C	p.R896C	COSM14066	NEGATIVE	SNV
ERBB2 R896H	p.R896H	COSM119971	NEGATIVE	SNV
ERBB2 S310F	p.S310F	COSM48358	NEGATIVE	SNV
ERBB2 S310P	p.S310P	COSM1563863	NEGATIVE	SNV
ERBB2 S310Y	p.S310Y	COSM6906440	NEGATIVE	mnp
ERBB2 S310Y	p.S310Y	COSM94225	NEGATIVE	SNV
ERBB2 T733I	p.T733I	COSM14059	NEGATIVE	SNV
ERBB2 T798I	p.T798I	OM3210	NEGATIVE	SNV
ERBB2 V659E	p.V659E	COSM3724566	NEGATIVE	mnp
ERBB2 V659E	p.V659E	COSM6503262	NEGATIVE	mnp
ERBB2 V777A	p.V777A	COSM35496	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM14062	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM436500	NEGATIVE	SNV
ERBB2 V777M	p.V777M	COSM14064	NEGATIVE	SNV
ERBB2 V842E	p.V842E	COSM6005745	NEGATIVE	SNV
ERBB2 V842I	p.V842I	COSM14065	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.Y772_V773insMMAY	OMINDEL820	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.A775_G776insVVMA	SPRNV01	NEGATIVE	ins
ERBB2 exon 20 insertion	p.M774dup	OMINDEL823	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insYLMA	SPRNV02	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_G776insV	MAN307	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	MAN300	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.A775_G776insV	MAN308	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN309	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12552	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12553	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsLC	COSM12554	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM19875	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM20895	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsVC	COSM5235349	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsAVGC	COSM6907033	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM85995	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_V777delinsSVA	G776_V777delinsSVA_3	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_S779insCVG	OMINDEL1179	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsIC	OMINDEL826	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	SPRNV03	NEGATIVE	complex
ERBB2 exon 20 insertion	p.V777_S779dup	COSM681	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVV	COSM6977433	NEGATIVE	complex
ERBB2 exon 20 insertion	p.V777dup	COSM9869865	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.G776_G778delinsVVGD	OMINDEL1189	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_P780dup	COSM12555	NEGATIVE	ins
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	COSM303948	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_P780dup	COSM6865893	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPG	COSM7449770	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insCPG	OMINDEL1180	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_G778insC	OMINDEL827	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPS	COSM5802314	NEGATIVE	ins
ERBB3 D297H	p.D297H	COSM1935392	NEGATIVE	SNV
ERBB3 D297N	p.D297N	COSM941489	NEGATIVE	SNV
ERBB3 D297V	p.D297V	COSM941490	NEGATIVE	SNV
ERBB3 D297Y	p.D297Y	COSM160822	NEGATIVE	SNV
ERBB3 E332K	p.E332K	COSM254677	NEGATIVE	SNV
ERBB3 E332Q	p.E332Q	COSM4838033	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM5413554	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM941488	NEGATIVE	SNV
ERBB3 M60K	p.M60K	COSM254678	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB3 M60L	p.M60L	COSM1606366	NEGATIVE	SNV
ERBB3 M60R	p.M60R	COSM941484	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM122890	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM1299636	NEGATIVE	SNV
ERBB3 R103G	p.R103G	COSM6907312	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM160824	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM191840	NEGATIVE	SNV
ERBB3 V104M	p.V104M	COSM172423	NEGATIVE	SNV
ERBB4 R711C	p.R711C	COSM160827	NEGATIVE	SNV
ERBB4 R711L	p.R711L	OM3150	NEGATIVE	SNV
ESR1 D538G	p.D538G	COSM94250	NEGATIVE	SNV
ESR1 E380Q	p.E380Q	COSM3829320	NEGATIVE	SNV
ESR1 K303R	p.K303R	COSM4745827	NEGATIVE	SNV
ESR1 L536Q	p.L536Q	COSM4766050	NEGATIVE	mnp
ESR1 L536R	p.L536R	COSM4774826	NEGATIVE	SNV
ESR1 P535H	p.P535H	BT24	NEGATIVE	SNV
ESR1 P535T	p.P535T	COSM5666097	NEGATIVE	SNV
ESR1 R555C	p.R555C	BT32	NEGATIVE	SNV
ESR1 S463P	p.S463P	COSM4771561	NEGATIVE	SNV
ESR1 V392I	p.V392I	COSM1545070	NEGATIVE	SNV
ESR1 Y537C	p.Y537C	COSM1074637	NEGATIVE	SNV
ESR1 Y537N	p.Y537N	COSM1074635	NEGATIVE	SNV
ESR1 Y537S	p.Y537S	COSM1074639	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR1 K687E	p.K687E	COSM35673	NEGATIVE	SNV
FGFR1 K687M	p.K687M	COSM4771556	NEGATIVE	SNV
FGFR1 K687N	p.K687N	COSM6578115	NEGATIVE	SNV
FGFR1 V592M	p.V592M	COSM6840954	NEGATIVE	SNV
FGFR2 A314D	p.A314D	COSM49171	NEGATIVE	SNV
FGFR2 A315T	p.A315T	COSM30777	NEGATIVE	SNV
FGFR2 A648T	p.A648T	COSM29836	NEGATIVE	SNV
FGFR2 C382G	p.C382G	COSM6197913	NEGATIVE	SNV
FGFR2 C382R	p.C382R	COSM36906	NEGATIVE	SNV
FGFR2 C382Y	p.C382Y	COSM915493	NEGATIVE	SNV
FGFR2 E565A	p.E565A	OM3292	NEGATIVE	SNV
FGFR2 E565G	p.E565G	OM3563	NEGATIVE	SNV
FGFR2 G613V	p.G613V	COSM94318	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM1474415	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM48382	NEGATIVE	SNV
FGFR2 I547V	p.I547V	COSM36913	NEGATIVE	SNV
FGFR2 K310R	p.K310R	COSM36901	NEGATIVE	SNV
FGFR2 K641R	p.K641R	COSM5010388	NEGATIVE	SNV
FGFR2 K659E	p.K659E	COSM36909	NEGATIVE	SNV
FGFR2 K659M	p.K659M	COSM49175	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM49173	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM683054	NEGATIVE	SNV
FGFR2 L617M	p.L617M	OM3562	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR2 L617V	p.L617V	OM3291	NEGATIVE	SNV
FGFR2 M391R	p.M391R	COSM36907	NEGATIVE	SNV
FGFR2 N549D	p.N549D	COSM4604460	NEGATIVE	SNV
FGFR2 N549H	p.N549H	COSM250083	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36902	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36912	NEGATIVE	SNV
FGFR2 N549S	p.N549S	COSM3665553	NEGATIVE	SNV
FGFR2 N549T	p.N549T	COSM6952624	NEGATIVE	SNV
FGFR2 P253L	p.P253L	COSM537801	NEGATIVE	SNV
FGFR2 P253R	p.P253R	COSM49170	NEGATIVE	SNV
FGFR2 P253S	p.P253S	COSM1346291	NEGATIVE	SNV
FGFR2 S252W	p.S252W	COSM36903	NEGATIVE	SNV
FGFR2 V395D	p.V395D	COSM250081	NEGATIVE	mnp
FGFR2 V564F	p.V564F	OM3293	NEGATIVE	SNV
FGFR2 V564I	p.V564I	OM3247	NEGATIVE	SNV
FGFR2 V564L	p.V564L	OM3564	NEGATIVE	SNV
FGFR2 V679I	p.V679I	COSM4822956	NEGATIVE	SNV
FGFR2 Y375C	p.Y375C	COSM36904	NEGATIVE	SNV
FGFR2 Y375H	p.Y375H	COSM1560916	NEGATIVE	SNV
FGFR3 A391E	p.A391E	COSM721	NEGATIVE	SNV
FGFR3 A391T	p.A391T	OM3151	NEGATIVE	SNV
FGFR3 A391V	p.A391V	COSM1737909	NEGATIVE	SNV
FGFR3 D641G	p.D641G	COSM1428721	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 D641N	p.D641N	COSM29438	NEGATIVE	SNV
FGFR3 G370C	p.G370C	COSM716	NEGATIVE	SNV
FGFR3 G370D	p.G370D	COSM5966955	NEGATIVE	SNV
FGFR3 G380E	p.G380E	COSM51545	NEGATIVE	SNV
FGFR3 G380R	p.G380R	COSM24842	NEGATIVE	SNV
FGFR3 K650E	p.K650E	COSM719	NEGATIVE	SNV
FGFR3 K650M	p.K650M	COSM720	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM1428730	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM3993568	NEGATIVE	SNV
FGFR3 K650Q	p.K650Q	COSM726	NEGATIVE	SNV
FGFR3 K650R	p.K650R	OM3224	NEGATIVE	SNV
FGFR3 K650T	p.K650T	COSM731	NEGATIVE	SNV
FGFR3 K715M	p.K715M	COSM732992	NEGATIVE	SNV
FGFR3 R248C	p.R248C	COSM714	NEGATIVE	SNV
FGFR3 R248H	p.R248H	COSM1757310	NEGATIVE	SNV
FGFR3 R399C	p.R399C	COSM296687	NEGATIVE	SNV
FGFR3 S249C	p.S249C	COSM715	NEGATIVE	SNV
FGFR3 S249T	p.S249T	COSM29431	NEGATIVE	SNV
FGFR3 S371C	p.S371C	COSM17461	NEGATIVE	SNV
FGFR3 V553M	p.V553M	OM3223	NEGATIVE	SNV
FGFR3 V555M	p.V555M	COSM303914	NEGATIVE	SNV
FGFR3 V677I	p.V677I	COSM178247	NEGATIVE	SNV
FGFR3 Y373C	p.Y373C	COSM718	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR4 V550E	p.V550E	COSM9215998	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM1066172	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM3738141	NEGATIVE	SNV
FGFR4 V550M	p.V550M	COSM6163	NEGATIVE	SNV
FLT3 D835A	p.D835A	COSM27650	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM787	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM788	NEGATIVE	SNV
FLT3 D835F	p.D835F	COSM19844	NEGATIVE	mnp
FLT3 D835G	p.D835G	COSM1366188	NEGATIVE	SNV
FLT3 D835H	p.D835H	COSM785	NEGATIVE	SNV
FLT3 D835K	p.D835K	OM3570	NEGATIVE	mnp
FLT3 D835K	p.D835K	OM3571	NEGATIVE	mnp
FLT3 D835L	p.D835L	OM3572	NEGATIVE	mnp
FLT3 D835N	p.D835N	COSM789	NEGATIVE	SNV
FLT3 D835V	p.D835V	COSM784	NEGATIVE	SNV
FLT3 D835Y	p.D835Y	COSM783	NEGATIVE	SNV
FLT3 D835_I836del	p.D835_I836del	COSM5879385	NEGATIVE	del
FLT3 D835_I836delinsV	p.D835_I836delinsV	COSM27651	NEGATIVE	del
FLT3 D835del	p.D835del	COSM854	NEGATIVE	del
FLT3 D839E	p.D839E	COSM5945287	NEGATIVE	SNV
FLT3 D839G	p.D839G	COSM1166729	NEGATIVE	SNV
FLT3 D839H	p.D839H	COSM3399304	NEGATIVE	SNV
FLT3 D839N	p.D839N	COSM6005768	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FLT3 I836D	p.I836D	OM3569	NEGATIVE	mnp
FLT3 I836L	p.I836L	COSM19649	NEGATIVE	SNV
FLT3 I836S	p.I836S	COSM19843	NEGATIVE	SNV
FLT3 I836T	p.I836T	COSM5879386	NEGATIVE	SNV
FLT3 I836del	p.I836del	COSM19836	NEGATIVE	del
GNAS Q227E	p.Q227E	COSM4962873	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM27900	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM4168960	NEGATIVE	SNV
GNAS Q227K	p.Q227K	COSM28618	NEGATIVE	SNV
GNAS Q227L	p.Q227L	COSM27888	NEGATIVE	SNV
GNAS Q227R	p.Q227R	COSM27896	NEGATIVE	SNV
GNAS R201C	p.R201C	COSM27887	NEGATIVE	SNV
GNAS R201G	p.R201G	COSM1757313	NEGATIVE	SNV
GNAS R201H	p.R201H	COSM27895	NEGATIVE	SNV
GNAS R201L	p.R201L	COSM99221	NEGATIVE	SNV
GNAS R201S	p.R201S	COSM27899	NEGATIVE	SNV
HRAS G12A	p.G12A	COSM485	NEGATIVE	SNV
HRAS G12C	p.G12C	COSM481	NEGATIVE	SNV
HRAS G12D	p.G12D	COSM484	NEGATIVE	SNV
HRAS G12N	p.G12N	COSM4517473	NEGATIVE	mnp
HRAS G12R	p.G12R	COSM482	NEGATIVE	SNV
HRAS G12S	p.G12S	COSM480	NEGATIVE	SNV
HRAS G12V	p.G12V	COSM483	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
HRAS G13C	p.G13C	COSM488	NEGATIVE	SNV
HRAS G13D	p.G13D	COSM490	NEGATIVE	SNV
HRAS G13R	p.G13R	COSM486	NEGATIVE	SNV
HRAS G13S	p.G13S	COSM487	NEGATIVE	SNV
HRAS G13V	p.G13V	COSM489	NEGATIVE	SNV
HRAS Q61E	p.Q61E	COSM497	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM502	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM503	NEGATIVE	SNV
HRAS Q61K	p.Q61K	COSM496	NEGATIVE	SNV
HRAS Q61L	p.Q61L	COSM52978	NEGATIVE	mnp
HRAS Q61L	p.Q61L	COSM498	NEGATIVE	SNV
HRAS Q61P	p.Q61P	COSM500	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM33695	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM501	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM499	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM52979	NEGATIVE	mnp
IDH1 G70D	p.G70D	COSM96531	NEGATIVE	SNV
IDH1 R132C	p.R132C	COSM28747	NEGATIVE	SNV
IDH1 R132G	p.R132G	COSM28749	NEGATIVE	SNV
IDH1 R132H	p.R132H	COSM86993	NEGATIVE	mnp
IDH1 R132H	p.R132H	COSM28746	NEGATIVE	SNV
IDH1 R132L	p.R132L	COSM28750	NEGATIVE	SNV
IDH1 R132Q	p.R132Q	OM3221	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
IDH1 R132Q	p.R132Q	OM3222	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM1169216	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM28748	NEGATIVE	SNV
IDH1 R132V	p.R132V	COSM28751	NEGATIVE	mnp
IDH1 S280F	p.S280F	OM3315	NEGATIVE	SNV
IDH2 I319M	p.I319M	OM3319	NEGATIVE	SNV
IDH2 Q316E	p.Q316E	OM3320	NEGATIVE	SNV
IDH2 R140G	p.R140G	COSM1737874	NEGATIVE	SNV
IDH2 R140L	p.R140L	COSM41875	NEGATIVE	SNV
IDH2 R140Q	p.R140Q	COSM41590	NEGATIVE	SNV
IDH2 R140W	p.R140W	COSM41877	NEGATIVE	SNV
IDH2 R172G	p.R172G	COSM33731	NEGATIVE	SNV
IDH2 R172K	p.R172K	COSM33733	NEGATIVE	SNV
IDH2 R172M	p.R172M	COSM33732	NEGATIVE	SNV
IDH2 R172N	p.R172N	COSM256046	NEGATIVE	mnp
IDH2 R172S	p.R172S	COSM133672	NEGATIVE	SNV
IDH2 R172S	p.R172S	COSM34090	NEGATIVE	SNV
IDH2 R172T	p.R172T	COSM1636997	NEGATIVE	SNV
IDH2 R172V	p.R172V	OM3327	NEGATIVE	mnp
IDH2 R172W	p.R172W	COSM34039	NEGATIVE	SNV
KEAP1 D422N	p.D422N	COSM710198	NEGATIVE	SNV
KEAP1 G333C	p.G333C	COSM1193323	NEGATIVE	SNV
KEAP1 M409*	p.M409*	COSM6916353	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 R320L	p.R320L	COSM2812614	NEGATIVE	SNV
KEAP1 R320P	p.R320P	COSM346494	NEGATIVE	SNV
KEAP1 R320Q	p.R320Q	COSM710195	NEGATIVE	SNV
KEAP1 R320W	p.R320W	COSM1524062	NEGATIVE	SNV
KEAP1 R415G	p.R415G	COSM6196677	NEGATIVE	SNV
KEAP1 R470C	p.R470C	COSM564847	NEGATIVE	SNV
KEAP1 R470G	p.R470G	OM3724	NEGATIVE	SNV
KEAP1 R470H	p.R470H	COSM1524064	NEGATIVE	SNV
KEAP1 R470S	p.R470S	COSM564846	NEGATIVE	SNV
KEAP1 R554Q	p.R554Q	COSM1524066	NEGATIVE	SNV
KEAP1 S243C	p.S243C	COSM710191	NEGATIVE	SNV
KEAP1 V155F	p.V155F	COSM710187	NEGATIVE	SNV
KEAP1 V167F	p.V167F	COSM710188	NEGATIVE	SNV
KEAP1 Y584*	p.Y584*	COSM6201646	NEGATIVE	ins
KEAP1 c.1531+1G>T	p.?	COSM6956065	NEGATIVE	SNV
KEAP1 c.1531+2T>G	p.?	OM3723	NEGATIVE	SNV
KEAP1 c.1532-1G>T	p.?	COSM6969960	NEGATIVE	SNV
KEAP1 c.1532-2A>G	p.?	COSM381459	NEGATIVE	SNV
KEAP1 c.1708+1G>T	p.?	COSM7738757	NEGATIVE	SNV
KEAP1 c.1708+2T>A	p.?	COSM6149754	NEGATIVE	SNV
KEAP1 c.1708_1708+1delinsTT	p.?	COSM6927565	NEGATIVE	mnp
KEAP1 c.1709-1G>T	p.?	COSM7724088	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 c.1709-2A>G	p.?	COSM7088100	NEGATIVE	SNV
KEAP1 c.640-1G>A	p.?	COSM6972470	NEGATIVE	SNV
KEAP1 c.640-1G>C	p.?	OM3726	NEGATIVE	SNV
KEAP1 c.640-1G>T	p.?	OM3725	NEGATIVE	SNV
KEAP1 c.640-2A>C	p.?	OM3727	NEGATIVE	SNV
KIT A502_Y503dup	p.A502_Y503dup	COSM1326	NEGATIVE	ins
KIT A502_Y503insFA	p.A502_Y503insFA	COSM53306	NEGATIVE	ins
KIT A829P	p.A829P	COSM13172	NEGATIVE	SNV
KIT A829V	p.A829V	COSM133757	NEGATIVE	SNV
KIT D419_L421delinsF	p.D419_L421delinsF	COSM1152	NEGATIVE	complex
KIT D419_L421delinsH	p.D419_L421delinsH	COSM1578133	NEGATIVE	complex
KIT D419_R420del	p.D419_R420del	COSM1578132	NEGATIVE	del
KIT D419_R420insFF	p.D419_R420insFF	COSM36334	NEGATIVE	ins
KIT D419_R420insFL	p.D419_R420insFL	COSM36332	NEGATIVE	ins
KIT D419del	p.D419del	COSM29014	NEGATIVE	del
KIT D419del	p.D419del	COSM1600400	NEGATIVE	complex
KIT D816A	p.D816A	COSM24675	NEGATIVE	SNV
KIT D816E	p.D816E	COSM19285	NEGATIVE	SNV
KIT D816E	p.D816E	COSM5749965	NEGATIVE	SNV
KIT D816F	p.D816F	COSM1312	NEGATIVE	mnp
KIT D816G	p.D816G	COSM12711	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1311	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1600404	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT D816I	p.D816I	COSM21979	NEGATIVE	mnp
KIT D816N	p.D816N	COSM1313	NEGATIVE	SNV
KIT D816V	p.D816V	COSM1314	NEGATIVE	SNV
KIT D816Y	p.D816Y	COSM1310	NEGATIVE	SNV
KIT D820A	p.D820A	COSM133670	NEGATIVE	SNV
KIT D820E	p.D820E	COSM12709	NEGATIVE	SNV
KIT D820E	p.D820E	COSM19280	NEGATIVE	SNV
KIT D820G	p.D820G	COSM1316	NEGATIVE	SNV
KIT D820H	p.D820H	COSM22379	NEGATIVE	SNV
KIT D820N	p.D820N	COSM17947	NEGATIVE	SNV
KIT D820V	p.D820V	COSM1317	NEGATIVE	SNV
KIT D820Y	p.D820Y	COSM12710	NEGATIVE	SNV
KIT F522C	p.F522C	OM3237	NEGATIVE	SNV
KIT H697Y	p.H697Y	COSM36053	NEGATIVE	SNV
KIT I653T	p.I653T	COSM19326	NEGATIVE	SNV
KIT K550I	p.K550I	COSM1165	NEGATIVE	SNV
KIT K550N	p.K550N	COSM1167	NEGATIVE	SNV
KIT K550R	p.K550R	COSM1166	NEGATIVE	SNV
KIT K558N	p.K558N	OM3238	NEGATIVE	SNV
KIT K558N	p.K558N	COSM19216	NEGATIVE	SNV
KIT K642E	p.K642E	COSM1304	NEGATIVE	SNV
KIT K642N	p.K642N	COSM4125155	NEGATIVE	SNV
KIT K642Q	p.K642Q	COSM96871	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT L576F	p.L576F	COSM33966	NEGATIVE	SNV
KIT L576P	p.L576P	COSM1290	NEGATIVE	SNV
KIT L576R	p.L576R	COSM5945006	NEGATIVE	SNV
KIT N505I	p.N505I	OM3236	NEGATIVE	SNV
KIT N655K	p.N655K	COSM235682	NEGATIVE	SNV
KIT N655K	p.N655K	COSM4413464	NEGATIVE	SNV
KIT N655S	p.N655S	OM3302	NEGATIVE	SNV
KIT N655T	p.N655T	OM3303	NEGATIVE	SNV
KIT N680K	p.N680K	COSM5352270	NEGATIVE	SNV
KIT N822H	p.N822H	COSM1318	NEGATIVE	SNV
KIT N822I	p.N822I	COSM3604523	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1321	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1322	NEGATIVE	SNV
KIT N822S	p.N822S	COSM33975	NEGATIVE	SNV
KIT N822Y	p.N822Y	COSM19109	NEGATIVE	SNV
KIT P838L	p.P838L	OM3240	NEGATIVE	SNV
KIT R796A	p.R796A	COSM1272091	NEGATIVE	mnp
KIT R796G	p.R796G	COSM51381	NEGATIVE	SNV
KIT R796K	p.R796K	COSM1600411	NEGATIVE	SNV
KIT R796P	p.R796P	COSM1272092	NEGATIVE	mnp
KIT S501_A502dup	p.S501_A502dup	COSM5702501	NEGATIVE	ins
KIT S501_A502dup	p.S501_A502dup	COSM219786	NEGATIVE	ins
KIT T417_D419delinsI	p.T417_D419delinsI	COSM1148	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT T417_D419delinsRG	p.T417_D419delinsRG	COSM1149	NEGATIVE	complex
KIT T670A	p.T670A	OM3304	NEGATIVE	SNV
KIT T670E	p.T670E	COSM19293	NEGATIVE	mnp
KIT T670I	p.T670I	COSM12708	NEGATIVE	SNV
KIT V530I	p.V530I	COSM1155	NEGATIVE	SNV
KIT V559A	p.V559A	COSM1255	NEGATIVE	SNV
KIT V559C	p.V559C	OM3239	NEGATIVE	mnp
KIT V559D	p.V559D	COSM1252	NEGATIVE	SNV
KIT V559G	p.V559G	COSM1253	NEGATIVE	SNV
KIT V559I	p.V559I	COSM1251	NEGATIVE	SNV
KIT V559K	p.V559K	COSM96869	NEGATIVE	mnp
KIT V559P	p.V559P	COSM1272086	NEGATIVE	mnp
KIT V560A	p.V560A	COSM36302	NEGATIVE	SNV
KIT V560D	p.V560D	COSM1257	NEGATIVE	SNV
KIT V560E	p.V560E	COSM1333	NEGATIVE	mnp
KIT V560G	p.V560G	COSM1260	NEGATIVE	SNV
KIT V654A	p.V654A	COSM12706	NEGATIVE	SNV
KIT V654L	p.V654L	COSM5941904	NEGATIVE	SNV
KIT V825A	p.V825A	COSM1323	NEGATIVE	SNV
KIT V825I	p.V825I	COSM19110	NEGATIVE	SNV
KIT W557C	p.W557C	COSM19108	NEGATIVE	SNV
KIT W557C	p.W557C	COSM133700	NEGATIVE	SNV
KIT W557G	p.W557G	COSM1221	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT W557R	p.W557R	COSM1216	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1219	NEGATIVE	SNV
KIT W557S	p.W557S	COSM1227	NEGATIVE	SNV
KIT Y553C	p.Y553C	COSM4413463	NEGATIVE	SNV
KIT Y553N	p.Y553N	COSM133763	NEGATIVE	SNV
KIT Y570H	p.Y570H	COSM1430152	NEGATIVE	SNV
KIT Y578C	p.Y578C	COSM6503446	NEGATIVE	SNV
KIT Y823C	p.Y823C	COSM18682	NEGATIVE	SNV
KIT Y823D	p.Y823D	COSM18681	NEGATIVE	SNV
KIT Y823H	p.Y823H	COSM1651649	NEGATIVE	SNV
KIT Y823N	p.Y823N	COSM22378	NEGATIVE	SNV
KIT [K558S;V559S]	p.[K558S;V559S]	COSM28637	NEGATIVE	mnp
KIT [W557C;K558P]	p.[W557C;K558P]	COSM21978	NEGATIVE	mnp
KIT [W557C;K558T]	p.[W557C;K558T]	COSM28992	NEGATIVE	mnp
KIT [W557F;K558P]	p.[W557F;K558P]	COSM1224	NEGATIVE	mnp
KIT [W557S;K558S]	p.[W557S;K558S]	COSM96948	NEGATIVE	mnp
KIT exon 11 deletion	p.?	OMINDEL901	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1163	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1158	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1160	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1162	NEGATIVE	del
KIT exon 11 deletion	p.K550_V555delinsI	COSM1164	NEGATIVE	del
KIT exon 11 deletion	p.P551_E554del	COSM1172	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K550_V555delinsQ	COSM6938892	NEGATIVE	complex
KIT exon 11 deletion	p.K550_Q556delinsII	COSM1161	NEGATIVE	complex
KIT exon 11 deletion	p.P551_V555del	COSM1169	NEGATIVE	del
KIT exon 11 deletion	p.K550_W557delinsIL	COSM6936468	NEGATIVE	complex
KIT exon 11 deletion	p.K550_E554delinsI	COSM6953101	NEGATIVE	complex
KIT exon 11 deletion	p.K550_P551delinsI	COSM6971267	NEGATIVE	complex
KIT exon 11 deletion	p.K550_W557delinsIR	COSM6977972	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Q556del	COSM1170	NEGATIVE	del
KIT exon 11 deletion	p.P551del	COSM1171	NEGATIVE	del
KIT exon 11 deletion	p.P551_V555delinsL	COSM1174	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.P551_V555delinsTL	COSM30550	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Y553delinsH	COSM6934157	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1177	NEGATIVE	del
KIT exon 11 deletion	p.M552_V555del	COSM6957862	NEGATIVE	del
KIT exon 11 deletion	p.M552_Y553del	COSM96881	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsL	COSM1178	NEGATIVE	complex
KIT exon 11 deletion	p.M552_Y553del	COSM1179	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1180	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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.M552_V555del	COSM18827	NEGATIVE	del
KIT exon 11 deletion	p.M552_K558del	OMINDEL902	NEGATIVE	complex
KIT exon 11 deletion	p.M552_V555delinsI	COSM1185	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557delinsR	COSM133636	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.M552_Y553delinsN	COSM96863	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsK	COSM96868	NEGATIVE	del
KIT exon 11 deletion	p.M552_E561delinsK	COSM96880	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_K558del	COSM27909	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554delinsI	COSM6944981	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559del	COSM1191	NEGATIVE	del
KIT exon 11 deletion	p.Y553_E554del	COSM6923694	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559delinsE	COSM96946	NEGATIVE	complex
KIT exon 11 deletion	p.E554_N564del	COSM96947	NEGATIVE	del
KIT exon 11 deletion	p.E554_V559del	OMINDEL701	NEGATIVE	del
KIT exon 11 deletion	p.E554_K558del	COSM1192	NEGATIVE	del
KIT exon 11 deletion	p.V555_E562del	COSM1194	NEGATIVE	del
KIT exon 11 deletion	p.E554_E562del	COSM36310	NEGATIVE	del
KIT exon 11 deletion	p.V555_I563del	COSM1196	NEGATIVE	del
KIT exon 11 deletion	p.V555_G565del	COSM37027	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.Q556_V560del	COSM1198	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559del	COSM1199	NEGATIVE	del
KIT exon 11 deletion	p.V555_Q556del	COSM1201	NEGATIVE	del
KIT exon 11 deletion	p.V555_V560del	COSM1202	NEGATIVE	del
KIT exon 11 deletion	p.V555_N566delinsD	COSM96870	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1204	NEGATIVE	del
KIT exon 11 deletion	p.Q556_E561del	COSM1206	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558del	COSM23418	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.W557_K558del	COSM6986076	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsF	COSM1207	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558del	COSM1217	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561del	COSM1332	NEGATIVE	del
KIT exon 11 deletion	p.Q556_W557delinsR	COSM133642	NEGATIVE	del
KIT exon 11 deletion	p.Q556_I563delinsL	COSM1651755	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsTTF	COSM27904	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E562del	COSM36312	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsH	COSM1213	NEGATIVE	del
KIT exon 11 deletion	p.W557del	COSM1220	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559delinsH	COSM133641	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558delinsR	COSM28993	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsRG	COSM6954545	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.Q556_E561delinsHH	COSM1208	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V559delinsHT	COSM1214	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559del	COSM1218	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560del	COSM1223	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561delinsPS	COSM6922994	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E561delinsQ	COSM6954755	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsI	COSM1222	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsF	COSM1226	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560delinsF	COSM1230	NEGATIVE	del
KIT exon 11 deletion	p.W557_K558delinsE	COSM24748	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558delinsQ	COSM6946777	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V560delinsC	COSM1232	NEGATIVE	del
KIT exon 11 deletion	p.W557_V559delinsC	COSM1233	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.K558_V560del	COSM1234	NEGATIVE	del
KIT exon 11 deletion	p.K558_V559del	COSM1235	NEGATIVE	del
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.W557_K558delinsC	COSM96887	NEGATIVE	complex
KIT exon 11 deletion	p.K558_G565delinsR	COSM1240	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsI	COSM1241	NEGATIVE	del
KIT exon 11 deletion	p.V559_E561del	COSM1250	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K558_Y570delinsN	COSM28991	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsL	COSM6940807	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V560delinsM	COSM1242	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V559delinsN	COSM1244	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsN	COSM27069	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM6971633	NEGATIVE	del
KIT exon 11 deletion	p.V560del	COSM1247	NEGATIVE	del
KIT exon 11 deletion	p.V559_V560del	COSM1248	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM1249	NEGATIVE	del
KIT exon 11 deletion	p.V559_I571del	COSM36301	NEGATIVE	del
KIT exon 11 deletion	p.V560_I563del	COSM18897	NEGATIVE	del
KIT exon 11 deletion	p.V559_N566delinsD	COSM6977976	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1262	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1263	NEGATIVE	del
KIT exon 11 deletion	p.E562_V569delinsD	COSM1267	NEGATIVE	del
KIT exon 11 deletion	p.I563_D572del	COSM1268	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.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

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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.V569_D572del	COSM1282	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 deletion	p.I571_Y578del	OMINDEL1186	NEGATIVE	del
KIT exon 11 deletion	p.I571_L576del	COSM133754	NEGATIVE	del
KIT exon 11 deletion	p.Q575del	COSM96875	NEGATIVE	del
KIT exon 11 deletion	p.Q575_L576del	COSM6987753	NEGATIVE	del
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.Q575_P577delinsT	COSM96865	NEGATIVE	complex
KIT exon 11 deletion	p.L576del	COSM1289	NEGATIVE	del
KIT exon 11 deletion	p.P577_Y578del	COSM1291	NEGATIVE	del
KIT exon 11 deletion	p.P577del	COSM1684822	NEGATIVE	del
KIT exon 11 deletion	p.P577_D579del	COSM98379	NEGATIVE	del
KIT exon 11 deletion	p.D579del	COSM1294	NEGATIVE	del
KIT exon 11 deletion	p.H580del	COSM1295	NEGATIVE	del
KIT exon 11 insertion	p.K558delinsNP	COSM21976	NEGATIVE	ins
KIT exon 11 insertion	p.K558delinsNP	COSM1245	NEGATIVE	complex
KIT exon 11 insertion	p.I571_D579dup	COSM23417	NEGATIVE	ins
KIT exon 11 insertion	p.D572_D579dup	COSM308544	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
insertion				
KIT exon 11 insertion	p.P573_D579dup	COSM18668	NEGATIVE	ins
KIT exon 11 insertion	p.T574_D579dup	OMINDEL702	NEGATIVE	ins
KIT exon 11 insertion	p.Q575_L576dup	COSM1292	NEGATIVE	ins
KIT exon 11 insertion	p.Y578_P585dup	COSM41632	NEGATIVE	ins
KIT exon 11 insertion	p.P577_H580dup	COSM1296	NEGATIVE	ins
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
KRAS A146G	p.A146G	COSM1360828	NEGATIVE	SNV
KRAS A146P	p.A146P	COSM19905	NEGATIVE	SNV
KRAS A146S	p.A146S	COSM5967494	NEGATIVE	SNV
KRAS A146T	p.A146T	COSM19404	NEGATIVE	SNV
KRAS A146V	p.A146V	COSM19900	NEGATIVE	SNV
KRAS A59E	p.A59E	COSM547	NEGATIVE	SNV
KRAS A59G	p.A59G	COSM28518	NEGATIVE	SNV
KRAS A59S	p.A59S	COSM1235389	NEGATIVE	SNV
KRAS A59T	p.A59T	COSM546	NEGATIVE	SNV
KRAS G12A	p.G12A	COSM522	NEGATIVE	SNV
KRAS G12C	p.G12C	COSM516	NEGATIVE	SNV
KRAS G12D	p.G12D	COSM14209	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G12D	p.G12D	COSM521	NEGATIVE	SNV
KRAS G12E	p.G12E	COSM30566	NEGATIVE	mnp
KRAS G12E	p.G12E	COSM519	NEGATIVE	mnp
KRAS G12F	p.G12F	COSM512	NEGATIVE	mnp
KRAS G12I	p.G12I	COSM34144	NEGATIVE	mnp
KRAS G12L	p.G12L	COSM514	NEGATIVE	mnp
KRAS G12N	p.G12N	COSM13643	NEGATIVE	mnp
KRAS G12R	p.G12R	COSM518	NEGATIVE	SNV
KRAS G12S	p.G12S	COSM517	NEGATIVE	SNV
KRAS G12V	p.G12V	COSM515	NEGATIVE	mnp
KRAS G12V	p.G12V	COSM520	NEGATIVE	SNV
KRAS G12W	p.G12W	COSM36281	NEGATIVE	mnp
KRAS G12Y	p.G12Y	COSM25081	NEGATIVE	mnp
KRAS G13A	p.G13A	COSM533	NEGATIVE	SNV
KRAS G13C	p.G13C	COSM527	NEGATIVE	SNV
KRAS G13D	p.G13D	COSM531	NEGATIVE	mnp
KRAS G13D	p.G13D	COSM532	NEGATIVE	SNV
KRAS G13E	p.G13E	COSM30567	NEGATIVE	mnp
KRAS G13E	p.G13E	COSM87280	NEGATIVE	mnp
KRAS G13I	p.G13I	COSM525	NEGATIVE	mnp
KRAS G13N	p.G13N	COSM53283	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM526	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM529	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G13S	p.G13S	COSM528	NEGATIVE	SNV
KRAS G13V	p.G13V	COSM12721	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM530	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM534	NEGATIVE	SNV
KRAS G15D	p.G15D	COSM539	NEGATIVE	SNV
KRAS G15S	p.G15S	COSM538	NEGATIVE	SNV
KRAS K117E	p.K117E	COSM6854421	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM19940	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM28519	NEGATIVE	SNV
KRAS K117R	p.K117R	COSM4696721	NEGATIVE	SNV
KRAS Q61E	p.Q61E	COSM550	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM554	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM555	NEGATIVE	SNV
KRAS Q61K	p.Q61K	COSM549	NEGATIVE	SNV
KRAS Q61L	p.Q61L	COSM553	NEGATIVE	SNV
KRAS Q61P	p.Q61P	COSM551	NEGATIVE	SNV
KRAS Q61R	p.Q61R	COSM1168052	NEGATIVE	mnp
KRAS Q61R	p.Q61R	COSM552	NEGATIVE	SNV
KRAS [G12=;G13C]	p.[G12=;G13C]	COSM87281	NEGATIVE	mnp
KRAS [G13D;V14I]	p.[G13D;V14I]	COSM30568	NEGATIVE	mnp
KRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM87298	NEGATIVE	mnp
MAP2K1 C121S	p.C121S	COSM555601	NEGATIVE	SNV
MAP2K1 C121S	p.C121S	COSM1315829	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 E203K	p.E203K	COSM232755	NEGATIVE	SNV
MAP2K1 E203V	p.E203V	COSM3386991	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3249	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	COSM1235480	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3250	NEGATIVE	SNV
MAP2K1 F53C	p.F53C	COSM1562837	NEGATIVE	SNV
MAP2K1 F53I	p.F53I	COSM3503329	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM555604	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM1725008	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM2257208	NEGATIVE	SNV
MAP2K1 F53S	p.F53S	ClinVar13350	NEGATIVE	SNV
MAP2K1 F53V	p.F53V	COSM5077832	NEGATIVE	SNV
MAP2K1 F53Y	p.F53Y	COSM5588244	NEGATIVE	SNV
MAP2K1 G128D	p.G128D	COSM1374186	NEGATIVE	SNV
MAP2K1 G128V	p.G128V	COSM1517232	NEGATIVE	SNV
MAP2K1 K57E	p.K57E	COSM5369532	NEGATIVE	SNV
MAP2K1 K57M	p.K57M	MAN124	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM1235478	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM5520914	NEGATIVE	SNV
MAP2K1 K57Q	p.K57Q	OM3155	NEGATIVE	SNV
MAP2K1 K57T	p.K57T	COSM4756761	NEGATIVE	SNV
MAP2K1 L115P	p.L115P	OM3248	NEGATIVE	SNV
MAP2K1 L215P	p.L215P	OM3252	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 P124L	p.P124L	COSM1315861	NEGATIVE	SNV
MAP2K1 P124Q	p.P124Q	COSM1167912	NEGATIVE	SNV
MAP2K1 P124S	p.P124S	COSM235614	NEGATIVE	SNV
MAP2K1 Q56P	p.Q56P	COSM1235481	NEGATIVE	SNV
MAP2K1 V211D	p.V211D	OM3251	NEGATIVE	SNV
MAP2K1 V60E	p.V60E	COSM2257209	NEGATIVE	SNV
MAP2K1 V60M	p.V60M	OM3576	NEGATIVE	SNV
MAP2K2 C125S	p.C125S	COSM5855814	NEGATIVE	SNV
MAP2K2 N126D	p.N126D	COSM1731742	NEGATIVE	SNV
MAP2K2 V35M	p.V35M	COSM5855836	NEGATIVE	SNV
MET H1112L	p.H1112L	COSM698	NEGATIVE	SNV
MET H1112R	p.H1112R	COSM703	NEGATIVE	SNV
MET H1112Y	p.H1112Y	COSM696	NEGATIVE	SNV
MET Y1021F	p.Y1021F	COSM339515	NEGATIVE	SNV
MET Y1021S	p.Y1021S	COSM7450124	NEGATIVE	SNV
MET c.2942-20_2942-19delinsAGAG	p.?	OMINDEL804	NEGATIVE	complex
MET c.2942-21_2942-20delinsGC	p.?	COSM8631602	NEGATIVE	mnp
MET c.2942-29_2942-28delinsGG	p.?	OM3305	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM29635	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL802	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.?	OMINDEL674	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM49015	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6980941	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL709	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6056832	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL675	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL676	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL710	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.?	COSM6057022	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL640	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL697	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6947399	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	OMINDEL642	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL677	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL679	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL712	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6948341	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL680	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL681	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.?	COSM6976260	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL684	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL706	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL685	NEGATIVE	ins
MET exon 14 skipping (DNA)	p.?	OMINDEL808	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OM3177	NEGATIVE	SNV
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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6057025	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL686	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6937267	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6977526	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL654	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL655	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL811	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL656	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL658	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL688	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL687	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL689	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL662	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.?	COSM6965225	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL691	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL812	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	OMINDEL813	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.?	OMINDEL814	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.?	OMINDEL694	NEGATIVE	del
MET exon 14 skipping (DNA)	p.D1028IfsTer19	COSM24687	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.D1028Y	COSM6917314	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.D1028N	COSM6924587	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL695	NEGATIVE	complex
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.?	COSM6928344	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6940825	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6983140	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL815	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6108460	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6947995	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3185	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3217	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM744044	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3186	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3188	NEGATIVE	SNV
NRAS A146T	p.A146T	COSM27174	NEGATIVE	SNV
NRAS A146V	p.A146V	COSM4170228	NEGATIVE	SNV
NRAS A59D	p.A59D	COSM253327	NEGATIVE	SNV
NRAS A59T	p.A59T	COSM578	NEGATIVE	SNV
NRAS G12A	p.G12A	COSM565	NEGATIVE	SNV
NRAS G12C	p.G12C	COSM562	NEGATIVE	SNV
NRAS G12D	p.G12D	COSM564	NEGATIVE	SNV
NRAS G12E	p.G12E	COSM144577	NEGATIVE	mnp
NRAS G12N	p.G12N	COSM12723	NEGATIVE	mnp
NRAS G12P	p.G12P	COSM559	NEGATIVE	mnp
NRAS G12R	p.G12R	COSM561	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS G12S	p.G12S	COSM563	NEGATIVE	SNV
NRAS G12V	p.G12V	COSM566	NEGATIVE	SNV
NRAS G12Y	p.G12Y	COSM560	NEGATIVE	mnp
NRAS G13A	p.G13A	COSM575	NEGATIVE	SNV
NRAS G13C	p.G13C	COSM570	NEGATIVE	SNV
NRAS G13D	p.G13D	COSM573	NEGATIVE	SNV
NRAS G13N	p.G13N	COSM24668	NEGATIVE	mnp
NRAS G13R	p.G13R	COSM569	NEGATIVE	SNV
NRAS G13S	p.G13S	COSM571	NEGATIVE	SNV
NRAS G13V	p.G13V	COSM572	NEGATIVE	mnp
NRAS G13V	p.G13V	COSM574	NEGATIVE	SNV
NRAS G13Y	p.G13Y	COSM568	NEGATIVE	mnp
NRAS K117N	p.K117N	MAN13	NEGATIVE	SNV
NRAS Q61E	p.Q61E	COSM581	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM585	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM586	NEGATIVE	SNV
NRAS Q61K	p.Q61K	COSM53223	NEGATIVE	mnp
NRAS Q61K	p.Q61K	COSM580	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM30646	NEGATIVE	mnp
NRAS Q61L	p.Q61L	COSM583	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM12725	NEGATIVE	mnp
NRAS Q61P	p.Q61P	COSM582	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM33693	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS Q61R	p.Q61R	COSM584	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM579	NEGATIVE	mnp
NRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM12730	NEGATIVE	mnp
NRAS [Q61H;E62K]	p.[Q61H;E62K]	COSM26494	NEGATIVE	mnp
NTRK1 F589C	p.F589C	OM3255	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT106	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT107	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT108	NEGATIVE	SNV
NTRK1 F589Y	p.F589Y	BT105	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT109	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT110	NEGATIVE	SNV
NTRK1 G667A	p.G667A	OM3301	NEGATIVE	SNV
NTRK1 G667C	p.G667C	BT111	NEGATIVE	SNV
NTRK1 G667S	p.G667S	BT112	NEGATIVE	SNV
NTRK1 V573M	p.V573M	BT104	NEGATIVE	SNV
NTRK2 F633C	p.F633C	OM3278	NEGATIVE	SNV
NTRK2 F633I	p.F633I	OM3276	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3277	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3308	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3309	NEGATIVE	SNV
NTRK2 F633Y	p.F633Y	OM3307	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3279	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3280	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NTRK2 G709A	p.G709A	OM3310	NEGATIVE	SNV
NTRK2 Q612E	p.Q612E	OM3272	NEGATIVE	SNV
NTRK2 Q612P	p.Q612P	OM3273	NEGATIVE	SNV
NTRK2 R646K	p.R646K	OM3281	NEGATIVE	SNV
NTRK2 V617D	p.V617D	OM3306	NEGATIVE	SNV
NTRK2 V617G	p.V617G	OM3275	NEGATIVE	SNV
NTRK2 V617M	p.V617M	OM3274	NEGATIVE	mnp
NTRK3 F617L	p.F617L	OM3297	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3298	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3299	NEGATIVE	SNV
NTRK3 F617Y	p.F617Y	OM3311	NEGATIVE	SNV
NTRK3 G623R	p.G623R	COSM6951362	NEGATIVE	SNV
NTRK3 G623R	p.G623R	OM3253	NEGATIVE	SNV
NTRK3 G696A	p.G696A	OM3296	NEGATIVE	SNV
NTRK3 G696C	p.G696C	OM3294	NEGATIVE	mnp
NTRK3 G696C	p.G696C	OM3295	NEGATIVE	mnp
NTRK3 G696E	p.G696E	COSM5850004	NEGATIVE	SNV
NTRK3 V601F	p.V601F	OM3312	NEGATIVE	SNV
NTRK3 V601M	p.V601M	OM3300	NEGATIVE	mnp
PDGFRA A530_I543dup	p.A530_I543dup	COSM7002381	NEGATIVE	ins
PDGFRA D568N	p.D568N	OM3225	NEGATIVE	SNV
PDGFRA D842F	p.D842F	COSM29007	NEGATIVE	mnp
PDGFRA D842H	p.D842H	COSM734174	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA D842I	p.D842I	COSM12398	NEGATIVE	mnp
PDGFRA D842V	p.D842V	COSM736	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12396	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12397	NEGATIVE	mnp
PDGFRA D846Y	p.D846Y	COSM12399	NEGATIVE	SNV
PDGFRA G853D	p.G853D	OM3235	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3228	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3229	NEGATIVE	SNV
PDGFRA H845Y	p.H845Y	COSM96893	NEGATIVE	SNV
PDGFRA N659H	p.N659H	COSM51516	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22414	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22415	NEGATIVE	SNV
PDGFRA N659R	p.N659R	OM3230	NEGATIVE	mnp
PDGFRA N659S	p.N659S	COSM6960333	NEGATIVE	SNV
PDGFRA N659Y	p.N659Y	COSM22416	NEGATIVE	SNV
PDGFRA N848K	p.N848K	COSM28052	NEGATIVE	SNV
PDGFRA P577S	p.P577S	COSM6438084	NEGATIVE	SNV
PDGFRA Q579R	p.Q579R	OM3226	NEGATIVE	SNV
PDGFRA R841K	p.R841K	OM3233	NEGATIVE	SNV
PDGFRA S566_E571delinsR	p.S566_E571delinsR	COSM12418	NEGATIVE	del
PDGFRA V536E	p.V536E	COSM43059	NEGATIVE	SNV
PDGFRA V561A	p.V561A	COSM1430077	NEGATIVE	SNV
PDGFRA V561D	p.V561D	COSM739	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA W559_R560del	p.W559_R560del	OMINDEL700	NEGATIVE	del
PDGFRA Y555C	p.Y555C	COSM133645	NEGATIVE	SNV
PDGFRA Y849C	p.Y849C	COSM94952	NEGATIVE	SNV
PDGFRA Y849S	p.Y849S	OM3234	NEGATIVE	SNV
PDGFRA exon 18 deletion	p.D842_M844del	COSM96895	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_M844del	COSM12401	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842del	COSM12406	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_D846delinsN	COSM12411	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_I843delinsV	COSM405285	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_H845delinsY	COSM96896	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsRI	OMINDEL900	NEGATIVE	complex
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.I843del	COSM4969655	NEGATIVE	del
PDGFRA exon 18 deletion	p.I843_D846del	COSM12400	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_D846delinsE	COSM12408	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsEA	COSM12415	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA exon 18 deletion	p.I843_S847delinsT	COSM12407	NEGATIVE	del
PIK3CA A1046E	p.A1046E	COSM27156	NEGATIVE	SNV
PIK3CA A1046T	p.A1046T	COSM27273	NEGATIVE	SNV
PIK3CA A1046V	p.A1046V	COSM6146	NEGATIVE	SNV
PIK3CA C420R	p.C420R	COSM757	NEGATIVE	SNV
PIK3CA D1017H	p.D1017H	COSM27377	NEGATIVE	SNV
PIK3CA D1045G	p.D1045G	COSM1421003	NEGATIVE	SNV
PIK3CA D1045N	p.D1045N	COSM25086	NEGATIVE	SNV
PIK3CA D1045V	p.D1045V	COSM729813	NEGATIVE	SNV
PIK3CA D538G	p.D538G	COSM29123	NEGATIVE	SNV
PIK3CA D538N	p.D538N	COSM21467	NEGATIVE	SNV
PIK3CA D538Y	p.D538Y	COSM1716811	NEGATIVE	SNV
PIK3CA E109_I112delinsD	p.E109_I112delinsD	COSM445995	NEGATIVE	del
PIK3CA E110del	p.E110del	COSM24710	NEGATIVE	del
PIK3CA E39G	p.E39G	COSM1041451	NEGATIVE	SNV
PIK3CA E39K	p.E39K	COSM30625	NEGATIVE	SNV
PIK3CA E39V	p.E39V	COSM1420731	NEGATIVE	SNV
PIK3CA E418K	p.E418K	COSM12582	NEGATIVE	SNV
PIK3CA E542A	p.E542A	COSM1041494	NEGATIVE	SNV
PIK3CA E542G	p.E542G	COSM761	NEGATIVE	SNV
PIK3CA E542K	p.E542K	COSM760	NEGATIVE	SNV
PIK3CA E542Q	p.E542Q	COSM17442	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA E542R	p.E542R	COSM1659152	NEGATIVE	mnp
PIK3CA E542V	p.E542V	COSM762	NEGATIVE	SNV
PIK3CA E545A	p.E545A	COSM12458	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM27374	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM765	NEGATIVE	SNV
PIK3CA E545G	p.E545G	COSM764	NEGATIVE	SNV
PIK3CA E545K	p.E545K	COSM763	NEGATIVE	SNV
PIK3CA E545Q	p.E545Q	COSM27133	NEGATIVE	SNV
PIK3CA E545V	p.E545V	COSM27155	NEGATIVE	SNV
PIK3CA E547D	p.E547D	COSM729819	NEGATIVE	SNV
PIK3CA E547G	p.E547G	COSM1420864	NEGATIVE	SNV
PIK3CA E547K	p.E547K	COSM29315	NEGATIVE	SNV
PIK3CA E81A	p.E81A	COSM5347052	NEGATIVE	SNV
PIK3CA E81K	p.E81K	COSM27502	NEGATIVE	SNV
PIK3CA G1049A	p.G1049A	COSM27158	NEGATIVE	SNV
PIK3CA G1049C	p.G1049C	COSM1168056	NEGATIVE	SNV
PIK3CA G1049D	p.G1049D	COSM308548	NEGATIVE	SNV
PIK3CA G1049R	p.G1049R	COSM12597	NEGATIVE	SNV
PIK3CA G1049S	p.G1049S	COSM777	NEGATIVE	SNV
PIK3CA G106A	p.G106A	COSM166339	NEGATIVE	SNV
PIK3CA G106R	p.G106R	COSM86046	NEGATIVE	SNV
PIK3CA G106V	p.G106V	COSM748	NEGATIVE	SNV
PIK3CA G118D	p.G118D	COSM751	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA H1047L	p.H1047L	COSM776	NEGATIVE	SNV
PIK3CA H1047N	p.H1047N	COSM5029128	NEGATIVE	SNV
PIK3CA H1047P	p.H1047P	COSM249874	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM1041525	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM24714	NEGATIVE	SNV
PIK3CA H1047R	p.H1047R	COSM775	NEGATIVE	SNV
PIK3CA H1047Y	p.H1047Y	COSM774	NEGATIVE	SNV
PIK3CA H1048D	p.H1048D	COSM249881	NEGATIVE	SNV
PIK3CA H1048L	p.H1048L	COSM3734761	NEGATIVE	SNV
PIK3CA H1048N	p.H1048N	COSM1421010	NEGATIVE	SNV
PIK3CA H1048Q	p.H1048Q	COSM27157	NEGATIVE	SNV
PIK3CA H1048R	p.H1048R	COSM36289	NEGATIVE	SNV
PIK3CA H1048Y	p.H1048Y	COSM249875	NEGATIVE	SNV
PIK3CA K111E	p.K111E	COSM13570	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM12580	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM27505	NEGATIVE	SNV
PIK3CA K111R	p.K111R	COSM14052	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM29313	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM773	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM94984	NEGATIVE	SNV
PIK3CA M1043L	p.M1043L	COSM5731063	NEGATIVE	SNV
PIK3CA M1043T	p.M1043T	COSM12463	NEGATIVE	SNV
PIK3CA M1043V	p.M1043V	COSM12591	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA N1044D	p.N1044D	COSM27134	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM12592	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM27504	NEGATIVE	SNV
PIK3CA N1044S	p.N1044S	COSM22541	NEGATIVE	SNV
PIK3CA N1044Y	p.N1044Y	COSM36288	NEGATIVE	SNV
PIK3CA N345D	p.N345D	COSM1420788	NEGATIVE	SNV
PIK3CA N345H	p.N345H	COSM3846786	NEGATIVE	SNV
PIK3CA N345I	p.N345I	COSM94978	NEGATIVE	SNV
PIK3CA N345K	p.N345K	COSM754	NEGATIVE	SNV
PIK3CA N345S	p.N345S	COSM13476	NEGATIVE	SNV
PIK3CA N345T	p.N345T	COSM446002	NEGATIVE	SNV
PIK3CA P104L	p.P104L	COSM163485	NEGATIVE	SNV
PIK3CA P104R	p.P104R	COSM747	NEGATIVE	SNV
PIK3CA P104T	p.P104T	COSM1666660	NEGATIVE	SNV
PIK3CA P539R	p.P539R	COSM759	NEGATIVE	SNV
PIK3CA P539S	p.P539S	COSM249880	NEGATIVE	SNV
PIK3CA P539T	p.P539T	COSM41779	NEGATIVE	SNV
PIK3CA Q546E	p.Q546E	COSM6147	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM24712	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM5881064	NEGATIVE	SNV
PIK3CA Q546K	p.Q546K	COSM766	NEGATIVE	SNV
PIK3CA Q546L	p.Q546L	COSM25041	NEGATIVE	SNV
PIK3CA Q546P	p.Q546P	COSM767	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Q546R	p.Q546R	COSM12459	NEGATIVE	SNV
PIK3CA R108C	p.R108C	COSM6438095	NEGATIVE	SNV
PIK3CA R108H	p.R108H	COSM27497	NEGATIVE	SNV
PIK3CA R108L	p.R108L	COSM729828	NEGATIVE	SNV
PIK3CA R108P	p.R108P	COSM749	NEGATIVE	SNV
PIK3CA R38C	p.R38C	COSM744	NEGATIVE	SNV
PIK3CA R38G	p.R38G	COSM40945	NEGATIVE	SNV
PIK3CA R38H	p.R38H	COSM745	NEGATIVE	SNV
PIK3CA R38L	p.R38L	COSM729833	NEGATIVE	SNV
PIK3CA R38S	p.R38S	COSM87310	NEGATIVE	SNV
PIK3CA T1025A	p.T1025A	COSM771	NEGATIVE	SNV
PIK3CA T1025I	p.T1025I	COSM36285	NEGATIVE	SNV
PIK3CA T1025N	p.T1025N	COSM772	NEGATIVE	SNV
PIK3CA T1025P	p.T1025P	COSM6023925	NEGATIVE	SNV
PIK3CA T1025S	p.T1025S	COSM12590	NEGATIVE	SNV
PIK3CA V344A	p.V344A	COSM86951	NEGATIVE	SNV
PIK3CA V344E	p.V344E	COSM6746438	NEGATIVE	SNV
PIK3CA V344G	p.V344G	COSM22540	NEGATIVE	SNV
PIK3CA V344M	p.V344M	COSM253279	NEGATIVE	SNV
PIK3CA Y1021C	p.Y1021C	COSM12461	NEGATIVE	SNV
PIK3CA Y1021F	p.Y1021F	COSM328025	NEGATIVE	SNV
PIK3CA Y1021H	p.Y1021H	COSM17444	NEGATIVE	SNV
PIK3CA Y1021N	p.Y1021N	COSM6143	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN A126D	p.A126D	COSM28884	NEGATIVE	SNV
PTEN A126G	p.A126G	COSM4391544	NEGATIVE	SNV
PTEN A126P	p.A126P	COSM5211	NEGATIVE	SNV
PTEN A126S	p.A126S	COSM28889	NEGATIVE	SNV
PTEN A126T	p.A126T	COSM5051	NEGATIVE	SNV
PTEN A126V	p.A126V	COSM5041	NEGATIVE	SNV
PTEN C136F	p.C136F	COSM5228	NEGATIVE	SNV
PTEN C136R	p.C136R	COSM5096	NEGATIVE	SNV
PTEN C136Y	p.C136Y	COSM12734	NEGATIVE	SNV
PTEN C71Y	p.C71Y	COSM5102	NEGATIVE	SNV
PTEN D24E	p.D24E	COSM5198	NEGATIVE	SNV
PTEN D24G	p.D24G	COSM5107	NEGATIVE	SNV
PTEN D24H	p.D24H	COSM1603823	NEGATIVE	SNV
PTEN D24N	p.D24N	COSM5079	NEGATIVE	SNV
PTEN D24V	p.D24V	COSM4016448	NEGATIVE	SNV
PTEN D24Y	p.D24Y	COSM5237	NEGATIVE	SNV
PTEN F341V	p.F341V	COSM5255	NEGATIVE	SNV
PTEN G127E	p.G127E	COSM5143	NEGATIVE	SNV
PTEN G129E	p.G129E	COSM28917	NEGATIVE	SNV
PTEN G129R	p.G129R	COSM246853	NEGATIVE	SNV
PTEN G129V	p.G129V	COSM5276	NEGATIVE	SNV
PTEN L181P	p.L181P	MCH17	NEGATIVE	SNV
PTEN L345Q	p.L345Q	COSM5213	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN N48K	p.N48K	COSM4392678	NEGATIVE	SNV
PTEN R130*	p.R130*	COSM5152	NEGATIVE	SNV
PTEN R130G	p.R130G	COSM5219	NEGATIVE	SNV
PTEN R130L	p.R130L	COSM5216	NEGATIVE	SNV
PTEN R130P	p.R130P	COSM5277	NEGATIVE	SNV
PTEN R130Q	p.R130Q	COSM5033	NEGATIVE	SNV
PTEN R173C	p.R173C	COSM5089	NEGATIVE	SNV
PTEN R173H	p.R173H	COSM5039	NEGATIVE	SNV
PTEN R173P	p.R173P	COSM12735	NEGATIVE	SNV
PTEN S170N	p.S170N	COSM5045	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH20	NEGATIVE	SNV
PTEN S170R	p.S170R	COSM6006454	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH21	NEGATIVE	SNV
PTEN T131I	p.T131I	COSM5104	NEGATIVE	SNV
PTEN V133I	p.V133I	COSM5044	NEGATIVE	SNV
PTEN V343E	p.V343E	MCH18	NEGATIVE	SNV
PTEN Y155C	p.Y155C	COSM5144	NEGATIVE	SNV
PTEN Y16C	p.Y16C	COSM5133	NEGATIVE	SNV
PTEN Y174C	p.Y174C	COSM5575601	NEGATIVE	SNV
PTEN Y174N	p.Y174N	COSM5221	NEGATIVE	SNV
RAF1 S257L	p.S257L	COSM181063	NEGATIVE	SNV
RAF1 S257P	p.S257P	ClinVar40600	NEGATIVE	SNV
RAF1 S257W	p.S257W	COSM581519	NEGATIVE	SNV

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 S259F	p.S259F	COSM1536534	NEGATIVE	SNV
RAF1 S259P	p.S259P	COSM1418555	NEGATIVE	SNV
RAF1 S259T	p.S259T	ClinVar40601	NEGATIVE	SNV
RET A883F	p.A883F	COSM977	NEGATIVE	mnp
RET A883S	p.A883S	COSM133167	NEGATIVE	SNV
RET A883T	p.A883T	COSM100081	NEGATIVE	SNV
RET A883V	p.A883V	COSM4594153	NEGATIVE	SNV
RET A883Y	p.A883Y	OM3415	NEGATIVE	mnp
RET C618F	p.C618F	OM3401	NEGATIVE	SNV
RET C618G	p.C618G	COSM9214400	NEGATIVE	SNV
RET C618R	p.C618R	COSM29803	NEGATIVE	SNV
RET C618S	p.C618S	COSM87267	NEGATIVE	SNV
RET C618S	p.C618S	OM3402	NEGATIVE	SNV
RET C618T	p.C618T	OM3400	NEGATIVE	mnp
RET C618W	p.C618W	OM3403	NEGATIVE	SNV
RET C618Y	p.C618Y	COSM980	NEGATIVE	SNV
RET C620F	p.C620F	OM3405	NEGATIVE	SNV
RET C620G	p.C620G	OM3404	NEGATIVE	SNV
RET C620R	p.C620R	COSM29804	NEGATIVE	SNV
RET C620S	p.C620S	COSM5946160	NEGATIVE	SNV
RET C620S	p.C620S	COSM29805	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET C620W	p.C620W	COSM249815	NEGATIVE	SNV
RET C620Y	p.C620Y	COSM7403807	NEGATIVE	SNV
RET C630F	p.C630F	OM3406	NEGATIVE	SNV
RET C630G	p.C630G	COSM29806	NEGATIVE	SNV
RET C630R	p.C630R	COSM964	NEGATIVE	SNV
RET C630Y	p.C630Y	COSM9233214	NEGATIVE	SNV
RET C634F	p.C634F	COSM1237919	NEGATIVE	SNV
RET C634G	p.C634G	COSM1738369	NEGATIVE	SNV
RET C634L	p.C634L	OM3408	NEGATIVE	mnp
RET C634R	p.C634R	COSM966	NEGATIVE	SNV
RET C634S	p.C634S	COSM1237918	NEGATIVE	SNV
RET C634S	p.C634S	COSM1666664	NEGATIVE	SNV
RET C634W	p.C634W	COSM975	NEGATIVE	SNV
RET C634Y	p.C634Y	COSM974	NEGATIVE	SNV
RET C634_R635dup	p.C634_R635dup	OMINDEL1177	NEGATIVE	ins
RET C634_R635insHELC	p.C634_R635insHELC	OMINDEL1174	NEGATIVE	ins
RET C634_T636dup	p.C634_T636dup	COSM9213763	NEGATIVE	ins
RET D631Y	p.D631Y	COSM1738370	NEGATIVE	SNV
RET D631_C634del	p.D631_C634del	OMINDEL1172	NEGATIVE	del
RET D631_L633delinsE	p.D631_L633delinsE	COSM983	NEGATIVE	del
RET D631_L633delinsV	p.D631_L633delinsV	OMINDEL1175	NEGATIVE	del
RET D631del	p.D631del	OMINDEL1173	NEGATIVE	del
RET D898_E901del	p.D898_E901del	COSM962	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET E632K	p.E632K	COSM1702268	NEGATIVE	SNV
RET E632_C634delinsG	p.E632_C634delinsG	COSM973	NEGATIVE	del
RET E632_C634dup	p.E632_C634dup	OMINDEL1176	NEGATIVE	ins
RET E632_L633del	p.E632_L633del	COSM968	NEGATIVE	del
RET E632_L633delinsV	p.E632_L633delinsV	COSM982	NEGATIVE	del
RET E632_T636delinsSS	p.E632_T636delinsSS	COSM1048	NEGATIVE	complex
RET E768D	p.E768D	COSM21338	NEGATIVE	SNV
RET E768D	p.E768D	OM3413	NEGATIVE	SNV
RET E768G	p.E768G	COSM1347811	NEGATIVE	SNV
RET E768Q	p.E768Q	COSM1716312	NEGATIVE	SNV
RET E884K	p.E884K	COSM960	NEGATIVE	SNV
RET E884V	p.E884V	COSM1570338	NEGATIVE	SNV
RET G736A	p.G736A	OM3411	NEGATIVE	SNV
RET G810A	p.G810A	OM3290	NEGATIVE	SNV
RET G810C	p.G810C	OM3557	NEGATIVE	SNV
RET G810R	p.G810R	OM3289	NEGATIVE	SNV
RET G810S	p.G810S	OM3288	NEGATIVE	SNV
RET G810V	p.G810V	OM3558	NEGATIVE	SNV
RET G911D	p.G911D	COSM20888	NEGATIVE	SNV
RET I788N	p.I788N	OM3284	NEGATIVE	SNV
RET L730Q	p.L730Q	OM3409	NEGATIVE	SNV
RET L730R	p.L730R	OM3410	NEGATIVE	SNV
RET L760Q	p.L760Q	OM3412	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET L790F	p.L790F	rs75030001	NEGATIVE	SNV
RET L881V	p.L881V	OM3414	NEGATIVE	SNV
RET P628_L633del	p.P628_L633del	OMINDEL1171	NEGATIVE	del
RET R770Q	p.R770Q	COSM6006462	NEGATIVE	SNV
RET R886W	p.R886W	COSM6942691	NEGATIVE	SNV
RET R897*	p.R897*	COSM6983129	NEGATIVE	SNV
RET R912L	p.R912L	COSM188545	NEGATIVE	SNV
RET R912P	p.R912P	OM3419	NEGATIVE	SNV
RET R912Q	p.R912Q	COSM3437793	NEGATIVE	SNV
RET R912W	p.R912W	COSM3415038	NEGATIVE	SNV
RET S891A	p.S891A	rs75234356	NEGATIVE	SNV
RET S904C	p.S904C	OM3418	NEGATIVE	SNV
RET S904F	p.S904F	COSM6438204	NEGATIVE	SNV
RET V804G	p.V804G	OM3554	NEGATIVE	SNV
RET V804L	p.V804L	OM3285	NEGATIVE	SNV
RET V804L	p.V804L	OM3286	NEGATIVE	SNV
RET V804M	p.V804M	OM3195	NEGATIVE	SNV
RET V899_D903del	p.V899_D903del	COSM5991595	NEGATIVE	del
RET Y806C	p.Y806C	OM3287	NEGATIVE	SNV
RET Y806F	p.Y806F	OM3556	NEGATIVE	SNV
RET [D903E;S904P]	p.[D903E;S904P]	OM3416	NEGATIVE	mnp
RET [D903E;S904P]	p.[D903E;S904P]	OM3417	NEGATIVE	mnp
RET [E632D;L633V;C634R]	p.[E632D;L633V;C634R]	OM3407	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET [V882=;A883F]	p.[V882=;A883F]	COSM981	NEGATIVE	mnp
RET c.1880_1899delinsCTCAC	p.?	OMINDEL1170	NEGATIVE	complex
ROS1 D2033N	p.D2033N	COSM6924065	NEGATIVE	SNV
ROS1 G2032K	p.G2032K	OM3533	NEGATIVE	mnp
ROS1 G2032R	p.G2032R	MAN10	NEGATIVE	SNV
ROS1 G2032R	p.G2032R	MAN11	NEGATIVE	SNV
ROS1 L1951M	p.L1951M	COSM1072523	NEGATIVE	SNV
ROS1 L1951R	p.L1951R	OM3264	NEGATIVE	SNV
ROS1 L2026M	p.L2026M	BT151	NEGATIVE	SNV
ROS1 S1986F	p.S1986F	COSM6929870	NEGATIVE	SNV
ROS1 S1986Y	p.S1986Y	OM3170	NEGATIVE	SNV
STK11 A205T	p.A205T	COSM20953	NEGATIVE	SNV
STK11 D176N	p.D176N	COSM4827690	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3710	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3711	NEGATIVE	SNV
STK11 D194H	p.D194H	COSM5325765	NEGATIVE	SNV
STK11 D194N	p.D194N	COSM25847	NEGATIVE	SNV
STK11 D194V	p.D194V	COSM20957	NEGATIVE	SNV
STK11 D194Y	p.D194Y	COSM20944	NEGATIVE	SNV
STK11 D53Gfs*110	p.D53GfsTer110	COSM391251	NEGATIVE	ins
STK11 L290P	p.L290P	OM3719	NEGATIVE	SNV
STK11 L67P	p.L67P	OM3700	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 R304W	p.R304W	COSM29468	NEGATIVE	SNV
STK11 S240W	p.S240W	OM3714	NEGATIVE	SNV
STK11 c.1108+1G>A	p.?	OM3722	NEGATIVE	SNV
STK11 c.289_290+1delinsGAT	p.?	COSM6922318	NEGATIVE	mnp
STK11 c.289_290+2delAAGT	p.?	OMINDEL1000	NEGATIVE	del
STK11 c.290+1G>A	p.?	COSM6350392	NEGATIVE	SNV
STK11 c.290+1G>C	p.?	OM3701	NEGATIVE	SNV
STK11 c.290+1G>T	p.?	OM3702	NEGATIVE	SNV
STK11 c.290+1_290+2delGT	p.?	OMINDEL1001	NEGATIVE	del
STK11 c.290+2T>A	p.?	COSM6925006	NEGATIVE	SNV
STK11 c.290+2T>G	p.?	COSM25857	NEGATIVE	SNV
STK11 c.290+2delT	p.?	OMINDEL1002	NEGATIVE	del
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.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.375-1C>G	p.?	COSM4074282	NEGATIVE	SNV
STK11 c.375-1C>T	p.?	COSM6977173	NEGATIVE	SNV
STK11 c.375-2A>C	p.?	OM3706	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.375-2A>G	p.?	OM3707	NEGATIVE	SNV
STK11 c.375-2A>T	p.?	COSM6967966	NEGATIVE	SNV
STK11 c.375-2_386delACGTATATGGTGAT	p.?	OMINDEL1006	NEGATIVE	del
STK11 c.375-6_375-2delCCCCA	p.?	OMINDEL1005	NEGATIVE	del
STK11 c.465-1G>A	p.?	COSM25855	NEGATIVE	SNV
STK11 c.465-1G>C	p.?	OM3709	NEGATIVE	SNV
STK11 c.465-1G>T	p.?	COSM21570	NEGATIVE	SNV
STK11 c.465-1_465delinsTT	p.?	COSM6928792	NEGATIVE	mnp
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.465-2_465-1delAG	p.?	OMINDEL1009	NEGATIVE	del
STK11 c.465-2_465-1delinsTC	p.?	OM3708	NEGATIVE	mnp
STK11 c.465-4_472delGCAGGTACTTCT	p.?	OMINDEL1007	NEGATIVE	del
STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT	p.?	OMINDEL1008	NEGATIVE	del
STK11 c.598-1G>A	p.?	COSM3724562	NEGATIVE	SNV
STK11 c.598-1G>T	p.?	COSM51521	NEGATIVE	SNV
STK11 c.598-1_598delinsTT	p.?	OM3713	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.598-1_612delGGCACTGCACCCGTTC	p.?	OMINDEL1014	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.598-2_600delAGGCA	p.?	OMINDEL1012	NEGATIVE	del
STK11 c.727_734+9delGTCACCCTGTAAGTGCC	p.?	OMINDEL1015	NEGATIVE	del
STK11 c.731_734+9delCCCTGTAAGTGCC	p.?	OMINDEL1016	NEGATIVE	del
STK11 c.734+1G>A	p.?	COSM6916684	NEGATIVE	SNV
STK11 c.734+1G>C	p.?	COSM6970016	NEGATIVE	SNV
STK11 c.734+1G>T	p.?	COSM51523	NEGATIVE	SNV
STK11 c.734+2T>A	p.?	OM3715	NEGATIVE	SNV
STK11 c.734+2T>G	p.?	COSM6961577	NEGATIVE	SNV
STK11 c.734+2_734+10delTAAGTGCCC	p.?	OMINDEL1017	NEGATIVE	del
STK11 c.735-1G>A	p.?	COSM5732040	NEGATIVE	SNV
STK11 c.735-1G>T	p.?	COSM51522	NEGATIVE	SNV
STK11 c.735-2A>G	p.?	OM3716	NEGATIVE	SNV
STK11 c.735-2A>T	p.?	COSM25856	NEGATIVE	SNV
STK11 c.850_862+4delGACCTGCTGAAAGGTGG	p.?	OMINDEL1018	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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+2T>A	p.?	COSM5576087	NEGATIVE	SNV
STK11 c.862+2T>G	p.?	OM3718	NEGATIVE	SNV
STK11 c.862+3delG	p.?	COSM8554973	NEGATIVE	del
STK11 c.863-1G>A	p.?	COSM6916725	NEGATIVE	SNV
STK11 c.863-1G>T	p.?	COSM6925330	NEGATIVE	SNV
STK11 c.863-2A>C	p.?	COSM6912582	NEGATIVE	SNV
STK11 c.863-2A>T	p.?	COSM401786	NEGATIVE	SNV
STK11 c.918_920+11delCAGGTGAGCGGCCC	p.?	OMINDEL1019	NEGATIVE	del
STK11 c.920+1G>A	p.?	OM3720	NEGATIVE	SNV
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
TP53 A138V	p.A138V	COSM43818	NEGATIVE	SNV
TP53 C135F	p.C135F	COSM10647	NEGATIVE	SNV
TP53 C135G	p.C135G	COSM44829	NEGATIVE	SNV
TP53 C135R	p.C135R	COSM10684	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44643	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44910	NEGATIVE	SNV
TP53 C135W	p.C135W	COSM44219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C135Y	p.C135Y	COSM10801	NEGATIVE	SNV
TP53 C141F	p.C141F	COSM44911	NEGATIVE	SNV
TP53 C141G	p.C141G	COSM45794	NEGATIVE	SNV
TP53 C141R	p.C141R	COSM43901	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM293910	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM44564	NEGATIVE	SNV
TP53 C141W	p.C141W	COSM44204	NEGATIVE	SNV
TP53 C141Y	p.C141Y	COSM43708	NEGATIVE	SNV
TP53 C176F	p.C176F	COSM10645	NEGATIVE	SNV
TP53 C176G	p.C176G	COSM44692	NEGATIVE	SNV
TP53 C176R	p.C176R	COSM44948	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44645	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44146	NEGATIVE	SNV
TP53 C176W	p.C176W	COSM11114	NEGATIVE	SNV
TP53 C176Y	p.C176Y	COSM10687	NEGATIVE	SNV
TP53 C238F	p.C238F	COSM43778	NEGATIVE	SNV
TP53 C238G	p.C238G	COSM46336	NEGATIVE	SNV
TP53 C238R	p.C238R	COSM44321	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM44653	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM43700	NEGATIVE	SNV
TP53 C238W	p.C238W	COSM44676	NEGATIVE	SNV
TP53 C238Y	p.C238Y	COSM11059	NEGATIVE	SNV
TP53 C242F	p.C242F	COSM10810	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C242G	p.C242G	COSM44135	NEGATIVE	SNV
TP53 C242R	p.C242R	COSM11738	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM11133	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM44935	NEGATIVE	SNV
TP53 C242W	p.C242W	COSM11356	NEGATIVE	SNV
TP53 C242Y	p.C242Y	COSM10646	NEGATIVE	SNV
TP53 C275F	p.C275F	COSM10701	NEGATIVE	SNV
TP53 C275G	p.C275G	COSM11501	NEGATIVE	SNV
TP53 C275R	p.C275R	COSM43902	NEGATIVE	SNV
TP53 C275S	p.C275S	COSM45413	NEGATIVE	SNV
TP53 C275W	p.C275W	COSM43823	NEGATIVE	SNV
TP53 C275Y	p.C275Y	COSM10893	NEGATIVE	SNV
TP53 C277F	p.C277F	COSM10749	NEGATIVE	SNV
TP53 D281A	p.D281A	COSM11665	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43837	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43906	NEGATIVE	SNV
TP53 D281G	p.D281G	COSM11232	NEGATIVE	SNV
TP53 D281H	p.D281H	COSM10943	NEGATIVE	SNV
TP53 D281N	p.D281N	COSM43596	NEGATIVE	SNV
TP53 D281V	p.D281V	COSM45729	NEGATIVE	SNV
TP53 D281Y	p.D281Y	COSM11516	NEGATIVE	SNV
TP53 E258A	p.E258A	COSM44719	NEGATIVE	SNV
TP53 E258D	p.E258D	COSM44962	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 E258D	p.E258D	COSM46410	NEGATIVE	SNV
TP53 E258G	p.E258G	COSM44168	NEGATIVE	SNV
TP53 E258K	p.E258K	COSM10988	NEGATIVE	SNV
TP53 E258Q	p.E258Q	COSM10751	NEGATIVE	SNV
TP53 E258V	p.E258V	COSM44450	NEGATIVE	SNV
TP53 E285A	p.E285A	COSM43614	NEGATIVE	SNV
TP53 E285G	p.E285G	COSM45649	NEGATIVE	SNV
TP53 E285K	p.E285K	COSM10722	NEGATIVE	SNV
TP53 E285Q	p.E285Q	COSM45138	NEGATIVE	SNV
TP53 E285V	p.E285V	COSM44227	NEGATIVE	SNV
TP53 E286A	p.E286A	COSM45892	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45439	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45492	NEGATIVE	SNV
TP53 E286G	p.E286G	COSM43565	NEGATIVE	SNV
TP53 E286K	p.E286K	COSM10726	NEGATIVE	SNV
TP53 E286Q	p.E286Q	COSM44250	NEGATIVE	SNV
TP53 E286V	p.E286V	COSM43936	NEGATIVE	SNV
TP53 G154V	p.G154V	COSM6815	NEGATIVE	SNV
TP53 G244A	p.G244A	COSM12013	NEGATIVE	SNV
TP53 G244C	p.G244C	COSM11524	NEGATIVE	SNV
TP53 G244D	p.G244D	COSM10883	NEGATIVE	SNV
TP53 G244R	p.G244R	COSM44221	NEGATIVE	SNV
TP53 G244S	p.G244S	COSM10941	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 G244V	p.G244V	COSM43652	NEGATIVE	SNV
TP53 G245A	p.G245A	COSM43965	NEGATIVE	SNV
TP53 G245C	p.G245C	COSM11081	NEGATIVE	SNV
TP53 G245D	p.G245D	COSM43606	NEGATIVE	SNV
TP53 G245R	p.G245R	COSM10957	NEGATIVE	SNV
TP53 G245S	p.G245S	COSM6932	NEGATIVE	SNV
TP53 G245V	p.G245V	COSM11196	NEGATIVE	SNV
TP53 G262V	p.G262V	COSM11198	NEGATIVE	SNV
TP53 G266A	p.G266A	COSM45488	NEGATIVE	SNV
TP53 G266E	p.G266E	COSM10867	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM10794	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM11205	NEGATIVE	SNV
TP53 G266V	p.G266V	COSM10958	NEGATIVE	SNV
TP53 G279E	p.G279E	COSM43714	NEGATIVE	SNV
TP53 H179D	p.H179D	COSM44776	NEGATIVE	SNV
TP53 H179L	p.H179L	COSM43635	NEGATIVE	SNV
TP53 H179N	p.H179N	COSM44151	NEGATIVE	SNV
TP53 H179P	p.H179P	COSM44218	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM11249	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM44214	NEGATIVE	SNV
TP53 H179R	p.H179R	COSM10889	NEGATIVE	SNV
TP53 H179Y	p.H179Y	COSM10768	NEGATIVE	SNV
TP53 H193D	p.H193D	COSM44002	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 H193L	p.H193L	COSM11066	NEGATIVE	SNV
TP53 H193N	p.H193N	COSM43935	NEGATIVE	SNV
TP53 H193P	p.H193P	COSM43833	NEGATIVE	SNV
TP53 H193R	p.H193R	COSM10742	NEGATIVE	SNV
TP53 H193Y	p.H193Y	COSM10672	NEGATIVE	SNV
TP53 H214D	p.H214D	COSM45115	NEGATIVE	SNV
TP53 H214L	p.H214L	COSM5632535	NEGATIVE	SNV
TP53 H214P	p.H214P	COSM1640841	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM44407	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM46272	NEGATIVE	SNV
TP53 H214R	p.H214R	COSM43687	NEGATIVE	SNV
TP53 H214Y	p.H214Y	COSM44112	NEGATIVE	SNV
TP53 I195F	p.I195F	COSM44633	NEGATIVE	SNV
TP53 I195L	p.I195L	COSM45773	NEGATIVE	SNV
TP53 I195M	p.I195M	COSM562637	NEGATIVE	SNV
TP53 I195N	p.I195N	COSM44877	NEGATIVE	SNV
TP53 I195S	p.I195S	COSM44539	NEGATIVE	SNV
TP53 I195T	p.I195T	COSM11089	NEGATIVE	SNV
TP53 K132E	p.K132E	COSM10813	NEGATIVE	SNV
TP53 K132M	p.K132M	COSM43592	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM10991	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM43963	NEGATIVE	SNV
TP53 K132Q	p.K132Q	COSM11224	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 K132R	p.K132R	COSM11582	NEGATIVE	SNV
TP53 K132T	p.K132T	COSM43912	NEGATIVE	SNV
TP53 L145P	p.L145P	COSM43899	NEGATIVE	SNV
TP53 L194F	p.L194F	COSM10995	NEGATIVE	SNV
TP53 L194H	p.L194H	COSM43623	NEGATIVE	SNV
TP53 L194I	p.L194I	COSM45330	NEGATIVE	SNV
TP53 L194P	p.L194P	COSM43827	NEGATIVE	SNV
TP53 L194R	p.L194R	COSM44571	NEGATIVE	SNV
TP53 L194V	p.L194V	COSM46117	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM10834	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM11063	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM44415	NEGATIVE	SNV
TP53 M237K	p.M237K	COSM43952	NEGATIVE	SNV
TP53 M237L	p.M237L	COSM44965	NEGATIVE	SNV
TP53 M237R	p.M237R	COSM45032	NEGATIVE	SNV
TP53 M237T	p.M237T	COSM45329	NEGATIVE	SNV
TP53 M237V	p.M237V	COSM44525	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM10757	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM44310	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM46136	NEGATIVE	SNV
TP53 M246K	p.M246K	COSM44103	NEGATIVE	SNV
TP53 M246L	p.M246L	COSM45992	NEGATIVE	SNV
TP53 M246R	p.M246R	COSM11376	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 M246T	p.M246T	COSM11355	NEGATIVE	SNV
TP53 M246V	p.M246V	COSM43555	NEGATIVE	SNV
TP53 N239D	p.N239D	COSM10777	NEGATIVE	SNV
TP53 N239I	p.N239I	COSM307333	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44510	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44647	NEGATIVE	SNV
TP53 N239S	p.N239S	COSM44094	NEGATIVE	SNV
TP53 N239T	p.N239T	COSM43801	NEGATIVE	SNV
TP53 N239Y	p.N239Y	COSM45870	NEGATIVE	SNV
TP53 P151A	p.P151A	COSM44944	NEGATIVE	SNV
TP53 P151H	p.P151H	COSM11476	NEGATIVE	SNV
TP53 P151L	p.P151L	COSM44288	NEGATIVE	SNV
TP53 P151R	p.P151R	COSM44003	NEGATIVE	SNV
TP53 P151S	p.P151S	COSM10905	NEGATIVE	SNV
TP53 P151T	p.P151T	COSM43911	NEGATIVE	SNV
TP53 P152A	p.P152A	COSM44788	NEGATIVE	SNV
TP53 P152L	p.P152L	COSM10790	NEGATIVE	SNV
TP53 P152Q	p.P152Q	COSM44613	NEGATIVE	SNV
TP53 P152R	p.P152R	COSM45505	NEGATIVE	SNV
TP53 P152S	p.P152S	COSM43582	NEGATIVE	SNV
TP53 P152T	p.P152T	COSM44561	NEGATIVE	SNV
TP53 P177_C182del	p.P177_C182del	COSM43570	NEGATIVE	del
TP53 P190L	p.P190L	COSM43657	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 P191del	p.P191del	COSM45140	NEGATIVE	del
TP53 P250L	p.P250L	COSM10771	NEGATIVE	SNV
TP53 P278A	p.P278A	COSM10814	NEGATIVE	SNV
TP53 P278H	p.P278H	COSM43755	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 Q192*	p.Q192*	COSM10733	NEGATIVE	SNV
TP53 R156P	p.R156P	COSM10760	NEGATIVE	SNV
TP53 R175C	p.R175C	COSM43680	NEGATIVE	SNV
TP53 R175G	p.R175G	COSM10870	NEGATIVE	SNV
TP53 R175H	p.R175H	COSM10648	NEGATIVE	SNV
TP53 R175L	p.R175L	COSM10718	NEGATIVE	SNV
TP53 R175P	p.R175P	COSM45416	NEGATIVE	SNV
TP53 R175S	p.R175S	COSM43931	NEGATIVE	SNV
TP53 R196*	p.R196*	COSM10705	NEGATIVE	SNV
TP53 R213*	p.R213*	COSM10654	NEGATIVE	SNV
TP53 R213G	p.R213G	COSM44102	NEGATIVE	SNV
TP53 R213L	p.R213L	COSM43650	NEGATIVE	SNV
TP53 R213P	p.R213P	COSM11860	NEGATIVE	SNV
TP53 R213Q	p.R213Q	COSM10735	NEGATIVE	SNV
TP53 R248G	p.R248G	COSM11564	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R248L	p.R248L	COSM6549	NEGATIVE	SNV
TP53 R248P	p.R248P	COSM11491	NEGATIVE	SNV
TP53 R248Q	p.R248Q	COSM10662	NEGATIVE	SNV
TP53 R248W	p.R248W	COSM10656	NEGATIVE	SNV
TP53 R249G	p.R249G	COSM10668	NEGATIVE	SNV
TP53 R249K	p.R249K	COSM44091	NEGATIVE	SNV
TP53 R249M	p.R249M	COSM43871	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10785	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10817	NEGATIVE	SNV
TP53 R249T	p.R249T	COSM43665	NEGATIVE	SNV
TP53 R249W	p.R249W	COSM43629	NEGATIVE	SNV
TP53 R273C	p.R273C	COSM10659	NEGATIVE	SNV
TP53 R273G	p.R273G	COSM43843	NEGATIVE	SNV
TP53 R273H	p.R273H	COSM10660	NEGATIVE	SNV
TP53 R273L	p.R273L	COSM10779	NEGATIVE	SNV
TP53 R273P	p.R273P	COSM43896	NEGATIVE	SNV
TP53 R273S	p.R273S	COSM43909	NEGATIVE	SNV
TP53 R280G	p.R280G	COSM11123	NEGATIVE	SNV
TP53 R280I	p.R280I	COSM11287	NEGATIVE	SNV
TP53 R280K	p.R280K	COSM10728	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44171	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44233	NEGATIVE	SNV
TP53 R280T	p.R280T	COSM10724	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R282G	p.R282G	COSM10992	NEGATIVE	SNV
TP53 R282L	p.R282L	COSM44470	NEGATIVE	SNV
TP53 R282P	p.R282P	COSM44306	NEGATIVE	SNV
TP53 R282Q	p.R282Q	COSM44338	NEGATIVE	SNV
TP53 R282W	p.R282W	COSM10704	NEGATIVE	SNV
TP53 R283P	p.R283P	COSM10743	NEGATIVE	SNV
TP53 S240G	p.S240G	COSM43973	NEGATIVE	SNV
TP53 S241A	p.S241A	COSM44224	NEGATIVE	SNV
TP53 S241C	p.S241C	COSM10709	NEGATIVE	SNV
TP53 S241F	p.S241F	COSM10812	NEGATIVE	SNV
TP53 S241P	p.S241P	COSM44578	NEGATIVE	SNV
TP53 S241T	p.S241T	COSM44067	NEGATIVE	SNV
TP53 S241Y	p.S241Y	COSM10935	NEGATIVE	SNV
TP53 V157F	p.V157F	COSM10670	NEGATIVE	SNV
TP53 V157L	p.V157L	COSM45120	NEGATIVE	SNV
TP53 V172F	p.V172F	COSM44240	NEGATIVE	SNV
TP53 V173A	p.V173A	COSM44327	NEGATIVE	SNV
TP53 V173E	p.V173E	COSM45731	NEGATIVE	SNV
TP53 V173G	p.V173G	COSM44383	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM43559	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM44057	NEGATIVE	SNV
TP53 V173M	p.V173M	COSM11084	NEGATIVE	SNV
TP53 V197M	p.V197M	COSM43779	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 V216A	p.V216A	COSM44567	NEGATIVE	SNV
TP53 V216E	p.V216E	COSM44274	NEGATIVE	SNV
TP53 V216G	p.V216G	COSM43681	NEGATIVE	SNV
TP53 V216L	p.V216L	COSM11210	NEGATIVE	SNV
TP53 V216M	p.V216M	COSM10667	NEGATIVE	SNV
TP53 V272A	p.V272A	COSM44294	NEGATIVE	SNV
TP53 V272E	p.V272E	COSM44580	NEGATIVE	SNV
TP53 V272G	p.V272G	COSM44870	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM10859	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM45898	NEGATIVE	SNV
TP53 V272M	p.V272M	COSM10891	NEGATIVE	SNV
TP53 V274F	p.V274F	COSM10769	NEGATIVE	SNV
TP53 V274L	p.V274L	COSM44443	NEGATIVE	SNV
TP53 Y220C	p.Y220C	COSM10758	NEGATIVE	SNV
TP53 Y220D	p.Y220D	COSM11847	NEGATIVE	SNV
TP53 Y220F	p.Y220F	COSM1564197	NEGATIVE	SNV
TP53 Y220H	p.Y220H	COSM44637	NEGATIVE	SNV
TP53 Y220N	p.Y220N	COSM44672	NEGATIVE	SNV
TP53 Y220S	p.Y220S	COSM43850	NEGATIVE	SNV
TP53 Y234C	p.Y234C	COSM10725	NEGATIVE	SNV
TP53 Y234D	p.Y234D	COSM43768	NEGATIVE	SNV
TP53 Y234F	p.Y234F	COSM44315	NEGATIVE	SNV
TP53 Y234H	p.Y234H	COSM11152	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 Y234N	p.Y234N	COSM43956	NEGATIVE	SNV
TP53 Y234S	p.Y234S	COSM43865	NEGATIVE	SNV
TP53 Y236C	p.Y236C	COSM10731	NEGATIVE	SNV
TP53 Y236D	p.Y236D	COSM43602	NEGATIVE	SNV
TP53 Y236F	p.Y236F	COSM3958809	NEGATIVE	SNV
TP53 Y236H	p.Y236H	COSM44326	NEGATIVE	SNV
TP53 Y236N	p.Y236N	COSM43826	NEGATIVE	SNV
TP53 Y236S	p.Y236S	COSM44693	NEGATIVE	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
A2M-ALK fusion	A2M(22) - ALK(19)	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
AGAP1-ALK fusion	AGAP1(6) - ALK(18)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATIC-ALK fusion	ATIC(7) - ALK(20)	Fusion	NEGATIVE
ATRNL1-ALK fusion	ATRNL1(19) - ALK(20)	Fusion	NEGATIVE
BCL11A-ALK fusion	BCL11A(4) - ALK(20)	Fusion	NEGATIVE
BEND5-ALK fusion	BEND5(3) - ALK(20)	Fusion	NEGATIVE
BIRC6-ALK fusion	BIRC6(10) - ALK(20)	Fusion	NEGATIVE
CAD-ALK fusion	CAD(35) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CAD-ALK fusion	CAD(36) - ALK(20)	Fusion	NEGATIVE
CAMKMT-ALK fusion	CAMKMT(3) - ALK(20)	Fusion	NEGATIVE
CARS-ALK fusion	CARS(17) - ALK(20)	Fusion	NEGATIVE
CCDC149-ALK fusion	CCDC149(10) - ALK(20)	Fusion	NEGATIVE
CCDC61-ALK fusion	CCDC61(13) - ALK(20)	Fusion	NEGATIVE
CCDC88A-ALK fusion	CCDC88A(12) - ALK(20)	Fusion	NEGATIVE
CEBPZ-ALK fusion	CEBPZ(2) - ALK(20)	Fusion	NEGATIVE
CENPF-ALK fusion	CENPF(11) - ALK(20)	Fusion	NEGATIVE
CLIP1-ALK fusion	CLIP1(13) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(10) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(14) - ALK(17)	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
COL25A1-ALK fusion	COL25A1(3) - ALK(20)	Fusion	NEGATIVE
CPSF7-ALK fusion	CPSF7(5) - ALK(20)	Fusion	NEGATIVE
CREB1-ALK fusion	CREB1(1) - ALK(9)	Fusion	NEGATIVE
CTNND1-ALK fusion	CTNND1(16) - ALK(20)	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
DDX6-ALK fusion	DDX6(2) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DES-ALK fusion	DES(7) - ALK(19)	Fusion	NEGATIVE
DMD-ALK fusion	DMD(55) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(8) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(6) - ALK(20)	Fusion	NEGATIVE
EIF2AK3-ALK fusion	EIF2AK3(2) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(17) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(2) - 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(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(3) - 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(19)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(18)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(17)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(16)	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

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(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(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(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(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

Variant Name	Genes (Exons)	Variant Type	Test Result
EML4-ALK fusion	EML4(22) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(4) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(5) - ALK(20)	Fusion	NEGATIVE
FBXO36-ALK fusion	FBXO36(2) - 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
GALNT14-ALK fusion	GALNT14(1) - ALK(19)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(13) - ALK(20)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(19) - 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
HNRNPA1-ALK fusion	HNRNPA1(2) - ALK(18)	Fusion	NEGATIVE
IGFBP5-ALK fusion	IGFBP5(1) - ALK(19)	Fusion	NEGATIVE
INTS9-ALK fusion	INTS9(8) - ALK(20)	Fusion	NEGATIVE
ITSN2-ALK fusion	ITSN2(14) - ALK(2)	Fusion	NEGATIVE
KANK2-ALK fusion	KANK2(4) - ALK(16)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KCNQ5-ALK fusion	KCNQ5(1) - ALK(10)	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
KLC1-ALK fusion	KLC1(9) - ALK(20)	Fusion	NEGATIVE
KTN1-ALK fusion	KTN1(43) - ALK(19)	Fusion	NEGATIVE
LMNA-ALK fusion	LMNA(2) - ALK(20)	Fusion	NEGATIVE
LMO7-ALK fusion	LMO7(15) - ALK(20)	Fusion	NEGATIVE
LRPPRC-ALK fusion	LRPPRC(23) - ALK(20)	Fusion	NEGATIVE
LRRFIP2-ALK fusion	LRRFIP2(26) - ALK(20)	Fusion	NEGATIVE
LTBP1-ALK fusion	LTBP1(19) - ALK(20)	Fusion	NEGATIVE
MALAT1-ALK fusion	MALAT1(1) - ALK(16)	Fusion	NEGATIVE
MANBA-ALK fusion	MANBA(14) - ALK(20)	Fusion	NEGATIVE
MAPRE3-ALK fusion	MAPRE3(7) - ALK(20)	Fusion	NEGATIVE
MCFD2-ALK fusion	MCFD2(1) - ALK(20)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(2) - ALK(7)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(1) - ALK(2)	Fusion	NEGATIVE
MLPH-ALK fusion	MLPH(11) - ALK(20)	Fusion	NEGATIVE
MPRIP-ALK fusion	MPRIP(21) - ALK(20)	Fusion	NEGATIVE
MROH2B-ALK fusion	MROH2B(21) - ALK(12)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(9) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MYH10-ALK fusion	MYH10(38) - 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
MYO5C-ALK fusion	MYO5C(30) - ALK(20)	Fusion	NEGATIVE
MYT1L-ALK fusion	MYT1L(14) - ALK(20)	Fusion	NEGATIVE
NCOA1-ALK fusion	NCOA1(21) - ALK(1)	Fusion	NEGATIVE
NPM1-ALK fusion	NPM1(4) - ALK(20)	Fusion	NEGATIVE
NUMA1-ALK fusion	NUMA1(19) - ALK(20)	Fusion	NEGATIVE
OTOF-ALK fusion	OTOF(2) - ALK(3)	Fusion	NEGATIVE
PABPC1-ALK fusion	PABPC1(9) - ALK(20)	Fusion	NEGATIVE
PCM1-ALK fusion	PCM1(23) - ALK(20)	Fusion	NEGATIVE
PDE3A-ALK fusion	PDE3A(10) - ALK(8)	Fusion	NEGATIVE
PDE8B-ALK fusion	PDE8B(11) - ALK(20)	Fusion	NEGATIVE
PHACTR1-ALK fusion	PHACTR1(4) - ALK(20)	Fusion	NEGATIVE
PLEKHA7-ALK fusion	PLEKHA7(26) - ALK(19)	Fusion	NEGATIVE
PNPT1-ALK fusion	PNPT1(5) - ALK(4)	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
PPM1B-ALK fusion	PPM1B(1) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
PPP1R21-ALK fusion	PPP1R21(7) - ALK(20)	Fusion	NEGATIVE
PPP1R21-ALK fusion	PPP1R21(17) - ALK(20)	Fusion	NEGATIVE
PPP4R3B-ALK fusion	PPP4R3B(10) - ALK(2)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(2) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(5) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(10) - ALK(20)	Fusion	NEGATIVE
PRKAR1B-ALK fusion	PRKAR1B(4) - ALK(20)	Fusion	NEGATIVE
PRKAR2A-ALK fusion	PRKAR2A(2) - ALK(20)	Fusion	NEGATIVE
PRKCE-ALK fusion	PRKCE(1) - ALK(20)	Fusion	NEGATIVE
RANBP2-ALK fusion	RANBP2(18) - ALK(20)	Fusion	NEGATIVE
RNF213-ALK fusion	RNF213(20) - ALK(20)	Fusion	NEGATIVE
RRBP1-ALK fusion	RRBP1(20) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(6) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(5) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(22) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(21) - ALK(20)	Fusion	NEGATIVE
SFT2D1-ALK fusion	SFT2D1(1) - ALK(6)	Fusion	NEGATIVE
SLC34A2-ALK fusion	SLC34A2(1) - ALK(20)	Fusion	NEGATIVE
SLC8A1-ALK fusion	SLC8A1(1) - ALK(20)	Fusion	NEGATIVE
SLMAP-ALK fusion	SLMAP(11) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SP100-ALK fusion	SP100(4) - ALK(20)	Fusion	NEGATIVE
SPDYA-ALK fusion	SPDYA(6) - ALK(20)	Fusion	NEGATIVE
SPECC1L-ALK fusion	SPECC1L(9) - 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
SQSTM1-ALK fusion	SQSTM1(5) - ALK(20)	Fusion	NEGATIVE
SQSTM1-ALK fusion	SQSTM1(5) - ALK(19)	Fusion	NEGATIVE
STK32B-ALK fusion	STK32B(11) - ALK(20)	Fusion	NEGATIVE
STRN-ALK fusion	STRN(3) - ALK(20)	Fusion	NEGATIVE
TERT-ALK fusion	TERT(11) - ALK(5)	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(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(5) - ALK(19)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(6) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(19)	Fusion	NEGATIVE
THBS1-ALK fusion	THBS1(4) - ALK(19)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(20)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(19)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(32) - ALK(13)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(28) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(20)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(16) - ALK(17)	Fusion	NEGATIVE
TPM1-ALK fusion	TPM1(8) - ALK(20)	Fusion	NEGATIVE
TPM3-ALK fusion	TPM3(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(15) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(4) - ALK(20)	Fusion	NEGATIVE
TRAF1-ALK fusion	TRAF1(6) - ALK(20)	Fusion	NEGATIVE
TRMT61B-ALK fusion	TRMT61B(1) - ALK(9)	Fusion	NEGATIVE
VCL-ALK fusion	VCL(16) - ALK(20)	Fusion	NEGATIVE
YPEL5-ALK fusion	YPEL5(1) - ALK(18)	Fusion	NEGATIVE
ZNF2-ALK fusion	ZNF2(1) - ALK(20)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(11)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(9)	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
AGK-BRAF fusion	AGK(2) - BRAF(8)	Fusion	NEGATIVE
AGTRAP-BRAF fusion	AGTRAP(5) - BRAF(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
AP3B1-BRAF fusion	AP3B1(22) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(3) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(4) - BRAF(11)	Fusion	NEGATIVE
ATG7-BRAF fusion	ATG7(18) - BRAF(9)	Fusion	NEGATIVE
BAIAP2L1-BRAF fusion	BAIAP2L1(12) - BRAF(9)	Fusion	NEGATIVE
BBS9-BRAF fusion	BBS9(19) - BRAF(4)	Fusion	NEGATIVE
BCAP29-BRAF fusion	BCAP29(7) - BRAF(11)	Fusion	NEGATIVE
BCL2L11-BRAF fusion	BCL2L11(3) - BRAF(10)	Fusion	NEGATIVE
BTF3L4-BRAF fusion	BTF3L4(3) - BRAF(11)	Fusion	NEGATIVE
C7orf73-BRAF fusion	C7orf73(2) - BRAF(9)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(2) - BRAF(8)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(1) - BRAF(9)	Fusion	NEGATIVE
CCDC91-BRAF fusion	CCDC91(11) - BRAF(9)	Fusion	NEGATIVE
CCNY-BRAF fusion	CCNY(1) - BRAF(10)	Fusion	NEGATIVE
CDC27-BRAF fusion	CDC27(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CDC42BPB-BRAF fusion	CDC42BPB(36) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(8)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(12) - BRAF(11)	Fusion	NEGATIVE
CEP89-BRAF fusion	CEP89(16) - BRAF(9)	Fusion	NEGATIVE
CLCN6-BRAF fusion	CLCN6(2) - BRAF(11)	Fusion	NEGATIVE
CLIP2-BRAF fusion	CLIP2(6) - BRAF(11)	Fusion	NEGATIVE
COBL-BRAF fusion	COBL(7) - BRAF(10)	Fusion	NEGATIVE
CUL1-BRAF fusion	CUL1(7) - BRAF(9)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(8) - BRAF(8)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(10) - BRAF(9)	Fusion	NEGATIVE
DGKI-BRAF fusion	DGKI(7) - BRAF(10)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(5) - BRAF(9)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(3) - BRAF(10)	Fusion	NEGATIVE
DOCK4-BRAF fusion	DOCK4(41) - BRAF(9)	Fusion	NEGATIVE
DSTYK-BRAF fusion	DSTYK(1) - BRAF(10)	Fusion	NEGATIVE
DYNC1I2-BRAF fusion	DYNC1I2(7) - BRAF(10)	Fusion	NEGATIVE
EML4-BRAF fusion	EML4(6) - BRAF(10)	Fusion	NEGATIVE
EPS15-BRAF fusion	EPS15(22) - BRAF(10)	Fusion	NEGATIVE
EPS15L1-BRAF fusion	EPS15L1(16) - BRAF(9)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(12) - BRAF(10)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(17) - BRAF(8)	Fusion	NEGATIVE
ESYT2-BRAF fusion	ESYT2(8) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FAM114A2-BRAF fusion	FAM114A2(9) - BRAF(11)	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
FAM73A-BRAF fusion	FAM73A(7) - BRAF(9)	Fusion	NEGATIVE
FCHSD1-BRAF fusion	FCHSD1(13) - BRAF(9)	Fusion	NEGATIVE
FXR1-BRAF fusion	FXR1(13) - BRAF(10)	Fusion	NEGATIVE
GATM-BRAF fusion	GATM(2) - BRAF(11)	Fusion	NEGATIVE
GHR-BRAF fusion	GHR(1) - BRAF(10)	Fusion	NEGATIVE
GIPC2-BRAF fusion	GIPC2(3) - BRAF(10)	Fusion	NEGATIVE
GIT2-BRAF fusion	GIT2(14) - BRAF(9)	Fusion	NEGATIVE
GNAI1-BRAF fusion	GNAI1(1) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(4) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(20) - BRAF(10)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(4) - BRAF(7)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(6) - BRAF(7)	Fusion	NEGATIVE
KANK1-BRAF fusion	KANK1(7) - BRAF(9)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(3) - BRAF(8)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(4) - BRAF(8)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(12) - BRAF(4)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(11) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(18) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(17) - BRAF(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(10)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(8)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(8)	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
KLHL7-BRAF fusion	KLHL7(4) - BRAF(10)	Fusion	NEGATIVE
KLHL7-BRAF fusion	KLHL7(5) - BRAF(9)	Fusion	NEGATIVE
LMO7-BRAF fusion	LMO7(7) - BRAF(9)	Fusion	NEGATIVE
LSM12-BRAF fusion	LSM12(3) - BRAF(9)	Fusion	NEGATIVE
LSM14A-BRAF fusion	LSM14A(9) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(5) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(60) - BRAF(9)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(17) - BRAF(10)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MBP-BRAF fusion	MBP(2) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(11)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(10)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(8)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(3) - BRAF(10)	Fusion	NEGATIVE
MYRIP-BRAF fusion	MYRIP(16) - BRAF(9)	Fusion	NEGATIVE
MZT1-BRAF fusion	MZT1(2) - BRAF(11)	Fusion	NEGATIVE
NFIC-BRAF fusion	NFIC(6) - BRAF(10)	Fusion	NEGATIVE
NRF1-BRAF fusion	NRF1(5) - BRAF(9)	Fusion	NEGATIVE
NUB1-BRAF fusion	NUB1(3) - BRAF(9)	Fusion	NEGATIVE
NUDCD3-BRAF fusion	NUDCD3(4) - BRAF(9)	Fusion	NEGATIVE
NUP214-BRAF fusion	NUP214(21) - BRAF(10)	Fusion	NEGATIVE
OSBPL1A-BRAF fusion	OSBPL1A(16) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(11) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(14) - BRAF(9)	Fusion	NEGATIVE
PACSIN2-BRAF fusion	PACSIN2(7) - BRAF(9)	Fusion	NEGATIVE
PAPSS1-BRAF fusion	PAPSS1(5) - BRAF(9)	Fusion	NEGATIVE
PARP12-BRAF fusion	PARP12(9) - BRAF(11)	Fusion	NEGATIVE
PCM1-BRAF fusion	PCM1(37) - BRAF(9)	Fusion	NEGATIVE
PHTF2-BRAF fusion	PHTF2(17) - BRAF(9)	Fusion	NEGATIVE
PICALM-BRAF fusion	PICALM(19) - BRAF(9)	Fusion	NEGATIVE
PJA2-BRAF fusion	PJA2(7) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PLIN3-BRAF fusion	PLIN3(1) - BRAF(9)	Fusion	NEGATIVE
PLXNA2-BRAF fusion	PLXNA2(9) - BRAF(2)	Fusion	NEGATIVE
POR-BRAF fusion	POR(3) - BRAF(11)	Fusion	NEGATIVE
PPFIBP2-BRAF fusion	PPFIBP2(3) - BRAF(11)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(10) - BRAF(9)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(9) - BRAF(9)	Fusion	NEGATIVE
PRKAR2B-BRAF fusion	PRKAR2B(1) - BRAF(10)	Fusion	NEGATIVE
RAD18-BRAF fusion	RAD18(7) - BRAF(10)	Fusion	NEGATIVE
RBM33-BRAF fusion	RBM33(3) - BRAF(11)	Fusion	NEGATIVE
RBMS3-BRAF fusion	RBMS3(11) - BRAF(11)	Fusion	NEGATIVE
RNF11-BRAF fusion	RNF11(1) - BRAF(11)	Fusion	NEGATIVE
RNF130-BRAF fusion	RNF130(3) - BRAF(9)	Fusion	NEGATIVE
RP2-BRAF fusion	RP2(3) - BRAF(10)	Fusion	NEGATIVE
RUNDC1-BRAF fusion	RUNDC1(1) - BRAF(9)	Fusion	NEGATIVE
SCRIB-BRAF fusion	SCRIB(27) - BRAF(11)	Fusion	NEGATIVE
SLC12A7-BRAF fusion	SLC12A7(17) - BRAF(11)	Fusion	NEGATIVE
SLC45A3-BRAF fusion	SLC45A3(1) - 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(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(9) - BRAF(8)	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(11)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(8)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(6) - BRAF(9)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(5) - BRAF(9)	Fusion	NEGATIVE
STRN3-BRAF fusion	STRN3(3) - BRAF(10)	Fusion	NEGATIVE
STXBP6-BRAF fusion	STXBP6(4) - BRAF(10)	Fusion	NEGATIVE
TANK-BRAF fusion	TANK(4) - BRAF(9)	Fusion	NEGATIVE
TAX1BP1-BRAF fusion	TAX1BP1(8) - BRAF(11)	Fusion	NEGATIVE
TMEM106B-BRAF fusion	TMEM106B(4) - BRAF(8)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(1) - BRAF(10)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(2) - BRAF(9)	Fusion	NEGATIVE
TMPRSS2-BRAF fusion	TMPRSS2(3) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TOM1L2-BRAF fusion	TOM1L2(11) - BRAF(9)	Fusion	NEGATIVE
TPR-BRAF fusion	TPR(14) - BRAF(7)	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
TRIM4-BRAF fusion	TRIM4(6) - BRAF(10)	Fusion	NEGATIVE
UBN2-BRAF fusion	UBN2(3) - BRAF(11)	Fusion	NEGATIVE
ZBTB8A-BRAF fusion	ZBTB8A(4) - BRAF(9)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(7) - BRAF(11)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(3) - BRAF(10)	Fusion	NEGATIVE
ZKSCAN5-BRAF fusion	ZKSCAN5(2) - BRAF(9)	Fusion	NEGATIVE
ZNF207-BRAF fusion	ZNF207(3) - BRAF(10)	Fusion	NEGATIVE
ZNF767P-BRAF fusion	ZNF767P(1) - BRAF(11)	Fusion	NEGATIVE
ZSCAN30-BRAF fusion	ZSCAN30(3) - BRAF(10)	Fusion	NEGATIVE
EGFRvIII	EGFR(1) - EGFR(8)	RNAExonVariant	NEGATIVE
ESR1-ARMT1 fusion	ESR1(3) - ARMT1(4)	Fusion	NEGATIVE
ESR1-ARMT1 fusion	ESR1(6) - ARMT1(2)	Fusion	NEGATIVE
ESR1-ARNT2 fusion	ESR1(7) - ARNT2(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-ATP2B2 fusion	ESR1(4) - ATP2B2(4)	Fusion	NEGATIVE
ESR1-BMPER fusion	ESR1(6) - BMPER(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(2)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(5)	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(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(10)	Fusion	NEGATIVE
ESR1-CDK13 fusion	ESR1(7) - CDK13(13)	Fusion	NEGATIVE
ESR1-COA5 fusion	ESR1(4) - COA5(2)	Fusion	NEGATIVE
ESR1-DAB2 fusion	ESR1(6) - DAB2(3)	Fusion	NEGATIVE
ESR1-GYG2 fusion	ESR1(6) - GYG2(2)	Fusion	NEGATIVE
ESR1-MKL1 fusion	ESR1(6) - MKL1(7)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(6) - MTHFD1L(21)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(7) - MTHFD1L(27)	Fusion	NEGATIVE
ESR1-NKAIN2 fusion	ESR1(6) - NKAIN2(2)	Fusion	NEGATIVE
ESR1-NOP2 fusion	ESR1(6) - NOP2(16)	Fusion	NEGATIVE
ESR1-PCDH11X fusion	ESR1(6) - PCDH11X(6)	Fusion	NEGATIVE
ESR1-PDE10A fusion	ESR1(7) - PDE10A(6)	Fusion	NEGATIVE
ESR1-PLEKHG1 fusion	ESR1(6) - PLEKHG1(12)	Fusion	NEGATIVE
ESR1-POLH fusion	ESR1(6) - POLH(2)	Fusion	NEGATIVE
ESR1-RMND1 fusion	ESR1(6) - RMND1(9)	Fusion	NEGATIVE
ESR1-SOX9 fusion	ESR1(6) - SOX9(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-SYNE1 fusion	ESR1(6) - SYNE1(59)	Fusion	NEGATIVE
ESR1-TFG fusion	ESR1(6) - TFG(4)	Fusion	NEGATIVE
ESR1-TNRC6B fusion	ESR1(6) - TNRC6B(14)	Fusion	NEGATIVE
ESR1-YAP1 fusion	ESR1(6) - YAP1(4)	Fusion	NEGATIVE
PLEKHG1-ESR1 fusion	PLEKHG1(3) - ESR1(2)	Fusion	NEGATIVE
TNS3-ESR1 fusion	TNS3(16) - ESR1(5)	Fusion	NEGATIVE
ACPP-FGFR1 fusion	ACPP(10) - FGFR1(4)	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
BCR-FGFR1 fusion	BCR(4) - FGFR1(10)	Fusion	NEGATIVE
CNTRL-FGFR1 fusion	CNTRL(40) - FGFR1(10)	Fusion	NEGATIVE
CPSF6-FGFR1 fusion	CPSF6(8) - FGFR1(10)	Fusion	NEGATIVE
CUX1-FGFR1 fusion	CUX1(11) - FGFR1(10)	Fusion	NEGATIVE
EGFR-FGFR1 fusion	EGFR(24) - FGFR1(10)	Fusion	NEGATIVE
ERLIN2-FGFR1 fusion	ERLIN2(8) - FGFR1(2)	Fusion	NEGATIVE
ERVK3-1-FGFR1 fusion	ERVK3-1(3) - FGFR1(10)	Fusion	NEGATIVE
FGFR1-ADAM32 fusion	FGFR1(17) - ADAM32(14)	Fusion	NEGATIVE
FGFR1-EVI5 fusion	FGFR1(17) - EVI5(12)	Fusion	NEGATIVE
FGFR1-HOOK3 fusion	FGFR1(17) - HOOK3(5)	Fusion	NEGATIVE
FGFR1-TACC1 fusion	FGFR1(17) - TACC1(7)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(5) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(6) - FGFR1(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR1OP-FGFR1 fusion	FGFR1OP(7) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP2-FGFR1 fusion	FGFR1OP2(4) - 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(4)	Fusion	NEGATIVE
FN1-FGFR1 fusion	FN1(22) - FGFR1(3)	Fusion	NEGATIVE
KAT6A-FGFR1 fusion	KAT6A(5) - FGFR1(2)	Fusion	NEGATIVE
LRRFIP1-FGFR1 fusion	LRRFIP1(8) - FGFR1(10)	Fusion	NEGATIVE
MYO18A-FGFR1 fusion	MYO18A(33) - FGFR1(10)	Fusion	NEGATIVE
RANBP2-FGFR1 fusion	RANBP2(20) - FGFR1(10)	Fusion	NEGATIVE
SNX29-FGFR1 fusion	SNX29(1) - FGFR1(4)	Fusion	NEGATIVE
SQSTM1-FGFR1 fusion	SQSTM1(6) - FGFR1(10)	Fusion	NEGATIVE
TPR-FGFR1 fusion	TPR(22) - FGFR1(10)	Fusion	NEGATIVE
TRIM24-FGFR1 fusion	TRIM24(11) - FGFR1(10)	Fusion	NEGATIVE
WHSC1L1-FGFR1 fusion	WHSC1L1(14) - FGFR1(5)	Fusion	NEGATIVE
ZMYM2-FGFR1 fusion	ZMYM2(17) - FGFR1(10)	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
CD44-FGFR2 fusion	CD44(1) - FGFR2(3)	Fusion	NEGATIVE
COL25A1-FGFR2 fusion	COL25A1(3) - FGFR2(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CTNNB1-FGFR2 fusion	CTNNB1(1) - FGFR2(10)	Fusion	NEGATIVE
FGFR2-ACSL5 fusion	FGFR2(17) - ACSL5(15)	Fusion	NEGATIVE
FGFR2-AFF3 fusion	FGFR2(17) - AFF3(7)	Fusion	NEGATIVE
FGFR2-AHCYL1 fusion	FGFR2(17) - AHCYL1(2)	Fusion	NEGATIVE
FGFR2-ANKRD24 fusion	FGFR2(17) - ANKRD24(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(2)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(10)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(16)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(18)	Fusion	NEGATIVE
FGFR2-CASP7 fusion	FGFR2(17) - CASP7(2)	Fusion	NEGATIVE
FGFR2-CBX5 fusion	FGFR2(17) - CBX5(2)	Fusion	NEGATIVE
FGFR2-CCAR2 fusion	FGFR2(17) - CCAR2(4)	Fusion	NEGATIVE
FGFR2-CCDC186 fusion	FGFR2(17) - CCDC186(4)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(2)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(1)	Fusion	NEGATIVE
FGFR2-CELF2 fusion	FGFR2(17) - CELF2(4)	Fusion	NEGATIVE
FGFR2-CIT fusion	FGFR2(17) - CIT(23)	Fusion	NEGATIVE
FGFR2-CLIP2 fusion	FGFR2(17) - CLIP2(10)	Fusion	NEGATIVE
FGFR2-COL14A1 fusion	FGFR2(17) - COL14A1(34)	Fusion	NEGATIVE
FGFR2-CREB5 fusion	FGFR2(17) - CREB5(8)	Fusion	NEGATIVE
FGFR2-DDX21 fusion	FGFR2(17) - DDX21(11)	Fusion	NEGATIVE
FGFR2-EIF4A2 fusion	FGFR2(17) - EIF4A2(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-FAM76A fusion	FGFR2(17) - FAM76A(2)	Fusion	NEGATIVE
FGFR2-GAB2 fusion	FGFR2(17) - GAB2(2)	Fusion	NEGATIVE
FGFR2-INA fusion	FGFR2(17) - INA(2)	Fusion	NEGATIVE
FGFR2-KCTD1 fusion	FGFR2(17) - KCTD1(2)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(3)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(5)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(6)	Fusion	NEGATIVE
FGFR2-KRT8 fusion	FGFR2(17) - KRT8(6)	Fusion	NEGATIVE
FGFR2-LAMC1 fusion	FGFR2(17) - LAMC1(27)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MGEA5 fusion	FGFR2(17) - MGEA5(12)	Fusion	NEGATIVE
FGFR2-NOL4 fusion	FGFR2(17) - NOL4(7)	Fusion	NEGATIVE
FGFR2-NRAP fusion	FGFR2(17) - NRAP(24)	Fusion	NEGATIVE
FGFR2-NRBF2 fusion	FGFR2(17) - NRBF2(4)	Fusion	NEGATIVE
FGFR2-OFD1 fusion	FGFR2(17) - OFD1(3)	Fusion	NEGATIVE
FGFR2-OGDH fusion	FGFR2(17) - OGDH(3)	Fusion	NEGATIVE
FGFR2-OPTN fusion	FGFR2(17) - OPTN(5)	Fusion	NEGATIVE
FGFR2-PASD1 fusion	FGFR2(17) - PASD1(10)	Fusion	NEGATIVE
FGFR2-PCM1 fusion	FGFR2(17) - PCM1(7)	Fusion	NEGATIVE
FGFR2-PHC1 fusion	FGFR2(17) - PHC1(2)	Fusion	NEGATIVE
FGFR2-PKD2L1 fusion	FGFR2(17) - PKD2L1(7)	Fusion	NEGATIVE
FGFR2-PPHLN1 fusion	FGFR2(17) - PPHLN1(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-RABGAP1L fusion	FGFR2(17) - RABGAP1L(20)	Fusion	NEGATIVE
FGFR2-RASAL2 fusion	FGFR2(17) - RASAL2(3)	Fusion	NEGATIVE
FGFR2-ROCK1 fusion	FGFR2(17) - ROCK1(2)	Fusion	NEGATIVE
FGFR2-SBNO2 fusion	FGFR2(17) - SBNO2(19)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(7)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(5)	Fusion	NEGATIVE
FGFR2-SMN1 fusion	FGFR2(17) - SMN1(2)	Fusion	NEGATIVE
FGFR2-STK26 fusion	FGFR2(17) - STK26(3)	Fusion	NEGATIVE
FGFR2-TACC2 fusion	FGFR2(17) - TACC2(11)	Fusion	NEGATIVE
FGFR2-TACC3 fusion	FGFR2(17) - TACC3(11)	Fusion	NEGATIVE
FGFR2-TBC1D1 fusion	FGFR2(17) - TBC1D1(9)	Fusion	NEGATIVE
FGFR2-TFEC fusion	FGFR2(17) - TFEC(2)	Fusion	NEGATIVE
FGFR2-TJP2 fusion	FGFR2(17) - TJP2(5)	Fusion	NEGATIVE
FGFR2-TXLNA fusion	FGFR2(17) - TXLNA(6)	Fusion	NEGATIVE
FGFR2-UBQLN1 fusion	FGFR2(17) - UBQLN1(8)	Fusion	NEGATIVE
FGFR2-UTRN fusion	FGFR2(17) - UTRN(51)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(2)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(7)	Fusion	NEGATIVE
FGFR2-WAC fusion	FGFR2(17) - WAC(5)	Fusion	NEGATIVE
FGFR2-WARS fusion	FGFR2(17) - WARS(3)	Fusion	NEGATIVE
FGFR2-ZMYM4 fusion	FGFR2(17) - ZMYM4(10)	Fusion	NEGATIVE
KLK2-FGFR2 fusion	KLK2(1) - FGFR2(5)	Fusion	NEGATIVE
PARK2-FGFR2 fusion	PARK2(9) - FGFR2(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDHX-FGFR2 fusion	PDHX(1) - FGFR2(7)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(2)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(1)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(8) - FGFR2(7)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(7) - FGFR2(7)	Fusion	NEGATIVE
ETV6-FGFR3 fusion	ETV6(5) - FGFR3(9)	Fusion	NEGATIVE
FGFR3-ADD1 fusion	FGFR3(17) - ADD1(3)	Fusion	NEGATIVE
FGFR3-AES fusion	FGFR3(17) - AES(2)	Fusion	NEGATIVE
FGFR3-AMBRA1 fusion	FGFR3(17) - AMBRA1(14)	Fusion	NEGATIVE
FGFR3-BAIAP2L1 fusion	FGFR3(17) - BAIAP2L1(2)	Fusion	NEGATIVE
FGFR3-BRAP fusion	FGFR3(17) - BRAP(5)	Fusion	NEGATIVE
FGFR3-CAMK2A fusion	FGFR3(18) - CAMK2A(14)	Fusion	NEGATIVE
FGFR3-ELAVL3 fusion	FGFR3(17) - ELAVL3(2)	Fusion	NEGATIVE
FGFR3-FBXO28 fusion	FGFR3(17) - FBXO28(4)	Fusion	NEGATIVE
FGFR3-JAKMIP1 fusion	FGFR3(17) - JAKMIP1(4)	Fusion	NEGATIVE
FGFR3-NBR1 fusion	FGFR3(17) - NBR1(7)	Fusion	NEGATIVE
FGFR3-RBBP4 fusion	FGFR3(17) - RBBP4(12)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(6)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(8)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(11)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(18) - TACC3(11)	Fusion	NEGATIVE
BAIAP2L1-MET fusion	BAIAP2L1(9) - MET(15)	Fusion	NEGATIVE
C8orf34-MET fusion	C8orf34(2) - 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
CLIP2-MET fusion	CLIP2(11) - MET(15)	Fusion	NEGATIVE
DCTN1-MET fusion	DCTN1(26) - MET(15)	Fusion	NEGATIVE
EPS15-MET fusion	EPS15(21) - MET(15)	Fusion	NEGATIVE
ETV6-MET fusion	ETV6(4) - MET(15)	Fusion	NEGATIVE
GTF2I-MET fusion	GTF2I(6) - MET(15)	Fusion	NEGATIVE
HLA-DRB1-MET fusion	HLA-DRB1(5) - MET(15)	Fusion	NEGATIVE
KIF5B-MET fusion	KIF5B(24) - MET(15)	Fusion	NEGATIVE
LRRFIP1-MET fusion	LRRFIP1(19) - MET(15)	Fusion	NEGATIVE
MET KDD	MET(21) - MET(15)	RNAExonVariant	NEGATIVE
MET exon 14 skipping (RNA)	MET(13) - MET(15)	RNAExonVariant	NEGATIVE
OXR1-MET fusion	OXR1(9) - MET(13)	Fusion	NEGATIVE
OXR1-MET fusion	OXR1(1) - MET(13)	Fusion	NEGATIVE
PPFIBP1-MET fusion	PPFIBP1(9) - MET(15)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
SCAF11-MET fusion	SCAF11(1) - MET(2)	Fusion	NEGATIVE
SLC34A2-MET fusion	SLC34A2(4) - MET(15)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(2)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(11)	Fusion	NEGATIVE
STARD3NL-MET fusion	STARD3NL(6) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(5) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(7) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(36) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(4) - MET(15)	Fusion	NEGATIVE
TRIM4-MET fusion	TRIM4(6) - MET(15)	Fusion	NEGATIVE
ZKSCAN1-MET fusion	ZKSCAN1(3) - MET(15)	Fusion	NEGATIVE
AKAP13-NRG1 fusion	AKAP13(5) - NRG1(2)	Fusion	NEGATIVE
ATP1B1-NRG1 fusion	ATP1B1(2) - 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
CDH1-NRG1 fusion	CDH1(2) - NRG1(2)	Fusion	NEGATIVE
CLU-NRG1 fusion	CLU(2) - NRG1(6)	Fusion	NEGATIVE
COX10-AS1-NRG1 fusion	COX10-AS1(1) - NRG1(2)	Fusion	NEGATIVE
HMBOX1-NRG1 fusion	HMBOX1(1) - NRG1(6)	Fusion	NEGATIVE
KIF13B-NRG1 fusion	KIF13B(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
NOTCH2-NRG1 fusion	NOTCH2(4) - NRG1(6)	Fusion	NEGATIVE
PARP8-NRG1 fusion	PARP8(1) - NRG1(2)	Fusion	NEGATIVE
PDE7A-NRG1 fusion	PDE7A(3) - NRG1(6)	Fusion	NEGATIVE
RAB3IL1-NRG1 fusion	RAB3IL1(9) - NRG1(6)	Fusion	NEGATIVE
RALGAPA1-NRG1 fusion	RALGAPA1(20) - NRG1(6)	Fusion	NEGATIVE
ROCK1-NRG1 fusion	ROCK1(2) - NRG1(2)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(4) - NRG1(6)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(2) - NRG1(6)	Fusion	NEGATIVE
SETD4-NRG1 fusion	SETD4(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(5) - NRG1(6)	Fusion	NEGATIVE
SMAD4-NRG1 fusion	SMAD4(1) - NRG1(6)	Fusion	NEGATIVE
TBL1XR1-NRG1 fusion	TBL1XR1(1) - NRG1(2)	Fusion	NEGATIVE
TENM4-NRG1 fusion	TENM4(12) - NRG1(2)	Fusion	NEGATIVE
THAP7-NRG1 fusion	THAP7(3) - NRG1(2)	Fusion	NEGATIVE
THBS1-NRG1 fusion	THBS1(6) - NRG1(6)	Fusion	NEGATIVE
TNC-NRG1 fusion	TNC(10) - NRG1(6)	Fusion	NEGATIVE
TSHZ2-NRG1 fusion	TSHZ2(1) - NRG1(6)	Fusion	NEGATIVE
VAMP2-NRG1 fusion	VAMP2(4) - NRG1(4)	Fusion	NEGATIVE
VTCN1-NRG1 fusion	VTCN1(2) - NRG1(2)	Fusion	NEGATIVE
WHSC1L1-NRG1 fusion	WHSC1L1(1) - NRG1(2)	Fusion	NEGATIVE
WRN-NRG1 fusion	WRN(30) - NRG1(2)	Fusion	NEGATIVE
ZMYM2-NRG1 fusion	ZMYM2(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
AFAP1-NTRK1 fusion	AFAP1(4) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF2-NTRK1 fusion	ARHGEF2(21) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF25-NTRK1 fusion	ARHGEF25(11) - NTRK1(8)	Fusion	NEGATIVE
BANP-NTRK1 fusion	BANP(4) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(12) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(13) - NTRK1(11)	Fusion	NEGATIVE
CD74-NTRK1 fusion	CD74(7) - NTRK1(10)	Fusion	NEGATIVE
CEL-NTRK1 fusion	CEL(7) - NTRK1(9)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(10)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(11)	Fusion	NEGATIVE
CTRC-NTRK1 fusion	CTRC(1) - NTRK1(9)	Fusion	NEGATIVE
EPHB2-NTRK1 fusion	EPHB2(3) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
GON4L-NTRK1 fusion	GON4L(21) - NTRK1(9)	Fusion	NEGATIVE
GRIPAP1-NTRK1 fusion	GRIPAP1(22) - NTRK1(11)	Fusion	NEGATIVE
GSN-NTRK1 fusion	GSN(7) - NTRK1(10)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(10)	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
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(11)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(10)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(5) - NTRK1(11)	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(11)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(12)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(13)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(11) - NTRK1(11)	Fusion	NEGATIVE
LRRC71-NTRK1 fusion	LRRC71(1) - NTRK1(9)	Fusion	NEGATIVE
MDM4-NTRK1 fusion	MDM4(3) - NTRK1(5)	Fusion	NEGATIVE
MPRIP-NTRK1 fusion	MPRIP(21) - NTRK1(12)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
NFASC-NTRK1 fusion	NFASC(20) - NTRK1(10)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(16) - DYNC2H1(85)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(17) - DYNC2H1(85)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PEAR1-NTRK1 fusion	PEAR1(2) - NTRK1(9)	Fusion	NEGATIVE
PEAR1-NTRK1 fusion	PEAR1(15) - NTRK1(10)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(21) - NTRK1(9)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(16) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(11)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(13)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(12) - NTRK1(13)	Fusion	NEGATIVE
RABGAP1L-NTRK1 fusion	RABGAP1L(14) - NTRK1(16)	Fusion	NEGATIVE
RFWD2-NTRK1 fusion	RFWD2(2) - NTRK1(3)	Fusion	NEGATIVE
RNF213-NTRK1 fusion	RNF213(15) - NTRK1(12)	Fusion	NEGATIVE
SCYL3-NTRK1 fusion	SCYL3(11) - NTRK1(12)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(2) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(12)	Fusion	NEGATIVE
SSBP2-NTRK1 fusion	SSBP2(12) - NTRK1(12)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
TP53-NTRK1 fusion	TP53(8) - NTRK1(9)	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(9)	Fusion	NEGATIVE
TPM3-NTRK1 fusion	TPM3(7) - 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(6) - NTRK1(11)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(9)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(10)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(12)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(13)	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
TRIM63-NTRK1 fusion	TRIM63(8) - NTRK1(9)	Fusion	NEGATIVE
VANGL2-NTRK1 fusion	VANGL2(1) - NTRK1(12)	Fusion	NEGATIVE
AFAP1-NTRK2 fusion	AFAP1(14) - NTRK2(12)	Fusion	NEGATIVE
AGBL4-NTRK2 fusion	AGBL4(6) - NTRK2(16)	Fusion	NEGATIVE
BCR-NTRK2 fusion	BCR(1) - NTRK2(17)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DAB2IP-NTRK2 fusion	DAB2IP(1) - NTRK2(17)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(4) - NTRK2(16)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(15)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(16)	Fusion	NEGATIVE
GKAP1-NTRK2 fusion	GKAP1(9) - NTRK2(13)	Fusion	NEGATIVE
GNAQ-NTRK2 fusion	GNAQ(2) - NTRK2(16)	Fusion	NEGATIVE
KCTD8-NTRK2 fusion	KCTD8(1) - NTRK2(13)	Fusion	NEGATIVE
NACC2-NTRK2 fusion	NACC2(4) - NTRK2(13)	Fusion	NEGATIVE
NAV1-NTRK2 fusion	NAV1(15) - NTRK2(11)	Fusion	NEGATIVE
NOS1AP-NTRK2 fusion	NOS1AP(9) - NTRK2(11)	Fusion	NEGATIVE
PAN3-NTRK2 fusion	PAN3(1) - NTRK2(17)	Fusion	NEGATIVE
QKI-NTRK2 fusion	QKI(6) - NTRK2(16)	Fusion	NEGATIVE
SLMAP-NTRK2 fusion	SLMAP(14) - NTRK2(16)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(13)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(17)	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
STRN3-NTRK2 fusion	STRN3(7) - NTRK2(16)	Fusion	NEGATIVE
TBC1D2-NTRK2 fusion	TBC1D2(6) - NTRK2(12)	Fusion	NEGATIVE
TLE4-NTRK2 fusion	TLE4(7) - NTRK2(15)	Fusion	NEGATIVE
TP63-NTRK2 fusion	TP63(8) - NTRK2(12)	Fusion	NEGATIVE
TRAF2-NTRK2 fusion	TRAF2(9) - NTRK2(15)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(15)	Fusion	NEGATIVE
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(16)	Fusion	NEGATIVE
VCAN-NTRK2 fusion	VCAN(6) - NTRK2(10)	Fusion	NEGATIVE
VCL-NTRK2 fusion	VCL(16) - NTRK2(12)	Fusion	NEGATIVE
WNK2-NTRK2 fusion	WNK2(24) - NTRK2(16)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(14) - NTRK3(14)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(16) - NTRK3(15)	Fusion	NEGATIVE
BTBD1-NTRK3 fusion	BTBD1(4) - NTRK3(14)	Fusion	NEGATIVE
COX5A-NTRK3 fusion	COX5A(1) - NTRK3(15)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(2) - NTRK3(14)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(6) - NTRK3(14)	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(15)	Fusion	NEGATIVE
ETV6-NTRK3 fusion	ETV6(5) - NTRK3(14)	Fusion	NEGATIVE
FAT1-NTRK3 fusion	FAT1(2) - NTRK3(7)	Fusion	NEGATIVE
FBXL20-NTRK3 fusion	FBXL20(1) - NTRK3(6)	Fusion	NEGATIVE
HNRNPA2B1-NTRK3 fusion	HNRNPA2B1(7) - NTRK3(14)	Fusion	NEGATIVE
KANK1-NTRK3 fusion	KANK1(3) - NTRK3(14)	Fusion	NEGATIVE
LYN-NTRK3 fusion	LYN(8) - NTRK3(14)	Fusion	NEGATIVE
MYH9-NTRK3 fusion	MYH9(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(13)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(11)	Fusion	NEGATIVE
NTRK3-HOMER1 fusion	NTRK3(17) - HOMER1(2)	Fusion	NEGATIVE
NTRK3-HOMER2 fusion	NTRK3(17) - HOMER2(2)	Fusion	NEGATIVE
NTRK3-LOXL2 fusion	NTRK3(3) - LOXL2(5)	Fusion	NEGATIVE
RBPMS-NTRK3 fusion	RBPMS(5) - 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
TPM4-NTRK3 fusion	TPM4(7) - NTRK3(12)	Fusion	NEGATIVE
UBE2R2-NTRK3 fusion	UBE2R2(3) - NTRK3(4)	Fusion	NEGATIVE
ZNF710-NTRK3 fusion	ZNF710(1) - NTRK3(14)	Fusion	NEGATIVE
AFF1-NUTM1 fusion	AFF1(17) - NUTM1(3)	Fusion	NEGATIVE
BCORL1-NUTM1 fusion	BCORL1(1) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CIC-NUTM1 fusion	CIC(17) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(2)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(20) - NUTM1(5)	Fusion	NEGATIVE
MXD1-NUTM1 fusion	MXD1(5) - NUTM1(2)	Fusion	NEGATIVE
WHSC1L1-NUTM1 fusion	WHSC1L1(7) - NUTM1(2)	Fusion	NEGATIVE
ZNF618-NUTM1 fusion	ZNF618(12) - NUTM1(2)	Fusion	NEGATIVE
ABI3BP-RET fusion	ABI3BP(34) - RET(12)	Fusion	NEGATIVE
ACBD5-RET fusion	ACBD5(11) - RET(12)	Fusion	NEGATIVE
ADD3-RET fusion	ADD3(1) - RET(12)	Fusion	NEGATIVE
AFAP1-RET fusion	AFAP1(3) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(6) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(2) - RET(12)	Fusion	NEGATIVE
AKAP13-RET fusion	AKAP13(35) - RET(12)	Fusion	NEGATIVE
ANK3-RET fusion	ANK3(1) - RET(12)	Fusion	NEGATIVE
BCR-RET fusion	BCR(4) - RET(12)	Fusion	NEGATIVE
CCDC186-RET fusion	CCDC186(7) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CCDC6-RET fusion	CCDC6(1) - RET(10)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(12)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CUX1-RET fusion	CUX1(10) - RET(12)	Fusion	NEGATIVE
DLG5-RET fusion	DLG5(13) - RET(12)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(11)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(12)	Fusion	NEGATIVE
EPHA5-RET fusion	EPHA5(2) - 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
FGFR1OP-RET fusion	FGFR1OP(11) - RET(11)	Fusion	NEGATIVE
FGFR1OP-RET fusion	FGFR1OP(12) - RET(12)	Fusion	NEGATIVE
FKBP15-RET fusion	FKBP15(25) - RET(12)	Fusion	NEGATIVE
FRMD4A-RET fusion	FRMD4A(12) - RET(12)	Fusion	NEGATIVE
GOLGA5-RET fusion	GOLGA5(7) - RET(12)	Fusion	NEGATIVE
GPHN-RET fusion	GPHN(9) - RET(12)	Fusion	NEGATIVE
HOOK3-RET fusion	HOOK3(11) - RET(12)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(10)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1468-RET fusion	KIAA1468(10) - RET(12)	Fusion	NEGATIVE
KIF13A-RET fusion	KIF13A(18) - 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(8)	Fusion	NEGATIVE
KIF5B-RET fusion	KIF5B(23) - RET(12)	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
KIF5B-RET fusion	KIF5B(15) - RET(12)	Fusion	NEGATIVE
KTN1-RET fusion	KTN1(29) - RET(12)	Fusion	NEGATIVE
MPRIP-RET fusion	MPRIP(19) - RET(12)	Fusion	NEGATIVE
MYH10-RET fusion	MYH10(33) - RET(12)	Fusion	NEGATIVE
MYH13-RET fusion	MYH13(35) - RET(12)	Fusion	NEGATIVE
MYO5A-RET fusion	MYO5A(23) - RET(11)	Fusion	NEGATIVE
MYO5C-RET fusion	MYO5C(25) - 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
OLFM4-RET fusion	OLFM4(4) - RET(10)	Fusion	NEGATIVE
PCM1-RET fusion	PCM1(29) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDCD10-RET fusion	PDCD10(4) - RET(11)	Fusion	NEGATIVE
PICALM-RET fusion	PICALM(19) - RET(12)	Fusion	NEGATIVE
PPFIBP2-RET fusion	PPFIBP2(9) - RET(12)	Fusion	NEGATIVE
PRKAR1A-RET fusion	PRKAR1A(7) - RET(12)	Fusion	NEGATIVE
PRKG1-RET fusion	PRKG1(3) - RET(12)	Fusion	NEGATIVE
RASAL2-RET fusion	RASAL2(15) - RET(12)	Fusion	NEGATIVE
RASSF4-RET fusion	RASSF4(3) - RET(12)	Fusion	NEGATIVE
RRBP1-RET fusion	RRBP1(9) - RET(12)	Fusion	NEGATIVE
RUFY1-RET fusion	RUFY1(17) - RET(12)	Fusion	NEGATIVE
RUFY2-RET fusion	RUFY2(9) - RET(12)	Fusion	NEGATIVE
SATB1-RET fusion	SATB1(7) - RET(12)	Fusion	NEGATIVE
SNRNP70-RET fusion	SNRNP70(7) - RET(12)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(11)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(12)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(5) - RET(11)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(6) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(11)	Fusion	NEGATIVE
TFG-RET fusion	TFG(4) - RET(11)	Fusion	NEGATIVE
TNIP1-RET fusion	TNIP1(11) - RET(12)	Fusion	NEGATIVE
TNIP2-RET fusion	TNIP2(5) - RET(12)	Fusion	NEGATIVE
TRIM24-RET fusion	TRIM24(9) - RET(12)	Fusion	NEGATIVE
TRIM27-RET fusion	TRIM27(3) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
WAC-RET fusion	WAC(3) - RET(12)	Fusion	NEGATIVE
CCDC30-ROS1 fusion	CCDC30(10) - ROS1(36)	Fusion	NEGATIVE
CCDC6-ROS1 fusion	CCDC6(5) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(7) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(33)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(32)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(4) - ROS1(33)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(12) - ROS1(35)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(8) - ROS1(36)	Fusion	NEGATIVE
CLIP1-ROS1 fusion	CLIP1(20) - ROS1(36)	Fusion	NEGATIVE
CLTC-ROS1 fusion	CLTC(31) - ROS1(35)	Fusion	NEGATIVE
ERC1-ROS1 fusion	ERC1(11) - ROS1(36)	Fusion	NEGATIVE
EYS-ROS1 fusion	EYS(12) - ROS1(9)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(35)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(34)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(33)	Fusion	NEGATIVE
GOLGB1-ROS1 fusion	GOLGB1(9) - ROS1(35)	Fusion	NEGATIVE
GOPC-ROS1 fusion	GOPC(8) - ROS1(35)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
GOPC-ROS1 fusion	GOPC(4) - ROS1(36)	Fusion	NEGATIVE
HLA-A-ROS1 fusion	HLA-A(7) - ROS1(34)	Fusion	NEGATIVE
LIMA1-ROS1 fusion	LIMA1(10) - ROS1(36)	Fusion	NEGATIVE
LRIG3-ROS1 fusion	LRIG3(16) - ROS1(35)	Fusion	NEGATIVE
MSN-ROS1 fusion	MSN(9) - ROS1(34)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(32) - ROS1(35)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(23) - ROS1(35)	Fusion	NEGATIVE
MYO5C-ROS1 fusion	MYO5C(30) - ROS1(35)	Fusion	NEGATIVE
NCOR2-ROS1 fusion	NCOR2(7) - ROS1(36)	Fusion	NEGATIVE
NFKB2-ROS1 fusion	NFKB2(14) - ROS1(36)	Fusion	NEGATIVE
PPFIBP1-ROS1 fusion	PPFIBP1(9) - ROS1(35)	Fusion	NEGATIVE
PWWP2A-ROS1 fusion	PWWP2A(1) - ROS1(36)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(5) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(32)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(32)	Fusion	NEGATIVE
SHTN1-ROS1 fusion	SHTN1(11) - ROS1(36)	Fusion	NEGATIVE
SLC16A10-ROS1 fusion	SLC16A10(3) - ROS1(3)	Fusion	NEGATIVE
SLC22A16-ROS1 fusion	SLC22A16(7) - ROS1(16)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(35)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(32)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(33)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(32)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - 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
TMEM106B-ROS1 fusion	TMEM106B(3) - ROS1(35)	Fusion	NEGATIVE
TPD52L1-ROS1 fusion	TPD52L1(3) - ROS1(33)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(7) - ROS1(35)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(3) - ROS1(36)	Fusion	NEGATIVE
TSPAN3-ROS1 fusion	TSPAN3(5) - ROS1(31)	Fusion	NEGATIVE
YWHAE-ROS1 fusion	YWHAE(4) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(3) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(2) - ROS1(36)	Fusion	NEGATIVE
EIF3E-RSPO2 fusion	EIF3E(1) - RSPO2(2)	Fusion	NEGATIVE
GRHL2-RSPO2 fusion	GRHL2(8) - 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

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
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

QC Metrics

NTC QC - DNA

Metric Name	Value	Reference Range	QC Status
Average Base Coverage Depth	3	<=15	Passed
Hotspot Calls	0	<=0	Passed

NTC QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	0	<=2848	Passed
Total Fusion Calls	0	<=0	Passed

Sample QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	94944	>=6000	Passed
RNA Expression Ctrls Detected	7	>=5	Passed

Sample QC - DNA

Metric Name	Value	Reference Range	QC Status
MAPD	0.18	[0-0.5]	Passed
Mapped Reads	862563	>=100000	Passed
Mean AQ20 Read Length (bp)	88	>=64	Passed

Templating Control QC - CF-1

Metric Name	Value	Reference Range	QC Status
Average Reads Per Lane	26283	>=2383	Passed
Mean AQ20 Read Length (bp)	108	>=82	Passed

PositiveControl01 QC - DNA

Metric Name	Value	Reference Range	QC Status
MAPD	0.25	[0-0.5]	Passed
Mapped Reads	1250723	>=100000	Passed
Mean AQ20 Read Length (bp)	93	>=76	Passed
COSM1072523	POSITIVE	POSITIVE	Passed
COSM1072523_AF	0.1172	>=0.0511	Passed
COSM12376	POSITIVE	POSITIVE	Passed
COSM12376_AF	0.1278	>=0.02	Passed
COSM12428	POSITIVE	POSITIVE	Passed
COSM12428_AF	0.1465	>=0.0529	Passed
COSM12558	POSITIVE	POSITIVE	Passed
COSM12558_AF	0.1273	>=0.02	Passed
COSM133167	POSITIVE	POSITIVE	Passed
COSM133167_AF	0.1299	>=0.025	Passed
COSM144252	POSITIVE	POSITIVE	Passed
COSM144252_AF	0.1483	>=0.02	Passed
COSM256046	POSITIVE	POSITIVE	Passed
COSM256046_AF	0.1281	>=0.0489	Passed
COSM28055	POSITIVE	POSITIVE	Passed
COSM28055_AF	0.1201	>=0.0423	Passed
COSM28747	POSITIVE	POSITIVE	Passed
COSM28747_AF	0.0973	>=0.0448	Passed
COSM33765	POSITIVE	POSITIVE	Passed

Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** | Date of Birth: 30 MAR 1975 | Date: 13 JAN 2026

Metric Name	Value	Reference Range	QC Status
COSM33765_AF	0.1132	>=0.025	Passed
COSM41590	POSITIVE	POSITIVE	Passed
COSM41590_AF	0.1215	>=0.032	Passed
COSM476	POSITIVE	POSITIVE	Passed
COSM476_AF	0.1186	>=0.0394	Passed
COSM48358	POSITIVE	POSITIVE	Passed
COSM48358_AF	0.108	>=0.0539	Passed
COSM516	POSITIVE	POSITIVE	Passed
COSM516_AF	0.1053	>=0.025	Passed
COSM5219	POSITIVE	POSITIVE	Passed
COSM5219_AF	0.1412	>=0.0555	Passed
COSM6223	POSITIVE	POSITIVE	Passed
COSM6223_AF	0.1388	>=0.0348	Passed
COSM6224	POSITIVE	POSITIVE	Passed
COSM6224_AF	0.147	>=0.0471	Passed
COSM6240	POSITIVE	POSITIVE	Passed
COSM6240_AF	0.139	>=0.025	Passed
COSM683	POSITIVE	POSITIVE	Passed
COSM683_AF	0.1128	>=0.0297	Passed
COSM775	POSITIVE	POSITIVE	Passed
COSM775_AF	0.1155	>=0.0261	Passed
COSM962	POSITIVE	POSITIVE	Passed
COSM962_AF	0.1342	>=0.0309	Passed

Metric Name	Value	Reference Range	QC Status
OMINDEL1177	POSITIVE	POSITIVE	Passed
OMINDEL1177_AF	0.1305	>=0.0334	Passed
OMINDEL698	POSITIVE	POSITIVE	Passed
OMINDEL698_AF	0.1345	>=0.0456	Passed
ERBB2 CNV	POSITIVE	POSITIVE	Passed
ERBB2 Copy Number	9.6	>=5	Passed
MET CNV	POSITIVE	POSITIVE	Passed
MET Copy Number	9.9	>=7	Passed

PositiveControl01 QC - RNA

Metric Name	Value	Reference Range	QC Status
Mapped Fusion Reads	341314	>=50475	Passed
RNA Expression Ctrls Detected	7	>=7	Passed
ALK Targeted Isoform	POSITIVE	POSITIVE	Passed
AR Targeted Isoform	POSITIVE	POSITIVE	Passed
BRAF Targeted Isoform	POSITIVE	POSITIVE	Passed
EGFR Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR1 Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR2 Expression Imbalance	POSITIVE	POSITIVE	Passed
FGFR2 Targeted Isoform	POSITIVE	POSITIVE	Passed
FGFR3 Expression Imbalance	POSITIVE	POSITIVE	Passed
FGFR3 Targeted Isoform	POSITIVE	POSITIVE	Passed
MET Targeted Isoform	POSITIVE	POSITIVE	Passed
NRG1 Targeted Isoform	POSITIVE	POSITIVE	Passed

Metric Name	Value	Reference Range	QC Status
NTRK1 Expression Imbalance	POSITIVE	POSITIVE	Passed
NTRK1 Targeted Isoform	POSITIVE	POSITIVE	Passed
NTRK2 Targeted Isoform	POSITIVE	POSITIVE	Passed
NTRK3 Targeted Isoform	POSITIVE	POSITIVE	Passed
NUTM1 Targeted Isoform	POSITIVE	POSITIVE	Passed
RET Targeted Isoform	POSITIVE	POSITIVE	Passed
ROS1 Targeted Isoform	POSITIVE	POSITIVE	Passed
RSPO2 Targeted Isoform	POSITIVE	POSITIVE	Passed
RSPO3 Targeted Isoform	POSITIVE	POSITIVE	Passed

Sequencing Run Details

Assay Name: **Oncomine™ Dx Express Test - FFPE - RS**

Panel: **Oncomine™ Dx Express Test - FFPE Panel - RS v5.2.0**

Started On: **v5.2.0 15 FEB 2022 18:38**

Completed On: **06 NOV 2025 18:23**

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

Kit Barcodes

Sample Name	Nucleic Acid Type	Kit Type	Kit Information
FFPE01	DNA	Extraction	100101850
FFPE01	RNA	Extraction	100101851
FFPE02	DNA	Extraction	100101850
FFPE02	RNA	Extraction	100101851
FFPE03	DNA	Extraction	100101850
FFPE03	RNA	Extraction	100101851
FFPE04	DNA	Extraction	100101850
FFPE04	RNA	Extraction	100101851
FFPE05	DNA	Extraction	100101850
FFPE05	RNA	Extraction	100101851
FFPE06	DNA	Extraction	100101850
FFPE06	RNA	Extraction	100101851
FFPE07	DNA	Extraction	100101850
FFPE07	RNA	Extraction	100101851
FFPE08	DNA	Extraction	100101850
FFPE08	RNA	Extraction	100101851
FFPE09	DNA	Extraction	100101850

Clinical Laboratory Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** | Date of Birth: 30 MAR 1975 | Date: 13 JAN 2026

Sample Name	Nucleic Acid Type	Kit Type	Kit Information
FFPE09	RNA	Extraction	100101851
FFPE10	DNA	Extraction	100101850
FFPE10	RNA	Extraction	100101851
FFPE11	DNA	Extraction	100101850
FFPE11	RNA	Extraction	100101851
FFPE12	DNA	Extraction	100101850
FFPE12	RNA	Extraction	100101851
PositiveControl01	DNA	Control	A50834(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
PositiveControl01	RNA	Control	A50835(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
NTC	DNA	Control	A50836(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)
NTC	RNA	Control	A50836(Part #) - 30 JUN 2025(Expiration Date) - 2882442(Lot #)

Sample		**Patient**	
Sample Name:	**FFPE01**	Patient Id:	**PID01**
Cancer Type:	**Non-Small Cell Lung Cancer**	Gender:	**Male**
Cancer Stage:	**Stage III**	Date of Birth:	**30 MAR 1975**
Sample Type:	**FFPE**	Collection Date:	**22 FEB 2021**
%Cellularity:	**100**	Ordering Physician:	**Dr.Q**
%Necrosis:		Created On:	**06 NOV 2025 14:24**

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
EGFR	EGFR exon 20 insertion	p.?	c.2284-6_2284-5insTCCAGGAAGCCT	POSITIVE	COSM26720	ins
KRAS	KRAS G12C	p.G12C	c.34_36delGGTinsTGC	POSITIVE	COSM513	mnp

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
SQSTM1-NTRK3 fusion	SQSTM1(5) - NTRK3(14)	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
AKT1	AKT1 E17K	p.E17K	c.49G>A	POSITIVE	COSM33765	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
AR-V7	AR(3) - AR(4)	RNAExonVariant	POSITIVE

ⓘ 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 therapies 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)
NSCLC	ERBB2	ERBB2/HER2 activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions)	HYRNUO® (sevabertinib)

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 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 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 >70% necrotic tissue may yield unreliable sequencing results for the detection of *EGFR* exon 20 insertion and *ERBB2* 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).

30. For *MET* Exon 14 skipping results for cancer types other than non-small cell lung cancer, confirmatory testing using a clinically validated assay is recommended.

31. For NSCLC, the Oncomine™ Dx Express Test assay definition file includes prevalent but not all rare or newly identified *EGFR* exon 20 insertions, *ERBB2/HER2* exon 20 insertions, and *ERBB2/HER2* SNVs in exons 18-21 within the tyrosine kinase domain. The Oncomine™ Dx Express Test may miss rare or newly identified:

- *EGFR* exon 20 insertions carried by a subset of patients who may derive benefit from sunvozertinib
- *ERBB2/HER2* activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions) carried by a subset of patients who may derive benefit from sevabertinib

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 C77F	p.C77F	COSM5027916	NEGATIVE	SNV
AKT1 L52R	p.L52R	COSM93893	NEGATIVE	SNV
AKT1 P51L	p.P51L	COSM4468165	NEGATIVE	SNV
AKT1 P51S	p.P51S	COSM5991558	NEGATIVE	SNV

Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** Date of Birth: 30 MAR 1975 Date: 13 JAN 2026

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AKT1 P70_C77dup	p.P70_C77dup	COSM6568358	NEGATIVE	ins
AKT1 Q79K	p.Q79K	COSM159008	NEGATIVE	SNV
AKT1 R76_C77insWPNTFIIR	p.R76_C77insWPNTFIIR	COSM5045024	NEGATIVE	ins
AKT2 D324G	p.D324G	COSM474741	NEGATIVE	SNV
AKT2 D324H	p.D324H	COSM474742	NEGATIVE	SNV
AKT2 E17K	p.E17K	COSM159009	NEGATIVE	SNV
AKT3 E17K	p.E17K	COSM224779	NEGATIVE	SNV
AKT3 L51R	p.L51R	COSM309035	NEGATIVE	SNV
AKT3 Q78K	p.Q78K	COSM242892	NEGATIVE	SNV
ALK A1200V	p.A1200V	COSM317003	NEGATIVE	SNV
ALK A1280V	p.A1280V	COSM6914238	NEGATIVE	SNV
ALK C1156F	p.C1156F	BT144	NEGATIVE	SNV
ALK C1156T	p.C1156T	OM3350	NEGATIVE	mnp
ALK C1156Y	p.C1156Y	COSM99136	NEGATIVE	SNV
ALK D1203A	p.D1203A	OM3313	NEGATIVE	SNV
ALK D1203N	p.D1203N	COSM6191344	NEGATIVE	SNV
ALK E1129V	p.E1129V	BT117	NEGATIVE	SNV
ALK E1154Q	p.E1154Q	OM3324	NEGATIVE	SNV
ALK E1210K	p.E1210K	OM3161	NEGATIVE	SNV
ALK F1174C	p.F1174C	COSM28059	NEGATIVE	SNV
ALK F1174I	p.F1174I	COSM28491	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28055	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28061	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK F1174L	p.F1174L	COSM28057	NEGATIVE	SNV
ALK F1174S	p.F1174S	COSM53063	NEGATIVE	SNV
ALK F1174V	p.F1174V	COSM28054	NEGATIVE	SNV
ALK F1245C	p.F1245C	COSM28500	NEGATIVE	SNV
ALK F1245I	p.F1245I	COSM28492	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28062	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28493	NEGATIVE	SNV
ALK F1245V	p.F1245V	COSM28499	NEGATIVE	SNV
ALK G1123D	p.G1123D	OM3262	NEGATIVE	SNV
ALK G1123S	p.G1123S	COSM5045108	NEGATIVE	SNV
ALK G1128A	p.G1128A	COSM98475	NEGATIVE	SNV
ALK G1202R	p.G1202R	COSM144250	NEGATIVE	SNV
ALK G1202del	p.G1202del	OMINDEL698	NEGATIVE	del
ALK G1269A	p.G1269A	COSM1169707	NEGATIVE	SNV
ALK I1171H	p.I1171H	OM3163	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM1169448	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM28498	NEGATIVE	SNV
ALK I1171S	p.I1171S	COSM4440758	NEGATIVE	SNV
ALK I1171T	p.I1171T	COSM4381100	NEGATIVE	SNV
ALK I1268V	p.I1268V	BT149	NEGATIVE	SNV
ALK L1152P	p.L1152P	COSM1407659	NEGATIVE	SNV
ALK L1152R	p.L1152R	COSM97185	NEGATIVE	SNV
ALK L1152V	p.L1152V	OM3513	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK L1196M	p.L1196M	COSM99137	NEGATIVE	SNV
ALK L1196Q	p.L1196Q	COSM1169447	NEGATIVE	SNV
ALK L1198F	p.L1198F	COSM97028	NEGATIVE	SNV
ALK L1198P	p.L1198P	OM3162	NEGATIVE	SNV
ALK L1198V	p.L1198V	OM3314	NEGATIVE	SNV
ALK R1192P	p.R1192P	OM3259	NEGATIVE	SNV
ALK R1192Q	p.R1192Q	OM3372	NEGATIVE	SNV
ALK R1209Q	p.R1209Q	COSM327056	NEGATIVE	SNV
ALK R1231Q	p.R1231Q	COSM5967491	NEGATIVE	SNV
ALK R1231W	p.R1231W	BT148	NEGATIVE	SNV
ALK R1275L	p.R1275L	COSM28060	NEGATIVE	SNV
ALK R1275Q	p.R1275Q	COSM28056	NEGATIVE	SNV
ALK S1157F	p.S1157F	BT145	NEGATIVE	SNV
ALK S1206C	p.S1206C	BT147	NEGATIVE	SNV
ALK S1206F	p.S1206F	COSM6909200	NEGATIVE	SNV
ALK S1206Y	p.S1206Y	COSM144251	NEGATIVE	SNV
ALK T1151K	p.T1151K	OM3261	NEGATIVE	SNV
ALK T1151M	p.T1151M	COSM98478	NEGATIVE	SNV
ALK T1151R	p.T1151R	OM3260	NEGATIVE	SNV
ALK T1151dup	p.T1151dup	COSM144252	NEGATIVE	ins
ALK V1180L	p.V1180L	COSM4381101	NEGATIVE	SNV
ALK Y1278S	p.Y1278S	COSM28058	NEGATIVE	SNV
AR D880E	p.D880E	COSM5945750	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AR D891H	p.D891H	COSM5945767	NEGATIVE	SNV
AR E894K	p.E894K	COSM5945748	NEGATIVE	SNV
AR E898G	p.E898G	OM3322	NEGATIVE	SNV
AR F814V	p.F814V	COSM150793	NEGATIVE	SNV
AR F877L	p.F877L	COSM5945752	NEGATIVE	SNV
AR F877L	p.F877L	COSM5946164	NEGATIVE	SNV
AR H875Q	p.H875Q	COSM5945766	NEGATIVE	SNV
AR H875Y	p.H875Y	COSM238555	NEGATIVE	SNV
AR L702H	p.L702H	COSM238553	NEGATIVE	SNV
AR L882I	p.L882I	COSM5945751	NEGATIVE	SNV
AR M896T	p.M896T	OM3321	NEGATIVE	SNV
AR M896V	p.M896V	COSM5945749	NEGATIVE	SNV
AR S889G	p.S889G	COSM5945753	NEGATIVE	SNV
AR T878A	p.T878A	COSM236693	NEGATIVE	SNV
AR T878S	p.T878S	COSM5570418	NEGATIVE	SNV
AR V716M	p.V716M	COSM3094417	NEGATIVE	SNV
AR V731M	p.V731M	COSM1468984	NEGATIVE	SNV
AR W742C	p.W742C	COSM5572266	NEGATIVE	SNV
AR W742C	p.W742C	COSM5944171	NEGATIVE	SNV
AR W742L	p.W742L	COSM5944172	NEGATIVE	SNV
ARAF S214A	p.S214A	COSM1742787	NEGATIVE	SNV
ARAF S214C	p.S214C	COSM5044705	NEGATIVE	SNV
ARAF S214F	p.S214F	COSM612884	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ARAF S214T	p.S214T	COSM1558277	NEGATIVE	SNV
ARAF S214Y	p.S214Y	COSM5416443	NEGATIVE	SNV
BRAF A598T	p.A598T	COSM28505	NEGATIVE	SNV
BRAF A598V	p.A598V	COSM21549	NEGATIVE	SNV
BRAF D594A	p.D594A	COSM1583010	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM1121	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM253330	NEGATIVE	SNV
BRAF D594G	p.D594G	COSM467	NEGATIVE	SNV
BRAF D594H	p.D594H	COSM144576	NEGATIVE	SNV
BRAF D594N	p.D594N	COSM27639	NEGATIVE	SNV
BRAF D594V	p.D594V	COSM466	NEGATIVE	SNV
BRAF D594Y	p.D594Y	COSM6928839	NEGATIVE	SNV
BRAF D594_T599dup	p.D594_T599dup	COSM26504	NEGATIVE	ins
BRAF E586K	p.E586K	COSM463	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM468	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM53198	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM21612	NEGATIVE	SNV
BRAF F595S	p.F595S	COSM1123	NEGATIVE	SNV
BRAF G464E	p.G464E	COSM449	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1111	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1448615	NEGATIVE	SNV
BRAF G464V	p.G464V	COSM450	NEGATIVE	SNV
BRAF G466A	p.G466A	COSM452	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF G466E	p.G466E	COSM453	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM1112	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM253328	NEGATIVE	SNV
BRAF G466V	p.G466V	COSM451	NEGATIVE	SNV
BRAF G469A	p.G469A	COSM460	NEGATIVE	SNV
BRAF G469E	p.G469E	COSM461	NEGATIVE	SNV
BRAF G469L	p.G469L	COSM6056848	NEGATIVE	mnp
BRAF G469R	p.G469R	COSM455	NEGATIVE	SNV
BRAF G469R	p.G469R	COSM457	NEGATIVE	SNV
BRAF G469S	p.G469S	COSM1113	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM27912	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM458	NEGATIVE	mnp
BRAF G469V	p.G469V	COSM459	NEGATIVE	SNV
BRAF G596C	p.G596C	COSM6936824	NEGATIVE	SNV
BRAF G596D	p.G596D	COSM26506	NEGATIVE	SNV
BRAF G596R	p.G596R	COSM469	NEGATIVE	SNV
BRAF G596V	p.G596V	COSM9145962	NEGATIVE	SNV
BRAF I463S	p.I463S	COSM448	NEGATIVE	SNV
BRAF K601E	p.K601E	COSM478	NEGATIVE	SNV
BRAF K601I	p.K601I	COSM26491	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM1132	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM6265	NEGATIVE	SNV
BRAF K601R	p.K601R	COSM1568586	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF K601T	p.K601T	COSM3878760	NEGATIVE	SNV
BRAF L597P	p.L597P	COSM1448593	NEGATIVE	SNV
BRAF L597Q	p.L597Q	COSM1125	NEGATIVE	SNV
BRAF L597R	p.L597R	COSM471	NEGATIVE	SNV
BRAF L597S	p.L597S	COSM1126	NEGATIVE	mnp
BRAF L597V	p.L597V	COSM470	NEGATIVE	SNV
BRAF N581I	p.N581I	COSM26460	NEGATIVE	SNV
BRAF N581S	p.N581S	COSM462	NEGATIVE	SNV
BRAF N581T	p.N581T	COSM1699452	NEGATIVE	SNV
BRAF R462I	p.R462I	COSM447	NEGATIVE	SNV
BRAF T599I	p.T599I	COSM472	NEGATIVE	SNV
BRAF T599R	p.T599R	COSM4172020	NEGATIVE	SNV
BRAF T599_V600insEAT	p.T599_V600insEAT	COSM4166148	NEGATIVE	ins
BRAF T599_V600insETT	p.T599_V600insETT	COSM4172018	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM144982	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM30730	NEGATIVE	ins
BRAF V600A	p.V600A	COSM18443	NEGATIVE	SNV
BRAF V600D	p.V600D	COSM308550	NEGATIVE	mnp
BRAF V600D	p.V600D	COSM477	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM475	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM476	NEGATIVE	SNV
BRAF V600G	p.V600G	COSM6137	NEGATIVE	SNV
BRAF V600K	p.V600K	COSM473	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF V600L	p.V600L	COSM219798	NEGATIVE	SNV
BRAF V600L	p.V600L	COSM33808	NEGATIVE	SNV
BRAF V600M	p.V600M	COSM1130	NEGATIVE	SNV
BRAF V600Q	p.V600Q	COSM249889	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM1583011	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM474	NEGATIVE	mnp
BRAF V600_K601delinsE	p.V600_K601delinsE	COSM1133	NEGATIVE	del
BRAF Y472C	p.Y472C	COSM1133046	NEGATIVE	SNV
BRAF [G593=;D594N]	p.[G593=;D594N]	COSM211600	NEGATIVE	mnp
BRAF [T599=;V600R]	p.[T599=;V600R]	COSM1127	NEGATIVE	mnp
BRAF [T599I;V600K]	p.[T599I;V600K]	COSM1735763	NEGATIVE	mnp
BRAF [T599R;V600G]	p.[T599R;V600G]	OM3547	NEGATIVE	mnp
CDK4 K22M	p.K22M	COSM3463915	NEGATIVE	SNV
CDK4 K22Q	p.K22Q	OM3153	NEGATIVE	SNV
CDK4 K22R	p.K22R	COSM232013	NEGATIVE	SNV
CDK4 R24C	p.R24C	COSM1677139	NEGATIVE	SNV
CDK4 R24H	p.R24H	COSM1989836	NEGATIVE	SNV
CDK4 R24L	p.R24L	COSM363684	NEGATIVE	SNV
CDK4 R24S	p.R24S	COSM3463914	NEGATIVE	SNV
CHEK2 K373E	p.K373E	COSM42871	NEGATIVE	SNV
CHEK2 M381V	p.M381V	COSM1559601	NEGATIVE	SNV
CHEK2 R523H	p.R523H	COSM1559596	NEGATIVE	SNV
CHEK2 R523P	p.R523P	COSM1559595	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CHEK2 [S372=;K373E]	p.[S372=;K373E]	COSM1294063	NEGATIVE	mnp
CTNNB1 D32A	p.D32A	COSM5690	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM22939	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM5766	NEGATIVE	SNV
CTNNB1 D32G	p.D32G	COSM5681	NEGATIVE	SNV
CTNNB1 D32H	p.D32H	COSM5668	NEGATIVE	SNV
CTNNB1 D32N	p.D32N	COSM5672	NEGATIVE	SNV
CTNNB1 D32V	p.D32V	COSM5691	NEGATIVE	SNV
CTNNB1 D32Y	p.D32Y	COSM5661	NEGATIVE	SNV
CTNNB1 G34A	p.G34A	COSM5988492	NEGATIVE	SNV
CTNNB1 G34E	p.G34E	COSM5671	NEGATIVE	SNV
CTNNB1 G34I	p.G34I	COSM1666841	NEGATIVE	mnp
CTNNB1 G34R	p.G34R	COSM5684	NEGATIVE	SNV
CTNNB1 G34R	p.G34R	COSM5686	NEGATIVE	SNV
CTNNB1 G34V	p.G34V	COSM5670	NEGATIVE	SNV
CTNNB1 H36P	p.H36P	COSM5678	NEGATIVE	SNV
CTNNB1 H36R	p.H36R	COSM27378	NEGATIVE	SNV
CTNNB1 H36Y	p.H36Y	COSM5703	NEGATIVE	SNV
CTNNB1 I35K	p.I35K	COSM28727	NEGATIVE	mnp
CTNNB1 I35M	p.I35M	COSM28711	NEGATIVE	SNV
CTNNB1 I35N	p.I35N	COSM5695	NEGATIVE	SNV
CTNNB1 I35S	p.I35S	COSM5674	NEGATIVE	SNV
CTNNB1 I35T	p.I35T	COSM13168	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 I35V	p.I35V	COSM1566774	NEGATIVE	SNV
CTNNB1 K335I	p.K335I	COSM17797	NEGATIVE	SNV
CTNNB1 K335Q	p.K335Q	COSM4421324	NEGATIVE	SNV
CTNNB1 K335R	p.K335R	COSM5945941	NEGATIVE	SNV
CTNNB1 K335T	p.K335T	COSM1725761	NEGATIVE	SNV
CTNNB1 L46_S47del	p.L46_S47del	COSM6129	NEGATIVE	del
CTNNB1 L46_S47del	p.L46_S47del	COSM6132	NEGATIVE	del
CTNNB1 P44A	p.P44A	COSM17661	NEGATIVE	SNV
CTNNB1 P44L	p.P44L	COSM5761	NEGATIVE	SNV
CTNNB1 P44S	p.P44S	COSM5704	NEGATIVE	SNV
CTNNB1 S33A	p.S33A	COSM5683	NEGATIVE	SNV
CTNNB1 S33C	p.S33C	COSM5677	NEGATIVE	SNV
CTNNB1 S33F	p.S33F	COSM5669	NEGATIVE	SNV
CTNNB1 S33L	p.S33L	COSM6098	NEGATIVE	mnp
CTNNB1 S33N	p.S33N	COSM6099	NEGATIVE	mnp
CTNNB1 S33P	p.S33P	COSM5682	NEGATIVE	SNV
CTNNB1 S33T	p.S33T	COSM27311	NEGATIVE	SNV
CTNNB1 S33Y	p.S33Y	COSM5673	NEGATIVE	SNV
CTNNB1 S37A	p.S37A	COSM5675	NEGATIVE	SNV
CTNNB1 S37C	p.S37C	COSM5679	NEGATIVE	SNV
CTNNB1 S37F	p.S37F	COSM5662	NEGATIVE	SNV
CTNNB1 S37P	p.S37P	COSM5687	NEGATIVE	SNV
CTNNB1 S37T	p.S37T	COSM5729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 S37Y	p.S37Y	COSM5666	NEGATIVE	SNV
CTNNB1 S45A	p.S45A	COSM5685	NEGATIVE	SNV
CTNNB1 S45C	p.S45C	COSM5689	NEGATIVE	SNV
CTNNB1 S45F	p.S45F	COSM5667	NEGATIVE	SNV
CTNNB1 S45P	p.S45P	COSM5663	NEGATIVE	SNV
CTNNB1 S45T	p.S45T	COSM5719	NEGATIVE	SNV
CTNNB1 S45Y	p.S45Y	COSM5692	NEGATIVE	SNV
CTNNB1 S45_G48del	p.S45_G48del	COSM5992	NEGATIVE	del
CTNNB1 S45_S47del	p.S45_S47del	COSM1617582	NEGATIVE	del
CTNNB1 S45_S47delinsC	p.S45_S47delinsC	COSM28708	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6128	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6131	NEGATIVE	del
CTNNB1 T41A	p.T41A	COSM5664	NEGATIVE	SNV
CTNNB1 T41I	p.T41I	COSM5676	NEGATIVE	SNV
CTNNB1 T41N	p.T41N	COSM5730	NEGATIVE	SNV
CTNNB1 T41P	p.T41P	COSM5688	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5716	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5701	NEGATIVE	SNV
CTNNB1 [S45E;L46=]	p.[S45E;L46=]	COSM86011	NEGATIVE	mnp
EGFR A289D	p.A289D	COSM21685	NEGATIVE	SNV
EGFR A289T	p.A289T	COSM21686	NEGATIVE	SNV
EGFR A289V	p.A289V	COSM21687	NEGATIVE	SNV
EGFR A750P	p.A750P	COSM6219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR C775Y	p.C775Y	COSM22954	NEGATIVE	SNV
EGFR C797G	p.C797G	COSM6493936	NEGATIVE	SNV
EGFR C797N	p.C797N	OM3543	NEGATIVE	mnp
EGFR C797S	p.C797S	COSM6493937	NEGATIVE	SNV
EGFR C797S	p.C797S	COSM5945664	NEGATIVE	SNV
EGFR E709A	p.E709A	COSM13427	NEGATIVE	SNV
EGFR E709G	p.E709G	COSM13009	NEGATIVE	SNV
EGFR E709H	p.E709H	COSM12428	NEGATIVE	mnp
EGFR E709K	p.E709K	COSM12988	NEGATIVE	SNV
EGFR E709Q	p.E709Q	COSM116882	NEGATIVE	SNV
EGFR E709V	p.E709V	COSM12371	NEGATIVE	SNV
EGFR E709_T710delinsA	p.E709_T710delinsA	COSM85796	NEGATIVE	del
EGFR E709_T710delinsD	p.E709_T710delinsD	COSM51525	NEGATIVE	del
EGFR E709_T710delinsG	p.E709_T710delinsG	COSM48981	NEGATIVE	complex
EGFR E746G	p.E746G	COSM6849765	NEGATIVE	SNV
EGFR G465E	p.G465E	BT129	NEGATIVE	SNV
EGFR G465R	p.G465R	COSM601581	NEGATIVE	SNV
EGFR G598A	p.G598A	COSM3412196	NEGATIVE	SNV
EGFR G598V	p.G598V	COSM21690	NEGATIVE	SNV
EGFR G719A	p.G719A	COSM6239	NEGATIVE	SNV
EGFR G719C	p.G719C	COSM6253	NEGATIVE	SNV
EGFR G719D	p.G719D	COSM18425	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR G719S	p.G719S	COSM6252	NEGATIVE	SNV
EGFR G721D	p.G721D	COSM22993	NEGATIVE	SNV
EGFR G721R	p.G721R	OM3325	NEGATIVE	SNV
EGFR G721S	p.G721S	COSM22992	NEGATIVE	SNV
EGFR G724S	p.G724S	COSM13979	NEGATIVE	SNV
EGFR G796A	p.G796A	COSM27568	NEGATIVE	SNV
EGFR G796C	p.G796C	OM3318	NEGATIVE	SNV
EGFR G796R	p.G796R	COSM6493935	NEGATIVE	SNV
EGFR G796S	p.G796S	COSM20891	NEGATIVE	SNV
EGFR H835L	p.H835L	COSM6227	NEGATIVE	SNV
EGFR I491M	p.I491M	BT135	NEGATIVE	SNV
EGFR I744T	p.I744T	COSM87245	NEGATIVE	SNV
EGFR K467T	p.K467T	BT132	NEGATIVE	SNV
EGFR K757R	p.K757R	COSM133588	NEGATIVE	SNV
EGFR L718M	p.L718M	COSM1716255	NEGATIVE	SNV
EGFR L718Q	p.L718Q	COSM6503269	NEGATIVE	SNV
EGFR L718V	p.L718V	OM3316	NEGATIVE	SNV
EGFR L747P	p.L747P	COSM24267	NEGATIVE	mnp
EGFR L747S	p.L747S	COSM26704	NEGATIVE	SNV
EGFR L747V	p.L747V	OM3537	NEGATIVE	SNV
EGFR L792F	p.L792F	COSM6493933	NEGATIVE	SNV
EGFR L792H	p.L792H	COSM6493934	NEGATIVE	SNV
EGFR L792P	p.L792P	COSM13190	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR L792R	p.L792R	OM3267	NEGATIVE	SNV
EGFR L792V	p.L792V	OM3265	NEGATIVE	SNV
EGFR L792Y	p.L792Y	OM3266	NEGATIVE	mnp
EGFR L798I	p.L798I	OM3268	NEGATIVE	SNV
EGFR L833F	p.L833F	COSM219794	NEGATIVE	SNV
EGFR L833V	p.L833V	COSM13424	NEGATIVE	SNV
EGFR L833W	p.L833W	COSM133204	NEGATIVE	SNV
EGFR L858K	p.L858K	COSM24268	NEGATIVE	mnp
EGFR L858M	p.L858M	COSM12366	NEGATIVE	SNV
EGFR L858Q	p.L858Q	COSM29578	NEGATIVE	SNV
EGFR L858R	p.L858R	COSM13553	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM12429	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM133630	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM6224	NEGATIVE	SNV
EGFR L861Q	p.L861Q	COSM6213	NEGATIVE	SNV
EGFR L861R	p.L861R	COSM12374	NEGATIVE	SNV
EGFR L861V	p.L861V	COSM24848	NEGATIVE	SNV
EGFR N771T	p.N771T	OM3174	NEGATIVE	SNV
EGFR P848L	p.P848L	COSM22943	NEGATIVE	SNV
EGFR R108G	p.R108G	COSM1451536	NEGATIVE	SNV
EGFR R108K	p.R108K	COSM21683	NEGATIVE	SNV
EGFR R451C	p.R451C	COSM3266583	NEGATIVE	SNV
EGFR R836C	p.R836C	COSM28604	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR S464L	p.S464L	BT126	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236671	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236670	NEGATIVE	SNV
EGFR S720I	p.S720I	OM3317	NEGATIVE	mnp
EGFR S720P	p.S720P	COSM88729	NEGATIVE	SNV
EGFR S768G	p.S768G	COSM1451595	NEGATIVE	SNV
EGFR S768I	p.S768I	COSM6241	NEGATIVE	SNV
EGFR S768N	p.S768N	COSM12989	NEGATIVE	SNV
EGFR S768T	p.S768T	COSM291998	NEGATIVE	SNV
EGFR T790A	p.T790A	COSM1272071	NEGATIVE	SNV
EGFR T790M	p.T790M	COSM6240	NEGATIVE	SNV
EGFR T854A	p.T854A	COSM28537	NEGATIVE	SNV
EGFR T854I	p.T854I	COSM87246	NEGATIVE	SNV
EGFR T854S	p.T854S	COSM1235378	NEGATIVE	SNV
EGFR V292L	p.V292L	COSM43067	NEGATIVE	SNV
EGFR V769L	p.V769L	COSM6242	NEGATIVE	SNV
EGFR V769M	p.V769M	COSM13425	NEGATIVE	SNV
EGFR [H773L;V774M]	p.[H773L;V774M]	COSM6978402	NEGATIVE	mnp
EGFR [K745=;E746I;L747P]	p.[K745=;E746I;L747P]	OM3355	NEGATIVE	mnp
EGFR [L718=;G719C]	p.[L718=;G719C]	COSM18441	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM6984779	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM85750	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	OMINDEL1181	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR [V769G;D770I]	p.[V769G;D770I]	OM3540	NEGATIVE	mnp
EGFR exon 19 deletion	p.I744_E749delinsLKR	COSM133186	NEGATIVE	complex
EGFR exon 19 deletion	p.I744_A750delinsVK	COSM85798	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750del	COSM221565	NEGATIVE	del
EGFR exon 19 deletion	p.K745_E749del	COSM26038	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM3734668	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750delinsT	COSM1190791	NEGATIVE	del
EGFR exon 19 deletion	p.E746del	COSM18420	NEGATIVE	del
EGFR exon 19 deletion	p.E746_E749del	COSM28517	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751del	COSM24869	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.E746_T751del	COSM12728	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752del	COSM133189	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsA	COSM13549	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM13550	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM13551	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM13552	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_E749del	COSM6218	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM6225	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsFPS	COSM6506513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsA	COSM12367	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsRP	COSM12413	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsA	COSM12678	NEGATIVE	del
EGFR exon 19 deletion	p.E746_P753delinsLS	COSM13200	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133188	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133190	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsIS	COSM133191	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsQP	COSM13557	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsQ	COSM22999	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_K754del	COSM24970	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsS	COSM26513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM26680	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsL	COSM51502	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM51526	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_A750delinsP	COSM6947327	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM6966471	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsM	OMINDEL716	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751del	COSM12369	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752delinsV	COSM12384	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM12386	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM12416	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM133193	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsAP	COSM144207	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM18421	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.L747_P753del	COSM255211	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsVP	COSM28623	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsVQ	COSM51524	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM52935	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM53205	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsD	COSM6220	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_S752delinsV	COSM674057	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_T751delinsVS	COSM6980200	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750del	COSM9179903	NEGATIVE	del
EGFR exon 19 deletion	p.L747_T751delinsQ	COSM12419	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQH	COSM12421	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12422	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsDP	COSM18428	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM22944	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQ	COSM26441	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.E746_A750delinsD	COSM6974307	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM1235296	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_P753delinsS	COSM12370	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12382	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM12383	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM12387	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_S752delinsQH	COSM133196	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_A750delinsP	COSM4170220	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsS	COSM4170221	NEGATIVE	del
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM51501	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsN	COSM51503	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsS	COSM6210	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A755delinsAN	COSM85891	NEGATIVE	complex
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_E758delinsP	COSM26440	NEGATIVE	complex

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.T751_E758del	COSM133207	NEGATIVE	del
EGFR exon 19 deletion	p.A750_I759delinsGS	COSM5023005	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM1667024	NEGATIVE	complex
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.S752_I759del	COSM13556	NEGATIVE	del
EGFR exon 19 deletion	p.T751_I759delinsS	COSM1667027	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsREA	COSM22956	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM24270	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM96856	NEGATIVE	complex
EGFR exon 19 deletion	p.S752_I759del	COSM6256	NEGATIVE	del
EGFR exon 19 deletion	p.P753_I759del	COSM24269	NEGATIVE	del
EGFR exon 19 deletion	p.K754_I759del	COSM5271797	NEGATIVE	del
EGFR exon 19 insertion	p.I740_K745dup	COSM12423	NEGATIVE	ins
EGFR exon 19 insertion	p.I740_K745dup	COSM51504	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insVPVAIK	COSM26444	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insTPVAIK	COSM255152	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.?	OMINDEL1050	NEGATIVE	ins
EGFR exon 20 insertion	p.?	COSM26720	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_D770dup	COSM6962256	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_V765insHH	OMINDEL1051	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM12376	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_S768insTLA	COSM12425	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM20884	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVASV	OMINDEL1800	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.S768_P772dup	OMINDEL1053	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSSV	OMINDEL1054	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insTSV	OMINDEL1055	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.S768_V769insLDS	OMINDEL1802	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insRSV	OMINDEL952	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.S768_N771dup	S768_N771dup	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.V769_D770insCV	COSM12379	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRVDNP	OMINDEL1056	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insANV	COSM1651741	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	COSM18430	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_N771dup	COSM20885	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_P772dup	OMINDEL1057	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_V769delinsISW	OMINDEL1058	NEGATIVE	complex
EGFR exon 20 insertion	p.S768delinsRRGH	OMINDEL1059	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTV	OMINDEL1060	NEGATIVE	ins
EGFR exon 20 insertion	p.V769dup	COSM6506514	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	OMINDEL1187	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	OMINDEL1803	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTM	OMINDEL1061	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1062	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1063	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1064	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1065	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1066	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM6845099	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM85795	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGG	MATNV07	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_D770insGE	OMINDEL1069	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGQ	OMINDEL1070	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1071	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.V769_D770insERG	COSM1651742	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM1651745	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM22955	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insMASVD	COSM28638	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	MATNV03	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771insGTD	OMINDEL1074	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1075	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insP	OMINDEL1076	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	OMINDEL1804	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771dup	OMINDEL1805	NEGATIVE	ins
EGFR exon 20 insertion	p.D770dup	OMINDEL1806	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insY	COSM1238030	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	COSM12381	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGY	COSM12427	NEGATIVE	ins
EGFR exon 20 insertion	p.N771dup	COSM13003	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGTH	COSM6983510	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGNPH	D770_N771insGNPH	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_1	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_2	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.D770delins
ASVH

OMINDEL108
0

NEGATIVE

ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771insH	OMINDEL1081	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.N771_P772insHN	OMINDEL1084	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.D770_N771insH	OMINDEL1807	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	OMINDEL1808	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1809	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsAGG	COSM12737	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insG	COSM13004	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM13558	NEGATIVE	complex
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_N771insT	COSM5023008	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insGNPH	COSM51544	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	COSM6977296	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGV	D770delinsGV	NEGATIVE	complex

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.D770_N771delinsAGH	MATNV09	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insPNP	OMINDEL1087	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1088	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsGGT	OMINDEL1090	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsEF	OMINDEL1091	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsEG	OMINDEL1092	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
EGFR exon 20 insertion	p.D770_N771insP	OMINDEL1096	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1097	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insANP	OMINDEL1098	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_H773insNPNP	OMINDEL1101	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1102	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1103	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1104	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1105	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1106	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1107	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1108	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1109	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insYPNP	OMINDEL1110	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772delinsASVDNR	OMINDEL1801	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsAS	OMINDEL1810	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insVDSVDNP	OMINDEL953	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	COSM12378	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGT	COSM1238029	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insSVE	COSM1651743	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insAPW	COSM20886	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_N771insSVE	D770_N771insSVE	NEGATIVE	ins
EGFR exon 20	p.D770_N771insG	MATNV05	NEGATIVE	ins

insertion

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771delinsERG	MATNV06	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1111	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1112	NEGATIVE	ins
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.P772_H773insTRNP	OMINDEL1116	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_N771insF	OMINDEL1812	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRNP	OMINDEL1813	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	COSM12380	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insH	COSM1238031	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSGH	COSM1651744	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insRH	COSM166390	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsGF	COSM18431	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsTH	COSM22946	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSH	COSM24434	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insTHP	COSM48923	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsVH	COSM5023007	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGY	COSM53189	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSTH	COSM6920147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insHH	COSM6931207	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsPH	MATNV04	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsHH	OMINDEL1118	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGD	OMINDEL1119	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1120	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1121	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insHPH	OMINDEL1122	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1123	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1124	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1125	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1126	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1127	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1128	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1129	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1130	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSPH	OMINDEL1131	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1132	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1133	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1134	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1135	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.N771delinsKPH	OMINDEL1188	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsCH	OMINDEL1814	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSQRGH	OMINDEL1815	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsHHR	OMINDEL1816	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGC	OMINDEL904	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL954	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insPHPH	p.H773_V774insPHPH_2	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsSVD NR	COSM13554	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsKL	COSM6438147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insV	COSM6922328	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insT	OMINDEL1138	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1139	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSVDS	OMINDEL1140	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1141	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1142	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insG	OMINDEL1143	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1144	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1145	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1146	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1183	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPP	OMINDEL955	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_C775dup	OMINDEL956	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_V774dup	COSM6845098	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1147	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_P772insT	OMINDEL1148	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1149	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1150	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.H773_V774insTH	OMINDEL1151	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insT	OMINDEL1152	NEGATIVE	ins
EGFR exon 20 insertion	p.H773dup	COSM12377	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774dup	COSM12389	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insNH	COSM5023006	NEGATIVE	ins
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_H773insL	OMINDEL1155	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insR	OMINDEL1156	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTP	COSM12388	NEGATIVE	complex
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.H773_V774insPHPH	H773_V774insPHPH	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.H773_V774insGH	OMINDEL1157	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1158	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1159	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	OMINDEL1184	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1811	NEGATIVE	complex
EGFR exon 20 insertion	p.H773delinsPNPY	COSM1735761	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsRY	MATNV08	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	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.H773delinsPNPY	OMINDEL1165	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1166	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1167	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insY	OMINDEL1168	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insTQPP	COSM3727813	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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
ERBB2 C311R	p.C311R	COSM35763	NEGATIVE	SNV
ERBB2 D769H	p.D769H	COSM13170	NEGATIVE	SNV
ERBB2 D769N	p.D769N	COSM1302747	NEGATIVE	SNV
ERBB2 D769Y	p.D769Y	COSM1251412	NEGATIVE	SNV
ERBB2 E321G	p.E321G	COSM35764	NEGATIVE	SNV
ERBB2 G309A	p.G309A	COSM436496	NEGATIVE	SNV
ERBB2 G309E	p.G309E	COSM94224	NEGATIVE	SNV
ERBB2 G660D	p.G660D	COSM4681497	NEGATIVE	SNV
ERBB2 G776A	p.G776A	OBENV03	NEGATIVE	SNV
ERBB2 G776C	p.G776C	COSM303938	NEGATIVE	SNV
ERBB2 G776L	p.G776L	COSM144255	NEGATIVE	mnp
ERBB2 G776R	p.G776R	OM3421	NEGATIVE	SNV
ERBB2 G776S	p.G776S	COSM685	NEGATIVE	SNV
ERBB2 G776V	p.G776V	COSM18609	NEGATIVE	SNV
ERBB2 I767M	p.I767M	COSM51317	NEGATIVE	SNV
ERBB2 L755A	p.L755A	COSM6906940	NEGATIVE	mnp
ERBB2 L755F	p.L755F	OBENV01	NEGATIVE	SNV
ERBB2 L755F	p.L755F	OBENV02	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 L755M	p.L755M	COSM1205571	NEGATIVE	SNV
ERBB2 L755S	p.L755S	COSM14060	NEGATIVE	SNV
ERBB2 L755W	p.L755W	COSM436499	NEGATIVE	SNV
ERBB2 L755_T759del	p.L755_T759del	COSM7345568	NEGATIVE	del
ERBB2 L755_T759del	p.L755_T759del	COSM5029269	NEGATIVE	del
ERBB2 L869R	p.L869R	COSM249793	NEGATIVE	SNV
ERBB2 R678Q	p.R678Q	COSM436498	NEGATIVE	SNV
ERBB2 R868W	p.R868W	COSM289684	NEGATIVE	SNV
ERBB2 R896C	p.R896C	COSM14066	NEGATIVE	SNV
ERBB2 R896H	p.R896H	COSM119971	NEGATIVE	SNV
ERBB2 S310F	p.S310F	COSM48358	NEGATIVE	SNV
ERBB2 S310P	p.S310P	COSM1563863	NEGATIVE	SNV
ERBB2 S310Y	p.S310Y	COSM6906440	NEGATIVE	mnp
ERBB2 S310Y	p.S310Y	COSM94225	NEGATIVE	SNV
ERBB2 T733I	p.T733I	COSM14059	NEGATIVE	SNV
ERBB2 T798I	p.T798I	OM3210	NEGATIVE	SNV
ERBB2 V659E	p.V659E	COSM3724566	NEGATIVE	mnp
ERBB2 V659E	p.V659E	COSM6503262	NEGATIVE	mnp
ERBB2 V777A	p.V777A	COSM35496	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM14062	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM436500	NEGATIVE	SNV
ERBB2 V777M	p.V777M	COSM14064	NEGATIVE	SNV
ERBB2 V842E	p.V842E	COSM6005745	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 V842I	p.V842I	COSM14065	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.Y772_V773insMMAY	OMINDEL820	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.A775_G776insVVMA	SPRNV01	NEGATIVE	ins
ERBB2 exon 20 insertion	p.M774dup	OMINDEL823	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insYLMA	SPRNV02	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_G776insV	MAN307	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insVAG	OMINDEL817	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insC	OMINDEL825	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.A775_G776insV	MAN300	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN308	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN309	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12552	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12553	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsLC	COSM12554	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM19875	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM20895	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsVC	COSM5235349	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsAVGC	COSM6907033	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM85995	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_V777delinsSVA	G776_V777delinsSVA_3	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_S779insCVG	OMINDEL1179	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsIC	OMINDEL826	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	SPRNV03	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776_V777insL	COSM6438151	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_S779dup	COSM681	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVV	COSM6977433	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.V777dup	COSM9869865	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_G778delinsVVGD	OMINDEL1189	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_P780dup	COSM12555	NEGATIVE	ins
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	COSM303948	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_P780dup	COSM6865893	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPG	COSM7449770	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insCPG	OMINDEL1180	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_G778insC	OMINDEL827	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPS	COSM5802314	NEGATIVE	ins
ERBB3 D297H	p.D297H	COSM1935392	NEGATIVE	SNV
ERBB3 D297N	p.D297N	COSM941489	NEGATIVE	SNV
ERBB3 D297V	p.D297V	COSM941490	NEGATIVE	SNV
ERBB3 D297Y	p.D297Y	COSM160822	NEGATIVE	SNV
ERBB3 E332K	p.E332K	COSM254677	NEGATIVE	SNV
ERBB3 E332Q	p.E332Q	COSM4838033	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM5413554	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB3 G284R	p.G284R	COSM941488	NEGATIVE	SNV
ERBB3 M60K	p.M60K	COSM254678	NEGATIVE	SNV
ERBB3 M60L	p.M60L	COSM1606366	NEGATIVE	SNV
ERBB3 M60R	p.M60R	COSM941484	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM122890	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM1299636	NEGATIVE	SNV
ERBB3 R103G	p.R103G	COSM6907312	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM160824	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM191840	NEGATIVE	SNV
ERBB3 V104M	p.V104M	COSM172423	NEGATIVE	SNV
ERBB4 R711C	p.R711C	COSM160827	NEGATIVE	SNV
ERBB4 R711L	p.R711L	OM3150	NEGATIVE	SNV
ESR1 D538G	p.D538G	COSM94250	NEGATIVE	SNV
ESR1 E380Q	p.E380Q	COSM3829320	NEGATIVE	SNV
ESR1 K303R	p.K303R	COSM4745827	NEGATIVE	SNV
ESR1 L536Q	p.L536Q	COSM4766050	NEGATIVE	mnp
ESR1 L536R	p.L536R	COSM4774826	NEGATIVE	SNV
ESR1 P535H	p.P535H	BT24	NEGATIVE	SNV
ESR1 P535T	p.P535T	COSM5666097	NEGATIVE	SNV
ESR1 R555C	p.R555C	BT32	NEGATIVE	SNV
ESR1 S463P	p.S463P	COSM4771561	NEGATIVE	SNV
ESR1 V392I	p.V392I	COSM1545070	NEGATIVE	SNV
ESR1 Y537C	p.Y537C	COSM1074637	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ESR1 Y537N	p.Y537N	COSM1074635	NEGATIVE	SNV
ESR1 Y537S	p.Y537S	COSM1074639	NEGATIVE	SNV
FGFR1 K687E	p.K687E	COSM35673	NEGATIVE	SNV
FGFR1 K687M	p.K687M	COSM4771556	NEGATIVE	SNV
FGFR1 K687N	p.K687N	COSM6578115	NEGATIVE	SNV
FGFR1 V592M	p.V592M	COSM6840954	NEGATIVE	SNV
FGFR2 A314D	p.A314D	COSM49171	NEGATIVE	SNV
FGFR2 A315T	p.A315T	COSM30777	NEGATIVE	SNV
FGFR2 A648T	p.A648T	COSM29836	NEGATIVE	SNV
FGFR2 C382G	p.C382G	COSM6197913	NEGATIVE	SNV
FGFR2 C382R	p.C382R	COSM36906	NEGATIVE	SNV
FGFR2 C382Y	p.C382Y	COSM915493	NEGATIVE	SNV
FGFR2 E565A	p.E565A	OM3292	NEGATIVE	SNV
FGFR2 E565G	p.E565G	OM3563	NEGATIVE	SNV
FGFR2 G613V	p.G613V	COSM94318	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM1474415	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM48382	NEGATIVE	SNV
FGFR2 I547V	p.I547V	COSM36913	NEGATIVE	SNV
FGFR2 K310R	p.K310R	COSM36901	NEGATIVE	SNV
FGFR2 K641R	p.K641R	COSM5010388	NEGATIVE	SNV
FGFR2 K659E	p.K659E	COSM36909	NEGATIVE	SNV
FGFR2 K659M	p.K659M	COSM49175	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM49173	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR2 K659N	p.K659N	COSM683054	NEGATIVE	SNV
FGFR2 L617M	p.L617M	OM3562	NEGATIVE	SNV
FGFR2 L617V	p.L617V	OM3291	NEGATIVE	SNV
FGFR2 M391R	p.M391R	COSM36907	NEGATIVE	SNV
FGFR2 N549D	p.N549D	COSM4604460	NEGATIVE	SNV
FGFR2 N549H	p.N549H	COSM250083	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36902	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36912	NEGATIVE	SNV
FGFR2 N549S	p.N549S	COSM3665553	NEGATIVE	SNV
FGFR2 N549T	p.N549T	COSM6952624	NEGATIVE	SNV
FGFR2 P253L	p.P253L	COSM537801	NEGATIVE	SNV
FGFR2 P253R	p.P253R	COSM49170	NEGATIVE	SNV
FGFR2 P253S	p.P253S	COSM1346291	NEGATIVE	SNV
FGFR2 S252W	p.S252W	COSM36903	NEGATIVE	SNV
FGFR2 V395D	p.V395D	COSM250081	NEGATIVE	mnp
FGFR2 V564F	p.V564F	OM3293	NEGATIVE	SNV
FGFR2 V564I	p.V564I	OM3247	NEGATIVE	SNV
FGFR2 V564L	p.V564L	OM3564	NEGATIVE	SNV
FGFR2 V679I	p.V679I	COSM4822956	NEGATIVE	SNV
FGFR2 Y375C	p.Y375C	COSM36904	NEGATIVE	SNV
FGFR2 Y375H	p.Y375H	COSM1560916	NEGATIVE	SNV
FGFR3 A391E	p.A391E	COSM721	NEGATIVE	SNV
FGFR3 A391T	p.A391T	OM3151	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 A391V	p.A391V	COSM1737909	NEGATIVE	SNV
FGFR3 D641G	p.D641G	COSM1428721	NEGATIVE	SNV
FGFR3 D641N	p.D641N	COSM29438	NEGATIVE	SNV
FGFR3 G370C	p.G370C	COSM716	NEGATIVE	SNV
FGFR3 G370D	p.G370D	COSM5966955	NEGATIVE	SNV
FGFR3 G380E	p.G380E	COSM51545	NEGATIVE	SNV
FGFR3 G380R	p.G380R	COSM24842	NEGATIVE	SNV
FGFR3 K650E	p.K650E	COSM719	NEGATIVE	SNV
FGFR3 K650M	p.K650M	COSM720	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM1428730	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM3993568	NEGATIVE	SNV
FGFR3 K650Q	p.K650Q	COSM726	NEGATIVE	SNV
FGFR3 K650R	p.K650R	OM3224	NEGATIVE	SNV
FGFR3 K650T	p.K650T	COSM731	NEGATIVE	SNV
FGFR3 K715M	p.K715M	COSM732992	NEGATIVE	SNV
FGFR3 R248C	p.R248C	COSM714	NEGATIVE	SNV
FGFR3 R248H	p.R248H	COSM1757310	NEGATIVE	SNV
FGFR3 R399C	p.R399C	COSM296687	NEGATIVE	SNV
FGFR3 S249C	p.S249C	COSM715	NEGATIVE	SNV
FGFR3 S249T	p.S249T	COSM29431	NEGATIVE	SNV
FGFR3 S371C	p.S371C	COSM17461	NEGATIVE	SNV
FGFR3 V553M	p.V553M	OM3223	NEGATIVE	SNV
FGFR3 V555M	p.V555M	COSM303914	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 V677I	p.V677I	COSM178247	NEGATIVE	SNV
FGFR3 Y373C	p.Y373C	COSM718	NEGATIVE	SNV
FGFR4 V550E	p.V550E	COSM9215998	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM1066172	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM3738141	NEGATIVE	SNV
FGFR4 V550M	p.V550M	COSM6163	NEGATIVE	SNV
FLT3 D835A	p.D835A	COSM27650	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM787	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM788	NEGATIVE	SNV
FLT3 D835F	p.D835F	COSM19844	NEGATIVE	mnp
FLT3 D835G	p.D835G	COSM1366188	NEGATIVE	SNV
FLT3 D835H	p.D835H	COSM785	NEGATIVE	SNV
FLT3 D835K	p.D835K	OM3570	NEGATIVE	mnp
FLT3 D835K	p.D835K	OM3571	NEGATIVE	mnp
FLT3 D835L	p.D835L	OM3572	NEGATIVE	mnp
FLT3 D835N	p.D835N	COSM789	NEGATIVE	SNV
FLT3 D835V	p.D835V	COSM784	NEGATIVE	SNV
FLT3 D835Y	p.D835Y	COSM783	NEGATIVE	SNV
FLT3 D835_I836del	p.D835_I836del	COSM5879385	NEGATIVE	del
FLT3 D835_I836delinsV	p.D835_I836delinsV	COSM27651	NEGATIVE	del
FLT3 D835del	p.D835del	COSM854	NEGATIVE	del
FLT3 D839E	p.D839E	COSM5945287	NEGATIVE	SNV
FLT3 D839G	p.D839G	COSM1166729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FLT3 D839H	p.D839H	COSM3399304	NEGATIVE	SNV
FLT3 D839N	p.D839N	COSM6005768	NEGATIVE	SNV
FLT3 I836D	p.I836D	OM3569	NEGATIVE	mnp
FLT3 I836L	p.I836L	COSM19649	NEGATIVE	SNV
FLT3 I836S	p.I836S	COSM19843	NEGATIVE	SNV
FLT3 I836T	p.I836T	COSM5879386	NEGATIVE	SNV
FLT3 I836del	p.I836del	COSM19836	NEGATIVE	del
GNAS Q227E	p.Q227E	COSM4962873	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM27900	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM4168960	NEGATIVE	SNV
GNAS Q227K	p.Q227K	COSM28618	NEGATIVE	SNV
GNAS Q227L	p.Q227L	COSM27888	NEGATIVE	SNV
GNAS Q227R	p.Q227R	COSM27896	NEGATIVE	SNV
GNAS R201C	p.R201C	COSM27887	NEGATIVE	SNV
GNAS R201G	p.R201G	COSM1757313	NEGATIVE	SNV
GNAS R201H	p.R201H	COSM27895	NEGATIVE	SNV
GNAS R201L	p.R201L	COSM99221	NEGATIVE	SNV
GNAS R201S	p.R201S	COSM27899	NEGATIVE	SNV
HRAS G12A	p.G12A	COSM485	NEGATIVE	SNV
HRAS G12C	p.G12C	COSM481	NEGATIVE	SNV
HRAS G12D	p.G12D	COSM484	NEGATIVE	SNV
HRAS G12N	p.G12N	COSM4517473	NEGATIVE	mnp
HRAS G12R	p.G12R	COSM482	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
HRAS G12S	p.G12S	COSM480	NEGATIVE	SNV
HRAS G12V	p.G12V	COSM483	NEGATIVE	SNV
HRAS G13C	p.G13C	COSM488	NEGATIVE	SNV
HRAS G13D	p.G13D	COSM490	NEGATIVE	SNV
HRAS G13R	p.G13R	COSM486	NEGATIVE	SNV
HRAS G13S	p.G13S	COSM487	NEGATIVE	SNV
HRAS G13V	p.G13V	COSM489	NEGATIVE	SNV
HRAS Q61E	p.Q61E	COSM497	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM502	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM503	NEGATIVE	SNV
HRAS Q61K	p.Q61K	COSM496	NEGATIVE	SNV
HRAS Q61L	p.Q61L	COSM52978	NEGATIVE	mnp
HRAS Q61L	p.Q61L	COSM498	NEGATIVE	SNV
HRAS Q61P	p.Q61P	COSM500	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM33695	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM501	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM499	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM52979	NEGATIVE	mnp
IDH1 G70D	p.G70D	COSM96531	NEGATIVE	SNV
IDH1 R132C	p.R132C	COSM28747	NEGATIVE	SNV
IDH1 R132G	p.R132G	COSM28749	NEGATIVE	SNV
IDH1 R132H	p.R132H	COSM86993	NEGATIVE	mnp
IDH1 R132H	p.R132H	COSM28746	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
IDH1 R132L	p.R132L	COSM28750	NEGATIVE	SNV
IDH1 R132Q	p.R132Q	OM3221	NEGATIVE	mnp
IDH1 R132Q	p.R132Q	OM3222	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM1169216	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM28748	NEGATIVE	SNV
IDH1 R132V	p.R132V	COSM28751	NEGATIVE	mnp
IDH1 S280F	p.S280F	OM3315	NEGATIVE	SNV
IDH2 I319M	p.I319M	OM3319	NEGATIVE	SNV
IDH2 Q316E	p.Q316E	OM3320	NEGATIVE	SNV
IDH2 R140G	p.R140G	COSM1737874	NEGATIVE	SNV
IDH2 R140L	p.R140L	COSM41875	NEGATIVE	SNV
IDH2 R140Q	p.R140Q	COSM41590	NEGATIVE	SNV
IDH2 R140W	p.R140W	COSM41877	NEGATIVE	SNV
IDH2 R172G	p.R172G	COSM33731	NEGATIVE	SNV
IDH2 R172K	p.R172K	COSM33733	NEGATIVE	SNV
IDH2 R172M	p.R172M	COSM33732	NEGATIVE	SNV
IDH2 R172N	p.R172N	COSM256046	NEGATIVE	mnp
IDH2 R172S	p.R172S	COSM133672	NEGATIVE	SNV
IDH2 R172S	p.R172S	COSM34090	NEGATIVE	SNV
IDH2 R172T	p.R172T	COSM1636997	NEGATIVE	SNV
IDH2 R172V	p.R172V	OM3327	NEGATIVE	mnp
IDH2 R172W	p.R172W	COSM34039	NEGATIVE	SNV
KEAP1 D422N	p.D422N	COSM710198	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 G333C	p.G333C	COSM1193323	NEGATIVE	SNV
KEAP1 M409*	p.M409*	COSM6916353	NEGATIVE	del
KEAP1 R320L	p.R320L	COSM2812614	NEGATIVE	SNV
KEAP1 R320P	p.R320P	COSM346494	NEGATIVE	SNV
KEAP1 R320Q	p.R320Q	COSM710195	NEGATIVE	SNV
KEAP1 R320W	p.R320W	COSM1524062	NEGATIVE	SNV
KEAP1 R415G	p.R415G	COSM6196677	NEGATIVE	SNV
KEAP1 R470C	p.R470C	COSM564847	NEGATIVE	SNV
KEAP1 R470G	p.R470G	OM3724	NEGATIVE	SNV
KEAP1 R470H	p.R470H	COSM1524064	NEGATIVE	SNV
KEAP1 R470S	p.R470S	COSM564846	NEGATIVE	SNV
KEAP1 R554Q	p.R554Q	COSM1524066	NEGATIVE	SNV
KEAP1 S243C	p.S243C	COSM710191	NEGATIVE	SNV
KEAP1 V155F	p.V155F	COSM710187	NEGATIVE	SNV
KEAP1 V167F	p.V167F	COSM710188	NEGATIVE	SNV
KEAP1 Y584*	p.Y584*	COSM6201646	NEGATIVE	ins
KEAP1 c.1531+1G>T	p.?	COSM6956065	NEGATIVE	SNV
KEAP1 c.1531+2T>G	p.?	OM3723	NEGATIVE	SNV
KEAP1 c.1532-1G>T	p.?	COSM6969960	NEGATIVE	SNV
KEAP1 c.1532-2A>G	p.?	COSM381459	NEGATIVE	SNV
KEAP1 c.1708+1G>T	p.?	COSM7738757	NEGATIVE	SNV
KEAP1 c.1708+2T>A	p.?	COSM6149754	NEGATIVE	SNV
KEAP1 c.1708_1708+1delinsT	p.?	COSM6927565	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
T				
KEAP1 c.1709-1G>T	p.?	COSM7724088	NEGATIVE	SNV
KEAP1 c.1709-2A>G	p.?	COSM7088100	NEGATIVE	SNV
KEAP1 c.640-1G>A	p.?	COSM6972470	NEGATIVE	SNV
KEAP1 c.640-1G>C	p.?	OM3726	NEGATIVE	SNV
KEAP1 c.640-1G>T	p.?	OM3725	NEGATIVE	SNV
KEAP1 c.640-2A>C	p.?	OM3727	NEGATIVE	SNV
KIT A502_Y503dup	p.A502_Y503dup	COSM1326	NEGATIVE	ins
KIT A502_Y503insFA	p.A502_Y503insFA	COSM53306	NEGATIVE	ins
KIT A829P	p.A829P	COSM13172	NEGATIVE	SNV
KIT A829V	p.A829V	COSM133757	NEGATIVE	SNV
KIT D419_L421delinsF	p.D419_L421delinsF	COSM1152	NEGATIVE	complex
KIT D419_L421delinsH	p.D419_L421delinsH	COSM1578133	NEGATIVE	complex
KIT D419_R420del	p.D419_R420del	COSM1578132	NEGATIVE	del
KIT D419_R420insFF	p.D419_R420insFF	COSM36334	NEGATIVE	ins
KIT D419_R420insFL	p.D419_R420insFL	COSM36332	NEGATIVE	ins
KIT D419del	p.D419del	COSM29014	NEGATIVE	del
KIT D419del	p.D419del	COSM1600400	NEGATIVE	complex
KIT D816A	p.D816A	COSM24675	NEGATIVE	SNV
KIT D816E	p.D816E	COSM19285	NEGATIVE	SNV
KIT D816E	p.D816E	COSM5749965	NEGATIVE	SNV
KIT D816F	p.D816F	COSM1312	NEGATIVE	mnp
KIT D816G	p.D816G	COSM12711	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT D816H	p.D816H	COSM1311	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1600404	NEGATIVE	mnp
KIT D816I	p.D816I	COSM21979	NEGATIVE	mnp
KIT D816N	p.D816N	COSM1313	NEGATIVE	SNV
KIT D816V	p.D816V	COSM1314	NEGATIVE	SNV
KIT D816Y	p.D816Y	COSM1310	NEGATIVE	SNV
KIT D820A	p.D820A	COSM133670	NEGATIVE	SNV
KIT D820E	p.D820E	COSM12709	NEGATIVE	SNV
KIT D820E	p.D820E	COSM19280	NEGATIVE	SNV
KIT D820G	p.D820G	COSM1316	NEGATIVE	SNV
KIT D820H	p.D820H	COSM22379	NEGATIVE	SNV
KIT D820N	p.D820N	COSM17947	NEGATIVE	SNV
KIT D820V	p.D820V	COSM1317	NEGATIVE	SNV
KIT D820Y	p.D820Y	COSM12710	NEGATIVE	SNV
KIT F522C	p.F522C	OM3237	NEGATIVE	SNV
KIT H697Y	p.H697Y	COSM36053	NEGATIVE	SNV
KIT I653T	p.I653T	COSM19326	NEGATIVE	SNV
KIT K550I	p.K550I	COSM1165	NEGATIVE	SNV
KIT K550N	p.K550N	COSM1167	NEGATIVE	SNV
KIT K550R	p.K550R	COSM1166	NEGATIVE	SNV
KIT K558N	p.K558N	OM3238	NEGATIVE	SNV
KIT K558N	p.K558N	COSM19216	NEGATIVE	SNV
KIT K642E	p.K642E	COSM1304	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT K642N	p.K642N	COSM4125155	NEGATIVE	SNV
KIT K642Q	p.K642Q	COSM96871	NEGATIVE	SNV
KIT L576F	p.L576F	COSM33966	NEGATIVE	SNV
KIT L576P	p.L576P	COSM1290	NEGATIVE	SNV
KIT L576R	p.L576R	COSM5945006	NEGATIVE	SNV
KIT N505I	p.N505I	OM3236	NEGATIVE	SNV
KIT N655K	p.N655K	COSM235682	NEGATIVE	SNV
KIT N655K	p.N655K	COSM4413464	NEGATIVE	SNV
KIT N655S	p.N655S	OM3302	NEGATIVE	SNV
KIT N655T	p.N655T	OM3303	NEGATIVE	SNV
KIT N680K	p.N680K	COSM5352270	NEGATIVE	SNV
KIT N822H	p.N822H	COSM1318	NEGATIVE	SNV
KIT N822I	p.N822I	COSM3604523	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1321	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1322	NEGATIVE	SNV
KIT N822S	p.N822S	COSM33975	NEGATIVE	SNV
KIT N822Y	p.N822Y	COSM19109	NEGATIVE	SNV
KIT P838L	p.P838L	OM3240	NEGATIVE	SNV
KIT R796A	p.R796A	COSM1272091	NEGATIVE	mnp
KIT R796G	p.R796G	COSM51381	NEGATIVE	SNV
KIT R796K	p.R796K	COSM1600411	NEGATIVE	SNV
KIT R796P	p.R796P	COSM1272092	NEGATIVE	mnp
KIT S501_A502dup	p.S501_A502dup	COSM5702501	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT S501_A502dup	p.S501_A502dup	COSM219786	NEGATIVE	ins
KIT T417_D419delinsI	p.T417_D419delinsI	COSM1148	NEGATIVE	complex
KIT T417_D419delinsRG	p.T417_D419delinsRG	COSM1149	NEGATIVE	complex
KIT T670A	p.T670A	OM3304	NEGATIVE	SNV
KIT T670E	p.T670E	COSM19293	NEGATIVE	mnp
KIT T670I	p.T670I	COSM12708	NEGATIVE	SNV
KIT V530I	p.V530I	COSM1155	NEGATIVE	SNV
KIT V559A	p.V559A	COSM1255	NEGATIVE	SNV
KIT V559C	p.V559C	OM3239	NEGATIVE	mnp
KIT V559D	p.V559D	COSM1252	NEGATIVE	SNV
KIT V559G	p.V559G	COSM1253	NEGATIVE	SNV
KIT V559I	p.V559I	COSM1251	NEGATIVE	SNV
KIT V559K	p.V559K	COSM96869	NEGATIVE	mnp
KIT V559P	p.V559P	COSM1272086	NEGATIVE	mnp
KIT V560A	p.V560A	COSM36302	NEGATIVE	SNV
KIT V560D	p.V560D	COSM1257	NEGATIVE	SNV
KIT V560E	p.V560E	COSM1333	NEGATIVE	mnp
KIT V560G	p.V560G	COSM1260	NEGATIVE	SNV
KIT V654A	p.V654A	COSM12706	NEGATIVE	SNV
KIT V654L	p.V654L	COSM5941904	NEGATIVE	SNV
KIT V825A	p.V825A	COSM1323	NEGATIVE	SNV
KIT V825I	p.V825I	COSM19110	NEGATIVE	SNV
KIT W557C	p.W557C	COSM19108	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT W557C	p.W557C	COSM133700	NEGATIVE	SNV
KIT W557G	p.W557G	COSM1221	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1216	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1219	NEGATIVE	SNV
KIT W557S	p.W557S	COSM1227	NEGATIVE	SNV
KIT Y553C	p.Y553C	COSM4413463	NEGATIVE	SNV
KIT Y553N	p.Y553N	COSM133763	NEGATIVE	SNV
KIT Y570H	p.Y570H	COSM1430152	NEGATIVE	SNV
KIT Y578C	p.Y578C	COSM6503446	NEGATIVE	SNV
KIT Y823C	p.Y823C	COSM18682	NEGATIVE	SNV
KIT Y823D	p.Y823D	COSM18681	NEGATIVE	SNV
KIT Y823H	p.Y823H	COSM1651649	NEGATIVE	SNV
KIT Y823N	p.Y823N	COSM22378	NEGATIVE	SNV
KIT [K558S;V559S]	p.[K558S;V559S]	COSM28637	NEGATIVE	mnp
KIT [W557C;K558P]	p.[W557C;K558P]	COSM21978	NEGATIVE	mnp
KIT [W557C;K558T]	p.[W557C;K558T]	COSM28992	NEGATIVE	mnp
KIT [W557F;K558P]	p.[W557F;K558P]	COSM1224	NEGATIVE	mnp
KIT [W557S;K558S]	p.[W557S;K558S]	COSM96948	NEGATIVE	mnp
KIT exon 11 deletion	p.?	OMINDEL901	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1163	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1158	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1160	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1162	NEGATIVE	del

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.P551_E554del	COSM1172	NEGATIVE	del
KIT exon 11 deletion	p.K550_V555delinsQ	COSM6938892	NEGATIVE	complex
KIT exon 11 deletion	p.K550_Q556delinsII	COSM1161	NEGATIVE	complex
KIT exon 11 deletion	p.P551_V555del	COSM1169	NEGATIVE	del
KIT exon 11 deletion	p.K550_W557delinsIL	COSM6936468	NEGATIVE	complex
KIT exon 11 deletion	p.K550_E554delinsI	COSM6953101	NEGATIVE	complex
KIT exon 11 deletion	p.K550_P551delinsI	COSM6971267	NEGATIVE	complex
KIT exon 11 deletion	p.K550_W557delinsIR	COSM6977972	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Q556del	COSM1170	NEGATIVE	del
KIT exon 11 deletion	p.P551del	COSM1171	NEGATIVE	del
KIT exon 11 deletion	p.P551_V555delinsL	COSM1174	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.P551_V555delinsTL	COSM30550	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Y553delinsH	COSM6934157	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1177	NEGATIVE	del
KIT exon 11 deletion	p.M552_V555del	COSM6957862	NEGATIVE	del
KIT exon 11 deletion	p.M552_Y553del	COSM96881	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsL	COSM1178	NEGATIVE	complex
KIT exon 11 deletion	p.M552_Y553del	COSM1179	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1180	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554del	COSM1181	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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_V555del	COSM18827	NEGATIVE	del
KIT exon 11 deletion	p.M552_K558del	OMINDEL902	NEGATIVE	complex
KIT exon 11 deletion	p.M552_V555delinsI	COSM1185	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557delinsR	COSM133636	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.M552_Y553delinsN	COSM96863	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsK	COSM96868	NEGATIVE	del
KIT exon 11 deletion	p.M552_E561delinsK	COSM96880	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_K558del	COSM27909	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554delinsI	COSM6944981	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559del	COSM1191	NEGATIVE	del
KIT exon 11 deletion	p.Y553_E554del	COSM6923694	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559delinsE	COSM96946	NEGATIVE	complex
KIT exon 11 deletion	p.E554_N564del	COSM96947	NEGATIVE	del
KIT exon 11 deletion	p.E554_V559del	OMINDEL701	NEGATIVE	del
KIT exon 11 deletion	p.E554_K558del	COSM1192	NEGATIVE	del
KIT exon 11 deletion	p.V555_E562del	COSM1194	NEGATIVE	del
KIT exon 11 deletion	p.E554_E562del	COSM36310	NEGATIVE	del

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.V555_G565del	COSM37027	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1198	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559del	COSM1199	NEGATIVE	del
KIT exon 11 deletion	p.V555_Q556del	COSM1201	NEGATIVE	del
KIT exon 11 deletion	p.V555_V560del	COSM1202	NEGATIVE	del
KIT exon 11 deletion	p.V555_N566delinsD	COSM96870	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1204	NEGATIVE	del
KIT exon 11 deletion	p.Q556_E561del	COSM1206	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558del	COSM23418	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.W557_K558del	COSM6986076	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsF	COSM1207	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558del	COSM1217	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561del	COSM1332	NEGATIVE	del
KIT exon 11 deletion	p.Q556_W557delinsR	COSM133642	NEGATIVE	del
KIT exon 11 deletion	p.Q556_I563delinsL	COSM1651755	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsTTF	COSM27904	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E562del	COSM36312	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsH	COSM1213	NEGATIVE	del
KIT exon 11 deletion	p.W557del	COSM1220	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559delinsH	COSM133641	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_E561delinsHH	COSM1208	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V559delinsHT	COSM1214	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559del	COSM1218	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560del	COSM1223	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561delinsPS	COSM6922994	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E561delinsQ	COSM6954755	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsI	COSM1222	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsF	COSM1226	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560delinsF	COSM1230	NEGATIVE	del
KIT exon 11 deletion	p.W557_K558delinsE	COSM24748	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558delinsQ	COSM6946777	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V560delinsC	COSM1232	NEGATIVE	del
KIT exon 11 deletion	p.W557_V559delinsC	COSM1233	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.K558_V560del	COSM1234	NEGATIVE	del
KIT exon 11 deletion	p.K558_V559del	COSM1235	NEGATIVE	del
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.W557_K558delinsC	COSM96887	NEGATIVE	complex
KIT exon 11 deletion	p.K558_G565delinsR	COSM1240	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K558_V560delinsI	COSM1241	NEGATIVE	del
KIT exon 11 deletion	p.V559_E561del	COSM1250	NEGATIVE	del
KIT exon 11 deletion	p.K558_Y570delinsN	COSM28991	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsL	COSM6940807	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V560delinsM	COSM1242	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V559delinsN	COSM1244	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsN	COSM27069	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM6971633	NEGATIVE	del
KIT exon 11 deletion	p.V560del	COSM1247	NEGATIVE	del
KIT exon 11 deletion	p.V559_V560del	COSM1248	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM1249	NEGATIVE	del
KIT exon 11 deletion	p.V559_I571del	COSM36301	NEGATIVE	del
KIT exon 11 deletion	p.V560_I563del	COSM18897	NEGATIVE	del
KIT exon 11 deletion	p.V559_N566delinsD	COSM6977976	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1262	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1263	NEGATIVE	del
KIT exon 11 deletion	p.E562_V569delinsD	COSM1267	NEGATIVE	del
KIT exon 11 deletion	p.I563_D572del	COSM1268	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.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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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_Q575del	COSM1281	NEGATIVE	del
KIT exon 11 deletion	p.V569_D572del	COSM1282	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 deletion	p.I571_Y578del	OMINDEL1186	NEGATIVE	del
KIT exon 11 deletion	p.I571_L576del	COSM133754	NEGATIVE	del
KIT exon 11 deletion	p.Q575del	COSM96875	NEGATIVE	del
KIT exon 11 deletion	p.Q575_L576del	COSM6987753	NEGATIVE	del
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.Q575_P577delinsT	COSM96865	NEGATIVE	complex
KIT exon 11 deletion	p.L576del	COSM1289	NEGATIVE	del
KIT exon 11 deletion	p.P577_Y578del	COSM1291	NEGATIVE	del
KIT exon 11 deletion	p.P577del	COSM1684822	NEGATIVE	del
KIT exon 11 deletion	p.P577_D579del	COSM98379	NEGATIVE	del
KIT exon 11 deletion	p.D579del	COSM1294	NEGATIVE	del
KIT exon 11 deletion	p.H580del	COSM1295	NEGATIVE	del
KIT exon 11 insertion	p.K558delinsNP	COSM21976	NEGATIVE	ins
KIT exon 11 insertion	p.K558delinsNP	COSM1245	NEGATIVE	complex
KIT exon 11 insertion	p.I571_D579dup	COSM23417	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 insertion	p.D572_D579dup	COSM308544	NEGATIVE	ins
KIT exon 11 insertion	p.P573_H580dup	COSM34126	NEGATIVE	ins
KIT exon 11 insertion	p.P573_D579dup	COSM18668	NEGATIVE	ins
KIT exon 11 insertion	p.T574_D579dup	OMINDEL702	NEGATIVE	ins
KIT exon 11 insertion	p.Q575_L576dup	COSM1292	NEGATIVE	ins
KIT exon 11 insertion	p.Y578_P585dup	COSM41632	NEGATIVE	ins
KIT exon 11 insertion	p.P577_H580dup	COSM1296	NEGATIVE	ins
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
KRAS A146G	p.A146G	COSM1360828	NEGATIVE	SNV
KRAS A146P	p.A146P	COSM19905	NEGATIVE	SNV
KRAS A146S	p.A146S	COSM5967494	NEGATIVE	SNV
KRAS A146T	p.A146T	COSM19404	NEGATIVE	SNV
KRAS A146V	p.A146V	COSM19900	NEGATIVE	SNV
KRAS A59E	p.A59E	COSM547	NEGATIVE	SNV
KRAS A59G	p.A59G	COSM28518	NEGATIVE	SNV
KRAS A59S	p.A59S	COSM1235389	NEGATIVE	SNV
KRAS A59T	p.A59T	COSM546	NEGATIVE	SNV
KRAS G12A	p.G12A	COSM522	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G12C	p.G12C	COSM516	NEGATIVE	SNV
KRAS G12D	p.G12D	COSM14209	NEGATIVE	mnp
KRAS G12D	p.G12D	COSM521	NEGATIVE	SNV
KRAS G12E	p.G12E	COSM30566	NEGATIVE	mnp
KRAS G12E	p.G12E	COSM519	NEGATIVE	mnp
KRAS G12F	p.G12F	COSM512	NEGATIVE	mnp
KRAS G12I	p.G12I	COSM34144	NEGATIVE	mnp
KRAS G12L	p.G12L	COSM514	NEGATIVE	mnp
KRAS G12N	p.G12N	COSM13643	NEGATIVE	mnp
KRAS G12R	p.G12R	COSM518	NEGATIVE	SNV
KRAS G12S	p.G12S	COSM517	NEGATIVE	SNV
KRAS G12V	p.G12V	COSM515	NEGATIVE	mnp
KRAS G12V	p.G12V	COSM520	NEGATIVE	SNV
KRAS G12W	p.G12W	COSM36281	NEGATIVE	mnp
KRAS G12Y	p.G12Y	COSM25081	NEGATIVE	mnp
KRAS G13A	p.G13A	COSM533	NEGATIVE	SNV
KRAS G13C	p.G13C	COSM527	NEGATIVE	SNV
KRAS G13D	p.G13D	COSM531	NEGATIVE	mnp
KRAS G13D	p.G13D	COSM532	NEGATIVE	SNV
KRAS G13E	p.G13E	COSM30567	NEGATIVE	mnp
KRAS G13E	p.G13E	COSM87280	NEGATIVE	mnp
KRAS G13I	p.G13I	COSM525	NEGATIVE	mnp
KRAS G13N	p.G13N	COSM53283	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G13R	p.G13R	COSM526	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM529	NEGATIVE	SNV
KRAS G13S	p.G13S	COSM528	NEGATIVE	SNV
KRAS G13V	p.G13V	COSM12721	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM530	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM534	NEGATIVE	SNV
KRAS G15D	p.G15D	COSM539	NEGATIVE	SNV
KRAS G15S	p.G15S	COSM538	NEGATIVE	SNV
KRAS K117E	p.K117E	COSM6854421	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM19940	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM28519	NEGATIVE	SNV
KRAS K117R	p.K117R	COSM4696721	NEGATIVE	SNV
KRAS Q61E	p.Q61E	COSM550	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM554	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM555	NEGATIVE	SNV
KRAS Q61K	p.Q61K	COSM549	NEGATIVE	SNV
KRAS Q61L	p.Q61L	COSM553	NEGATIVE	SNV
KRAS Q61P	p.Q61P	COSM551	NEGATIVE	SNV
KRAS Q61R	p.Q61R	COSM1168052	NEGATIVE	mnp
KRAS Q61R	p.Q61R	COSM552	NEGATIVE	SNV
KRAS [G12=;G13C]	p.[G12=;G13C]	COSM87281	NEGATIVE	mnp
KRAS [G13D;V14I]	p.[G13D;V14I]	COSM30568	NEGATIVE	mnp
KRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM87298	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 C121S	p.C121S	COSM555601	NEGATIVE	SNV
MAP2K1 C121S	p.C121S	COSM1315829	NEGATIVE	SNV
MAP2K1 E203K	p.E203K	COSM232755	NEGATIVE	SNV
MAP2K1 E203V	p.E203V	COSM3386991	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3249	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	COSM1235480	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3250	NEGATIVE	SNV
MAP2K1 F53C	p.F53C	COSM1562837	NEGATIVE	SNV
MAP2K1 F53I	p.F53I	COSM3503329	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM555604	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM1725008	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM2257208	NEGATIVE	SNV
MAP2K1 F53S	p.F53S	ClinVar13350	NEGATIVE	SNV
MAP2K1 F53V	p.F53V	COSM5077832	NEGATIVE	SNV
MAP2K1 F53Y	p.F53Y	COSM5588244	NEGATIVE	SNV
MAP2K1 G128D	p.G128D	COSM1374186	NEGATIVE	SNV
MAP2K1 G128V	p.G128V	COSM1517232	NEGATIVE	SNV
MAP2K1 K57E	p.K57E	COSM5369532	NEGATIVE	SNV
MAP2K1 K57M	p.K57M	MAN124	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM1235478	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM5520914	NEGATIVE	SNV
MAP2K1 K57Q	p.K57Q	OM3155	NEGATIVE	SNV
MAP2K1 K57T	p.K57T	COSM4756761	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 L115P	p.L115P	OM3248	NEGATIVE	SNV
MAP2K1 L215P	p.L215P	OM3252	NEGATIVE	SNV
MAP2K1 P124L	p.P124L	COSM1315861	NEGATIVE	SNV
MAP2K1 P124Q	p.P124Q	COSM1167912	NEGATIVE	SNV
MAP2K1 P124S	p.P124S	COSM235614	NEGATIVE	SNV
MAP2K1 Q56P	p.Q56P	COSM1235481	NEGATIVE	SNV
MAP2K1 V211D	p.V211D	OM3251	NEGATIVE	SNV
MAP2K1 V60E	p.V60E	COSM2257209	NEGATIVE	SNV
MAP2K1 V60M	p.V60M	OM3576	NEGATIVE	SNV
MAP2K2 C125S	p.C125S	COSM5855814	NEGATIVE	SNV
MAP2K2 N126D	p.N126D	COSM1731742	NEGATIVE	SNV
MAP2K2 V35M	p.V35M	COSM5855836	NEGATIVE	SNV
MET H1112L	p.H1112L	COSM698	NEGATIVE	SNV
MET H1112R	p.H1112R	COSM703	NEGATIVE	SNV
MET H1112Y	p.H1112Y	COSM696	NEGATIVE	SNV
MET Y1021F	p.Y1021F	COSM339515	NEGATIVE	SNV
MET Y1021S	p.Y1021S	COSM7450124	NEGATIVE	SNV
MET c.2942-20_2942-19delinsAGAG	p.?	OMINDEL804	NEGATIVE	complex
MET c.2942-21_2942-20delinsGC	p.?	COSM8631602	NEGATIVE	mnp
MET c.2942-29_2942-28delinsGG	p.?	OM3305	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM29635	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.?	OMINDEL674	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM49015	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6980941	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL709	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6056832	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL675	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL676	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL710	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.?	COSM6057022	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL640	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL697	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6947399	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL642	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL677	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL679	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL712	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6948341	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL680	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL681	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.?	COSM6976260	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL684	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL706	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL685	NEGATIVE	ins
MET exon 14 skipping (DNA)	p.?	OMINDEL808	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OM3177	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3178	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL809	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.A1023IfsTer19	OMINDEL810	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6057025	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL686	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6937267	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6977526	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL654	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL655	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL811	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL656	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL658	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL688	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL687	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL689	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL662	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.?	COSM6965225	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.?	OMINDEL812	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL813	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.?	OMINDEL814	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.?	OMINDEL694	NEGATIVE	del
MET exon 14 skipping (DNA)	p.D1028IfsTer19	COSM24687	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.D1028Y	COSM6917314	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.D1028N	COSM6924587	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL695	NEGATIVE	complex
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.?	COSM6928344	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6940825	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6983140	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL815	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6108460	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6947995	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3185	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3217	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM744044	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3186	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3188	NEGATIVE	SNV
NRAS A146T	p.A146T	COSM27174	NEGATIVE	SNV
NRAS A146V	p.A146V	COSM4170228	NEGATIVE	SNV
NRAS A59D	p.A59D	COSM253327	NEGATIVE	SNV
NRAS A59T	p.A59T	COSM578	NEGATIVE	SNV
NRAS G12A	p.G12A	COSM565	NEGATIVE	SNV
NRAS G12C	p.G12C	COSM562	NEGATIVE	SNV
NRAS G12D	p.G12D	COSM564	NEGATIVE	SNV
NRAS G12E	p.G12E	COSM144577	NEGATIVE	mnp
NRAS G12N	p.G12N	COSM12723	NEGATIVE	mnp
NRAS G12P	p.G12P	COSM559	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS G12R	p.G12R	COSM561	NEGATIVE	SNV
NRAS G12S	p.G12S	COSM563	NEGATIVE	SNV
NRAS G12V	p.G12V	COSM566	NEGATIVE	SNV
NRAS G12Y	p.G12Y	COSM560	NEGATIVE	mnp
NRAS G13A	p.G13A	COSM575	NEGATIVE	SNV
NRAS G13C	p.G13C	COSM570	NEGATIVE	SNV
NRAS G13D	p.G13D	COSM573	NEGATIVE	SNV
NRAS G13N	p.G13N	COSM24668	NEGATIVE	mnp
NRAS G13R	p.G13R	COSM569	NEGATIVE	SNV
NRAS G13S	p.G13S	COSM571	NEGATIVE	SNV
NRAS G13V	p.G13V	COSM572	NEGATIVE	mnp
NRAS G13V	p.G13V	COSM574	NEGATIVE	SNV
NRAS G13Y	p.G13Y	COSM568	NEGATIVE	mnp
NRAS K117N	p.K117N	MAN13	NEGATIVE	SNV
NRAS Q61E	p.Q61E	COSM581	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM585	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM586	NEGATIVE	SNV
NRAS Q61K	p.Q61K	COSM53223	NEGATIVE	mnp
NRAS Q61K	p.Q61K	COSM580	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM30646	NEGATIVE	mnp
NRAS Q61L	p.Q61L	COSM583	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM12725	NEGATIVE	mnp
NRAS Q61P	p.Q61P	COSM582	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS Q61R	p.Q61R	COSM33693	NEGATIVE	mnp
NRAS Q61R	p.Q61R	COSM584	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM579	NEGATIVE	mnp
NRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM12730	NEGATIVE	mnp
NRAS [Q61H;E62K]	p.[Q61H;E62K]	COSM26494	NEGATIVE	mnp
NTRK1 F589C	p.F589C	OM3255	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT106	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT107	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT108	NEGATIVE	SNV
NTRK1 F589Y	p.F589Y	BT105	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT109	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT110	NEGATIVE	SNV
NTRK1 G667A	p.G667A	OM3301	NEGATIVE	SNV
NTRK1 G667C	p.G667C	BT111	NEGATIVE	SNV
NTRK1 G667S	p.G667S	BT112	NEGATIVE	SNV
NTRK1 V573M	p.V573M	BT104	NEGATIVE	SNV
NTRK2 F633C	p.F633C	OM3278	NEGATIVE	SNV
NTRK2 F633I	p.F633I	OM3276	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3277	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3308	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3309	NEGATIVE	SNV
NTRK2 F633Y	p.F633Y	OM3307	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3279	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NTRK2 G639R	p.G639R	OM3280	NEGATIVE	SNV
NTRK2 G709A	p.G709A	OM3310	NEGATIVE	SNV
NTRK2 Q612E	p.Q612E	OM3272	NEGATIVE	SNV
NTRK2 Q612P	p.Q612P	OM3273	NEGATIVE	SNV
NTRK2 R646K	p.R646K	OM3281	NEGATIVE	SNV
NTRK2 V617D	p.V617D	OM3306	NEGATIVE	SNV
NTRK2 V617G	p.V617G	OM3275	NEGATIVE	SNV
NTRK2 V617M	p.V617M	OM3274	NEGATIVE	mnp
NTRK3 F617L	p.F617L	OM3297	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3298	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3299	NEGATIVE	SNV
NTRK3 F617Y	p.F617Y	OM3311	NEGATIVE	SNV
NTRK3 G623R	p.G623R	COSM6951362	NEGATIVE	SNV
NTRK3 G623R	p.G623R	OM3253	NEGATIVE	SNV
NTRK3 G696A	p.G696A	OM3296	NEGATIVE	SNV
NTRK3 G696C	p.G696C	OM3294	NEGATIVE	mnp
NTRK3 G696C	p.G696C	OM3295	NEGATIVE	mnp
NTRK3 G696E	p.G696E	COSM5850004	NEGATIVE	SNV
NTRK3 V601F	p.V601F	OM3312	NEGATIVE	SNV
NTRK3 V601M	p.V601M	OM3300	NEGATIVE	mnp
PDGFRA A530_I543dup	p.A530_I543dup	COSM7002381	NEGATIVE	ins
PDGFRA D568N	p.D568N	OM3225	NEGATIVE	SNV
PDGFRA D842F	p.D842F	COSM29007	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA D842H	p.D842H	COSM734174	NEGATIVE	SNV
PDGFRA D842I	p.D842I	COSM12398	NEGATIVE	mnp
PDGFRA D842V	p.D842V	COSM736	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12396	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12397	NEGATIVE	mnp
PDGFRA D846Y	p.D846Y	COSM12399	NEGATIVE	SNV
PDGFRA G853D	p.G853D	OM3235	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3228	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3229	NEGATIVE	SNV
PDGFRA H845Y	p.H845Y	COSM96893	NEGATIVE	SNV
PDGFRA N659H	p.N659H	COSM51516	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22414	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22415	NEGATIVE	SNV
PDGFRA N659R	p.N659R	OM3230	NEGATIVE	mnp
PDGFRA N659S	p.N659S	COSM6960333	NEGATIVE	SNV
PDGFRA N659Y	p.N659Y	COSM22416	NEGATIVE	SNV
PDGFRA N848K	p.N848K	COSM28052	NEGATIVE	SNV
PDGFRA P577S	p.P577S	COSM6438084	NEGATIVE	SNV
PDGFRA Q579R	p.Q579R	OM3226	NEGATIVE	SNV
PDGFRA R841K	p.R841K	OM3233	NEGATIVE	SNV
PDGFRA S566_E571delinsR	p.S566_E571delinsR	COSM12418	NEGATIVE	del
PDGFRA V536E	p.V536E	COSM43059	NEGATIVE	SNV
PDGFRA V561A	p.V561A	COSM1430077	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA V561D	p.V561D	COSM739	NEGATIVE	SNV
PDGFRA W559_R560d el	p.W559_R560del	OMINDEL700	NEGATIVE	del
PDGFRA Y555C	p.Y555C	COSM133645	NEGATIVE	SNV
PDGFRA Y849C	p.Y849C	COSM94952	NEGATIVE	SNV
PDGFRA Y849S	p.Y849S	OM3234	NEGATIVE	SNV
PDGFRA exon 18 deletion	p.D842_M844del	COSM96895	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_M844del	COSM12401	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842del	COSM12406	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_D846delinsN	COSM12411	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_I843delinsV	COSM405285	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_H845delinsY	COSM96896	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsRI	OMINDEL900	NEGATIVE	complex
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.I843del	COSM4969655	NEGATIVE	del
PDGFRA exon 18 deletion	p.I843_D846del	COSM12400	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_D846delinsE	COSM12408	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA exon 18 deletion	p.D842_S847delinsEA	COSM12415	NEGATIVE	complex
PDGFRA exon 18 deletion	p.I843_S847delinsT	COSM12407	NEGATIVE	del
PIK3CA A1046E	p.A1046E	COSM27156	NEGATIVE	SNV
PIK3CA A1046T	p.A1046T	COSM27273	NEGATIVE	SNV
PIK3CA A1046V	p.A1046V	COSM6146	NEGATIVE	SNV
PIK3CA C420R	p.C420R	COSM757	NEGATIVE	SNV
PIK3CA D1017H	p.D1017H	COSM27377	NEGATIVE	SNV
PIK3CA D1045G	p.D1045G	COSM1421003	NEGATIVE	SNV
PIK3CA D1045N	p.D1045N	COSM25086	NEGATIVE	SNV
PIK3CA D1045V	p.D1045V	COSM729813	NEGATIVE	SNV
PIK3CA D538G	p.D538G	COSM29123	NEGATIVE	SNV
PIK3CA D538N	p.D538N	COSM21467	NEGATIVE	SNV
PIK3CA D538Y	p.D538Y	COSM1716811	NEGATIVE	SNV
PIK3CA E109_I112delinsD	p.E109_I112delinsD	COSM445995	NEGATIVE	del
PIK3CA E110del	p.E110del	COSM24710	NEGATIVE	del
PIK3CA E39G	p.E39G	COSM1041451	NEGATIVE	SNV
PIK3CA E39K	p.E39K	COSM30625	NEGATIVE	SNV
PIK3CA E39V	p.E39V	COSM1420731	NEGATIVE	SNV
PIK3CA E418K	p.E418K	COSM12582	NEGATIVE	SNV
PIK3CA E542A	p.E542A	COSM1041494	NEGATIVE	SNV
PIK3CA E542G	p.E542G	COSM761	NEGATIVE	SNV
PIK3CA E542K	p.E542K	COSM760	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA E542Q	p.E542Q	COSM17442	NEGATIVE	SNV
PIK3CA E542R	p.E542R	COSM1659152	NEGATIVE	mnp
PIK3CA E542V	p.E542V	COSM762	NEGATIVE	SNV
PIK3CA E545A	p.E545A	COSM12458	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM27374	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM765	NEGATIVE	SNV
PIK3CA E545G	p.E545G	COSM764	NEGATIVE	SNV
PIK3CA E545K	p.E545K	COSM763	NEGATIVE	SNV
PIK3CA E545Q	p.E545Q	COSM27133	NEGATIVE	SNV
PIK3CA E545V	p.E545V	COSM27155	NEGATIVE	SNV
PIK3CA E547D	p.E547D	COSM729819	NEGATIVE	SNV
PIK3CA E547G	p.E547G	COSM1420864	NEGATIVE	SNV
PIK3CA E547K	p.E547K	COSM29315	NEGATIVE	SNV
PIK3CA E81A	p.E81A	COSM5347052	NEGATIVE	SNV
PIK3CA E81K	p.E81K	COSM27502	NEGATIVE	SNV
PIK3CA G1049A	p.G1049A	COSM27158	NEGATIVE	SNV
PIK3CA G1049C	p.G1049C	COSM1168056	NEGATIVE	SNV
PIK3CA G1049D	p.G1049D	COSM308548	NEGATIVE	SNV
PIK3CA G1049R	p.G1049R	COSM12597	NEGATIVE	SNV
PIK3CA G1049S	p.G1049S	COSM777	NEGATIVE	SNV
PIK3CA G106A	p.G106A	COSM166339	NEGATIVE	SNV
PIK3CA G106R	p.G106R	COSM86046	NEGATIVE	SNV
PIK3CA G106V	p.G106V	COSM748	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA G118D	p.G118D	COSM751	NEGATIVE	SNV
PIK3CA H1047L	p.H1047L	COSM776	NEGATIVE	SNV
PIK3CA H1047N	p.H1047N	COSM5029128	NEGATIVE	SNV
PIK3CA H1047P	p.H1047P	COSM249874	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM1041525	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM24714	NEGATIVE	SNV
PIK3CA H1047R	p.H1047R	COSM775	NEGATIVE	SNV
PIK3CA H1047Y	p.H1047Y	COSM774	NEGATIVE	SNV
PIK3CA H1048D	p.H1048D	COSM249881	NEGATIVE	SNV
PIK3CA H1048L	p.H1048L	COSM3734761	NEGATIVE	SNV
PIK3CA H1048N	p.H1048N	COSM1421010	NEGATIVE	SNV
PIK3CA H1048Q	p.H1048Q	COSM27157	NEGATIVE	SNV
PIK3CA H1048R	p.H1048R	COSM36289	NEGATIVE	SNV
PIK3CA H1048Y	p.H1048Y	COSM249875	NEGATIVE	SNV
PIK3CA K111E	p.K111E	COSM13570	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM12580	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM27505	NEGATIVE	SNV
PIK3CA K111R	p.K111R	COSM14052	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM29313	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM773	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM94984	NEGATIVE	SNV
PIK3CA M1043L	p.M1043L	COSM5731063	NEGATIVE	SNV
PIK3CA M1043T	p.M1043T	COSM12463	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA M1043V	p.M1043V	COSM12591	NEGATIVE	SNV
PIK3CA N1044D	p.N1044D	COSM27134	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM12592	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM27504	NEGATIVE	SNV
PIK3CA N1044S	p.N1044S	COSM22541	NEGATIVE	SNV
PIK3CA N1044Y	p.N1044Y	COSM36288	NEGATIVE	SNV
PIK3CA N345D	p.N345D	COSM1420788	NEGATIVE	SNV
PIK3CA N345H	p.N345H	COSM3846786	NEGATIVE	SNV
PIK3CA N345I	p.N345I	COSM94978	NEGATIVE	SNV
PIK3CA N345K	p.N345K	COSM754	NEGATIVE	SNV
PIK3CA N345S	p.N345S	COSM13476	NEGATIVE	SNV
PIK3CA N345T	p.N345T	COSM446002	NEGATIVE	SNV
PIK3CA P104L	p.P104L	COSM163485	NEGATIVE	SNV
PIK3CA P104R	p.P104R	COSM747	NEGATIVE	SNV
PIK3CA P104T	p.P104T	COSM1666660	NEGATIVE	SNV
PIK3CA P539R	p.P539R	COSM759	NEGATIVE	SNV
PIK3CA P539S	p.P539S	COSM249880	NEGATIVE	SNV
PIK3CA P539T	p.P539T	COSM41779	NEGATIVE	SNV
PIK3CA Q546E	p.Q546E	COSM6147	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM24712	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM5881064	NEGATIVE	SNV
PIK3CA Q546K	p.Q546K	COSM766	NEGATIVE	SNV
PIK3CA Q546L	p.Q546L	COSM25041	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Q546P	p.Q546P	COSM767	NEGATIVE	SNV
PIK3CA Q546R	p.Q546R	COSM12459	NEGATIVE	SNV
PIK3CA R108C	p.R108C	COSM6438095	NEGATIVE	SNV
PIK3CA R108H	p.R108H	COSM27497	NEGATIVE	SNV
PIK3CA R108L	p.R108L	COSM729828	NEGATIVE	SNV
PIK3CA R108P	p.R108P	COSM749	NEGATIVE	SNV
PIK3CA R38C	p.R38C	COSM744	NEGATIVE	SNV
PIK3CA R38G	p.R38G	COSM40945	NEGATIVE	SNV
PIK3CA R38H	p.R38H	COSM745	NEGATIVE	SNV
PIK3CA R38L	p.R38L	COSM729833	NEGATIVE	SNV
PIK3CA R38S	p.R38S	COSM87310	NEGATIVE	SNV
PIK3CA T1025A	p.T1025A	COSM771	NEGATIVE	SNV
PIK3CA T1025I	p.T1025I	COSM36285	NEGATIVE	SNV
PIK3CA T1025N	p.T1025N	COSM772	NEGATIVE	SNV
PIK3CA T1025P	p.T1025P	COSM6023925	NEGATIVE	SNV
PIK3CA T1025S	p.T1025S	COSM12590	NEGATIVE	SNV
PIK3CA V344A	p.V344A	COSM86951	NEGATIVE	SNV
PIK3CA V344E	p.V344E	COSM6746438	NEGATIVE	SNV
PIK3CA V344G	p.V344G	COSM22540	NEGATIVE	SNV
PIK3CA V344M	p.V344M	COSM253279	NEGATIVE	SNV
PIK3CA Y1021C	p.Y1021C	COSM12461	NEGATIVE	SNV
PIK3CA Y1021F	p.Y1021F	COSM328025	NEGATIVE	SNV
PIK3CA Y1021H	p.Y1021H	COSM17444	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Y1021N	p.Y1021N	COSM6143	NEGATIVE	SNV
PTEN A126D	p.A126D	COSM28884	NEGATIVE	SNV
PTEN A126G	p.A126G	COSM4391544	NEGATIVE	SNV
PTEN A126P	p.A126P	COSM5211	NEGATIVE	SNV
PTEN A126S	p.A126S	COSM28889	NEGATIVE	SNV
PTEN A126T	p.A126T	COSM5051	NEGATIVE	SNV
PTEN A126V	p.A126V	COSM5041	NEGATIVE	SNV
PTEN C136F	p.C136F	COSM5228	NEGATIVE	SNV
PTEN C136R	p.C136R	COSM5096	NEGATIVE	SNV
PTEN C136Y	p.C136Y	COSM12734	NEGATIVE	SNV
PTEN C71Y	p.C71Y	COSM5102	NEGATIVE	SNV
PTEN D24E	p.D24E	COSM5198	NEGATIVE	SNV
PTEN D24G	p.D24G	COSM5107	NEGATIVE	SNV
PTEN D24H	p.D24H	COSM1603823	NEGATIVE	SNV
PTEN D24N	p.D24N	COSM5079	NEGATIVE	SNV
PTEN D24V	p.D24V	COSM4016448	NEGATIVE	SNV
PTEN D24Y	p.D24Y	COSM5237	NEGATIVE	SNV
PTEN F341V	p.F341V	COSM5255	NEGATIVE	SNV
PTEN G127E	p.G127E	COSM5143	NEGATIVE	SNV
PTEN G129E	p.G129E	COSM28917	NEGATIVE	SNV
PTEN G129R	p.G129R	COSM246853	NEGATIVE	SNV
PTEN G129V	p.G129V	COSM5276	NEGATIVE	SNV
PTEN L181P	p.L181P	MCH17	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN L345Q	p.L345Q	COSM5213	NEGATIVE	SNV
PTEN N48K	p.N48K	COSM4392678	NEGATIVE	SNV
PTEN R130*	p.R130*	COSM5152	NEGATIVE	SNV
PTEN R130G	p.R130G	COSM5219	NEGATIVE	SNV
PTEN R130L	p.R130L	COSM5216	NEGATIVE	SNV
PTEN R130P	p.R130P	COSM5277	NEGATIVE	SNV
PTEN R130Q	p.R130Q	COSM5033	NEGATIVE	SNV
PTEN R173C	p.R173C	COSM5089	NEGATIVE	SNV
PTEN R173H	p.R173H	COSM5039	NEGATIVE	SNV
PTEN R173P	p.R173P	COSM12735	NEGATIVE	SNV
PTEN S170N	p.S170N	COSM5045	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH20	NEGATIVE	SNV
PTEN S170R	p.S170R	COSM6006454	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH21	NEGATIVE	SNV
PTEN T131I	p.T131I	COSM5104	NEGATIVE	SNV
PTEN V133I	p.V133I	COSM5044	NEGATIVE	SNV
PTEN V343E	p.V343E	MCH18	NEGATIVE	SNV
PTEN Y155C	p.Y155C	COSM5144	NEGATIVE	SNV
PTEN Y16C	p.Y16C	COSM5133	NEGATIVE	SNV
PTEN Y174C	p.Y174C	COSM5575601	NEGATIVE	SNV
PTEN Y174N	p.Y174N	COSM5221	NEGATIVE	SNV
RAF1 S257L	p.S257L	COSM181063	NEGATIVE	SNV
RAF1 S257P	p.S257P	ClinVar40600	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RAF1 S257W	p.S257W	COSM581519	NEGATIVE	SNV
RAF1 S259A	p.S259A	COSM20741	NEGATIVE	SNV
RAF1 S259C	p.S259C	COSM5719965	NEGATIVE	SNV
RAF1 S259F	p.S259F	COSM1536534	NEGATIVE	SNV
RAF1 S259P	p.S259P	COSM1418555	NEGATIVE	SNV
RAF1 S259T	p.S259T	ClinVar40601	NEGATIVE	SNV
RET A883F	p.A883F	COSM977	NEGATIVE	mnp
RET A883S	p.A883S	COSM133167	NEGATIVE	SNV
RET A883T	p.A883T	COSM100081	NEGATIVE	SNV
RET A883V	p.A883V	COSM4594153	NEGATIVE	SNV
RET A883Y	p.A883Y	OM3415	NEGATIVE	mnp
RET C618F	p.C618F	OM3401	NEGATIVE	SNV
RET C618G	p.C618G	COSM9214400	NEGATIVE	SNV
RET C618R	p.C618R	COSM29803	NEGATIVE	SNV
RET C618S	p.C618S	COSM87267	NEGATIVE	SNV
RET C618S	p.C618S	OM3402	NEGATIVE	SNV
RET C618T	p.C618T	OM3400	NEGATIVE	mnp
RET C618W	p.C618W	OM3403	NEGATIVE	SNV
RET C618Y	p.C618Y	COSM980	NEGATIVE	SNV
RET C620F	p.C620F	OM3405	NEGATIVE	SNV
RET C620G	p.C620G	OM3404	NEGATIVE	SNV
RET C620R	p.C620R	COSM29804	NEGATIVE	SNV
RET C620S	p.C620S	COSM5946160	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET C620S	p.C620S	COSM29805	NEGATIVE	SNV
RET C620W	p.C620W	COSM249815	NEGATIVE	SNV
RET C620Y	p.C620Y	COSM7403807	NEGATIVE	SNV
RET C630F	p.C630F	OM3406	NEGATIVE	SNV
RET C630G	p.C630G	COSM29806	NEGATIVE	SNV
RET C630R	p.C630R	COSM964	NEGATIVE	SNV
RET C630Y	p.C630Y	COSM9233214	NEGATIVE	SNV
RET C634F	p.C634F	COSM1237919	NEGATIVE	SNV
RET C634G	p.C634G	COSM1738369	NEGATIVE	SNV
RET C634L	p.C634L	OM3408	NEGATIVE	mnp
RET C634R	p.C634R	COSM966	NEGATIVE	SNV
RET C634S	p.C634S	COSM1237918	NEGATIVE	SNV
RET C634S	p.C634S	COSM1666664	NEGATIVE	SNV
RET C634W	p.C634W	COSM975	NEGATIVE	SNV
RET C634Y	p.C634Y	COSM974	NEGATIVE	SNV
RET C634_R635dup	p.C634_R635dup	OMINDEL1177	NEGATIVE	ins
RET C634_R635insHELC	p.C634_R635insHELC	OMINDEL1174	NEGATIVE	ins
RET C634_T636dup	p.C634_T636dup	COSM9213763	NEGATIVE	ins
RET D631Y	p.D631Y	COSM1738370	NEGATIVE	SNV
RET D631_C634del	p.D631_C634del	OMINDEL1172	NEGATIVE	del
RET D631_L633delinsE	p.D631_L633delinsE	COSM983	NEGATIVE	del
RET D631_L633delinsV	p.D631_L633delinsV	OMINDEL1175	NEGATIVE	del
RET D631del	p.D631del	OMINDEL1173	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET D898_E901del	p.D898_E901del	COSM962	NEGATIVE	del
RET E632K	p.E632K	COSM1702268	NEGATIVE	SNV
RET E632_C634delinsG	p.E632_C634delinsG	COSM973	NEGATIVE	del
RET E632_C634dup	p.E632_C634dup	OMINDEL1176	NEGATIVE	ins
RET E632_L633del	p.E632_L633del	COSM968	NEGATIVE	del
RET E632_L633delinsV	p.E632_L633delinsV	COSM982	NEGATIVE	del
RET E632_T636delinsSS	p.E632_T636delinsSS	COSM1048	NEGATIVE	complex
RET E768D	p.E768D	COSM21338	NEGATIVE	SNV
RET E768D	p.E768D	OM3413	NEGATIVE	SNV
RET E768G	p.E768G	COSM1347811	NEGATIVE	SNV
RET E768Q	p.E768Q	COSM1716312	NEGATIVE	SNV
RET E884K	p.E884K	COSM960	NEGATIVE	SNV
RET E884V	p.E884V	COSM1570338	NEGATIVE	SNV
RET G736A	p.G736A	OM3411	NEGATIVE	SNV
RET G810A	p.G810A	OM3290	NEGATIVE	SNV
RET G810C	p.G810C	OM3557	NEGATIVE	SNV
RET G810R	p.G810R	OM3289	NEGATIVE	SNV
RET G810S	p.G810S	OM3288	NEGATIVE	SNV
RET G810V	p.G810V	OM3558	NEGATIVE	SNV
RET G911D	p.G911D	COSM20888	NEGATIVE	SNV
RET I788N	p.I788N	OM3284	NEGATIVE	SNV
RET L730Q	p.L730Q	OM3409	NEGATIVE	SNV
RET L730R	p.L730R	OM3410	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET L760Q	p.L760Q	OM3412	NEGATIVE	SNV
RET L790F	p.L790F	rs75030001	NEGATIVE	SNV
RET L881V	p.L881V	OM3414	NEGATIVE	SNV
RET P628_L633del	p.P628_L633del	OMINDEL1171	NEGATIVE	del
RET R770Q	p.R770Q	COSM6006462	NEGATIVE	SNV
RET R886W	p.R886W	COSM6942691	NEGATIVE	SNV
RET R897*	p.R897*	COSM6983129	NEGATIVE	SNV
RET R912L	p.R912L	COSM188545	NEGATIVE	SNV
RET R912P	p.R912P	OM3419	NEGATIVE	SNV
RET R912Q	p.R912Q	COSM3437793	NEGATIVE	SNV
RET R912W	p.R912W	COSM3415038	NEGATIVE	SNV
RET S891A	p.S891A	rs75234356	NEGATIVE	SNV
RET S904C	p.S904C	OM3418	NEGATIVE	SNV
RET S904F	p.S904F	COSM6438204	NEGATIVE	SNV
RET V804G	p.V804G	OM3554	NEGATIVE	SNV
RET V804L	p.V804L	OM3285	NEGATIVE	SNV
RET V804L	p.V804L	OM3286	NEGATIVE	SNV
RET V804M	p.V804M	OM3195	NEGATIVE	SNV
RET V899_D903del	p.V899_D903del	COSM5991595	NEGATIVE	del
RET Y806C	p.Y806C	OM3287	NEGATIVE	SNV
RET Y806F	p.Y806F	OM3556	NEGATIVE	SNV
RET [D903E;S904P]	p.[D903E;S904P]	OM3416	NEGATIVE	mnp
RET [D903E;S904P]	p.[D903E;S904P]	OM3417	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET [E632D;L633V;C634R]	p.[E632D;L633V;C634R]	OM3407	NEGATIVE	mnp
RET [V882=;A883F]	p.[V882=;A883F]	COSM981	NEGATIVE	mnp
RET c.1880_1899delinsCTCAC	p.?	OMINDEL1170	NEGATIVE	complex
ROS1 D2033N	p.D2033N	COSM6924065	NEGATIVE	SNV
ROS1 G2032K	p.G2032K	OM3533	NEGATIVE	mnp
ROS1 G2032R	p.G2032R	MAN10	NEGATIVE	SNV
ROS1 G2032R	p.G2032R	MAN11	NEGATIVE	SNV
ROS1 L1951M	p.L1951M	COSM1072523	NEGATIVE	SNV
ROS1 L1951R	p.L1951R	OM3264	NEGATIVE	SNV
ROS1 L2026M	p.L2026M	BT151	NEGATIVE	SNV
ROS1 S1986F	p.S1986F	COSM6929870	NEGATIVE	SNV
ROS1 S1986Y	p.S1986Y	OM3170	NEGATIVE	SNV
STK11 A205T	p.A205T	COSM20953	NEGATIVE	SNV
STK11 D176N	p.D176N	COSM4827690	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3710	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3711	NEGATIVE	SNV
STK11 D194H	p.D194H	COSM5325765	NEGATIVE	SNV
STK11 D194N	p.D194N	COSM25847	NEGATIVE	SNV
STK11 D194V	p.D194V	COSM20957	NEGATIVE	SNV
STK11 D194Y	p.D194Y	COSM20944	NEGATIVE	SNV
STK11 D53Gfs*110	p.D53GfsTer110	COSM391251	NEGATIVE	ins
STK11 L290P	p.L290P	OM3719	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 L67P	p.L67P	OM3700	NEGATIVE	SNV
STK11 R304W	p.R304W	COSM29468	NEGATIVE	SNV
STK11 S240W	p.S240W	OM3714	NEGATIVE	SNV
STK11 c.1108+1G>A	p.?	OM3722	NEGATIVE	SNV
STK11 c.289_290+1delinsGAT	p.?	COSM6922318	NEGATIVE	mnp
STK11 c.289_290+2delAAGT	p.?	OMINDEL1000	NEGATIVE	del
STK11 c.290+1G>A	p.?	COSM6350392	NEGATIVE	SNV
STK11 c.290+1G>C	p.?	OM3701	NEGATIVE	SNV
STK11 c.290+1G>T	p.?	OM3702	NEGATIVE	SNV
STK11 c.290+1_290+2delGT	p.?	OMINDEL1001	NEGATIVE	del
STK11 c.290+2T>A	p.?	COSM6925006	NEGATIVE	SNV
STK11 c.290+2T>G	p.?	COSM25857	NEGATIVE	SNV
STK11 c.290+2delT	p.?	OMINDEL1002	NEGATIVE	del
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.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.375-1C>G	p.?	COSM4074282	NEGATIVE	SNV
STK11 c.375-1C>T	p.?	COSM6977173	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.375-2A>C	p.?	OM3706	NEGATIVE	SNV
STK11 c.375-2A>G	p.?	OM3707	NEGATIVE	SNV
STK11 c.375-2A>T	p.?	COSM6967966	NEGATIVE	SNV
STK11 c.375-2_386delACGTATATGGTGAT	p.?	OMINDEL1006	NEGATIVE	del
STK11 c.375-6_375-2delCCCCA	p.?	OMINDEL1005	NEGATIVE	del
STK11 c.465-1G>A	p.?	COSM25855	NEGATIVE	SNV
STK11 c.465-1G>C	p.?	OM3709	NEGATIVE	SNV
STK11 c.465-1G>T	p.?	COSM21570	NEGATIVE	SNV
STK11 c.465-1_465delinsTT	p.?	COSM6928792	NEGATIVE	mnp
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.465-2_465-1delAG	p.?	OMINDEL1009	NEGATIVE	del
STK11 c.465-2_465-1delinsTC	p.?	OM3708	NEGATIVE	mnp
STK11 c.465-4_472delGCAGGTACTTCT	p.?	OMINDEL1007	NEGATIVE	del
STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT	p.?	OMINDEL1008	NEGATIVE	del
STK11 c.598-1G>A	p.?	COSM3724562	NEGATIVE	SNV
STK11 c.598-1G>T	p.?	COSM51521	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.598-1_598delinsTT	p.?	OM3713	NEGATIVE	mnp
STK11 c.598-1_612delGGCACTGCACCCGTTC	p.?	OMINDEL1014	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.598-2_600delAGGCA	p.?	OMINDEL1012	NEGATIVE	del
STK11 c.727_734+9delGTCACCCTGTAAGTGCC	p.?	OMINDEL1015	NEGATIVE	del
STK11 c.731_734+9delCCCTGTAAGTGCC	p.?	OMINDEL1016	NEGATIVE	del
STK11 c.734+1G>A	p.?	COSM6916684	NEGATIVE	SNV
STK11 c.734+1G>C	p.?	COSM6970016	NEGATIVE	SNV
STK11 c.734+1G>T	p.?	COSM51523	NEGATIVE	SNV
STK11 c.734+2T>A	p.?	OM3715	NEGATIVE	SNV
STK11 c.734+2T>G	p.?	COSM6961577	NEGATIVE	SNV
STK11 c.734+2_734+10delTAAGTGCCC	p.?	OMINDEL1017	NEGATIVE	del
STK11 c.735-1G>A	p.?	COSM5732040	NEGATIVE	SNV
STK11 c.735-1G>T	p.?	COSM51522	NEGATIVE	SNV
STK11 c.735-2A>G	p.?	OM3716	NEGATIVE	SNV
STK11 c.735-2A>T	p.?	COSM25856	NEGATIVE	SNV
STK11 c.850_862+4delGACCTG	p.?	OMINDEL1018	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTGAAAGGTGG				
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+2T>A	p.?	COSM5576087	NEGATIVE	SNV
STK11 c.862+2T>G	p.?	OM3718	NEGATIVE	SNV
STK11 c.862+3delG	p.?	COSM8554973	NEGATIVE	del
STK11 c.863-1G>A	p.?	COSM6916725	NEGATIVE	SNV
STK11 c.863-1G>T	p.?	COSM6925330	NEGATIVE	SNV
STK11 c.863-2A>C	p.?	COSM6912582	NEGATIVE	SNV
STK11 c.863-2A>T	p.?	COSM401786	NEGATIVE	SNV
STK11 c.918_920+11delCAGGT GAGCGGCCC	p.?	OMINDEL1019	NEGATIVE	del
STK11 c.920+1G>A	p.?	OM3720	NEGATIVE	SNV
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
TP53 A138V	p.A138V	COSM43818	NEGATIVE	SNV
TP53 C135F	p.C135F	COSM10647	NEGATIVE	SNV
TP53 C135G	p.C135G	COSM44829	NEGATIVE	SNV
TP53 C135R	p.C135R	COSM10684	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44643	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44910	NEGATIVE	SNV
TP53 C135W	p.C135W	COSM44219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C135Y	p.C135Y	COSM10801	NEGATIVE	SNV
TP53 C141F	p.C141F	COSM44911	NEGATIVE	SNV
TP53 C141G	p.C141G	COSM45794	NEGATIVE	SNV
TP53 C141R	p.C141R	COSM43901	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM293910	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM44564	NEGATIVE	SNV
TP53 C141W	p.C141W	COSM44204	NEGATIVE	SNV
TP53 C141Y	p.C141Y	COSM43708	NEGATIVE	SNV
TP53 C176F	p.C176F	COSM10645	NEGATIVE	SNV
TP53 C176G	p.C176G	COSM44692	NEGATIVE	SNV
TP53 C176R	p.C176R	COSM44948	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44645	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44146	NEGATIVE	SNV
TP53 C176W	p.C176W	COSM11114	NEGATIVE	SNV
TP53 C176Y	p.C176Y	COSM10687	NEGATIVE	SNV
TP53 C238F	p.C238F	COSM43778	NEGATIVE	SNV
TP53 C238G	p.C238G	COSM46336	NEGATIVE	SNV
TP53 C238R	p.C238R	COSM44321	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM44653	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM43700	NEGATIVE	SNV
TP53 C238W	p.C238W	COSM44676	NEGATIVE	SNV
TP53 C238Y	p.C238Y	COSM11059	NEGATIVE	SNV
TP53 C242F	p.C242F	COSM10810	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C242G	p.C242G	COSM44135	NEGATIVE	SNV
TP53 C242R	p.C242R	COSM11738	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM11133	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM44935	NEGATIVE	SNV
TP53 C242W	p.C242W	COSM11356	NEGATIVE	SNV
TP53 C242Y	p.C242Y	COSM10646	NEGATIVE	SNV
TP53 C275F	p.C275F	COSM10701	NEGATIVE	SNV
TP53 C275G	p.C275G	COSM11501	NEGATIVE	SNV
TP53 C275R	p.C275R	COSM43902	NEGATIVE	SNV
TP53 C275S	p.C275S	COSM45413	NEGATIVE	SNV
TP53 C275W	p.C275W	COSM43823	NEGATIVE	SNV
TP53 C275Y	p.C275Y	COSM10893	NEGATIVE	SNV
TP53 C277F	p.C277F	COSM10749	NEGATIVE	SNV
TP53 D281A	p.D281A	COSM11665	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43837	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43906	NEGATIVE	SNV
TP53 D281G	p.D281G	COSM11232	NEGATIVE	SNV
TP53 D281H	p.D281H	COSM10943	NEGATIVE	SNV
TP53 D281N	p.D281N	COSM43596	NEGATIVE	SNV
TP53 D281V	p.D281V	COSM45729	NEGATIVE	SNV
TP53 D281Y	p.D281Y	COSM11516	NEGATIVE	SNV
TP53 E258A	p.E258A	COSM44719	NEGATIVE	SNV
TP53 E258D	p.E258D	COSM44962	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 E258D	p.E258D	COSM46410	NEGATIVE	SNV
TP53 E258G	p.E258G	COSM44168	NEGATIVE	SNV
TP53 E258K	p.E258K	COSM10988	NEGATIVE	SNV
TP53 E258Q	p.E258Q	COSM10751	NEGATIVE	SNV
TP53 E258V	p.E258V	COSM44450	NEGATIVE	SNV
TP53 E285A	p.E285A	COSM43614	NEGATIVE	SNV
TP53 E285G	p.E285G	COSM45649	NEGATIVE	SNV
TP53 E285K	p.E285K	COSM10722	NEGATIVE	SNV
TP53 E285Q	p.E285Q	COSM45138	NEGATIVE	SNV
TP53 E285V	p.E285V	COSM44227	NEGATIVE	SNV
TP53 E286A	p.E286A	COSM45892	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45439	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45492	NEGATIVE	SNV
TP53 E286G	p.E286G	COSM43565	NEGATIVE	SNV
TP53 E286K	p.E286K	COSM10726	NEGATIVE	SNV
TP53 E286Q	p.E286Q	COSM44250	NEGATIVE	SNV
TP53 E286V	p.E286V	COSM43936	NEGATIVE	SNV
TP53 G154V	p.G154V	COSM6815	NEGATIVE	SNV
TP53 G244A	p.G244A	COSM12013	NEGATIVE	SNV
TP53 G244C	p.G244C	COSM11524	NEGATIVE	SNV
TP53 G244D	p.G244D	COSM10883	NEGATIVE	SNV
TP53 G244R	p.G244R	COSM44221	NEGATIVE	SNV
TP53 G244S	p.G244S	COSM10941	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 G244V	p.G244V	COSM43652	NEGATIVE	SNV
TP53 G245A	p.G245A	COSM43965	NEGATIVE	SNV
TP53 G245C	p.G245C	COSM11081	NEGATIVE	SNV
TP53 G245D	p.G245D	COSM43606	NEGATIVE	SNV
TP53 G245R	p.G245R	COSM10957	NEGATIVE	SNV
TP53 G245S	p.G245S	COSM6932	NEGATIVE	SNV
TP53 G245V	p.G245V	COSM11196	NEGATIVE	SNV
TP53 G262V	p.G262V	COSM11198	NEGATIVE	SNV
TP53 G266A	p.G266A	COSM45488	NEGATIVE	SNV
TP53 G266E	p.G266E	COSM10867	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM10794	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM11205	NEGATIVE	SNV
TP53 G266V	p.G266V	COSM10958	NEGATIVE	SNV
TP53 G279E	p.G279E	COSM43714	NEGATIVE	SNV
TP53 H179D	p.H179D	COSM44776	NEGATIVE	SNV
TP53 H179L	p.H179L	COSM43635	NEGATIVE	SNV
TP53 H179N	p.H179N	COSM44151	NEGATIVE	SNV
TP53 H179P	p.H179P	COSM44218	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM11249	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM44214	NEGATIVE	SNV
TP53 H179R	p.H179R	COSM10889	NEGATIVE	SNV
TP53 H179Y	p.H179Y	COSM10768	NEGATIVE	SNV
TP53 H193D	p.H193D	COSM44002	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 H193L	p.H193L	COSM11066	NEGATIVE	SNV
TP53 H193N	p.H193N	COSM43935	NEGATIVE	SNV
TP53 H193P	p.H193P	COSM43833	NEGATIVE	SNV
TP53 H193R	p.H193R	COSM10742	NEGATIVE	SNV
TP53 H193Y	p.H193Y	COSM10672	NEGATIVE	SNV
TP53 H214D	p.H214D	COSM45115	NEGATIVE	SNV
TP53 H214L	p.H214L	COSM5632535	NEGATIVE	SNV
TP53 H214P	p.H214P	COSM1640841	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM44407	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM46272	NEGATIVE	SNV
TP53 H214R	p.H214R	COSM43687	NEGATIVE	SNV
TP53 H214Y	p.H214Y	COSM44112	NEGATIVE	SNV
TP53 I195F	p.I195F	COSM44633	NEGATIVE	SNV
TP53 I195L	p.I195L	COSM45773	NEGATIVE	SNV
TP53 I195M	p.I195M	COSM562637	NEGATIVE	SNV
TP53 I195N	p.I195N	COSM44877	NEGATIVE	SNV
TP53 I195S	p.I195S	COSM44539	NEGATIVE	SNV
TP53 I195T	p.I195T	COSM11089	NEGATIVE	SNV
TP53 K132E	p.K132E	COSM10813	NEGATIVE	SNV
TP53 K132M	p.K132M	COSM43592	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM10991	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM43963	NEGATIVE	SNV
TP53 K132Q	p.K132Q	COSM11224	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 K132R	p.K132R	COSM11582	NEGATIVE	SNV
TP53 K132T	p.K132T	COSM43912	NEGATIVE	SNV
TP53 L145P	p.L145P	COSM43899	NEGATIVE	SNV
TP53 L194F	p.L194F	COSM10995	NEGATIVE	SNV
TP53 L194H	p.L194H	COSM43623	NEGATIVE	SNV
TP53 L194I	p.L194I	COSM45330	NEGATIVE	SNV
TP53 L194P	p.L194P	COSM43827	NEGATIVE	SNV
TP53 L194R	p.L194R	COSM44571	NEGATIVE	SNV
TP53 L194V	p.L194V	COSM46117	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM10834	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM11063	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM44415	NEGATIVE	SNV
TP53 M237K	p.M237K	COSM43952	NEGATIVE	SNV
TP53 M237L	p.M237L	COSM44965	NEGATIVE	SNV
TP53 M237R	p.M237R	COSM45032	NEGATIVE	SNV
TP53 M237T	p.M237T	COSM45329	NEGATIVE	SNV
TP53 M237V	p.M237V	COSM44525	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM10757	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM44310	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM46136	NEGATIVE	SNV
TP53 M246K	p.M246K	COSM44103	NEGATIVE	SNV
TP53 M246L	p.M246L	COSM45992	NEGATIVE	SNV
TP53 M246R	p.M246R	COSM11376	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 M246T	p.M246T	COSM11355	NEGATIVE	SNV
TP53 M246V	p.M246V	COSM43555	NEGATIVE	SNV
TP53 N239D	p.N239D	COSM10777	NEGATIVE	SNV
TP53 N239I	p.N239I	COSM307333	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44510	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44647	NEGATIVE	SNV
TP53 N239S	p.N239S	COSM44094	NEGATIVE	SNV
TP53 N239T	p.N239T	COSM43801	NEGATIVE	SNV
TP53 N239Y	p.N239Y	COSM45870	NEGATIVE	SNV
TP53 P151A	p.P151A	COSM44944	NEGATIVE	SNV
TP53 P151H	p.P151H	COSM11476	NEGATIVE	SNV
TP53 P151L	p.P151L	COSM44288	NEGATIVE	SNV
TP53 P151R	p.P151R	COSM44003	NEGATIVE	SNV
TP53 P151S	p.P151S	COSM10905	NEGATIVE	SNV
TP53 P151T	p.P151T	COSM43911	NEGATIVE	SNV
TP53 P152A	p.P152A	COSM44788	NEGATIVE	SNV
TP53 P152L	p.P152L	COSM10790	NEGATIVE	SNV
TP53 P152Q	p.P152Q	COSM44613	NEGATIVE	SNV
TP53 P152R	p.P152R	COSM45505	NEGATIVE	SNV
TP53 P152S	p.P152S	COSM43582	NEGATIVE	SNV
TP53 P152T	p.P152T	COSM44561	NEGATIVE	SNV
TP53 P177_C182del	p.P177_C182del	COSM43570	NEGATIVE	del
TP53 P190L	p.P190L	COSM43657	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 P191del	p.P191del	COSM45140	NEGATIVE	del
TP53 P250L	p.P250L	COSM10771	NEGATIVE	SNV
TP53 P278A	p.P278A	COSM10814	NEGATIVE	SNV
TP53 P278H	p.P278H	COSM43755	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 Q192*	p.Q192*	COSM10733	NEGATIVE	SNV
TP53 R156P	p.R156P	COSM10760	NEGATIVE	SNV
TP53 R175C	p.R175C	COSM43680	NEGATIVE	SNV
TP53 R175G	p.R175G	COSM10870	NEGATIVE	SNV
TP53 R175H	p.R175H	COSM10648	NEGATIVE	SNV
TP53 R175L	p.R175L	COSM10718	NEGATIVE	SNV
TP53 R175P	p.R175P	COSM45416	NEGATIVE	SNV
TP53 R175S	p.R175S	COSM43931	NEGATIVE	SNV
TP53 R196*	p.R196*	COSM10705	NEGATIVE	SNV
TP53 R213*	p.R213*	COSM10654	NEGATIVE	SNV
TP53 R213G	p.R213G	COSM44102	NEGATIVE	SNV
TP53 R213L	p.R213L	COSM43650	NEGATIVE	SNV
TP53 R213P	p.R213P	COSM11860	NEGATIVE	SNV
TP53 R213Q	p.R213Q	COSM10735	NEGATIVE	SNV
TP53 R248G	p.R248G	COSM11564	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R248L	p.R248L	COSM6549	NEGATIVE	SNV
TP53 R248P	p.R248P	COSM11491	NEGATIVE	SNV
TP53 R248Q	p.R248Q	COSM10662	NEGATIVE	SNV
TP53 R248W	p.R248W	COSM10656	NEGATIVE	SNV
TP53 R249G	p.R249G	COSM10668	NEGATIVE	SNV
TP53 R249K	p.R249K	COSM44091	NEGATIVE	SNV
TP53 R249M	p.R249M	COSM43871	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10785	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10817	NEGATIVE	SNV
TP53 R249T	p.R249T	COSM43665	NEGATIVE	SNV
TP53 R249W	p.R249W	COSM43629	NEGATIVE	SNV
TP53 R273C	p.R273C	COSM10659	NEGATIVE	SNV
TP53 R273G	p.R273G	COSM43843	NEGATIVE	SNV
TP53 R273H	p.R273H	COSM10660	NEGATIVE	SNV
TP53 R273L	p.R273L	COSM10779	NEGATIVE	SNV
TP53 R273P	p.R273P	COSM43896	NEGATIVE	SNV
TP53 R273S	p.R273S	COSM43909	NEGATIVE	SNV
TP53 R280G	p.R280G	COSM11123	NEGATIVE	SNV
TP53 R280I	p.R280I	COSM11287	NEGATIVE	SNV
TP53 R280K	p.R280K	COSM10728	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44171	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44233	NEGATIVE	SNV
TP53 R280T	p.R280T	COSM10724	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R282G	p.R282G	COSM10992	NEGATIVE	SNV
TP53 R282L	p.R282L	COSM44470	NEGATIVE	SNV
TP53 R282P	p.R282P	COSM44306	NEGATIVE	SNV
TP53 R282Q	p.R282Q	COSM44338	NEGATIVE	SNV
TP53 R282W	p.R282W	COSM10704	NEGATIVE	SNV
TP53 R283P	p.R283P	COSM10743	NEGATIVE	SNV
TP53 S240G	p.S240G	COSM43973	NEGATIVE	SNV
TP53 S241A	p.S241A	COSM44224	NEGATIVE	SNV
TP53 S241C	p.S241C	COSM10709	NEGATIVE	SNV
TP53 S241F	p.S241F	COSM10812	NEGATIVE	SNV
TP53 S241P	p.S241P	COSM44578	NEGATIVE	SNV
TP53 S241T	p.S241T	COSM44067	NEGATIVE	SNV
TP53 S241Y	p.S241Y	COSM10935	NEGATIVE	SNV
TP53 V157F	p.V157F	COSM10670	NEGATIVE	SNV
TP53 V157L	p.V157L	COSM45120	NEGATIVE	SNV
TP53 V172F	p.V172F	COSM44240	NEGATIVE	SNV
TP53 V173A	p.V173A	COSM44327	NEGATIVE	SNV
TP53 V173E	p.V173E	COSM45731	NEGATIVE	SNV
TP53 V173G	p.V173G	COSM44383	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM43559	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM44057	NEGATIVE	SNV
TP53 V173M	p.V173M	COSM11084	NEGATIVE	SNV
TP53 V197M	p.V197M	COSM43779	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 V216A	p.V216A	COSM44567	NEGATIVE	SNV
TP53 V216E	p.V216E	COSM44274	NEGATIVE	SNV
TP53 V216G	p.V216G	COSM43681	NEGATIVE	SNV
TP53 V216L	p.V216L	COSM11210	NEGATIVE	SNV
TP53 V216M	p.V216M	COSM10667	NEGATIVE	SNV
TP53 V272A	p.V272A	COSM44294	NEGATIVE	SNV
TP53 V272E	p.V272E	COSM44580	NEGATIVE	SNV
TP53 V272G	p.V272G	COSM44870	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM10859	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM45898	NEGATIVE	SNV
TP53 V272M	p.V272M	COSM10891	NEGATIVE	SNV
TP53 V274F	p.V274F	COSM10769	NEGATIVE	SNV
TP53 V274L	p.V274L	COSM44443	NEGATIVE	SNV
TP53 Y220C	p.Y220C	COSM10758	NEGATIVE	SNV
TP53 Y220D	p.Y220D	COSM11847	NEGATIVE	SNV
TP53 Y220F	p.Y220F	COSM1564197	NEGATIVE	SNV
TP53 Y220H	p.Y220H	COSM44637	NEGATIVE	SNV
TP53 Y220N	p.Y220N	COSM44672	NEGATIVE	SNV
TP53 Y220S	p.Y220S	COSM43850	NEGATIVE	SNV
TP53 Y234C	p.Y234C	COSM10725	NEGATIVE	SNV
TP53 Y234D	p.Y234D	COSM43768	NEGATIVE	SNV
TP53 Y234F	p.Y234F	COSM44315	NEGATIVE	SNV
TP53 Y234H	p.Y234H	COSM11152	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 Y234N	p.Y234N	COSM43956	NEGATIVE	SNV
TP53 Y234S	p.Y234S	COSM43865	NEGATIVE	SNV
TP53 Y236C	p.Y236C	COSM10731	NEGATIVE	SNV
TP53 Y236D	p.Y236D	COSM43602	NEGATIVE	SNV
TP53 Y236F	p.Y236F	COSM3958809	NEGATIVE	SNV
TP53 Y236H	p.Y236H	COSM44326	NEGATIVE	SNV
TP53 Y236N	p.Y236N	COSM43826	NEGATIVE	SNV
TP53 Y236S	p.Y236S	COSM44693	NEGATIVE	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
A2M-ALK fusion	A2M(22) - ALK(19)	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
AGAP1-ALK fusion	AGAP1(6) - ALK(18)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATIC-ALK fusion	ATIC(7) - ALK(20)	Fusion	NEGATIVE
ATRNL1-ALK fusion	ATRNL1(19) - ALK(20)	Fusion	NEGATIVE
BCL11A-ALK fusion	BCL11A(4) - ALK(20)	Fusion	NEGATIVE
BEND5-ALK fusion	BEND5(3) - ALK(20)	Fusion	NEGATIVE
BIRC6-ALK fusion	BIRC6(10) - ALK(20)	Fusion	NEGATIVE
CAD-ALK fusion	CAD(35) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CAD-ALK fusion	CAD(36) - ALK(20)	Fusion	NEGATIVE
CAMKMT-ALK fusion	CAMKMT(3) - ALK(20)	Fusion	NEGATIVE
CARS-ALK fusion	CARS(17) - ALK(20)	Fusion	NEGATIVE
CCDC149-ALK fusion	CCDC149(10) - ALK(20)	Fusion	NEGATIVE
CCDC61-ALK fusion	CCDC61(13) - ALK(20)	Fusion	NEGATIVE
CCDC88A-ALK fusion	CCDC88A(12) - ALK(20)	Fusion	NEGATIVE
CEBPZ-ALK fusion	CEBPZ(2) - ALK(20)	Fusion	NEGATIVE
CENPF-ALK fusion	CENPF(11) - ALK(20)	Fusion	NEGATIVE
CLIP1-ALK fusion	CLIP1(13) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(10) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(14) - ALK(17)	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
COL25A1-ALK fusion	COL25A1(3) - ALK(20)	Fusion	NEGATIVE
CPSF7-ALK fusion	CPSF7(5) - ALK(20)	Fusion	NEGATIVE
CREB1-ALK fusion	CREB1(1) - ALK(9)	Fusion	NEGATIVE
CTNND1-ALK fusion	CTNND1(16) - ALK(20)	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
DDX6-ALK fusion	DDX6(2) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DES-ALK fusion	DES(7) - ALK(19)	Fusion	NEGATIVE
DMD-ALK fusion	DMD(55) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(8) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(6) - ALK(20)	Fusion	NEGATIVE
EIF2AK3-ALK fusion	EIF2AK3(2) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(17) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(2) - 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(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(3) - 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(19)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(18)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(17)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(16)	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

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(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(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(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(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

Variant Name	Genes (Exons)	Variant Type	Test Result
EML4-ALK fusion	EML4(22) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(4) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(5) - ALK(20)	Fusion	NEGATIVE
FBXO36-ALK fusion	FBXO36(2) - 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
GALNT14-ALK fusion	GALNT14(1) - ALK(19)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(13) - ALK(20)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(19) - 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
HNRNPA1-ALK fusion	HNRNPA1(2) - ALK(18)	Fusion	NEGATIVE
IGFBP5-ALK fusion	IGFBP5(1) - ALK(19)	Fusion	NEGATIVE
INTS9-ALK fusion	INTS9(8) - ALK(20)	Fusion	NEGATIVE
ITSN2-ALK fusion	ITSN2(14) - ALK(2)	Fusion	NEGATIVE
KANK2-ALK fusion	KANK2(4) - ALK(16)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KCNQ5-ALK fusion	KCNQ5(1) - ALK(10)	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
KLC1-ALK fusion	KLC1(9) - ALK(20)	Fusion	NEGATIVE
KTN1-ALK fusion	KTN1(43) - ALK(19)	Fusion	NEGATIVE
LMNA-ALK fusion	LMNA(2) - ALK(20)	Fusion	NEGATIVE
LMO7-ALK fusion	LMO7(15) - ALK(20)	Fusion	NEGATIVE
LRPPRC-ALK fusion	LRPPRC(23) - ALK(20)	Fusion	NEGATIVE
LRRFIP2-ALK fusion	LRRFIP2(26) - ALK(20)	Fusion	NEGATIVE
LTBP1-ALK fusion	LTBP1(19) - ALK(20)	Fusion	NEGATIVE
MALAT1-ALK fusion	MALAT1(1) - ALK(16)	Fusion	NEGATIVE
MANBA-ALK fusion	MANBA(14) - ALK(20)	Fusion	NEGATIVE
MAPRE3-ALK fusion	MAPRE3(7) - ALK(20)	Fusion	NEGATIVE
MCFD2-ALK fusion	MCFD2(1) - ALK(20)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(2) - ALK(7)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(1) - ALK(2)	Fusion	NEGATIVE
MLPH-ALK fusion	MLPH(11) - ALK(20)	Fusion	NEGATIVE
MPRIP-ALK fusion	MPRIP(21) - ALK(20)	Fusion	NEGATIVE
MROH2B-ALK fusion	MROH2B(21) - ALK(12)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(9) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MYH10-ALK fusion	MYH10(38) - 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
MYO5C-ALK fusion	MYO5C(30) - ALK(20)	Fusion	NEGATIVE
MYT1L-ALK fusion	MYT1L(14) - ALK(20)	Fusion	NEGATIVE
NCOA1-ALK fusion	NCOA1(21) - ALK(1)	Fusion	NEGATIVE
NPM1-ALK fusion	NPM1(4) - ALK(20)	Fusion	NEGATIVE
NUMA1-ALK fusion	NUMA1(19) - ALK(20)	Fusion	NEGATIVE
OTOF-ALK fusion	OTOF(2) - ALK(3)	Fusion	NEGATIVE
PABPC1-ALK fusion	PABPC1(9) - ALK(20)	Fusion	NEGATIVE
PCM1-ALK fusion	PCM1(23) - ALK(20)	Fusion	NEGATIVE
PDE3A-ALK fusion	PDE3A(10) - ALK(8)	Fusion	NEGATIVE
PDE8B-ALK fusion	PDE8B(11) - ALK(20)	Fusion	NEGATIVE
PHACTR1-ALK fusion	PHACTR1(4) - ALK(20)	Fusion	NEGATIVE
PLEKHA7-ALK fusion	PLEKHA7(26) - ALK(19)	Fusion	NEGATIVE
PNPT1-ALK fusion	PNPT1(5) - ALK(4)	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
PPM1B-ALK fusion	PPM1B(1) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
PPP1R21-ALK fusion	PPP1R21(7) - ALK(20)	Fusion	NEGATIVE
PPP1R21-ALK fusion	PPP1R21(17) - ALK(20)	Fusion	NEGATIVE
PPP4R3B-ALK fusion	PPP4R3B(10) - ALK(2)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(2) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(5) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(10) - ALK(20)	Fusion	NEGATIVE
PRKAR1B-ALK fusion	PRKAR1B(4) - ALK(20)	Fusion	NEGATIVE
PRKAR2A-ALK fusion	PRKAR2A(2) - ALK(20)	Fusion	NEGATIVE
PRKCE-ALK fusion	PRKCE(1) - ALK(20)	Fusion	NEGATIVE
RANBP2-ALK fusion	RANBP2(18) - ALK(20)	Fusion	NEGATIVE
RNF213-ALK fusion	RNF213(20) - ALK(20)	Fusion	NEGATIVE
RRBP1-ALK fusion	RRBP1(20) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(6) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(5) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(22) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(21) - ALK(20)	Fusion	NEGATIVE
SFT2D1-ALK fusion	SFT2D1(1) - ALK(6)	Fusion	NEGATIVE
SLC34A2-ALK fusion	SLC34A2(1) - ALK(20)	Fusion	NEGATIVE
SLC8A1-ALK fusion	SLC8A1(1) - ALK(20)	Fusion	NEGATIVE
SLMAP-ALK fusion	SLMAP(11) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SP100-ALK fusion	SP100(4) - ALK(20)	Fusion	NEGATIVE
SPDYA-ALK fusion	SPDYA(6) - ALK(20)	Fusion	NEGATIVE
SPECC1L-ALK fusion	SPECC1L(9) - 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
SQSTM1-ALK fusion	SQSTM1(5) - ALK(20)	Fusion	NEGATIVE
SQSTM1-ALK fusion	SQSTM1(5) - ALK(19)	Fusion	NEGATIVE
STK32B-ALK fusion	STK32B(11) - ALK(20)	Fusion	NEGATIVE
STRN-ALK fusion	STRN(3) - ALK(20)	Fusion	NEGATIVE
TERT-ALK fusion	TERT(11) - ALK(5)	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(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(5) - ALK(19)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(6) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(19)	Fusion	NEGATIVE
THBS1-ALK fusion	THBS1(4) - ALK(19)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(20)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(19)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(32) - ALK(13)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(28) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(20)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(16) - ALK(17)	Fusion	NEGATIVE
TPM1-ALK fusion	TPM1(8) - ALK(20)	Fusion	NEGATIVE
TPM3-ALK fusion	TPM3(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(15) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(4) - ALK(20)	Fusion	NEGATIVE
TRAF1-ALK fusion	TRAF1(6) - ALK(20)	Fusion	NEGATIVE
TRMT61B-ALK fusion	TRMT61B(1) - ALK(9)	Fusion	NEGATIVE
VCL-ALK fusion	VCL(16) - ALK(20)	Fusion	NEGATIVE
YPEL5-ALK fusion	YPEL5(1) - ALK(18)	Fusion	NEGATIVE
ZNF2-ALK fusion	ZNF2(1) - ALK(20)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(11)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(9)	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
AGK-BRAF fusion	AGK(2) - BRAF(8)	Fusion	NEGATIVE
AGTRAP-BRAF fusion	AGTRAP(5) - BRAF(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
AP3B1-BRAF fusion	AP3B1(22) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(3) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(4) - BRAF(11)	Fusion	NEGATIVE
ATG7-BRAF fusion	ATG7(18) - BRAF(9)	Fusion	NEGATIVE
BAIAP2L1-BRAF fusion	BAIAP2L1(12) - BRAF(9)	Fusion	NEGATIVE
BBS9-BRAF fusion	BBS9(19) - BRAF(4)	Fusion	NEGATIVE
BCAP29-BRAF fusion	BCAP29(7) - BRAF(11)	Fusion	NEGATIVE
BCL2L11-BRAF fusion	BCL2L11(3) - BRAF(10)	Fusion	NEGATIVE
BTF3L4-BRAF fusion	BTF3L4(3) - BRAF(11)	Fusion	NEGATIVE
C7orf73-BRAF fusion	C7orf73(2) - BRAF(9)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(2) - BRAF(8)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(1) - BRAF(9)	Fusion	NEGATIVE
CCDC91-BRAF fusion	CCDC91(11) - BRAF(9)	Fusion	NEGATIVE
CCNY-BRAF fusion	CCNY(1) - BRAF(10)	Fusion	NEGATIVE
CDC27-BRAF fusion	CDC27(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CDC42BPB-BRAF fusion	CDC42BPB(36) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(8)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(12) - BRAF(11)	Fusion	NEGATIVE
CEP89-BRAF fusion	CEP89(16) - BRAF(9)	Fusion	NEGATIVE
CLCN6-BRAF fusion	CLCN6(2) - BRAF(11)	Fusion	NEGATIVE
CLIP2-BRAF fusion	CLIP2(6) - BRAF(11)	Fusion	NEGATIVE
COBL-BRAF fusion	COBL(7) - BRAF(10)	Fusion	NEGATIVE
CUL1-BRAF fusion	CUL1(7) - BRAF(9)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(8) - BRAF(8)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(10) - BRAF(9)	Fusion	NEGATIVE
DGKI-BRAF fusion	DGKI(7) - BRAF(10)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(5) - BRAF(9)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(3) - BRAF(10)	Fusion	NEGATIVE
DOCK4-BRAF fusion	DOCK4(41) - BRAF(9)	Fusion	NEGATIVE
DSTYK-BRAF fusion	DSTYK(1) - BRAF(10)	Fusion	NEGATIVE
DYNC1I2-BRAF fusion	DYNC1I2(7) - BRAF(10)	Fusion	NEGATIVE
EML4-BRAF fusion	EML4(6) - BRAF(10)	Fusion	NEGATIVE
EPS15-BRAF fusion	EPS15(22) - BRAF(10)	Fusion	NEGATIVE
EPS15L1-BRAF fusion	EPS15L1(16) - BRAF(9)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(12) - BRAF(10)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(17) - BRAF(8)	Fusion	NEGATIVE
ESYT2-BRAF fusion	ESYT2(8) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FAM114A2-BRAF fusion	FAM114A2(9) - BRAF(11)	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
FAM73A-BRAF fusion	FAM73A(7) - BRAF(9)	Fusion	NEGATIVE
FCHSD1-BRAF fusion	FCHSD1(13) - BRAF(9)	Fusion	NEGATIVE
FXR1-BRAF fusion	FXR1(13) - BRAF(10)	Fusion	NEGATIVE
GATM-BRAF fusion	GATM(2) - BRAF(11)	Fusion	NEGATIVE
GHR-BRAF fusion	GHR(1) - BRAF(10)	Fusion	NEGATIVE
GIPC2-BRAF fusion	GIPC2(3) - BRAF(10)	Fusion	NEGATIVE
GIT2-BRAF fusion	GIT2(14) - BRAF(9)	Fusion	NEGATIVE
GNAI1-BRAF fusion	GNAI1(1) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(4) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(20) - BRAF(10)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(4) - BRAF(7)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(6) - BRAF(7)	Fusion	NEGATIVE
KANK1-BRAF fusion	KANK1(7) - BRAF(9)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(3) - BRAF(8)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(4) - BRAF(8)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(12) - BRAF(4)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(11) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(18) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(17) - BRAF(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(10)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(8)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(8)	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
KLHL7-BRAF fusion	KLHL7(4) - BRAF(10)	Fusion	NEGATIVE
KLHL7-BRAF fusion	KLHL7(5) - BRAF(9)	Fusion	NEGATIVE
LMO7-BRAF fusion	LMO7(7) - BRAF(9)	Fusion	NEGATIVE
LSM12-BRAF fusion	LSM12(3) - BRAF(9)	Fusion	NEGATIVE
LSM14A-BRAF fusion	LSM14A(9) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(5) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(60) - BRAF(9)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(17) - BRAF(10)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MBP-BRAF fusion	MBP(2) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(11)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(10)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(8)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(3) - BRAF(10)	Fusion	NEGATIVE
MYRIP-BRAF fusion	MYRIP(16) - BRAF(9)	Fusion	NEGATIVE
MZT1-BRAF fusion	MZT1(2) - BRAF(11)	Fusion	NEGATIVE
NFIC-BRAF fusion	NFIC(6) - BRAF(10)	Fusion	NEGATIVE
NRF1-BRAF fusion	NRF1(5) - BRAF(9)	Fusion	NEGATIVE
NUB1-BRAF fusion	NUB1(3) - BRAF(9)	Fusion	NEGATIVE
NUDCD3-BRAF fusion	NUDCD3(4) - BRAF(9)	Fusion	NEGATIVE
NUP214-BRAF fusion	NUP214(21) - BRAF(10)	Fusion	NEGATIVE
OSBPL1A-BRAF fusion	OSBPL1A(16) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(11) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(14) - BRAF(9)	Fusion	NEGATIVE
PACSIN2-BRAF fusion	PACSIN2(7) - BRAF(9)	Fusion	NEGATIVE
PAPSS1-BRAF fusion	PAPSS1(5) - BRAF(9)	Fusion	NEGATIVE
PARP12-BRAF fusion	PARP12(9) - BRAF(11)	Fusion	NEGATIVE
PCM1-BRAF fusion	PCM1(37) - BRAF(9)	Fusion	NEGATIVE
PHTF2-BRAF fusion	PHTF2(17) - BRAF(9)	Fusion	NEGATIVE
PICALM-BRAF fusion	PICALM(19) - BRAF(9)	Fusion	NEGATIVE
PJA2-BRAF fusion	PJA2(7) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PLIN3-BRAF fusion	PLIN3(1) - BRAF(9)	Fusion	NEGATIVE
PLXNA2-BRAF fusion	PLXNA2(9) - BRAF(2)	Fusion	NEGATIVE
POR-BRAF fusion	POR(3) - BRAF(11)	Fusion	NEGATIVE
PPFIBP2-BRAF fusion	PPFIBP2(3) - BRAF(11)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(10) - BRAF(9)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(9) - BRAF(9)	Fusion	NEGATIVE
PRKAR2B-BRAF fusion	PRKAR2B(1) - BRAF(10)	Fusion	NEGATIVE
RAD18-BRAF fusion	RAD18(7) - BRAF(10)	Fusion	NEGATIVE
RBM33-BRAF fusion	RBM33(3) - BRAF(11)	Fusion	NEGATIVE
RBMS3-BRAF fusion	RBMS3(11) - BRAF(11)	Fusion	NEGATIVE
RNF11-BRAF fusion	RNF11(1) - BRAF(11)	Fusion	NEGATIVE
RNF130-BRAF fusion	RNF130(3) - BRAF(9)	Fusion	NEGATIVE
RP2-BRAF fusion	RP2(3) - BRAF(10)	Fusion	NEGATIVE
RUNDC1-BRAF fusion	RUNDC1(1) - BRAF(9)	Fusion	NEGATIVE
SCRIB-BRAF fusion	SCRIB(27) - BRAF(11)	Fusion	NEGATIVE
SLC12A7-BRAF fusion	SLC12A7(17) - BRAF(11)	Fusion	NEGATIVE
SLC45A3-BRAF fusion	SLC45A3(1) - 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(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(9) - BRAF(8)	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(11)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(8)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(6) - BRAF(9)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(5) - BRAF(9)	Fusion	NEGATIVE
STRN3-BRAF fusion	STRN3(3) - BRAF(10)	Fusion	NEGATIVE
STXBP6-BRAF fusion	STXBP6(4) - BRAF(10)	Fusion	NEGATIVE
TANK-BRAF fusion	TANK(4) - BRAF(9)	Fusion	NEGATIVE
TAX1BP1-BRAF fusion	TAX1BP1(8) - BRAF(11)	Fusion	NEGATIVE
TMEM106B-BRAF fusion	TMEM106B(4) - BRAF(8)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(1) - BRAF(10)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(2) - BRAF(9)	Fusion	NEGATIVE
TMPRSS2-BRAF fusion	TMPRSS2(3) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TOM1L2-BRAF fusion	TOM1L2(11) - BRAF(9)	Fusion	NEGATIVE
TPR-BRAF fusion	TPR(14) - BRAF(7)	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
TRIM4-BRAF fusion	TRIM4(6) - BRAF(10)	Fusion	NEGATIVE
UBN2-BRAF fusion	UBN2(3) - BRAF(11)	Fusion	NEGATIVE
ZBTB8A-BRAF fusion	ZBTB8A(4) - BRAF(9)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(7) - BRAF(11)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(3) - BRAF(10)	Fusion	NEGATIVE
ZKSCAN5-BRAF fusion	ZKSCAN5(2) - BRAF(9)	Fusion	NEGATIVE
ZNF207-BRAF fusion	ZNF207(3) - BRAF(10)	Fusion	NEGATIVE
ZNF767P-BRAF fusion	ZNF767P(1) - BRAF(11)	Fusion	NEGATIVE
ZSCAN30-BRAF fusion	ZSCAN30(3) - BRAF(10)	Fusion	NEGATIVE
EGFRvIII	EGFR(1) - EGFR(8)	RNAExonVariant	NEGATIVE
ESR1-ARMT1 fusion	ESR1(3) - ARMT1(4)	Fusion	NEGATIVE
ESR1-ARMT1 fusion	ESR1(6) - ARMT1(2)	Fusion	NEGATIVE
ESR1-ARNT2 fusion	ESR1(7) - ARNT2(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-ATP2B2 fusion	ESR1(4) - ATP2B2(4)	Fusion	NEGATIVE
ESR1-BMPER fusion	ESR1(6) - BMPER(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(2)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(5)	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(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(10)	Fusion	NEGATIVE
ESR1-CDK13 fusion	ESR1(7) - CDK13(13)	Fusion	NEGATIVE
ESR1-COA5 fusion	ESR1(4) - COA5(2)	Fusion	NEGATIVE
ESR1-DAB2 fusion	ESR1(6) - DAB2(3)	Fusion	NEGATIVE
ESR1-GYG2 fusion	ESR1(6) - GYG2(2)	Fusion	NEGATIVE
ESR1-MKL1 fusion	ESR1(6) - MKL1(7)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(6) - MTHFD1L(21)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(7) - MTHFD1L(27)	Fusion	NEGATIVE
ESR1-NKAIN2 fusion	ESR1(6) - NKAIN2(2)	Fusion	NEGATIVE
ESR1-NOP2 fusion	ESR1(6) - NOP2(16)	Fusion	NEGATIVE
ESR1-PCDH11X fusion	ESR1(6) - PCDH11X(6)	Fusion	NEGATIVE
ESR1-PDE10A fusion	ESR1(7) - PDE10A(6)	Fusion	NEGATIVE
ESR1-PLEKHG1 fusion	ESR1(6) - PLEKHG1(12)	Fusion	NEGATIVE
ESR1-POLH fusion	ESR1(6) - POLH(2)	Fusion	NEGATIVE
ESR1-RMND1 fusion	ESR1(6) - RMND1(9)	Fusion	NEGATIVE
ESR1-SOX9 fusion	ESR1(6) - SOX9(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-SYNE1 fusion	ESR1(6) - SYNE1(59)	Fusion	NEGATIVE
ESR1-TFG fusion	ESR1(6) - TFG(4)	Fusion	NEGATIVE
ESR1-TNRC6B fusion	ESR1(6) - TNRC6B(14)	Fusion	NEGATIVE
ESR1-YAP1 fusion	ESR1(6) - YAP1(4)	Fusion	NEGATIVE
PLEKHG1-ESR1 fusion	PLEKHG1(3) - ESR1(2)	Fusion	NEGATIVE
TNS3-ESR1 fusion	TNS3(16) - ESR1(5)	Fusion	NEGATIVE
ACPP-FGFR1 fusion	ACPP(10) - FGFR1(4)	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
BCR-FGFR1 fusion	BCR(4) - FGFR1(10)	Fusion	NEGATIVE
CNTRL-FGFR1 fusion	CNTRL(40) - FGFR1(10)	Fusion	NEGATIVE
CPSF6-FGFR1 fusion	CPSF6(8) - FGFR1(10)	Fusion	NEGATIVE
CUX1-FGFR1 fusion	CUX1(11) - FGFR1(10)	Fusion	NEGATIVE
EGFR-FGFR1 fusion	EGFR(24) - FGFR1(10)	Fusion	NEGATIVE
ERLIN2-FGFR1 fusion	ERLIN2(8) - FGFR1(2)	Fusion	NEGATIVE
ERVK3-1-FGFR1 fusion	ERVK3-1(3) - FGFR1(10)	Fusion	NEGATIVE
FGFR1-ADAM32 fusion	FGFR1(17) - ADAM32(14)	Fusion	NEGATIVE
FGFR1-EVI5 fusion	FGFR1(17) - EVI5(12)	Fusion	NEGATIVE
FGFR1-HOOK3 fusion	FGFR1(17) - HOOK3(5)	Fusion	NEGATIVE
FGFR1-TACC1 fusion	FGFR1(17) - TACC1(7)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(5) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(6) - FGFR1(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR1OP-FGFR1 fusion	FGFR1OP(7) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP2-FGFR1 fusion	FGFR1OP2(4) - 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(4)	Fusion	NEGATIVE
FN1-FGFR1 fusion	FN1(22) - FGFR1(3)	Fusion	NEGATIVE
KAT6A-FGFR1 fusion	KAT6A(5) - FGFR1(2)	Fusion	NEGATIVE
LRRFIP1-FGFR1 fusion	LRRFIP1(8) - FGFR1(10)	Fusion	NEGATIVE
MYO18A-FGFR1 fusion	MYO18A(33) - FGFR1(10)	Fusion	NEGATIVE
RANBP2-FGFR1 fusion	RANBP2(20) - FGFR1(10)	Fusion	NEGATIVE
SNX29-FGFR1 fusion	SNX29(1) - FGFR1(4)	Fusion	NEGATIVE
SQSTM1-FGFR1 fusion	SQSTM1(6) - FGFR1(10)	Fusion	NEGATIVE
TPR-FGFR1 fusion	TPR(22) - FGFR1(10)	Fusion	NEGATIVE
TRIM24-FGFR1 fusion	TRIM24(11) - FGFR1(10)	Fusion	NEGATIVE
WHSC1L1-FGFR1 fusion	WHSC1L1(14) - FGFR1(5)	Fusion	NEGATIVE
ZMYM2-FGFR1 fusion	ZMYM2(17) - FGFR1(10)	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
CD44-FGFR2 fusion	CD44(1) - FGFR2(3)	Fusion	NEGATIVE
COL25A1-FGFR2 fusion	COL25A1(3) - FGFR2(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CTNNB1-FGFR2 fusion	CTNNB1(1) - FGFR2(10)	Fusion	NEGATIVE
FGFR2-ACSL5 fusion	FGFR2(17) - ACSL5(15)	Fusion	NEGATIVE
FGFR2-AFF3 fusion	FGFR2(17) - AFF3(7)	Fusion	NEGATIVE
FGFR2-AHCYL1 fusion	FGFR2(17) - AHCYL1(2)	Fusion	NEGATIVE
FGFR2-ANKRD24 fusion	FGFR2(17) - ANKRD24(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(2)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(10)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(16)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(18)	Fusion	NEGATIVE
FGFR2-CASP7 fusion	FGFR2(17) - CASP7(2)	Fusion	NEGATIVE
FGFR2-CBX5 fusion	FGFR2(17) - CBX5(2)	Fusion	NEGATIVE
FGFR2-CCAR2 fusion	FGFR2(17) - CCAR2(4)	Fusion	NEGATIVE
FGFR2-CCDC186 fusion	FGFR2(17) - CCDC186(4)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(2)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(1)	Fusion	NEGATIVE
FGFR2-CELF2 fusion	FGFR2(17) - CELF2(4)	Fusion	NEGATIVE
FGFR2-CIT fusion	FGFR2(17) - CIT(23)	Fusion	NEGATIVE
FGFR2-CLIP2 fusion	FGFR2(17) - CLIP2(10)	Fusion	NEGATIVE
FGFR2-COL14A1 fusion	FGFR2(17) - COL14A1(34)	Fusion	NEGATIVE
FGFR2-CREB5 fusion	FGFR2(17) - CREB5(8)	Fusion	NEGATIVE
FGFR2-DDX21 fusion	FGFR2(17) - DDX21(11)	Fusion	NEGATIVE
FGFR2-EIF4A2 fusion	FGFR2(17) - EIF4A2(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-FAM76A fusion	FGFR2(17) - FAM76A(2)	Fusion	NEGATIVE
FGFR2-GAB2 fusion	FGFR2(17) - GAB2(2)	Fusion	NEGATIVE
FGFR2-INA fusion	FGFR2(17) - INA(2)	Fusion	NEGATIVE
FGFR2-KCTD1 fusion	FGFR2(17) - KCTD1(2)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(3)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(5)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(6)	Fusion	NEGATIVE
FGFR2-KRT8 fusion	FGFR2(17) - KRT8(6)	Fusion	NEGATIVE
FGFR2-LAMC1 fusion	FGFR2(17) - LAMC1(27)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MGEA5 fusion	FGFR2(17) - MGEA5(12)	Fusion	NEGATIVE
FGFR2-NOL4 fusion	FGFR2(17) - NOL4(7)	Fusion	NEGATIVE
FGFR2-NRAP fusion	FGFR2(17) - NRAP(24)	Fusion	NEGATIVE
FGFR2-NRBF2 fusion	FGFR2(17) - NRBF2(4)	Fusion	NEGATIVE
FGFR2-OFD1 fusion	FGFR2(17) - OFD1(3)	Fusion	NEGATIVE
FGFR2-OGDH fusion	FGFR2(17) - OGDH(3)	Fusion	NEGATIVE
FGFR2-OPTN fusion	FGFR2(17) - OPTN(5)	Fusion	NEGATIVE
FGFR2-PASD1 fusion	FGFR2(17) - PASD1(10)	Fusion	NEGATIVE
FGFR2-PCM1 fusion	FGFR2(17) - PCM1(7)	Fusion	NEGATIVE
FGFR2-PHC1 fusion	FGFR2(17) - PHC1(2)	Fusion	NEGATIVE
FGFR2-PKD2L1 fusion	FGFR2(17) - PKD2L1(7)	Fusion	NEGATIVE
FGFR2-PPHLN1 fusion	FGFR2(17) - PPHLN1(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-RABGAP1L fusion	FGFR2(17) - RABGAP1L(20)	Fusion	NEGATIVE
FGFR2-RASAL2 fusion	FGFR2(17) - RASAL2(3)	Fusion	NEGATIVE
FGFR2-ROCK1 fusion	FGFR2(17) - ROCK1(2)	Fusion	NEGATIVE
FGFR2-SBNO2 fusion	FGFR2(17) - SBNO2(19)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(7)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(5)	Fusion	NEGATIVE
FGFR2-SMN1 fusion	FGFR2(17) - SMN1(2)	Fusion	NEGATIVE
FGFR2-STK26 fusion	FGFR2(17) - STK26(3)	Fusion	NEGATIVE
FGFR2-TACC2 fusion	FGFR2(17) - TACC2(11)	Fusion	NEGATIVE
FGFR2-TACC3 fusion	FGFR2(17) - TACC3(11)	Fusion	NEGATIVE
FGFR2-TBC1D1 fusion	FGFR2(17) - TBC1D1(9)	Fusion	NEGATIVE
FGFR2-TFEC fusion	FGFR2(17) - TFEC(2)	Fusion	NEGATIVE
FGFR2-TJP2 fusion	FGFR2(17) - TJP2(5)	Fusion	NEGATIVE
FGFR2-TXLNA fusion	FGFR2(17) - TXLNA(6)	Fusion	NEGATIVE
FGFR2-UBQLN1 fusion	FGFR2(17) - UBQLN1(8)	Fusion	NEGATIVE
FGFR2-UTRN fusion	FGFR2(17) - UTRN(51)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(2)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(7)	Fusion	NEGATIVE
FGFR2-WAC fusion	FGFR2(17) - WAC(5)	Fusion	NEGATIVE
FGFR2-WARS fusion	FGFR2(17) - WARS(3)	Fusion	NEGATIVE
FGFR2-ZMYM4 fusion	FGFR2(17) - ZMYM4(10)	Fusion	NEGATIVE
KLK2-FGFR2 fusion	KLK2(1) - FGFR2(5)	Fusion	NEGATIVE
PARK2-FGFR2 fusion	PARK2(9) - FGFR2(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDHX-FGFR2 fusion	PDHX(1) - FGFR2(7)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(2)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(1)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(8) - FGFR2(7)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(7) - FGFR2(7)	Fusion	NEGATIVE
ETV6-FGFR3 fusion	ETV6(5) - FGFR3(9)	Fusion	NEGATIVE
FGFR3-ADD1 fusion	FGFR3(17) - ADD1(3)	Fusion	NEGATIVE
FGFR3-AES fusion	FGFR3(17) - AES(2)	Fusion	NEGATIVE
FGFR3-AMBRA1 fusion	FGFR3(17) - AMBRA1(14)	Fusion	NEGATIVE
FGFR3-BAIAP2L1 fusion	FGFR3(17) - BAIAP2L1(2)	Fusion	NEGATIVE
FGFR3-BRAP fusion	FGFR3(17) - BRAP(5)	Fusion	NEGATIVE
FGFR3-CAMK2A fusion	FGFR3(18) - CAMK2A(14)	Fusion	NEGATIVE
FGFR3-ELAVL3 fusion	FGFR3(17) - ELAVL3(2)	Fusion	NEGATIVE
FGFR3-FBXO28 fusion	FGFR3(17) - FBXO28(4)	Fusion	NEGATIVE
FGFR3-JAKMIP1 fusion	FGFR3(17) - JAKMIP1(4)	Fusion	NEGATIVE
FGFR3-NBR1 fusion	FGFR3(17) - NBR1(7)	Fusion	NEGATIVE
FGFR3-RBBP4 fusion	FGFR3(17) - RBBP4(12)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(6)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(8)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(11)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(18) - TACC3(11)	Fusion	NEGATIVE
BAIAP2L1-MET fusion	BAIAP2L1(9) - MET(15)	Fusion	NEGATIVE
C8orf34-MET fusion	C8orf34(2) - 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
CLIP2-MET fusion	CLIP2(11) - MET(15)	Fusion	NEGATIVE
DCTN1-MET fusion	DCTN1(26) - MET(15)	Fusion	NEGATIVE
EPS15-MET fusion	EPS15(21) - MET(15)	Fusion	NEGATIVE
ETV6-MET fusion	ETV6(4) - MET(15)	Fusion	NEGATIVE
GTF2I-MET fusion	GTF2I(6) - MET(15)	Fusion	NEGATIVE
HLA-DRB1-MET fusion	HLA-DRB1(5) - MET(15)	Fusion	NEGATIVE
KIF5B-MET fusion	KIF5B(24) - MET(15)	Fusion	NEGATIVE
LRRFIP1-MET fusion	LRRFIP1(19) - MET(15)	Fusion	NEGATIVE
MET KDD	MET(21) - MET(15)	RNAExonVariant	NEGATIVE
MET exon 14 skipping (RNA)	MET(13) - MET(15)	RNAExonVariant	NEGATIVE
OXR1-MET fusion	OXR1(9) - MET(13)	Fusion	NEGATIVE
OXR1-MET fusion	OXR1(1) - MET(13)	Fusion	NEGATIVE
PPFIBP1-MET fusion	PPFIBP1(9) - MET(15)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
SCAF11-MET fusion	SCAF11(1) - MET(2)	Fusion	NEGATIVE
SLC34A2-MET fusion	SLC34A2(4) - MET(15)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(2)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(11)	Fusion	NEGATIVE
STARD3NL-MET fusion	STARD3NL(6) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(5) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(7) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(36) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(4) - MET(15)	Fusion	NEGATIVE
TRIM4-MET fusion	TRIM4(6) - MET(15)	Fusion	NEGATIVE
ZKSCAN1-MET fusion	ZKSCAN1(3) - MET(15)	Fusion	NEGATIVE
AKAP13-NRG1 fusion	AKAP13(5) - NRG1(2)	Fusion	NEGATIVE
ATP1B1-NRG1 fusion	ATP1B1(2) - 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
CDH1-NRG1 fusion	CDH1(2) - NRG1(2)	Fusion	NEGATIVE
CLU-NRG1 fusion	CLU(2) - NRG1(6)	Fusion	NEGATIVE
COX10-AS1-NRG1 fusion	COX10-AS1(1) - NRG1(2)	Fusion	NEGATIVE
HMBOX1-NRG1 fusion	HMBOX1(1) - NRG1(6)	Fusion	NEGATIVE
KIF13B-NRG1 fusion	KIF13B(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
NOTCH2-NRG1 fusion	NOTCH2(4) - NRG1(6)	Fusion	NEGATIVE
PARP8-NRG1 fusion	PARP8(1) - NRG1(2)	Fusion	NEGATIVE
PDE7A-NRG1 fusion	PDE7A(3) - NRG1(6)	Fusion	NEGATIVE
RAB3IL1-NRG1 fusion	RAB3IL1(9) - NRG1(6)	Fusion	NEGATIVE
RALGAPA1-NRG1 fusion	RALGAPA1(20) - NRG1(6)	Fusion	NEGATIVE
ROCK1-NRG1 fusion	ROCK1(2) - NRG1(2)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(4) - NRG1(6)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(2) - NRG1(6)	Fusion	NEGATIVE
SETD4-NRG1 fusion	SETD4(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(5) - NRG1(6)	Fusion	NEGATIVE
SMAD4-NRG1 fusion	SMAD4(1) - NRG1(6)	Fusion	NEGATIVE
TBL1XR1-NRG1 fusion	TBL1XR1(1) - NRG1(2)	Fusion	NEGATIVE
TENM4-NRG1 fusion	TENM4(12) - NRG1(2)	Fusion	NEGATIVE
THAP7-NRG1 fusion	THAP7(3) - NRG1(2)	Fusion	NEGATIVE
THBS1-NRG1 fusion	THBS1(6) - NRG1(6)	Fusion	NEGATIVE
TNC-NRG1 fusion	TNC(10) - NRG1(6)	Fusion	NEGATIVE
TSHZ2-NRG1 fusion	TSHZ2(1) - NRG1(6)	Fusion	NEGATIVE
VAMP2-NRG1 fusion	VAMP2(4) - NRG1(4)	Fusion	NEGATIVE
VTCN1-NRG1 fusion	VTCN1(2) - NRG1(2)	Fusion	NEGATIVE
WHSC1L1-NRG1 fusion	WHSC1L1(1) - NRG1(2)	Fusion	NEGATIVE
WRN-NRG1 fusion	WRN(30) - NRG1(2)	Fusion	NEGATIVE
ZMYM2-NRG1 fusion	ZMYM2(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
AFAP1-NTRK1 fusion	AFAP1(4) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF2-NTRK1 fusion	ARHGEF2(21) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF25-NTRK1 fusion	ARHGEF25(11) - NTRK1(8)	Fusion	NEGATIVE
BANP-NTRK1 fusion	BANP(4) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(12) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(13) - NTRK1(11)	Fusion	NEGATIVE
CD74-NTRK1 fusion	CD74(7) - NTRK1(10)	Fusion	NEGATIVE
CEL-NTRK1 fusion	CEL(7) - NTRK1(9)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(10)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(11)	Fusion	NEGATIVE
CTRC-NTRK1 fusion	CTRC(1) - NTRK1(9)	Fusion	NEGATIVE
EPHB2-NTRK1 fusion	EPHB2(3) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
GON4L-NTRK1 fusion	GON4L(21) - NTRK1(9)	Fusion	NEGATIVE
GRIPAP1-NTRK1 fusion	GRIPAP1(22) - NTRK1(11)	Fusion	NEGATIVE
GSN-NTRK1 fusion	GSN(7) - NTRK1(10)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(10)	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
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(11)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(10)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(5) - NTRK1(11)	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(11)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(12)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(13)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(11) - NTRK1(11)	Fusion	NEGATIVE
LRRC71-NTRK1 fusion	LRRC71(1) - NTRK1(9)	Fusion	NEGATIVE
MDM4-NTRK1 fusion	MDM4(3) - NTRK1(5)	Fusion	NEGATIVE
MPRIP-NTRK1 fusion	MPRIP(21) - NTRK1(12)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
NFASC-NTRK1 fusion	NFASC(20) - NTRK1(10)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(16) - DYNC2H1(85)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(17) - DYNC2H1(85)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PEAR1-NTRK1 fusion	PEAR1(2) - NTRK1(9)	Fusion	NEGATIVE
PEAR1-NTRK1 fusion	PEAR1(15) - NTRK1(10)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(21) - NTRK1(9)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(16) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(11)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(13)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(12) - NTRK1(13)	Fusion	NEGATIVE
RABGAP1L-NTRK1 fusion	RABGAP1L(14) - NTRK1(16)	Fusion	NEGATIVE
RFWD2-NTRK1 fusion	RFWD2(2) - NTRK1(3)	Fusion	NEGATIVE
RNF213-NTRK1 fusion	RNF213(15) - NTRK1(12)	Fusion	NEGATIVE
SCYL3-NTRK1 fusion	SCYL3(11) - NTRK1(12)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(2) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(12)	Fusion	NEGATIVE
SSBP2-NTRK1 fusion	SSBP2(12) - NTRK1(12)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
TP53-NTRK1 fusion	TP53(8) - NTRK1(9)	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(9)	Fusion	NEGATIVE
TPM3-NTRK1 fusion	TPM3(7) - 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(6) - NTRK1(11)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(9)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(10)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(12)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(13)	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
TRIM63-NTRK1 fusion	TRIM63(8) - NTRK1(9)	Fusion	NEGATIVE
VANGL2-NTRK1 fusion	VANGL2(1) - NTRK1(12)	Fusion	NEGATIVE
AFAP1-NTRK2 fusion	AFAP1(14) - NTRK2(12)	Fusion	NEGATIVE
AGBL4-NTRK2 fusion	AGBL4(6) - NTRK2(16)	Fusion	NEGATIVE
BCR-NTRK2 fusion	BCR(1) - NTRK2(17)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DAB2IP-NTRK2 fusion	DAB2IP(1) - NTRK2(17)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(4) - NTRK2(16)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(15)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(16)	Fusion	NEGATIVE
GKAP1-NTRK2 fusion	GKAP1(9) - NTRK2(13)	Fusion	NEGATIVE
GNAQ-NTRK2 fusion	GNAQ(2) - NTRK2(16)	Fusion	NEGATIVE
KCTD8-NTRK2 fusion	KCTD8(1) - NTRK2(13)	Fusion	NEGATIVE
NACC2-NTRK2 fusion	NACC2(4) - NTRK2(13)	Fusion	NEGATIVE
NAV1-NTRK2 fusion	NAV1(15) - NTRK2(11)	Fusion	NEGATIVE
NOS1AP-NTRK2 fusion	NOS1AP(9) - NTRK2(11)	Fusion	NEGATIVE
PAN3-NTRK2 fusion	PAN3(1) - NTRK2(17)	Fusion	NEGATIVE
QKI-NTRK2 fusion	QKI(6) - NTRK2(16)	Fusion	NEGATIVE
SLMAP-NTRK2 fusion	SLMAP(14) - NTRK2(16)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(13)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(17)	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
STRN3-NTRK2 fusion	STRN3(7) - NTRK2(16)	Fusion	NEGATIVE
TBC1D2-NTRK2 fusion	TBC1D2(6) - NTRK2(12)	Fusion	NEGATIVE
TLE4-NTRK2 fusion	TLE4(7) - NTRK2(15)	Fusion	NEGATIVE
TP63-NTRK2 fusion	TP63(8) - NTRK2(12)	Fusion	NEGATIVE
TRAF2-NTRK2 fusion	TRAF2(9) - NTRK2(15)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(15)	Fusion	NEGATIVE
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(16)	Fusion	NEGATIVE
VCAN-NTRK2 fusion	VCAN(6) - NTRK2(10)	Fusion	NEGATIVE
VCL-NTRK2 fusion	VCL(16) - NTRK2(12)	Fusion	NEGATIVE
WNK2-NTRK2 fusion	WNK2(24) - NTRK2(16)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(14) - NTRK3(14)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(16) - NTRK3(15)	Fusion	NEGATIVE
BTBD1-NTRK3 fusion	BTBD1(4) - NTRK3(14)	Fusion	NEGATIVE
COX5A-NTRK3 fusion	COX5A(1) - NTRK3(15)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(2) - NTRK3(14)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(6) - NTRK3(14)	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(15)	Fusion	NEGATIVE
ETV6-NTRK3 fusion	ETV6(5) - NTRK3(14)	Fusion	NEGATIVE
FAT1-NTRK3 fusion	FAT1(2) - NTRK3(7)	Fusion	NEGATIVE
FBXL20-NTRK3 fusion	FBXL20(1) - NTRK3(6)	Fusion	NEGATIVE
HNRNPA2B1-NTRK3 fusion	HNRNPA2B1(7) - NTRK3(14)	Fusion	NEGATIVE
KANK1-NTRK3 fusion	KANK1(3) - NTRK3(14)	Fusion	NEGATIVE
LYN-NTRK3 fusion	LYN(8) - NTRK3(14)	Fusion	NEGATIVE
MYH9-NTRK3 fusion	MYH9(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(13)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(11)	Fusion	NEGATIVE
NTRK3-HOMER1 fusion	NTRK3(17) - HOMER1(2)	Fusion	NEGATIVE
NTRK3-HOMER2 fusion	NTRK3(17) - HOMER2(2)	Fusion	NEGATIVE
NTRK3-LOXL2 fusion	NTRK3(3) - LOXL2(5)	Fusion	NEGATIVE
RBPMS-NTRK3 fusion	RBPMS(5) - 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
TPM4-NTRK3 fusion	TPM4(7) - NTRK3(12)	Fusion	NEGATIVE
UBE2R2-NTRK3 fusion	UBE2R2(3) - NTRK3(4)	Fusion	NEGATIVE
ZNF710-NTRK3 fusion	ZNF710(1) - NTRK3(14)	Fusion	NEGATIVE
AFF1-NUTM1 fusion	AFF1(17) - NUTM1(3)	Fusion	NEGATIVE
BCORL1-NUTM1 fusion	BCORL1(1) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CIC-NUTM1 fusion	CIC(17) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(2)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(20) - NUTM1(5)	Fusion	NEGATIVE
MXD1-NUTM1 fusion	MXD1(5) - NUTM1(2)	Fusion	NEGATIVE
WHSC1L1-NUTM1 fusion	WHSC1L1(7) - NUTM1(2)	Fusion	NEGATIVE
ZNF618-NUTM1 fusion	ZNF618(12) - NUTM1(2)	Fusion	NEGATIVE
ABI3BP-RET fusion	ABI3BP(34) - RET(12)	Fusion	NEGATIVE
ACBD5-RET fusion	ACBD5(11) - RET(12)	Fusion	NEGATIVE
ADD3-RET fusion	ADD3(1) - RET(12)	Fusion	NEGATIVE
AFAP1-RET fusion	AFAP1(3) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(6) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(2) - RET(12)	Fusion	NEGATIVE
AKAP13-RET fusion	AKAP13(35) - RET(12)	Fusion	NEGATIVE
ANK3-RET fusion	ANK3(1) - RET(12)	Fusion	NEGATIVE
BCR-RET fusion	BCR(4) - RET(12)	Fusion	NEGATIVE
CCDC186-RET fusion	CCDC186(7) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CCDC6-RET fusion	CCDC6(1) - RET(10)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(12)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CUX1-RET fusion	CUX1(10) - RET(12)	Fusion	NEGATIVE
DLG5-RET fusion	DLG5(13) - RET(12)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(11)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(12)	Fusion	NEGATIVE
EPHA5-RET fusion	EPHA5(2) - 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
FGFR1OP-RET fusion	FGFR1OP(11) - RET(11)	Fusion	NEGATIVE
FGFR1OP-RET fusion	FGFR1OP(12) - RET(12)	Fusion	NEGATIVE
FKBP15-RET fusion	FKBP15(25) - RET(12)	Fusion	NEGATIVE
FRMD4A-RET fusion	FRMD4A(12) - RET(12)	Fusion	NEGATIVE
GOLGA5-RET fusion	GOLGA5(7) - RET(12)	Fusion	NEGATIVE
GPHN-RET fusion	GPHN(9) - RET(12)	Fusion	NEGATIVE
HOOK3-RET fusion	HOOK3(11) - RET(12)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(10)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1468-RET fusion	KIAA1468(10) - RET(12)	Fusion	NEGATIVE
KIF13A-RET fusion	KIF13A(18) - 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(8)	Fusion	NEGATIVE
KIF5B-RET fusion	KIF5B(23) - RET(12)	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
KIF5B-RET fusion	KIF5B(15) - RET(12)	Fusion	NEGATIVE
KTN1-RET fusion	KTN1(29) - RET(12)	Fusion	NEGATIVE
MPRIP-RET fusion	MPRIP(19) - RET(12)	Fusion	NEGATIVE
MYH10-RET fusion	MYH10(33) - RET(12)	Fusion	NEGATIVE
MYH13-RET fusion	MYH13(35) - RET(12)	Fusion	NEGATIVE
MYO5A-RET fusion	MYO5A(23) - RET(11)	Fusion	NEGATIVE
MYO5C-RET fusion	MYO5C(25) - 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
OLFM4-RET fusion	OLFM4(4) - RET(10)	Fusion	NEGATIVE
PCM1-RET fusion	PCM1(29) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDCD10-RET fusion	PDCD10(4) - RET(11)	Fusion	NEGATIVE
PICALM-RET fusion	PICALM(19) - RET(12)	Fusion	NEGATIVE
PPFIBP2-RET fusion	PPFIBP2(9) - RET(12)	Fusion	NEGATIVE
PRKAR1A-RET fusion	PRKAR1A(7) - RET(12)	Fusion	NEGATIVE
PRKG1-RET fusion	PRKG1(3) - RET(12)	Fusion	NEGATIVE
RASAL2-RET fusion	RASAL2(15) - RET(12)	Fusion	NEGATIVE
RASSF4-RET fusion	RASSF4(3) - RET(12)	Fusion	NEGATIVE
RRBP1-RET fusion	RRBP1(9) - RET(12)	Fusion	NEGATIVE
RUFY1-RET fusion	RUFY1(17) - RET(12)	Fusion	NEGATIVE
RUFY2-RET fusion	RUFY2(9) - RET(12)	Fusion	NEGATIVE
SATB1-RET fusion	SATB1(7) - RET(12)	Fusion	NEGATIVE
SNRNP70-RET fusion	SNRNP70(7) - RET(12)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(11)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(12)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(5) - RET(11)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(6) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(11)	Fusion	NEGATIVE
TFG-RET fusion	TFG(4) - RET(11)	Fusion	NEGATIVE
TNIP1-RET fusion	TNIP1(11) - RET(12)	Fusion	NEGATIVE
TNIP2-RET fusion	TNIP2(5) - RET(12)	Fusion	NEGATIVE
TRIM24-RET fusion	TRIM24(9) - RET(12)	Fusion	NEGATIVE
TRIM27-RET fusion	TRIM27(3) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
WAC-RET fusion	WAC(3) - RET(12)	Fusion	NEGATIVE
CCDC30-ROS1 fusion	CCDC30(10) - ROS1(36)	Fusion	NEGATIVE
CCDC6-ROS1 fusion	CCDC6(5) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(7) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(33)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(32)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(4) - ROS1(33)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(12) - ROS1(35)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(8) - ROS1(36)	Fusion	NEGATIVE
CLIP1-ROS1 fusion	CLIP1(20) - ROS1(36)	Fusion	NEGATIVE
CLTC-ROS1 fusion	CLTC(31) - ROS1(35)	Fusion	NEGATIVE
ERC1-ROS1 fusion	ERC1(11) - ROS1(36)	Fusion	NEGATIVE
EYS-ROS1 fusion	EYS(12) - ROS1(9)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(35)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(34)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(33)	Fusion	NEGATIVE
GOLGB1-ROS1 fusion	GOLGB1(9) - ROS1(35)	Fusion	NEGATIVE
GOPC-ROS1 fusion	GOPC(8) - ROS1(35)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
GOPC-ROS1 fusion	GOPC(4) - ROS1(36)	Fusion	NEGATIVE
HLA-A-ROS1 fusion	HLA-A(7) - ROS1(34)	Fusion	NEGATIVE
LIMA1-ROS1 fusion	LIMA1(10) - ROS1(36)	Fusion	NEGATIVE
LRIG3-ROS1 fusion	LRIG3(16) - ROS1(35)	Fusion	NEGATIVE
MSN-ROS1 fusion	MSN(9) - ROS1(34)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(32) - ROS1(35)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(23) - ROS1(35)	Fusion	NEGATIVE
MYO5C-ROS1 fusion	MYO5C(30) - ROS1(35)	Fusion	NEGATIVE
NCOR2-ROS1 fusion	NCOR2(7) - ROS1(36)	Fusion	NEGATIVE
NFKB2-ROS1 fusion	NFKB2(14) - ROS1(36)	Fusion	NEGATIVE
PPFIBP1-ROS1 fusion	PPFIBP1(9) - ROS1(35)	Fusion	NEGATIVE
PWWP2A-ROS1 fusion	PWWP2A(1) - ROS1(36)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(5) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(32)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(32)	Fusion	NEGATIVE
SHTN1-ROS1 fusion	SHTN1(11) - ROS1(36)	Fusion	NEGATIVE
SLC16A10-ROS1 fusion	SLC16A10(3) - ROS1(3)	Fusion	NEGATIVE
SLC22A16-ROS1 fusion	SLC22A16(7) - ROS1(16)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(35)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(32)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(33)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(32)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - 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
TMEM106B-ROS1 fusion	TMEM106B(3) - ROS1(35)	Fusion	NEGATIVE
TPD52L1-ROS1 fusion	TPD52L1(3) - ROS1(33)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(7) - ROS1(35)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(3) - ROS1(36)	Fusion	NEGATIVE
TSPAN3-ROS1 fusion	TSPAN3(5) - ROS1(31)	Fusion	NEGATIVE
YWHAE-ROS1 fusion	YWHAE(4) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(3) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(2) - ROS1(36)	Fusion	NEGATIVE
EIF3E-RSPO2 fusion	EIF3E(1) - RSPO2(2)	Fusion	NEGATIVE
GRHL2-RSPO2 fusion	GRHL2(8) - 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

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
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

180 Oyster Point Blvd
South San Francisco, CA 94080

Clinical Test Report: Oncomine™ Dx Express Test - FFPE - RS v5.2.0

Patient Id: **PID01** Date of Birth: **30 MAR 1975** Date: 13 JAN 2026

Sample		Patient	
Sample Name:	**FFPE01**	Patient Id:	**PID01**
Cancer Type:	**Non-Small Cell Lung Cancer**	Gender:	**Male**
Cancer Stage:	**Stage III**	Date of Birth:	**30 MAR 1975**
Sample Type:	**FFPE**	Collection Date:	**22 FEB 2021**
%Cellularity:	**100**	Ordering Physician:	**Dr.Q**
%Necrosis:		Created On:	**06 NOV 2025 14:24**

Companion Diagnostics (CDx) Biomarkers

SNVs/Indels

Gene	Variant Name	AA Change	Nuc Change	Test Result	Variant ID	Associated Therapy
ERBB2	ERBB2 L755P	p.L755P	c.2263_2264delTTinsCC	POSITIVE	COSM683	HYRNUO® (sevabertinib)
ERBB2	ERBB2 exon 20 insertion	p.G776_V777insL	c.2328_2329insCTT	POSITIVE	COSM6438151	HYRNUO® (sevabertinib)

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
EGFR	EGFR exon 20 insertion	p.	c.2284-6_2284-5insTCCAGGAAGCCT	POSITIVE	COSM26720	ins
KRAS	KRAS G12C	p.G12C	c.34_36delGGTinsTGC	POSITIVE	COSM513	mnp

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
SQSTM1-NTRK3 fusion	SQSTM1(5) - NTRK3(14)	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
AKT1	AKT1 E17K	p.E17K	c.49G>A	POSITIVE	COSM33765	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
AR-V7	AR(3) - AR(4)	RNAExonVariant	POSITIVE

ⓘ 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 therapies 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)
NSCLC	ERBB2	ERBB2/HER2 activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions)	HYRNUO® (sevabertinib)

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 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 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 >70% necrotic tissue may yield unreliable sequencing results for the detection of *EGFR* exon 20 insertion and *ERBB2* 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).

30. For *MET* Exon 14 skipping results for cancer types other than non-small cell lung cancer, confirmatory testing using a clinically validated assay is recommended.

31. For NSCLC, the Oncomine™ Dx Express Test assay definition file includes prevalent but not all rare or newly identified *EGFR* exon 20 insertions, *ERBB2/HER2* exon 20 insertions, and *ERBB2/HER2* SNVs in exons 18-21 within the tyrosine kinase domain. The Oncomine™ Dx Express Test may miss rare or newly identified:

- *EGFR* exon 20 insertions carried by a subset of patients who may derive benefit from sunvozertinib
- *ERBB2/HER2* activating mutations (SNVs in exons 18-21 within the tyrosine kinase domain and exon 20 insertions) carried by a subset of patients who may derive benefit from sevabertinib

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 C77F	p.C77F	COSM5027916	NEGATIVE	SNV
AKT1 L52R	p.L52R	COSM93893	NEGATIVE	SNV
AKT1 P51L	p.P51L	COSM4468165	NEGATIVE	SNV
AKT1 P51S	p.P51S	COSM5991558	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AKT1 P70_C77dup	p.P70_C77dup	COSM6568358	NEGATIVE	ins
AKT1 Q79K	p.Q79K	COSM159008	NEGATIVE	SNV
AKT1 R76_C77insWPNTFIIR	p.R76_C77insWPNTFIIR	COSM5045024	NEGATIVE	ins
AKT2 D324G	p.D324G	COSM474741	NEGATIVE	SNV
AKT2 D324H	p.D324H	COSM474742	NEGATIVE	SNV
AKT2 E17K	p.E17K	COSM159009	NEGATIVE	SNV
AKT3 E17K	p.E17K	COSM224779	NEGATIVE	SNV
AKT3 L51R	p.L51R	COSM309035	NEGATIVE	SNV
AKT3 Q78K	p.Q78K	COSM242892	NEGATIVE	SNV
ALK A1200V	p.A1200V	COSM317003	NEGATIVE	SNV
ALK A1280V	p.A1280V	COSM6914238	NEGATIVE	SNV
ALK C1156F	p.C1156F	BT144	NEGATIVE	SNV
ALK C1156T	p.C1156T	OM3350	NEGATIVE	mnp
ALK C1156Y	p.C1156Y	COSM99136	NEGATIVE	SNV
ALK D1203A	p.D1203A	OM3313	NEGATIVE	SNV
ALK D1203N	p.D1203N	COSM6191344	NEGATIVE	SNV
ALK E1129V	p.E1129V	BT117	NEGATIVE	SNV
ALK E1154Q	p.E1154Q	OM3324	NEGATIVE	SNV
ALK E1210K	p.E1210K	OM3161	NEGATIVE	SNV
ALK F1174C	p.F1174C	COSM28059	NEGATIVE	SNV
ALK F1174I	p.F1174I	COSM28491	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28055	NEGATIVE	SNV
ALK F1174L	p.F1174L	COSM28061	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK F1174L	p.F1174L	COSM28057	NEGATIVE	SNV
ALK F1174S	p.F1174S	COSM53063	NEGATIVE	SNV
ALK F1174V	p.F1174V	COSM28054	NEGATIVE	SNV
ALK F1245C	p.F1245C	COSM28500	NEGATIVE	SNV
ALK F1245I	p.F1245I	COSM28492	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28062	NEGATIVE	SNV
ALK F1245L	p.F1245L	COSM28493	NEGATIVE	SNV
ALK F1245V	p.F1245V	COSM28499	NEGATIVE	SNV
ALK G1123D	p.G1123D	OM3262	NEGATIVE	SNV
ALK G1123S	p.G1123S	COSM5045108	NEGATIVE	SNV
ALK G1128A	p.G1128A	COSM98475	NEGATIVE	SNV
ALK G1202R	p.G1202R	COSM144250	NEGATIVE	SNV
ALK G1202del	p.G1202del	OMINDEL698	NEGATIVE	del
ALK G1269A	p.G1269A	COSM1169707	NEGATIVE	SNV
ALK I1171H	p.I1171H	OM3163	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM1169448	NEGATIVE	mnp
ALK I1171N	p.I1171N	COSM28498	NEGATIVE	SNV
ALK I1171S	p.I1171S	COSM4440758	NEGATIVE	SNV
ALK I1171T	p.I1171T	COSM4381100	NEGATIVE	SNV
ALK I1268V	p.I1268V	BT149	NEGATIVE	SNV
ALK L1152P	p.L1152P	COSM1407659	NEGATIVE	SNV
ALK L1152R	p.L1152R	COSM97185	NEGATIVE	SNV
ALK L1152V	p.L1152V	OM3513	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ALK L1196M	p.L1196M	COSM99137	NEGATIVE	SNV
ALK L1196Q	p.L1196Q	COSM1169447	NEGATIVE	SNV
ALK L1198F	p.L1198F	COSM97028	NEGATIVE	SNV
ALK L1198P	p.L1198P	OM3162	NEGATIVE	SNV
ALK L1198V	p.L1198V	OM3314	NEGATIVE	SNV
ALK R1192P	p.R1192P	OM3259	NEGATIVE	SNV
ALK R1192Q	p.R1192Q	OM3372	NEGATIVE	SNV
ALK R1209Q	p.R1209Q	COSM327056	NEGATIVE	SNV
ALK R1231Q	p.R1231Q	COSM5967491	NEGATIVE	SNV
ALK R1231W	p.R1231W	BT148	NEGATIVE	SNV
ALK R1275L	p.R1275L	COSM28060	NEGATIVE	SNV
ALK R1275Q	p.R1275Q	COSM28056	NEGATIVE	SNV
ALK S1157F	p.S1157F	BT145	NEGATIVE	SNV
ALK S1206C	p.S1206C	BT147	NEGATIVE	SNV
ALK S1206F	p.S1206F	COSM6909200	NEGATIVE	SNV
ALK S1206Y	p.S1206Y	COSM144251	NEGATIVE	SNV
ALK T1151K	p.T1151K	OM3261	NEGATIVE	SNV
ALK T1151M	p.T1151M	COSM98478	NEGATIVE	SNV
ALK T1151R	p.T1151R	OM3260	NEGATIVE	SNV
ALK T1151dup	p.T1151dup	COSM144252	NEGATIVE	ins
ALK V1180L	p.V1180L	COSM4381101	NEGATIVE	SNV
ALK Y1278S	p.Y1278S	COSM28058	NEGATIVE	SNV
AR D880E	p.D880E	COSM5945750	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
AR D891H	p.D891H	COSM5945767	NEGATIVE	SNV
AR E894K	p.E894K	COSM5945748	NEGATIVE	SNV
AR E898G	p.E898G	OM3322	NEGATIVE	SNV
AR F814V	p.F814V	COSM150793	NEGATIVE	SNV
AR F877L	p.F877L	COSM5945752	NEGATIVE	SNV
AR F877L	p.F877L	COSM5946164	NEGATIVE	SNV
AR H875Q	p.H875Q	COSM5945766	NEGATIVE	SNV
AR H875Y	p.H875Y	COSM238555	NEGATIVE	SNV
AR L702H	p.L702H	COSM238553	NEGATIVE	SNV
AR L882I	p.L882I	COSM5945751	NEGATIVE	SNV
AR M896T	p.M896T	OM3321	NEGATIVE	SNV
AR M896V	p.M896V	COSM5945749	NEGATIVE	SNV
AR S889G	p.S889G	COSM5945753	NEGATIVE	SNV
AR T878A	p.T878A	COSM236693	NEGATIVE	SNV
AR T878S	p.T878S	COSM5570418	NEGATIVE	SNV
AR V716M	p.V716M	COSM3094417	NEGATIVE	SNV
AR V731M	p.V731M	COSM1468984	NEGATIVE	SNV
AR W742C	p.W742C	COSM5572266	NEGATIVE	SNV
AR W742C	p.W742C	COSM5944171	NEGATIVE	SNV
AR W742L	p.W742L	COSM5944172	NEGATIVE	SNV
ARAF S214A	p.S214A	COSM1742787	NEGATIVE	SNV
ARAF S214C	p.S214C	COSM5044705	NEGATIVE	SNV
ARAF S214F	p.S214F	COSM612884	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ARAF S214T	p.S214T	COSM1558277	NEGATIVE	SNV
ARAF S214Y	p.S214Y	COSM5416443	NEGATIVE	SNV
BRAF A598T	p.A598T	COSM28505	NEGATIVE	SNV
BRAF A598V	p.A598V	COSM21549	NEGATIVE	SNV
BRAF D594A	p.D594A	COSM1583010	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM1121	NEGATIVE	SNV
BRAF D594E	p.D594E	COSM253330	NEGATIVE	SNV
BRAF D594G	p.D594G	COSM467	NEGATIVE	SNV
BRAF D594H	p.D594H	COSM144576	NEGATIVE	SNV
BRAF D594N	p.D594N	COSM27639	NEGATIVE	SNV
BRAF D594V	p.D594V	COSM466	NEGATIVE	SNV
BRAF D594Y	p.D594Y	COSM6928839	NEGATIVE	SNV
BRAF D594_T599dup	p.D594_T599dup	COSM26504	NEGATIVE	ins
BRAF E586K	p.E586K	COSM463	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM468	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM53198	NEGATIVE	SNV
BRAF F595L	p.F595L	COSM21612	NEGATIVE	SNV
BRAF F595S	p.F595S	COSM1123	NEGATIVE	SNV
BRAF G464E	p.G464E	COSM449	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1111	NEGATIVE	SNV
BRAF G464R	p.G464R	COSM1448615	NEGATIVE	SNV
BRAF G464V	p.G464V	COSM450	NEGATIVE	SNV
BRAF G466A	p.G466A	COSM452	NEGATIVE	SNV

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Patient Id: **PID01** Date of Birth: 30 MAR 1975 Date: 13 JAN 2026

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF G466E	p.G466E	COSM453	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM1112	NEGATIVE	SNV
BRAF G466R	p.G466R	COSM253328	NEGATIVE	SNV
BRAF G466V	p.G466V	COSM451	NEGATIVE	SNV
BRAF G469A	p.G469A	COSM460	NEGATIVE	SNV
BRAF G469E	p.G469E	COSM461	NEGATIVE	SNV
BRAF G469L	p.G469L	COSM6056848	NEGATIVE	mnp
BRAF G469R	p.G469R	COSM455	NEGATIVE	SNV
BRAF G469R	p.G469R	COSM457	NEGATIVE	SNV
BRAF G469S	p.G469S	COSM1113	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM27912	NEGATIVE	mnp
BRAF G469S	p.G469S	COSM458	NEGATIVE	mnp
BRAF G469V	p.G469V	COSM459	NEGATIVE	SNV
BRAF G596C	p.G596C	COSM6936824	NEGATIVE	SNV
BRAF G596D	p.G596D	COSM26506	NEGATIVE	SNV
BRAF G596R	p.G596R	COSM469	NEGATIVE	SNV
BRAF G596V	p.G596V	COSM9145962	NEGATIVE	SNV
BRAF I463S	p.I463S	COSM448	NEGATIVE	SNV
BRAF K601E	p.K601E	COSM478	NEGATIVE	SNV
BRAF K601I	p.K601I	COSM26491	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM1132	NEGATIVE	SNV
BRAF K601N	p.K601N	COSM6265	NEGATIVE	SNV
BRAF K601R	p.K601R	COSM1568586	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF K601T	p.K601T	COSM3878760	NEGATIVE	SNV
BRAF L597P	p.L597P	COSM1448593	NEGATIVE	SNV
BRAF L597Q	p.L597Q	COSM1125	NEGATIVE	SNV
BRAF L597R	p.L597R	COSM471	NEGATIVE	SNV
BRAF L597S	p.L597S	COSM1126	NEGATIVE	mnp
BRAF L597V	p.L597V	COSM470	NEGATIVE	SNV
BRAF N581I	p.N581I	COSM26460	NEGATIVE	SNV
BRAF N581S	p.N581S	COSM462	NEGATIVE	SNV
BRAF N581T	p.N581T	COSM1699452	NEGATIVE	SNV
BRAF R462I	p.R462I	COSM447	NEGATIVE	SNV
BRAF T599I	p.T599I	COSM472	NEGATIVE	SNV
BRAF T599R	p.T599R	COSM4172020	NEGATIVE	SNV
BRAF T599_V600insEAT	p.T599_V600insEAT	COSM4166148	NEGATIVE	ins
BRAF T599_V600insETT	p.T599_V600insETT	COSM4172018	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM144982	NEGATIVE	ins
BRAF T599dup	p.T599dup	COSM30730	NEGATIVE	ins
BRAF V600A	p.V600A	COSM18443	NEGATIVE	SNV
BRAF V600D	p.V600D	COSM308550	NEGATIVE	mnp
BRAF V600D	p.V600D	COSM477	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM475	NEGATIVE	mnp
BRAF V600E	p.V600E	COSM476	NEGATIVE	SNV
BRAF V600G	p.V600G	COSM6137	NEGATIVE	SNV
BRAF V600K	p.V600K	COSM473	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
BRAF V600L	p.V600L	COSM219798	NEGATIVE	SNV
BRAF V600L	p.V600L	COSM33808	NEGATIVE	SNV
BRAF V600M	p.V600M	COSM1130	NEGATIVE	SNV
BRAF V600Q	p.V600Q	COSM249889	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM1583011	NEGATIVE	mnp
BRAF V600R	p.V600R	COSM474	NEGATIVE	mnp
BRAF V600_K601delinsE	p.V600_K601delinsE	COSM1133	NEGATIVE	del
BRAF Y472C	p.Y472C	COSM1133046	NEGATIVE	SNV
BRAF [G593=;D594N]	p.[G593=;D594N]	COSM211600	NEGATIVE	mnp
BRAF [T599=;V600R]	p.[T599=;V600R]	COSM1127	NEGATIVE	mnp
BRAF [T599I;V600K]	p.[T599I;V600K]	COSM1735763	NEGATIVE	mnp
BRAF [T599R;V600G]	p.[T599R;V600G]	OM3547	NEGATIVE	mnp
CDK4 K22M	p.K22M	COSM3463915	NEGATIVE	SNV
CDK4 K22Q	p.K22Q	OM3153	NEGATIVE	SNV
CDK4 K22R	p.K22R	COSM232013	NEGATIVE	SNV
CDK4 R24C	p.R24C	COSM1677139	NEGATIVE	SNV
CDK4 R24H	p.R24H	COSM1989836	NEGATIVE	SNV
CDK4 R24L	p.R24L	COSM363684	NEGATIVE	SNV
CDK4 R24S	p.R24S	COSM3463914	NEGATIVE	SNV
CHEK2 K373E	p.K373E	COSM42871	NEGATIVE	SNV
CHEK2 M381V	p.M381V	COSM1559601	NEGATIVE	SNV
CHEK2 R523H	p.R523H	COSM1559596	NEGATIVE	SNV
CHEK2 R523P	p.R523P	COSM1559595	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CHEK2 [S372=;K373E]	p.[S372=;K373E]	COSM1294063	NEGATIVE	mnp
CTNNB1 D32A	p.D32A	COSM5690	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM22939	NEGATIVE	SNV
CTNNB1 D32E	p.D32E	COSM5766	NEGATIVE	SNV
CTNNB1 D32G	p.D32G	COSM5681	NEGATIVE	SNV
CTNNB1 D32H	p.D32H	COSM5668	NEGATIVE	SNV
CTNNB1 D32N	p.D32N	COSM5672	NEGATIVE	SNV
CTNNB1 D32V	p.D32V	COSM5691	NEGATIVE	SNV
CTNNB1 D32Y	p.D32Y	COSM5661	NEGATIVE	SNV
CTNNB1 G34A	p.G34A	COSM5988492	NEGATIVE	SNV
CTNNB1 G34E	p.G34E	COSM5671	NEGATIVE	SNV
CTNNB1 G34I	p.G34I	COSM1666841	NEGATIVE	mnp
CTNNB1 G34R	p.G34R	COSM5684	NEGATIVE	SNV
CTNNB1 G34R	p.G34R	COSM5686	NEGATIVE	SNV
CTNNB1 G34V	p.G34V	COSM5670	NEGATIVE	SNV
CTNNB1 H36P	p.H36P	COSM5678	NEGATIVE	SNV
CTNNB1 H36R	p.H36R	COSM27378	NEGATIVE	SNV
CTNNB1 H36Y	p.H36Y	COSM5703	NEGATIVE	SNV
CTNNB1 I35K	p.I35K	COSM28727	NEGATIVE	mnp
CTNNB1 I35M	p.I35M	COSM28711	NEGATIVE	SNV
CTNNB1 I35N	p.I35N	COSM5695	NEGATIVE	SNV
CTNNB1 I35S	p.I35S	COSM5674	NEGATIVE	SNV
CTNNB1 I35T	p.I35T	COSM13168	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 I35V	p.I35V	COSM1566774	NEGATIVE	SNV
CTNNB1 K335I	p.K335I	COSM17797	NEGATIVE	SNV
CTNNB1 K335Q	p.K335Q	COSM4421324	NEGATIVE	SNV
CTNNB1 K335R	p.K335R	COSM5945941	NEGATIVE	SNV
CTNNB1 K335T	p.K335T	COSM1725761	NEGATIVE	SNV
CTNNB1 L46_S47del	p.L46_S47del	COSM6129	NEGATIVE	del
CTNNB1 L46_S47del	p.L46_S47del	COSM6132	NEGATIVE	del
CTNNB1 P44A	p.P44A	COSM17661	NEGATIVE	SNV
CTNNB1 P44L	p.P44L	COSM5761	NEGATIVE	SNV
CTNNB1 P44S	p.P44S	COSM5704	NEGATIVE	SNV
CTNNB1 S33A	p.S33A	COSM5683	NEGATIVE	SNV
CTNNB1 S33C	p.S33C	COSM5677	NEGATIVE	SNV
CTNNB1 S33F	p.S33F	COSM5669	NEGATIVE	SNV
CTNNB1 S33L	p.S33L	COSM6098	NEGATIVE	mnp
CTNNB1 S33N	p.S33N	COSM6099	NEGATIVE	mnp
CTNNB1 S33P	p.S33P	COSM5682	NEGATIVE	SNV
CTNNB1 S33T	p.S33T	COSM27311	NEGATIVE	SNV
CTNNB1 S33Y	p.S33Y	COSM5673	NEGATIVE	SNV
CTNNB1 S37A	p.S37A	COSM5675	NEGATIVE	SNV
CTNNB1 S37C	p.S37C	COSM5679	NEGATIVE	SNV
CTNNB1 S37F	p.S37F	COSM5662	NEGATIVE	SNV
CTNNB1 S37P	p.S37P	COSM5687	NEGATIVE	SNV
CTNNB1 S37T	p.S37T	COSM5729	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
CTNNB1 S37Y	p.S37Y	COSM5666	NEGATIVE	SNV
CTNNB1 S45A	p.S45A	COSM5685	NEGATIVE	SNV
CTNNB1 S45C	p.S45C	COSM5689	NEGATIVE	SNV
CTNNB1 S45F	p.S45F	COSM5667	NEGATIVE	SNV
CTNNB1 S45P	p.S45P	COSM5663	NEGATIVE	SNV
CTNNB1 S45T	p.S45T	COSM5719	NEGATIVE	SNV
CTNNB1 S45Y	p.S45Y	COSM5692	NEGATIVE	SNV
CTNNB1 S45_G48del	p.S45_G48del	COSM5992	NEGATIVE	del
CTNNB1 S45_S47del	p.S45_S47del	COSM1617582	NEGATIVE	del
CTNNB1 S45_S47delinsC	p.S45_S47delinsC	COSM28708	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6128	NEGATIVE	del
CTNNB1 S45del	p.S45del	COSM6131	NEGATIVE	del
CTNNB1 T41A	p.T41A	COSM5664	NEGATIVE	SNV
CTNNB1 T41I	p.T41I	COSM5676	NEGATIVE	SNV
CTNNB1 T41N	p.T41N	COSM5730	NEGATIVE	SNV
CTNNB1 T41P	p.T41P	COSM5688	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5716	NEGATIVE	SNV
CTNNB1 T41S	p.T41S	COSM5701	NEGATIVE	SNV
CTNNB1 [S45E;L46=]	p.[S45E;L46=]	COSM86011	NEGATIVE	mnp
EGFR A289D	p.A289D	COSM21685	NEGATIVE	SNV
EGFR A289T	p.A289T	COSM21686	NEGATIVE	SNV
EGFR A289V	p.A289V	COSM21687	NEGATIVE	SNV
EGFR A750P	p.A750P	COSM6219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR C775Y	p.C775Y	COSM22954	NEGATIVE	SNV
EGFR C797G	p.C797G	COSM6493936	NEGATIVE	SNV
EGFR C797N	p.C797N	OM3543	NEGATIVE	mnp
EGFR C797S	p.C797S	COSM6493937	NEGATIVE	SNV
EGFR C797S	p.C797S	COSM5945664	NEGATIVE	SNV
EGFR E709A	p.E709A	COSM13427	NEGATIVE	SNV
EGFR E709G	p.E709G	COSM13009	NEGATIVE	SNV
EGFR E709H	p.E709H	COSM12428	NEGATIVE	mnp
EGFR E709K	p.E709K	COSM12988	NEGATIVE	SNV
EGFR E709Q	p.E709Q	COSM116882	NEGATIVE	SNV
EGFR E709V	p.E709V	COSM12371	NEGATIVE	SNV
EGFR E709_T710delinsA	p.E709_T710delinsA	COSM85796	NEGATIVE	del
EGFR E709_T710delinsD	p.E709_T710delinsD	COSM51525	NEGATIVE	del
EGFR E709_T710delinsG	p.E709_T710delinsG	COSM48981	NEGATIVE	complex
EGFR E746G	p.E746G	COSM6849765	NEGATIVE	SNV
EGFR G465E	p.G465E	BT129	NEGATIVE	SNV
EGFR G465R	p.G465R	COSM601581	NEGATIVE	SNV
EGFR G598A	p.G598A	COSM3412196	NEGATIVE	SNV
EGFR G598V	p.G598V	COSM21690	NEGATIVE	SNV
EGFR G719A	p.G719A	COSM6239	NEGATIVE	SNV
EGFR G719C	p.G719C	COSM6253	NEGATIVE	SNV
EGFR G719D	p.G719D	COSM18425	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR G719S	p.G719S	COSM6252	NEGATIVE	SNV
EGFR G721D	p.G721D	COSM22993	NEGATIVE	SNV
EGFR G721R	p.G721R	OM3325	NEGATIVE	SNV
EGFR G721S	p.G721S	COSM22992	NEGATIVE	SNV
EGFR G724S	p.G724S	COSM13979	NEGATIVE	SNV
EGFR G796A	p.G796A	COSM27568	NEGATIVE	SNV
EGFR G796C	p.G796C	OM3318	NEGATIVE	SNV
EGFR G796R	p.G796R	COSM6493935	NEGATIVE	SNV
EGFR G796S	p.G796S	COSM20891	NEGATIVE	SNV
EGFR H835L	p.H835L	COSM6227	NEGATIVE	SNV
EGFR I491M	p.I491M	BT135	NEGATIVE	SNV
EGFR I744T	p.I744T	COSM87245	NEGATIVE	SNV
EGFR K467T	p.K467T	BT132	NEGATIVE	SNV
EGFR K757R	p.K757R	COSM133588	NEGATIVE	SNV
EGFR L718M	p.L718M	COSM1716255	NEGATIVE	SNV
EGFR L718Q	p.L718Q	COSM6503269	NEGATIVE	SNV
EGFR L718V	p.L718V	OM3316	NEGATIVE	SNV
EGFR L747P	p.L747P	COSM24267	NEGATIVE	mnp
EGFR L747S	p.L747S	COSM26704	NEGATIVE	SNV
EGFR L747V	p.L747V	OM3537	NEGATIVE	SNV
EGFR L792F	p.L792F	COSM6493933	NEGATIVE	SNV
EGFR L792H	p.L792H	COSM6493934	NEGATIVE	SNV
EGFR L792P	p.L792P	COSM13190	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR L792R	p.L792R	OM3267	NEGATIVE	SNV
EGFR L792V	p.L792V	OM3265	NEGATIVE	SNV
EGFR L792Y	p.L792Y	OM3266	NEGATIVE	mnp
EGFR L798I	p.L798I	OM3268	NEGATIVE	SNV
EGFR L833F	p.L833F	COSM219794	NEGATIVE	SNV
EGFR L833V	p.L833V	COSM13424	NEGATIVE	SNV
EGFR L833W	p.L833W	COSM133204	NEGATIVE	SNV
EGFR L858K	p.L858K	COSM24268	NEGATIVE	mnp
EGFR L858M	p.L858M	COSM12366	NEGATIVE	SNV
EGFR L858Q	p.L858Q	COSM29578	NEGATIVE	SNV
EGFR L858R	p.L858R	COSM13553	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM12429	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM133630	NEGATIVE	mnp
EGFR L858R	p.L858R	COSM6224	NEGATIVE	SNV
EGFR L861Q	p.L861Q	COSM6213	NEGATIVE	SNV
EGFR L861R	p.L861R	COSM12374	NEGATIVE	SNV
EGFR L861V	p.L861V	COSM24848	NEGATIVE	SNV
EGFR N771T	p.N771T	OM3174	NEGATIVE	SNV
EGFR P848L	p.P848L	COSM22943	NEGATIVE	SNV
EGFR R108G	p.R108G	COSM1451536	NEGATIVE	SNV
EGFR R108K	p.R108K	COSM21683	NEGATIVE	SNV
EGFR R451C	p.R451C	COSM3266583	NEGATIVE	SNV
EGFR R836C	p.R836C	COSM28604	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR S464L	p.S464L	BT126	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236671	NEGATIVE	SNV
EGFR S492R	p.S492R	COSM236670	NEGATIVE	SNV
EGFR S720I	p.S720I	OM3317	NEGATIVE	mnp
EGFR S720P	p.S720P	COSM88729	NEGATIVE	SNV
EGFR S768G	p.S768G	COSM1451595	NEGATIVE	SNV
EGFR S768I	p.S768I	COSM6241	NEGATIVE	SNV
EGFR S768N	p.S768N	COSM12989	NEGATIVE	SNV
EGFR S768T	p.S768T	COSM291998	NEGATIVE	SNV
EGFR T790A	p.T790A	COSM1272071	NEGATIVE	SNV
EGFR T790M	p.T790M	COSM6240	NEGATIVE	SNV
EGFR T854A	p.T854A	COSM28537	NEGATIVE	SNV
EGFR T854I	p.T854I	COSM87246	NEGATIVE	SNV
EGFR T854S	p.T854S	COSM1235378	NEGATIVE	SNV
EGFR V292L	p.V292L	COSM43067	NEGATIVE	SNV
EGFR V769L	p.V769L	COSM6242	NEGATIVE	SNV
EGFR V769M	p.V769M	COSM13425	NEGATIVE	SNV
EGFR [H773L;V774M]	p.[H773L;V774M]	COSM6978402	NEGATIVE	mnp
EGFR [K745=;E746I;L747P]	p.[K745=;E746I;L747P]	OM3355	NEGATIVE	mnp
EGFR [L718=;G719C]	p.[L718=;G719C]	COSM18441	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM6984779	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	COSM85750	NEGATIVE	mnp
EGFR [S768I;V769L]	p.[S768I;V769L]	OMINDEL1181	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR [V769G;D770I]	p.[V769G;D770I]	OM3540	NEGATIVE	mnp
EGFR exon 19 deletion	p.I744_E749delinsLKR	COSM133186	NEGATIVE	complex
EGFR exon 19 deletion	p.I744_A750delinsVK	COSM85798	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750del	COSM221565	NEGATIVE	del
EGFR exon 19 deletion	p.K745_E749del	COSM26038	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM3734668	NEGATIVE	complex
EGFR exon 19 deletion	p.K745_A750delinsT	COSM1190791	NEGATIVE	del
EGFR exon 19 deletion	p.E746del	COSM18420	NEGATIVE	del
EGFR exon 19 deletion	p.E746_E749del	COSM28517	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751del	COSM24869	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.E746_T751del	COSM12728	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752del	COSM133189	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsA	COSM13549	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM13550	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM13551	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM13552	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_E749del	COSM6218	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750del	COSM6225	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsFPS	COSM6506513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsA	COSM12367	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsRP	COSM12413	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsA	COSM12678	NEGATIVE	del
EGFR exon 19 deletion	p.E746_P753delinsLS	COSM13200	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133188	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsI	COSM133190	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsIS	COSM133191	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsQP	COSM13557	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsQ	COSM22999	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_K754del	COSM24970	NEGATIVE	del
EGFR exon 19 deletion	p.E746_T751delinsS	COSM26513	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsI	COSM26680	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsL	COSM51502	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsIP	COSM51526	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_A750delinsP	COSM6947327	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsIP	COSM6966471	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsM	OMINDEL716	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751del	COSM12369	NEGATIVE	del
EGFR exon 19 deletion	p.E746_S752delinsV	COSM12384	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM12386	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM12416	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsV	COSM133193	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsAP	COSM144207	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM18421	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.L747_P753del	COSM255211	NEGATIVE	del
EGFR exon 19 deletion	p.E746_A750delinsVP	COSM28623	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_P753delinsVQ	COSM51524	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVP	COSM52935	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_T751delinsVA	COSM53205	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_S752delinsD	COSM6220	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.E746_S752delinsV	COSM674057	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_T751delinsVS	COSM6980200	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750del	COSM9179903	NEGATIVE	del
EGFR exon 19 deletion	p.L747_T751delinsQ	COSM12419	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQH	COSM12421	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12422	NEGATIVE	complex
EGFR exon 19 deletion	p.E746_A750delinsDP	COSM18428	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM22944	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_S752delinsQ	COSM26441	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.E746_A750delinsD	COSM6974307	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsP	COSM1235296	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_P753delinsS	COSM12370	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A750delinsP	COSM12382	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsP	COSM12383	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM12387	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_S752delinsQH	COSM133196	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_A750delinsP	COSM4170220	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_A750delinsS	COSM4170221	NEGATIVE	del
EGFR exon 19 deletion	p.L747_P753delinsQ	COSM51501	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsN	COSM51503	NEGATIVE	complex
EGFR exon 19 deletion	p.L747_T751delinsS	COSM6210	NEGATIVE	del
EGFR exon 19 deletion	p.L747_A755delinsAN	COSM85891	NEGATIVE	complex
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_E758delinsP	COSM26440	NEGATIVE	complex

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.T751_E758del	COSM133207	NEGATIVE	del
EGFR exon 19 deletion	p.A750_I759delinsGS	COSM5023005	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM1667024	NEGATIVE	complex
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.S752_I759del	COSM13556	NEGATIVE	del
EGFR exon 19 deletion	p.T751_I759delinsS	COSM1667027	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsREA	COSM22956	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM24270	NEGATIVE	complex
EGFR exon 19 deletion	p.T751_I759delinsN	COSM96856	NEGATIVE	complex
EGFR exon 19 deletion	p.S752_I759del	COSM6256	NEGATIVE	del
EGFR exon 19 deletion	p.P753_I759del	COSM24269	NEGATIVE	del
EGFR exon 19 deletion	p.K754_I759del	COSM5271797	NEGATIVE	del
EGFR exon 19 insertion	p.I740_K745dup	COSM12423	NEGATIVE	ins
EGFR exon 19 insertion	p.I740_K745dup	COSM51504	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insVPVAIK	COSM26444	NEGATIVE	ins
EGFR exon 19 insertion	p.K745_E746insTPVAIK	COSM255152	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.?	OMINDEL1050	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_D770dup	COSM6962256	NEGATIVE	ins
EGFR exon 20 insertion	p.Y764_V765insHH	OMINDEL1051	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM12376	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_S768insTLA	COSM12425	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM20884	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVASV	OMINDEL1800	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.S768_P772dup	OMINDEL1053	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSSV	OMINDEL1054	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insTSV	OMINDEL1055	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.S768_V769insLDS	OMINDEL1802	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insRSV	OMINDEL952	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_N771dup	S768_N771dup	NEGATIVE	ins

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.A767_S768insYVM	COSM1651740	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	COSM12379	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRVDNP	OMINDEL1056	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insANV	COSM1651741	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	COSM18430	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_N771dup	COSM20885	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_P772dup	OMINDEL1057	NEGATIVE	ins
EGFR exon 20 insertion	p.S768_V769delinsISW	OMINDEL1058	NEGATIVE	complex
EGFR exon 20 insertion	p.S768delinsRRGH	OMINDEL1059	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTV	OMINDEL1060	NEGATIVE	ins
EGFR exon 20 insertion	p.V769dup	COSM6506514	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGVV	OMINDEL1187	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	OMINDEL1803	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTM	OMINDEL1061	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1062	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insM	OMINDEL1063	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1064	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1065	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insSTL	OMINDEL1066	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM6845099	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM85795	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGG	MATNV07	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_D770insGE	OMINDEL1069	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGQ	OMINDEL1070	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insGTR	OMINDEL1071	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.V769_D770insERG	COSM1651742	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772dup	COSM1651745	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	COSM22955	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insMASVD	COSM28638	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	MATNV03	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGTD	OMINDEL1074	NEGATIVE	ins

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.V769_D770insP	OMINDEL1076	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGD	OMINDEL1804	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771dup	OMINDEL1805	NEGATIVE	ins
EGFR exon 20 insertion	p.D770dup	OMINDEL1806	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insY	COSM1238030	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	COSM12381	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGY	COSM12427	NEGATIVE	ins
EGFR exon 20 insertion	p.N771dup	COSM13003	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGTH	COSM6983510	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGNPH	D770_N771insGNPH	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_1	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insKH	D770_N771insKH_2	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.D770delinsASVH	OMINDEL1080	NEGATIVE	ins

EGFR exon 20 insertion	p.D770_N771insH	OMINDEL1081	NEGATIVE	ins	

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.N771_P772insHN	OMINDEL1084	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.D770_N771insH	OMINDEL1807	NEGATIVE	ins
EGFR exon 20 insertion	p.V769_D770insCV	OMINDEL1808	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1809	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsAGG	COSM12737	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insG	COSM13004	NEGATIVE	ins
EGFR exon 20 insertion	p.A767_V769dup	COSM13558	NEGATIVE	complex
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_N771insT	COSM5023008	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insGNPH	COSM51544	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	COSM6977296	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsGV	D770delinsGV	NEGATIVE	complex
EGFR exon 20 insertion	p.N771_P772dup	MAN123	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.D770_N771delinsAGH	MATNV09	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insPNP	OMINDEL1087	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGNP	OMINDEL1088	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771delinsGGT	OMINDEL1090	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsEF	OMINDEL1091	NEGATIVE	ins
EGFR exon 20 insertion	p.D770delinsEG	OMINDEL1092	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
EGFR exon 20 insertion	p.D770_N771insP	OMINDEL1096	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_V774dup	OMINDEL1097	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insANP	OMINDEL1098	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_H773insNPNP	OMINDEL1101	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1102	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1103	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insSNP	OMINDEL1104	NEGATIVE	ins

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.P772_H773insTPNP	OMINDEL1106	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTPNP	OMINDEL1107	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1108	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTRNP	OMINDEL1109	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insYPNP	OMINDEL1110	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_P772delinsASVDNR	OMINDEL1801	NEGATIVE	complex
EGFR exon 20 insertion	p.D770delinsAS	OMINDEL1810	NEGATIVE	complex
EGFR exon 20 insertion	p.D770_N771insVDSVDNP	OMINDEL953	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	COSM12378	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insGT	COSM1238029	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insSVE	COSM1651743	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insAPW	COSM20886	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_N771insSVE	D770_N771insSVE	NEGATIVE	ins
EGFR exon 20 insertion	p.D770_N771insG	MATNV05	NEGATIVE	ins
EGFR exon 20	p.D770_N771delinsERG	MATNV06	NEGATIVE	complex

Insertion

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.N771_V774dup	OMINDEL1112	NEGATIVE	ins
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.P772_H773insTRNP	OMINDEL1116	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_N771insF	OMINDEL1812	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insRNP	OMINDEL1813	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	COSM12380	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insH	COSM1238031	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSGH	COSM1651744	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insRH	COSM166390	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsGF	COSM18431	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsTH	COSM22946	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSH	COSM24434	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTHP	COSM48923	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsVH	COSM5023007	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGY	COSM53189	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSTH	COSM6920147	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insHH	COSM6931207	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsPH	MATNV04	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsHH	OMINDEL1118	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGD	OMINDEL1119	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1120	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1121	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insHPH	OMINDEL1122	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1123	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKH	OMINDEL1124	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1125	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPPH	OMINDEL1126	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1127	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRD	OMINDEL1128	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1129	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsRH	OMINDEL1130	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.N771delinsSPH	OMINDEL1131	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1132	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL1133	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1134	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insGHP	OMINDEL1135	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.N771delinsKPH	OMINDEL1188	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsCH	OMINDEL1814	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsSQRGH	OMINDEL1815	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsHHR	OMINDEL1816	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsGC	OMINDEL904	NEGATIVE	complex
EGFR exon 20 insertion	p.P772_H773insGDP	OMINDEL954	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insPHPH	p.H773_V774insPHPH_2	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772delinsSVDNR	COSM13554	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKL	COSM6438147	NEGATIVE	ins

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.N771_P772insT	OMINDEL1138	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insL	OMINDEL1139	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsSVDS	OMINDEL1140	NEGATIVE	complex
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1141	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKG	OMINDEL1142	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_P772insG	OMINDEL1143	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1144	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1145	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1146	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTTP	OMINDEL1183	NEGATIVE	ins
EGFR exon 20 insertion	p.N771delinsKPP	OMINDEL955	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_C775dup	OMINDEL956	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_V774dup	COSM6845098	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.N771_P772insL	OMINDEL1149	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.H773_V774insAPH	OMINDEL1150	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.H773_V774insTH	OMINDEL1151	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insT	OMINDEL1152	NEGATIVE	ins
EGFR exon 20 insertion	p.H773dup	COSM12377	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774dup	COSM12389	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insNH	COSM5023006	NEGATIVE	ins
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_H773insL	OMINDEL1155	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insR	OMINDEL1156	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insTP	COSM12388	NEGATIVE	complex
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.H773_V774insPHPH	H773_V774insPHPH	NEGATIVE	complex
EGFR exon 20 insertion	p.H773_V774insGH	OMINDEL1157	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1158	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773insQ	OMINDEL1159	NEGATIVE	ins
EGFR exon 20 insertion	p.P772_H773dup	OMINDEL1184	NEGATIVE	ins
EGFR exon 20 insertion	p.N771_H773dup	OMINDEL1811	NEGATIVE	complex
EGFR exon 20 insertion	p.H773delinsPNPY	COSM1735761	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsRY	MATNV08	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	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.H773delinsPNPY	OMINDEL1165	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1166	NEGATIVE	ins
EGFR exon 20 insertion	p.H773delinsQY	OMINDEL1167	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insY	OMINDEL1168	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insTQPP	COSM3727813	NEGATIVE	ins
EGFR exon 20 insertion	p.H773_V774insHV	OMINDEL1169	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
EGFR exon20 insertion	p.H773_V774insQ	COSM131552	NEGATIVE	ins
EGFR exon20 insertion	p.V774_C775insPR	COSM4170223	NEGATIVE	ins
ERBB2 C311R	p.C311R	COSM35763	NEGATIVE	SNV
ERBB2 D769H	p.D769H	COSM13170	NEGATIVE	SNV
ERBB2 D769N	p.D769N	COSM1302747	NEGATIVE	SNV
ERBB2 D769Y	p.D769Y	COSM1251412	NEGATIVE	SNV
ERBB2 E321G	p.E321G	COSM35764	NEGATIVE	SNV
ERBB2 G309A	p.G309A	COSM436496	NEGATIVE	SNV
ERBB2 G309E	p.G309E	COSM94224	NEGATIVE	SNV
ERBB2 G660D	p.G660D	COSM4681497	NEGATIVE	SNV
ERBB2 G776A	p.G776A	OBENV03	NEGATIVE	SNV
ERBB2 G776C	p.G776C	COSM303938	NEGATIVE	SNV
ERBB2 G776L	p.G776L	COSM144255	NEGATIVE	mnp
ERBB2 G776R	p.G776R	OM3421	NEGATIVE	SNV
ERBB2 G776S	p.G776S	COSM685	NEGATIVE	SNV
ERBB2 G776V	p.G776V	COSM18609	NEGATIVE	SNV
ERBB2 I767M	p.I767M	COSM51317	NEGATIVE	SNV
ERBB2 L755A	p.L755A	COSM6906940	NEGATIVE	mnp
ERBB2 L755F	p.L755F	OBENV01	NEGATIVE	SNV
ERBB2 L755F	p.L755F	OBENV02	NEGATIVE	SNV
ERBB2 L755M	p.L755M	COSM1205571	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 L755S	p.L755S	COSM14060	NEGATIVE	SNV
ERBB2 L755W	p.L755W	COSM436499	NEGATIVE	SNV
ERBB2 L755_T759del	p.L755_T759del	COSM7345568	NEGATIVE	del
ERBB2 L755_T759del	p.L755_T759del	COSM5029269	NEGATIVE	del
ERBB2 L869R	p.L869R	COSM249793	NEGATIVE	SNV
ERBB2 R678Q	p.R678Q	COSM436498	NEGATIVE	SNV
ERBB2 R868W	p.R868W	COSM289684	NEGATIVE	SNV
ERBB2 R896C	p.R896C	COSM14066	NEGATIVE	SNV
ERBB2 R896H	p.R896H	COSM119971	NEGATIVE	SNV
ERBB2 S310F	p.S310F	COSM48358	NEGATIVE	SNV
ERBB2 S310P	p.S310P	COSM1563863	NEGATIVE	SNV
ERBB2 S310Y	p.S310Y	COSM6906440	NEGATIVE	mnp
ERBB2 S310Y	p.S310Y	COSM94225	NEGATIVE	SNV
ERBB2 T733I	p.T733I	COSM14059	NEGATIVE	SNV
ERBB2 T798I	p.T798I	OM3210	NEGATIVE	SNV
ERBB2 V659E	p.V659E	COSM3724566	NEGATIVE	mnp
ERBB2 V659E	p.V659E	COSM6503262	NEGATIVE	mnp
ERBB2 V777A	p.V777A	COSM35496	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM14062	NEGATIVE	SNV
ERBB2 V777L	p.V777L	COSM436500	NEGATIVE	SNV
ERBB2 V777M	p.V777M	COSM14064	NEGATIVE	SNV
ERBB2 V842E	p.V842E	COSM6005745	NEGATIVE	SNV
ERBB2 V842I	p.V842I	COSM14065	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.Y772_V773insMMAY	OMINDEL820	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.A775_G776insVVMA	SPRNV01	NEGATIVE	ins
ERBB2 exon 20 insertion	p.M774dup	OMINDEL823	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insYLMA	SPRNV02	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_G776insV	MAN307	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	MAN300	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.A775_G776insV	MAN308	NEGATIVE	ins
ERBB2 exon 20 insertion	p.A775_G776insV	MAN309	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12552	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM12553	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsLC	COSM12554	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM19875	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	COSM20895	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsVC	COSM5235349	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsAVGC	COSM6907033	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVC	COSM85995	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776_V777delinsSVA	G776_V777delinsSVA_3	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_S779insCVG	OMINDEL1179	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsIC	OMINDEL826	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G776delinsLC	SPRNV03	NEGATIVE	complex
ERBB2 exon 20 insertion	p.V777_S779dup	COSM681	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G776delinsVV	COSM6977433	NEGATIVE	complex
ERBB2 exon 20 insertion	p.V777dup	COSM9869865	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB2 exon 20 insertion	p.G776_G778delinsVVGD	OMINDEL1189	NEGATIVE	complex
ERBB2 exon 20 insertion	p.G778_P780dup	COSM12555	NEGATIVE	ins
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	COSM303948	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_P780dup	COSM6865893	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPG	COSM7449770	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insCPG	OMINDEL1180	NEGATIVE	ins
ERBB2 exon 20 insertion	p.V777_G778insC	OMINDEL827	NEGATIVE	ins
ERBB2 exon 20 insertion	p.G778_S779insLPS	COSM5802314	NEGATIVE	ins
ERBB3 D297H	p.D297H	COSM1935392	NEGATIVE	SNV
ERBB3 D297N	p.D297N	COSM941489	NEGATIVE	SNV
ERBB3 D297V	p.D297V	COSM941490	NEGATIVE	SNV
ERBB3 D297Y	p.D297Y	COSM160822	NEGATIVE	SNV
ERBB3 E332K	p.E332K	COSM254677	NEGATIVE	SNV
ERBB3 E332Q	p.E332Q	COSM4838033	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM5413554	NEGATIVE	SNV
ERBB3 G284R	p.G284R	COSM941488	NEGATIVE	SNV
ERBB3 M60K	p.M60K	COSM254678	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
ERBB3 M60L	p.M60L	COSM1606366	NEGATIVE	SNV
ERBB3 M60R	p.M60R	COSM941484	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM122890	NEGATIVE	SNV
ERBB3 M91I	p.M91I	COSM1299636	NEGATIVE	SNV
ERBB3 R103G	p.R103G	COSM6907312	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM160824	NEGATIVE	SNV
ERBB3 V104L	p.V104L	COSM191840	NEGATIVE	SNV
ERBB3 V104M	p.V104M	COSM172423	NEGATIVE	SNV
ERBB4 R711C	p.R711C	COSM160827	NEGATIVE	SNV
ERBB4 R711L	p.R711L	OM3150	NEGATIVE	SNV
ESR1 D538G	p.D538G	COSM94250	NEGATIVE	SNV
ESR1 E380Q	p.E380Q	COSM3829320	NEGATIVE	SNV
ESR1 K303R	p.K303R	COSM4745827	NEGATIVE	SNV
ESR1 L536Q	p.L536Q	COSM4766050	NEGATIVE	mnp
ESR1 L536R	p.L536R	COSM4774826	NEGATIVE	SNV
ESR1 P535H	p.P535H	BT24	NEGATIVE	SNV
ESR1 P535T	p.P535T	COSM5666097	NEGATIVE	SNV
ESR1 R555C	p.R555C	BT32	NEGATIVE	SNV
ESR1 S463P	p.S463P	COSM4771561	NEGATIVE	SNV
ESR1 V392I	p.V392I	COSM1545070	NEGATIVE	SNV
ESR1 Y537C	p.Y537C	COSM1074637	NEGATIVE	SNV
ESR1 Y537N	p.Y537N	COSM1074635	NEGATIVE	SNV
ESR1 Y537S	p.Y537S	COSM1074639	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR1 K687E	p.K687E	COSM35673	NEGATIVE	SNV
FGFR1 K687M	p.K687M	COSM4771556	NEGATIVE	SNV
FGFR1 K687N	p.K687N	COSM6578115	NEGATIVE	SNV
FGFR1 V592M	p.V592M	COSM6840954	NEGATIVE	SNV
FGFR2 A314D	p.A314D	COSM49171	NEGATIVE	SNV
FGFR2 A315T	p.A315T	COSM30777	NEGATIVE	SNV
FGFR2 A648T	p.A648T	COSM29836	NEGATIVE	SNV
FGFR2 C382G	p.C382G	COSM6197913	NEGATIVE	SNV
FGFR2 C382R	p.C382R	COSM36906	NEGATIVE	SNV
FGFR2 C382Y	p.C382Y	COSM915493	NEGATIVE	SNV
FGFR2 E565A	p.E565A	OM3292	NEGATIVE	SNV
FGFR2 E565G	p.E565G	OM3563	NEGATIVE	SNV
FGFR2 G613V	p.G613V	COSM94318	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM1474415	NEGATIVE	SNV
FGFR2 H544Q	p.H544Q	COSM48382	NEGATIVE	SNV
FGFR2 I547V	p.I547V	COSM36913	NEGATIVE	SNV
FGFR2 K310R	p.K310R	COSM36901	NEGATIVE	SNV
FGFR2 K641R	p.K641R	COSM5010388	NEGATIVE	SNV
FGFR2 K659E	p.K659E	COSM36909	NEGATIVE	SNV
FGFR2 K659M	p.K659M	COSM49175	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM49173	NEGATIVE	SNV
FGFR2 K659N	p.K659N	COSM683054	NEGATIVE	SNV
FGFR2 L617M	p.L617M	OM3562	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR2 L617V	p.L617V	OM3291	NEGATIVE	SNV
FGFR2 M391R	p.M391R	COSM36907	NEGATIVE	SNV
FGFR2 N549D	p.N549D	COSM4604460	NEGATIVE	SNV
FGFR2 N549H	p.N549H	COSM250083	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36902	NEGATIVE	SNV
FGFR2 N549K	p.N549K	COSM36912	NEGATIVE	SNV
FGFR2 N549S	p.N549S	COSM3665553	NEGATIVE	SNV
FGFR2 N549T	p.N549T	COSM6952624	NEGATIVE	SNV
FGFR2 P253L	p.P253L	COSM537801	NEGATIVE	SNV
FGFR2 P253R	p.P253R	COSM49170	NEGATIVE	SNV
FGFR2 P253S	p.P253S	COSM1346291	NEGATIVE	SNV
FGFR2 S252W	p.S252W	COSM36903	NEGATIVE	SNV
FGFR2 V395D	p.V395D	COSM250081	NEGATIVE	mnp
FGFR2 V564F	p.V564F	OM3293	NEGATIVE	SNV
FGFR2 V564I	p.V564I	OM3247	NEGATIVE	SNV
FGFR2 V564L	p.V564L	OM3564	NEGATIVE	SNV
FGFR2 V679I	p.V679I	COSM4822956	NEGATIVE	SNV
FGFR2 Y375C	p.Y375C	COSM36904	NEGATIVE	SNV
FGFR2 Y375H	p.Y375H	COSM1560916	NEGATIVE	SNV
FGFR3 A391E	p.A391E	COSM721	NEGATIVE	SNV
FGFR3 A391T	p.A391T	OM3151	NEGATIVE	SNV
FGFR3 A391V	p.A391V	COSM1737909	NEGATIVE	SNV
FGFR3 D641G	p.D641G	COSM1428721	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR3 D641N	p.D641N	COSM29438	NEGATIVE	SNV
FGFR3 G370C	p.G370C	COSM716	NEGATIVE	SNV
FGFR3 G370D	p.G370D	COSM5966955	NEGATIVE	SNV
FGFR3 G380E	p.G380E	COSM51545	NEGATIVE	SNV
FGFR3 G380R	p.G380R	COSM24842	NEGATIVE	SNV
FGFR3 K650E	p.K650E	COSM719	NEGATIVE	SNV
FGFR3 K650M	p.K650M	COSM720	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM1428730	NEGATIVE	SNV
FGFR3 K650N	p.K650N	COSM3993568	NEGATIVE	SNV
FGFR3 K650Q	p.K650Q	COSM726	NEGATIVE	SNV
FGFR3 K650R	p.K650R	OM3224	NEGATIVE	SNV
FGFR3 K650T	p.K650T	COSM731	NEGATIVE	SNV
FGFR3 K715M	p.K715M	COSM732992	NEGATIVE	SNV
FGFR3 R248C	p.R248C	COSM714	NEGATIVE	SNV
FGFR3 R248H	p.R248H	COSM1757310	NEGATIVE	SNV
FGFR3 R399C	p.R399C	COSM296687	NEGATIVE	SNV
FGFR3 S249C	p.S249C	COSM715	NEGATIVE	SNV
FGFR3 S249T	p.S249T	COSM29431	NEGATIVE	SNV
FGFR3 S371C	p.S371C	COSM17461	NEGATIVE	SNV
FGFR3 V553M	p.V553M	OM3223	NEGATIVE	SNV
FGFR3 V555M	p.V555M	COSM303914	NEGATIVE	SNV
FGFR3 V677I	p.V677I	COSM178247	NEGATIVE	SNV
FGFR3 Y373C	p.Y373C	COSM718	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FGFR4 V550E	p.V550E	COSM9215998	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM1066172	NEGATIVE	SNV
FGFR4 V550L	p.V550L	COSM3738141	NEGATIVE	SNV
FGFR4 V550M	p.V550M	COSM6163	NEGATIVE	SNV
FLT3 D835A	p.D835A	COSM27650	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM787	NEGATIVE	SNV
FLT3 D835E	p.D835E	COSM788	NEGATIVE	SNV
FLT3 D835F	p.D835F	COSM19844	NEGATIVE	mnp
FLT3 D835G	p.D835G	COSM1366188	NEGATIVE	SNV
FLT3 D835H	p.D835H	COSM785	NEGATIVE	SNV
FLT3 D835K	p.D835K	OM3570	NEGATIVE	mnp
FLT3 D835K	p.D835K	OM3571	NEGATIVE	mnp
FLT3 D835L	p.D835L	OM3572	NEGATIVE	mnp
FLT3 D835N	p.D835N	COSM789	NEGATIVE	SNV
FLT3 D835V	p.D835V	COSM784	NEGATIVE	SNV
FLT3 D835Y	p.D835Y	COSM783	NEGATIVE	SNV
FLT3 D835_I836del	p.D835_I836del	COSM5879385	NEGATIVE	del
FLT3 D835_I836delinsV	p.D835_I836delinsV	COSM27651	NEGATIVE	del
FLT3 D835del	p.D835del	COSM854	NEGATIVE	del
FLT3 D839E	p.D839E	COSM5945287	NEGATIVE	SNV
FLT3 D839G	p.D839G	COSM1166729	NEGATIVE	SNV
FLT3 D839H	p.D839H	COSM3399304	NEGATIVE	SNV
FLT3 D839N	p.D839N	COSM6005768	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
FLT3 I836D	p.I836D	OM3569	NEGATIVE	mnp
FLT3 I836L	p.I836L	COSM19649	NEGATIVE	SNV
FLT3 I836S	p.I836S	COSM19843	NEGATIVE	SNV
FLT3 I836T	p.I836T	COSM5879386	NEGATIVE	SNV
FLT3 I836del	p.I836del	COSM19836	NEGATIVE	del
GNAS Q227E	p.Q227E	COSM4962873	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM27900	NEGATIVE	SNV
GNAS Q227H	p.Q227H	COSM4168960	NEGATIVE	SNV
GNAS Q227K	p.Q227K	COSM28618	NEGATIVE	SNV
GNAS Q227L	p.Q227L	COSM27888	NEGATIVE	SNV
GNAS Q227R	p.Q227R	COSM27896	NEGATIVE	SNV
GNAS R201C	p.R201C	COSM27887	NEGATIVE	SNV
GNAS R201G	p.R201G	COSM1757313	NEGATIVE	SNV
GNAS R201H	p.R201H	COSM27895	NEGATIVE	SNV
GNAS R201L	p.R201L	COSM99221	NEGATIVE	SNV
GNAS R201S	p.R201S	COSM27899	NEGATIVE	SNV
HRAS G12A	p.G12A	COSM485	NEGATIVE	SNV
HRAS G12C	p.G12C	COSM481	NEGATIVE	SNV
HRAS G12D	p.G12D	COSM484	NEGATIVE	SNV
HRAS G12N	p.G12N	COSM4517473	NEGATIVE	mnp
HRAS G12R	p.G12R	COSM482	NEGATIVE	SNV
HRAS G12S	p.G12S	COSM480	NEGATIVE	SNV
HRAS G12V	p.G12V	COSM483	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
HRAS G13C	p.G13C	COSM488	NEGATIVE	SNV
HRAS G13D	p.G13D	COSM490	NEGATIVE	SNV
HRAS G13R	p.G13R	COSM486	NEGATIVE	SNV
HRAS G13S	p.G13S	COSM487	NEGATIVE	SNV
HRAS G13V	p.G13V	COSM489	NEGATIVE	SNV
HRAS Q61E	p.Q61E	COSM497	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM502	NEGATIVE	SNV
HRAS Q61H	p.Q61H	COSM503	NEGATIVE	SNV
HRAS Q61K	p.Q61K	COSM496	NEGATIVE	SNV
HRAS Q61L	p.Q61L	COSM52978	NEGATIVE	mnp
HRAS Q61L	p.Q61L	COSM498	NEGATIVE	SNV
HRAS Q61P	p.Q61P	COSM500	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM33695	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM501	NEGATIVE	mnp
HRAS Q61R	p.Q61R	COSM499	NEGATIVE	SNV
HRAS Q61R	p.Q61R	COSM52979	NEGATIVE	mnp
IDH1 G70D	p.G70D	COSM96531	NEGATIVE	SNV
IDH1 R132C	p.R132C	COSM28747	NEGATIVE	SNV
IDH1 R132G	p.R132G	COSM28749	NEGATIVE	SNV
IDH1 R132H	p.R132H	COSM86993	NEGATIVE	mnp
IDH1 R132H	p.R132H	COSM28746	NEGATIVE	SNV
IDH1 R132L	p.R132L	COSM28750	NEGATIVE	SNV
IDH1 R132Q	p.R132Q	OM3221	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
IDH1 R132Q	p.R132Q	OM3222	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM1169216	NEGATIVE	mnp
IDH1 R132S	p.R132S	COSM28748	NEGATIVE	SNV
IDH1 R132V	p.R132V	COSM28751	NEGATIVE	mnp
IDH1 S280F	p.S280F	OM3315	NEGATIVE	SNV
IDH2 I319M	p.I319M	OM3319	NEGATIVE	SNV
IDH2 Q316E	p.Q316E	OM3320	NEGATIVE	SNV
IDH2 R140G	p.R140G	COSM1737874	NEGATIVE	SNV
IDH2 R140L	p.R140L	COSM41875	NEGATIVE	SNV
IDH2 R140Q	p.R140Q	COSM41590	NEGATIVE	SNV
IDH2 R140W	p.R140W	COSM41877	NEGATIVE	SNV
IDH2 R172G	p.R172G	COSM33731	NEGATIVE	SNV
IDH2 R172K	p.R172K	COSM33733	NEGATIVE	SNV
IDH2 R172M	p.R172M	COSM33732	NEGATIVE	SNV
IDH2 R172N	p.R172N	COSM256046	NEGATIVE	mnp
IDH2 R172S	p.R172S	COSM133672	NEGATIVE	SNV
IDH2 R172S	p.R172S	COSM34090	NEGATIVE	SNV
IDH2 R172T	p.R172T	COSM1636997	NEGATIVE	SNV
IDH2 R172V	p.R172V	OM3327	NEGATIVE	mnp
IDH2 R172W	p.R172W	COSM34039	NEGATIVE	SNV
KEAP1 D422N	p.D422N	COSM710198	NEGATIVE	SNV
KEAP1 G333C	p.G333C	COSM1193323	NEGATIVE	SNV
KEAP1 M409*	p.M409*	COSM6916353	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 R320L	p.R320L	COSM2812614	NEGATIVE	SNV
KEAP1 R320P	p.R320P	COSM346494	NEGATIVE	SNV
KEAP1 R320Q	p.R320Q	COSM710195	NEGATIVE	SNV
KEAP1 R320W	p.R320W	COSM1524062	NEGATIVE	SNV
KEAP1 R415G	p.R415G	COSM6196677	NEGATIVE	SNV
KEAP1 R470C	p.R470C	COSM564847	NEGATIVE	SNV
KEAP1 R470G	p.R470G	OM3724	NEGATIVE	SNV
KEAP1 R470H	p.R470H	COSM1524064	NEGATIVE	SNV
KEAP1 R470S	p.R470S	COSM564846	NEGATIVE	SNV
KEAP1 R554Q	p.R554Q	COSM1524066	NEGATIVE	SNV
KEAP1 S243C	p.S243C	COSM710191	NEGATIVE	SNV
KEAP1 V155F	p.V155F	COSM710187	NEGATIVE	SNV
KEAP1 V167F	p.V167F	COSM710188	NEGATIVE	SNV
KEAP1 Y584*	p.Y584*	COSM6201646	NEGATIVE	ins
KEAP1 c.1531+1G>T	p.?	COSM6956065	NEGATIVE	SNV
KEAP1 c.1531+2T>G	p.?	OM3723	NEGATIVE	SNV
KEAP1 c.1532-1G>T	p.?	COSM6969960	NEGATIVE	SNV
KEAP1 c.1532-2A>G	p.?	COSM381459	NEGATIVE	SNV
KEAP1 c.1708+1G>T	p.?	COSM7738757	NEGATIVE	SNV
KEAP1 c.1708+2T>A	p.?	COSM6149754	NEGATIVE	SNV
KEAP1 c.1708_1708+1delinsTT	p.?	COSM6927565	NEGATIVE	mnp
KEAP1 c.1709-1G>T	p.?	COSM7724088	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KEAP1 c.1709-2A>G	p.?	COSM7088100	NEGATIVE	SNV
KEAP1 c.640-1G>A	p.?	COSM6972470	NEGATIVE	SNV
KEAP1 c.640-1G>C	p.?	OM3726	NEGATIVE	SNV
KEAP1 c.640-1G>T	p.?	OM3725	NEGATIVE	SNV
KEAP1 c.640-2A>C	p.?	OM3727	NEGATIVE	SNV
KIT A502_Y503dup	p.A502_Y503dup	COSM1326	NEGATIVE	ins
KIT A502_Y503insFA	p.A502_Y503insFA	COSM53306	NEGATIVE	ins
KIT A829P	p.A829P	COSM13172	NEGATIVE	SNV
KIT A829V	p.A829V	COSM133757	NEGATIVE	SNV
KIT D419_L421delinsF	p.D419_L421delinsF	COSM1152	NEGATIVE	complex
KIT D419_L421delinsH	p.D419_L421delinsH	COSM1578133	NEGATIVE	complex
KIT D419_R420del	p.D419_R420del	COSM1578132	NEGATIVE	del
KIT D419_R420insFF	p.D419_R420insFF	COSM36334	NEGATIVE	ins
KIT D419_R420insFL	p.D419_R420insFL	COSM36332	NEGATIVE	ins
KIT D419del	p.D419del	COSM29014	NEGATIVE	del
KIT D419del	p.D419del	COSM1600400	NEGATIVE	complex
KIT D816A	p.D816A	COSM24675	NEGATIVE	SNV
KIT D816E	p.D816E	COSM19285	NEGATIVE	SNV
KIT D816E	p.D816E	COSM5749965	NEGATIVE	SNV
KIT D816F	p.D816F	COSM1312	NEGATIVE	mnp
KIT D816G	p.D816G	COSM12711	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1311	NEGATIVE	SNV
KIT D816H	p.D816H	COSM1600404	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT D816I	p.D816I	COSM21979	NEGATIVE	mnp
KIT D816N	p.D816N	COSM1313	NEGATIVE	SNV
KIT D816V	p.D816V	COSM1314	NEGATIVE	SNV
KIT D816Y	p.D816Y	COSM1310	NEGATIVE	SNV
KIT D820A	p.D820A	COSM133670	NEGATIVE	SNV
KIT D820E	p.D820E	COSM12709	NEGATIVE	SNV
KIT D820E	p.D820E	COSM19280	NEGATIVE	SNV
KIT D820G	p.D820G	COSM1316	NEGATIVE	SNV
KIT D820H	p.D820H	COSM22379	NEGATIVE	SNV
KIT D820N	p.D820N	COSM17947	NEGATIVE	SNV
KIT D820V	p.D820V	COSM1317	NEGATIVE	SNV
KIT D820Y	p.D820Y	COSM12710	NEGATIVE	SNV
KIT F522C	p.F522C	OM3237	NEGATIVE	SNV
KIT H697Y	p.H697Y	COSM36053	NEGATIVE	SNV
KIT I653T	p.I653T	COSM19326	NEGATIVE	SNV
KIT K550I	p.K550I	COSM1165	NEGATIVE	SNV
KIT K550N	p.K550N	COSM1167	NEGATIVE	SNV
KIT K550R	p.K550R	COSM1166	NEGATIVE	SNV
KIT K558N	p.K558N	OM3238	NEGATIVE	SNV
KIT K558N	p.K558N	COSM19216	NEGATIVE	SNV
KIT K642E	p.K642E	COSM1304	NEGATIVE	SNV
KIT K642N	p.K642N	COSM4125155	NEGATIVE	SNV
KIT K642Q	p.K642Q	COSM96871	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT L576F	p.L576F	COSM33966	NEGATIVE	SNV
KIT L576P	p.L576P	COSM1290	NEGATIVE	SNV
KIT L576R	p.L576R	COSM5945006	NEGATIVE	SNV
KIT N505I	p.N505I	OM3236	NEGATIVE	SNV
KIT N655K	p.N655K	COSM235682	NEGATIVE	SNV
KIT N655K	p.N655K	COSM4413464	NEGATIVE	SNV
KIT N655S	p.N655S	OM3302	NEGATIVE	SNV
KIT N655T	p.N655T	OM3303	NEGATIVE	SNV
KIT N680K	p.N680K	COSM5352270	NEGATIVE	SNV
KIT N822H	p.N822H	COSM1318	NEGATIVE	SNV
KIT N822I	p.N822I	COSM3604523	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1321	NEGATIVE	SNV
KIT N822K	p.N822K	COSM1322	NEGATIVE	SNV
KIT N822S	p.N822S	COSM33975	NEGATIVE	SNV
KIT N822Y	p.N822Y	COSM19109	NEGATIVE	SNV
KIT P838L	p.P838L	OM3240	NEGATIVE	SNV
KIT R796A	p.R796A	COSM1272091	NEGATIVE	mnp
KIT R796G	p.R796G	COSM51381	NEGATIVE	SNV
KIT R796K	p.R796K	COSM1600411	NEGATIVE	SNV
KIT R796P	p.R796P	COSM1272092	NEGATIVE	mnp
KIT S501_A502dup	p.S501_A502dup	COSM5702501	NEGATIVE	ins
KIT S501_A502dup	p.S501_A502dup	COSM219786	NEGATIVE	ins
KIT T417_D419delinsI	p.T417_D419delinsI	COSM1148	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT T417_D419delinsRG	p.T417_D419delinsRG	COSM1149	NEGATIVE	complex
KIT T670A	p.T670A	OM3304	NEGATIVE	SNV
KIT T670E	p.T670E	COSM19293	NEGATIVE	mnp
KIT T670I	p.T670I	COSM12708	NEGATIVE	SNV
KIT V530I	p.V530I	COSM1155	NEGATIVE	SNV
KIT V559A	p.V559A	COSM1255	NEGATIVE	SNV
KIT V559C	p.V559C	OM3239	NEGATIVE	mnp
KIT V559D	p.V559D	COSM1252	NEGATIVE	SNV
KIT V559G	p.V559G	COSM1253	NEGATIVE	SNV
KIT V559I	p.V559I	COSM1251	NEGATIVE	SNV
KIT V559K	p.V559K	COSM96869	NEGATIVE	mnp
KIT V559P	p.V559P	COSM1272086	NEGATIVE	mnp
KIT V560A	p.V560A	COSM36302	NEGATIVE	SNV
KIT V560D	p.V560D	COSM1257	NEGATIVE	SNV
KIT V560E	p.V560E	COSM1333	NEGATIVE	mnp
KIT V560G	p.V560G	COSM1260	NEGATIVE	SNV
KIT V654A	p.V654A	COSM12706	NEGATIVE	SNV
KIT V654L	p.V654L	COSM5941904	NEGATIVE	SNV
KIT V825A	p.V825A	COSM1323	NEGATIVE	SNV
KIT V825I	p.V825I	COSM19110	NEGATIVE	SNV
KIT W557C	p.W557C	COSM19108	NEGATIVE	SNV
KIT W557C	p.W557C	COSM133700	NEGATIVE	SNV
KIT W557G	p.W557G	COSM1221	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT W557R	p.W557R	COSM1216	NEGATIVE	SNV
KIT W557R	p.W557R	COSM1219	NEGATIVE	SNV
KIT W557S	p.W557S	COSM1227	NEGATIVE	SNV
KIT Y553C	p.Y553C	COSM4413463	NEGATIVE	SNV
KIT Y553N	p.Y553N	COSM133763	NEGATIVE	SNV
KIT Y570H	p.Y570H	COSM1430152	NEGATIVE	SNV
KIT Y578C	p.Y578C	COSM6503446	NEGATIVE	SNV
KIT Y823C	p.Y823C	COSM18682	NEGATIVE	SNV
KIT Y823D	p.Y823D	COSM18681	NEGATIVE	SNV
KIT Y823H	p.Y823H	COSM1651649	NEGATIVE	SNV
KIT Y823N	p.Y823N	COSM22378	NEGATIVE	SNV
KIT [K558S;V559S]	p.[K558S;V559S]	COSM28637	NEGATIVE	mnp
KIT [W557C;K558P]	p.[W557C;K558P]	COSM21978	NEGATIVE	mnp
KIT [W557C;K558T]	p.[W557C;K558T]	COSM28992	NEGATIVE	mnp
KIT [W557F;K558P]	p.[W557F;K558P]	COSM1224	NEGATIVE	mnp
KIT [W557S;K558S]	p.[W557S;K558S]	COSM96948	NEGATIVE	mnp
KIT exon 11 deletion	p.?	OMINDEL901	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1163	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1158	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1160	NEGATIVE	del
KIT exon 11 deletion	p.?	COSM1162	NEGATIVE	del
KIT exon 11 deletion	p.K550_V555delinsI	COSM1164	NEGATIVE	del
KIT exon 11 deletion	p.P551_E554del	COSM1172	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K550_V555delinsQ	COSM6938892	NEGATIVE	complex
KIT exon 11 deletion	p.K550_Q556delinsII	COSM1161	NEGATIVE	complex
KIT exon 11 deletion	p.P551_V555del	COSM1169	NEGATIVE	del
KIT exon 11 deletion	p.K550_W557delinsIL	COSM6936468	NEGATIVE	complex
KIT exon 11 deletion	p.K550_E554delinsI	COSM6953101	NEGATIVE	complex
KIT exon 11 deletion	p.K550_P551delinsI	COSM6971267	NEGATIVE	complex
KIT exon 11 deletion	p.K550_W557delinsIR	COSM6977972	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Q556del	COSM1170	NEGATIVE	del
KIT exon 11 deletion	p.P551del	COSM1171	NEGATIVE	del
KIT exon 11 deletion	p.P551_V555delinsL	COSM1174	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.P551_V555delinsTL	COSM30550	NEGATIVE	complex
KIT exon 11 deletion	p.P551_Y553delinsH	COSM6934157	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1177	NEGATIVE	del
KIT exon 11 deletion	p.M552_V555del	COSM6957862	NEGATIVE	del
KIT exon 11 deletion	p.M552_Y553del	COSM96881	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsL	COSM1178	NEGATIVE	complex
KIT exon 11 deletion	p.M552_Y553del	COSM1179	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557del	COSM1180	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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.M552_V555del	COSM18827	NEGATIVE	del
KIT exon 11 deletion	p.M552_K558del	OMINDEL902	NEGATIVE	complex
KIT exon 11 deletion	p.M552_V555delinsI	COSM1185	NEGATIVE	del
KIT exon 11 deletion	p.M552_W557delinsR	COSM133636	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.M552_Y553delinsN	COSM96863	NEGATIVE	del
KIT exon 11 deletion	p.M552_Q556delinsK	COSM96868	NEGATIVE	del
KIT exon 11 deletion	p.M552_E561delinsK	COSM96880	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_K558del	COSM27909	NEGATIVE	del
KIT exon 11 deletion	p.M552_E554delinsI	COSM6944981	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559del	COSM1191	NEGATIVE	del
KIT exon 11 deletion	p.Y553_E554del	COSM6923694	NEGATIVE	del
KIT exon 11 deletion	p.Y553_V559delinsE	COSM96946	NEGATIVE	complex
KIT exon 11 deletion	p.E554_N564del	COSM96947	NEGATIVE	del
KIT exon 11 deletion	p.E554_V559del	OMINDEL701	NEGATIVE	del
KIT exon 11 deletion	p.E554_K558del	COSM1192	NEGATIVE	del
KIT exon 11 deletion	p.V555_E562del	COSM1194	NEGATIVE	del
KIT exon 11 deletion	p.E554_E562del	COSM36310	NEGATIVE	del
KIT exon 11 deletion	p.V555_I563del	COSM1196	NEGATIVE	del
KIT exon 11 deletion	p.V555_G565del	COSM37027	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.Q556_V560del	COSM1198	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559del	COSM1199	NEGATIVE	del
KIT exon 11 deletion	p.V555_Q556del	COSM1201	NEGATIVE	del
KIT exon 11 deletion	p.V555_V560del	COSM1202	NEGATIVE	del
KIT exon 11 deletion	p.V555_N566delinsD	COSM96870	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560del	COSM1204	NEGATIVE	del
KIT exon 11 deletion	p.Q556_E561del	COSM1206	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558del	COSM23418	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.W557_K558del	COSM6986076	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsF	COSM1207	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558del	COSM1217	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561del	COSM1332	NEGATIVE	del
KIT exon 11 deletion	p.Q556_W557delinsR	COSM133642	NEGATIVE	del
KIT exon 11 deletion	p.Q556_I563delinsL	COSM1651755	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsTTF	COSM27904	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E562del	COSM36312	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V560delinsH	COSM1213	NEGATIVE	del
KIT exon 11 deletion	p.W557del	COSM1220	NEGATIVE	del
KIT exon 11 deletion	p.Q556_V559delinsH	COSM133641	NEGATIVE	del
KIT exon 11 deletion	p.Q556_K558delinsR	COSM28993	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V560delinsRG	COSM6954545	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.Q556_E561delinsHH	COSM1208	NEGATIVE	complex
KIT exon 11 deletion	p.Q556_V559delinsHT	COSM1214	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559del	COSM1218	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560del	COSM1223	NEGATIVE	del
KIT exon 11 deletion	p.W557_E561delinsPS	COSM6922994	NEGATIVE	complex
KIT exon 11 deletion	p.W557_E561delinsQ	COSM6954755	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsI	COSM1222	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V559delinsF	COSM1226	NEGATIVE	del
KIT exon 11 deletion	p.W557_V560delinsF	COSM1230	NEGATIVE	del
KIT exon 11 deletion	p.W557_K558delinsE	COSM24748	NEGATIVE	complex
KIT exon 11 deletion	p.W557_K558delinsQ	COSM6946777	NEGATIVE	complex
KIT exon 11 deletion	p.W557_V560delinsC	COSM1232	NEGATIVE	del
KIT exon 11 deletion	p.W557_V559delinsC	COSM1233	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.K558_V560del	COSM1234	NEGATIVE	del
KIT exon 11 deletion	p.K558_V559del	COSM1235	NEGATIVE	del
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.W557_K558delinsC	COSM96887	NEGATIVE	complex
KIT exon 11 deletion	p.K558_G565delinsR	COSM1240	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsI	COSM1241	NEGATIVE	del
KIT exon 11 deletion	p.V559_E561del	COSM1250	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KIT exon 11 deletion	p.K558_Y570delinsN	COSM28991	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsL	COSM6940807	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V560delinsM	COSM1242	NEGATIVE	complex
KIT exon 11 deletion	p.K558_V559delinsN	COSM1244	NEGATIVE	del
KIT exon 11 deletion	p.K558_V560delinsN	COSM27069	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM6971633	NEGATIVE	del
KIT exon 11 deletion	p.V560del	COSM1247	NEGATIVE	del
KIT exon 11 deletion	p.V559_V560del	COSM1248	NEGATIVE	del
KIT exon 11 deletion	p.V559_G565del	COSM1249	NEGATIVE	del
KIT exon 11 deletion	p.V559_I571del	COSM36301	NEGATIVE	del
KIT exon 11 deletion	p.V560_I563del	COSM18897	NEGATIVE	del
KIT exon 11 deletion	p.V559_N566delinsD	COSM6977976	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1262	NEGATIVE	del
KIT exon 11 deletion	p.E562del	COSM1263	NEGATIVE	del
KIT exon 11 deletion	p.E562_V569delinsD	COSM1267	NEGATIVE	del
KIT exon 11 deletion	p.I563_D572del	COSM1268	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.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

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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.V569_D572del	COSM1282	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 deletion	p.I571_Y578del	OMINDEL1186	NEGATIVE	del
KIT exon 11 deletion	p.I571_L576del	COSM133754	NEGATIVE	del
KIT exon 11 deletion	p.Q575del	COSM96875	NEGATIVE	del
KIT exon 11 deletion	p.Q575_L576del	COSM6987753	NEGATIVE	del
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.Q575_P577delinsT	COSM96865	NEGATIVE	complex
KIT exon 11 deletion	p.L576del	COSM1289	NEGATIVE	del
KIT exon 11 deletion	p.P577_Y578del	COSM1291	NEGATIVE	del
KIT exon 11 deletion	p.P577del	COSM1684822	NEGATIVE	del
KIT exon 11 deletion	p.P577_D579del	COSM98379	NEGATIVE	del
KIT exon 11 deletion	p.D579del	COSM1294	NEGATIVE	del
KIT exon 11 deletion	p.H580del	COSM1295	NEGATIVE	del
KIT exon 11 insertion	p.K558delinsNP	COSM21976	NEGATIVE	ins
KIT exon 11 insertion	p.K558delinsNP	COSM1245	NEGATIVE	complex
KIT exon 11 insertion	p.I571_D579dup	COSM23417	NEGATIVE	ins
KIT exon 11 insertion	p.D572_D579dup	COSM308544	NEGATIVE	ins

Variant Name	AA Change	Variant ID	Test Result	Variant Type
insertion				
KIT exon 11 insertion	p.P573_D579dup	COSM18668	NEGATIVE	ins
KIT exon 11 insertion	p.T574_D579dup	OMINDEL702	NEGATIVE	ins
KIT exon 11 insertion	p.Q575_L576dup	COSM1292	NEGATIVE	ins
KIT exon 11 insertion	p.Y578_P585dup	COSM41632	NEGATIVE	ins
KIT exon 11 insertion	p.P577_H580dup	COSM1296	NEGATIVE	ins
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
KRAS A146G	p.A146G	COSM1360828	NEGATIVE	SNV
KRAS A146P	p.A146P	COSM19905	NEGATIVE	SNV
KRAS A146S	p.A146S	COSM5967494	NEGATIVE	SNV
KRAS A146T	p.A146T	COSM19404	NEGATIVE	SNV
KRAS A146V	p.A146V	COSM19900	NEGATIVE	SNV
KRAS A59E	p.A59E	COSM547	NEGATIVE	SNV
KRAS A59G	p.A59G	COSM28518	NEGATIVE	SNV
KRAS A59S	p.A59S	COSM1235389	NEGATIVE	SNV
KRAS A59T	p.A59T	COSM546	NEGATIVE	SNV
KRAS G12A	p.G12A	COSM522	NEGATIVE	SNV
KRAS G12C	p.G12C	COSM516	NEGATIVE	SNV
KRAS G12D	p.G12D	COSM14209	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G12D	p.G12D	COSM521	NEGATIVE	SNV
KRAS G12E	p.G12E	COSM30566	NEGATIVE	mnp
KRAS G12E	p.G12E	COSM519	NEGATIVE	mnp
KRAS G12F	p.G12F	COSM512	NEGATIVE	mnp
KRAS G12I	p.G12I	COSM34144	NEGATIVE	mnp
KRAS G12L	p.G12L	COSM514	NEGATIVE	mnp
KRAS G12N	p.G12N	COSM13643	NEGATIVE	mnp
KRAS G12R	p.G12R	COSM518	NEGATIVE	SNV
KRAS G12S	p.G12S	COSM517	NEGATIVE	SNV
KRAS G12V	p.G12V	COSM515	NEGATIVE	mnp
KRAS G12V	p.G12V	COSM520	NEGATIVE	SNV
KRAS G12W	p.G12W	COSM36281	NEGATIVE	mnp
KRAS G12Y	p.G12Y	COSM25081	NEGATIVE	mnp
KRAS G13A	p.G13A	COSM533	NEGATIVE	SNV
KRAS G13C	p.G13C	COSM527	NEGATIVE	SNV
KRAS G13D	p.G13D	COSM531	NEGATIVE	mnp
KRAS G13D	p.G13D	COSM532	NEGATIVE	SNV
KRAS G13E	p.G13E	COSM30567	NEGATIVE	mnp
KRAS G13E	p.G13E	COSM87280	NEGATIVE	mnp
KRAS G13I	p.G13I	COSM525	NEGATIVE	mnp
KRAS G13N	p.G13N	COSM53283	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM526	NEGATIVE	mnp
KRAS G13R	p.G13R	COSM529	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
KRAS G13S	p.G13S	COSM528	NEGATIVE	SNV
KRAS G13V	p.G13V	COSM12721	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM530	NEGATIVE	mnp
KRAS G13V	p.G13V	COSM534	NEGATIVE	SNV
KRAS G15D	p.G15D	COSM539	NEGATIVE	SNV
KRAS G15S	p.G15S	COSM538	NEGATIVE	SNV
KRAS K117E	p.K117E	COSM6854421	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM19940	NEGATIVE	SNV
KRAS K117N	p.K117N	COSM28519	NEGATIVE	SNV
KRAS K117R	p.K117R	COSM4696721	NEGATIVE	SNV
KRAS Q61E	p.Q61E	COSM550	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM554	NEGATIVE	SNV
KRAS Q61H	p.Q61H	COSM555	NEGATIVE	SNV
KRAS Q61K	p.Q61K	COSM549	NEGATIVE	SNV
KRAS Q61L	p.Q61L	COSM553	NEGATIVE	SNV
KRAS Q61P	p.Q61P	COSM551	NEGATIVE	SNV
KRAS Q61R	p.Q61R	COSM1168052	NEGATIVE	mnp
KRAS Q61R	p.Q61R	COSM552	NEGATIVE	SNV
KRAS [G12=;G13C]	p.[G12=;G13C]	COSM87281	NEGATIVE	mnp
KRAS [G13D;V14I]	p.[G13D;V14I]	COSM30568	NEGATIVE	mnp
KRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM87298	NEGATIVE	mnp
MAP2K1 C121S	p.C121S	COSM555601	NEGATIVE	SNV
MAP2K1 C121S	p.C121S	COSM1315829	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 E203K	p.E203K	COSM232755	NEGATIVE	SNV
MAP2K1 E203V	p.E203V	COSM3386991	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3249	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	COSM1235480	NEGATIVE	SNV
MAP2K1 F129L	p.F129L	OM3250	NEGATIVE	SNV
MAP2K1 F53C	p.F53C	COSM1562837	NEGATIVE	SNV
MAP2K1 F53I	p.F53I	COSM3503329	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM555604	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM1725008	NEGATIVE	SNV
MAP2K1 F53L	p.F53L	COSM2257208	NEGATIVE	SNV
MAP2K1 F53S	p.F53S	ClinVar13350	NEGATIVE	SNV
MAP2K1 F53V	p.F53V	COSM5077832	NEGATIVE	SNV
MAP2K1 F53Y	p.F53Y	COSM5588244	NEGATIVE	SNV
MAP2K1 G128D	p.G128D	COSM1374186	NEGATIVE	SNV
MAP2K1 G128V	p.G128V	COSM1517232	NEGATIVE	SNV
MAP2K1 K57E	p.K57E	COSM5369532	NEGATIVE	SNV
MAP2K1 K57M	p.K57M	MAN124	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM1235478	NEGATIVE	SNV
MAP2K1 K57N	p.K57N	COSM5520914	NEGATIVE	SNV
MAP2K1 K57Q	p.K57Q	OM3155	NEGATIVE	SNV
MAP2K1 K57T	p.K57T	COSM4756761	NEGATIVE	SNV
MAP2K1 L115P	p.L115P	OM3248	NEGATIVE	SNV
MAP2K1 L215P	p.L215P	OM3252	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MAP2K1 P124L	p.P124L	COSM1315861	NEGATIVE	SNV
MAP2K1 P124Q	p.P124Q	COSM1167912	NEGATIVE	SNV
MAP2K1 P124S	p.P124S	COSM235614	NEGATIVE	SNV
MAP2K1 Q56P	p.Q56P	COSM1235481	NEGATIVE	SNV
MAP2K1 V211D	p.V211D	OM3251	NEGATIVE	SNV
MAP2K1 V60E	p.V60E	COSM2257209	NEGATIVE	SNV
MAP2K1 V60M	p.V60M	OM3576	NEGATIVE	SNV
MAP2K2 C125S	p.C125S	COSM5855814	NEGATIVE	SNV
MAP2K2 N126D	p.N126D	COSM1731742	NEGATIVE	SNV
MAP2K2 V35M	p.V35M	COSM5855836	NEGATIVE	SNV
MET H1112L	p.H1112L	COSM698	NEGATIVE	SNV
MET H1112R	p.H1112R	COSM703	NEGATIVE	SNV
MET H1112Y	p.H1112Y	COSM696	NEGATIVE	SNV
MET Y1021F	p.Y1021F	COSM339515	NEGATIVE	SNV
MET Y1021S	p.Y1021S	COSM7450124	NEGATIVE	SNV
MET c.2942-20_2942-19delinsAGAG	p.?	OMINDEL804	NEGATIVE	complex
MET c.2942-21_2942-20delinsGC	p.?	COSM8631602	NEGATIVE	mnp
MET c.2942-29_2942-28delinsGG	p.?	OM3305	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM29635	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL802	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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.?	OMINDEL674	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM49015	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6980941	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL709	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6056832	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL675	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL676	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL710	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.?	COSM6057022	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL640	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL697	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6947399	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	OMINDEL642	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL677	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL679	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL712	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	COSM6948341	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL680	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL681	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.?	COSM6976260	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL684	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL706	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL685	NEGATIVE	ins
MET exon 14 skipping (DNA)	p.?	OMINDEL808	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OM3177	NEGATIVE	SNV
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

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6057025	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL686	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6937267	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6977526	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL654	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL655	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL811	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL656	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL658	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL688	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL687	NEGATIVE	complex
MET exon 14 skipping (DNA)	p.?	OMINDEL689	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL662	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.?	COSM6965225	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL691	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL812	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	OMINDEL813	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.?	OMINDEL814	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.?	OMINDEL694	NEGATIVE	del
MET exon 14 skipping (DNA)	p.D1028IfsTer19	COSM24687	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.D1028Y	COSM6917314	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.D1028N	COSM6924587	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OMINDEL695	NEGATIVE	complex
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.?	COSM6928344	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6940825	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
MET exon 14 skipping (DNA)	p.?	COSM6983140	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	OMINDEL815	NEGATIVE	del
MET exon 14 skipping (DNA)	p.?	COSM6108460	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	COSM6947995	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3185	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3217	NEGATIVE	mnp
MET exon 14 skipping (DNA)	p.?	COSM744044	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3186	NEGATIVE	SNV
MET exon 14 skipping (DNA)	p.?	OM3188	NEGATIVE	SNV
NRAS A146T	p.A146T	COSM27174	NEGATIVE	SNV
NRAS A146V	p.A146V	COSM4170228	NEGATIVE	SNV
NRAS A59D	p.A59D	COSM253327	NEGATIVE	SNV
NRAS A59T	p.A59T	COSM578	NEGATIVE	SNV
NRAS G12A	p.G12A	COSM565	NEGATIVE	SNV
NRAS G12C	p.G12C	COSM562	NEGATIVE	SNV
NRAS G12D	p.G12D	COSM564	NEGATIVE	SNV
NRAS G12E	p.G12E	COSM144577	NEGATIVE	mnp
NRAS G12N	p.G12N	COSM12723	NEGATIVE	mnp
NRAS G12P	p.G12P	COSM559	NEGATIVE	mnp
NRAS G12R	p.G12R	COSM561	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS G12S	p.G12S	COSM563	NEGATIVE	SNV
NRAS G12V	p.G12V	COSM566	NEGATIVE	SNV
NRAS G12Y	p.G12Y	COSM560	NEGATIVE	mnp
NRAS G13A	p.G13A	COSM575	NEGATIVE	SNV
NRAS G13C	p.G13C	COSM570	NEGATIVE	SNV
NRAS G13D	p.G13D	COSM573	NEGATIVE	SNV
NRAS G13N	p.G13N	COSM24668	NEGATIVE	mnp
NRAS G13R	p.G13R	COSM569	NEGATIVE	SNV
NRAS G13S	p.G13S	COSM571	NEGATIVE	SNV
NRAS G13V	p.G13V	COSM572	NEGATIVE	mnp
NRAS G13V	p.G13V	COSM574	NEGATIVE	SNV
NRAS G13Y	p.G13Y	COSM568	NEGATIVE	mnp
NRAS K117N	p.K117N	MAN13	NEGATIVE	SNV
NRAS Q61E	p.Q61E	COSM581	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM585	NEGATIVE	SNV
NRAS Q61H	p.Q61H	COSM586	NEGATIVE	SNV
NRAS Q61K	p.Q61K	COSM53223	NEGATIVE	mnp
NRAS Q61K	p.Q61K	COSM580	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM30646	NEGATIVE	mnp
NRAS Q61L	p.Q61L	COSM583	NEGATIVE	SNV
NRAS Q61L	p.Q61L	COSM12725	NEGATIVE	mnp
NRAS Q61P	p.Q61P	COSM582	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM33693	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NRAS Q61R	p.Q61R	COSM584	NEGATIVE	SNV
NRAS Q61R	p.Q61R	COSM579	NEGATIVE	mnp
NRAS [G60=;Q61K]	p.[G60=;Q61K]	COSM12730	NEGATIVE	mnp
NRAS [Q61H;E62K]	p.[Q61H;E62K]	COSM26494	NEGATIVE	mnp
NTRK1 F589C	p.F589C	OM3255	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT106	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT107	NEGATIVE	SNV
NTRK1 F589L	p.F589L	BT108	NEGATIVE	SNV
NTRK1 F589Y	p.F589Y	BT105	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT109	NEGATIVE	SNV
NTRK1 G595R	p.G595R	BT110	NEGATIVE	SNV
NTRK1 G667A	p.G667A	OM3301	NEGATIVE	SNV
NTRK1 G667C	p.G667C	BT111	NEGATIVE	SNV
NTRK1 G667S	p.G667S	BT112	NEGATIVE	SNV
NTRK1 V573M	p.V573M	BT104	NEGATIVE	SNV
NTRK2 F633C	p.F633C	OM3278	NEGATIVE	SNV
NTRK2 F633I	p.F633I	OM3276	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3277	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3308	NEGATIVE	SNV
NTRK2 F633L	p.F633L	OM3309	NEGATIVE	SNV
NTRK2 F633Y	p.F633Y	OM3307	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3279	NEGATIVE	SNV
NTRK2 G639R	p.G639R	OM3280	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
NTRK2 G709A	p.G709A	OM3310	NEGATIVE	SNV
NTRK2 Q612E	p.Q612E	OM3272	NEGATIVE	SNV
NTRK2 Q612P	p.Q612P	OM3273	NEGATIVE	SNV
NTRK2 R646K	p.R646K	OM3281	NEGATIVE	SNV
NTRK2 V617D	p.V617D	OM3306	NEGATIVE	SNV
NTRK2 V617G	p.V617G	OM3275	NEGATIVE	SNV
NTRK2 V617M	p.V617M	OM3274	NEGATIVE	mnp
NTRK3 F617L	p.F617L	OM3297	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3298	NEGATIVE	SNV
NTRK3 F617L	p.F617L	OM3299	NEGATIVE	SNV
NTRK3 F617Y	p.F617Y	OM3311	NEGATIVE	SNV
NTRK3 G623R	p.G623R	COSM6951362	NEGATIVE	SNV
NTRK3 G623R	p.G623R	OM3253	NEGATIVE	SNV
NTRK3 G696A	p.G696A	OM3296	NEGATIVE	SNV
NTRK3 G696C	p.G696C	OM3294	NEGATIVE	mnp
NTRK3 G696C	p.G696C	OM3295	NEGATIVE	mnp
NTRK3 G696E	p.G696E	COSM5850004	NEGATIVE	SNV
NTRK3 V601F	p.V601F	OM3312	NEGATIVE	SNV
NTRK3 V601M	p.V601M	OM3300	NEGATIVE	mnp
PDGFRA A530_I543dup	p.A530_I543dup	COSM7002381	NEGATIVE	ins
PDGFRA D568N	p.D568N	OM3225	NEGATIVE	SNV
PDGFRA D842F	p.D842F	COSM29007	NEGATIVE	mnp
PDGFRA D842H	p.D842H	COSM734174	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA D842I	p.D842I	COSM12398	NEGATIVE	mnp
PDGFRA D842V	p.D842V	COSM736	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12396	NEGATIVE	SNV
PDGFRA D842Y	p.D842Y	COSM12397	NEGATIVE	mnp
PDGFRA D846Y	p.D846Y	COSM12399	NEGATIVE	SNV
PDGFRA G853D	p.G853D	OM3235	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3228	NEGATIVE	SNV
PDGFRA H650Q	p.H650Q	OM3229	NEGATIVE	SNV
PDGFRA H845Y	p.H845Y	COSM96893	NEGATIVE	SNV
PDGFRA N659H	p.N659H	COSM51516	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22414	NEGATIVE	SNV
PDGFRA N659K	p.N659K	COSM22415	NEGATIVE	SNV
PDGFRA N659R	p.N659R	OM3230	NEGATIVE	mnp
PDGFRA N659S	p.N659S	COSM6960333	NEGATIVE	SNV
PDGFRA N659Y	p.N659Y	COSM22416	NEGATIVE	SNV
PDGFRA N848K	p.N848K	COSM28052	NEGATIVE	SNV
PDGFRA P577S	p.P577S	COSM6438084	NEGATIVE	SNV
PDGFRA Q579R	p.Q579R	OM3226	NEGATIVE	SNV
PDGFRA R841K	p.R841K	OM3233	NEGATIVE	SNV
PDGFRA S566_E571delinsR	p.S566_E571delinsR	COSM12418	NEGATIVE	del
PDGFRA V536E	p.V536E	COSM43059	NEGATIVE	SNV
PDGFRA V561A	p.V561A	COSM1430077	NEGATIVE	SNV
PDGFRA V561D	p.V561D	COSM739	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA W559_R560d el	p.W559_R560del	OMINDEL700	NEGATIVE	del
PDGFRA Y555C	p.Y555C	COSM133645	NEGATIVE	SNV
PDGFRA Y849C	p.Y849C	COSM94952	NEGATIVE	SNV
PDGFRA Y849S	p.Y849S	OM3234	NEGATIVE	SNV
PDGFRA exon 18 deletion	p.D842_M844del	COSM96895	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_M844del	COSM12401	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842del	COSM12406	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_D846delinsN	COSM12411	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_I843delinsV	COSM405285	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_H845delinsY	COSM96896	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsRI	OMINDEL900	NEGATIVE	complex
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.I843del	COSM4969655	NEGATIVE	del
PDGFRA exon 18 deletion	p.I843_D846del	COSM12400	NEGATIVE	del
PDGFRA exon 18 deletion	p.D842_D846delinsE	COSM12408	NEGATIVE	complex
PDGFRA exon 18 deletion	p.D842_S847delinsEA	COSM12415	NEGATIVE	complex

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PDGFRA exon 18 deletion	p.I843_S847delinsT	COSM12407	NEGATIVE	del
PIK3CA A1046E	p.A1046E	COSM27156	NEGATIVE	SNV
PIK3CA A1046T	p.A1046T	COSM27273	NEGATIVE	SNV
PIK3CA A1046V	p.A1046V	COSM6146	NEGATIVE	SNV
PIK3CA C420R	p.C420R	COSM757	NEGATIVE	SNV
PIK3CA D1017H	p.D1017H	COSM27377	NEGATIVE	SNV
PIK3CA D1045G	p.D1045G	COSM1421003	NEGATIVE	SNV
PIK3CA D1045N	p.D1045N	COSM25086	NEGATIVE	SNV
PIK3CA D1045V	p.D1045V	COSM729813	NEGATIVE	SNV
PIK3CA D538G	p.D538G	COSM29123	NEGATIVE	SNV
PIK3CA D538N	p.D538N	COSM21467	NEGATIVE	SNV
PIK3CA D538Y	p.D538Y	COSM1716811	NEGATIVE	SNV
PIK3CA E109_I112delinsD	p.E109_I112delinsD	COSM445995	NEGATIVE	del
PIK3CA E110del	p.E110del	COSM24710	NEGATIVE	del
PIK3CA E39G	p.E39G	COSM1041451	NEGATIVE	SNV
PIK3CA E39K	p.E39K	COSM30625	NEGATIVE	SNV
PIK3CA E39V	p.E39V	COSM1420731	NEGATIVE	SNV
PIK3CA E418K	p.E418K	COSM12582	NEGATIVE	SNV
PIK3CA E542A	p.E542A	COSM1041494	NEGATIVE	SNV
PIK3CA E542G	p.E542G	COSM761	NEGATIVE	SNV
PIK3CA E542K	p.E542K	COSM760	NEGATIVE	SNV
PIK3CA E542Q	p.E542Q	COSM17442	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA E542R	p.E542R	COSM1659152	NEGATIVE	mnp
PIK3CA E542V	p.E542V	COSM762	NEGATIVE	SNV
PIK3CA E545A	p.E545A	COSM12458	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM27374	NEGATIVE	SNV
PIK3CA E545D	p.E545D	COSM765	NEGATIVE	SNV
PIK3CA E545G	p.E545G	COSM764	NEGATIVE	SNV
PIK3CA E545K	p.E545K	COSM763	NEGATIVE	SNV
PIK3CA E545Q	p.E545Q	COSM27133	NEGATIVE	SNV
PIK3CA E545V	p.E545V	COSM27155	NEGATIVE	SNV
PIK3CA E547D	p.E547D	COSM729819	NEGATIVE	SNV
PIK3CA E547G	p.E547G	COSM1420864	NEGATIVE	SNV
PIK3CA E547K	p.E547K	COSM29315	NEGATIVE	SNV
PIK3CA E81A	p.E81A	COSM5347052	NEGATIVE	SNV
PIK3CA E81K	p.E81K	COSM27502	NEGATIVE	SNV
PIK3CA G1049A	p.G1049A	COSM27158	NEGATIVE	SNV
PIK3CA G1049C	p.G1049C	COSM1168056	NEGATIVE	SNV
PIK3CA G1049D	p.G1049D	COSM308548	NEGATIVE	SNV
PIK3CA G1049R	p.G1049R	COSM12597	NEGATIVE	SNV
PIK3CA G1049S	p.G1049S	COSM777	NEGATIVE	SNV
PIK3CA G106A	p.G106A	COSM166339	NEGATIVE	SNV
PIK3CA G106R	p.G106R	COSM86046	NEGATIVE	SNV
PIK3CA G106V	p.G106V	COSM748	NEGATIVE	SNV
PIK3CA G118D	p.G118D	COSM751	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA H1047L	p.H1047L	COSM776	NEGATIVE	SNV
PIK3CA H1047N	p.H1047N	COSM5029128	NEGATIVE	SNV
PIK3CA H1047P	p.H1047P	COSM249874	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM1041525	NEGATIVE	SNV
PIK3CA H1047Q	p.H1047Q	COSM24714	NEGATIVE	SNV
PIK3CA H1047R	p.H1047R	COSM775	NEGATIVE	SNV
PIK3CA H1047Y	p.H1047Y	COSM774	NEGATIVE	SNV
PIK3CA H1048D	p.H1048D	COSM249881	NEGATIVE	SNV
PIK3CA H1048L	p.H1048L	COSM3734761	NEGATIVE	SNV
PIK3CA H1048N	p.H1048N	COSM1421010	NEGATIVE	SNV
PIK3CA H1048Q	p.H1048Q	COSM27157	NEGATIVE	SNV
PIK3CA H1048R	p.H1048R	COSM36289	NEGATIVE	SNV
PIK3CA H1048Y	p.H1048Y	COSM249875	NEGATIVE	SNV
PIK3CA K111E	p.K111E	COSM13570	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM12580	NEGATIVE	SNV
PIK3CA K111N	p.K111N	COSM27505	NEGATIVE	SNV
PIK3CA K111R	p.K111R	COSM14052	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM29313	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM773	NEGATIVE	SNV
PIK3CA M1043I	p.M1043I	COSM94984	NEGATIVE	SNV
PIK3CA M1043L	p.M1043L	COSM5731063	NEGATIVE	SNV
PIK3CA M1043T	p.M1043T	COSM12463	NEGATIVE	SNV
PIK3CA M1043V	p.M1043V	COSM12591	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA N1044D	p.N1044D	COSM27134	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM12592	NEGATIVE	SNV
PIK3CA N1044K	p.N1044K	COSM27504	NEGATIVE	SNV
PIK3CA N1044S	p.N1044S	COSM22541	NEGATIVE	SNV
PIK3CA N1044Y	p.N1044Y	COSM36288	NEGATIVE	SNV
PIK3CA N345D	p.N345D	COSM1420788	NEGATIVE	SNV
PIK3CA N345H	p.N345H	COSM3846786	NEGATIVE	SNV
PIK3CA N345I	p.N345I	COSM94978	NEGATIVE	SNV
PIK3CA N345K	p.N345K	COSM754	NEGATIVE	SNV
PIK3CA N345S	p.N345S	COSM13476	NEGATIVE	SNV
PIK3CA N345T	p.N345T	COSM446002	NEGATIVE	SNV
PIK3CA P104L	p.P104L	COSM163485	NEGATIVE	SNV
PIK3CA P104R	p.P104R	COSM747	NEGATIVE	SNV
PIK3CA P104T	p.P104T	COSM1666660	NEGATIVE	SNV
PIK3CA P539R	p.P539R	COSM759	NEGATIVE	SNV
PIK3CA P539S	p.P539S	COSM249880	NEGATIVE	SNV
PIK3CA P539T	p.P539T	COSM41779	NEGATIVE	SNV
PIK3CA Q546E	p.Q546E	COSM6147	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM24712	NEGATIVE	SNV
PIK3CA Q546H	p.Q546H	COSM5881064	NEGATIVE	SNV
PIK3CA Q546K	p.Q546K	COSM766	NEGATIVE	SNV
PIK3CA Q546L	p.Q546L	COSM25041	NEGATIVE	SNV
PIK3CA Q546P	p.Q546P	COSM767	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PIK3CA Q546R	p.Q546R	COSM12459	NEGATIVE	SNV
PIK3CA R108C	p.R108C	COSM6438095	NEGATIVE	SNV
PIK3CA R108H	p.R108H	COSM27497	NEGATIVE	SNV
PIK3CA R108L	p.R108L	COSM729828	NEGATIVE	SNV
PIK3CA R108P	p.R108P	COSM749	NEGATIVE	SNV
PIK3CA R38C	p.R38C	COSM744	NEGATIVE	SNV
PIK3CA R38G	p.R38G	COSM40945	NEGATIVE	SNV
PIK3CA R38H	p.R38H	COSM745	NEGATIVE	SNV
PIK3CA R38L	p.R38L	COSM729833	NEGATIVE	SNV
PIK3CA R38S	p.R38S	COSM87310	NEGATIVE	SNV
PIK3CA T1025A	p.T1025A	COSM771	NEGATIVE	SNV
PIK3CA T1025I	p.T1025I	COSM36285	NEGATIVE	SNV
PIK3CA T1025N	p.T1025N	COSM772	NEGATIVE	SNV
PIK3CA T1025P	p.T1025P	COSM6023925	NEGATIVE	SNV
PIK3CA T1025S	p.T1025S	COSM12590	NEGATIVE	SNV
PIK3CA V344A	p.V344A	COSM86951	NEGATIVE	SNV
PIK3CA V344E	p.V344E	COSM6746438	NEGATIVE	SNV
PIK3CA V344G	p.V344G	COSM22540	NEGATIVE	SNV
PIK3CA V344M	p.V344M	COSM253279	NEGATIVE	SNV
PIK3CA Y1021C	p.Y1021C	COSM12461	NEGATIVE	SNV
PIK3CA Y1021F	p.Y1021F	COSM328025	NEGATIVE	SNV
PIK3CA Y1021H	p.Y1021H	COSM17444	NEGATIVE	SNV
PIK3CA Y1021N	p.Y1021N	COSM6143	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN A126D	p.A126D	COSM28884	NEGATIVE	SNV
PTEN A126G	p.A126G	COSM4391544	NEGATIVE	SNV
PTEN A126P	p.A126P	COSM5211	NEGATIVE	SNV
PTEN A126S	p.A126S	COSM28889	NEGATIVE	SNV
PTEN A126T	p.A126T	COSM5051	NEGATIVE	SNV
PTEN A126V	p.A126V	COSM5041	NEGATIVE	SNV
PTEN C136F	p.C136F	COSM5228	NEGATIVE	SNV
PTEN C136R	p.C136R	COSM5096	NEGATIVE	SNV
PTEN C136Y	p.C136Y	COSM12734	NEGATIVE	SNV
PTEN C71Y	p.C71Y	COSM5102	NEGATIVE	SNV
PTEN D24E	p.D24E	COSM5198	NEGATIVE	SNV
PTEN D24G	p.D24G	COSM5107	NEGATIVE	SNV
PTEN D24H	p.D24H	COSM1603823	NEGATIVE	SNV
PTEN D24N	p.D24N	COSM5079	NEGATIVE	SNV
PTEN D24V	p.D24V	COSM4016448	NEGATIVE	SNV
PTEN D24Y	p.D24Y	COSM5237	NEGATIVE	SNV
PTEN F341V	p.F341V	COSM5255	NEGATIVE	SNV
PTEN G127E	p.G127E	COSM5143	NEGATIVE	SNV
PTEN G129E	p.G129E	COSM28917	NEGATIVE	SNV
PTEN G129R	p.G129R	COSM246853	NEGATIVE	SNV
PTEN G129V	p.G129V	COSM5276	NEGATIVE	SNV
PTEN L181P	p.L181P	MCH17	NEGATIVE	SNV
PTEN L345Q	p.L345Q	COSM5213	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
PTEN N48K	p.N48K	COSM4392678	NEGATIVE	SNV
PTEN R130*	p.R130*	COSM5152	NEGATIVE	SNV
PTEN R130G	p.R130G	COSM5219	NEGATIVE	SNV
PTEN R130L	p.R130L	COSM5216	NEGATIVE	SNV
PTEN R130P	p.R130P	COSM5277	NEGATIVE	SNV
PTEN R130Q	p.R130Q	COSM5033	NEGATIVE	SNV
PTEN R173C	p.R173C	COSM5089	NEGATIVE	SNV
PTEN R173H	p.R173H	COSM5039	NEGATIVE	SNV
PTEN R173P	p.R173P	COSM12735	NEGATIVE	SNV
PTEN S170N	p.S170N	COSM5045	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH20	NEGATIVE	SNV
PTEN S170R	p.S170R	COSM6006454	NEGATIVE	SNV
PTEN S170R	p.S170R	MCH21	NEGATIVE	SNV
PTEN T131I	p.T131I	COSM5104	NEGATIVE	SNV
PTEN V133I	p.V133I	COSM5044	NEGATIVE	SNV
PTEN V343E	p.V343E	MCH18	NEGATIVE	SNV
PTEN Y155C	p.Y155C	COSM5144	NEGATIVE	SNV
PTEN Y16C	p.Y16C	COSM5133	NEGATIVE	SNV
PTEN Y174C	p.Y174C	COSM5575601	NEGATIVE	SNV
PTEN Y174N	p.Y174N	COSM5221	NEGATIVE	SNV
RAF1 S257L	p.S257L	COSM181063	NEGATIVE	SNV
RAF1 S257P	p.S257P	ClinVar40600	NEGATIVE	SNV
RAF1 S257W	p.S257W	COSM581519	NEGATIVE	SNV

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 S259F	p.S259F	COSM1536534	NEGATIVE	SNV
RAF1 S259P	p.S259P	COSM1418555	NEGATIVE	SNV
RAF1 S259T	p.S259T	ClinVar40601	NEGATIVE	SNV
RET A883F	p.A883F	COSM977	NEGATIVE	mnp
RET A883S	p.A883S	COSM133167	NEGATIVE	SNV
RET A883T	p.A883T	COSM100081	NEGATIVE	SNV
RET A883V	p.A883V	COSM4594153	NEGATIVE	SNV
RET A883Y	p.A883Y	OM3415	NEGATIVE	mnp
RET C618F	p.C618F	OM3401	NEGATIVE	SNV
RET C618G	p.C618G	COSM9214400	NEGATIVE	SNV
RET C618R	p.C618R	COSM29803	NEGATIVE	SNV
RET C618S	p.C618S	COSM87267	NEGATIVE	SNV
RET C618S	p.C618S	OM3402	NEGATIVE	SNV
RET C618T	p.C618T	OM3400	NEGATIVE	mnp
RET C618W	p.C618W	OM3403	NEGATIVE	SNV
RET C618Y	p.C618Y	COSM980	NEGATIVE	SNV
RET C620F	p.C620F	OM3405	NEGATIVE	SNV
RET C620G	p.C620G	OM3404	NEGATIVE	SNV
RET C620R	p.C620R	COSM29804	NEGATIVE	SNV
RET C620S	p.C620S	COSM5946160	NEGATIVE	SNV
RET C620S	p.C620S	COSM29805	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET C620W	p.C620W	COSM249815	NEGATIVE	SNV
RET C620Y	p.C620Y	COSM7403807	NEGATIVE	SNV
RET C630F	p.C630F	OM3406	NEGATIVE	SNV
RET C630G	p.C630G	COSM29806	NEGATIVE	SNV
RET C630R	p.C630R	COSM964	NEGATIVE	SNV
RET C630Y	p.C630Y	COSM9233214	NEGATIVE	SNV
RET C634F	p.C634F	COSM1237919	NEGATIVE	SNV
RET C634G	p.C634G	COSM1738369	NEGATIVE	SNV
RET C634L	p.C634L	OM3408	NEGATIVE	mnp
RET C634R	p.C634R	COSM966	NEGATIVE	SNV
RET C634S	p.C634S	COSM1237918	NEGATIVE	SNV
RET C634S	p.C634S	COSM1666664	NEGATIVE	SNV
RET C634W	p.C634W	COSM975	NEGATIVE	SNV
RET C634Y	p.C634Y	COSM974	NEGATIVE	SNV
RET C634_R635dup	p.C634_R635dup	OMINDEL1177	NEGATIVE	ins
RET C634_R635insHELC	p.C634_R635insHELC	OMINDEL1174	NEGATIVE	ins
RET C634_T636dup	p.C634_T636dup	COSM9213763	NEGATIVE	ins
RET D631Y	p.D631Y	COSM1738370	NEGATIVE	SNV
RET D631_C634del	p.D631_C634del	OMINDEL1172	NEGATIVE	del
RET D631_L633delinsE	p.D631_L633delinsE	COSM983	NEGATIVE	del
RET D631_L633delinsV	p.D631_L633delinsV	OMINDEL1175	NEGATIVE	del
RET D631del	p.D631del	OMINDEL1173	NEGATIVE	del
RET D898_E901del	p.D898_E901del	COSM962	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET E632K	p.E632K	COSM1702268	NEGATIVE	SNV
RET E632_C634delinsG	p.E632_C634delinsG	COSM973	NEGATIVE	del
RET E632_C634dup	p.E632_C634dup	OMINDEL1176	NEGATIVE	ins
RET E632_L633del	p.E632_L633del	COSM968	NEGATIVE	del
RET E632_L633delinsV	p.E632_L633delinsV	COSM982	NEGATIVE	del
RET E632_T636delinsSS	p.E632_T636delinsSS	COSM1048	NEGATIVE	complex
RET E768D	p.E768D	COSM21338	NEGATIVE	SNV
RET E768D	p.E768D	OM3413	NEGATIVE	SNV
RET E768G	p.E768G	COSM1347811	NEGATIVE	SNV
RET E768Q	p.E768Q	COSM1716312	NEGATIVE	SNV
RET E884K	p.E884K	COSM960	NEGATIVE	SNV
RET E884V	p.E884V	COSM1570338	NEGATIVE	SNV
RET G736A	p.G736A	OM3411	NEGATIVE	SNV
RET G810A	p.G810A	OM3290	NEGATIVE	SNV
RET G810C	p.G810C	OM3557	NEGATIVE	SNV
RET G810R	p.G810R	OM3289	NEGATIVE	SNV
RET G810S	p.G810S	OM3288	NEGATIVE	SNV
RET G810V	p.G810V	OM3558	NEGATIVE	SNV
RET G911D	p.G911D	COSM20888	NEGATIVE	SNV
RET I788N	p.I788N	OM3284	NEGATIVE	SNV
RET L730Q	p.L730Q	OM3409	NEGATIVE	SNV
RET L730R	p.L730R	OM3410	NEGATIVE	SNV
RET L760Q	p.L760Q	OM3412	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET L790F	p.L790F	rs75030001	NEGATIVE	SNV
RET L881V	p.L881V	OM3414	NEGATIVE	SNV
RET P628_L633del	p.P628_L633del	OMINDEL1171	NEGATIVE	del
RET R770Q	p.R770Q	COSM6006462	NEGATIVE	SNV
RET R886W	p.R886W	COSM6942691	NEGATIVE	SNV
RET R897*	p.R897*	COSM6983129	NEGATIVE	SNV
RET R912L	p.R912L	COSM188545	NEGATIVE	SNV
RET R912P	p.R912P	OM3419	NEGATIVE	SNV
RET R912Q	p.R912Q	COSM3437793	NEGATIVE	SNV
RET R912W	p.R912W	COSM3415038	NEGATIVE	SNV
RET S891A	p.S891A	rs75234356	NEGATIVE	SNV
RET S904C	p.S904C	OM3418	NEGATIVE	SNV
RET S904F	p.S904F	COSM6438204	NEGATIVE	SNV
RET V804G	p.V804G	OM3554	NEGATIVE	SNV
RET V804L	p.V804L	OM3285	NEGATIVE	SNV
RET V804L	p.V804L	OM3286	NEGATIVE	SNV
RET V804M	p.V804M	OM3195	NEGATIVE	SNV
RET V899_D903del	p.V899_D903del	COSM5991595	NEGATIVE	del
RET Y806C	p.Y806C	OM3287	NEGATIVE	SNV
RET Y806F	p.Y806F	OM3556	NEGATIVE	SNV
RET [D903E;S904P]	p.[D903E;S904P]	OM3416	NEGATIVE	mnp
RET [D903E;S904P]	p.[D903E;S904P]	OM3417	NEGATIVE	mnp
RET [E632D;L633V;C634R]	p.[E632D;L633V;C634R]	OM3407	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
RET [V882=;A883F]	p.[V882=;A883F]	COSM981	NEGATIVE	mnp
RET c.1880_1899delinsCTCAC	p.?	OMINDEL1170	NEGATIVE	complex
ROS1 D2033N	p.D2033N	COSM6924065	NEGATIVE	SNV
ROS1 G2032K	p.G2032K	OM3533	NEGATIVE	mnp
ROS1 G2032R	p.G2032R	MAN10	NEGATIVE	SNV
ROS1 G2032R	p.G2032R	MAN11	NEGATIVE	SNV
ROS1 L1951M	p.L1951M	COSM1072523	NEGATIVE	SNV
ROS1 L1951R	p.L1951R	OM3264	NEGATIVE	SNV
ROS1 L2026M	p.L2026M	BT151	NEGATIVE	SNV
ROS1 S1986F	p.S1986F	COSM6929870	NEGATIVE	SNV
ROS1 S1986Y	p.S1986Y	OM3170	NEGATIVE	SNV
STK11 A205T	p.A205T	COSM20953	NEGATIVE	SNV
STK11 D176N	p.D176N	COSM4827690	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3710	NEGATIVE	SNV
STK11 D194E	p.D194E	OM3711	NEGATIVE	SNV
STK11 D194H	p.D194H	COSM5325765	NEGATIVE	SNV
STK11 D194N	p.D194N	COSM25847	NEGATIVE	SNV
STK11 D194V	p.D194V	COSM20957	NEGATIVE	SNV
STK11 D194Y	p.D194Y	COSM20944	NEGATIVE	SNV
STK11 D53Gfs*110	p.D53GfsTer110	COSM391251	NEGATIVE	ins
STK11 L290P	p.L290P	OM3719	NEGATIVE	SNV
STK11 L67P	p.L67P	OM3700	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 R304W	p.R304W	COSM29468	NEGATIVE	SNV
STK11 S240W	p.S240W	OM3714	NEGATIVE	SNV
STK11 c.1108+1G>A	p.?	OM3722	NEGATIVE	SNV
STK11 c.289_290+1delinsGAT	p.?	COSM6922318	NEGATIVE	mnp
STK11 c.289_290+2delAAGT	p.?	OMINDEL1000	NEGATIVE	del
STK11 c.290+1G>A	p.?	COSM6350392	NEGATIVE	SNV
STK11 c.290+1G>C	p.?	OM3701	NEGATIVE	SNV
STK11 c.290+1G>T	p.?	OM3702	NEGATIVE	SNV
STK11 c.290+1_290+2delGT	p.?	OMINDEL1001	NEGATIVE	del
STK11 c.290+2T>A	p.?	COSM6925006	NEGATIVE	SNV
STK11 c.290+2T>G	p.?	COSM25857	NEGATIVE	SNV
STK11 c.290+2delT	p.?	OMINDEL1002	NEGATIVE	del
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.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.375-1C>G	p.?	COSM4074282	NEGATIVE	SNV
STK11 c.375-1C>T	p.?	COSM6977173	NEGATIVE	SNV
STK11 c.375-2A>C	p.?	OM3706	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.375-2A>G	p.?	OM3707	NEGATIVE	SNV
STK11 c.375-2A>T	p.?	COSM6967966	NEGATIVE	SNV
STK11 c.375-2_386delACGTATATGGTGAT	p.?	OMINDEL1006	NEGATIVE	del
STK11 c.375-6_375-2delCCCCA	p.?	OMINDEL1005	NEGATIVE	del
STK11 c.465-1G>A	p.?	COSM25855	NEGATIVE	SNV
STK11 c.465-1G>C	p.?	OM3709	NEGATIVE	SNV
STK11 c.465-1G>T	p.?	COSM21570	NEGATIVE	SNV
STK11 c.465-1_465delinsTT	p.?	COSM6928792	NEGATIVE	mnp
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.465-2_465-1delAG	p.?	OMINDEL1009	NEGATIVE	del
STK11 c.465-2_465-1delinsTC	p.?	OM3708	NEGATIVE	mnp
STK11 c.465-4_472delGCAGGTACTTCT	p.?	OMINDEL1007	NEGATIVE	del
STK11 c.465-4_479delGCAGGTACTTCTGTCAGCT	p.?	OMINDEL1008	NEGATIVE	del
STK11 c.598-1G>A	p.?	COSM3724562	NEGATIVE	SNV
STK11 c.598-1G>T	p.?	COSM51521	NEGATIVE	SNV
STK11 c.598-1_598delinsTT	p.?	OM3713	NEGATIVE	mnp

Variant Name	AA Change	Variant ID	Test Result	Variant Type
STK11 c.598-1_612delGGCACTGCACCCGTTC	p.?	OMINDEL1014	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.598-2_600delAGGCA	p.?	OMINDEL1012	NEGATIVE	del
STK11 c.727_734+9delGTCACCCTGTAAGTGCC	p.?	OMINDEL1015	NEGATIVE	del
STK11 c.731_734+9delCCCTGTAAGTGCC	p.?	OMINDEL1016	NEGATIVE	del
STK11 c.734+1G>A	p.?	COSM6916684	NEGATIVE	SNV
STK11 c.734+1G>C	p.?	COSM6970016	NEGATIVE	SNV
STK11 c.734+1G>T	p.?	COSM51523	NEGATIVE	SNV
STK11 c.734+2T>A	p.?	OM3715	NEGATIVE	SNV
STK11 c.734+2T>G	p.?	COSM6961577	NEGATIVE	SNV
STK11 c.734+2_734+10delTAAGTGCCC	p.?	OMINDEL1017	NEGATIVE	del
STK11 c.735-1G>A	p.?	COSM5732040	NEGATIVE	SNV
STK11 c.735-1G>T	p.?	COSM51522	NEGATIVE	SNV
STK11 c.735-2A>G	p.?	OM3716	NEGATIVE	SNV
STK11 c.735-2A>T	p.?	COSM25856	NEGATIVE	SNV
STK11 c.850_862+4delGACCTGCTGAAAGGTGG	p.?	OMINDEL1018	NEGATIVE	del

Variant Name	AA Change	Variant ID	Test Result	Variant Type
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+2T>A	p.?	COSM5576087	NEGATIVE	SNV
STK11 c.862+2T>G	p.?	OM3718	NEGATIVE	SNV
STK11 c.862+3delG	p.?	COSM8554973	NEGATIVE	del
STK11 c.863-1G>A	p.?	COSM6916725	NEGATIVE	SNV
STK11 c.863-1G>T	p.?	COSM6925330	NEGATIVE	SNV
STK11 c.863-2A>C	p.?	COSM6912582	NEGATIVE	SNV
STK11 c.863-2A>T	p.?	COSM401786	NEGATIVE	SNV
STK11 c.918_920+11delCAGGTGAGCGGCCC	p.?	OMINDEL1019	NEGATIVE	del
STK11 c.920+1G>A	p.?	OM3720	NEGATIVE	SNV
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
TP53 A138V	p.A138V	COSM43818	NEGATIVE	SNV
TP53 C135F	p.C135F	COSM10647	NEGATIVE	SNV
TP53 C135G	p.C135G	COSM44829	NEGATIVE	SNV
TP53 C135R	p.C135R	COSM10684	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44643	NEGATIVE	SNV
TP53 C135S	p.C135S	COSM44910	NEGATIVE	SNV
TP53 C135W	p.C135W	COSM44219	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C135Y	p.C135Y	COSM10801	NEGATIVE	SNV
TP53 C141F	p.C141F	COSM44911	NEGATIVE	SNV
TP53 C141G	p.C141G	COSM45794	NEGATIVE	SNV
TP53 C141R	p.C141R	COSM43901	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM293910	NEGATIVE	SNV
TP53 C141S	p.C141S	COSM44564	NEGATIVE	SNV
TP53 C141W	p.C141W	COSM44204	NEGATIVE	SNV
TP53 C141Y	p.C141Y	COSM43708	NEGATIVE	SNV
TP53 C176F	p.C176F	COSM10645	NEGATIVE	SNV
TP53 C176G	p.C176G	COSM44692	NEGATIVE	SNV
TP53 C176R	p.C176R	COSM44948	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44645	NEGATIVE	SNV
TP53 C176S	p.C176S	COSM44146	NEGATIVE	SNV
TP53 C176W	p.C176W	COSM11114	NEGATIVE	SNV
TP53 C176Y	p.C176Y	COSM10687	NEGATIVE	SNV
TP53 C238F	p.C238F	COSM43778	NEGATIVE	SNV
TP53 C238G	p.C238G	COSM46336	NEGATIVE	SNV
TP53 C238R	p.C238R	COSM44321	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM44653	NEGATIVE	SNV
TP53 C238S	p.C238S	COSM43700	NEGATIVE	SNV
TP53 C238W	p.C238W	COSM44676	NEGATIVE	SNV
TP53 C238Y	p.C238Y	COSM11059	NEGATIVE	SNV
TP53 C242F	p.C242F	COSM10810	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 C242G	p.C242G	COSM44135	NEGATIVE	SNV
TP53 C242R	p.C242R	COSM11738	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM11133	NEGATIVE	SNV
TP53 C242S	p.C242S	COSM44935	NEGATIVE	SNV
TP53 C242W	p.C242W	COSM11356	NEGATIVE	SNV
TP53 C242Y	p.C242Y	COSM10646	NEGATIVE	SNV
TP53 C275F	p.C275F	COSM10701	NEGATIVE	SNV
TP53 C275G	p.C275G	COSM11501	NEGATIVE	SNV
TP53 C275R	p.C275R	COSM43902	NEGATIVE	SNV
TP53 C275S	p.C275S	COSM45413	NEGATIVE	SNV
TP53 C275W	p.C275W	COSM43823	NEGATIVE	SNV
TP53 C275Y	p.C275Y	COSM10893	NEGATIVE	SNV
TP53 C277F	p.C277F	COSM10749	NEGATIVE	SNV
TP53 D281A	p.D281A	COSM11665	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43837	NEGATIVE	SNV
TP53 D281E	p.D281E	COSM43906	NEGATIVE	SNV
TP53 D281G	p.D281G	COSM11232	NEGATIVE	SNV
TP53 D281H	p.D281H	COSM10943	NEGATIVE	SNV
TP53 D281N	p.D281N	COSM43596	NEGATIVE	SNV
TP53 D281V	p.D281V	COSM45729	NEGATIVE	SNV
TP53 D281Y	p.D281Y	COSM11516	NEGATIVE	SNV
TP53 E258A	p.E258A	COSM44719	NEGATIVE	SNV
TP53 E258D	p.E258D	COSM44962	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 E258D	p.E258D	COSM46410	NEGATIVE	SNV
TP53 E258G	p.E258G	COSM44168	NEGATIVE	SNV
TP53 E258K	p.E258K	COSM10988	NEGATIVE	SNV
TP53 E258Q	p.E258Q	COSM10751	NEGATIVE	SNV
TP53 E258V	p.E258V	COSM44450	NEGATIVE	SNV
TP53 E285A	p.E285A	COSM43614	NEGATIVE	SNV
TP53 E285G	p.E285G	COSM45649	NEGATIVE	SNV
TP53 E285K	p.E285K	COSM10722	NEGATIVE	SNV
TP53 E285Q	p.E285Q	COSM45138	NEGATIVE	SNV
TP53 E285V	p.E285V	COSM44227	NEGATIVE	SNV
TP53 E286A	p.E286A	COSM45892	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45439	NEGATIVE	SNV
TP53 E286D	p.E286D	COSM45492	NEGATIVE	SNV
TP53 E286G	p.E286G	COSM43565	NEGATIVE	SNV
TP53 E286K	p.E286K	COSM10726	NEGATIVE	SNV
TP53 E286Q	p.E286Q	COSM44250	NEGATIVE	SNV
TP53 E286V	p.E286V	COSM43936	NEGATIVE	SNV
TP53 G154V	p.G154V	COSM6815	NEGATIVE	SNV
TP53 G244A	p.G244A	COSM12013	NEGATIVE	SNV
TP53 G244C	p.G244C	COSM11524	NEGATIVE	SNV
TP53 G244D	p.G244D	COSM10883	NEGATIVE	SNV
TP53 G244R	p.G244R	COSM44221	NEGATIVE	SNV
TP53 G244S	p.G244S	COSM10941	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 G244V	p.G244V	COSM43652	NEGATIVE	SNV
TP53 G245A	p.G245A	COSM43965	NEGATIVE	SNV
TP53 G245C	p.G245C	COSM11081	NEGATIVE	SNV
TP53 G245D	p.G245D	COSM43606	NEGATIVE	SNV
TP53 G245R	p.G245R	COSM10957	NEGATIVE	SNV
TP53 G245S	p.G245S	COSM6932	NEGATIVE	SNV
TP53 G245V	p.G245V	COSM11196	NEGATIVE	SNV
TP53 G262V	p.G262V	COSM11198	NEGATIVE	SNV
TP53 G266A	p.G266A	COSM45488	NEGATIVE	SNV
TP53 G266E	p.G266E	COSM10867	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM10794	NEGATIVE	SNV
TP53 G266R	p.G266R	COSM11205	NEGATIVE	SNV
TP53 G266V	p.G266V	COSM10958	NEGATIVE	SNV
TP53 G279E	p.G279E	COSM43714	NEGATIVE	SNV
TP53 H179D	p.H179D	COSM44776	NEGATIVE	SNV
TP53 H179L	p.H179L	COSM43635	NEGATIVE	SNV
TP53 H179N	p.H179N	COSM44151	NEGATIVE	SNV
TP53 H179P	p.H179P	COSM44218	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM11249	NEGATIVE	SNV
TP53 H179Q	p.H179Q	COSM44214	NEGATIVE	SNV
TP53 H179R	p.H179R	COSM10889	NEGATIVE	SNV
TP53 H179Y	p.H179Y	COSM10768	NEGATIVE	SNV
TP53 H193D	p.H193D	COSM44002	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 H193L	p.H193L	COSM11066	NEGATIVE	SNV
TP53 H193N	p.H193N	COSM43935	NEGATIVE	SNV
TP53 H193P	p.H193P	COSM43833	NEGATIVE	SNV
TP53 H193R	p.H193R	COSM10742	NEGATIVE	SNV
TP53 H193Y	p.H193Y	COSM10672	NEGATIVE	SNV
TP53 H214D	p.H214D	COSM45115	NEGATIVE	SNV
TP53 H214L	p.H214L	COSM5632535	NEGATIVE	SNV
TP53 H214P	p.H214P	COSM1640841	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM44407	NEGATIVE	SNV
TP53 H214Q	p.H214Q	COSM46272	NEGATIVE	SNV
TP53 H214R	p.H214R	COSM43687	NEGATIVE	SNV
TP53 H214Y	p.H214Y	COSM44112	NEGATIVE	SNV
TP53 I195F	p.I195F	COSM44633	NEGATIVE	SNV
TP53 I195L	p.I195L	COSM45773	NEGATIVE	SNV
TP53 I195M	p.I195M	COSM562637	NEGATIVE	SNV
TP53 I195N	p.I195N	COSM44877	NEGATIVE	SNV
TP53 I195S	p.I195S	COSM44539	NEGATIVE	SNV
TP53 I195T	p.I195T	COSM11089	NEGATIVE	SNV
TP53 K132E	p.K132E	COSM10813	NEGATIVE	SNV
TP53 K132M	p.K132M	COSM43592	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM10991	NEGATIVE	SNV
TP53 K132N	p.K132N	COSM43963	NEGATIVE	SNV
TP53 K132Q	p.K132Q	COSM11224	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 K132R	p.K132R	COSM11582	NEGATIVE	SNV
TP53 K132T	p.K132T	COSM43912	NEGATIVE	SNV
TP53 L145P	p.L145P	COSM43899	NEGATIVE	SNV
TP53 L194F	p.L194F	COSM10995	NEGATIVE	SNV
TP53 L194H	p.L194H	COSM43623	NEGATIVE	SNV
TP53 L194I	p.L194I	COSM45330	NEGATIVE	SNV
TP53 L194P	p.L194P	COSM43827	NEGATIVE	SNV
TP53 L194R	p.L194R	COSM44571	NEGATIVE	SNV
TP53 L194V	p.L194V	COSM46117	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM10834	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM11063	NEGATIVE	SNV
TP53 M237I	p.M237I	COSM44415	NEGATIVE	SNV
TP53 M237K	p.M237K	COSM43952	NEGATIVE	SNV
TP53 M237L	p.M237L	COSM44965	NEGATIVE	SNV
TP53 M237R	p.M237R	COSM45032	NEGATIVE	SNV
TP53 M237T	p.M237T	COSM45329	NEGATIVE	SNV
TP53 M237V	p.M237V	COSM44525	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM10757	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM44310	NEGATIVE	SNV
TP53 M246I	p.M246I	COSM46136	NEGATIVE	SNV
TP53 M246K	p.M246K	COSM44103	NEGATIVE	SNV
TP53 M246L	p.M246L	COSM45992	NEGATIVE	SNV
TP53 M246R	p.M246R	COSM11376	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 M246T	p.M246T	COSM11355	NEGATIVE	SNV
TP53 M246V	p.M246V	COSM43555	NEGATIVE	SNV
TP53 N239D	p.N239D	COSM10777	NEGATIVE	SNV
TP53 N239I	p.N239I	COSM307333	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44510	NEGATIVE	SNV
TP53 N239K	p.N239K	COSM44647	NEGATIVE	SNV
TP53 N239S	p.N239S	COSM44094	NEGATIVE	SNV
TP53 N239T	p.N239T	COSM43801	NEGATIVE	SNV
TP53 N239Y	p.N239Y	COSM45870	NEGATIVE	SNV
TP53 P151A	p.P151A	COSM44944	NEGATIVE	SNV
TP53 P151H	p.P151H	COSM11476	NEGATIVE	SNV
TP53 P151L	p.P151L	COSM44288	NEGATIVE	SNV
TP53 P151R	p.P151R	COSM44003	NEGATIVE	SNV
TP53 P151S	p.P151S	COSM10905	NEGATIVE	SNV
TP53 P151T	p.P151T	COSM43911	NEGATIVE	SNV
TP53 P152A	p.P152A	COSM44788	NEGATIVE	SNV
TP53 P152L	p.P152L	COSM10790	NEGATIVE	SNV
TP53 P152Q	p.P152Q	COSM44613	NEGATIVE	SNV
TP53 P152R	p.P152R	COSM45505	NEGATIVE	SNV
TP53 P152S	p.P152S	COSM43582	NEGATIVE	SNV
TP53 P152T	p.P152T	COSM44561	NEGATIVE	SNV
TP53 P177_C182del	p.P177_C182del	COSM43570	NEGATIVE	del
TP53 P190L	p.P190L	COSM43657	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 P191del	p.P191del	COSM45140	NEGATIVE	del
TP53 P250L	p.P250L	COSM10771	NEGATIVE	SNV
TP53 P278A	p.P278A	COSM10814	NEGATIVE	SNV
TP53 P278H	p.P278H	COSM43755	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 Q192*	p.Q192*	COSM10733	NEGATIVE	SNV
TP53 R156P	p.R156P	COSM10760	NEGATIVE	SNV
TP53 R175C	p.R175C	COSM43680	NEGATIVE	SNV
TP53 R175G	p.R175G	COSM10870	NEGATIVE	SNV
TP53 R175H	p.R175H	COSM10648	NEGATIVE	SNV
TP53 R175L	p.R175L	COSM10718	NEGATIVE	SNV
TP53 R175P	p.R175P	COSM45416	NEGATIVE	SNV
TP53 R175S	p.R175S	COSM43931	NEGATIVE	SNV
TP53 R196*	p.R196*	COSM10705	NEGATIVE	SNV
TP53 R213*	p.R213*	COSM10654	NEGATIVE	SNV
TP53 R213G	p.R213G	COSM44102	NEGATIVE	SNV
TP53 R213L	p.R213L	COSM43650	NEGATIVE	SNV
TP53 R213P	p.R213P	COSM11860	NEGATIVE	SNV
TP53 R213Q	p.R213Q	COSM10735	NEGATIVE	SNV
TP53 R248G	p.R248G	COSM11564	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R248L	p.R248L	COSM6549	NEGATIVE	SNV
TP53 R248P	p.R248P	COSM11491	NEGATIVE	SNV
TP53 R248Q	p.R248Q	COSM10662	NEGATIVE	SNV
TP53 R248W	p.R248W	COSM10656	NEGATIVE	SNV
TP53 R249G	p.R249G	COSM10668	NEGATIVE	SNV
TP53 R249K	p.R249K	COSM44091	NEGATIVE	SNV
TP53 R249M	p.R249M	COSM43871	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10785	NEGATIVE	SNV
TP53 R249S	p.R249S	COSM10817	NEGATIVE	SNV
TP53 R249T	p.R249T	COSM43665	NEGATIVE	SNV
TP53 R249W	p.R249W	COSM43629	NEGATIVE	SNV
TP53 R273C	p.R273C	COSM10659	NEGATIVE	SNV
TP53 R273G	p.R273G	COSM43843	NEGATIVE	SNV
TP53 R273H	p.R273H	COSM10660	NEGATIVE	SNV
TP53 R273L	p.R273L	COSM10779	NEGATIVE	SNV
TP53 R273P	p.R273P	COSM43896	NEGATIVE	SNV
TP53 R273S	p.R273S	COSM43909	NEGATIVE	SNV
TP53 R280G	p.R280G	COSM11123	NEGATIVE	SNV
TP53 R280I	p.R280I	COSM11287	NEGATIVE	SNV
TP53 R280K	p.R280K	COSM10728	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44171	NEGATIVE	SNV
TP53 R280S	p.R280S	COSM44233	NEGATIVE	SNV
TP53 R280T	p.R280T	COSM10724	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 R282G	p.R282G	COSM10992	NEGATIVE	SNV
TP53 R282L	p.R282L	COSM44470	NEGATIVE	SNV
TP53 R282P	p.R282P	COSM44306	NEGATIVE	SNV
TP53 R282Q	p.R282Q	COSM44338	NEGATIVE	SNV
TP53 R282W	p.R282W	COSM10704	NEGATIVE	SNV
TP53 R283P	p.R283P	COSM10743	NEGATIVE	SNV
TP53 S240G	p.S240G	COSM43973	NEGATIVE	SNV
TP53 S241A	p.S241A	COSM44224	NEGATIVE	SNV
TP53 S241C	p.S241C	COSM10709	NEGATIVE	SNV
TP53 S241F	p.S241F	COSM10812	NEGATIVE	SNV
TP53 S241P	p.S241P	COSM44578	NEGATIVE	SNV
TP53 S241T	p.S241T	COSM44067	NEGATIVE	SNV
TP53 S241Y	p.S241Y	COSM10935	NEGATIVE	SNV
TP53 V157F	p.V157F	COSM10670	NEGATIVE	SNV
TP53 V157L	p.V157L	COSM45120	NEGATIVE	SNV
TP53 V172F	p.V172F	COSM44240	NEGATIVE	SNV
TP53 V173A	p.V173A	COSM44327	NEGATIVE	SNV
TP53 V173E	p.V173E	COSM45731	NEGATIVE	SNV
TP53 V173G	p.V173G	COSM44383	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM43559	NEGATIVE	SNV
TP53 V173L	p.V173L	COSM44057	NEGATIVE	SNV
TP53 V173M	p.V173M	COSM11084	NEGATIVE	SNV
TP53 V197M	p.V197M	COSM43779	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 V216A	p.V216A	COSM44567	NEGATIVE	SNV
TP53 V216E	p.V216E	COSM44274	NEGATIVE	SNV
TP53 V216G	p.V216G	COSM43681	NEGATIVE	SNV
TP53 V216L	p.V216L	COSM11210	NEGATIVE	SNV
TP53 V216M	p.V216M	COSM10667	NEGATIVE	SNV
TP53 V272A	p.V272A	COSM44294	NEGATIVE	SNV
TP53 V272E	p.V272E	COSM44580	NEGATIVE	SNV
TP53 V272G	p.V272G	COSM44870	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM10859	NEGATIVE	SNV
TP53 V272L	p.V272L	COSM45898	NEGATIVE	SNV
TP53 V272M	p.V272M	COSM10891	NEGATIVE	SNV
TP53 V274F	p.V274F	COSM10769	NEGATIVE	SNV
TP53 V274L	p.V274L	COSM44443	NEGATIVE	SNV
TP53 Y220C	p.Y220C	COSM10758	NEGATIVE	SNV
TP53 Y220D	p.Y220D	COSM11847	NEGATIVE	SNV
TP53 Y220F	p.Y220F	COSM1564197	NEGATIVE	SNV
TP53 Y220H	p.Y220H	COSM44637	NEGATIVE	SNV
TP53 Y220N	p.Y220N	COSM44672	NEGATIVE	SNV
TP53 Y220S	p.Y220S	COSM43850	NEGATIVE	SNV
TP53 Y234C	p.Y234C	COSM10725	NEGATIVE	SNV
TP53 Y234D	p.Y234D	COSM43768	NEGATIVE	SNV
TP53 Y234F	p.Y234F	COSM44315	NEGATIVE	SNV
TP53 Y234H	p.Y234H	COSM11152	NEGATIVE	SNV

Variant Name	AA Change	Variant ID	Test Result	Variant Type
TP53 Y234N	p.Y234N	COSM43956	NEGATIVE	SNV
TP53 Y234S	p.Y234S	COSM43865	NEGATIVE	SNV
TP53 Y236C	p.Y236C	COSM10731	NEGATIVE	SNV
TP53 Y236D	p.Y236D	COSM43602	NEGATIVE	SNV
TP53 Y236F	p.Y236F	COSM3958809	NEGATIVE	SNV
TP53 Y236H	p.Y236H	COSM44326	NEGATIVE	SNV
TP53 Y236N	p.Y236N	COSM43826	NEGATIVE	SNV
TP53 Y236S	p.Y236S	COSM44693	NEGATIVE	SNV

Fusions

Variant Name	Genes (Exons)	Variant Type	Test Result
A2M-ALK fusion	A2M(22) - ALK(19)	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
AGAP1-ALK fusion	AGAP1(6) - ALK(18)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATG16L1-ALK fusion	ATG16L1(2) - ALK(20)	Fusion	NEGATIVE
ATIC-ALK fusion	ATIC(7) - ALK(20)	Fusion	NEGATIVE
ATRNL1-ALK fusion	ATRNL1(19) - ALK(20)	Fusion	NEGATIVE
BCL11A-ALK fusion	BCL11A(4) - ALK(20)	Fusion	NEGATIVE
BEND5-ALK fusion	BEND5(3) - ALK(20)	Fusion	NEGATIVE
BIRC6-ALK fusion	BIRC6(10) - ALK(20)	Fusion	NEGATIVE
CAD-ALK fusion	CAD(35) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CAD-ALK fusion	CAD(36) - ALK(20)	Fusion	NEGATIVE
CAMKMT-ALK fusion	CAMKMT(3) - ALK(20)	Fusion	NEGATIVE
CARS-ALK fusion	CARS(17) - ALK(20)	Fusion	NEGATIVE
CCDC149-ALK fusion	CCDC149(10) - ALK(20)	Fusion	NEGATIVE
CCDC61-ALK fusion	CCDC61(13) - ALK(20)	Fusion	NEGATIVE
CCDC88A-ALK fusion	CCDC88A(12) - ALK(20)	Fusion	NEGATIVE
CEBPZ-ALK fusion	CEBPZ(2) - ALK(20)	Fusion	NEGATIVE
CENPF-ALK fusion	CENPF(11) - ALK(20)	Fusion	NEGATIVE
CLIP1-ALK fusion	CLIP1(13) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(10) - ALK(20)	Fusion	NEGATIVE
CLIP4-ALK fusion	CLIP4(14) - ALK(17)	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
COL25A1-ALK fusion	COL25A1(3) - ALK(20)	Fusion	NEGATIVE
CPSF7-ALK fusion	CPSF7(5) - ALK(20)	Fusion	NEGATIVE
CREB1-ALK fusion	CREB1(1) - ALK(9)	Fusion	NEGATIVE
CTNND1-ALK fusion	CTNND1(16) - ALK(20)	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
DDX6-ALK fusion	DDX6(2) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DES-ALK fusion	DES(7) - ALK(19)	Fusion	NEGATIVE
DMD-ALK fusion	DMD(55) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(8) - ALK(20)	Fusion	NEGATIVE
EEF1G-ALK fusion	EEF1G(6) - ALK(20)	Fusion	NEGATIVE
EIF2AK3-ALK fusion	EIF2AK3(2) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(17) - ALK(20)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(2) - 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(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(3) - 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(19)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(18)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(17)	Fusion	NEGATIVE
EML4-ALK fusion	EML4(6) - ALK(16)	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

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(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(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(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(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

Variant Name	Genes (Exons)	Variant Type	Test Result
EML4-ALK fusion	EML4(22) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(4) - ALK(20)	Fusion	NEGATIVE
ETV6-ALK fusion	ETV6(5) - ALK(20)	Fusion	NEGATIVE
FBXO36-ALK fusion	FBXO36(2) - 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
GALNT14-ALK fusion	GALNT14(1) - ALK(19)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(13) - ALK(20)	Fusion	NEGATIVE
GCC2-ALK fusion	GCC2(19) - 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
HNRNPA1-ALK fusion	HNRNPA1(2) - ALK(18)	Fusion	NEGATIVE
IGFBP5-ALK fusion	IGFBP5(1) - ALK(19)	Fusion	NEGATIVE
INTS9-ALK fusion	INTS9(8) - ALK(20)	Fusion	NEGATIVE
ITSN2-ALK fusion	ITSN2(14) - ALK(2)	Fusion	NEGATIVE
KANK2-ALK fusion	KANK2(4) - ALK(16)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KCNQ5-ALK fusion	KCNQ5(1) - ALK(10)	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
KLC1-ALK fusion	KLC1(9) - ALK(20)	Fusion	NEGATIVE
KTN1-ALK fusion	KTN1(43) - ALK(19)	Fusion	NEGATIVE
LMNA-ALK fusion	LMNA(2) - ALK(20)	Fusion	NEGATIVE
LMO7-ALK fusion	LMO7(15) - ALK(20)	Fusion	NEGATIVE
LRPPRC-ALK fusion	LRPPRC(23) - ALK(20)	Fusion	NEGATIVE
LRRFIP2-ALK fusion	LRRFIP2(26) - ALK(20)	Fusion	NEGATIVE
LTBP1-ALK fusion	LTBP1(19) - ALK(20)	Fusion	NEGATIVE
MALAT1-ALK fusion	MALAT1(1) - ALK(16)	Fusion	NEGATIVE
MANBA-ALK fusion	MANBA(14) - ALK(20)	Fusion	NEGATIVE
MAPRE3-ALK fusion	MAPRE3(7) - ALK(20)	Fusion	NEGATIVE
MCFD2-ALK fusion	MCFD2(1) - ALK(20)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(2) - ALK(7)	Fusion	NEGATIVE
MEMO1-ALK fusion	MEMO1(1) - ALK(2)	Fusion	NEGATIVE
MLPH-ALK fusion	MLPH(11) - ALK(20)	Fusion	NEGATIVE
MPRIP-ALK fusion	MPRIP(21) - ALK(20)	Fusion	NEGATIVE
MROH2B-ALK fusion	MROH2B(21) - ALK(12)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(9) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MSN-ALK fusion	MSN(11) - ALK(20)	Fusion	NEGATIVE
MYH10-ALK fusion	MYH10(38) - 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
MYO5C-ALK fusion	MYO5C(30) - ALK(20)	Fusion	NEGATIVE
MYT1L-ALK fusion	MYT1L(14) - ALK(20)	Fusion	NEGATIVE
NCOA1-ALK fusion	NCOA1(21) - ALK(1)	Fusion	NEGATIVE
NPM1-ALK fusion	NPM1(4) - ALK(20)	Fusion	NEGATIVE
NUMA1-ALK fusion	NUMA1(19) - ALK(20)	Fusion	NEGATIVE
OTOF-ALK fusion	OTOF(2) - ALK(3)	Fusion	NEGATIVE
PABPC1-ALK fusion	PABPC1(9) - ALK(20)	Fusion	NEGATIVE
PCM1-ALK fusion	PCM1(23) - ALK(20)	Fusion	NEGATIVE
PDE3A-ALK fusion	PDE3A(10) - ALK(8)	Fusion	NEGATIVE
PDE8B-ALK fusion	PDE8B(11) - ALK(20)	Fusion	NEGATIVE
PHACTR1-ALK fusion	PHACTR1(4) - ALK(20)	Fusion	NEGATIVE
PLEKHA7-ALK fusion	PLEKHA7(26) - ALK(19)	Fusion	NEGATIVE
PNPT1-ALK fusion	PNPT1(5) - ALK(4)	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
PPM1B-ALK fusion	PPM1B(1) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
PPP1R21-ALK fusion	PPP1R21(7) - ALK(20)	Fusion	NEGATIVE
PPP1R21-ALK fusion	PPP1R21(17) - ALK(20)	Fusion	NEGATIVE
PPP4R3B-ALK fusion	PPP4R3B(10) - ALK(2)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(2) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(5) - ALK(20)	Fusion	NEGATIVE
PRKAR1A-ALK fusion	PRKAR1A(10) - ALK(20)	Fusion	NEGATIVE
PRKAR1B-ALK fusion	PRKAR1B(4) - ALK(20)	Fusion	NEGATIVE
PRKAR2A-ALK fusion	PRKAR2A(2) - ALK(20)	Fusion	NEGATIVE
PRKCE-ALK fusion	PRKCE(1) - ALK(20)	Fusion	NEGATIVE
RANBP2-ALK fusion	RANBP2(18) - ALK(20)	Fusion	NEGATIVE
RNF213-ALK fusion	RNF213(20) - ALK(20)	Fusion	NEGATIVE
RRBP1-ALK fusion	RRBP1(20) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(6) - ALK(20)	Fusion	NEGATIVE
SATB1-ALK fusion	SATB1(5) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(22) - ALK(20)	Fusion	NEGATIVE
SEC31A-ALK fusion	SEC31A(21) - ALK(20)	Fusion	NEGATIVE
SFT2D1-ALK fusion	SFT2D1(1) - ALK(6)	Fusion	NEGATIVE
SLC34A2-ALK fusion	SLC34A2(1) - ALK(20)	Fusion	NEGATIVE
SLC8A1-ALK fusion	SLC8A1(1) - ALK(20)	Fusion	NEGATIVE
SLMAP-ALK fusion	SLMAP(11) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SP100-ALK fusion	SP100(4) - ALK(20)	Fusion	NEGATIVE
SPDYA-ALK fusion	SPDYA(6) - ALK(20)	Fusion	NEGATIVE
SPECC1L-ALK fusion	SPECC1L(9) - 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
SQSTM1-ALK fusion	SQSTM1(5) - ALK(20)	Fusion	NEGATIVE
SQSTM1-ALK fusion	SQSTM1(5) - ALK(19)	Fusion	NEGATIVE
STK32B-ALK fusion	STK32B(11) - ALK(20)	Fusion	NEGATIVE
STRN-ALK fusion	STRN(3) - ALK(20)	Fusion	NEGATIVE
TERT-ALK fusion	TERT(11) - ALK(5)	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(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(5) - ALK(19)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(6) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(20)	Fusion	NEGATIVE
TFG-ALK fusion	TFG(7) - ALK(19)	Fusion	NEGATIVE
THBS1-ALK fusion	THBS1(4) - ALK(19)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(20)	Fusion	NEGATIVE
TIMP3-ALK fusion	TIMP3(1) - ALK(19)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(32) - ALK(13)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(28) - ALK(20)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(20)	Fusion	NEGATIVE
TNS1-ALK fusion	TNS1(16) - ALK(17)	Fusion	NEGATIVE
TPM1-ALK fusion	TPM1(8) - ALK(20)	Fusion	NEGATIVE
TPM3-ALK fusion	TPM3(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPM4-ALK fusion	TPM4(7) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(15) - ALK(20)	Fusion	NEGATIVE
TPR-ALK fusion	TPR(4) - ALK(20)	Fusion	NEGATIVE
TRAF1-ALK fusion	TRAF1(6) - ALK(20)	Fusion	NEGATIVE
TRMT61B-ALK fusion	TRMT61B(1) - ALK(9)	Fusion	NEGATIVE
VCL-ALK fusion	VCL(16) - ALK(20)	Fusion	NEGATIVE
YPEL5-ALK fusion	YPEL5(1) - ALK(18)	Fusion	NEGATIVE
ZNF2-ALK fusion	ZNF2(1) - ALK(20)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(11)	Fusion	NEGATIVE
AGAP3-BRAF fusion	AGAP3(8) - BRAF(9)	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
AGK-BRAF fusion	AGK(2) - BRAF(8)	Fusion	NEGATIVE
AGTRAP-BRAF fusion	AGTRAP(5) - BRAF(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
AP3B1-BRAF fusion	AP3B1(22) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(3) - BRAF(9)	Fusion	NEGATIVE
ARMC10-BRAF fusion	ARMC10(4) - BRAF(11)	Fusion	NEGATIVE
ATG7-BRAF fusion	ATG7(18) - BRAF(9)	Fusion	NEGATIVE
BAIAP2L1-BRAF fusion	BAIAP2L1(12) - BRAF(9)	Fusion	NEGATIVE
BBS9-BRAF fusion	BBS9(19) - BRAF(4)	Fusion	NEGATIVE
BCAP29-BRAF fusion	BCAP29(7) - BRAF(11)	Fusion	NEGATIVE
BCL2L11-BRAF fusion	BCL2L11(3) - BRAF(10)	Fusion	NEGATIVE
BTF3L4-BRAF fusion	BTF3L4(3) - BRAF(11)	Fusion	NEGATIVE
C7orf73-BRAF fusion	C7orf73(2) - BRAF(9)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(2) - BRAF(8)	Fusion	NEGATIVE
CCDC6-BRAF fusion	CCDC6(1) - BRAF(9)	Fusion	NEGATIVE
CCDC91-BRAF fusion	CCDC91(11) - BRAF(9)	Fusion	NEGATIVE
CCNY-BRAF fusion	CCNY(1) - BRAF(10)	Fusion	NEGATIVE
CDC27-BRAF fusion	CDC27(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CDC42BPB-BRAF fusion	CDC42BPB(36) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(9)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(13) - BRAF(8)	Fusion	NEGATIVE
CDK5RAP2-BRAF fusion	CDK5RAP2(12) - BRAF(11)	Fusion	NEGATIVE
CEP89-BRAF fusion	CEP89(16) - BRAF(9)	Fusion	NEGATIVE
CLCN6-BRAF fusion	CLCN6(2) - BRAF(11)	Fusion	NEGATIVE
CLIP2-BRAF fusion	CLIP2(6) - BRAF(11)	Fusion	NEGATIVE
COBL-BRAF fusion	COBL(7) - BRAF(10)	Fusion	NEGATIVE
CUL1-BRAF fusion	CUL1(7) - BRAF(9)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(8) - BRAF(8)	Fusion	NEGATIVE
CUX1-BRAF fusion	CUX1(10) - BRAF(9)	Fusion	NEGATIVE
DGKI-BRAF fusion	DGKI(7) - BRAF(10)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(5) - BRAF(9)	Fusion	NEGATIVE
DLG1-BRAF fusion	DLG1(3) - BRAF(10)	Fusion	NEGATIVE
DOCK4-BRAF fusion	DOCK4(41) - BRAF(9)	Fusion	NEGATIVE
DSTYK-BRAF fusion	DSTYK(1) - BRAF(10)	Fusion	NEGATIVE
DYNC1I2-BRAF fusion	DYNC1I2(7) - BRAF(10)	Fusion	NEGATIVE
EML4-BRAF fusion	EML4(6) - BRAF(10)	Fusion	NEGATIVE
EPS15-BRAF fusion	EPS15(22) - BRAF(10)	Fusion	NEGATIVE
EPS15L1-BRAF fusion	EPS15L1(16) - BRAF(9)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(12) - BRAF(10)	Fusion	NEGATIVE
ERC1-BRAF fusion	ERC1(17) - BRAF(8)	Fusion	NEGATIVE
ESYT2-BRAF fusion	ESYT2(8) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FAM114A2-BRAF fusion	FAM114A2(9) - BRAF(11)	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
FAM73A-BRAF fusion	FAM73A(7) - BRAF(9)	Fusion	NEGATIVE
FCHSD1-BRAF fusion	FCHSD1(13) - BRAF(9)	Fusion	NEGATIVE
FXR1-BRAF fusion	FXR1(13) - BRAF(10)	Fusion	NEGATIVE
GATM-BRAF fusion	GATM(2) - BRAF(11)	Fusion	NEGATIVE
GHR-BRAF fusion	GHR(1) - BRAF(10)	Fusion	NEGATIVE
GIPC2-BRAF fusion	GIPC2(3) - BRAF(10)	Fusion	NEGATIVE
GIT2-BRAF fusion	GIT2(14) - BRAF(9)	Fusion	NEGATIVE
GNAI1-BRAF fusion	GNAI1(1) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(4) - BRAF(10)	Fusion	NEGATIVE
GTF2I-BRAF fusion	GTF2I(20) - BRAF(10)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(4) - BRAF(7)	Fusion	NEGATIVE
HERPUD1-BRAF fusion	HERPUD1(6) - BRAF(7)	Fusion	NEGATIVE
KANK1-BRAF fusion	KANK1(7) - BRAF(9)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(3) - BRAF(8)	Fusion	NEGATIVE
KCTD7-BRAF fusion	KCTD7(4) - BRAF(8)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(12) - BRAF(4)	Fusion	NEGATIVE
KDM7A-BRAF fusion	KDM7A(11) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(18) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(17) - BRAF(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(10)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(15) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(14) - BRAF(8)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(11)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(9)	Fusion	NEGATIVE
KIAA1549-BRAF fusion	KIAA1549(13) - BRAF(8)	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
KLHL7-BRAF fusion	KLHL7(4) - BRAF(10)	Fusion	NEGATIVE
KLHL7-BRAF fusion	KLHL7(5) - BRAF(9)	Fusion	NEGATIVE
LMO7-BRAF fusion	LMO7(7) - BRAF(9)	Fusion	NEGATIVE
LSM12-BRAF fusion	LSM12(3) - BRAF(9)	Fusion	NEGATIVE
LSM14A-BRAF fusion	LSM14A(9) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(5) - BRAF(9)	Fusion	NEGATIVE
MACF1-BRAF fusion	MACF1(60) - BRAF(9)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(17) - BRAF(10)	Fusion	NEGATIVE
MAD1L1-BRAF fusion	MAD1L1(16) - BRAF(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MBP-BRAF fusion	MBP(2) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(11)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(10)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(9)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(4) - BRAF(8)	Fusion	NEGATIVE
MKRN1-BRAF fusion	MKRN1(3) - BRAF(10)	Fusion	NEGATIVE
MYRIP-BRAF fusion	MYRIP(16) - BRAF(9)	Fusion	NEGATIVE
MZT1-BRAF fusion	MZT1(2) - BRAF(11)	Fusion	NEGATIVE
NFIC-BRAF fusion	NFIC(6) - BRAF(10)	Fusion	NEGATIVE
NRF1-BRAF fusion	NRF1(5) - BRAF(9)	Fusion	NEGATIVE
NUB1-BRAF fusion	NUB1(3) - BRAF(9)	Fusion	NEGATIVE
NUDCD3-BRAF fusion	NUDCD3(4) - BRAF(9)	Fusion	NEGATIVE
NUP214-BRAF fusion	NUP214(21) - BRAF(10)	Fusion	NEGATIVE
OSBPL1A-BRAF fusion	OSBPL1A(16) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(11) - BRAF(10)	Fusion	NEGATIVE
OSBPL9-BRAF fusion	OSBPL9(14) - BRAF(9)	Fusion	NEGATIVE
PACSIN2-BRAF fusion	PACSIN2(7) - BRAF(9)	Fusion	NEGATIVE
PAPSS1-BRAF fusion	PAPSS1(5) - BRAF(9)	Fusion	NEGATIVE
PARP12-BRAF fusion	PARP12(9) - BRAF(11)	Fusion	NEGATIVE
PCM1-BRAF fusion	PCM1(37) - BRAF(9)	Fusion	NEGATIVE
PHTF2-BRAF fusion	PHTF2(17) - BRAF(9)	Fusion	NEGATIVE
PICALM-BRAF fusion	PICALM(19) - BRAF(9)	Fusion	NEGATIVE
PJA2-BRAF fusion	PJA2(7) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PLIN3-BRAF fusion	PLIN3(1) - BRAF(9)	Fusion	NEGATIVE
PLXNA2-BRAF fusion	PLXNA2(9) - BRAF(2)	Fusion	NEGATIVE
POR-BRAF fusion	POR(3) - BRAF(11)	Fusion	NEGATIVE
PPFIBP2-BRAF fusion	PPFIBP2(3) - BRAF(11)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(10) - BRAF(9)	Fusion	NEGATIVE
PRKAR1B-BRAF fusion	PRKAR1B(9) - BRAF(9)	Fusion	NEGATIVE
PRKAR2B-BRAF fusion	PRKAR2B(1) - BRAF(10)	Fusion	NEGATIVE
RAD18-BRAF fusion	RAD18(7) - BRAF(10)	Fusion	NEGATIVE
RBM33-BRAF fusion	RBM33(3) - BRAF(11)	Fusion	NEGATIVE
RBMS3-BRAF fusion	RBMS3(11) - BRAF(11)	Fusion	NEGATIVE
RNF11-BRAF fusion	RNF11(1) - BRAF(11)	Fusion	NEGATIVE
RNF130-BRAF fusion	RNF130(3) - BRAF(9)	Fusion	NEGATIVE
RP2-BRAF fusion	RP2(3) - BRAF(10)	Fusion	NEGATIVE
RUNDC1-BRAF fusion	RUNDC1(1) - BRAF(9)	Fusion	NEGATIVE
SCRIB-BRAF fusion	SCRIB(27) - BRAF(11)	Fusion	NEGATIVE
SLC12A7-BRAF fusion	SLC12A7(17) - BRAF(11)	Fusion	NEGATIVE
SLC45A3-BRAF fusion	SLC45A3(1) - 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(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(9) - BRAF(8)	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(11)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(17) - BRAF(9)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(10)	Fusion	NEGATIVE
SND1-BRAF fusion	SND1(18) - BRAF(8)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(6) - BRAF(9)	Fusion	NEGATIVE
SOX6-BRAF fusion	SOX6(5) - BRAF(9)	Fusion	NEGATIVE
STRN3-BRAF fusion	STRN3(3) - BRAF(10)	Fusion	NEGATIVE
STXBP6-BRAF fusion	STXBP6(4) - BRAF(10)	Fusion	NEGATIVE
TANK-BRAF fusion	TANK(4) - BRAF(9)	Fusion	NEGATIVE
TAX1BP1-BRAF fusion	TAX1BP1(8) - BRAF(11)	Fusion	NEGATIVE
TMEM106B-BRAF fusion	TMEM106B(4) - BRAF(8)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(1) - BRAF(10)	Fusion	NEGATIVE
TMEM178B-BRAF fusion	TMEM178B(2) - BRAF(9)	Fusion	NEGATIVE
TMPRSS2-BRAF fusion	TMPRSS2(3) - BRAF(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TOM1L2-BRAF fusion	TOM1L2(11) - BRAF(9)	Fusion	NEGATIVE
TPR-BRAF fusion	TPR(14) - BRAF(7)	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
TRIM4-BRAF fusion	TRIM4(6) - BRAF(10)	Fusion	NEGATIVE
UBN2-BRAF fusion	UBN2(3) - BRAF(11)	Fusion	NEGATIVE
ZBTB8A-BRAF fusion	ZBTB8A(4) - BRAF(9)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(7) - BRAF(11)	Fusion	NEGATIVE
ZC3HAV1-BRAF fusion	ZC3HAV1(3) - BRAF(10)	Fusion	NEGATIVE
ZKSCAN5-BRAF fusion	ZKSCAN5(2) - BRAF(9)	Fusion	NEGATIVE
ZNF207-BRAF fusion	ZNF207(3) - BRAF(10)	Fusion	NEGATIVE
ZNF767P-BRAF fusion	ZNF767P(1) - BRAF(11)	Fusion	NEGATIVE
ZSCAN30-BRAF fusion	ZSCAN30(3) - BRAF(10)	Fusion	NEGATIVE
EGFRvIII	EGFR(1) - EGFR(8)	RNAExonVariant	NEGATIVE
ESR1-ARMT1 fusion	ESR1(3) - ARMT1(4)	Fusion	NEGATIVE
ESR1-ARMT1 fusion	ESR1(6) - ARMT1(2)	Fusion	NEGATIVE
ESR1-ARNT2 fusion	ESR1(7) - ARNT2(9)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-ATP2B2 fusion	ESR1(4) - ATP2B2(4)	Fusion	NEGATIVE
ESR1-BMPER fusion	ESR1(6) - BMPER(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(2)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(5)	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(8)	Fusion	NEGATIVE
ESR1-CCDC170 fusion	ESR1(2) - CCDC170(10)	Fusion	NEGATIVE
ESR1-CDK13 fusion	ESR1(7) - CDK13(13)	Fusion	NEGATIVE
ESR1-COA5 fusion	ESR1(4) - COA5(2)	Fusion	NEGATIVE
ESR1-DAB2 fusion	ESR1(6) - DAB2(3)	Fusion	NEGATIVE
ESR1-GYG2 fusion	ESR1(6) - GYG2(2)	Fusion	NEGATIVE
ESR1-MKL1 fusion	ESR1(6) - MKL1(7)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(6) - MTHFD1L(21)	Fusion	NEGATIVE
ESR1-MTHFD1L fusion	ESR1(7) - MTHFD1L(27)	Fusion	NEGATIVE
ESR1-NKAIN2 fusion	ESR1(6) - NKAIN2(2)	Fusion	NEGATIVE
ESR1-NOP2 fusion	ESR1(6) - NOP2(16)	Fusion	NEGATIVE
ESR1-PCDH11X fusion	ESR1(6) - PCDH11X(6)	Fusion	NEGATIVE
ESR1-PDE10A fusion	ESR1(7) - PDE10A(6)	Fusion	NEGATIVE
ESR1-PLEKHG1 fusion	ESR1(6) - PLEKHG1(12)	Fusion	NEGATIVE
ESR1-POLH fusion	ESR1(6) - POLH(2)	Fusion	NEGATIVE
ESR1-RMND1 fusion	ESR1(6) - RMND1(9)	Fusion	NEGATIVE
ESR1-SOX9 fusion	ESR1(6) - SOX9(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
ESR1-SYNE1 fusion	ESR1(6) - SYNE1(59)	Fusion	NEGATIVE
ESR1-TFG fusion	ESR1(6) - TFG(4)	Fusion	NEGATIVE
ESR1-TNRC6B fusion	ESR1(6) - TNRC6B(14)	Fusion	NEGATIVE
ESR1-YAP1 fusion	ESR1(6) - YAP1(4)	Fusion	NEGATIVE
PLEKHG1-ESR1 fusion	PLEKHG1(3) - ESR1(2)	Fusion	NEGATIVE
TNS3-ESR1 fusion	TNS3(16) - ESR1(5)	Fusion	NEGATIVE
ACPP-FGFR1 fusion	ACPP(10) - FGFR1(4)	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
BCR-FGFR1 fusion	BCR(4) - FGFR1(10)	Fusion	NEGATIVE
CNTRL-FGFR1 fusion	CNTRL(40) - FGFR1(10)	Fusion	NEGATIVE
CPSF6-FGFR1 fusion	CPSF6(8) - FGFR1(10)	Fusion	NEGATIVE
CUX1-FGFR1 fusion	CUX1(11) - FGFR1(10)	Fusion	NEGATIVE
EGFR-FGFR1 fusion	EGFR(24) - FGFR1(10)	Fusion	NEGATIVE
ERLIN2-FGFR1 fusion	ERLIN2(8) - FGFR1(2)	Fusion	NEGATIVE
ERVK3-1-FGFR1 fusion	ERVK3-1(3) - FGFR1(10)	Fusion	NEGATIVE
FGFR1-ADAM32 fusion	FGFR1(17) - ADAM32(14)	Fusion	NEGATIVE
FGFR1-EVI5 fusion	FGFR1(17) - EVI5(12)	Fusion	NEGATIVE
FGFR1-HOOK3 fusion	FGFR1(17) - HOOK3(5)	Fusion	NEGATIVE
FGFR1-TACC1 fusion	FGFR1(17) - TACC1(7)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(5) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP-FGFR1 fusion	FGFR1OP(6) - FGFR1(10)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR1OP-FGFR1 fusion	FGFR1OP(7) - FGFR1(10)	Fusion	NEGATIVE
FGFR1OP2-FGFR1 fusion	FGFR1OP2(4) - 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(4)	Fusion	NEGATIVE
FN1-FGFR1 fusion	FN1(22) - FGFR1(3)	Fusion	NEGATIVE
KAT6A-FGFR1 fusion	KAT6A(5) - FGFR1(2)	Fusion	NEGATIVE
LRRFIP1-FGFR1 fusion	LRRFIP1(8) - FGFR1(10)	Fusion	NEGATIVE
MYO18A-FGFR1 fusion	MYO18A(33) - FGFR1(10)	Fusion	NEGATIVE
RANBP2-FGFR1 fusion	RANBP2(20) - FGFR1(10)	Fusion	NEGATIVE
SNX29-FGFR1 fusion	SNX29(1) - FGFR1(4)	Fusion	NEGATIVE
SQSTM1-FGFR1 fusion	SQSTM1(6) - FGFR1(10)	Fusion	NEGATIVE
TPR-FGFR1 fusion	TPR(22) - FGFR1(10)	Fusion	NEGATIVE
TRIM24-FGFR1 fusion	TRIM24(11) - FGFR1(10)	Fusion	NEGATIVE
WHSC1L1-FGFR1 fusion	WHSC1L1(14) - FGFR1(5)	Fusion	NEGATIVE
ZMYM2-FGFR1 fusion	ZMYM2(17) - FGFR1(10)	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
CD44-FGFR2 fusion	CD44(1) - FGFR2(3)	Fusion	NEGATIVE
COL25A1-FGFR2 fusion	COL25A1(3) - FGFR2(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
CTNNB1-FGFR2 fusion	CTNNB1(1) - FGFR2(10)	Fusion	NEGATIVE
FGFR2-ACSL5 fusion	FGFR2(17) - ACSL5(15)	Fusion	NEGATIVE
FGFR2-AFF3 fusion	FGFR2(17) - AFF3(7)	Fusion	NEGATIVE
FGFR2-AHCYL1 fusion	FGFR2(17) - AHCYL1(2)	Fusion	NEGATIVE
FGFR2-ANKRD24 fusion	FGFR2(17) - ANKRD24(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(2)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(3)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(10)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(16)	Fusion	NEGATIVE
FGFR2-BICC1 fusion	FGFR2(17) - BICC1(18)	Fusion	NEGATIVE
FGFR2-CASP7 fusion	FGFR2(17) - CASP7(2)	Fusion	NEGATIVE
FGFR2-CBX5 fusion	FGFR2(17) - CBX5(2)	Fusion	NEGATIVE
FGFR2-CCAR2 fusion	FGFR2(17) - CCAR2(4)	Fusion	NEGATIVE
FGFR2-CCDC186 fusion	FGFR2(17) - CCDC186(4)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(2)	Fusion	NEGATIVE
FGFR2-CCDC6 fusion	FGFR2(17) - CCDC6(1)	Fusion	NEGATIVE
FGFR2-CELF2 fusion	FGFR2(17) - CELF2(4)	Fusion	NEGATIVE
FGFR2-CIT fusion	FGFR2(17) - CIT(23)	Fusion	NEGATIVE
FGFR2-CLIP2 fusion	FGFR2(17) - CLIP2(10)	Fusion	NEGATIVE
FGFR2-COL14A1 fusion	FGFR2(17) - COL14A1(34)	Fusion	NEGATIVE
FGFR2-CREB5 fusion	FGFR2(17) - CREB5(8)	Fusion	NEGATIVE
FGFR2-DDX21 fusion	FGFR2(17) - DDX21(11)	Fusion	NEGATIVE
FGFR2-EIF4A2 fusion	FGFR2(17) - EIF4A2(8)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-FAM76A fusion	FGFR2(17) - FAM76A(2)	Fusion	NEGATIVE
FGFR2-GAB2 fusion	FGFR2(17) - GAB2(2)	Fusion	NEGATIVE
FGFR2-INA fusion	FGFR2(17) - INA(2)	Fusion	NEGATIVE
FGFR2-KCTD1 fusion	FGFR2(17) - KCTD1(2)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(3)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(5)	Fusion	NEGATIVE
FGFR2-KIAA1217 fusion	FGFR2(17) - KIAA1217(6)	Fusion	NEGATIVE
FGFR2-KRT8 fusion	FGFR2(17) - KRT8(6)	Fusion	NEGATIVE
FGFR2-LAMC1 fusion	FGFR2(17) - LAMC1(27)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MCU fusion	FGFR2(17) - MCU(4)	Fusion	NEGATIVE
FGFR2-MGEA5 fusion	FGFR2(17) - MGEA5(12)	Fusion	NEGATIVE
FGFR2-NOL4 fusion	FGFR2(17) - NOL4(7)	Fusion	NEGATIVE
FGFR2-NRAP fusion	FGFR2(17) - NRAP(24)	Fusion	NEGATIVE
FGFR2-NRBF2 fusion	FGFR2(17) - NRBF2(4)	Fusion	NEGATIVE
FGFR2-OFD1 fusion	FGFR2(17) - OFD1(3)	Fusion	NEGATIVE
FGFR2-OGDH fusion	FGFR2(17) - OGDH(3)	Fusion	NEGATIVE
FGFR2-OPTN fusion	FGFR2(17) - OPTN(5)	Fusion	NEGATIVE
FGFR2-PASD1 fusion	FGFR2(17) - PASD1(10)	Fusion	NEGATIVE
FGFR2-PCM1 fusion	FGFR2(17) - PCM1(7)	Fusion	NEGATIVE
FGFR2-PHC1 fusion	FGFR2(17) - PHC1(2)	Fusion	NEGATIVE
FGFR2-PKD2L1 fusion	FGFR2(17) - PKD2L1(7)	Fusion	NEGATIVE
FGFR2-PPHLN1 fusion	FGFR2(17) - PPHLN1(3)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR2-RABGAP1L fusion	FGFR2(17) - RABGAP1L(20)	Fusion	NEGATIVE
FGFR2-RASAL2 fusion	FGFR2(17) - RASAL2(3)	Fusion	NEGATIVE
FGFR2-ROCK1 fusion	FGFR2(17) - ROCK1(2)	Fusion	NEGATIVE
FGFR2-SBNO2 fusion	FGFR2(17) - SBNO2(19)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(7)	Fusion	NEGATIVE
FGFR2-SHTN1 fusion	FGFR2(17) - SHTN1(5)	Fusion	NEGATIVE
FGFR2-SMN1 fusion	FGFR2(17) - SMN1(2)	Fusion	NEGATIVE
FGFR2-STK26 fusion	FGFR2(17) - STK26(3)	Fusion	NEGATIVE
FGFR2-TACC2 fusion	FGFR2(17) - TACC2(11)	Fusion	NEGATIVE
FGFR2-TACC3 fusion	FGFR2(17) - TACC3(11)	Fusion	NEGATIVE
FGFR2-TBC1D1 fusion	FGFR2(17) - TBC1D1(9)	Fusion	NEGATIVE
FGFR2-TFEC fusion	FGFR2(17) - TFEC(2)	Fusion	NEGATIVE
FGFR2-TJP2 fusion	FGFR2(17) - TJP2(5)	Fusion	NEGATIVE
FGFR2-TXLNA fusion	FGFR2(17) - TXLNA(6)	Fusion	NEGATIVE
FGFR2-UBQLN1 fusion	FGFR2(17) - UBQLN1(8)	Fusion	NEGATIVE
FGFR2-UTRN fusion	FGFR2(17) - UTRN(51)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(2)	Fusion	NEGATIVE
FGFR2-VCL fusion	FGFR2(17) - VCL(7)	Fusion	NEGATIVE
FGFR2-WAC fusion	FGFR2(17) - WAC(5)	Fusion	NEGATIVE
FGFR2-WARS fusion	FGFR2(17) - WARS(3)	Fusion	NEGATIVE
FGFR2-ZMYM4 fusion	FGFR2(17) - ZMYM4(10)	Fusion	NEGATIVE
KLK2-FGFR2 fusion	KLK2(1) - FGFR2(5)	Fusion	NEGATIVE
PARK2-FGFR2 fusion	PARK2(9) - FGFR2(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDHX-FGFR2 fusion	PDHX(1) - FGFR2(7)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(2)	Fusion	NEGATIVE
SLC45A3-FGFR2 fusion	SLC45A3(1) - FGFR2(1)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(8) - FGFR2(7)	Fusion	NEGATIVE
SNX19-FGFR2 fusion	SNX19(7) - FGFR2(7)	Fusion	NEGATIVE
ETV6-FGFR3 fusion	ETV6(5) - FGFR3(9)	Fusion	NEGATIVE
FGFR3-ADD1 fusion	FGFR3(17) - ADD1(3)	Fusion	NEGATIVE
FGFR3-AES fusion	FGFR3(17) - AES(2)	Fusion	NEGATIVE
FGFR3-AMBRA1 fusion	FGFR3(17) - AMBRA1(14)	Fusion	NEGATIVE
FGFR3-BAIAP2L1 fusion	FGFR3(17) - BAIAP2L1(2)	Fusion	NEGATIVE
FGFR3-BRAP fusion	FGFR3(17) - BRAP(5)	Fusion	NEGATIVE
FGFR3-CAMK2A fusion	FGFR3(18) - CAMK2A(14)	Fusion	NEGATIVE
FGFR3-ELAVL3 fusion	FGFR3(17) - ELAVL3(2)	Fusion	NEGATIVE
FGFR3-FBXO28 fusion	FGFR3(17) - FBXO28(4)	Fusion	NEGATIVE
FGFR3-JAKMIP1 fusion	FGFR3(17) - JAKMIP1(4)	Fusion	NEGATIVE
FGFR3-NBR1 fusion	FGFR3(17) - NBR1(7)	Fusion	NEGATIVE
FGFR3-RBBP4 fusion	FGFR3(17) - RBBP4(12)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(4)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(6)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(7)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(8)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(10)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(11)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	Fusion	NEGATIVE
FGFR3-TACC3 fusion	FGFR3(17) - TACC3(14)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
FGFR3-TACC3 fusion	FGFR3(18) - TACC3(11)	Fusion	NEGATIVE
BAIAP2L1-MET fusion	BAIAP2L1(9) - MET(15)	Fusion	NEGATIVE
C8orf34-MET fusion	C8orf34(2) - 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
CLIP2-MET fusion	CLIP2(11) - MET(15)	Fusion	NEGATIVE
DCTN1-MET fusion	DCTN1(26) - MET(15)	Fusion	NEGATIVE
EPS15-MET fusion	EPS15(21) - MET(15)	Fusion	NEGATIVE
ETV6-MET fusion	ETV6(4) - MET(15)	Fusion	NEGATIVE
GTF2I-MET fusion	GTF2I(6) - MET(15)	Fusion	NEGATIVE
HLA-DRB1-MET fusion	HLA-DRB1(5) - MET(15)	Fusion	NEGATIVE
KIF5B-MET fusion	KIF5B(24) - MET(15)	Fusion	NEGATIVE
LRRFIP1-MET fusion	LRRFIP1(19) - MET(15)	Fusion	NEGATIVE
MET KDD	MET(21) - MET(15)	RNAExonVariant	NEGATIVE
MET exon 14 skipping (RNA)	MET(13) - MET(15)	RNAExonVariant	NEGATIVE
OXR1-MET fusion	OXR1(9) - MET(13)	Fusion	NEGATIVE
OXR1-MET fusion	OXR1(1) - MET(13)	Fusion	NEGATIVE
PPFIBP1-MET fusion	PPFIBP1(9) - MET(15)	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

Variant Name	Genes (Exons)	Variant Type	Test Result
SCAF11-MET fusion	SCAF11(1) - MET(2)	Fusion	NEGATIVE
SLC34A2-MET fusion	SLC34A2(4) - MET(15)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(2)	Fusion	NEGATIVE
ST7-MET fusion	ST7(1) - MET(11)	Fusion	NEGATIVE
STARD3NL-MET fusion	STARD3NL(6) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(5) - MET(15)	Fusion	NEGATIVE
TFG-MET fusion	TFG(7) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(36) - MET(15)	Fusion	NEGATIVE
TPR-MET fusion	TPR(4) - MET(15)	Fusion	NEGATIVE
TRIM4-MET fusion	TRIM4(6) - MET(15)	Fusion	NEGATIVE
ZKSCAN1-MET fusion	ZKSCAN1(3) - MET(15)	Fusion	NEGATIVE
AKAP13-NRG1 fusion	AKAP13(5) - NRG1(2)	Fusion	NEGATIVE
ATP1B1-NRG1 fusion	ATP1B1(2) - 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
CDH1-NRG1 fusion	CDH1(2) - NRG1(2)	Fusion	NEGATIVE
CLU-NRG1 fusion	CLU(2) - NRG1(6)	Fusion	NEGATIVE
COX10-AS1-NRG1 fusion	COX10-AS1(1) - NRG1(2)	Fusion	NEGATIVE
HMBOX1-NRG1 fusion	HMBOX1(1) - NRG1(6)	Fusion	NEGATIVE
KIF13B-NRG1 fusion	KIF13B(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
NOTCH2-NRG1 fusion	NOTCH2(4) - NRG1(6)	Fusion	NEGATIVE
PARP8-NRG1 fusion	PARP8(1) - NRG1(2)	Fusion	NEGATIVE
PDE7A-NRG1 fusion	PDE7A(3) - NRG1(6)	Fusion	NEGATIVE
RAB3IL1-NRG1 fusion	RAB3IL1(9) - NRG1(6)	Fusion	NEGATIVE
RALGAPA1-NRG1 fusion	RALGAPA1(20) - NRG1(6)	Fusion	NEGATIVE
ROCK1-NRG1 fusion	ROCK1(2) - NRG1(2)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(4) - NRG1(6)	Fusion	NEGATIVE
SDC4-NRG1 fusion	SDC4(2) - NRG1(6)	Fusion	NEGATIVE
SETD4-NRG1 fusion	SETD4(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(2) - NRG1(2)	Fusion	NEGATIVE
SLC3A2-NRG1 fusion	SLC3A2(5) - NRG1(6)	Fusion	NEGATIVE
SMAD4-NRG1 fusion	SMAD4(1) - NRG1(6)	Fusion	NEGATIVE
TBL1XR1-NRG1 fusion	TBL1XR1(1) - NRG1(2)	Fusion	NEGATIVE
TENM4-NRG1 fusion	TENM4(12) - NRG1(2)	Fusion	NEGATIVE
THAP7-NRG1 fusion	THAP7(3) - NRG1(2)	Fusion	NEGATIVE
THBS1-NRG1 fusion	THBS1(6) - NRG1(6)	Fusion	NEGATIVE
TNC-NRG1 fusion	TNC(10) - NRG1(6)	Fusion	NEGATIVE
TSHZ2-NRG1 fusion	TSHZ2(1) - NRG1(6)	Fusion	NEGATIVE
VAMP2-NRG1 fusion	VAMP2(4) - NRG1(4)	Fusion	NEGATIVE
VTCN1-NRG1 fusion	VTCN1(2) - NRG1(2)	Fusion	NEGATIVE
WHSC1L1-NRG1 fusion	WHSC1L1(1) - NRG1(2)	Fusion	NEGATIVE
WRN-NRG1 fusion	WRN(30) - NRG1(2)	Fusion	NEGATIVE
ZMYM2-NRG1 fusion	ZMYM2(2) - NRG1(2)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
AFAP1-NTRK1 fusion	AFAP1(4) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF2-NTRK1 fusion	ARHGEF2(21) - NTRK1(10)	Fusion	NEGATIVE
ARHGEF25-NTRK1 fusion	ARHGEF25(11) - NTRK1(8)	Fusion	NEGATIVE
BANP-NTRK1 fusion	BANP(4) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(12) - NTRK1(10)	Fusion	NEGATIVE
BCAN-NTRK1 fusion	BCAN(13) - NTRK1(11)	Fusion	NEGATIVE
CD74-NTRK1 fusion	CD74(7) - NTRK1(10)	Fusion	NEGATIVE
CEL-NTRK1 fusion	CEL(7) - NTRK1(9)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(10)	Fusion	NEGATIVE
CHTOP-NTRK1 fusion	CHTOP(5) - NTRK1(11)	Fusion	NEGATIVE
CTRC-NTRK1 fusion	CTRC(1) - NTRK1(9)	Fusion	NEGATIVE
EPHB2-NTRK1 fusion	EPHB2(3) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
EPS15-NTRK1 fusion	EPS15(21) - NTRK1(9)	Fusion	NEGATIVE
GON4L-NTRK1 fusion	GON4L(21) - NTRK1(9)	Fusion	NEGATIVE
GRIPAP1-NTRK1 fusion	GRIPAP1(22) - NTRK1(11)	Fusion	NEGATIVE
GSN-NTRK1 fusion	GSN(7) - NTRK1(10)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(10)	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
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(11)	Fusion	NEGATIVE
IRF2BP2-NTRK1 fusion	IRF2BP2(1) - NTRK1(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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(10)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(5) - NTRK1(11)	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(11)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(12)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(10) - NTRK1(13)	Fusion	NEGATIVE
LMNA-NTRK1 fusion	LMNA(11) - NTRK1(11)	Fusion	NEGATIVE
LRRC71-NTRK1 fusion	LRRC71(1) - NTRK1(9)	Fusion	NEGATIVE
MDM4-NTRK1 fusion	MDM4(3) - NTRK1(5)	Fusion	NEGATIVE
MPRIP-NTRK1 fusion	MPRIP(21) - NTRK1(12)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
MRPL24-NTRK1 fusion	MRPL24(1) - NTRK1(9)	Fusion	NEGATIVE
NFASC-NTRK1 fusion	NFASC(20) - NTRK1(10)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(16) - DYNC2H1(85)	Fusion	NEGATIVE
NTRK1-DYNC2H1 fusion	NTRK1(17) - DYNC2H1(85)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PEAR1-NTRK1 fusion	PEAR1(2) - NTRK1(9)	Fusion	NEGATIVE
PEAR1-NTRK1 fusion	PEAR1(15) - NTRK1(10)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(21) - NTRK1(9)	Fusion	NEGATIVE
PLEKHA6-NTRK1 fusion	PLEKHA6(16) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(10)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(11)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(22) - NTRK1(13)	Fusion	NEGATIVE
PPL-NTRK1 fusion	PPL(12) - NTRK1(13)	Fusion	NEGATIVE
RABGAP1L-NTRK1 fusion	RABGAP1L(14) - NTRK1(16)	Fusion	NEGATIVE
RFWD2-NTRK1 fusion	RFWD2(2) - NTRK1(3)	Fusion	NEGATIVE
RNF213-NTRK1 fusion	RNF213(15) - NTRK1(12)	Fusion	NEGATIVE
SCYL3-NTRK1 fusion	SCYL3(11) - NTRK1(12)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(2) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(10)	Fusion	NEGATIVE
SQSTM1-NTRK1 fusion	SQSTM1(5) - NTRK1(12)	Fusion	NEGATIVE
SSBP2-NTRK1 fusion	SSBP2(12) - NTRK1(12)	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
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

Variant Name	Genes (Exons)	Variant Type	Test Result
TP53-NTRK1 fusion	TP53(8) - NTRK1(9)	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(9)	Fusion	NEGATIVE
TPM3-NTRK1 fusion	TPM3(7) - 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(6) - NTRK1(11)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(9)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(10)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(12)	Fusion	NEGATIVE
TPR-NTRK1 fusion	TPR(21) - NTRK1(13)	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
TRIM63-NTRK1 fusion	TRIM63(8) - NTRK1(9)	Fusion	NEGATIVE
VANGL2-NTRK1 fusion	VANGL2(1) - NTRK1(12)	Fusion	NEGATIVE
AFAP1-NTRK2 fusion	AFAP1(14) - NTRK2(12)	Fusion	NEGATIVE
AGBL4-NTRK2 fusion	AGBL4(6) - NTRK2(16)	Fusion	NEGATIVE
BCR-NTRK2 fusion	BCR(1) - NTRK2(17)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
DAB2IP-NTRK2 fusion	DAB2IP(1) - NTRK2(17)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(4) - NTRK2(16)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(15)	Fusion	NEGATIVE
ETV6-NTRK2 fusion	ETV6(5) - NTRK2(16)	Fusion	NEGATIVE
GKAP1-NTRK2 fusion	GKAP1(9) - NTRK2(13)	Fusion	NEGATIVE
GNAQ-NTRK2 fusion	GNAQ(2) - NTRK2(16)	Fusion	NEGATIVE
KCTD8-NTRK2 fusion	KCTD8(1) - NTRK2(13)	Fusion	NEGATIVE
NACC2-NTRK2 fusion	NACC2(4) - NTRK2(13)	Fusion	NEGATIVE
NAV1-NTRK2 fusion	NAV1(15) - NTRK2(11)	Fusion	NEGATIVE
NOS1AP-NTRK2 fusion	NOS1AP(9) - NTRK2(11)	Fusion	NEGATIVE
PAN3-NTRK2 fusion	PAN3(1) - NTRK2(17)	Fusion	NEGATIVE
QKI-NTRK2 fusion	QKI(6) - NTRK2(16)	Fusion	NEGATIVE
SLMAP-NTRK2 fusion	SLMAP(14) - NTRK2(16)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(13)	Fusion	NEGATIVE
SQSTM1-NTRK2 fusion	SQSTM1(5) - NTRK2(17)	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
STRN3-NTRK2 fusion	STRN3(7) - NTRK2(16)	Fusion	NEGATIVE
TBC1D2-NTRK2 fusion	TBC1D2(6) - NTRK2(12)	Fusion	NEGATIVE
TLE4-NTRK2 fusion	TLE4(7) - NTRK2(15)	Fusion	NEGATIVE
TP63-NTRK2 fusion	TP63(8) - NTRK2(12)	Fusion	NEGATIVE
TRAF2-NTRK2 fusion	TRAF2(9) - NTRK2(15)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(15)	Fusion	NEGATIVE
TRIM24-NTRK2 fusion	TRIM24(12) - NTRK2(16)	Fusion	NEGATIVE
VCAN-NTRK2 fusion	VCAN(6) - NTRK2(10)	Fusion	NEGATIVE
VCL-NTRK2 fusion	VCL(16) - NTRK2(12)	Fusion	NEGATIVE
WNK2-NTRK2 fusion	WNK2(24) - NTRK2(16)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(14) - NTRK3(14)	Fusion	NEGATIVE
AKAP13-NTRK3 fusion	AKAP13(16) - NTRK3(15)	Fusion	NEGATIVE
BTBD1-NTRK3 fusion	BTBD1(4) - NTRK3(14)	Fusion	NEGATIVE
COX5A-NTRK3 fusion	COX5A(1) - NTRK3(15)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(2) - NTRK3(14)	Fusion	NEGATIVE
EML4-NTRK3 fusion	EML4(6) - NTRK3(14)	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(15)	Fusion	NEGATIVE
ETV6-NTRK3 fusion	ETV6(5) - NTRK3(14)	Fusion	NEGATIVE
FAT1-NTRK3 fusion	FAT1(2) - NTRK3(7)	Fusion	NEGATIVE
FBXL20-NTRK3 fusion	FBXL20(1) - NTRK3(6)	Fusion	NEGATIVE
HNRNPA2B1-NTRK3 fusion	HNRNPA2B1(7) - NTRK3(14)	Fusion	NEGATIVE
KANK1-NTRK3 fusion	KANK1(3) - NTRK3(14)	Fusion	NEGATIVE
LYN-NTRK3 fusion	LYN(8) - NTRK3(14)	Fusion	NEGATIVE
MYH9-NTRK3 fusion	MYH9(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(13)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
MYO5A-NTRK3 fusion	MYO5A(32) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(14)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(12)	Fusion	NEGATIVE
MYO5A-NTRK3 fusion	MYO5A(23) - NTRK3(11)	Fusion	NEGATIVE
NTRK3-HOMER1 fusion	NTRK3(17) - HOMER1(2)	Fusion	NEGATIVE
NTRK3-HOMER2 fusion	NTRK3(17) - HOMER2(2)	Fusion	NEGATIVE
NTRK3-LOXL2 fusion	NTRK3(3) - LOXL2(5)	Fusion	NEGATIVE
RBPMS-NTRK3 fusion	RBPMS(5) - 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
TPM4-NTRK3 fusion	TPM4(7) - NTRK3(12)	Fusion	NEGATIVE
UBE2R2-NTRK3 fusion	UBE2R2(3) - NTRK3(4)	Fusion	NEGATIVE
ZNF710-NTRK3 fusion	ZNF710(1) - NTRK3(14)	Fusion	NEGATIVE
AFF1-NUTM1 fusion	AFF1(17) - NUTM1(3)	Fusion	NEGATIVE
BCORL1-NUTM1 fusion	BCORL1(1) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CIC-NUTM1 fusion	CIC(17) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(2)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(18) - NUTM1(5)	Fusion	NEGATIVE
CIC-NUTM1 fusion	CIC(20) - NUTM1(5)	Fusion	NEGATIVE
MXD1-NUTM1 fusion	MXD1(5) - NUTM1(2)	Fusion	NEGATIVE
WHSC1L1-NUTM1 fusion	WHSC1L1(7) - NUTM1(2)	Fusion	NEGATIVE
ZNF618-NUTM1 fusion	ZNF618(12) - NUTM1(2)	Fusion	NEGATIVE
ABI3BP-RET fusion	ABI3BP(34) - RET(12)	Fusion	NEGATIVE
ACBD5-RET fusion	ACBD5(11) - RET(12)	Fusion	NEGATIVE
ADD3-RET fusion	ADD3(1) - RET(12)	Fusion	NEGATIVE
AFAP1-RET fusion	AFAP1(3) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(6) - RET(12)	Fusion	NEGATIVE
AFAP1L2-RET fusion	AFAP1L2(2) - RET(12)	Fusion	NEGATIVE
AKAP13-RET fusion	AKAP13(35) - RET(12)	Fusion	NEGATIVE
ANK3-RET fusion	ANK3(1) - RET(12)	Fusion	NEGATIVE
BCR-RET fusion	BCR(4) - RET(12)	Fusion	NEGATIVE
CCDC186-RET fusion	CCDC186(7) - 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

Variant Name	Genes (Exons)	Variant Type	Test Result
CCDC6-RET fusion	CCDC6(1) - RET(10)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(12)	Fusion	NEGATIVE
CCDC6-RET fusion	CCDC6(1) - RET(11)	Fusion	NEGATIVE
CUX1-RET fusion	CUX1(10) - RET(12)	Fusion	NEGATIVE
DLG5-RET fusion	DLG5(13) - RET(12)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(11)	Fusion	NEGATIVE
EML4-RET fusion	EML4(19) - RET(12)	Fusion	NEGATIVE
EPHA5-RET fusion	EPHA5(2) - 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
FGFR1OP-RET fusion	FGFR1OP(11) - RET(11)	Fusion	NEGATIVE
FGFR1OP-RET fusion	FGFR1OP(12) - RET(12)	Fusion	NEGATIVE
FKBP15-RET fusion	FKBP15(25) - RET(12)	Fusion	NEGATIVE
FRMD4A-RET fusion	FRMD4A(12) - RET(12)	Fusion	NEGATIVE
GOLGA5-RET fusion	GOLGA5(7) - RET(12)	Fusion	NEGATIVE
GPHN-RET fusion	GPHN(9) - RET(12)	Fusion	NEGATIVE
HOOK3-RET fusion	HOOK3(11) - RET(12)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(10)	Fusion	NEGATIVE
KIAA1217-RET fusion	KIAA1217(11) - RET(11)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
KIAA1468-RET fusion	KIAA1468(10) - RET(12)	Fusion	NEGATIVE
KIF13A-RET fusion	KIF13A(18) - 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(8)	Fusion	NEGATIVE
KIF5B-RET fusion	KIF5B(23) - RET(12)	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
KIF5B-RET fusion	KIF5B(15) - RET(12)	Fusion	NEGATIVE
KTN1-RET fusion	KTN1(29) - RET(12)	Fusion	NEGATIVE
MPRIP-RET fusion	MPRIP(19) - RET(12)	Fusion	NEGATIVE
MYH10-RET fusion	MYH10(33) - RET(12)	Fusion	NEGATIVE
MYH13-RET fusion	MYH13(35) - RET(12)	Fusion	NEGATIVE
MYO5A-RET fusion	MYO5A(23) - RET(11)	Fusion	NEGATIVE
MYO5C-RET fusion	MYO5C(25) - 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
OLFM4-RET fusion	OLFM4(4) - RET(10)	Fusion	NEGATIVE
PCM1-RET fusion	PCM1(29) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
PDCD10-RET fusion	PDCD10(4) - RET(11)	Fusion	NEGATIVE
PICALM-RET fusion	PICALM(19) - RET(12)	Fusion	NEGATIVE
PPFIBP2-RET fusion	PPFIBP2(9) - RET(12)	Fusion	NEGATIVE
PRKAR1A-RET fusion	PRKAR1A(7) - RET(12)	Fusion	NEGATIVE
PRKG1-RET fusion	PRKG1(3) - RET(12)	Fusion	NEGATIVE
RASAL2-RET fusion	RASAL2(15) - RET(12)	Fusion	NEGATIVE
RASSF4-RET fusion	RASSF4(3) - RET(12)	Fusion	NEGATIVE
RRBP1-RET fusion	RRBP1(9) - RET(12)	Fusion	NEGATIVE
RUFY1-RET fusion	RUFY1(17) - RET(12)	Fusion	NEGATIVE
RUFY2-RET fusion	RUFY2(9) - RET(12)	Fusion	NEGATIVE
SATB1-RET fusion	SATB1(7) - RET(12)	Fusion	NEGATIVE
SNRNP70-RET fusion	SNRNP70(7) - RET(12)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(11)	Fusion	NEGATIVE
SPECC1L-RET fusion	SPECC1L(10) - RET(12)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(5) - RET(11)	Fusion	NEGATIVE
SQSTM1-RET fusion	SQSTM1(6) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(12)	Fusion	NEGATIVE
TBL1XR1-RET fusion	TBL1XR1(9) - RET(11)	Fusion	NEGATIVE
TFG-RET fusion	TFG(4) - RET(11)	Fusion	NEGATIVE
TNIP1-RET fusion	TNIP1(11) - RET(12)	Fusion	NEGATIVE
TNIP2-RET fusion	TNIP2(5) - RET(12)	Fusion	NEGATIVE
TRIM24-RET fusion	TRIM24(9) - RET(12)	Fusion	NEGATIVE
TRIM27-RET fusion	TRIM27(3) - RET(12)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
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
WAC-RET fusion	WAC(3) - RET(12)	Fusion	NEGATIVE
CCDC30-ROS1 fusion	CCDC30(10) - ROS1(36)	Fusion	NEGATIVE
CCDC6-ROS1 fusion	CCDC6(5) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(7) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(35)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(34)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(33)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(6) - ROS1(32)	Fusion	NEGATIVE
CD74-ROS1 fusion	CD74(4) - ROS1(33)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(12) - ROS1(35)	Fusion	NEGATIVE
CEP85L-ROS1 fusion	CEP85L(8) - ROS1(36)	Fusion	NEGATIVE
CLIP1-ROS1 fusion	CLIP1(20) - ROS1(36)	Fusion	NEGATIVE
CLTC-ROS1 fusion	CLTC(31) - ROS1(35)	Fusion	NEGATIVE
ERC1-ROS1 fusion	ERC1(11) - ROS1(36)	Fusion	NEGATIVE
EYS-ROS1 fusion	EYS(12) - ROS1(9)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(35)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(34)	Fusion	NEGATIVE
EZR-ROS1 fusion	EZR(10) - ROS1(33)	Fusion	NEGATIVE
GOLGB1-ROS1 fusion	GOLGB1(9) - ROS1(35)	Fusion	NEGATIVE
GOPC-ROS1 fusion	GOPC(8) - ROS1(35)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
GOPC-ROS1 fusion	GOPC(4) - ROS1(36)	Fusion	NEGATIVE
HLA-A-ROS1 fusion	HLA-A(7) - ROS1(34)	Fusion	NEGATIVE
LIMA1-ROS1 fusion	LIMA1(10) - ROS1(36)	Fusion	NEGATIVE
LRIG3-ROS1 fusion	LRIG3(16) - ROS1(35)	Fusion	NEGATIVE
MSN-ROS1 fusion	MSN(9) - ROS1(34)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(32) - ROS1(35)	Fusion	NEGATIVE
MYO5A-ROS1 fusion	MYO5A(23) - ROS1(35)	Fusion	NEGATIVE
MYO5C-ROS1 fusion	MYO5C(30) - ROS1(35)	Fusion	NEGATIVE
NCOR2-ROS1 fusion	NCOR2(7) - ROS1(36)	Fusion	NEGATIVE
NFKB2-ROS1 fusion	NFKB2(14) - ROS1(36)	Fusion	NEGATIVE
PPFIBP1-ROS1 fusion	PPFIBP1(9) - ROS1(35)	Fusion	NEGATIVE
PWWP2A-ROS1 fusion	PWWP2A(1) - ROS1(36)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(5) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(4) - ROS1(32)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(34)	Fusion	NEGATIVE
SDC4-ROS1 fusion	SDC4(2) - ROS1(32)	Fusion	NEGATIVE
SHTN1-ROS1 fusion	SHTN1(11) - ROS1(36)	Fusion	NEGATIVE
SLC16A10-ROS1 fusion	SLC16A10(3) - ROS1(3)	Fusion	NEGATIVE
SLC22A16-ROS1 fusion	SLC22A16(7) - ROS1(16)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(35)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(4) - ROS1(32)	Fusion	NEGATIVE

Variant Name	Genes (Exons)	Variant Type	Test Result
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(34)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(33)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - ROS1(32)	Fusion	NEGATIVE
SLC34A2-ROS1 fusion	SLC34A2(13) - 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
TMEM106B-ROS1 fusion	TMEM106B(3) - ROS1(35)	Fusion	NEGATIVE
TPD52L1-ROS1 fusion	TPD52L1(3) - ROS1(33)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(7) - ROS1(35)	Fusion	NEGATIVE
TPM3-ROS1 fusion	TPM3(3) - ROS1(36)	Fusion	NEGATIVE
TSPAN3-ROS1 fusion	TSPAN3(5) - ROS1(31)	Fusion	NEGATIVE
YWHAE-ROS1 fusion	YWHAE(4) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(3) - ROS1(36)	Fusion	NEGATIVE
ZCCHC8-ROS1 fusion	ZCCHC8(2) - ROS1(36)	Fusion	NEGATIVE
EIF3E-RSPO2 fusion	EIF3E(1) - RSPO2(2)	Fusion	NEGATIVE
GRHL2-RSPO2 fusion	GRHL2(8) - 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

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
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