510(k) SUBSTANTIAL EQUIVALENCE DETERMINATION
DECISION SUMMARY

A. 510(k) Number:

K190661

B. Purpose for Submission:

New Device

C. Measurand:

Somatic single nucleotide variants, insertions, deletions, and tumor mutational burden (TMB) in human genomic DNA obtained from formalin-fixed, paraffin-embedded tumor tissue. Refer to Appendix 1a for a complete list of the genes in the assay.

D. Type of Test:

Next generation sequencing tumor profiling test

E. Applicant:

NantHealth, Inc.

F. Proprietary and Established Names:

Trade Name: Omics Core
Common Name: NantHealth Next Generating Sequencing Tumor Profiling Test

G. Regulatory Information:

1. **Regulation section:**

   21 CFR 866.6080

2. **Classification:**

   Class II

3. **Product code:**

   PZM

4. **Panel:**

   88-Pathology

H. Intended Use:

1. Indications for use:

The Omics Core assay is a qualitative in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed paraffin-embedded tumor tissue matched with normal specimens from patients with solid malignant neoplasms to detect tumor gene alterations in a broad multi gene panel. The test is intended to provide information on somatic mutations (point mutations and small insertions and deletions) and tumor mutational burden (TMB) for use by qualified health care professionals in accordance with professional guidelines, and is not conclusive or prescriptive for labeled use of any specific therapeutic product. Omics Core is a single-site assay performed at NantHealth, Inc.

2. Special conditions for use statement(s):

For *in vitro* diagnostic use only

For prescription use only

3. Special instrument requirements:

Illumina NovaSeq6000 (qualified by NantHealth)

I. Device Description:

A description of required equipment, software, reagents, vendors, and storage conditions were provided, and are described in the product labeling (NantHealth Omics Core manual). NantHealth assumes responsibility for the device.The device is a NGS-based tumor profiling assay, which sequences both tumor and matched normal specimens to detect somatic mutations.

1. Specimen Preparation

The tumor volume and minimum tumor content needed to obtain sufficient DNA for testing to achieve the necessary quality performance are shown in the Table 1 below:

Table 1: Specimen Handling and Processing FFPE Tissue

Tissue Type	Volume	Macrodissection requirements and Minimum Tumor Proportion;	Limitations	Storage
Formalin-fixed, paraffin-embedded (FFPE) Tumor tissue blocks or slides.	5-20 unstained section, 10 micron thick	Yes; more than 10% of tumor cells; sections containing > 20% viable tumor are preferred. For TMB testing ≥ 20% tumor cells.	Decalcified tissues are unsuitable for analysis	Room temp-erature

Genomic DNA is extracted from tissue specimens using Qiagen's QIAsymphony DSP DNA MiniK it per manufacturer's protocol. Matched normal control samples are

processed from blood (2 x 2.5ml in PAxGene Blood DNA collection tubes). DNA is quantified and concentrated if necessary. The amount of DNA required to perform the test is 50-300ng. Genomic DNA is stored at –20°C until assay initiation. DNA shearing is conducted per protocol and a quality control check is reviewed. Average fragment size for sheared DNA should be approximately ~200bp for tissue and approximately ~300bp for blood. Sheared DNA proceeds directly to library preparation.

2. **Library Preparation**

Sequence libraries are prepared using KAPA Biosystems Library Preparation Reagents by first producing blunt-ended, 5'-phosphorylated fragments. To the 3' ends of the dsDNA library fragments, dAMP is added (A-tailing). Next, dsDNA adapters with 3'dTMP is ligated to the A-tailed library fragments. Library fragments with appropriate adapter sequences are amplified via ligation-mediated pre-capture PCR. A quality control review on the amplified DNA libraries is performed: Samples should be a smear with average fragment peak size for tissue as approximately ~200bp and average blood peak is approximately ~300bp with a fluorescence above 20. The concentration should be10-300ng/µL to ensure adequate hybridization for capture.

3. **Hybrid Capture**

Library capture is conducted using IDT Capture reagents. Sequencing libraries are hybridized to the vendor oligo pool. Capture beads are used to pull down the complex of capture oligos and genomic DNA fragments. Unbound fragments are washed away. The enriched fragment pool is amplified by ligation mediated-PCR (LM-PCR). The success of the enrichment is measured as a quality control step: Samples should be a smear with an average peak fragment size for tissue at approximately ~200bp and an average peak fragment size for blood at approximately ~300bp with a fluorescence above 20. The concentration of the amplified DNA library should be 1-45ng/µL; the LM-PCR yield should be ≥ 25ng. Reactions can be stored at 4°C until ready for purification, up to 72 hours.

4. **Sequencing and Data Analysis**

Sequencing is conducted using the Illumina NovaSeq 6000 sequencing instruments and reagents along with PhiX Control v3. The sequencing process uses multiple quality checks.

   a. **Data Management System (DMS):** Automated sample tracking and archival of run-associated metadata (barcode, samples accession number, requisition number, source (class), and specimen type) is conducted with the following key functions: Tracking sample status through various stages of data analysis; tracking iterations of analysis applied to a given sample; recording versions of databases and algorithms used in analysis; archival of selected pipeline output files (FASTQ, BAM, VCF) and sequencing run statistics (e.g. yield, error rate, unassigned read indices).

   b. **Demultiplexing and FASTQ Generation:** The analysis pipeline uses software provided by Illumina. Multiple pairs of FASTQ files are generated per sample corresponding to full length forward and reverse reads. Demultiplexing quality

control includes quality metrics for per-base sequence quality, sequence content, GC content and sequence length distribution, and relative percentages of unmatched indices.

c. Indexing QC Check: The potential for index or sample contamination is managed during mutation calling and determined by looking for the presence of common polymorphisms in tumor samples that are different from the polymorphisms present in the matched-normal equivalent sample.

d. Read Alignment and BAM Generation: Spurious adapter sequences are trimmed prior to read alignment. Reads are aligned in paired-end mode to the Genome Reference Consortium Human Build 37 (GRCh37, aka hg19) version of the human genome. Aligned reads are written to a Sequence Alignment Map (SAM) file, which is then converted into Binary Alignment Map (BAM) format. PCR duplicates are flagged and excluded. Each base within a read is assigned a base quality score by the sequencing software, which reflects the probability an error was made with the base call. To account for systemic biases that may not accurately reflect the actual error probabilities observed empirically, the analysis pipeline uses another tool to adjust the reported quality scores based on the selected covariates. Control for low quality bases during identification as part of variant calling results in low quality reads being excluded.

e. Sample QC Checks: The baits used for hybridization capture include probes targeting >200,000 regions throughout the genome containing common single nucleotide polymorphisms (SNPs). The unique combination of SNPs specific to a given sample serves as a 'fingerprint' for the identity of the corresponding patient, and serves to identify potential sample mix-ups and contamination between samples and barcodes. QC checks involve the use of fingerprint' SNPs.

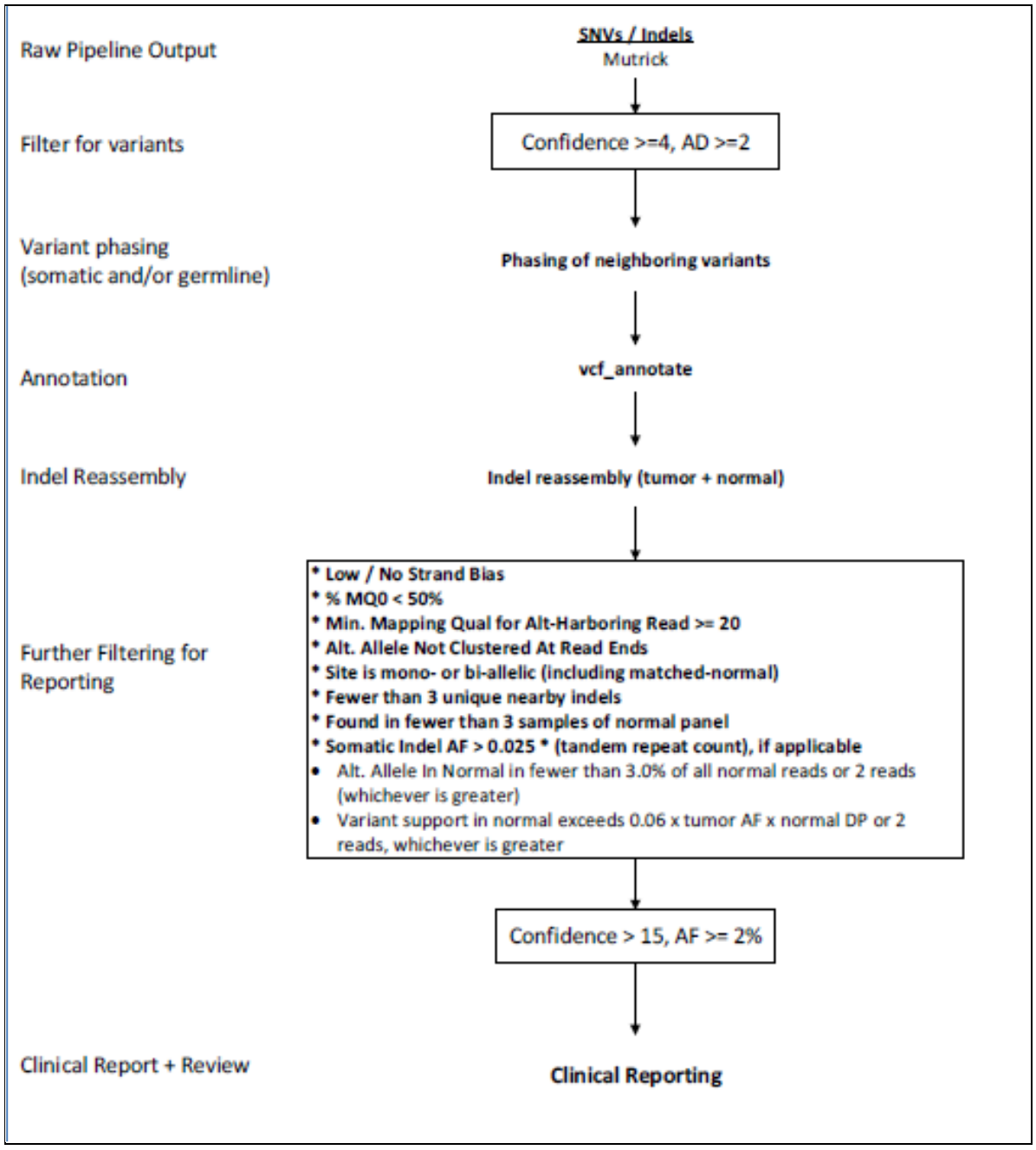
i. ***Sample mix-up check:*** The analysis pipeline computes the % Similarity between the normal and tumor samples associated with the same patient, where % Similarity is defined as the percentage "compatible" genotypes shared by the two samples among a random set of up to 1,000 dbSNP loci with sufficient coverage (coverage depth ≥60 in both tumor and normal samples). If the % Similarity falls below 97%, the tumor and normal samples are flagged for review as potentially mismatched samples.

ii. ***Sample contamination checks*:** The percentage of somatic variants that overlap common dbSNP loci (where "common" is defined as a polymorphism with a global population allele frequency > 5%) is calculated (% Common). If % Common exceeds 5% of the total somatic variant burden, the sample is flagged for review as potentially contaminated with unrelated DNA.

iii. ***Check for presence of tumor in normal:*** Tumor contamination in matched-normal samples is assessed by calculating the number and percentage of somatic calls that meet the following criteria:
1. Two (2) or more reads supporting mutant allele in matched-normal, and
2. Mutant allele fraction > 5% in matched-normal sample.

f. Mutation calling – SNVs and Indels: The analysis pipeline identifies two classes of mutations: (1) single nucleotide variants (SNVs) and (2) indels. Paired sample mutation calling is performed on tumor samples and their respective matched normal controls. Filtering is performed to remove low quality sequence data, sources of sequencing artifacts, and germline results. Summary of mutation filtering scheme is provided in Figure 1.

Figure 1: Summary of the mutation filtering scheme for Omics Core



i. ***Analysis of positive and negative controls*:** Data from controls is used to confirm lack of contamination as well as analytical sensitivity.

ii. ***Filters on sample coverage***: Sequencing coverage of ≥ 500X is required for tumor samples and sequencing coverage of ≥ 150X is required for normal samples.

iii. ***Filtering for high confidence mutations:*** Raw SNV and indel calls are subjected to a series of filtering steps to ensure only high-confidence calls are reported. These parameters include (1) evidence of it being a somatic mutations (i.e., log odds of tumor data at site explained by presence of somatic mutation vs. sequencing error > 6.3 (>4.3 for indels) in tumor, log odds of normal data at site explained by sequencing error vs. presence of somatic mutation > 2.2, (2) reference set of common artifacts found in 3 or more normal samples from a panel of >4,000 normal blood samples, (3) technical characteristics that use coverage depth (DP), number of mutant reads (AD), mutation allele fraction (AF), strand bias (SB), characteristics of reads supporting mutant allele (mismatch quality sum, nearby indels, mapping quality), and characteristics of region where mutant is located (adjacent simple tandem repeat, overlapping dbSNP markers). Omics Core does not report mutations below 2%.

g. Mutation Annotation Predicted functional effect and clinical interpretation for each mutation is curated by automated software using information from several databases.

5. <u>Controls</u>

a. Matched Normal Control: Genomic DNA is extracted from patient-matched normal whole blood. NantHealth requires a normal sample to perform the test.

b. Positive Control: The positive control sample (commercial vendor qualified by NantHealth) contains different confirmed mutations, representing a range of mutation allele frequencies. Results are compared against a negative control as an unmatched normal. Data generated from the positive control sample is analyzed using the pipeline, and frequencies of the detected mutations are reviewed to determine if (1) the known mutations are among those called, and (2) the observed frequencies for the known mutations match their expected values within 10%. The percentage agreement for scenarios (1) and (2) is expected to be 95% or higher.

c. Negative Control: The negative control sample is a purchased reference sample, verified to be free of tumor contamination and germline copy number mutations in a majority of genes. Single Nucleotide Polymorphisms (SNPs) present in the negative control have been identified in prior analyses. The observed genotypes are compared against the expected genotypes for 420 common SNPs, and the degree of concordance is measured by identifying the percentage of SNPs that are identical or compatible. The percentage agreement between expected and observed mutations is expected to be 97% or higher.

d. PCR Reagent Control – No Template Control (NTC): The NTC control should have a quantification measurement of < 1.0ng/μL. If this is not the case, the measurement may be high due to the presence of unincorporated primers carried over

from the LM-PCR reaction and not an indication of possible contamination amplified sample libraries. If the quantification measurement is ≥ 1ng/μL and determined not to be due to dimers, the sample will be flagged and reviewed to determine if a re-run or sequencing run is necessary.

6. Result Reporting:

- Oncopanel results are reported out under one of the two categories: "Cancer Mutations with Evidence of Clinical Significance" or "Cancer Mutations with Potential Clinical Significance". The two categories are based on the supporting level of clinical evidence. Refer to the Clinical Performance Section for more information.
- Results are reported for point mutations and small insertions and deletions in protein-coding exons of the 468 gene panel. Refer to Appendix 1a for a list of genes
- The Omics Core does not report mutations in 203 exons due to low sequence coverage, poor mapping quality, or high GC content. Refer to Appendix 1b for a list of excluded exons.
- Tumor Mutational Burden (TMB) Reporting: TMB is measured by counting the total number of somatic, non-synonymous exonic variants per the total number of genes (19,396) surveyed by the Omics Core assay. TMB is measured as mutations per megabase (mut/Mb), reported as a rate and included in the category for "Cancer Mutations with Potential Clinical Significance"

Reporting takes in account the following quality metrics in the Table 2 below.

Table 2. Flowcell and Sample Level Quality Control Metrics

QC Metrics	Acceptance Criteria
Flowcell Metrics	
Sequencing Output S2	> 850 Gb
Error Rate %	< 2
Base Quality Q30 %	> 75
Sample Metrics	
Average Target Coverage	≥ 500X
Coverage Uniformity	≥ 95% target exons above 100X
Mutation Calling Threshold	AF ≥ 0.02, Conf > 15
Normal DNA: Homozygosity of X	Male > 75% Female < 75%
Tumor DNA: % Interchromosomal	< 16
Provenance: % Similarity	> 90
Contamination: % Common	> 5% total and > 10 common site somatic mutations
Criteria for calling test failure	If a sample presents with mean coverage across all exons < 50X and no mutations are detected due to the low overall coverage, the test is deemed "failed" for the sample.

J. Substantial Equivalence Information:

1. Predicate device name(s):

MSK-IMPACT (Integrated Mutation Profiling of Actionable Cancer Targets)

2. Predicate 510(k) number(s):

DEN170058

3. Comparison with predicate:

Table 3: Comparison of Omics Core with Predicate Device

Characteristic	Predicate Device: MSK-IMPACT (DEN170058)	Subject Device: Omics Core
Similarities		
Indications For Use	The MSK-IMPACT assay is a qualitative in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed paraffin-embedded tumor tissue matched with normal specimens from patients with solid malignant neoplasms to detect tumor gene alterations in a broad multi gene panel. The test is intended to provide information on somatic mutations (point mutations and small insertions and deletions) and microsatellite instability for use by qualified health care professionals in accordance with professional guidelines, and is not conclusive or prescriptive for labeled use of any specific therapeutic product. MSK-IMPACT is a single-site assay performed at Memorial Sloan Kettering Cancer Center.	The Omics Core assay is a qualitative in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed paraffin-embedded tumor tissue matched with normal specimens from patients with solid malignant neoplasms to detect tumor gene alterations in a broad multi gene panel. The test is intended to provide information on somatic mutations (point mutations and small insertions and deletions) and tumor mutational burden (TMB) for use by qualified health care professionals in accordance with professional guidelines, and is not conclusive or prescriptive for labeled use of any specific therapeutic product. Omics Core is a single-site assay performed at NantHealth, Inc.
Technology	Hybrid Capture	Same
Specimen Types	Formalin-fixed, paraffin-embedded (FFPE) tumor tissue matched with normal specimens from patients with solid malignant neoplasms	Same
Target Population	Patients with solid malignant neoplasms	Same

Characteristic	**Predicate Device:** **MSK-IMPACT (DEN170058)**	**Subject Device:** **Omics Core**
Similarities		
Genes on Panel	468	Same
Test Environment	Single-site assay (performed at Memorial Sloan Kettering Cancer Center)	Same. Single-site assay (performed at NantHealth, Inc.)
Controls	• Matched normal • Positive control • Negative control • No template control (NTC)	Omics Core uses the same types of controls.
Result Report Format	Oncopanel results are reported under one of these two categories: • "Cancer Mutations with Evidence of Clinical Significance" or • "Cancer Mutations with Potential Clinical Significance."	Same

Characteristic	Predicate (MSK IMPACT)	New Device (Omics Core)
Differences		
Black List	73 exons	203 exons
Variant types	Intended to provide information on somatic mutations (point mutations and small insertions and deletions), and microsatellite instability	Same except Omics Core provides information on tumor mutational burden (TMB) and not microsatellite instability (MSI).
Instrument	Illumina HiSeq® 2500 Sequencing System	Illumina NovaSeq™ 6000 Sequencing System
Determination of Pipeline Thresholds	• Based on >200X target coverage, • 100X for ≥ 98% target exons, • hotspot mutation calling threshold (mutation coverage (DP) ≥ 20, mutant reads (AD) ≥ 8, mutation frequency (VF) ≥ 2%, and non-hotspot mutation threshold (DP ≥ 20, AD ≥ 10, VF ≥ 5%)	Based on ≥ 500X target coverage, ≥ 100X for 95% of target exons, and a mutation calling threshold of allele frequency (AF) ≥ 2% with Conf > 15 and heuristic filters, The minimum read depth for variants in the Omics Core assay are allele depth (AD) ≥ 2 and overall depth (DP) ≥ 4.
Assay cut-off	MSK-IMPACT does not report mutations below 2% for known hotspot mutations and 5% for non-hotspot mutations.	Omics Core does not report mutations below 2% for all mutations

Clinical Evidence Curation	Classification criteria were developed by MSK using the in-house OncoKB database. OncoKB undergoes periodic updates through the review of new information by a panel of experts	Classification criteria were developed by NantHealth, Inc. NantHealth periodically updates Omics Core through the review of new information available.

K. Standard/Guidance Document Referenced (if applicable):

Not applicable

L. Test Principle:

The NantHealth Omics Core assay is a custom targeted sequencing platform, utilizing solution-phase exon capture and sequencing, to detect somatic alterations (point mutations, small insertions and deletions), and overall tumor mutation burden in tumor specimens. The NantHealth Omics Core assay involves hybridization capture and deep sequencing of 468 cancer-associated genes for reporting of variants. TMB is reported on the basis of SNVs in the protein-coding exons of 19,396 genes including all known oncogenes and tumor suppressors. Omics Core reports on 468 genes from within the larger panel, reporting cancer mutations with evidence of clinical significance and cancer mutations with potential for clinical significance.The assay is not cleared for whole exome reporting of individual variants outside of the 468 genes. DNA probes are synthesized by a secondary manufacturer and are biotinylated to enable sequence enrichment through capture by streptavidin-conjugated beads. In total, the probes target approximately 39Mb of the human genome.

Genomic DNA is extracted from both a tumor and a patient-matched normal control sample. Sequence libraries are prepared through a series of enzymatic steps including shearing of double-stranded DNA, end repair, A-base addition, ligation of barcoded sequence adaptors, and low cycle PCR amplification. Single barcoded sequence libraries are captured using the biotinylated probes. Captured DNA fragments are then pooled and sequenced on an Illumina NovaSeq 6000 as paired-end reads. Sequence reads are then aligned to the reference human genome. By comparing the identity of bases from the tumor DNA to the matched normal DNA and the reference human genome, somatic alterations are identified in the tumor.

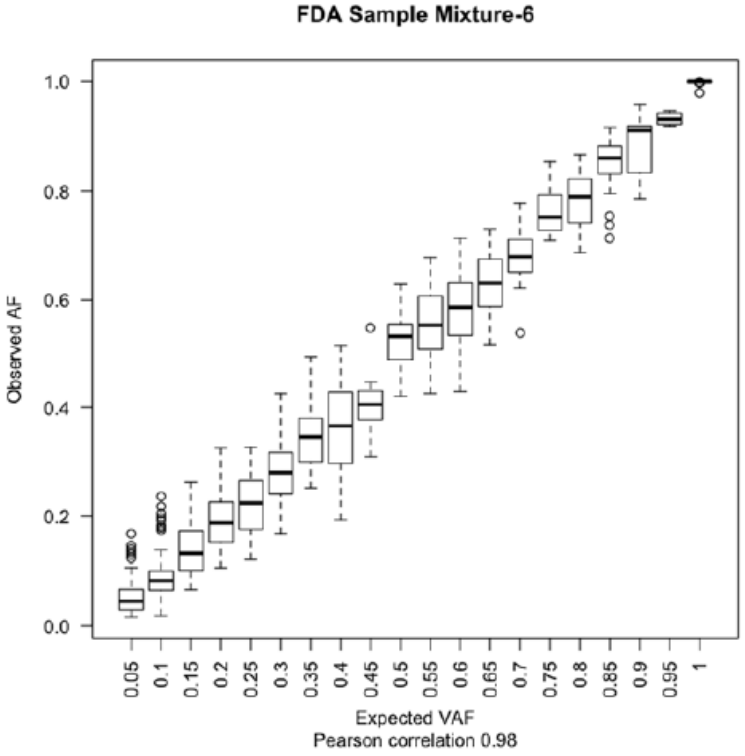
M. Performance:

1. **<u>Determination of pipeline thresholds</u>**
   a. **Requirements on exon coverage establishment:** A power analysis to compute the coverage or total number of reads needed to detect a mutation with true underlying mutation frequency 2% or greater, for varying levels of power (0.8 to 0.99), assuming a fixed alpha (Type I error rate) of 0.05 was conducted. Additionally, the 95% confidence interval ranges of observed mutation frequency as a function of coverage was also calculated. When the mutation is present at 10%, the 95% confidence interval with a coverage of 500X is expected to fall between 7.5% and 13%. When the overall coverage is 100X, the 95% CI for a mutation at 10% is estimated to fall

between 5.0% and 17.6%. To confirm these estimates, empirical data was obtained to measure the range of observed VF to expected VF using DNA from 10 normal FFPE samples from unrelated individuals which was mixed in equimass parts so as to create a range of SNPs with expected frequencies as low as 5%. A total of 672 common SNPs were considered for this experiment. A boxplot for one of the mixtures, mixture 6, showing the observed mutation frequencies for the 672 common SNPs genotyped in the pooled normal sample binned by their true underlying mutation frequency is shown in Figure 2. The results demonstrated that an observed VAF range of 4.2% to 20.3% for a SNP with true underlying mutation frequency of 10% when the mean coverage of the sample was 968X. This range in values is wider than the expected range of 7.5% to 13% and likely due to slight variations in the true mixing percentages. This data provided support for using a 5% as the lower limit for reporting mutations detected with true underlying frequency of 10%. The increased VAF range observed at the expected 10% frequency was also shown to be due to the different protocols and methods used for specimens sent to NantHealth e.g., fixation time, storage conditions, etc. These processing variations manifest as variable DNA damage and impact the sample DNA to library conversion rate (conversion efficiency), and thus result in differing sample representation within both a mixture of samples and the final sequencing data.

The boxplot in Figure 2 shows a correlation of 0.983, with a slope of .997 and intercept of -0.0096. Consistent correlation is established as >0.9 as for the whole pool analyzed.

Figure 2. Observed vs Expected Variant Frequency



b. Requirements on sample coverage: Ten normal (diploid) FFPE samples were profiled in duplicate using the Omics Core assay (total = 20 replicates) to generate summary statistics across all targeted exons. The mean coverage across all targeted exons for the normal FFPE samples was 810X (SD = 106X). Summary statistics were also computed on coverage values per exon normalized by per-sample coverage. There were exons that presented with consistently low coverage values, due to sequence similarity with other loci and high GC content; however, none of those exons were among the clinical validation genes and were removed from the Omics Core assay. Of the remaining exons across all genes, 99.9% were sequenced to a depth of 100X or greater while 91.2% were sequenced to a depth of 250X or greater. This analysis of normal FFPE samples indicates that with a mean sample coverage of 810X, 98% of exons are sequenced with coverage greater than 170X, or with normalized coverage greater than 0.21. (The 'mean-normalized coverage' is the coverage of the mutation divided by the mean coverage across all exons; it serves as a measure of how deeply the validation exon was sequenced relative to the overall coverage of the sample. A mean-normalized coverage below 1 indicates the exon coverage is below average; conversely if greater than 1, it indicates above average coverage.) The data are shown in Figure 3a and 3b.

Figure 3a. Distribution of mean coverage values for targeted exons. (Dashed line indicates coverage at 100X).

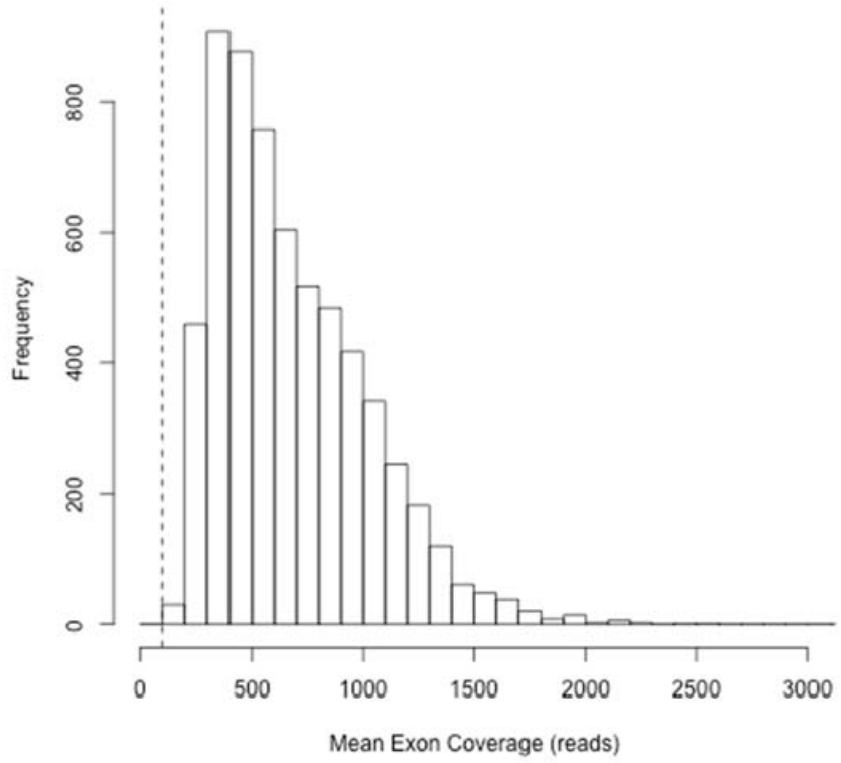
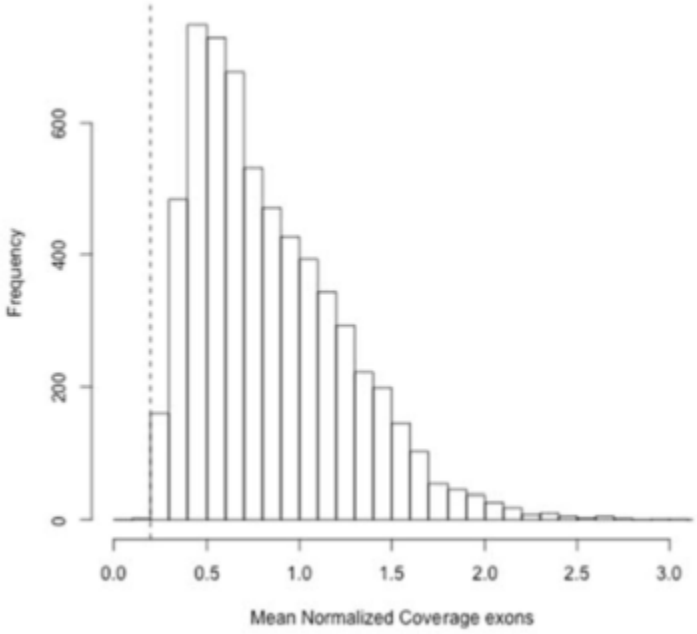


Figure 3a. Distribution of mean coverage values for targeted exons normalized by per- sample coverage. (Dashed line indicates 20% of mean sample coverage).



Based on the calculations, 98% of exons can be expected to be sequenced to coverage greater than 100X, when mean sample coverage is 476X (0.21 * 476X = 100X). (A 100X minimum coverage threshold per exon is required based on the power calculations, which showed 100X coverage was necessary to call mutations with true underlying mutation frequency 10% or greater, with 95% power at an alpha level of 0.05).

To be conservative, a threshold of 500X on mean sample coverage is used to determine if a sample is sequenced to sufficient depth for subsequent analysis. A sample is flagged as being at increased risk of false negatives it its mean coverage is below 500X.

To provide empirical data for these requirements, NantHealth utilizes the pooled normal FFPE sample with known expected single nucleotide mutations (n = 974) and the underlying mutation allele fractions (MAF). In silico downsampling analysis was conducted with a pooled normal mixture down to 46% where the sample coverage decreased from 968X to 450X. At this coverage level, 135 of the 139 (97%; 93-99% CI) mutations with expected underlying MAF of 10% were called.

c. **Requirements on mutation calling threshold:** Permissive standard filters were used to intentionally generate false positives to identify suitable thresholds for parameters such as variant frequency (VF) and heuristic filters to optimize specificity. The following criteria allows optimal rejection of false positive SNVs and indel calls, while maintaining the ability to detect true positive events with underlying frequency of 10% (5-17.6% observable). Potential strand-bias and heuristic filters (ctl, laf, mq0, np, str, end, tri, idl, grm, phs) are also evaluated in the standard somatic mutation

calling pipeline. An example of the number of false positive events detected pre and post filtering for the mixture sample using the standard somatic mutation calling pipeline is shown in Table 4.

Table 4: Sample Error Correction Pre and Post filtering on Metrcs

	Mutations	
	SNVs	Indels
Pre-filter	147	136
Post-filter	1	0
Rejection Rate	0.993	1.0

2. Pre-analytical Performance

Table 5 below displays the invalid rates across 42 different tumor types (in FFPE) gathered from in-house historical data from testing >2000 samples. The data shows the separate invalid rates for the different steps involved in the assay including the percentage of specimens with insufficient tumor, the percentage with failed library prep and the percentage that failed the sequencing run per cancer type. The range of invalid rates was 0.0% to 33..33% which were principally due to insufficient tumor and related to the specimen source. Invalid results due to sequencing failures were below 7.2% except for pancreas which was twice that but still acceptable. The data shows that the DNA extraction is valid across tumor types and interference effects from different specimens are not significant across different tumor types supporting the performance of the pan-cancer specimen handling.

Table 5. Omics Core Invalid Rates from Historical Data

Tumor Type	**Number of Tests**	**Pre-Run Invalids**	**Pre-Run Invalids**	**Pre-Run Invalids**	**Post-Run Invalids**	**Percent Invalids**
		Insufficient Tumor (Tumor % < 20%)	**Insufficient DNA (DNA yield <50ng)**	**Library Prep QC Failure**	**Sequencing QC Failure**	
Adrenal	8	0	2	0	0	25.00%
Ampulla of Vater	3	0	0	0	0	0.00%
Anal	12	2	0	0	1	25.00%
Bile Duct (extrahepatic)	11	1	0	0	0	9.09%
Biliary Tract (intrahepatic)	11	2	0	0	1	27.27%
Bladder	35	1	0	0	1	5.71%

Tumor Type	Number of Tests	Pre-Run Invalids	Pre-Run Invalids	Pre-Run Invalids	Post-Run Invalids	Percent Invalids
		Insufficient Tumor (Tumor % < 20%)	Insufficient DNA (DNA yield <50ng)	Library Prep QC Failure	Sequencing QC Failure	
Bone and Soft Tissue Cancers (including Sarcoma)	120	4	4	0	5	10.83%
Brain	87	1	1	0	3	5.75%
Breast	362	33	10	1	26	19.34%
Cervical	27	0	0	0	1	3.70%
Colon	191	15	1	2	13	16.23%
Esophageal	41	4	1	0	2	17.07%
Gall Bladder	18	0	1	0	1	11.11%
Gastric (Stomach)	41	2	0	0	4	14.63%
Gastrointestinal Stromal Tumor (GIST)	3	1	0	0	0	33.33%
Head and Neck	76	4	1	0	5	13.16%
Kidney	21	1	1	0	3	23.81%
Liver	36	0	0	0	1	2.78%
Lung	234	58	5	1	14	33.33%
Melanoma	39	2	1	0	2	12.82%
Mesothelioma	4	0	0	0	0	0.00%
Oral and Throat Cancers (Including Thyroid)	31	0	1	1	3	16.13%
Ovarian	121	5	0	1	5	9.09%
Pancreatic	151	20	1	0	22	28.48%
Penile	2	0	0	0	0	0.00%
Prostate	96	11	4	0	4	19.79%
Rectal	56	7	0	2	3	21.43%
Renal Pelvis and Ureter Cancers	16	0	1	0	1	12.50%
Skin (Non-Melanoma)	16	2	0	0	1	18.75%
Small Intestine	12	0	0	0	0	0.00%

Tumor Type	Number of Tests	Pre-Run Invalids	Pre-Run Invalids	Pre-Run Invalids	Post-Run Invalids	Percent Invalids
		Insufficient Tumor (Tumor % < 20%)	Insufficient DNA (DNA yield <50ng)	Library Prep QC Failure	Sequencing QC Failure	
Thymic	6	0	0	0	0	0.00%
Thyroid	2	0	0	0	0	0.00%
Unknown Primary	35	5	0	0	0	14.29%
Uterine (Endometrial)	77	2	0	0	4	7.79%
Vulvar	2	0	0	0	0	0.00%
Urethral	2	0	0	0	0	0.00%
Eye	1	0	0	0	0	0.00%
Metastasis	2	0	0	0	0	0.00%
Testicular	1	0	0	0	0	0.00%
Vaginal	3	0	0	0	0	0.00%
Soft Tissue	3	0	0	0	0	0.00%
Totals	2,189	204	38	9	141	17.91%
% Invalid by Category	N/A	9.32%	1.74%	0.41%	6.44%	17.91%

3. Analytical Performance:

The hybridization-capture-based targeted sequencing assay is designed to detect point mutations [also referred to as single nucleotide variants (SNVs)] as well as small insertions/deletions (indels) < 21 bp in length in the coding exons of 468 genes (See Appendix 1 and 2). A paired-sample analysis pipeline (tumor vs. matched normal blood) is used to identify somatic mutations in the targeted exons. A representative approach to validation of the SNVs and indels targeted in this panel was submitted at the variant level, and asseside quality metrics (data not shown).

a. Precision Studies

Reproducibility and repeatability were assessed in precision studies for Omics Core. Extracted DNA was run once per day for 3 non-consecutive days using different barcodes for inter-day assessment (n = 3). For one run, a sample was run in triplicate for intra-day assessment, resulting in a total of 3+1+1=5 replicates. For each replicate tested, all observed mutations in the selected genes were reported and assessed for precision. Details of the studies are described below.

i. *Precision Panel:* The precision of Omics Core was assessed using 12 FFPE clinical tumor samples and 1 commercial cell line (n = 13). The panel represented different tumor types, different mutation types, and a range of mutant allele frequencies. The specimen panel was selected based on known mutations corresponding to "Cancer Mutations with Evidence of Clinical Significance." Table 6 shows the list of specimens used in this study. A total of 530 variants were identified in the clinical and cell line samples that included 483 SNVs, 32 deletions and 15 insertions. Precision of each variant is reported across all samples to provide an assessment of the error rate.

Table 6. Summary of the Specimens and Allele Frequencies in the Precision Studies

Tumor Types	Mutation Type	Mutation	Gene / Exon	cDNA Change	Amino Acid Change	Mutation Allele Frequency
Non-Small Cell Lung Cancer	DEL	In-Frame Deletion	EGFR Exon 19	c.2235_2249delGGAA	p.E746_A750del	~8
Colon Cancer	SNV	Missense	BRAF Exon 15	c.1799T>A	p.V600E	~30
Pancreatic Cancer	SNV	Missense	KRAS Exon 2	c.35G>A	p.G12D	~19
Mullerian Ovarian Cancer	SNV	Missense	PIK3CA Exon 10	c.1624G>A	p.E542K	~80[1]
Endometrial Cancer	SNV	Missense	CTCF Exon 3	c.263C>A	p.A88D	~36
Adenoid Cystic Carcinoma	INS	In-Frame Insertion	BCOR Exon 4	c.2936_2937insATTCAAATG	p.K978_F977ins*	~31
Malignant Gastric Gist	INS	In-Frame Insertion	KIT Exon 9	c.1504_1509dupGCCTAT	p.Y503_A502ins	~36
Colon Cancer	INS	Frame Shift Ins.	PTEN Exon 5	c.440dupA	p.A148Gfs*32	~57
Colon Cancer	DEL	Frame Shift Del.	PTEN Exon 5	c.262_264delinsAG	p.Y88Sfs*11	~62
Metastatic Breast Cancer	DEL	In-Frame Deletion	PIK3CA Exon 2	c.312_317delAGTAGG	p.V105_G106del	~62
Glioblastoma	SNV	Missense	PIK3C2G Exon 20	c.2090T>C	p.L697P	~35
Melanoma	SNV	Missense	BRAF Exon 15	c.1798_1799delinsAA	p.V600K	~29

[1] Due to copy number gain of the gene, the MAF was the same as the tumor purity.

Tumor Types	Mutation Type	Mutation	Gene / Exon	cDNA Change	Amino Acid Change	Mutation Allele Frequency
Commercial FFPE cell line	DEL, SNV	Hotspot mutations in BRAF, KIT, EGFR, KRAS, NRAS, PIK3CA				~3 - 25

ii. *Precision – Panel-Wide Reproducibility:* The precision was reported for all detected mutants in all replicates for each sample. The results demonstrate that the overall positive call rate for all variants analyzed across the 12 FFPE clinical samples and one commercial cell line was 2607/2650, or 98.4% (97.8-98.8% CI). The coefficient of variation (%CV) for the mutation allele frequency was calculated for all 5 replicates. Three-hundred and ninety-eight (398) of 521 (76%) mutations in the clinical specimens had %CV ≤ 10%; 91 of 521 (17%) were between 10 and 20%; and 32 of 521 (6%) were > 20%. Discordance was observed for variants in the following genes as summarized in Table 7 below. Each specimen is separated by a dark gray line. Known mutation within each specimen are in **bold**. Discordant cases are denoted in light grey. All runs passed the quality metrics criteria.

Table7 : Panel-wide precision summary for all 5 replicates Abbreviations: NC (normalized coverage); MAF (Mutant allele frequency)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
EGFR Exon 19	**c.2235_2249delGGAATTAAGAGAAGC**	**p.E746_A750del**	**0.67-0.82**	**0.03-0.06**	**0.05**	**0.05**	**0.01**	**23.25 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
TP53 Exon 8	c.818G>A	p.R273H	0.98-1.07	0.07-0.11	0.09	0.09	0.01	15.23 %	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 1[a]	c.1379C>G	p.A460G	2.43-2.92	0.01-0.03	0.02	0.01	0.01	39.35 %	4 of 5	80.0% (28.4%, 99.5%)
PIK3CA Exon 10	c.1633G>A	p.E545K	0.15-0.23	0.05-0.14	0.08	0.08	0.03	36.52 %	5 of 5	100.0% (47.8%, 100.0%)
STK11 Exon 1	c.179dupA	p.Y60*	1.45-1.56	0.08-0.11	0.10	0.10	0.01	13.40 %	5 of 5	100.0% (47.8%, 100.0%)
PHOX2B Exon 3	c.756_776delGGCGGCAGCGGCAGCGGCGGC	p.A254_A260del	1.00-1.30	0.03-0.08	0.06	0.07	0.02	31.70 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ARID5B Exon 10	c.3131A>T	p.E1044V	1.60-2.44	0.03-0.04	0.04	0.04	0.00	12.45 %	5 of 5	100.0% (47.8%, 100.0%)
PARP1 Exon 4	c.433G>C	p.D145H	0.96-1.15	0.10-0.13	0.12	0.12	0.01	8.82%	5 of 5	100.0% (47.8%, 100.0%)
PTEN Exon 5	**c.440dupA**	**p.A148Gfs*32**	**0.17-0.52**	**0.51-0.59**	**0.55**	**0.55**	**0.03**	**4.64%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
NRAS Exon 2	c.35G>A	p.G12D	0.23-0.31	0.49-0.57	0.54	0.55	0.03	5.64%	5 of 5	100.0% (47.8%, 100.0%)
BMPR1A Exon 6	c.423dupT	p.V142Cfs*7	0.16-0.43	0.39-0.63	0.53	0.55	0.08	14.46 %	5 of 5	100.0% (47.8%, 100.0%)
MAP2K1 Exon 3	c.383G>A	p.G128D	0.69-0.82	0.11-0.15	0.13	0.12	0.02	14.02 %	5 of 5	100.0% (47.8%, 100.0%)
EPHA5 Exon 14	c.2402T>G	p.L801R	0.29-0.36	0.27-0.33	0.30	0.31	0.02	7.17%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.3640A>C	p.T1214P	1.07-1.68	0.12-0.17	0.15	0.15	0.02	11.84 %	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 2	**c.312_317delAGTAGG**	**p.V105_G106del**	**0.64-0.76**	**0.51-0.54**	**0.52**	**0.52**	**0.01**	**2.08%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
DCUN1D1 Exon 7	c.748C>T	p.Q250*	0.75-0.90	0.17-0.20	0.18	0.18	0.01	6.19%	5 of 5	100.0% (47.8%, 100.0%)
GATA3 Exon 6	c.1087C>T	p.Q363*	0.96-1.12	0.39-0.43	0.40	0.40	0.01	3.26%	5 of 5	100.0% (47.8%, 100.0%)
PRKD1 Exon 16[a]	c.2429A>C	p.H810P	0.58-0.58	0.08-0.08	0.08	0.08	0.00	0.00%	1 of 5	20.0% (0.5%, 71.6%)
NCOR1 Exon 37	c.5765G>C	p.R1922T	1.13-1.27	0.34-0.40	0.38	0.39	0.02	5.62%	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 6	c.584T>C	p.I195T	0.53-0.59	0.33-0.42	0.38	0.39	0.03	8.22%	5 of 5	100.0% (47.8%, 100.0%)
KEAP1 Exon 2	c.233A>G	p.D78G	1.31-1.65	0.21-0.23	0.22	0.22	0.01	4.03%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
RB1 Exon 19	c.1901C>A	p.S634*	0.08-0.75	0.29-0.41	0.35	0.34	0.04	10.99 %	5 of 5	100.0% (47.8%, 100.0%)
NSD1 Exon 5	c.3768G>C	p.L1256F	0.92-1.05	0.25-0.29	0.27	0.28	0.01	5.12%	5 of 5	100.0% (47.8%, 100.0%)
BAP1 Exon 5	c.295G>A	p.V99M	0.47-0.98	0.37-0.43	0.40	0.39	0.02	4.97%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CG Exon 4	c.2182A>T	p.M728L	0.69-0.76	0.17-0.21	0.19	0.19	0.01	7.55%	5 of 5	100.0% (47.8%, 100.0%)
TBX3 Exon 7	c.1339C>G	p.R447G	1.74-2.14	0.26-0.29	0.27	0.27	0.01	3.79%	5 of 5	100.0% (47.8%, 100.0%)
PPM1D Exon 1	c.254C>G	p.S85W	1.63-2.80	0.21-0.24	0.22	0.22	0.01	3.27%	5 of 5	100.0% (47.8%, 100.0%)
KDM5A Exon 7	c.820G>A	p.A274T	0.26-0.33	0.27-0.37	0.31	0.30	0.04	11.54 %	5 of 5	100.0% (47.8%, 100.0%)
ARID1A Exon 20	c.5548delG	p.D1850Tfs*33	2.04-2.33	0.28-0.30	0.30	0.30	0.01	2.87%	5 of 5	100.0% (47.8%, 100.0%)
PALB2 Exon 12	c.3260G>T	p.S1087I	0.23-0.58	0.13-0.26	0.20	0.20	0.04	21.07 %	5 of 5	100.0% (47.8%, 100.0%)
BCL6 Exon 3	c.76C>T	p.R26W	0.79-1.65	0.27-0.31	0.29	0.29	0.01	4.60%	5 of 5	100.0% (47.8%, 100.0%)
MGA Exon 17	c.5907_5911delinsAAGAT	p.E1971*	0.63-0.73	0.32-0.99	0.73	0.99	0.32	43.07 %	5 of 5	100.0% (47.8%, 100.0%)
ARID1A Exon 1	c.780_782delCTC	p.S265del	2.06-2.65	0.30-0.35	0.32	0.31	0.02	5.09%	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 8	c.817C>T	p.R273C	1.38-1.53	0.26-0.29	0.27	0.27	0.01	4.05%	5 of 5	100.0% (47.8%, 100.0%)
RNF43 Exon 9	c.1976delG	p.G659Vfs*41	1.52-1.81	0.62-0.63	0.63	0.62	0.01	0.98%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 37	c.7964G>A	p.C2655Y	1.42-1.67	0.28-0.32	0.30	0.30	0.01	3.63%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ZFHX3 Exon 9	c.4805A>T	p.N1602I	1.75-1.97	0.34-0.38	0.37	0.38	0.02	4.51%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K1 Exon 14	c.3651_3653delCAT	p.I1219del	0.58-0.66	0.30-0.37	0.34	0.35	0.02	6.90%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K14 Exon 12	c.2101G>T	p.G701C	1.55-2.00	0.30-0.32	0.32	0.32	0.01	2.24%	5 of 5	100.0% (47.8%, 100.0%)
SOX17 Exon 1	c.167C>A	p.A56D	1.61-2.05	0.33-0.36	0.34	0.34	0.02	4.50%	5 of 5	100.0% (47.8%, 100.0%)
MED12 Exon 42[a]	c.6226C>T	p.Q2076*	0.99-1.02	0.01-0.02	0.02	0.02	0.00	26.55 %	2 of 5	40.0% (5.3%, 85.3%)
PAK1 Exon 7	c.607C>T	p.R203W	0.47-0.60	0.23-0.29	0.26	0.26	0.02	8.07%	5 of 5	100.0% (47.8%, 100.0%)
BRAF Exon 15	**c.1799T>A**	**p.V600E**	**0.17-0.56**	**0.28-0.37**	**0.32**	**0.32**	**0.04**	**11.14 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
PTPRD Exon 44	c.5452G>T	p.E1818*	1.34-1.47	0.29-0.34	0.32	0.33	0.02	4.98%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 15	c.2186C>T	p.A729V	0.55-1.19	0.05-0.10	0.07	0.07	0.02	28.08 %	5 of 5	100.0% (47.8%, 100.0%)
NOTCH1 Exon 34	c.7455delC	p.S2486Rfs*103	1.58-2.01	0.31-0.35	0.33	0.33	0.02	4.63%	5 of 5	100.0% (47.8%, 100.0%)
FBXW7 Exon 10	c.1429G>A	p.G477S	0.52-0.59	0.33-0.38	0.36	0.36	0.02	5.43%	5 of 5	100.0% (47.8%, 100.0%)
BRD4 Exon 10	c.1988C>G	p.T663R	1.41-1.75	0.04-0.06	0.05	0.05	0.01	10.52 %	5 of 5	100.0% (47.8%, 100.0%)
NBN Exon 13[a]	c.1932G>T	p.Q644H	0.42-0.57	0.02-0.04	0.03	0.03	0.01	21.22 %	4 of 5	80.0% (28.4%, 99.5%)
NCOR1 Exon 15	c.1584delA	p.E529Kfs*26	0.52-0.70	0.27-0.32	0.29	0.28	0.02	7.77%	5 of 5	100.0% (47.8%, 100.0%)
PAK7 Exon 10	c.1945delC	p.L649Sfs*15	0.54-0.60	0.27-0.32	0.31	0.32	0.02	6.03%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
KMT2B Exon 27	c.5636delG	p.G1879Vfs*16	1.15-1.48	0.31-0.35	0.33	0.34	0.01	3.94%	5 of 5	100.0% (47.8%, 100.0%)
RBM10 Exon 10	c.994C>A	p.R332S	0.29-0.79	0.62-0.68	0.65	0.66	0.02	2.65%	5 of 5	100.0% (47.8%, 100.0%)
BRCA1 Exon 21[a]	c.5347A>T	p.M1783L	0.19-0.42	0.04-0.10	0.06	0.05	0.02	40.44 %	4 of 5	80.0% (28.4%, 99.5%)
ARID5B Exon 10	c.2263G>A	p.V755I	1.23-1.88	0.30-0.37	0.35	0.37	0.03	7.63%	5 of 5	100.0% (47.8%, 100.0%)
NF1 Exon 16	c.1738delT	p.Y580Tfs*6	0.47-0.60	0.30-0.41	0.36	0.38	0.04	11.03 %	5 of 5	100.0% (47.8%, 100.0%)
NKX2-1 Exon 3	c.1015G>A	p.G339S	1.32-1.61	0.24-0.35	0.30	0.32	0.04	13.52 %	5 of 5	100.0% (47.8%, 100.0%)
GRIN2A Exon 3	c.244C>A	p.L82I	0.98-1.33	0.35-0.38	0.36	0.37	0.01	2.47%	5 of 5	100.0% (47.8%, 100.0%)
MDM2 Exon 11	c.1111delA	p.T371Lfs*2	0.60-0.70	0.32-0.34	0.33	0.33	0.01	2.78%	5 of 5	100.0% (47.8%, 100.0%)
RRAGC Exon 2	c.328G>A	p.V110M	0.41-0.43	0.27-0.31	0.28	0.28	0.01	4.55%	5 of 5	100.0% (47.8%, 100.0%)
ATM Exon 58	c.8558C>T	p.T2853M	0.26-0.71	0.28-0.34	0.30	0.30	0.02	6.70%	5 of 5	100.0% (47.8%, 100.0%)
PHOX2B Exon 3	c.451G>A	p.A151T	0.68-0.93	0.02-0.05	0.03	0.03	0.01	29.43 %	5 of 5	100.0% (47.8%, 100.0%)
BCL10 Exon 3	c.686C>G	p.T229S	0.29-0.63	0.29-0.38	0.34	0.34	0.03	9.63%	5 of 5	100.0% (47.8%, 100.0%)
ARID2 Exon 2	c.109delA	p.I37Sfs*21	0.70-1.33	0.29-0.38	0.34	0.33	0.03	9.91%	5 of 5	100.0% (47.8%, 100.0%)
KIT Exon 3	c.590C>T	p.S197L	0.98-1.09	0.17-0.20	0.19	0.19	0.01	5.77%	5 of 5	100.0% (47.8%, 100.0%)
FLT1 Exon 9	c.1107-1G>A	None	0.37-0.49	0.24-0.36	0.30	0.32	0.04	13.80 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
CCND3 Exon 2	c.373_374delTG	p.C125Hfs*34	0.99-1.23	0.25-0.32	0.29	0.29	0.02	8.24%	5 of 5	100.0% (47.8%, 100.0%)
MSH6 Exon 5	c.3425C>T	p.T1142M	0.65-0.73	0.31-0.36	0.35	0.35	0.02	5.01%	5 of 5	100.0% (47.8%, 100.0%)
STAT3 Exon 7	c.641G>T	p.R214L	1.10-1.32	0.30-0.34	0.31	0.30	0.01	4.82%	5 of 5	100.0% (47.8%, 100.0%)
BMPR1A Exon 4	c.176T>A	p.L59*	0.31-0.67	0.22-0.28	0.25	0.27	0.03	10.26 %	5 of 5	100.0% (47.8%, 100.0%)
RNF43 Exon 9	c.1586G>T	p.R529L	1.52-1.81	0.05-0.07	0.06	0.05	0.01	13.32 %	5 of 5	100.0% (47.8%, 100.0%)
ROS1 Exon 39	c.6175C>A	p.L2059M	0.23-0.25	0.27-0.31	0.30	0.30	0.02	5.69%	5 of 5	100.0% (47.8%, 100.0%)
ERCC3 Exon 6	c.777A>T	p.E259D	0.61-0.65	0.06-0.10	0.08	0.08	0.01	15.20 %	5 of 5	100.0% (47.8%, 100.0%)
STAT3 Exon 3	c.272A>G	p.Q91R	0.80-0.93	0.03-0.08	0.06	0.06	0.01	26.29 %	5 of 5	100.0% (47.8%, 100.0%)
KDR Exon 15	c.2194G>A	p.E732K	0.32-0.43	0.31-0.38	0.33	0.33	0.03	7.66%	5 of 5	100.0% (47.8%, 100.0%)
TBX3 Exon 7	c.1292C>T	p.A431V	2.06-2.40	0.08-0.09	0.08	0.08	0.01	7.40%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 2	c.11G>A	p.R4Q	0.42-0.49	0.22-0.26	0.23	0.23	0.02	7.09%	5 of 5	100.0% (47.8%, 100.0%)
MSI1 Exon 9	c.587C>A	p.A196D	1.04-1.26	0.28-0.32	0.30	0.30	0.01	4.97%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K14 Exon 7	c.1294G>A	p.G432S	1.28-1.61	0.04-0.06	0.05	0.05	0.01	13.55 %	5 of 5	100.0% (47.8%, 100.0%)
WT1 Exon N/A	c.20C>T	p.T7M	0	0.37-0.43	0.39	0.39	0.02	5.81%	5 of 5	100.0% (47.8%, 100.0%)
TP53BP1 Exon 12	c.2672C>T	p.A891V	0.46-0.55	0.02-0.03	0.02	0.02	0.00	20.89 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
KMT2C Exon 38[a]	c.7443-2_7443-1insAA	None	0.89-0.90	0.06-0.06	0.06	0.06	0.00	3.99%	2 of 5	40.0% (5.3%, 85.3%)
IGF1R Exon 3	c.668C>T	p.A223V	1.48-1.76	0.25-0.30	0.28	0.28	0.02	6.83%	5 of 5	100.0% (47.8%, 100.0%)
EPCAM Exon 5	c.545C>T	p.T182M	0.26-0.34	0.30-0.34	0.32	0.32	0.01	4.46%	5 of 5	100.0% (47.8%, 100.0%)
MGA Exon 3	c.1626delA	p.K542Nfs*5	0.54-0.59	0.30-0.41	0.37	0.39	0.04	10.41 %	5 of 5	100.0% (47.8%, 100.0%)
SMARCA4 Exon 35	c.4933C>T	p.R1645W	1.17-1.43	0.35-0.39	0.36	0.35	0.02	4.40%	5 of 5	100.0% (47.8%, 100.0%)
SMARCA4 Exon 26	c.3727C>T	p.R1243W	1.00-1.15	0.31-0.37	0.34	0.35	0.02	6.01%	5 of 5	100.0% (47.8%, 100.0%)
AXIN2 Exon 8	c.1994_1995insG	p.N666Qfs*41	1.18-2.32	0.30-0.33	0.31	0.31	0.01	2.92%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K13 Exon 5	c.806A>G	p.Y269C	0.39-0.46	0.33-0.41	0.37	0.35	0.03	8.01%	5 of 5	100.0% (47.8%, 100.0%)
SMAD4 Exon 12	c.1607T>C	p.L536P	0.41-0.45	0.51-0.59	0.54	0.54	0.04	6.56%	5 of 5	100.0% (47.8%, 100.0%)
IRS1 Exon 1	c.2059_2060insGCA	p.S686_S680ins	1.39-1.65	0.24-0.27	0.25	0.25	0.01	4.89%	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 1	c.81G>T	p.E27D	0.91-2.12	0.30-0.34	0.32	0.33	0.02	4.85%	5 of 5	100.0% (47.8%, 100.0%)
JAK3 Exon 16	c.2172G>A	p.M724I	1.37-1.63	0.29-0.34	0.32	0.33	0.02	5.94%	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 6	c.637C>T	p.R213*	0.88-0.97	0.36-0.42	0.39	0.38	0.03	6.49%	5 of 5	100.0% (47.8%, 100.0%)
KMT2C Exon 43	c.11248_11249insTAAGGTAGAAGGAAACGCTGTAG	p.A3750Vfs*3	1.09-1.17	0.11-0.16	0.13	0.12	0.02	11.97 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ARID1B Exon 1	c.1379C>G	p.A460G	1.19-2.39	0.01-0.02	0.02	0.02	0.00	19.28 %	5 of 5	100.0% (47.8%, 100.0%)
MUTYH Exon 16	c.1613C>G	p.S538C	1.23-1.51	0.35-0.43	0.38	0.36	0.03	7.10%	5 of 5	100.0% (47.8%, 100.0%)
BCOR Exon 4	**c.2936_2937insATTCAAATG**	**p.K978_F977ins***	**1.47-1.68**	**0.28-0.34**	**0.31**	**0.32**	**0.02**	**6.16%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
RHEB Exon 2	c.104A>T	p.Y35F	0.50-0.60	0.04-0.05	0.04	0.04	0.00	9.99%	5 of 5	100.0% (47.8%, 100.0%)
ASXL2 Exon 1	c.38G>A	p.W13*	1.21-1.53	0.76-0.80	0.77	0.77	0.02	2.05%	5 of 5	100.0% (47.8%, 100.0%)
EZH2 Exon 8	c.893G>A	p.R298H	0.34-0.38	0.31-0.36	0.34	0.33	0.02	4.92%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CB Exon 18	c.2638G>A	p.A880T	0.25-0.32	0.25-0.29	0.26	0.26	0.01	4.73%	5 of 5	100.0% (47.8%, 100.0%)
RAD51 Exon 9	c.868A>G	p.N290D	0.24-0.89	0.26-0.42	0.32	0.32	0.05	15.51 %	5 of 5	100.0% (47.8%, 100.0%)
FLT4 Exon 14	c.2131T>C	p.W711R	1.12-1.31	0.27-0.35	0.32	0.33	0.03	8.97%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH2 Exon 24	c.3933G>A	p.M1311I	0.76-0.97	0.26-0.30	0.28	0.28	0.02	5.44%	5 of 5	100.0% (47.8%, 100.0%)
NSD1 Exon 23	c.7975G>A	p.A2659T	1.59-1.77	0.31-0.34	0.33	0.32	0.01	3.81%	5 of 5	100.0% (47.8%, 100.0%)
NCOR1 Exon 19	c.2063G>A	p.R688Q	0.29-0.32	0.31-0.38	0.34	0.34	0.03	7.88%	5 of 5	100.0% (47.8%, 100.0%)
MGA Exon 11	c.3736C>T	p.R1246*	0.73-0.84	0.29-0.35	0.31	0.30	0.02	7.83%	5 of 5	100.0% (47.8%, 100.0%)
PBRM1 Exon 4	c.340G>A	p.A114T	0.29-0.39	0.27-0.35	0.30	0.29	0.03	9.07%	5 of 5	100.0% (47.8%, 100.0%)
FUBP1 Exon 11	c.857C>A	p.A286D	0.13-0.21	0.20-0.37	0.30	0.33	0.06	18.92 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ARID2 Exon 15	c.4640C>T	p.A1547V	0.81-1.61	0.30-0.36	0.33	0.34	0.02	6.99%	5 of 5	100.0% (47.8%, 100.0%)
CSDE1 Exon 13[a]	c.1333A>G	p.M445V	0.54-0.54	0.02-0.02	0.02	0.02	0.00	0.00%	1 of 5	20.0% (0.5%, 71.6%)
RNF43 Exon 5	c.520G>A	p.E174K	1.18-1.40	0.31-0.34	0.33	0.34	0.01	3.22%	5 of 5	100.0% (47.8%, 100.0%)
POLD1 Exon 7	c.795G>A	p.W265*	0.97-1.16	0.02-0.03	0.02	0.02	0.00	19.27 %	5 of 5	100.0% (47.8%, 100.0%)
FOXP1 Exon 21	c.1945C>A	p.L649M	0.80-0.90	0.31-0.36	0.33	0.33	0.02	4.98%	5 of 5	100.0% (47.8%, 100.0%)
SMO Exon 12	c.2014A>G	p.K672E	1.02-1.24	0.31-0.34	0.33	0.33	0.01	2.99%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 5	c.239G>A	p.S80N	0.56-0.66	0.32-0.37	0.34	0.34	0.02	5.19%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 37	c.5695C>T	p.R1899W	0.88-1.41	0.02-0.03	0.02	0.02	0.01	22.12 %	5 of 5	100.0% (47.8%, 100.0%)
ALOX12B Exon 12	c.1534T>C	p.Y512H	1.30-1.55	0.32-0.36	0.34	0.33	0.02	4.70%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 24	c.5105A>G	p.Q1702R	1.33-1.48	0.31-0.35	0.33	0.33	0.01	4.39%	5 of 5	100.0% (47.8%, 100.0%)
SOS1 Exon 10	c.1575A>G	p.I525M	0.21-0.27	0.29-0.37	0.33	0.32	0.03	8.75%	5 of 5	100.0% (47.8%, 100.0%)
RECQL Exon 15	c.1679G>A	p.S560N	0.16-0.22	0.24-0.34	0.29	0.30	0.03	11.62 %	5 of 5	100.0% (47.8%, 100.0%)
GLI1 Exon 4	c.299G>A	p.R100H	0.60-0.69	0.29-0.33	0.31	0.31	0.02	5.50%	5 of 5	100.0% (47.8%, 100.0%)
TET2 Exon 3	c.2548C>G	p.H850D	0.77-0.89	0.29-0.35	0.32	0.32	0.02	6.65%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 11	c.3589C>T	p.P1197S	0.87-0.98	0.30-0.34	0.32	0.33	0.01	3.78%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
DDR2 Exon 17	c.2141G>A	p.R714Q	0.81-0.95	0.29-0.34	0.31	0.31	0.02	5.46%	5 of 5	100.0% (47.8%, 100.0%)
SOS1 Exon 23	c.3716G>A	p.G1239D	1.36-1.48	0.29-0.33	0.31	0.31	0.01	4.74%	5 of 5	100.0% (47.8%, 100.0%)
ATM Exon 39	c.5784delT	p.F1928Lfs*9	0.21-0.65	0.29-0.34	0.31	0.31	0.02	6.06%	5 of 5	100.0% (47.8%, 100.0%)
GLI1 Exon 12	c.2030A>G	p.D677G	0.93-1.09	0.31-0.36	0.33	0.33	0.02	5.14%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K1 Exon 13	c.2253G>A	p.W751*	0.33-0.38	0.29-0.37	0.33	0.32	0.03	8.23%	5 of 5	100.0% (47.8%, 100.0%)
CASP8 Exon 3	c.52G>T	p.D18Y	0.71-0.83	0.27-0.35	0.31	0.31	0.03	8.45%	5 of 5	100.0% (47.8%, 100.0%)
SPEN Exon 11	c.9808C>T	p.L3270F	1.44-1.57	0.31-0.34	0.33	0.34	0.01	3.89%	5 of 5	100.0% (47.8%, 100.0%)
REL Exon 3	c.254G>A	p.R85K	0.41-0.50	0.31-0.34	0.33	0.33	0.01	3.41%	5 of 5	100.0% (47.8%, 100.0%)
GATA3 Exon 2	c.178G>A	p.V60I	1.58-1.99	0.31-0.34	0.32	0.32	0.01	3.50%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH2 Exon 5	c.874+1G>A	None	0.25-0.35	0.28-0.40	0.34	0.34	0.04	11.54 %	5 of 5	100.0% (47.8%, 100.0%)
CDK8 Exon 8	c.818T>C	p.M273T	0.16-0.23	0.24-0.37	0.30	0.29	0.05	15.36 %	5 of 5	100.0% (47.8%, 100.0%)
HRAS Exon 4	c.295C>T	p.Q99*	1.10-1.44	0.28-0.33	0.30	0.30	0.02	5.80%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 22	c.4103G>A	p.R1368H	0.66-0.77	0.28-0.35	0.32	0.33	0.02	7.18%	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 25	c.3754delA	p.T1252Qfs*4	0.63-0.72	0.30-0.34	0.32	0.33	0.01	4.64%	5 of 5	100.0% (47.8%, 100.0%)
PREX2 Exon 23	c.2626G>T	p.V876L	0.36-0.50	0.28-0.36	0.32	0.31	0.03	9.82%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
JAK1 Exon 14	c.1978G>A	p.D660N	0.63-0.75	0.30-0.35	0.33	0.33	0.02	6.62%	5 of 5	100.0% (47.8%, 100.0%)
KMT2A Exon 3[a]	c.1102G>T	p.D368Y	0.80-0.90	0.30-0.32	0.31	0.31	0.01	3.28%	3 of 5	60.0% (14.7%, 94.7%)
ATM Exon 25	c.3637C>A	p.H1213N	0.25-0.65	0.23-0.34	0.28	0.28	0.04	13.44 %	5 of 5	100.0% (47.8%, 100.0%)
RICTOR Exon 37	c.4898G>A	p.R1633H	0.32-0.38	0.29-0.42	0.32	0.30	0.05	15.83 %	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 1	c.862G>A	p.E288K	1.24-2.63	0.28-0.33	0.31	0.32	0.02	5.12%	5 of 5	100.0% (47.8%, 100.0%)
BRCA1 Exon 12	c.4238A>G	p.E1413G	0.21-0.48	0.29-0.36	0.31	0.31	0.02	7.49%	5 of 5	100.0% (47.8%, 100.0%)
AXIN1 Exon 6	c.1321G>A	p.A441T	1.17-2.07	0.31-0.35	0.34	0.34	0.01	3.85%	5 of 5	100.0% (47.8%, 100.0%)
ATM Exon 25	c.3602T>C	p.F1201S	0.25-0.65	0.32-0.38	0.35	0.35	0.02	5.57%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH1 Exon 34	c.7034G>T	p.G2345V	1.92-2.22	0.33-0.36	0.34	0.35	0.01	2.77%	5 of 5	100.0% (47.8%, 100.0%)
STAG2 Exon 29	c.2925-3C>T	None	0.30-0.36	0.27-0.33	0.31	0.31	0.02	7.35%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.7468G>A	p.V2490M	1.23-1.92	0.31-0.36	0.33	0.33	0.02	5.38%	5 of 5	100.0% (47.8%, 100.0%)
IFNGR1 Exon 7	c.1100C>T	p.P367L	0.19-0.23	0.26-0.38	0.32	0.33	0.04	13.26 %	5 of 5	100.0% (47.8%, 100.0%)
ATRX Exon 9	c.2999A>C	p.K1000T	0.13-0.32	0.31-0.35	0.32	0.32	0.02	4.69%	5 of 5	100.0% (47.8%, 100.0%)
RUNX1 Exon 9	c.1163C>T	p.S388L	1.70-2.02	0.34-0.35	0.35	0.35	0.01	1.48%	5 of 5	100.0% (47.8%, 100.0%)
FLT4 Exon 30	c.3982G>T	p.G1328*	0.93-1.17	0.31-0.39	0.34	0.34	0.03	8.39%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
SF3B1 Exon 7	c.809G>A	p.R270Q	0.74-0.86	0.29-0.32	0.30	0.31	0.01	4.22%	5 of 5	100.0% (47.8%, 100.0%)
FLT1 Exon 11	c.1523G>A	p.R508H	0.21-0.24	0.28-0.38	0.33	0.31	0.04	11.31 %	5 of 5	100.0% (47.8%, 100.0%)
PPM1D Exon 6	c.1573G>T	p.E525*	0.21-0.61	0.30-0.36	0.32	0.31	0.02	7.49%	5 of 5	100.0% (47.8%, 100.0%)
INSR Exon 2	c.211G>A	p.E71K	0.79-0.92	0.28-0.36	0.32	0.32	0.03	8.69%	5 of 5	100.0% (47.8%, 100.0%)
ATM Exon 10	c.1339C>T	p.R447*	0.40-0.84	0.28-0.34	0.31	0.31	0.02	5.68%	5 of 5	100.0% (47.8%, 100.0%)
PTEN Exon 7	c.146C>T	p.P49L	0.23-0.57	0.18-0.32	0.27	0.28	0.05	17.50 %	5 of 5	100.0% (47.8%, 100.0%)
BCOR Exon 8	c.3625C>T	p.R1209C	1.22-1.38	0.32-0.33	0.33	0.33	0.01	1.81%	5 of 5	100.0% (47.8%, 100.0%)
TRAF2 Exon 2	c.88G>A	p.A30T	1.46-1.66	0.30-0.35	0.32	0.31	0.02	6.41%	5 of 5	100.0% (47.8%, 100.0%)
SLX4 Exon 3	c.751G>A	p.A251T	1.12-1.27	0.02-0.04	0.04	0.04	0.01	18.61 %	5 of 5	100.0% (47.8%, 100.0%)
IRS1 Exon 1	c.2685A>C	p.E895D	1.69-1.96	0.35-0.39	0.37	0.37	0.01	3.66%	5 of 5	100.0% (47.8%, 100.0%)
PTCH1 Exon 15	c.2427A>C	p.K809N	0.52-1.34	0.29-0.38	0.33	0.33	0.03	9.75%	5 of 5	100.0% (47.8%, 100.0%)
SETD2 Exon 12	c.5721G>T	p.E1907D	0.64-0.71	0.30-0.36	0.32	0.32	0.02	5.66%	5 of 5	100.0% (47.8%, 100.0%)
TSHR Exon 10	c.2011T>A	p.S671T	1.59-1.76	0.33-0.37	0.35	0.34	0.01	4.21%	5 of 5	100.0% (47.8%, 100.0%)
CTCF Exon 3	**c.263C>A**	**p.A88D**	**0.75-0.85**	**0.29-0.34**	**0.31**	**0.31**	**0.02**	**5.20%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
SYK Exon 2	c.202C>T	p.R68W	1.39-1.69	0.33-0.35	0.34	0.34	0.00	1.45%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
TNFAIP3 Exon 7	c.1477G>A	p.G493R	1.94-2.02	0.31-0.34	0.32	0.32	0.01	3.54%	5 of 5	100.0% (47.8%, 100.0%)
GRIN2A Exon 3[a]	c.97_99delinsACA	p.A33T	1.00-1.21	0.28-0.31	0.29	0.30	0.01	3.63%	4 of 5	80.0% (28.4%, 99.5%)
NF1 Exon 33	c.4336C>A	p.L1446I	0.20-0.23	0.22-0.36	0.29	0.31	0.05	16.96 %	5 of 5	100.0% (47.8%, 100.0%)
FLT4 Exon 25[a]	c.3391G>A	p.G1131S	0.64-0.79	0.02-0.02	0.02	0.02	0.00	11.82 %	4 of 5	80.0% (28.4%, 99.5%)
ERBB4 Exon 28	c.3581G>A	p.G1194D	1.15-1.28	0.29-0.35	0.32	0.33	0.02	6.67%	5 of 5	100.0% (47.8%, 100.0%)
TET1 Exon 12	c.6205G>A	p.E2069K	1.70-1.85	0.33-0.36	0.34	0.34	0.01	3.23%	5 of 5	100.0% (47.8%, 100.0%)
DNMT1 Exon 41	c.4879G>T	p.E1627*	0.59-0.73	0.30-0.35	0.32	0.32	0.02	5.70%	5 of 5	100.0% (47.8%, 100.0%)
TCF3 Exon 2	c.42G>T	p.K14N	0.81-1.15	0.28-0.38	0.33	0.32	0.03	10.22 %	5 of 5	100.0% (47.8%, 100.0%)
FLT1 Exon 19[a]	c.2669A>G	p.N890S	0.70-0.85	0.02-0.03	0.02	0.02	0.00	21.87 %	4 of 5	80.0% (28.4%, 99.5%)
INPPL1 Exon 21	c.2339G>A	p.S780N	0.59-0.69	0.27-0.34	0.30	0.30	0.03	9.21%	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 13	c.2153T>C	p.L718P	1.21-1.37	0.31-0.36	0.33	0.33	0.02	5.02%	5 of 5	100.0% (47.8%, 100.0%)
ERG Exon 6	c.413C>A	p.P138H	0.95-1.12	0.29-0.36	0.32	0.33	0.02	7.05%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K13 Exon 12	c.2147G>A	p.G716D	1.31-1.62	0.32-0.35	0.34	0.34	0.01	3.64%	5 of 5	100.0% (47.8%, 100.0%)
PGR Exon 1	c.538G>A	p.A180T	2.08-2.56	0.50-0.54	0.52	0.52	0.01	2.43%	5 of 5	100.0% (47.8%, 100.0%)
JAK3 Exon 9	c.1208G>A	p.R403H	0.44-0.60	0.27-0.33	0.31	0.31	0.02	7.41%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
FOXO1 Exon 2	c.1000G>A	p.E334K	1.22-1.34	0.33-0.34	0.33	0.33	0.00	1.32%	5 of 5	100.0% (47.8%, 100.0%)
CREBBP Exon 2	c.458C>T	p.P153L	1.36-1.58	0.31-0.34	0.32	0.32	0.01	2.65%	5 of 5	100.0% (47.8%, 100.0%)
IGF2 Exon 3	c.223G>A	p.A75T	0.78-1.06	0.31-0.35	0.33	0.33	0.01	3.95%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH2 Exon 33	c.5978C>A	p.T1993N	0.35-0.42	0.27-0.34	0.29	0.28	0.03	9.88%	5 of 5	100.0% (47.8%, 100.0%)
CDC73 Exon 14	c.1159G>A	p.V387I	0.50-0.72	0.29-0.37	0.32	0.32	0.03	9.88%	5 of 5	100.0% (47.8%, 100.0%)
AXIN2 Exon 4	c.1016G>A	p.R339H	0.42-0.91	0.21-0.26	0.24	0.26	0.02	8.82%	5 of 5	100.0% (47.8%, 100.0%)
GRIN2A Exon 14	c.4357G>A	p.V1453M	0.94-1.08	0.27-0.33	0.31	0.32	0.02	6.25%	5 of 5	100.0% (47.8%, 100.0%)
EPCAM Exon 4	c.458G>T	p.R153I	0.30-0.37	0.33-0.38	0.36	0.36	0.02	6.49%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 6	c.437C>T	p.A146V	0.21-0.26	0.26-0.31	0.29	0.30	0.02	5.16%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 17	c.7286C>A	p.S2429Y	0.52-0.63	0.24-0.35	0.30	0.29	0.04	12.66%	5 of 5	100.0% (47.8%, 100.0%)
EPHB1 Exon 15	c.2725G>T	p.D909Y	1.17-1.44	0.31-0.34	0.33	0.33	0.01	3.22%	5 of 5	100.0% (47.8%, 100.0%)
KEAP1 Exon 2	c.626T>C	p.M209T	1.09-1.32	0.31-0.35	0.33	0.33	0.02	5.14%	5 of 5	100.0% (47.8%, 100.0%)
PTPRT Exon 4	c.535G>A	p.V179M	0.47-0.85	0.29-0.36	0.33	0.34	0.02	7.18%	5 of 5	100.0% (47.8%, 100.0%)
ARID1A Exon 11	c.3177G>T	p.K1059N	1.27-1.49	0.02-0.03	0.03	0.02	0.00	15.50%	5 of 5	100.0% (47.8%, 100.0%)
TSC2 Exon 17	c.1832G>A	p.R611Q	1.52-1.79	0.35-0.36	0.35	0.35	0.01	1.52%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
BCL10 Exon 3	c.595G>T	p.G199W	0.29-0.70	0.28-0.34	0.31	0.31	0.02	6.36%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 2	c.58G>A	p.A20T	0.34-0.38	0.25-0.35	0.29	0.30	0.04	12.07 %	5 of 5	100.0% (47.8%, 100.0%)
EP300 Exon 31	c.5458C>T	p.H1820Y	2.03-2.27	0.32-0.35	0.33	0.33	0.01	2.73%	5 of 5	100.0% (47.8%, 100.0%)
HGF Exon 17	c.1900C>A	p.L634I	0.20-0.25	0.27-0.36	0.31	0.32	0.03	10.22 %	5 of 5	100.0% (47.8%, 100.0%)
FAT1 Exon 27	c.13382C>T	p.S4461F	1.49-1.71	0.30-0.31	0.30	0.31	0.00	1.07%	5 of 5	100.0% (47.8%, 100.0%)
RAF1 Exon 2[a]	c.125C>T	p.A42V	0.52-0.71	0.02-0.03	0.03	0.03	0.01	19.09 %	3 of 5	60.0% (14.7%, 94.7%)
CIC Exon 3	c.406G>T	p.G136*	1.64-1.96	0.34-0.35	0.35	0.35	0.01	1.84%	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 10[a]	c.1436_1438delinsCA	p.D479Afs*35	1.20-1.20	0.92-0.92	0.92	0.92	0.00	0.00%	1 of 5	20.0% (0.5%, 71.6%)
CTNNB1 Exon 12	c.1856G>C	p.C619S	0.32-0.39	0.28-0.34	0.31	0.31	0.02	7.91%	5 of 5	100.0% (47.8%, 100.0%)
CHEK2 Exon 2	c.153G>T	p.Q51H	1.30-1.50	0.23-0.26	0.24	0.24	0.01	3.78%	5 of 5	100.0% (47.8%, 100.0%)
PTCH1 Exon 23	c.4115C>T	p.T1372M	1.36-2.56	0.32-0.35	0.33	0.33	0.01	3.08%	5 of 5	100.0% (47.8%, 100.0%)
SYK Exon 4	c.650G>A	p.R217H	1.63-1.81	0.32-0.35	0.33	0.33	0.01	3.24%	5 of 5	100.0% (47.8%, 100.0%)
SETD2 Exon 3	c.2749_2750insA	p.S917Kfs*18	0.49-0.60	0.24-0.32	0.29	0.29	0.03	9.77%	5 of 5	100.0% (47.8%, 100.0%)
ERCC4 Exon 8	c.1468C>T	p.R490W	0.43-0.49	0.27-0.37	0.30	0.30	0.03	11.19 %	5 of 5	100.0% (47.8%, 100.0%)
JUN Exon 1	c.686C>T	p.P229L	1.60-1.91	0.29-0.34	0.32	0.32	0.02	4.79%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
EPAS1 Exon 16	c.2512G>A	p.E838K	1.49-1.88	0.32-0.34	0.33	0.33	0.01	2.00%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 14	c.3890G>A	p.C1297Y	0.53-0.65	0.32-0.35	0.33	0.34	0.01	3.32%	5 of 5	100.0% (47.8%, 100.0%)
ERBB2 Exon 29	c.2927G>A	p.R976H	0.75-0.89	0.31-0.34	0.32	0.32	0.01	3.12%	5 of 5	100.0% (47.8%, 100.0%)
FGFR4 Exon 17	c.2201C>A	p.P734H	0.83-0.97	0.31-0.34	0.32	0.33	0.01	3.23%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 14	c.3896C>T	p.A1299V	0.53-0.65	0.29-0.34	0.31	0.30	0.02	5.51%	5 of 5	100.0% (47.8%, 100.0%)
NTRK1 Exon 7	c.844G>A	p.V282I	1.02-1.21	0.31-0.35	0.34	0.35	0.02	4.84%	5 of 5	100.0% (47.8%, 100.0%)
KMT2D Exon 6	c.804G>A	p.W268*	1.31-1.51	0.30-0.33	0.32	0.32	0.01	3.79%	5 of 5	100.0% (47.8%, 100.0%)
ERBB4 Exon 25	c.2999_3002delinsGCAA	p.D1000_S1001delinsGN	0.17-0.24	0.25-0.37	0.30	0.29	0.04	13.06 %	5 of 5	100.0% (47.8%, 100.0%)
CARD11 Exon 8	c.1094T>C	p.M365T	0.83-0.99	0.30-0.36	0.32	0.31	0.02	7.69%	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 20	c.5968C>T	p.R1990*	1.16-2.18	0.32-0.34	0.33	0.33	0.01	3.02%	5 of 5	100.0% (47.8%, 100.0%)
CIC Exon 10	c.1526delC	p.P509Hfs*14	1.62-1.93	0.31-0.33	0.32	0.33	0.01	2.26%	5 of 5	100.0% (47.8%, 100.0%)
DNAJB1 Exon 2	c.293C>A	p.P98H	1.74-1.91	0.30-0.33	0.31	0.32	0.01	4.65%	5 of 5	100.0% (47.8%, 100.0%)
HNF1A Exon 2	c.341G>A	p.R114H	0.89-1.11	0.29-0.33	0.31	0.31	0.02	5.28%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CB Exon 12	c.1771C>A	p.L591I	0.43-0.51	0.27-0.29	0.28	0.28	0.01	2.28%	5 of 5	100.0% (47.8%, 100.0%)
CDK12 Exon 4	c.2228A>C	p.K743T	0.35-0.50	0.28-0.36	0.32	0.31	0.03	9.04%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
MYD88 Exon 1	c.17C>T	p.A6V	1.96-2.42	0.32-0.34	0.33	0.33	0.01	2.37%	5 of 5	100.0% (47.8%, 100.0%)
KMT2D Exon 10	c.1315C>T	p.P439S	1.38-1.64	0.31-0.33	0.32	0.32	0.01	2.43%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.4610G>A	p.G1537D	1.23-1.92	0.32-0.34	0.33	0.33	0.01	2.28%	5 of 5	100.0% (47.8%, 100.0%)
ETV1 Exon 8	c.680C>T	p.A227V	1.14-1.27	0.30-0.36	0.32	0.33	0.02	7.00%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CG Exon 11	c.3197delA	p.K1066Sfs*42	0.24-0.27	0.25-0.35	0.30	0.29	0.03	11.69%	5 of 5	100.0% (47.8%, 100.0%)
CDC42 Exon 6	c.402G>T	p.Q134H	0.31-0.36	0.28-0.34	0.30	0.29	0.02	7.63%	5 of 5	100.0% (47.8%, 100.0%)
PPP2R1A Exon 4	c.375G>T	p.E125D	1.09-1.61	0.31-0.35	0.33	0.33	0.01	3.57%	5 of 5	100.0% (47.8%, 100.0%)
SH2B3 Exon 6	c.1160G>A	p.G387E	0.77-0.90	0.27-0.33	0.30	0.31	0.02	6.84%	5 of 5	100.0% (47.8%, 100.0%)
MPL Exon 5	c.785G>T	p.G262V	1.03-1.23	0.31-0.37	0.33	0.33	0.02	5.85%	5 of 5	100.0% (47.8%, 100.0%)
PAK1 Exon 14	c.1468C>A	p.L490M	0.20-0.22	0.27-0.36	0.31	0.31	0.03	9.87%	5 of 5	100.0% (47.8%, 100.0%)
BCL6 Exon 5	c.1129A>G	p.K377E	1.20-2.52	0.30-0.35	0.33	0.33	0.02	5.56%	5 of 5	100.0% (47.8%, 100.0%)
RIT1 Exon 6	c.503G>A	p.R168H	0.37-0.46	0.29-0.33	0.31	0.32	0.01	4.40%	5 of 5	100.0% (47.8%, 100.0%)
JAK3 Exon 4	c.366G>T	p.K122N	0.56-0.66	0.30-0.35	0.32	0.33	0.02	5.71%	5 of 5	100.0% (47.8%, 100.0%)
PNRC1 Exon 1	c.169C>A	p.L57I	1.38-1.84	0.30-0.33	0.32	0.31	0.01	3.23%	5 of 5	100.0% (47.8%, 100.0%)
RPTOR Exon 22	c.2621C>T	p.A874V	0.97-1.33	0.28-0.33	0.31	0.30	0.02	5.53%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
BRCA1 Exon 10	c.3784T>C	p.S1262P	0.19-0.49	0.26-0.37	0.32	0.32	0.04	12.74 %	5 of 5	100.0% (47.8%, 100.0%)
SMARCD1 Exon 6	c.699C>A	p.F233L	0.18-0.25	0.24-0.30	0.28	0.28	0.02	7.81%	5 of 5	100.0% (47.8%, 100.0%)
MDM4 Exon 4	c.266G>A	p.S89N	0.46-0.55	0.25-0.30	0.27	0.28	0.02	6.50%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 8	c.694C>T	p.R232*	0.47-0.53	0.31-0.36	0.33	0.33	0.02	5.25%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CG Exon 2	c.1426C>T	p.R476C	1.68-1.89	0.30-0.34	0.31	0.32	0.01	4.77%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 28	c.5943G>T	p.E1981D	1.40-1.65	0.31-0.33	0.32	0.32	0.01	1.87%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH2 Exon 7	c.1175C>T	p.T392I	1.04-1.14	0.24-0.34	0.31	0.32	0.04	11.57 %	5 of 5	100.0% (47.8%, 100.0%)
SETD2 Exon 3	c.4305G>T	p.E1435D	0.49-0.60	0.26-0.32	0.28	0.28	0.02	7.34%	5 of 5	100.0% (47.8%, 100.0%)
BAP1 Exon 6	c.437+3G>C	None	0.80-1.66	0.32-0.34	0.32	0.32	0.01	2.32%	5 of 5	100.0% (47.8%, 100.0%)
RICTOR Exon 22	c.2054T>A	p.L685H	0.20-0.31	0.28-0.39	0.33	0.29	0.05	14.93 %	5 of 5	100.0% (47.8%, 100.0%)
ESR1 Exon 6	c.767G>A	p.R256Q	1.30-1.46	0.30-0.35	0.32	0.32	0.02	4.69%	5 of 5	100.0% (47.8%, 100.0%)
TGFBR1 Exon 2	c.166G>A	p.V56I	0.55-0.66	0.33-0.34	0.33	0.33	0.00	1.28%	5 of 5	100.0% (47.8%, 100.0%)
STK11 Exon 1	c.83G>A	p.R28H	1.70-2.00	0.31-0.34	0.33	0.34	0.01	4.07%	5 of 5	100.0% (47.8%, 100.0%)
ARID5B Exon 1	c.10A>G	p.N4D	0.51-0.78	0.26-0.36	0.32	0.33	0.03	9.90%	5 of 5	100.0% (47.8%, 100.0%)
MPL Exon 12	c.1697G>A	p.S566N	1.28-1.48	0.30-0.36	0.32	0.32	0.02	6.55%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
STAG2 Exon 30	c.3197G>A	p.R1066Q	0.41-0.44	0.27-0.35	0.31	0.32	0.03	9.60%	5 of 5	100.0% (47.8%, 100.0%)
MSH2 Exon 4	c.687dupA	p.A230Sfs*2	0.34-0.46	0.29-0.34	0.32	0.32	0.02	6.35%	5 of 5	100.0% (47.8%, 100.0%)
NBN Exon 3	c.200C>T	p.T67I	0.23-0.34	0.27-0.45	0.32	0.28	0.07	21.98 %	5 of 5	100.0% (47.8%, 100.0%)
EPHA5 Exon 18	c.3103G>A	p.V1035M	0.18-0.23	0.25-0.33	0.30	0.31	0.03	9.81%	5 of 5	100.0% (47.8%, 100.0%)
AXIN1 Exon 10	c.2350G>A	p.V784I	0.79-1.34	0.32-0.35	0.33	0.34	0.01	2.94%	5 of 5	100.0% (47.8%, 100.0%)
PAK1 Exon 13	c.1313G>A	p.R438Q	0.47-0.52	0.27-0.30	0.28	0.28	0.01	3.89%	5 of 5	100.0% (47.8%, 100.0%)
PDGFRA Exon 18	c.2532G>A	p.M844I	0.46-0.56	0.27-0.35	0.31	0.31	0.03	9.09%	5 of 5	100.0% (47.8%, 100.0%)
RECQL Exon 12	c.1217-1G>T	None	0.20-0.27	0.26-0.32	0.28	0.27	0.02	8.79%	5 of 5	100.0% (47.8%, 100.0%)
NCOR1 Exon 12	c.1297G>A	p.D433N	0.46-0.50	0.34-0.36	0.35	0.34	0.01	2.39%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 2	c.241G>A	p.E81K	0.26-0.30	0.27-0.32	0.29	0.29	0.02	5.74%	5 of 5	100.0% (47.8%, 100.0%)
EP300 Exon 30	c.4897C>A	p.L1633M	1.49-1.64	0.30-0.33	0.31	0.31	0.01	3.48%	5 of 5	100.0% (47.8%, 100.0%)
SMARCA4 Exon 29	c.4047G>T	p.K1349N	0.81-1.02	0.33-0.36	0.34	0.33	0.01	3.52%	5 of 5	100.0% (47.8%, 100.0%)
CIC Exon 4	c.494C>T	p.P165L	1.60-1.80	0.30-0.35	0.32	0.31	0.02	5.05%	5 of 5	100.0% (47.8%, 100.0%)
SYK Exon 4	c.625C>T	p.H209Y	1.63-1.81	0.31-0.35	0.32	0.32	0.02	4.80%	5 of 5	100.0% (47.8%, 100.0%)
SMO Exon 10	c.1798G>A	p.V600M	0.76-0.91	0.31-0.37	0.35	0.35	0.02	5.97%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
CREBBP Exon 31	c.5597G>A	p.R1866H	1.96-2.33	0.33-0.34	0.33	0.33	0.00	1.37%	5 of 5	100.0% (47.8%, 100.0%)
ASXL2 Exon 8	c.799G>A	p.A267T	0.78-0.86	0.30-0.34	0.32	0.32	0.02	6.00%	5 of 5	100.0% (47.8%, 100.0%)
FLT4 Exon 3	c.266C>T	p.A89V	1.15-1.34	0.14-0.18	0.16	0.16	0.01	8.97%	5 of 5	100.0% (47.8%, 100.0%)
FLT3 Exon 2	c.143G>A	p.G48E	0.32-0.40	0.26-0.30	0.28	0.29	0.02	6.24%	5 of 5	100.0% (47.8%, 100.0%)
CIC Exon 10	c.2153C>A	p.P718H	1.62-1.93	0.32-0.35	0.33	0.33	0.01	3.52%	5 of 5	100.0% (47.8%, 100.0%)
MSH6 Exon 4	c.3104G>A	p.R1035Q	0.37-0.46	0.26-0.36	0.31	0.32	0.03	10.45 %	5 of 5	100.0% (47.8%, 100.0%)
INSR Exon 10	c.2195T>C	p.F732S	0.50-0.53	0.29-0.35	0.32	0.31	0.02	5.93%	5 of 5	100.0% (47.8%, 100.0%)
SDHA Exon 5	c.501G>T	p.K167N	1.50-1.68	0.22-0.25	0.23	0.22	0.01	5.07%	5 of 5	100.0% (47.8%, 100.0%)
SH2B3 Exon 2	c.446G>A	p.R149H	0.90-1.05	0.23-0.26	0.25	0.25	0.01	5.29%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 14	c.2044C>A	p.L682M	0.72-1.24	0.31-0.35	0.34	0.34	0.02	4.51%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 17	c.5002G>A	p.E1668K	0.52-0.63	0.05-0.12	0.07	0.06	0.02	32.23 %	5 of 5	100.0% (47.8%, 100.0%)
ASXL2 Exon 6	c.545G>A	p.C182Y	0.94-1.14	0.30-0.33	0.31	0.30	0.01	4.10%	5 of 5	100.0% (47.8%, 100.0%)
SETD2 Exon 12	c.6034C>T	p.L2012F	0.64-0.71	0.30-0.36	0.32	0.30	0.03	9.70%	5 of 5	100.0% (47.8%, 100.0%)
MSH6 Exon 4	c.2039C>T	p.A680V	0.37-0.46	0.26-0.39	0.33	0.34	0.04	13.47 %	5 of 5	100.0% (47.8%, 100.0%)
KDM6A Exon 24	c.3493G>A	p.V1165I	0.24-0.28	0.31-0.37	0.35	0.35	0.02	6.09%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
MRE11A Exon 5	c.364G>T	p.V122L	0.23-0.27	0.26-0.33	0.29	0.29	0.02	8.28%	5 of 5	100.0% (47.8%, 100.0%)
EP300 Exon 31	c.7180G>T	p.G2394*	2.03-2.27	0.31-0.32	0.31	0.31	0.00	1.16%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 11	c.1204C>T	p.R402C	1.18-1.41	0.29-0.35	0.32	0.31	0.02	6.59%	5 of 5	100.0% (47.8%, 100.0%)
WHSC1 Exon 12	c.2266G>A	p.G756S	0.94-1.07	0.48-0.52	0.49	0.48	0.01	3.03%	5 of 5	100.0% (47.8%, 100.0%)
POLE Exon 34	c.4411C>T	p.R1471C	1.13-1.37	0.31-0.32	0.31	0.31	0.01	1.89%	5 of 5	100.0% (47.8%, 100.0%)
ARAF Exon 9	c.856G>A	p.D286N	2.32-2.68	0.31-0.33	0.32	0.33	0.01	2.18%	5 of 5	100.0% (47.8%, 100.0%)
PPM1D Exon 6	c.1322G>A	p.R441H	0.21-0.61	0.28-0.35	0.31	0.33	0.03	9.52%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.5921C>T	p.P1974L	1.23-1.92	0.32-0.33	0.32	0.32	0.01	1.72%	5 of 5	100.0% (47.8%, 100.0%)
RPS6KB2 Exon 14	c.1166A>G	p.Y389C	0.79-0.98	0.24-0.27	0.26	0.25	0.01	4.24%	5 of 5	100.0% (47.8%, 100.0%)
SESN3 Exon 10	c.1428G>A	p.M476I	0.17-0.24	0.25-0.38	0.33	0.35	0.05	16.17 %	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.2785A>G	p.S929G	1.23-1.92	0.35-0.38	0.36	0.36	0.01	2.63%	5 of 5	100.0% (47.8%, 100.0%)
INPP4A Exon 17	c.1671G>T	p.E557D	1.35-1.59	0.33-0.35	0.34	0.34	0.01	1.85%	5 of 5	100.0% (47.8%, 100.0%)
KEAP1 Exon 3	c.1321G>A	p.E441K	1.00-1.22	0.14-0.17	0.16	0.16	0.01	8.34%	5 of 5	100.0% (47.8%, 100.0%)
DROSHA Exon 14	c.1921delT	p.C641Vfs*2	0.30-0.38	0.28-0.38	0.32	0.32	0.04	11.93 %	5 of 5	100.0% (47.8%, 100.0%)
MSH2 Exon 11	c.1738G>T	p.E580*	0.23-0.30	0.58-0.70	0.64	0.65	0.05	7.53%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
EPHA3 Exon 10	c.1865C>T	p.S622F	0.34-0.37	0.27-0.34	0.30	0.29	0.03	9.31%	5 of 5	100.0% (47.8%, 100.0%)
WT1 Exon N/A	c.202C>T	p.Q68*	0	0.31-0.34	0.33	0.32	0.01	3.67%	5 of 5	100.0% (47.8%, 100.0%)
ERBB4 Exon 13	c.1538G>T	p.W513L	0.73-0.84	0.03-0.04	0.04	0.04	0.01	18.25 %	5 of 5	100.0% (47.8%, 100.0%)
PTPRT Exon 28	c.3824C>T	p.T1275I	0.80-1.27	0.31-0.35	0.34	0.34	0.01	4.06%	5 of 5	100.0% (47.8%, 100.0%)
DNMT1 Exon 8	c.662G>A	p.R221H	0.48-0.58	0.29-0.33	0.31	0.30	0.02	5.22%	5 of 5	100.0% (47.8%, 100.0%)
MAP2K2 Exon 7	c.824T>C	p.L275P	0.97-1.38	0.31-0.35	0.33	0.33	0.02	5.99%	5 of 5	100.0% (47.8%, 100.0%)
SMAD3 Exon 8	c.1010-1G>T	None	1.41-1.55	0.25-0.28	0.26	0.26	0.01	5.14%	5 of 5	100.0% (47.8%, 100.0%)
CTNNB1 Exon 4	c.437C>T	p.A146V	0.45-0.51	0.27-0.35	0.30	0.30	0.03	9.05%	5 of 5	100.0% (47.8%, 100.0%)
RTEL1 Exon 6	c.594C>A	p.F198L	0.78-0.99	0.02-0.03	0.02	0.02	0.00	20.74 %	5 of 5	100.0% (47.8%, 100.0%)
KDM5C Exon 21	c.3233C>T	p.A1078V	0.93-1.03	0.32-0.36	0.34	0.33	0.01	3.85%	5 of 5	100.0% (47.8%, 100.0%)
ALOX12B Exon 6	c.743C>A	p.S248Y	0.75-0.85	0.25-0.32	0.30	0.31	0.02	7.73%	5 of 5	100.0% (47.8%, 100.0%)
BCOR Exon 4	c.494C>T	p.A165V	1.81-2.01	0.33-0.36	0.34	0.34	0.01	3.28%	5 of 5	100.0% (47.8%, 100.0%)
NCOA3 Exon 18	c.3434A>G	p.Q1145R	0.72-0.85	0.34-0.37	0.35	0.36	0.01	3.78%	5 of 5	100.0% (47.8%, 100.0%)
CREBBP Exon 2	c.475G>A	p.V159M	1.36-1.58	0.32-0.37	0.34	0.33	0.02	4.73%	5 of 5	100.0% (47.8%, 100.0%)
MAP2K2 Exon 9	c.989C>A	p.P330H	0.73-1.06	0.06-0.11	0.08	0.08	0.02	20.47 %	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
KDM5C Exon 4	c.446T>A	p.I149N	1.00-1.13	0.01-0.02	0.01	0.01	0.01	39.69 %	5 of 5	100.0% (47.8%, 100.0%)
NTRK2 Exon 19	c.2036G>A	p.G679D	1.11-1.45	0.32-0.36	0.34	0.34	0.01	3.34%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 5	c.385G>T	p.E129*	0.56-0.66	0.29-0.34	0.31	0.31	0.02	6.13%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 21	c.3140A>G	p.H1047R	0.21-0.27	0.25-0.31	0.28	0.29	0.02	8.68%	5 of 5	100.0% (47.8%, 100.0%)
TSC2 Exon 3	c.176G>A	p.R59Q	0.61-0.63	0.30-0.35	0.33	0.34	0.02	6.54%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 19	c.3151C>A	p.L1051M	0.21-0.61	0.27-0.38	0.32	0.32	0.04	12.58 %	5 of 5	100.0% (47.8%, 100.0%)
ERBB3 Exon 28	c.3748C>T	p.P1250S	0.88-0.98	0.30-0.35	0.32	0.32	0.02	6.18%	5 of 5	100.0% (47.8%, 100.0%)
ERBB4 Exon 14	c.1676G>A	p.C559Y	0.18-0.22	0.26-0.32	0.29	0.28	0.02	6.51%	5 of 5	100.0% (47.8%, 100.0%)
KMT2D Exon 10	c.1432C>A	p.P478T	1.38-1.64	0.29-0.34	0.32	0.33	0.02	5.18%	5 of 5	100.0% (47.8%, 100.0%)
AXIN2 Exon 4	c.1058C>T	p.P353L	0.42-0.91	0.44-0.57	0.50	0.52	0.05	9.11%	5 of 5	100.0% (47.8%, 100.0%)
FAT1 Exon 17	c.10315G>A	p.V3439I	0.74-0.87	0.32-0.35	0.34	0.34	0.01	2.63%	5 of 5	100.0% (47.8%, 100.0%)
RECQL Exon 7	c.505C>T	p.H169Y	0.17-0.24	0.22-0.36	0.27	0.27	0.05	17.64 %	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 31	c.4694C>T	p.T1565M	0.75-1.56	0.33-0.36	0.35	0.35	0.01	2.63%	5 of 5	100.0% (47.8%, 100.0%)
TSC2 Exon 42	c.5419G>A	p.V1807M	1.59-1.87	0.31-0.36	0.34	0.34	0.02	4.84%	5 of 5	100.0% (47.8%, 100.0%)
SUFU Exon 2	c.196G>T	p.D66Y	0.95-1.06	0.30-0.34	0.32	0.33	0.01	3.91%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
IRF4 Exon 4	c.426G>T	p.E142D	0.90-1.15	0.20-0.25	0.22	0.22	0.02	8.48%	5 of 5	100.0% (47.8%, 100.0%)
CSDE1 Exon 17	c.1898G>T	p.G633V	0.44-0.55	0.27-0.35	0.32	0.33	0.03	8.79%	5 of 5	100.0% (47.8%, 100.0%)
FUBP1 Exon 12	c.992G>A	p.R331Q	0.12-0.17	0.29-0.45	0.35	0.33	0.05	15.32 %	5 of 5	100.0% (47.8%, 100.0%)
POLE Exon 30	c.3788C>T	p.T1263I	0.96-1.10	0.31-0.36	0.34	0.35	0.02	5.59%	5 of 5	100.0% (47.8%, 100.0%)
CCND2 Exon 5	c.778C>T	p.Q260*	0.93-1.05	0.29-0.34	0.31	0.30	0.02	5.62%	5 of 5	100.0% (47.8%, 100.0%)
ATRX Exon 32	c.6975G>T	p.E2325D	0.13-0.27	0.27-0.38	0.30	0.29	0.04	13.46 %	5 of 5	100.0% (47.8%, 100.0%)
PALB2 Exon 12	c.3296C>T	p.T1099M	0.24-0.69	0.28-0.30	0.29	0.29	0.01	3.33%	5 of 5	100.0% (47.8%, 100.0%)
TET1 Exon 11	c.5315C>T	p.A1772V	0.95-0.99	0.27-0.33	0.30	0.31	0.02	6.92%	5 of 5	100.0% (47.8%, 100.0%)
ARAF Exon 2	c.94G>A	p.V32M	1.04-1.25	0.32-0.35	0.34	0.35	0.01	3.09%	5 of 5	100.0% (47.8%, 100.0%)
ABL1 Exon 11	c.2750C>T	p.A917V	1.76-2.03	0.30-0.33	0.32	0.32	0.01	2.46%	5 of 5	100.0% (47.8%, 100.0%)
ICOSLG Exon 7	c.649G>A	p.V217I	1.25-1.44	0.31-0.33	0.32	0.32	0.01	2.05%	5 of 5	100.0% (47.8%, 100.0%)
ESR1 Exon 10[a]	c.1673C>T	p.A558V	1.18-1.25	0.01-0.02	0.02	0.02	0.01	32.21 %	3 of 5	60.0% (14.7%, 94.7%)
ZFHX3 Exon 9	c.4174C>T	p.P1392S	2.11-2.25	0.25-0.26	0.25	0.26	0.00	1.91%	5 of 5	100.0% (47.8%, 100.0%)
ERCC5 Exon 8	c.1352T>C	p.V451A	2.09-2.29	0.30-0.33	0.31	0.31	0.01	3.45%	5 of 5	100.0% (47.8%, 100.0%)
XPO1 Exon 15	c.1711G>A	p.E571K	0.64-0.79	0.32-0.38	0.34	0.33	0.02	6.34%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
KMT2B Exon 31	c.7287G>T	p.E2429D	1.24-1.34	0.32-0.36	0.33	0.32	0.02	4.75%	5 of 5	100.0% (47.8%, 100.0%)
PTPRD Exon 21	c.1229C>T	p.T410I	1.29-1.45	0.02-0.03	0.02	0.02	0.00	16.16 %	5 of 5	100.0% (47.8%, 100.0%)
RPS6KA4 Exon 6	c.596G>A	p.G199D	1.02-1.28	0.31-0.35	0.33	0.33	0.02	5.01%	5 of 5	100.0% (47.8%, 100.0%)
CDK12 Exon 4	c.2164C>T	p.R722C	0.35-0.50	0.29-0.35	0.31	0.31	0.02	6.46%	5 of 5	100.0% (47.8%, 100.0%)
KEAP1 Exon 2	c.556G>A	p.G186S	1.09-1.32	0.01-0.03	0.02	0.02	0.01	25.60 %	5 of 5	100.0% (47.8%, 100.0%)
ERCC2 Exon 4	c.227C>T	p.T76I	0.62-0.79	0.31-0.38	0.34	0.33	0.03	7.69%	5 of 5	100.0% (47.8%, 100.0%)
PRKAR1A Exon 4	c.379G>A	p.A127T	0.08-0.28	0.26-0.34	0.30	0.31	0.03	8.73%	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 18	c.4801C>A	p.P1601T	1.29-2.30	0.30-0.35	0.33	0.33	0.02	5.32%	5 of 5	100.0% (47.8%, 100.0%)
FAT1 Exon 2	c.3265+1G>A	None	1.30-1.44	0.30-0.34	0.32	0.31	0.02	5.86%	5 of 5	100.0% (47.8%, 100.0%)
MGA Exon 17	c.6641T>C	p.V2214A	0.54-0.64	0.28-0.36	0.32	0.31	0.03	8.46%	5 of 5	100.0% (47.8%, 100.0%)
CTNNB1 Exon 3	c.101G>A	p.G34E	0.32-0.37	0.25-0.30	0.28	0.29	0.02	5.93%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K1 Exon 4	c.966A>G	p.I322M	1.16-1.42	0.31-0.33	0.32	0.32	0.01	2.18%	5 of 5	100.0% (47.8%, 100.0%)
FH Exon 9	c.1270G>A	p.G424R	0.40-0.46	0.30-0.35	0.32	0.32	0.02	5.93%	5 of 5	100.0% (47.8%, 100.0%)
PPP2R1A Exon 10	c.1285_1286insA	p.L429Hfs*12	0.71-1.87	0.29-0.33	0.31	0.30	0.01	4.45%	5 of 5	100.0% (47.8%, 100.0%)
STAT5A Exon 20	c.2290G>A	p.V764M	1.27-1.40	0.31-0.35	0.33	0.32	0.01	4.59%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
CARD11 Exon 10	c.1423_1426delinsAACA	p.D475_D476delinsNN	0.88-1.05	0.28-0.33	0.31	0.30	0.02	5.64%	5 of 5	100.0% (47.8%, 100.0%)
PIK3C2G Exon 5	c.347G>A	p.G116D	0.14-0.23	0.28-0.32	0.30	0.30	0.02	5.17%	5 of 5	100.0% (47.8%, 100.0%)
STAT5A Exon 10	c.1102delC	p.Q368Rfs*2	2.12-2.33	0.30-0.33	0.32	0.31	0.01	3.94%	5 of 5	100.0% (47.8%, 100.0%)
IRS1 Exon 1	c.637C>T	p.R213C	1.69-1.96	0.32-0.34	0.33	0.33	0.01	1.86%	5 of 5	100.0% (47.8%, 100.0%)
ARID1B Exon 1	c.1180G>A	p.A394T	1.24-2.63	0.34-0.37	0.35	0.36	0.01	3.18%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 5	c.562C>T	p.R188*	0.51-1.03	0.34-0.37	0.36	0.35	0.01	3.70%	5 of 5	100.0% (47.8%, 100.0%)
PDGFRA Exon 23	c.3229G>A	p.G1077S	0.51-0.57	0.22-0.31	0.27	0.28	0.03	12.76 %	5 of 5	100.0% (47.8%, 100.0%)
JAK1 Exon 3	c.169G>A	p.A57T	0.84-1.06	0.28-0.32	0.30	0.30	0.01	4.27%	5 of 5	100.0% (47.8%, 100.0%)
MSH6 Exon 5	c.3173-1G>T	None	0.79-0.88	0.29-0.35	0.32	0.32	0.02	6.18%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 22	c.4160T>C	p.V1387A	0.66-0.77	0.29-0.34	0.33	0.33	0.02	5.25%	5 of 5	100.0% (47.8%, 100.0%)
HRAS Exon 4	c.340G>A	p.V114M	1.10-1.44	0.02-0.03	0.02	0.02	0.01	22.76 %	5 of 5	100.0% (47.8%, 100.0%)
ESR1 Exon 3	c.405G>T	p.E135D	1.66-2.03	0.34-0.37	0.35	0.35	0.01	3.32%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 20	c.3106G>T	p.E1036*	0.73-0.80	0.21-0.29	0.25	0.25	0.03	11.53 %	5 of 5	100.0% (47.8%, 100.0%)
ERCC5 Exon 13	c.2694A>C	p.E898D	1.38-1.53	0.28-0.33	0.31	0.32	0.02	6.97%	5 of 5	100.0% (47.8%, 100.0%)
POLE Exon 13	c.1231G>T	p.V411L	0.91-1.04	0.31-0.35	0.32	0.32	0.02	4.98%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
NTRK3 Exon 17	c.1930C>T	p.P644S	1.86-2.16	0.01-0.02	0.01	0.01	0.00	31.13 %	5 of 5	100.0% (47.8%, 100.0%)
AR Exon 1	c.457C>T	p.P153S	2.16-2.37	0.30-0.35	0.32	0.32	0.02	5.50%	5 of 5	100.0% (47.8%, 100.0%)
MYD88 Exon 5	c.937G>A	p.A313T	1.45-1.55	0.30-0.33	0.32	0.32	0.01	3.14%	5 of 5	100.0% (47.8%, 100.0%)
ZFHX3 Exon 6	c.3562G>A	p.D1188N	1.70-1.94	0.34-0.37	0.35	0.35	0.01	2.86%	5 of 5	100.0% (47.8%, 100.0%)
NOTCH3 Exon 26	c.4777C>T	p.Q1593*	0.74-0.93	0.31-0.34	0.33	0.33	0.01	4.21%	5 of 5	100.0% (47.8%, 100.0%)
RICTOR Exon 22	c.2126G>T	p.R709I	0.20-0.31	0.25-0.31	0.28	0.29	0.02	7.75%	5 of 5	100.0% (47.8%, 100.0%)
PTCH1 Exon 22	c.3559G>A	p.A1187T	1.09-2.35	0.30-0.36	0.34	0.34	0.02	5.63%	5 of 5	100.0% (47.8%, 100.0%)
GRIN2A Exon 3[a]	c.96_99delinsACA	p.A33Hfs*2	1.22-1.22	0.29-0.29	0.29	0.29	0.00	0.00%	1 of 5	20.0% (0.5%, 71.6%)
SMARCA4 Exon 12	c.1855G>A	p.V619M	0.68-0.82	0.31-0.34	0.33	0.33	0.01	4.06%	5 of 5	100.0% (47.8%, 100.0%)
SPRED1 Exon 7	c.1175C>T	p.S392L	0.62-0.77	0.27-0.32	0.29	0.30	0.02	5.99%	5 of 5	100.0% (47.8%, 100.0%)
KLF4 Exon 3	c.521G>A	p.G174D	1.57-1.88	0.32-0.35	0.33	0.33	0.01	2.42%	5 of 5	100.0% (47.8%, 100.0%)
TP63 Exon 12	c.1555C>A	p.L519I	1.96-2.28	0.29-0.32	0.31	0.31	0.01	3.41%	5 of 5	100.0% (47.8%, 100.0%)
IKBKE Exon 13	c.1367G>A	p.R456Q	0.58-0.68	0.22-0.33	0.30	0.31	0.04	12.89 %	5 of 5	100.0% (47.8%, 100.0%)
AXL Exon 7	c.895C>T	p.L299F	0.52-1.21	0.31-0.34	0.33	0.33	0.01	3.30%	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 4	c.371A>G	p.E124G	0.98-1.21	0.32-0.35	0.34	0.33	0.01	4.29%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ZFHX3 Exon 10	c.10750G>A	p.A3584T	2.09-2.27	0.29-0.30	0.30	0.30	0.01	2.37%	5 of 5	100.0% (47.8%, 100.0%)
RAD21 Exon 13	c.1686G>T	p.Q562H	0.15-0.37	0.49-0.58	0.53	0.54	0.03	6.53%	5 of 5	100.0% (47.8%, 100.0%)
SMARCA4 Exon 26	c.3656A>G	p.K1219R	0.83-0.98	0.33-0.35	0.34	0.34	0.01	2.29%	5 of 5	100.0% (47.8%, 100.0%)
PREX2 Exon 26	c.3147-2A>T	None	0.55-0.68	0.25-0.36	0.30	0.28	0.04	13.36 %	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 27[a]	c.4150C>T	p.R1384W	1.25-1.30	0.01-0.02	0.02	0.02	0.01	32.59 %	2 of 5	40.0% (5.3%, 85.3%)
BRCA2 Exon 8	c.674C>T	p.T225I	0.09-0.23	0.31-0.42	0.34	0.32	0.04	11.78 %	5 of 5	100.0% (47.8%, 100.0%)
CD276 Exon 8	c.1546G>T	p.A516S	0.85-0.99	0.21-0.26	0.23	0.24	0.02	6.77%	5 of 5	100.0% (47.8%, 100.0%)
ASXL1 Exon 12	c.3940C>A	p.L1314I	2.12-2.37	0.32-0.36	0.34	0.33	0.02	4.70%	5 of 5	100.0% (47.8%, 100.0%)
HNF1A Exon 4	c.864delG	p.P291Qfs*51	1.07-1.38	0.32-0.33	0.32	0.32	0.00	1.43%	5 of 5	100.0% (47.8%, 100.0%)
PGR Exon 6	c.2409G>T	p.Q803H	0.18-0.24	0.29-0.36	0.34	0.35	0.02	7.06%	5 of 5	100.0% (47.8%, 100.0%)
ABL1 Exon 11	c.2080G>A	p.E694K	1.76-2.03	0.33-0.35	0.34	0.35	0.00	1.39%	5 of 5	100.0% (47.8%, 100.0%)
EPHA5 Exon 3	c.646G>A	p.D216N	1.16-1.19	0.30-0.36	0.32	0.32	0.02	5.64%	5 of 5	100.0% (47.8%, 100.0%)
RPS6KA4 Exon 10	c.1072-3C>T	None	1.38-1.70	0.30-0.35	0.33	0.33	0.02	5.48%	5 of 5	100.0% (47.8%, 100.0%)
XPO1 Exon 13	c.1345G>T	p.D449Y	0.34-0.51	0.26-0.37	0.33	0.36	0.04	12.86 %	5 of 5	100.0% (47.8%, 100.0%)
PTCH1 Exon 14	c.2151C>A	p.D717E	0.89-1.99	0.31-0.35	0.33	0.34	0.01	3.75%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
EPAS1 Exon 13	c.2128C>T	p.R710*	1.61-1.92	0.31-0.32	0.31	0.31	0.00	1.17%	5 of 5	100.0% (47.8%, 100.0%)
PTEN Exon 6	c.531T>G	p.Y177*	0.19-0.51	0.28-0.42	0.34	0.34	0.05	13.51 %	5 of 5	100.0% (47.8%, 100.0%)
PLK2 Exon 7	c.757G>A	p.G253S	0.20-0.24	0.46-0.54	0.49	0.49	0.03	5.59%	5 of 5	100.0% (47.8%, 100.0%)
MGA Exon 2	c.116G>T	p.G39V	0.49-0.56	0.22-0.27	0.25	0.25	0.02	7.15%	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 10[a]	c.1437_1438delinsCA	p.A480T	1.18-1.38	0.01-0.94	0.47	0.47	0.46	97.05 %	4 of 5	80.0% (28.4%, 99.5%)
CARM1 Exon 2	c.265G>T	p.E89*	0.88-1.00	0.33-0.37	0.35	0.35	0.02	4.39%	5 of 5	100.0% (47.8%, 100.0%)
SPEN Exon 11	c.5671G>A	p.E1891K	1.44-1.57	0.30-0.33	0.32	0.32	0.01	3.38%	5 of 5	100.0% (47.8%, 100.0%)
BCOR Exon 4	c.2818A>G	p.T940A	1.81-2.01	0.33-0.35	0.34	0.34	0.01	2.37%	5 of 5	100.0% (47.8%, 100.0%)
FANCA Exon 11	c.898G>A	p.G300R	0.45-0.54	0.25-0.28	0.26	0.25	0.01	5.68%	5 of 5	100.0% (47.8%, 100.0%)
VEGFA Exon 6	c.966delA	p.K322Nfs*32	1.62-1.86	0.30-0.33	0.32	0.32	0.01	2.59%	5 of 5	100.0% (47.8%, 100.0%)
MED12 Exon 8	c.1111C>A	p.L371M	1.14-1.21	0.31-0.36	0.33	0.34	0.02	6.02%	5 of 5	100.0% (47.8%, 100.0%)
CBL Exon 4	c.610A>G	p.S204G	0.48-0.56	0.30-0.38	0.33	0.33	0.02	7.34%	5 of 5	100.0% (47.8%, 100.0%)
NTHL1 Exon 3	c.440C>T	p.T147M	1.40-1.74	0.31-0.35	0.33	0.33	0.02	5.12%	5 of 5	100.0% (47.8%, 100.0%)
EZH2 Exon 17	c.1976G>A	p.R659K	0.51-0.54	0.25-0.34	0.29	0.29	0.03	9.83%	5 of 5	100.0% (47.8%, 100.0%)
IGF1R Exon 12	c.2608G>A	p.G870R	0.70-0.79	0.47-0.52	0.49	0.49	0.01	3.02%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
SMARCB1 Exon 9[a]	c.1129C>T	p.R377C	0.77-0.94	0.01-0.02	0.02	0.01	0.01	32.44 %	4 of 5	80.0% (28.4%, 99.5%)
AXIN2 Exon 2	c.520G>A	p.A174T	1.23-2.28	0.45-0.49	0.48	0.49	0.02	3.76%	5 of 5	100.0% (47.8%, 100.0%)
PTEN Exon 2	c.164+1G>A	None	0.04-0.24	0.29-0.43	0.35	0.34	0.05	14.39 %	5 of 5	100.0% (47.8%, 100.0%)
SPEN Exon 11	c.1888C>T	p.R630C	1.44-1.57	0.29-0.34	0.32	0.33	0.02	5.80%	5 of 5	100.0% (47.8%, 100.0%)
PIK3R3 Exon 4	c.233A>G	p.K78R	0.17-0.25	0.20-0.31	0.24	0.21	0.04	17.30 %	5 of 5	100.0% (47.8%, 100.0%)
CUL3 Exon 12	c.1637G>A	p.R546Q	0.21-0.24	0.20-0.32	0.28	0.30	0.04	14.97 %	5 of 5	100.0% (47.8%, 100.0%)
MTOR Exon 35	c.4955C>T	p.T1652I	1.42-1.58	0.31-0.34	0.32	0.31	0.01	4.32%	5 of 5	100.0% (47.8%, 100.0%)
PARP1 Exon 12	c.1678G>A	p.V560M	0.94-1.13	0.31-0.37	0.34	0.33	0.02	6.08%	5 of 5	100.0% (47.8%, 100.0%)
KMT2C Exon 38	c.7913C>A	p.S2638Y	0.95-1.11	0.33-0.34	0.33	0.33	0.01	1.74%	5 of 5	100.0% (47.8%, 100.0%)
ROS1 Exon 29	c.4766A>C	p.E1589A	0.21-0.28	0.28-0.40	0.33	0.31	0.04	12.04 %	5 of 5	100.0% (47.8%, 100.0%)
MAP2K4 Exon 1	c.46A>G	p.S16G	1.10-1.54	0.34-0.38	0.36	0.36	0.02	4.46%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 9	c.1411C>T	p.P471S	0.28-0.39	0.03-0.08	0.05	0.05	0.01	30.95 %	5 of 5	100.0% (47.8%, 100.0%)
POLD1 Exon 13	c.1574G>A	p.R525Q	0.65-0.86	0.31-0.36	0.34	0.34	0.02	4.64%	5 of 5	100.0% (47.8%, 100.0%)
CSDE1 Exon 11	c.994C>T	p.R332C	0.51-0.59	0.30-0.36	0.33	0.32	0.02	6.88%	5 of 5	100.0% (47.8%, 100.0%)
FAT1 Exon 2	c.145G>T	p.A49S	1.30-1.44	0.29-0.32	0.31	0.30	0.01	4.60%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ARID1B Exon 18	c.4726C>T	p.P1576S	1.29-2.30	0.30-0.35	0.32	0.31	0.02	5.35%	5 of 5	100.0% (47.8%, 100.0%)
KDM5A Exon 22	c.3276delA	p.V1093*	0.48-0.58	0.28-0.36	0.32	0.31	0.03	8.37%	5 of 5	100.0% (47.8%, 100.0%)
IRF4 Exon 6	c.680A>C	p.E227A	1.04-1.40	0.44-0.50	0.47	0.48	0.02	5.09%	5 of 5	100.0% (47.8%, 100.0%)
RECQL Exon 11	c.1151G>T	p.R384M	0.17-0.25	0.23-0.40	0.32	0.34	0.07	21.78 %	5 of 5	100.0% (47.8%, 100.0%)
DNMT3A Exon 19	c.2206C>T	p.R736C	1.57-1.76	0.28-0.31	0.30	0.31	0.01	4.62%	5 of 5	100.0% (47.8%, 100.0%)
NCOR1 Exon 20	c.2561A>G	p.E854G	1.48-1.60	0.34-0.35	0.34	0.34	0.01	1.53%	5 of 5	100.0% (47.8%, 100.0%)
NTRK1 Exon 4	c.360-1G>T	None	0.73-0.81	0.31-0.34	0.32	0.32	0.01	4.41%	5 of 5	100.0% (47.8%, 100.0%)
AKT1 Exon 4	c.175+1G>A	None	1.38-1.69	0.31-0.34	0.32	0.32	0.01	3.18%	5 of 5	100.0% (47.8%, 100.0%)
INPP4A Exon 23	c.2446A>G	p.T816A	0.34-0.41	0.25-0.33	0.28	0.28	0.03	10.47 %	5 of 5	100.0% (47.8%, 100.0%)
AKT1 Exon 7	c.445G>A	p.E149K	1.61-1.89	0.33-0.35	0.34	0.34	0.01	2.18%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K1 Exon 14	c.3202G>T	p.G1068C	0.77-0.91	0.31-0.34	0.32	0.31	0.02	4.72%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 12	c.7599G>T	p.E2533D	0.96-1.47	0.32-0.37	0.34	0.33	0.02	4.92%	5 of 5	100.0% (47.8%, 100.0%)
SMAD4 Exon 7	c.809G>A	p.G270E	0.58-0.68	0.28-0.31	0.29	0.29	0.01	3.12%	5 of 5	100.0% (47.8%, 100.0%)
POLE Exon 20	c.2224C>T	p.R742C	0.77-0.89	0.32-0.35	0.33	0.33	0.01	3.73%	5 of 5	100.0% (47.8%, 100.0%)
MED12 Exon 3	c.244C>T	p.R82C	0.97-1.11	0.14-0.16	0.15	0.16	0.01	5.30%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ATM Exon 42	c.6100C>T	p.R2034*	0.22-0.51	0.24-0.32	0.29	0.29	0.03	10.55 %	5 of 5	100.0% (47.8%, 100.0%)
FAT1 Exon 9	c.4708C>T	p.R1570W	1.40-1.69	0.27-0.29	0.28	0.28	0.01	2.70%	5 of 5	100.0% (47.8%, 100.0%)
CREBBP Exon 31	c.5348G>A	p.C1783Y	1.96-2.33	0.30-0.36	0.33	0.33	0.02	5.83%	5 of 5	100.0% (47.8%, 100.0%)
STK40 Exon 6	c.346C>A	p.R116S	1.37-1.45	0.27-0.31	0.29	0.29	0.02	5.39%	5 of 5	100.0% (47.8%, 100.0%)
RAB35 Exon 5	c.451G>A	p.A151T	1.13-1.55	0.63-0.70	0.66	0.65	0.02	3.14%	5 of 5	100.0% (47.8%, 100.0%)
PIK3R1 Exon 12	c.1507C>T	p.R503W	0.25-0.37	0.27-0.34	0.30	0.29	0.03	8.40%	5 of 5	100.0% (47.8%, 100.0%)
RUNX1 Exon 9	c.1041G>A	p.M347I	1.70-2.02	0.31-0.36	0.32	0.32	0.02	5.06%	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 33	c.5128C>T	p.R1710C	0.56-1.14	0.30-0.33	0.32	0.32	0.01	3.32%	5 of 5	100.0% (47.8%, 100.0%)
KMT2B Exon 3	c.817G>A	p.G273S	1.36-1.58	0.33-0.35	0.33	0.33	0.01	2.22%	5 of 5	100.0% (47.8%, 100.0%)
PRKCI Exon 3	c.235A>G	p.T79A	0.17-0.37	0.27-0.35	0.30	0.31	0.03	10.53 %	5 of 5	100.0% (47.8%, 100.0%)
DOT1L Exon 24	c.3307G>A	p.V1103M	1.88-2.34	0.32-0.37	0.34	0.34	0.02	4.68%	5 of 5	100.0% (47.8%, 100.0%)
FLT3 Exon 22	c.2728C>T	p.Q910*	0.35-0.44	0.29-0.39	0.33	0.33	0.03	10.19 %	5 of 5	100.0% (47.8%, 100.0%)
PTEN Exon 6	c.518G>A	p.R173H	0.19-0.51	0.24-0.38	0.30	0.30	0.05	15.26 %	5 of 5	100.0% (47.8%, 100.0%)
PTPRS Exon 32	c.5041G>A	p.E1681K	1.02-1.76	0.28-0.34	0.32	0.33	0.02	6.41%	5 of 5	100.0% (47.8%, 100.0%)
BCOR Exon 14	c.4964C>A	p.S1655Y	0.98-1.05	0.32-0.36	0.34	0.34	0.02	5.18%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
SMARCB1 Exon 4	c.457A>G	p.R153G	0.65-0.82	0.31-0.38	0.35	0.36	0.02	7.16%	5 of 5	100.0% (47.8%, 100.0%)
POLE Exon 45	c.6326G>A	p.C2109Y	0.68-0.76	0.30-0.37	0.34	0.34	0.03	8.10%	5 of 5	100.0% (47.8%, 100.0%)
KMT2A Exon 3	c.2318delC	p.P773Rfs*8	0.80-0.90	0.28-0.33	0.30	0.29	0.02	6.88%	5 of 5	100.0% (47.8%, 100.0%)
ANKRD11 Exon 10	c.4135G>A	p.D1379N	1.23-1.92	0.31-0.34	0.33	0.32	0.01	3.10%	5 of 5	100.0% (47.8%, 100.0%)
ERCC5 Exon 8	c.1405G>A	p.D469N	2.09-2.29	0.30-0.33	0.31	0.31	0.01	2.92%	5 of 5	100.0% (47.8%, 100.0%)
PRDM1 Exon 5	c.1000A>G	p.T334A	1.09-2.41	0.30-0.33	0.31	0.31	0.01	3.03%	5 of 5	100.0% (47.8%, 100.0%)
KDM6A Exon 18	c.2093_2094delGT	p.G698Afs*9	0.45-0.51	0.15-0.24	0.18	0.18	0.03	16.06 %	5 of 5	100.0% (47.8%, 100.0%)
KRAS Exon 2	**c.35G>A**	**p.G12D**	**0.49-0.59**	**0.14-0.22**	**0.18**	**0.18**	**0.02**	**12.90 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
TEK Exon 9	c.1327G>A	p.V443I	0.74-0.83	0.05-0.08	0.07	0.08	0.01	15.25 %	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 7	c.742C>T	p.R248W	0.51-0.62	0.15-0.23	0.19	0.19	0.03	14.04 %	5 of 5	100.0% (47.8%, 100.0%)
RPTOR Exon 16	c.1786G>A	p.V596M	1.15-1.37	0.10-0.13	0.12	0.11	0.01	8.22%	5 of 5	100.0% (47.8%, 100.0%)
H3F3C Exon 1	c.155G>A	p.R52H	1.41-1.57	0.03-0.05	0.04	0.04	0.01	13.58 %	5 of 5	100.0% (47.8%, 100.0%)
PIK3C2G Exon 20	**c.2090T>C**	**p.L697P**	**0.25-0.32**	**0.33-0.39**	**0.36**	**0.35**	**0.02**	**5.82%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
AR Exon 1[a]	c.1420_1421insGCG	p.G473_G451ins	1.01-1.01	0.48-0.48	0.48	0.48	0.00	0.00%	1 of 5	20.0% (0.5%, 71.6%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
KIT Exon 9	**c.1504_1509dupGCCTAT**	**p.Y503_A502ins**	**0.81-0.95**	**0.32-0.38**	**0.36**	**0.35**	**0.02**	**5.97%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
PTEN Exon 5	**c.262_264delinsAG**	**p.Y88Sfs*11**	**0.29-0.71**	**0.37-0.54**	**0.48**	**0.49**	**0.06**	**12.63 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
APC Exon 17	c.7028C>G	p.S2343C	0.93-1.00	0.01-0.02	0.01	0.01	0.00	32.26 %	5 of 5	100.0% (47.8%, 100.0%)
KRAS Exon 2	c.38G>A	p.G13D	0.76-0.81	0.19-0.27	0.23	0.22	0.03	13.94 %	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 17	c.2971G>T	p.E991*	0.93-1.00	0.19-0.23	0.21	0.22	0.01	6.44%	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 5	c.473G>A	p.R158H	0.81-0.88	0.48-0.52	0.51	0.51	0.02	3.28%	5 of 5	100.0% (47.8%, 100.0%)
DICER1 Exon 23	c.4921T>A	p.C1641S	0.87-0.94	0.45-0.54	0.52	0.53	0.03	6.48%	5 of 5	100.0% (47.8%, 100.0%)
APC Exon 7	c.536C>T	p.S179F	0.48-0.58	0.05-0.10	0.08	0.08	0.02	22.94 %	5 of 5	100.0% (47.8%, 100.0%)
CD79B Exon 3	c.295G>A	p.E99K	0.74-0.87	0.08-0.10	0.09	0.09	0.01	7.09%	5 of 5	100.0% (47.8%, 100.0%)
BRAF Exon 15	**c.1798_1799delinsAA**	**p.V600K**	**0.24-0.62**	**0.21-0.36**	**0.31**	**0.32**	**0.05**	**16.92 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
NF1 Exon 25	c.3208C>T	p.Q1070*	0.43-0.51	0.05-0.11	0.08	0.08	0.02	24.10 %	5 of 5	100.0% (47.8%, 100.0%)
TBX3 Exon 1	c.193A>C	p.M65L	1.41-1.78	0.04-0.05	0.04	0.04	0.00	8.53%	5 of 5	100.0% (47.8%, 100.0%)
NSD1 Exon 5	c.2345G>A	p.S782N	0.96-1.06	0.08-0.12	0.09	0.09	0.02	18.50 %	5 of 5	100.0% (47.8%, 100.0%)
MAP3K14 Exon 12	c.2264C>T	p.T755I	1.07-1.45	0.07-0.10	0.09	0.09	0.01	13.36 %	5 of 5	100.0% (47.8%, 100.0%)
ZFHX3 Exon 2	c.869C>T	p.S290F	1.77-2.07	0.07-0.10	0.09	0.09	0.01	8.94%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
ATRX Exon 9	c.1426C>T	p.Q476*	0.40-0.88	0.08-0.09	0.09	0.09	0.01	7.65%	5 of 5	100.0% (47.8%, 100.0%)
KMT2A Exon 3	c.2503C>T	p.L835F	1.07-1.18	0.09-0.11	0.10	0.09	0.01	7.75%	5 of 5	100.0% (47.8%, 100.0%)
ARID2 Exon 11	c.1474C>T	p.Q492*	0.60-1.24	0.07-0.10	0.09	0.09	0.01	11.03 %	5 of 5	100.0% (47.8%, 100.0%)
EPHA7 Exon 16	c.2824G>A	p.D942N	0.69-0.78	0.08-0.11	0.10	0.09	0.01	11.94 %	5 of 5	100.0% (47.8%, 100.0%)
ASXL2 Exon 12	c.3445C>T	p.R1149C	1.50-1.67	0.09-0.10	0.09	0.09	0.00	4.30%	5 of 5	100.0% (47.8%, 100.0%)
PREX2 Exon 9	c.962G>A	p.G321E	0.60-0.67	0.09-0.12	0.10	0.09	0.01	13.12 %	5 of 5	100.0% (47.8%, 100.0%)
CSF1R Exon 17	c.2251G>A	p.E751K	0.74-0.88	0.07-0.11	0.09	0.09	0.01	12.66 %	5 of 5	100.0% (47.8%, 100.0%)
CARD11 Exon 13	c.1696G>A	p.E566K	0.76-0.98	0.12-0.18	0.14	0.14	0.02	13.80 %	5 of 5	100.0% (47.8%, 100.0%)
PGR Exon 8	c.2724G>A	p.M908I	0.50-0.57	0.07-0.10	0.08	0.07	0.01	11.63 %	5 of 5	100.0% (47.8%, 100.0%)
ZFHX3 Exon 2	c.2099C>T	p.S700F	1.77-2.07	0.08-0.10	0.09	0.09	0.01	11.52 %	5 of 5	100.0% (47.8%, 100.0%)
NEGR1 Exon 7	c.962G>A	p.G321E	0.42-0.54	0.06-0.09	0.07	0.08	0.01	15.58 %	5 of 5	100.0% (47.8%, 100.0%)
CDK12 Exon 8	c.2669G>A	p.R890H	0.41-0.52	0.20-0.23	0.21	0.21	0.01	5.18%	5 of 5	100.0% (47.8%, 100.0%)
SOX17 Exon 2	c.1134dupC	p.Y379Lfs*5	2.79-3.74	0.48-0.52	0.49	0.48	0.02	3.17%	5 of 5	100.0% (47.8%, 100.0%)
ATM Exon 7	c.769G>T	p.E257*	0.25-0.61	0.30-0.39	0.36	0.38	0.03	8.50%	5 of 5	100.0% (47.8%, 100.0%)
FBXW7 Exon 10	c.1513C>G	p.R505G	0.47-0.51	0.58-0.66	0.62	0.63	0.02	3.87%	5 of 5	100.0% (47.8%, 100.0%)

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
TP53 Exon 4	c.102_103insC	p.P36Afs*7	0.96-1.13	0.37-0.42	0.40	0.39	0.02	4.72%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 10	**c.1624G>A**	**p.E542K**	**0.25-0.38**	**0.74-0.85**	**0.80**	**0.82**	**0.04**	**4.98%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
KMT2C Exon 29	c.4430_4433delATCA	p.N1477Rfs*6	0.39-0.47	0.20-0.29	0.24	0.25	0.03	14.02 %	5 of 5	100.0% (47.8%, 100.0%)
PTPRD Exon 28	c.2989C>T	p.H997Y	1.67-1.85	0.33-0.38	0.35	0.35	0.02	4.85%	5 of 5	100.0% (47.8%, 100.0%)
MAP3K1 Exon 13	c.2367delC	p.R790Gfs*9	0.48-0.61	0.41-0.46	0.43	0.43	0.02	3.89%	5 of 5	100.0% (47.8%, 100.0%)
FGFR3 Exon 10	c.1321G>A	p.A441T	1.15-1.46	0.18-0.20	0.19	0.19	0.01	3.59%	5 of 5	100.0% (47.8%, 100.0%)
TP53 Exon 6	c.637C>T	p.R213*	0.63-0.73	0.33-0.41	0.37	0.37	0.03	7.91%	5 of 5	100.0% (47.8%, 100.0%)
PIK3CA Exon 21	**c.3140A>G**	**p.H1047R**	**0.44-0.52**	**0.15-0.20**	**0.17**	**0.17**	**0.02**	**8.87%**	**5 of 5**	**100.0% (47.8%, 100.0%)**
BRAF Exon 15	**c.1799T>A**	**p.V600E**	**0.23-0.60**	**0.10-0.13**	**0.12**	**0.13**	**0.01**	**10.62 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
KRAS Exon 2	**c.38G>A**	**p.G13D**	**0.57-0.69**	**0.12-0.21**	**0.15**	**0.14**	**0.03**	**21.55 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
EGFR Exon 21	**c.2573T>G**	**p.L858R**	**1.58-1.78**	**0.02-0.03**	**0.03**	**0.03**	**0.00**	**10.70 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
KRAS Exon 2	**c.35G>A**	**p.G12D**	**0.57-0.69**	**0.04-0.07**	**0.06**	**0.06**	**0.01**	**18.26 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
PIK3CA Exon 10	**c.1633G>A**	**p.E545K**	**0.22-0.28**	**0.10-0.18**	**0.14**	**0.15**	**0.03**	**18.85 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
KIT Exon 17	**c.2447A>T**	**p.D816V**	**0.30-0.38**	**0.09-0.16**	**0.13**	**0.13**	**0.02**	**17.36 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**
EGFR Exon 18	**c.2155G>A**	**p.G719S**	**1.50-1.69**	**0.21-0.26**	**0.23**	**0.23**	**0.02**	**7.13%**	**5 of 5**	**100.0% (47.8%, 100.0%)**

Gene/ Exon	Coding Change	Protein Change	NC Range	MAF Range	MAF Mean	MAF Median	MAF (SD)	MAF (%CV)	Positive/ Total Calls	Positive Call Rate (two-sided 95% CI)
NRAS Exon 3	**c.181C>A**	**p.Q61K**	**0.50-0.56**	**0.10-0.15**	**0.12**	**0.12**	**0.02**	**14.78 %**	**5 of 5**	**100.0% (47.8%, 100.0%)**

[a] Discordant

iii. *Precision – Per-Specimen Precision:* Precision was calculated for each individual specimen as shown in Table 8. Results from the precision studies were combined across all reportable genes for each specimen. The positive call rate was calculated based on the total number of mutations along with the two-sided 95% confidence interval. The results showed that all mutations have 100% concordance for 511 out of 530 unique mutations, or 96.4% (94.5%-97.8% CI).

The results showed that all mutations have 100% concordance in all replicates except for 19 mutations in 5 of the clinical specimens. In the clinical specimens, discordance was observed for 15 SNVs, 2 deletions, and 2 insertions.

Table 8. Precision per specimen across all reportable mutations (N – 5 replicates)

Specimen	Total No. Unique Mutations Detected Across All 5 Replicates	Positive Call Rate Per Mutation	Positive Call Rate[1] (two-sided 95% CI)	Negative Call Rate[2] (two-sided 95% CI)
1	5	5/5 for 4, 4/5 for 1	24/25 96.0% (79.6%-99.9%)	-
2	67	5/5 for 63, 4/5 for 2, 2/5 for 2	327/335 97.6% (95.3%-99.0%)	6/10 60.0% (26.2%-87.8%)
3	5	5/5 for 5	25/25 100.0% (86.3%-100.0%)	-
4	11	5/5 for 11	55/55 100.0% (93.5%-100.0%)	-
5	376	5/5 for 364 4/5 for 5 3/5 for 3 2/5 for 1 1/5 for 3	1854/1880 98.6% (98.0%-99.1%)	15/20 75.0% (50.9%-91.3%)

Specimen	Total No. Unique Mutations Detected Across All 5 Replicates	Positive Call Rate Per Mutation	Positive Call Rate[1] (two-sided 95% CI)	Negative Call Rate[2] (two-sided 95% CI)
6	6	5/5 for 6	30/30 100.0% (88.4%-100.0%)	-
7	1	5/5 for 1	5/5 100.0% (47.8%-100.0%)	-
8	9	5/5 for 9	45/45 100.0% (92.1%-100.0%)	-
9	6	5/5 for 6	30/30 100.0% (88.4%-100.0%)	-
10	13	5/5 for 12, 1/5 for 1	61/65 93.8% (85.0%-98.3%)	4/5 80.0% (28.4%-99.5%)
11	3	5/5 for 2, 1/5 for 1	11/15 73.3% (44.9%-92.2%)	4/5 80.0% (28.4%-99.5%)
12	19	5/5 for 19	95/95 100.0% (96.2%-100.0%)	-
13	9	5/5 for 9	45/45 100.0% (92.1%-100.0%)	-

[1] Positive call rate was calculated based on variants with majority call detected as positive.
[2] Negative call rate was calculated based on variants detected at least once, but with majority call as negative. For all other locations, the negative call rates are 100%.

iv. *Precision – Well-Characterized Reference Material:* The precision of Omics Core was also assessed through repeated measurements of the reference material NA12878 (pre-extracted DNA provided by Coriell Institute). Table 9 below summarizes precision data (agreement on variant calls across all replicates) binned by GC content (e.g., 0-5%, 5-10%, 10-25% 25-50%, >50%) and provide additional precision data on key quality metrics. The positive call rate for all 420 variants across all 20 passing replicates was 8,388 out of 8400, or 99.86% (99.75%-99.93% CI). Differentiated by zygosity, the positive call rate was 5610 out of 5620, or 99.82% (99.67%-99.91%) for heterozygous loci and 2778 out of 2780, or 99.93% (99.74%-99.99%) for homozygous loci.

Table 9. Performance across All Runs of the Reference Material

GC Bin	# Identical	Total (over all runs)	% Identical	Min DP*	Max DP*	Avg DP*	Std Dev DP*
[25,30)	80	80	100.00	90	445	238.3	93.1
[30,35)	679	680	99.85	46	876	287.5	133.6
[35,40)	820	820	100.00	70	904	382.3	164.3
[40,45)	980	980	100.00	74	1369	425.4	200.5
[45,50)	960	960	100.00	75	1224	411.1	198.4
[50,55)	919	920	99.89	157	1616	487.8	231.8
[55,60)	1258	1260	99.84	142	1907	549.7	256.3
[60,65)	1237	1240	99.76	147	1726	590.5	245
[65,70)	915	920	99.46	185	2363	675.5	280
[70,75)	400	400	100.00	169	2060	750.9	365.8
[75,80)	120	120	100.00	156	1211	519.4	205.4
[80,85)	20	20	100.00	315	754	468.6	125.9

*DP (Total coverage depth of reads at a site)

v. *Precision – Tumor Mutational Burden (TMB)* TMB is reported as mutations per megabase (MUT/MB). Table 10 summarizes TMB precision analysis based on all 13 samples assessed. Specimens spanned the range of TMB scores and included multiple tumor types. The tumor purity for these samples ranged from 25% to 90%. The per sample TMB analysis demonstrated repeatable and reproducible TMB rates with a % CV <10% for all 13 samples.

Table 10. TMB Precision Analysis

Sample #	Tumor Type	Tumor Purity	Mean TMB Value	# of Valid Results	SD	%CV	Average Mean Exon Coverage
1	Colon	35%	56.30	5	0.17	0.30	698.69
2	Pancreatic	35%	2.56	5	0.08	3.06	671.37
3	Mullerian Ovarian	75%	3.91	5	0.03	0.77	753.40
4	Endometrial	65%	384.13	5	0.59	0.15	773.13

Sample #	Tumor Type	Tumor Purity	Mean TMB Value	# of Valid Results	SD	%CV	Average Mean Exon Coverage
5	Adenoid Cystic Carcinoma	80%	0.90	5	0.06	6.10	730.67
6	Malignant Gastric GIST	90%	1.05	5	0.06	6.07	707.26
7	Colon	55%	5.23	5	0.10	1.94	681.81
8	Colon	55%	3.62	5	0.09	2.62	654.99
9	Metastatic Breast	60%	9.34	5	0.13	1.36	802.78
10	Glioblastoma	80%	1.82	5	0.14	7.75	765.82
11	Melanoma	65%	25.48	5	0.15	0.58	767.87
12	NSCLC	25%	1.56	5	0.04	2.59	794.63
13	Commercial Cell Line		880.79	5	2.67	0.30	747.01

b. Analytical Sensitivity - Limit of Detection (LoD)

i. LoD –SNVs and Indels

The LoD of the Omics Core assay is defined as the mutant allele fraction at which 95% of replicates for a variant type are reliably detected, of the Omics Core assay was evaluated by assessing a minimum of ten (10) FFPE clinical tumor samples from clinical cases across a diverse set of cancers. Specimens were selected for mutations with evidence of clinical significance for SNVs, small insertions and deletions. The LoD study was comprised of two steps: LoD establishment, sequencing of a specific variant over a dilution series, and LoD confirmation, sequencing of a specific variant at the established LoD.

Part1: Dilution Series: For LoD establishment, mutations in 5 exons with the lowest and highest coverage were assessed based on the mean normalized coverage for all exons within the 10 FFPE clinical samples.The 5 exons with lowest coverage correspond to 3 exons harboring SNVs, ATM exon 17 (F858L), TP53 exon 11 (R248W), and BRAF exon 17 (V600E), and 2 exons harboring indels, KIT exon 9 and PIK3CA exon 2. The 5 exons with highest coverage correspond to 3 exons harboring SNVs, FGFR3 exon 7 (S249C), ANKRD11 exon 13 (S1790L), and ASXL2 exon 11 (E722Q), and 2 exons harboring indels, MSH3 exon 1 and SOX9 exon 3. Table 11 sumarizes the samples used for this evaluation.

Table 11: Summary of the Specimens and Allele Frequencies for LoD Confirmation

Tissue Type	Mutation Type	Mutation	Gene / Exon	cDNA Change	Amino Acid Change
Endometrial Cancer	SNV	Missense	TSC2 Exon 22	c.2395C>T	p.R799C
Endometrial Cancer	SNV	Missense	ASXL1 Exon 12	c.4236A>C	p.K1412N
Metastatic Breast Cancer	SNV	Missense	BAP1 Exon 16	c.295G>A	p.V99M
Colon Cancer	SNV	Missense	IGF1R Exon 3	c.668C>T	p.A223V
Endometrial Cancer	SNV	Missense	NOTCH2 Exon 33	c.3752G>A	p.R1251H
Pancreatic Cancer	SNV	Missense	TP53 Exon 11	c.742C>T	p.R248W
Colon Cancer	DEL	In-Frame Deletion	ARID1A Exon 1	c.780_782delCTC	p.S265del
Colon Cancer	DEL	Frame Shift Deletion	CCND3 Exon 4	c.373_374delTG	p.C125Hfs*34
Mullerian Ovarian Cancer	DEL	In-Frame Deletion	MAP3K1 Exon 13	c.2367delC	p.R790Gfs*9
Adenoid Cystic Carcinoma	INS	In-Frame Insertion	BCOR Exon 14	c.2936_2937insATTCAAATG	p.K978_F977ins*
Liver CA	INS	Frame Shift Insertion	TBX3 Exon 7	c.326_327insC	p.K110*
Endometrial Cancer	INS	Frame Shift Insertion	PPP2R1A Exon 10	c.1285_1286insA	p.L429Hfs*12
Pancreatic Ductal Adenocarcinoma	INS	In-Frame Insertion	MSH3 Exon 1	c.195_203dupGCCCCCAGC	p.P69_A62ins

Five serial dilutions were prepared using patient samples positive for the mutations listed above, where tumor samples were diluted with a previously sequenced, unmatched normal FFPE sample created from a well-characterized cell line. One replicate at each dilution was tested and the ability to detect the mutation of interest was measured.

The results of the dilution series established precedence for Omics Core to call variants down to the 5% mutant allele frequency. A fully ranked list of all variants by their expected allele frequencies is shown in Tables 12A-12J , which demonstrate the successful detection of all variants estimated at 3.75% and above.

Table 12A Limit of Detection–Part 1

SNV ATM Exon 17 (Sample 14)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.2572T>C	p.F858L	143	73	51.05%	Called
1:2			113	27	23.89%	Called
1:4			107	9	8.41%	Called
1:8			77	9	11.69%	Called
1:16			WT			

Table 12B

SNV TP53 Exon 11 (Sample 15)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.742C>T	p.R248W	279	35	12.54%	Called
1:2			268	24	8.96%	Called
1:4			237	10	4.22%	Called
1:8			WT			
1:16			WT			

Table 12C

SNV BRAF Exon 17 (Sample 16)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.1799T>A	p.V600E	83	25	30.12%	Called
1:2			95	16	16.84%	Called
1:4			80	8	10.00%	Called
1:8			WT			
1:16			WT			

Table 12D

INS KIT Exon 9 (Sample 17)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.1504_1509dup GCCTAT	p.Y503_A502ins	380	147	38.68%	Called
1:2			287	59	20.56%	Called
1:4			276	34	12.32%	Called
1:8			239	21	8.79%	Called
1:16			199	11	5.53%	Called

Table 12E

DEL PIK3CA Exon 2 (Sample 18)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.312_317del AGTAGG	p.V105_G106del	316	179	56.65%	Called
1:2			295	117	39.66%	Called
1:4			359	101	28.13%	Called
1:8			224	32	14.29%	Called
1:16			176	12	6.82%	Called

Table 12F

SNV FGFR3 Exon 7 (Sample 19)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.746C>G	p.S249C	832	175	21.03%	Called
1:2			662	63	9.52%	Called
1:4			657	34	5.18%	Called
1:8			698	18	2.58%	Called
1:16			WT			

Table 12G

SNV ANKRD11 Exon 13 (Sample 20)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.5369C>T	p.S1790L	904	84	9.29%	Called
1:2			707	17	2.40%	Called
1:4			WT			
1:8			WT			
1:16			WT			

Table 12H

SNV ASXL2 Exon 11 (Sample 21)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.2164G>C	p.E722Q	1068	47	4.40%	Called
1:2			1151	39	3.39%	Called
1:4			1385	27	1.95%	Called
1:8			1143	13	1.14%	Called
1:16			1399	14	1.00%	Called

Table 12I

INS MSH3 Exon 1 (Sample 22)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.195_203dupGC CCCCAGC	p.P69_A62ins	512	147	28.71%	Called
1:2			704	120	17.05%	Called
1:4			722	65	9.00%	Called
1:8			WT			
1:16			WT			

Table 12J

DEL SOX9 Exon 3 (Sample 23)						
Dilution	**cDNA Change**	**AA Change**	**DP**	**AD**	**VF**	**Result**
Neat	c.1162delG	p.E388Sfs*15	686	182	26.53%	Called
1:2			877	67	7.64%	Called
1:4			873	24	2.75%	Called
1:8			939	13	1.38%	Called
1:16			WT			

Part 2: Confirmation of the LoD. Based on the LoD dilution series, a total of 5 replicates were tested for each of the 3 deletions, 4 insertions, and 6 SNVs at 5% mutant allele frequency. Table 13 summarizes the specimens and genes assessed for the LoD confirmation study. In the LoD confirmation study, replicates for all variants had 100% positive call rates except for one replicate for a SNV on BAP1 exon 16. This replicate was detected but failed to meet the variant confidence threshold established for reporting. A summary of the data per variant evaluated, including the mutation, the gene and exon, range for DP, AD, MAF, NormDP, and positive call rate is shown in Table 13. The results of the LoD establishment and confirmation study demonstrate Omics Core's ability to detect and reliably call each variant class at the 5% mutant allele frequency with a success rate of ≥ 95%. SNVs were called at 96.7% (82.8%-99.9% CI), insertions at 100% (83.2%-100.0% CI) and deletions at 100.0% (78.2%-100.0% CI).

Table 13: Limit of Detection–Part 2

Type	Mutation	Gene / Exon	Range DP	Range AD	Range MAF	Range NormDP	Positive Call Rate
SNV	Missense Mutation c.295G>A p.V99M	BAP1 Exon 16	250-832	7-53	0.021-0.078	0.83-1.63	80%
SNV	Missense Mutation c.668C>T p.A223V	IGF1R Exon 3	744-1356	40-55	0.041-0.068	2.32-2.66	100%
SNV	Missense Mutation c.742C>T p.R248W	TP53 Exon 11	216-284	12-15	0.051-0.065	0.52-0.58	100%
SNV	Missense Mutation c.4236A>C p.K1412N	ASXL1 Exon 12	625-975	14-38	0.02-0.046	1.89-2.01	100%
SNV	Missense Mutation c.2395C>T p.R799C	TSC2 Exon 22	439-825	11-32	0.013-0.049	1.39-1.73	100%
SNV	Missense Mutation c.3752G>A p.R1251H	NOTCH2 Exon 33	576-820	26-53	0.045-0.067	1.28-1.63	100%
DEL	In-Frame Deletion c.780_782delCTC p.S265del	ARID1A Exon 1	851-1622	71-104	0.06-0.106	2.58-2.94	100%
DEL	Frame Shift Deletion c.373_374delTG p.C125Hfs*34	CCND3 Exon 4	358-758	36-52	0.068-0.105	1.03-1.24	100%
DEL	Frame Shift Deletion c.2367delC p.R790Gfs*9	MAP3K1 Exon 13	168-214	14-29	0.083-0.163	0.47-0.61	100%
INS	In-Frame Insertion c.2936_2937insATTCAAATG p.K978_F977ins*	BCOR Exon 14	574-787	33-52	0.056-0.071	1.65-1.82	100%

Type	Mutation	Gene / Exon	Range DP	Range AD	Range MAF	Range NormDP	Positive Call Rate
INS	Frame Shift Insertion c.1285_1286insA p.L429Hfs*12	PPP2R1A Exon 10	316-910	37-140	0.083-0.154	0.82-2.01	100%
INS	In-Frame Insertion c.195_203dupGCCCCCAGC p.P69_A62ins	MSH3 Exon 1	460-662	33-93	0.072-0.16	1.25-1.38	100%
INS	Frame Shift Insertion c.326_327insC p.K110*	TBX3 Exon 7	666-1125	54-118	0.058-0.105	2.12-2.61	100%

ii. LoD – Tumor Mutation Burden (TMB):

Ten (10) FFPE tumor specimens with a tumor purity of ≤ 20%, as assessed by a board certified pathologist, were selected for TMB analysis. Five (5) replicates of each tumor specimen were analyzed by the Omics Core assay and TMB was calculated for all fifty (50) replicates surveyed by the Omics Core assay.

Table 14 demonstrates consistent reproducibility and repeatability of the Omics Core TMB assay with minimum tumor content representing tumor purities as low as 10%. The Omics Core assay will evaluate and report TMB rates for clinical samples with tumor purities ≥ 20%. All samples evaluated in the precision analyses had a %CV < 10%.

Table 14. TMB Analysis with Low Tumor Purity Samples

Tumor Type	Tumor Purity	Mean TMB Value	# of Valid Results	SD	%CV	Average Mean Exon Coverage
Gastric Adenocarcinoma	10%	3.2	5	0.11	3.30	833.46
Renal Cell Cancer	10%	2.36	5	0.03	1.28	807.69
Colon Cancer	10%	5.33	5	0.05	0.97	809.08
Lung Cancer	15%	6.06	5	0.13	2.11	901.64
Adenocarcinoma of Bladder	15%	2.82	5	0.03	1.23	847.58
Neoplasm of Head of Pancreas	15-20%	2.36	5	0.1	4.26	798.86
Gallbladder Adenocarcinoma	15-20%	5.41	5	0.13	2.31	898.5
Malignant Neoplasm of Border of Tongue	20%	3.05	5	0.17	5.52	925.04
Metastatic Neuroendocrine Carcinoma	20%	1.03	5	0.07	6.49	850.07
Breast Cancer	20%	8.15	5	0.12	1.50	828.07

iii. DNA-Input:

A DNA Input study demonstrated the analytical performance of the Omics Core assay across a range of DNA input (50–300 ng) in representative FFPE tumor types. A total of 11 FFPE clinical samples and one commercial cell line were assessed at six different DNA inputs (50, 100, 150, 200, 250, and 300 ng).

Analytical performance was assessed for mutations identified in the precision study. The positive call rate was assessed for each DNA input from 11 FFPE clinical samples and a commercial cell line. A summary assessment of analytical performance for each DNA input range is shown in Table 14.

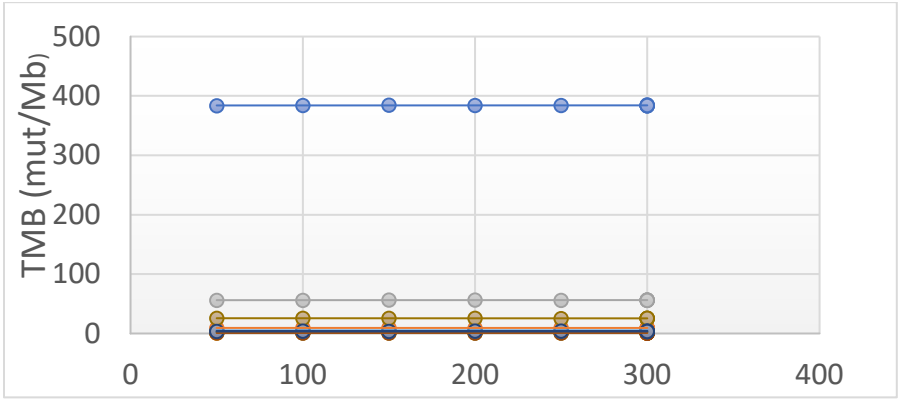
Table 14: Sequencing Failures Relative to DNA Input

DNA Input	Success (%)	Failure (%)
300 ng	98.40	1.60
250 ng	97.87	2.13
200 ng	97.87	2.13
150 ng	97.49	2.51
100 ng	97.68	2.32
50 ng	98.07	1.93

iv. DNA Input – TMB:

TMB was measured using 11 undiluted samples across a range of DNA inputs (50 - 300 ng) (Table 15). A total of 110 sequencing runs demonstrated a %CV of <10% for all 11 samples assessed (10 replicates each) (Results shown in Table 10). Figure 4 presents TMB calculation for each of the 11 samples at the respective DNA inputs in a scatter plot showing TMB score as a function of DNA input. The data demonstrate that the Omics Core TMB calculation is consistent when evaluating multiple tumor types across varying DNA inputs.

Figure 4: TMB vs DNA Input for 11 Representative FFPE Samples



c. Linearity /assay reportable range:

Not applicable

d. Traceability, Stability, Expected Values (controls, calibrators, or methods):

i. Traceability:

The Omics Core is not traceable to any known standard. Controls and quality metrics are described in the device description section.

ii. Stability:

Reagent stability is based on manufacturer expiration dating and is supported through NantHealth verification protocols. Stability of the reagents is also monitored through the use of the quality metrics and consistent output for controls.

iii. Expected values:

The laboratory has established baseline quality control values and metrics for the reporting of somatic variants claimed by this assay.

e. Analytical Specificity

High analytical specificity is maintained by paired/tumor matched normal sequencing and was established during assay optimization.

Interference: The Omics Core assay is designed to minimize interference from exogenous sources. The invalid sequencing rates across 42 tumor types supports that interference is minimized.

f. Assay Cut-off

The Omics Core does not report mutations below 2% variant allele frequency. Mutations included in the calculation of TMB must be present at 5% allele frequency or greater.

g. Comparison Studies

i. Method Comparison-SNVs, Insertions and Deletions

The accuracy of the Omics Core assay was assessed by comparing Omics Core results to results obtained from the orthogonal method. A total of 401 FFPE tumor samples from clinical cases across a diverse set of cancers were used in this analysis, representing mutations covering 2,634 SNVs, 125 small insertions, and 313 small deletions.

The results of the comparison study demonstrate the accuracy of the Omics Core assay by successfully detecting mutations in 401 out of the 401 samples, representing an accuracy of 100% (99.08-100.00% CI). There were 3 false positives across 2 genes and 8 false negatives across 8 genes. A total of 3072

unique mutations in 401 FFPE tumor specimens representing 1835 exons in 471 genes were tested and are listed in Appendix 2.

Omics Core accuracy study[2] included 2634 unique SNVs demonstrated a PPA of 99.76% (99.50-99.90% CI), PPV of 99.93% (99.75-99.99% CI); 125 unique small insertions (ranging from 1 bp to 12 bp) demonstrated a PPA of 100% (97.20-100.00% CI), PPV of 100% (97.20-100.00% CI); and 313 unique small deletions (ranging from 1 bp to 61 bp) demonstrated a PPA of 99.71% (98.38-99.99% CI), PPV of 99.71% (98.38-99.99% CI). To adjust for sampling prevalence, adjusted PPA was provided (All data 100% except where indicated). Calculating adjusted PPA requires estimation of the prevalence of positives which was estimated for the set of all variants tested, as well as a per-gene for the set of variants in a singe gene. Results are shown in Tables 15A-15C below separated by SNVs, insertions and deletions.

Table 15A: Percent Positive Agreement for SNVs by Gene

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
ABL1	4	7	5	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
ACVR1	3	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
AGO2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
AKT1	4	5	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
AKT2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
AKT3	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
ALK	12	17	15	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
ALOX12B	6	6	5	100.00% (54.07%-100.00%)	85.71% (42.13%-99.64%) 76.79% (52.19%-100.00%)
ANKRD11	4	22	14	100.00% (84.56%-100.00%)	100.00% (84.56%-100.00%)
APC	9	51	44	100.00% (93.94%-100.00%)	100.00% (93.94%-100.00%)
AR	4	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)

[2] Performance may be overestimated because specimens were selected based on Omics core original results and then confirmed by the orthogonal methods (i.e., the specimen set may lack challenging specimens).

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
ARAF	4	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
ARID1A	14	31	29	100.00% (88.78%-100.00%)	96.88% (83.78%-99.92%) 94.20% (84.19%-100.00%)
ARID1B	7	19	15	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
ARID2	10	15	13	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
ARID5B	3	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
ASXL1	4	13	11	100.00% (75.29%-100.00%)	100.00% (75.29%-100.00%)
ASXL2	6	15	5	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
ATM	20	30	24	100.00% (88.78%-100.00%)	100.00% (88.78%-100.00%)
ATR	10	12	7	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
ATRX	11	15	13	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
AURKA	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
AXIN1	4	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
AXIN2	5	6	4	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
AXL	6	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
B2M	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BAP1	9	12	11	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
BARD1	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BCL10	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BCL2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
BCL2L11	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BCL6	3	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
BCOR	5	15	11	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
BIRC3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
BLM	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BMPR1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BRAF	3	4	18	100.00% (81.47%-100.00%)	100.00% (81.47%-100.00%)
BRCA1	6	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
BRCA2	8	12	12	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
BRD4	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
BRIP1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BTK	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
CARD11	4	7	5	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
CARM1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CASP8	5	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
CBFB	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
CBL	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
CCND1	2	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
CCND2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
CCND3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CCNE1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CD274	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CD276	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CD79A	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CD79B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDC73	4	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
CDH1	9	12	12	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
CDK12	5	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
CDK4	0	0	0	N/A	N/A
CDK8	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDKN1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDKN1B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDKN2B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDKN2C	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CEBPA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CHEK1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CHEK2	4	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
CIC	4	5	3	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
CREBBP	12	21	17	100.00% (83.89%-100.00%)	100.00% (83.89%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
CRKL	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CSDE1	7	8	6	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
CSF1R	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
CSF3R	5	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
CTCF	7	10	10	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
CTNNB1	10	21	21	100.00% (86.77%-100.00%)	100.00% (86.77%-100.00%)
CUL3	4	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
CXCR4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CYLD	6	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
CYSLTR2	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
DDR2	7	9	6	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
DICER1	7	10	7	100.00% (69.15%-100.00%)	90.91% (58.72%-99.77%) 89.65% (74.08%-100.00%)
DIS3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
DNAJB1	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
DNMT1	5	5	3	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
DNMT3A	7	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
DNMT3B	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
DOT1L	6	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
DROSHA	8	9	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
DUSP4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
E2F3	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EED	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EGFL7	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EGFR	8	8	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
EIF1AX	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EIF4A2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ELF3	2	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
EP300	10	16	12	100.00% (79.41%-100.00%)	100.00% (79.41%-100.00%)
EPAS1	1	2	1	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EPCAM	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EPHA3	7	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
EPHA5	9	14	11	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
EPHA7	8	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
EPHB1	7	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
ERBB2	8	17	21	100.00% (84.56%-100.00%)	100.00% (84.56%-100.00%)
ERBB3	3	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
ERBB4	14	18	14	100.00% (81.47%-100.00%)	100.00% (81.47%-100.00%)
ERCC2	6	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
ERCC3	4	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
ERCC4	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
ERCC5	2	4	2	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
ERF	4	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
ERG	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
ESR1	5	9	11	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
ETV1	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
ETV6	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EZH1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EZH2	5	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
FAM46C	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FAM58A	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FANCA	10	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
FANCC	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FAT1	12	27	15	100.00% (87.23%-100.00%)	100.00% (87.23%-100.00%)
FBXW7	6	14	14	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
FGF19	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FGF3	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
FGF4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
FGFR1	5	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
FGFR2	5	7	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
FGFR3	5	6	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
FGFR4	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
FH	3	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
FLCN	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
FLT1	10	10	10	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
FLT3	8	8	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
FLT4	7	14	11	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
FOXA1	1	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
FOXL2	1	7	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
FOXO1	2	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
FOXP1	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
FUBP1	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
FYN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
GATA1	3	4	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
GATA2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
GATA3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
GLI1	4	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
GNA11	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
GNAS	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
GPS2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
GRIN2A	10	25	21	100.00% (86.28%-100.00%)	100.00% (86.28%-100.00%)
GSK3B	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
H3F3A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
H3F3B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
H3F3C	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
HGF	13	14	14	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
HIST1H1C	1	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
HIST1H3A	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
HIST1H3B	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
HIST1H3C	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
HIST1H3D	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
HIST1H3H	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
HIST1H3I	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
HIST1H3J	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
HNF1A	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
HOXB13	2	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
HRAS	3	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
ICOSLG	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
ID3	1	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
IDH1	1	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
IFNGR1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
IGF1R	5	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
IGF2	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
IKBKE	3	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
IKZF1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
IL10	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
IL7R	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
INHA	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
INHBA	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
INPP4A	6	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
INPP4B	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
INPPL1	8	10	10	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
INSR	3	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
IRF4	4	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
IRS1	1	2	1	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
JAK1	6	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
JAK2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
JAK3	7	7	6	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
JUN	1	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
KDM5A	8	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
KDM5C	6	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
KDM6A	7	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
KDR	10	12	11	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
KEAP1	4	12	11	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
KIT	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
KLF4	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
KMT2A	8	20	14	100.00% (83.16%-100.00%)	100.00% (83.16%-100.00%)
KMT2B	10	20	14	100.00% (83.16%-100.00%)	100.00% (83.16%-100.00%)
KMT2C	17	36	32	100.00% (90.26%-100.00%)	97.30% (85.84%-99.93%)
KMT2D	22	41	30	100.00% (91.40%-100.00%)	100.00% (91.40%-100.00%)
KRAS	5	17	78	97.53% (91.36%-99.70%)	100.00% (95.44%-100.00%)
LATS1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
LATS2	4	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
LMO1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
LYN	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MALT1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MAP2K2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MAP2K4	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MAP3K1	6	7	5	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
MAP3K13	5	14	14	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
MAP3K14	7	7	6	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
MAPK1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MAPK3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MAPKAP1	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
MAX	3	5	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
MCL1	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MDM2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MDM4	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MED12	15	17	14	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
MEF2B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MEN1	3	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
MET	7	13	11	100.00% (75.29%-100.00%)	100.00% (75.29%-100.00%)
MGA	9	15	15	100.00% (79.41%-100.00%)	94.12% (71.31%-99.85%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
MITF	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
MLH1	6	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
MPL	3	3	2	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MRE11A	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MSH2	6	6	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
MSH3	3	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
MSH6	6	14	10	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
MSI1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MSI2	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MST1	0	0	0	N/A	N/A
MST1R	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
MTOR	8	11	8	100.00% (71.51%-100.00%)	100.00% (71.51%-100.00%)
MUTYH	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
MYC	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MYCN	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MYD88	2	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
MYOD1	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
NBN	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
NCOA3	6	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
NCOR1	12	19	14	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
NEGR1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
NF1	11	13	12	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)
NF2	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
NFE2L2	3	6	4	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
NFKBIA	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NKX2-1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NKX3-1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NOTCH 1	12	18	18	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
NOTCH 2	10	17	12	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
NOTCH 3	8	12	11	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
NPM1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NRAS	2	7	7	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
NSD1	8	22	19	100.00% (84.56%-100.00%)	100.00% (84.56%-100.00%)
NTHL1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NTRK1	4	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
NTRK2	4	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
NTRK3	7	11	11	100.00% (71.51%-100.00%)	100.00% (71.51%-100.00%)
NUF2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NUP93	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
PAK1	3	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
PAK7	5	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
PALB2	5	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
PARK2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PARP1	7	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
PAX5	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PBRM1	11	13	12	100.00% (75.29%-100.00%)	100.00% (75.29%-100.00%)
PDCD1	3	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
PDCD1LG2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PDGFRA	6	11	10	100.00% (71.51%-100.00%)	100.00% (71.51%-100.00%)
PDGFRB	8	11	11	100.00% (71.51%-100.00%)	100.00% (71.51%-100.00%)
PDPK1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PGR	4	8	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
PHOX2B	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PIK3C2G	6	7	6	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
PIK3C3	3	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
PIK3CA	7	25	59	100.00% (94.40%-100.00%)	100.00% (94.40%-100.00%)
PIK3CB	8	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
PIK3CD	3	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
PIK3CG	5	10	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
PIK3R1	7	9	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
PIK3R2	3	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
PIK3R3	2	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
PIM1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PLCG2	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
PLK2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
PMS1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PMS2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PNRC1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
POLD1	8	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
POLE	14	18	13	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
PPARG	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
PPM1D	2	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
PPP2R1A	6	8	9	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
PPP4R2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PRDM1	3	7	8	100.00% (59.04%-100.00%)	87.50% (47.35%-99.68%) 86.24% (67.46%-100.00%)
PRDM14	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
PREX2	12	14	11	100.00% (76.84%-100.00%)	100.00% (76.84%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
PRKAR1A	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
PRKCI	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
PRKD1	6	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
PTCH1	8	10	7	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
PTEN	8	17	15	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
PTP4A1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PTPN11	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PTPRD	11	16	15	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
PTPRS	12	17	11	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
PTPRT	11	19	17	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
RAB35	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
RAC2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RAD21	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RAD50	5	5	3	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
RAD51	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
RAD51C	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RAD54L	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
RAF1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
RASA1	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
RB1	9	12	9	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
RBM10	6	8	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
RECQL	4	4	1	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
RECQL4	6	8	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
REL	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RET	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
RFWD2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
RHOA	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
RICTOR	6	8	7	100.00% (63.06%-100.00%)	88.89% (51.75%-99.72%) 87.63% (70.03%-100.00%)
RIT1	3	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
RNF43	5	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
ROS1	12	15	13	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
RPS6KA4	3	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
RPS6KB2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RPTOR	5	9	10	100.00% (69.15%-100.00%)	100.00% (69.15%-100.00%)
RRAGC	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RRAS	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RRAS2	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
RTEL1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
RUNX1	3	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
RXRA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RYBP	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SDHA	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SDHAF2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SESN1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SESN3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SETD2	7	15	12	100.00% (78.20%-100.00%)	100.00% (78.20%-100.00%)
SF3B1	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
SH2B3	5	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
SH2D1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SHQ1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SLX4	3	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
SMAD2	5	6	5	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
SMAD3	5	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
SMAD4	8	16	17	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
SMARCA4	13	19	13	100.00% (82.35%-100.00%)	100.00% (82.35%-100.00%)
SMARCB1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SMARCD1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
SMO	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SMYD3	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SOCS1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SOS1	4	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
SOX17	2	7	7	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
SOX2	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
SOX9	2	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
SPEN	5	17	12	100.00% (80.49%-100.00%)	100.00% (80.49%-100.00%)
SPOP	6	11	12	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
SPRED1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SRC	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SRSF2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
STAG2	7	7	6	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
STAT3	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
STAT5A	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
STAT5B	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
STK11	4	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
STK40	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SUFU	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
SUZ12	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SYK	3	4	2	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
TBX3	4	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
TCF3	7	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
TCF7L2	5	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
TEK	6	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
TERT	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
TET1	5	12	9	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
TET2	4	13	13	100.00% (75.29%-100.00%)	100.00% (75.29%-100.00%)
TGFBR1	4	7	6	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
TGFBR2	3	7	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
TMEM127	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TMPRSS2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
TNFAIP3	6	8	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
TNFRSF14	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
TOP1	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
TP53	10	105	154	100.00% (97.83%-100.00%)	100.00% (97.83%-100.00%)
TP53BP1	7	12	10	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
TP63	6	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
TRAF2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
TRAF7	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
TSC1	7	7	8	100.00% (63.06%-100.00%)	100.00% (63.06%-100.00%)
TSC2	8	12	10	100.00% (75.29%-100.00%)	100.00% (75.29%-100.00%)
TSHR	2	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
UPF1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
VEGFA	0	0	0	N/A	N/A
VHL	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
VTCN1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
WHSC1	5	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
WT1	2	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
WWTR1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
XIAP	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
XPO1	4	4	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
YES1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ZFHX3	6	21	20	100.00% (84.56%-100.00%)	100.00% (84.56%-100.00%)

Table 15B: Percent Positive Agreement for Insertions by Gene

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI)
APC	2	9	12	100.00% (73.54%-100.00%)	100.00% (73.54%-100.00%)
ARID1A	8	9	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
ARID2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ASXL1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ATM	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ATRX	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
AXIN2	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
BBC3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BCL2L1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BMPR1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BRCA1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BRCA2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CALR	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CBFB	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CBL	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CCND1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CCND3	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CDH1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDK12	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI)
CDKN1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CREBBP	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CSF3R	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CTNNB1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ELF3	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
EPHA3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ERBB2	1	1	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ERF	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FAT1	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
FBXW7	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FOXA1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FOXO1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
GATA3	1	3	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
IRF4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
IRS1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
JAK1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
JAK2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
KDM6A	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
KIT	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
KMT2C	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI)
KMT2D	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MSH2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MST1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NBN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NCOR1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NF1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NFKBIA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NKX2-1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PIK3R1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PPP2R1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PTEN	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
RAD21	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RAD51	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RASA1	3	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
RB1	4	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
RECQL4	0	0	0	N/A	N/A
RNF43	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
RUNX1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SETD2	2	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SMAD3	0	0	0	N/A	N/A
SMAD4	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI)
SOX17	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
SOX9	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SPEN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SPRED1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
STK11	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SUFU	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TBX3	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
TCF7L2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TP53	4	6	6	100.00% (54.07%-100.00%)	100.00% (54.07%-100.00%)
TSC1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TSC2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Table 15C: Percent Positive Agreement for Deletions by Gene

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
AGO2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ANKRD11	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
APC	2	26	23	100.00% (87.66%-100.00%)	100.00% (87.66%-100.00%)
ARID1A	9	20	19	100.00% (85.18%-100.00%)	100.00% (85.18%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
ARID1B	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
ARID2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ASXL1	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ASXL2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ATM	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ATRX	3	7	7	100.00% (59.04%-100.00%)	100.00% (59.04%-100.00%)
AXIN2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
B2M	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BAP1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BARD1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BCL2L11	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BCOR	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
BIRC3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BRAF	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
BRCA2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CASP8	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CBFB	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
CCND3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CD79A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CDH1	4	4	4	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
CDK12	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
CDKN1A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CIC	3	3	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
CREBBP	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
CSDE1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CSF3R	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CTCF	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CTNNB1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
CUL3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
DDR2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
DICER1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
DNAJB1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
DROSHA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
EGFR	2	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
ELF3	4	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)
EP300	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
EPCAM	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
EPHB1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ERBB2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ERCC4	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
ERF	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ERG	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ERRFI1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ETV1	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
EZH2	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FAT1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FBXW7	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FLT3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
FOXA1	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
FOXP1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
HIST1H3J	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
HNF1A	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
INPP4A	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
INPPL1	3	4	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
IRS1	1	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
JAK1	1	1	5	100.00% (39.76%-100.00%)	80.00% (28.36%-99.49%) 23.34% (9.15%-100.00%)
KDM5A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
KDM5C	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
KDM6A	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
KEAP1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
KIT	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
KMT2A	2	2	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
KMT2B	1	1	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
KMT2C	6	9	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
KMT2D	5	9	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
LYN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MAP3K1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MAX	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MDM2	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
MEN1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MGA	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
MLH1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MRE11A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MSH3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
MSH6	1	1	5	80.00% (28.36%-99.49%)	100.00% (39.76%-100.00%)
MTOR	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NBN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NCOR1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
NF1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NOTCH1	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NOTCH3	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
NSD1	4	4	3	100.00% (39.76%-100.00%)	100.00% (39.76%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
PAK7	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PALB2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PBRM1	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
PDCD1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PGR	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PIK3CA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PIK3CG	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
PIK3R1	5	9	9	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
PIK3R3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PMS1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
POLD1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PPARG	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PPM1D	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
PTEN	2	4	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
PTPRS	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RAD51B	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RB1	4	5	5	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)

Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
RECQL4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RNF43	3	5	8	100.00% (66.37%-100.00%)	100.00% (66.37%-100.00%)
RUNX1	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
RXRA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
RYBP	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SESN2	1	1	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
SETD2	3	3	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SLX4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SMAD3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SMAD4	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
SMARCA4	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SOX17	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
SOX9	2	3	2	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
SPEN	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
SPRED1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
STAG2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
STAT5A	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)

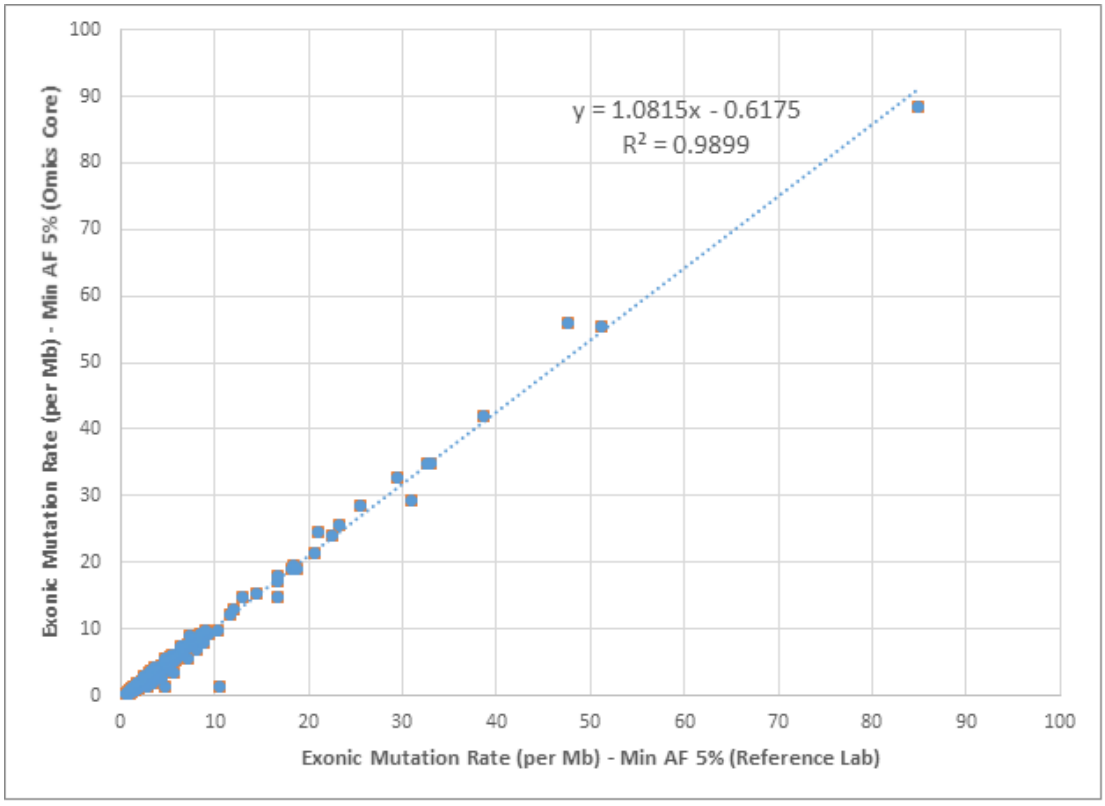
Gene	Number of Exons	Number of Unique Mutations	Number of Samples	PPV (95% CI)	PPA (95% CI) Adjusted PPA (95% CI)
STK11	1	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
TBX3	4	5	4	100.00% (47.82%-100.00%)	100.00% (47.82%-100.00%)
TCF7L2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TGFBR2	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
TNFAIP3	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
TP53	8	26	25	100.00% (87.23%-100.00%)	100.00% (87.23%-100.00%)
TSC2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
VEGFA	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
VHL	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)
WHSC1	1	1	3	100.00% (29.24%-100.00%)	100.00% (29.24%-100.00%)
XRCC2	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
YAP1	1	1	1	100.00% (2.50%-100.00%)	100.00% (2.50%-100.00%)
ZFHX3	2	2	2	100.00% (15.81%-100.00%)	100.00% (15.81%-100.00%)

ii. Accuracy (Method Comparison- Tumor Mutation Burden (TMB)

Tumor mutation burden (TMB) by Omics Core is reported as an estimate of mutation rate by counting all somatic, synonymous and non-synonymous variants detected in gene coding regions and dividing by the approximate size of the exome (33.7 Mb). TMB is reported as mutations per megabase (mut/Mb) unit. Mutations included in the calculation of TMB must be present at 5% allele frequency or greater after direct comparison of tumor

DNA with matched normal DNA. TMB accuracy was assessed by comparing the TMB rates generated by Omics Core and the orthogonal method. A linear regression was performed to calculate slope, intercept, and correlation between samples. This analysis identified a number of samples with higher than expected mutation rates after sequencing and variant calling by a Reference Call Lab. Further analysis revealed 58 of the 401 samples had homopolymer insertions (runs of A, C, G, or T with a minimum length of 4 bases, e.g. NF8:c.450_451insAAAAAAAAAAAAAA) unique to the GENEWIZ sample at rates higher than expected (> 10%), indicating a potential sample quality issue in the extended exome beyond the 468 genes of interest being used for reporting on Omics Core. The resulting 343 samples were then used to compare, showing higher concordance between Omics Core and GENEWIZ, with R2 = 0.9778. NantHealth excluded samples under the 250X exome coverage target designed in the study and observed a higher correlation between both sets of samples, with R2 = 0.9986. A scatter plot demonstrating the high correlation (R^2 = 0.9899) between Omics Core and the orthogonal method, is presented in Figure 5.

Figure 5: Omics Core Exonic Mutation Rate per Megabase Using Orthogonal Method Variant Calls



The following tables (Tables 16A-16D) provide a comparison of whole exome calls between the samples sequenced by Omics Core and the Reference Lab used in the final TMB analysis. In order to account for the lower targeted depth and lack of matched-normal in the Reference Lab callset, the following filters were used to exclude calls made by either Omics Core or the Reference Lab: AF < 5%, AD <= 2 reads, present in Matched-

Normal (using matched-normal germline sequencing data as determined by Omics Core), and % of MQ0 reads > 50%. The calls that remained for Omics Core or the Reference Lab were classified as "Positive" calls, while calls not made by one platform (but called by the other) were classified as "Negative" if the position was within a region targeted by the platform's exome kit (i.e., had sequencing data covering the position) or "No Call" if the position was not (i.e. no sequencing data covered the position). The sets of Positive, Negative, and No Call variants were then split by variant type (All variants, SNVs, Insertions, and Deletions) and intersected to determine the numbers in the following tables (16A- 16D) and calculated the PPA, NPA, and their respective confidence intervals.

Table 16A: TMB Accuracy – All Variants

ALL Variants	Reference Lab Positive Call	Reference Lab Negative Call	Reference Lab No Call
Omics Core Positive Call	46767	9104	1891
Omics Core Negative Call	1381	7636598912	1148979849
Omics Core No Call	25828	1078107398	676340898066
PPA (%)	97.14% (96.98%- 97.28% CI)		
NPA (%)	100.00% (100.00%- 100.00% CI)		

Table 16B: TMB Accuracy – SNV

SNV	Reference Lab Positive Call	Reference Lab Negative Call	Reference Lab No Call
Omics Core Positive Call	44637	8536	1796
Omics Core Negative Call	1029	7636601962	1148979944
Omics Core No Call	18041	1078115185	676340898066
PPA (%)	97.75 (97.61%-97.88% CI)		
NPA (%)	100.00% (100.00%- 100.00% CI)		

Table 16C: TMB Accuracy – INSERTIONS

INS	Reference Lab Positive Call	Reference Lab Negative Call	Reference Lab No Call
Omics Core Positive Call	487	119	29
Omics Core Negative Call	79	7636655479	1148981711
Omics Core No Call	6118	1078127108	676340898066
PPA (%)	86.04% (82.91%-88.79% CI)		
NPA (%)	100.00% (100.00%- 100.00% CI)		

Table 16D: TMB Accuracy – DELETIONS

DEL	Reference Lab Positive Call	Reference Lab Negative Call	Reference Lab No Call
Omics Core Positive Call	1643	449	66
Omics Core Negative Call	273	7636653799	1148981674
Omics Core No Call	1669	1078131557	676340898066
PPA (%)	85.75% (84.11%-87.29% CI)		
NPA (%)	100.00% (100.00%- 100.00% CI)		

iii. Supplemental Method Comparison Study for Wild-type Calls

A supplemental analysis was conducted to assess accuracy for a set of 32 hotspot variant positions from 12 genes within all samples. Omics Core results for all variant positions were compared to results obtained with the orthogonal method. Within the 401 specimens, there were 220 mutations across all samples and 12,612 wild-type calls. Discordant variants included 1 mutation observed in Omics Core and 1 mutation observed in the orthogonal method. The supplemental assay, which analyzed 220 positive mutations and 12,612 wild-type calls, demonstrated a PPA of 99.54% (97.48-99.99% CI) and NPA of 99.99% (99.99-100.00% CI).

4. <u>Clinical Performance:</u>

The Omics Core assay is a qualitative in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed paraffin-embedded tumor tissue matched with a

normal blood specimen from the patient with solid malignant neoplasms to detect tumor gene alterations in a broad multi gene panel. The genes in the panel include those that play a role in cancer pathogenesis and tumor suppression, or for clinical or mechanistic information of relevance in the management of cancer patients. The assay reports mutations under two categories: “Cancer Mutations with Evidence of Clinical Significance” and “Cancer Mutations with Potential Clinical Significance” consistent with the intended use clinical settings. Mutations with evidence of clinical significance are represented in professional guidelines as established by consensus opinion of experts in the healthcare community.

The NantHealth Omics Core report uses structured information stored within the Omics Core curation database, a manually curated knowledge base that includes biologic, clinical and therapeutic information curated from professional guidelines and recommendations, therapeutic labeling, disease specific expert and advocacy group recommendations, and medical literature. Classification criteria were developed by Omics Core to communicate the level of clinical evidence available for individual mutations including TMB in the test report. The mutations are reported under the two categories (i.e., “Cancer Mutations with Evidence of Clinical Significance” and “Cancer Mutations with Potential Clinical Significance”) based on pre-specified curtation classification criteria.NantHealth periodically updates Omics Core through the review of new information available.

5. **Clinical cut-off:**

Not applicable

6. **Expected values /Reference range:**

Not applicable

N. Instrument Name:

Illumina NovaSeq™ 6000 Sequencing System (qualified by NantHealth)

O. System Descriptions:

1. Modes of Operation:
The Illumina NovaSeq™ 6000 is a high throughput sequencing system using Sequencing-By-Synthesis chemistry.

2. Software:
FDA has reviewed applicant’s Hazard Analysis and software development processes for this line of product types:
Yes X or No ___

3. Level of Concern:
Moderate

4. Specimen Handling:
Refer to Device Description section above.

5. Calibration and Quality Controls:
Refer to Device Description section above.

P. Other Supportive Instrument Performance Characteristics Data Not Covered In The "Performance Characteristics" Section Above:

To support the continuous implementation of process improvements to the existing 468-gene Omics Core panel, protocols with specific procedures and acceptance criteria for modifications that could be anticipated at the time of submission were provided, reviewed by FDA, and cleared as part of this clearance. Future modifications by NantHealth for the specified types of changes below that are made in accordance with the applicable validation strategy and pre-specified success criteria do not require a new premarket notification. Significant changes such as adding new genes or variant types to the panel would require a new submission with appropriate validation.

Type of change	**Validation Strategy**	**Pre-specified success criteria**
New pre-analytical protocol, kits or reagents	Sequence at least 10 specimens with known mutations. Measure sequence coverage distribution, and call somatic mutations in all samples.	For cases sequenced to >500x, ensure that 95% of exons are covered to 100x or more. Concordance for known mutations should be >95%.
New library preparation protocol, kits, or reagents	Sequence at least 40 DNA specimens (tumor / normal pairs). Measure sequence coverage distribution, and call somatic mutations in all samples.	For cases sequenced to >500x, ensure that 95% of exons are covered to 100x or more. Concordance for calling somatic mutations with variant allele fraction >10% should be >98%.
Changes to probes for already analytically validated genes	Re-capture existing sequence libraries from at least 40 samples with new probes, sequence, and analyze.	For cases sequenced to >500x, ensure that 95% of exons in analytically validated genes are covered to 100x or

Type of change		Validation Strategy	Pre-specified success criteria
			more. Concordance for calling somatic mutations with variant allele fraction >10% should be >98%.
New sequencing instrument or reagents using similar chemistry and technology, and the sequence depth and read length are not changed from previous platform.		Re-sequence existing captured libraries from at least 3 runs, and call somatic mutations in all samples.	Sequence coverage distribution and GC bias across targeted regions should be within 5% of prior sequencing runs. Concordance for calling somatic mutations with variant allele fraction >10% should be >98%
Bioinformatics pipeline	Update to underlying annotation database or transcript isoforms	Reanalyze BAM files (sequencing data) from at least 40 samples. Compare variants calls between the clinical analysis results and the current modified results.	Confirm the changes do not change the variant call results. Confirm the annotations for the unaffected transcripts do not change. Confirm the annotations for the affected transcripts are modified as expected.
Bioinformatics pipeline	Update to data management system and system database	Reanalyze FASTQ files (raw sequencing reads) from at least 40 samples in production mode. Compare variants calls between the clinical analysis results and the current modified results.	Ensure that all previously called mutations are recovered and the variants in the database of results are concordant with the variants in the pipeline output files.
Bioinformatics pipeline	Modification to an existing component of the analysis pipeline (e.g., tool or algorithm) where the underlying algorithm or	Reanalyze FASTQ files (raw sequencing reads) from at least 40 samples. Compare variants calls between the clinical analysis results and the current modified results.	Ensure that all previously called mutations are recovered and that newly detected mutations can be explained by pipeline modifications.

Type of change		Validation Strategy	Pre-specified success criteria
	main parameter settings (e.g. minimal coverage/VAF threshold for SNV/indel calling) are not changed.		

Q. Proposed Labeling:

The labeling is sufficient and it satisfies the requirements of 21 CFR Parts 801 and 809, as applicable, and the special controls for this device type.

R. Patient Perspectives

This submission did not include specific information on patient perspectives for this device.

S. Conclusion:

The submitted information in this 510(k) notification supports the Indications For Use for Omics Core and demonstrates that the Omics Core assay is as safe and effective as the predicate device and therefore supports a substantial equivalence conclusion.

Appendix 1a : List of genes included on the Omics Core panel

Gene Name	Transcript ID
ABL1	NM_005157.5
ACVR1	NM_001111067.2
AGO2	NM_012154.3
AKT1	NM_001014431.1
AKT2	NM_001626.5
AKT3	NM_005465.4
ALK	NM_004304.4
ALOX12B	NM_001139.2
AMER1	NM_152424.3
ANKRD11	NM_013275.5
APC	NM_000038.5
AR	NM_000044.3
ARAF	NM_001654.4
ARID1A	NM_006015.4
ARID1B	NM_020732.3
ARID2	NM_152641.2
ARID5B	NM_032199.2
ASXL1	NM_015338.5
ASXL2	NM_018263.4
ATM	NM_000051.3
ATR	NM_001184.3
ATRX	NM_000489.4
AURKA	NM_003600.2
AURKB	NM_004217.3
AXIN1	NM_003502.3
AXIN2	NM_004655.3
AXL	NM_021913.4
B2M	NM_004048.2
BABAM1	NM_001033549.2
BAP1	NM_004656.3
BARD1	NM_000465.3
BBC3	NM_001127240.2

Gene Name	Transcript ID
BCL10	NM_003921.4
BCL2	NM_000633.2
BCL2L1	NM_138578.1
BCL2L11	NM_138621.4
BCL6	NM_001706.4
BCOR	NM_001123385.1
BIRC3	NM_182962.2
BLM	NM_000057.3
BMPR1A	NM_004329.2
BRAF	NM_004333.4
BRCA1	NM_007294.3
BRCA2	NM_000059.3
BRD4	NM_058243.2
BRIP1	NM_032043.2
BTK	NM_000061.2
CALR	NM_004343.3
CARD11	NM_032415.5
CARM1	NM_199141.1
CASP8	NM_001080125.1
CBFB	NM_022845.2
CBL	NM_005188.3
CCND1	NM_053056.2
CCND2	NM_001759.3
CCND3	NM_001760.4
CCNE1	NM_001238.2
CD274	NM_014143.3
CD276	NM_001024736.1
CD79A	NM_001783.3
CD79B	NM_001039933.1
CDC42	NM_001791.3
CDC73	NM_024529.4
CDH1	NM_004360.3

Gene Name	Transcript ID
CDK12	NM_016507.3
CDK4	NM_000075.3
CDK6	NM_001145306.1
CDK8	NM_001260.1
CDKN1A	NM_078467.2
CDKN1B	NM_004064.4
CDKN2A	NM_000077.4
CDKN2A	NM_058195.3
CDKN2B	NM_004936.3
CDKN2C	NM_078626.2
CEBPA	NM_004364.4
CENPA	NM_001809.3
CHEK1	NM_001274.5
CHEK2	NM_007194.3
CIC	NM_015125.4
CREBBP	NM_004380.2
CRKL	NM_005207.3
CRLF2	NM_022148.3
CSDE1	NM_001242891.1
CSF1R	NM_005211.3
CSF3R	NM_000760.3
CTCF	NM_006565.3
CTLA4	NM_005214.4
CTNNB1	NM_001904.3
CUL3	NM_003590.4
CXCR4	NM_003467.2
CYLD	NM_001042355.1
CYSLTR2	NM_020377.3
DAXX	NM_001141970.1
DCUN1D1	NM_020640.3
DDR2	NM_006182.2
DICER1	NM_030621.4
DIS3	NM_014953.4
DNAJB1	NM_006145.2

Gene Name	Transcript ID
DNMT1	NM_001379.2
DNMT3A	NM_022552.4
DNMT3B	NM_006892.3
DOT1L	NM_032482.2
DROSHA	NM_013235.4
DUSP4	NM_001394.6
E2F3	NM_001949.4
EED	NM_003797.4
EGFL7	NM_201446.2
EGFR	NM_005228.3
EIF1AX	NM_001412.3
EIF4A2	NM_001967.3
EIF4E	NM_001130678.1
ELF3	NM_004433.4
EP300	NM_001429.3
EPAS1	NM_001430.4
EPCAM	NM_002354.2
EPHA3	NM_005233.5
EPHA5	NM_004439.6
EPHA7	NM_004440.3
EPHB1	NM_004441.4
ERBB2	NM_004448.3
ERBB3	NM_001982.3
ERBB4	NM_005235.2
ERCC2	NM_000400.3
ERCC3	NM_000122.1
ERCC4	NM_005236.2
ERCC5	NM_000123.3
ERF	NM_006494.3
ERG	NM_182918.3
ERRFI1	NM_018948.3
ESR1	NM_001122740.1
ETV1	NM_001163147.1
ETV6	NM_001987.4

Gene Name	Transcript ID
EZH1	NM_001991.3
EZH2	NM_004456.4
FAM175A	NM_139076.2
FAM46C	NM_017709.3
FAM58A	NM_152274.4
FANCA	NM_000135.2
FANCC	NM_000136.2
FAT1	NM_005245.3
FBXW7	NM_033632.3
FGF19	NM_005117.2
FGF3	NM_005247.2
FGF4	NM_002007.2
FGFR1	NM_001174067.1
FGFR2	NM_000141.4
FGFR3	NM_000142.4
FGFR4	NM_213647.2
FH	NM_000143.3
FLCN	NM_144997.5
FLT1	NM_002019.4
FLT3	NM_004119.2
FLT4	NM_182925.4
FOXA1	NM_004496.3
FOXL2	NM_023067.3
FOXO1	NM_002015.3
FOXP1	NM_001244814.1
FUBP1	NM_003902.4
FYN	NM_153047.3
GATA1	NM_002049.3
GATA2	NM_032638.4
GATA3	NM_002051.2
GLI1	NM_005269.2
GNA11	NM_002067.4
GNAQ	NM_002072.4
GNAS	NM_000516.5

Gene Name	Transcript ID
GPS2	NM_004489.4
GREM1	NM_013372.6
GRIN2A	NM_001134407.2
GSK3B	NM_002093.3
H3F3A	NM_002107.4
H3F3B	NM_005324.4
H3F3C	NM_001013699.2
HGF	NM_000601.4
HIST1H1C	NM_005319.3
HIST1H2BD	NM_021063.3
HIST1H3A	NM_003529.2
HIST1H3B	NM_003537.3
HIST1H3C	NM_003531.2
HIST1H3D	NM_003530.4
HIST1H3E	NM_003532.2
HIST1H3F	NM_021018.2
HIST1H3G	NM_003534.2
HIST1H3H	NM_003536.2
HIST1H3I	NM_003533.2
HIST1H3J	NM_003535.2
HIST2H3C	NM_021059.2
HIST2H3D	NM_001123375.2
HIST3H3	NM_003493.2
HLA-A	NM_001242758.1
HLA-B	NM_005514.6
HNF1A	NM_000545.6
HOXB13	NM_006361.5
HRAS	NM_001130442.1
ICOSLG	NM_015259.5
ID3	NM_002167.4
IDH1	NM_005896.3
IDH2	NM_002168.3
IFNGR1	NM_000416.2
IGF1	NM_001111283.1

Gene Name	Transcript ID
IGF1R	NM_000875.4
IGF2	NM_001127598.2
IKBKE	NM_014002.3
IKZF1	NM_006060.5
IL10	NM_000572.2
IL7R	NM_002185.3
INHA	NM_002191.3
INHBA	NM_002192.2
INPP4A	NM_001134224.1
INPP4B	NM_001101669.1
INPPL1	NM_001567.3
INSR	NM_000208.2
IRF4	NM_002460.3
IRS1	NM_005544.2
IRS2	NM_003749.2
JAK1	NM_002227.2
JAK2	NM_004972.3
JAK3	NM_000215.3
JUN	NM_002228.3
KDM5A	NM_001042603.2
KDM5C	NM_004187.3
KDM6A	NM_021140.3
KDR	NM_002253.2
KEAP1	NM_203500.1
KIT	NM_000222.2
KLF4	NM_004235.4
KMT2A	NM_001197104.1
KMT2B	NM_014727.2
KMT2C	NM_170606.2
KMT2D	NM_003482.3
KMT5A	NM_020382.3
KNSTRN	NM_033286.3
KRAS	NM_033360.3
LATS1	NM_004690.3

Gene Name	Transcript ID
LATS2	NM_014572.2
LMO1	NM_002315.2
LYN	NM_002350.3
MALT1	NM_006785.3
MAP2K1	NM_002755.3
MAP2K2	NM_030662.3
MAP2K4	NM_003010.3
MAP3K1	NM_005921.1
MAP3K13	NM_004721.4
MAP3K14	NM_003954.4
MAPK1	NM_002745.4
MAPK3	NM_002746.2
MAPKAP1	NM_001006617.1
MAX	NM_002382.4
MCL1	NM_021960.4
MDC1	NM_014641.2
MDM2	NM_002392.5
MDM4	NM_002393.4
MED12	NM_005120.2
MEF2B	NM_001145785.1
MEN1	NM_000244.3
MET	NM_000245.2
MGA	NM_001164273.1
MITF	NM_198159.2
MLH1	NM_000249.3
MPL	NM_005373.2
MRE11A	NM_005591.3
MSH2	NM_000251.2
MSH3	NM_002439.4
MSH6	NM_000179.2
MSI1	NM_002442.3
MSI2	NM_138962.2
MST1	NM_020998.3
MST1R	NM_002447.2

Gene Name	Transcript ID
MTOR	NM_004958.3
MUTYH	NM_001128425.1
MYC	NM_002467.4
MYCL	NM_001033082.2
MYCN	NM_005378.5
MYD88	NM_002468.4
MYOD1	NM_002478.4
NBN	NM_002485.4
NCOA3	NM_181659.2
NCOR1	NM_006311.3
NEGR1	NM_173808.2
NF1	NM_001042492.2
NF2	NM_000268.3
NFE2L2	NM_006164.4
NFKBIA	NM_020529.2
NKX2-1	NM_001079668.2
NKX3-1	NM_006167.3
NOTCH1	NM_017617.3
NOTCH2	NM_024408.3
NOTCH3	NM_000435.2
NOTCH4	NM_004557.3
NPM1	NM_002520.6
NRAS	NM_002524.4
NSD1	NM_022455.4
NTHL1	NM_002528.5
NTRK1	NM_002529.3
NTRK2	NM_006180.4
NTRK3	NM_001012338.2
NUF2	NM_031423.3
NUP93	NM_014669.4
PAK1	NM_002576.4
PAK7	NM_177990.2
PALB2	NM_024675.3
PARK2	NM_004562.2

Gene Name	Transcript ID
PARP1	NM_001618.3
PAX5	NM_016734.2
PBRM1	NM_018313.4
PDCD1	NM_005018.2
PDCD1LG2	NM_025239.3
PDGFRA	NM_006206.4
PDGFRB	NM_002609.3
PDPK1	NM_002613.4
PGR	NM_000926.4
PHOX2B	NM_003924.3
PIK3C2G	NM_004570.5
PIK3C3	NM_002647.3
PIK3CA	NM_006218.2
PIK3CB	NM_006219.2
PIK3CD	NM_005026.3
PIK3CG	NM_002649.3
PIK3R1	NM_181523.2
PIK3R2	NM_005027.3
PIK3R3	NM_003629.3
PIM1	NM_002648.3
PLCG2	NM_002661.4
PLK2	NM_006622.3
PMAIP1	NM_021127.2
PMS1	NM_000534.4
PMS2	NM_000535.5
PNRC1	NM_006813.2
POLD1	NM_002691.3
POLE	NM_006231.3
PPARG	NM_015869.4
PPM1D	NM_003620.3
PPP2R1A	NM_014225.5
PPP4R2	NM_174907.2
PPP6C	NM_002721.4
PRDM1	NM_001198.3

Gene Name	Transcript ID
PRDM14	NM_024504.3
PREX2	NM_024870.2
PRKAR1A	NM_212471.2
PRKCI	NM_002740.5
PRKD1	NM_002742.2
PTCH1	NM_000264.3
PTEN	NM_000314.6
PTP4A1	NM_003463.4
PTPN11	NM_002834.3
PTPRD	NM_002839.3
PTPRS	NM_002850.3
PTPRT	NM_133170.3
RAB35	NM_006861.6
RAC1	NM_018890.3
RAC2	NM_002872.4
RAD21	NM_006265.2
RAD50	NM_005732.3
RAD51	NM_002875.4
RAD51B	NM_133509.3
RAD51C	NM_058216.2
RAD51D	NM_133629.2
RAD52	NM_134424.3
RAD54L	NM_001142548.1
RAF1	NM_002880.3
RARA	NM_000964.3
RASA1	NM_002890.2
RB1	NM_000321.2
RBM10	NM_001204468.1
RECQL	NM_032941.2
RECQL4	NM_004260.3
REL	NM_002908.3
RET	NM_020975.4
RFWD2	NM_022457.6
RHEB	NM_005614.3

Gene Name	Transcript ID
RHOA	NM_001664.2
RICTOR	NM_152756.4
RIT1	NM_006912.5
RNF43	NM_017763.5
ROS1	NM_002944.2
RPS6KA4	NM_003942.2
RPS6KB2	NM_003952.2
RPTOR	NM_020761.2
RRAGC	NM_022157.3
RRAS	NM_006270.4
RRAS2	NM_012250.5
RTEL1	NM_032957.4
RUNX1	NM_001754.4
RXRA	NM_002957.5
RYBP	NM_012234.6
SDHA	NM_004168.3
SDHAF2	NM_017841.2
SDHB	NM_003000.2
SDHC	NM_003001.3
SDHD	NM_003002.3
SESN1	NM_014454.2
SESN2	NM_031459.4
SESN3	NM_144665.3
SETD2	NM_014159.6
SF3B1	NM_012433.3
SH2B3	NM_005475.2
SH2D1A	NM_002351.4
SHOC2	NM_007373.3
SHQ1	NM_018130.2
SLX4	NM_032444.2
SMAD2	NM_001003652.3
SMAD3	NM_005902.3
SMAD4	NM_005359.5
SMARCA4	NM_003072.3

Gene Name	Transcript ID
SMARCB1	NM_003073.3
SMARCD1	NM_003076.4
SMO	NM_005631.4
SMYD3	NM_001167740.1
SOCS1	NM_003745.1
SOS1	NM_005633.3
SOX17	NM_022454.3
SOX2	NM_003106.3
SOX9	NM_000346.3
SPEN	NM_015001.2
SPOP	NM_001007228.1
SPRED1	NM_152594.2
SRC	NM_198291.2
SRSF2	NM_003016.4
STAG2	NM_001042749.2
STAT3	NM_139276.2
STAT5A	NM_003152.3
STAT5B	NM_012448.3
STK11	NM_000455.4
STK19	NM_004197.1
STK40	NM_032017.2
SUFU	NM_016169.3
SUZ12	NM_015355.2
SYK	NM_003177.6
TAP1	NM_000593.5
TAP2	NM_018833.2
TBX3	NM_016569.3
TCEB1	NM_005648.3
TCF3	NM_001136139.2
TCF7L2	NM_001146274.1
TEK	NM_000459.4
TERT	NM_198253.2
TET1	NM_030625.2

Gene Name	Transcript ID
TET2	NM_001127208.2
TGFBR1	NM_004612.3
TGFBR2	NM_001024847.2
TMEM127	NM_001193304.2
TMPRSS2	NM_001135099.1
TNFAIP3	NM_006290.3
TNFRSF14	NM_003820.3
TOP1	NM_003286.2
TP53	NM_000546.5
TP53BP1	NM_001141980.1
TP63	NM_003722.4
TRAF2	NM_021138.3
TRAF7	NM_032271.2
TSC1	NM_000368.4
TSC2	NM_000548.3
TSHR	NM_000369.2
U2AF1	NM_006758.2
UPF1	NM_002911.3
VEGFA	NM_001171623.1
VHL	NM_000551.3
VTCN1	NM_024626.3
WHSC1	NM_001042424.2
WHSC1L1	NM_023034.1
WT1	NM_024426.4
WWTR1	NM_001168280.1
XIAP	NM_001167.3
XPO1	NM_003400.3
XRCC2	NM_005431.1
YAP1	NM_001130145.2
YES1	NM_005433.3
ZFHX3	NM_006885.3

Appendix 1b : LIST OF EXONS EXCLUDED FROM REPORTING DUE TO CONSISTENTLY LOW COVERAGE

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
AMER1	NM_152424.3	X:63409758-63413166	1	1_3408	1_1136
ANKRD11	NM_013275.5	16:89334885-89335071	12	7807_7992	2603_2664
ATRX	NM_000489.4	X:76776265-76776394	33	7072_7200	2358_2400
ATRX	NM_000489.4	X:76814139-76814317	28	6327_6504	2109_2168
ATRX	NM_000489.4	X:76829714-76829823	27	6218_6326	2073_2109
ATRX	NM_000489.4	X:76849165-76849319	25	5957_6110	1986_2037
ATRX	NM_000489.4	X:76952064-76952192	4	243_370	81_124
ATRX	NM_000489.4	X:76972607-76972720	1	21_133	7_45
BCL2	NM_000633.2	18:60795857-60795992	2	586_720	196_240
BCOR	NM_001123385.1	X:39930889-39930943	4	2998_3051	1000_1017
BIRC3	NM_182962.2	11:102199627-102199676	5	1033_1081	345_361
BMPR1A	NM_004329.2	10:88676890-88677083	8	676_868	226_290
BRD4	NM_058243.2	19:15349187-15349256	19	4021_4089	1341_1363
BRD4	NM_058243.2	19:15349553-15349791	18	3783_4020	1261_1340
BRD4	NM_058243.2	19:15349869-15350075	17	3577_3782	1193_1261
BRD4	NM_058243.2	19:15350202-15350333	16	3446_3576	1149_1192
BRD4	NM_058243.2	19:15350469-15350632	15	3283_3445	1095_1149
BRD4	NM_058243.2	19:15350720-15350833	14	3170_3282	1057_1094
BRD4	NM_058243.2	19:15353710-15354298	13	2582_3169	861_1057
BRD4	NM_058243.2	19:15355041-15355411	12	2212_2581	738_861
BRD4	NM_058243.2	19:15355520-15355573	11	2159_2211	720_737
BTK	NM_000061.2	X:100615075-100615138	8	777_839	259_280
CARM1	NM_199141.1	19:10982378-10982598	0	1_220	1_74
CHEK2	NM_007194.3	22:29085122-29085203	13	1462_1542	488_514
CRLF2	NM_022148.3	X:1314893-1315014	5	647_767	216_256
CRLF2	NM_022148.3	X:1317418-1317581	4	484_646	162_216
CRLF2	NM_022148.3	X:1321271-1321405	3	350_483	117_161
CRLF2	NM_022148.3	X:1325325-1325492	2	183_349	61_117
CRLF2	NM_022148.3	X:1327698-1327801	1	80_182	27_61
CRLF2	NM_022148.3	X:1331448-1331527	0	1_79	1_27
DAXX	NM_001141970.1	6:33286519-33286579	7	2200_2259	734_753

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
DAXX	NM_001141970.1	6:33286773-33286996	6	1977_2199	659_733
DAXX	NM_001141970.1	6:33287156-33287631	5	1502_1976	501_659
DAXX	NM_001141970.1	6:33287787-33288001	4	1288_1501	430_501
DAXX	NM_001141970.1	6:33288156-33288368	3	1076_1287	359_429
DAXX	NM_001141970.1	6:33288512-33289344	2	244_1075	82_359
DAXX	NM_001141970.1	6:33289495-33289685	1	54_243	18_81
DAXX	NM_001141970.1	6:33290638-33290691	0	1_53	1_18
EIF4E	NM_001130678.1	4:99802178-99802293	6	600_714	200_238
GNAS	NM_000516.5	20:57466781-57466920	0	1_139	1_47
H3F3A	NM_002107.4	1:226259051-226259180	3	283_411	95_137
HIST2H3C	NM_021059.2	1:149824216-149824627	0	1_411	1_137
HIST2H3D	NM_001123375.2	1:149784825-149785236	0	1_411	1_137
HLA-A	NM_001242758.1	6:29910330-29910403	0	1_73	1_25
HLA-A	NM_001242758.1	6:29910533-29910803	1	74_343	25_115
HLA-A	NM_001242758.1	6:29911044-29911320	2	344_619	115_207
HLA-A	NM_001242758.1	6:29911898-29912174	3	620_895	207_299
HLA-A	NM_001242758.1	6:29912276-29912393	4	896_1012	299_338
HLA-A	NM_001242758.1	6:29912835-29912868	5	1013_1045	338_349
HLA-A	NM_001242758.1	6:29913010-29913058	6	1046_1093	349_365
HLA-A	NM_001242758.1	6:29913227-29913232	7	1094_1098	365_366
HLA-B	NM_005514.6	6:31322259-31322303	6	1046_1089	349_363
HLA-B	NM_005514.6	6:31322409-31322442	5	1013_1045	338_349
HLA-B	NM_005514.6	6:31322883-31323000	4	896_1012	299_338
HLA-B	NM_005514.6	6:31323093-31323369	3	620_895	207_299
HLA-B	NM_005514.6	6:31323943-31324219	2	344_619	115_207
HLA-B	NM_005514.6	6:31324464-31324734	1	74_343	25_115
HLA-B	NM_005514.6	6:31324862-31324935	0	1_73	1_25
IGF1	NM_001111283.1	12:102796318-102796344	4	452_477	151_159
IKZF1	NM_006060.5	7:50444230-50444491	3	161_421	54_141
IKZF1	NM_006060.5	7:50450237-50450405	4	422_589	141_197
IKZF1	NM_006060.5	7:50455042-50455168	5	590_715	197_239
IKZF1	NM_006060.5	7:50459426-50459561	6	716_850	239_284
IKZF1	NM_006060.5	7:50467615-50468325	7	851_1560	284_520
IRS2	NM_003749.2	13:110434388-110438400	0	1_4012	1_1338

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
KMT2C	NM_170606.2	7:151904384-151904513	23	3713_3841	1238_1281
KMT2C	NM_170606.2	7:151921519-151921701	18	2977_3158	993_1053
KMT2C	NM_170606.2	7:151927007-151927112	17	2872_2976	958_992
KMT2C	NM_170606.2	7:151932901-151933018	15	2653_2769	885_923
KMT2C	NM_170606.2	7:151935791-151935911	14	2533_2652	845_884
KMT2C	NM_170606.2	7:151962122-151962294	7	1013_1184	338_395
KMT2C	NM_170606.2	7:151970789-151970952	6	850_1012	284_338
LATS1	NM_004690.3	6:150004214-150005728	3	497_2010	166_670
MDC1	NM_014641.2	6:30668241-30668409	14	6103_6270	2035_2090
MDC1	NM_014641.2	6:30670329-30670440	13	5992_6102	1998_2034
MDC1	NM_014641.2	6:30670528-30670654	12	5866_5991	1956_1997
MDC1	NM_014641.2	6:30670880-30671063	11	5683_5865	1895_1955
MDC1	NM_014641.2	6:30671194-30671314	10	5563_5682	1855_1894
MDC1	NM_014641.2	6:30671397-30673875	9	3085_5562	1029_1854
MDC1	NM_014641.2	6:30675160-30675231	8	3014_3084	1005_1028
MDC1	NM_014641.2	6:30675342-30676134	7	2222_3013	741_1005
MDC1	NM_014641.2	6:30679188-30679281	6	2129_2221	710_741
MDC1	NM_014641.2	6:30679443-30679503	5	2069_2128	690_710
MDC1	NM_014641.2	6:30679650-30681131	4	588_2068	196_690
MDC1	NM_014641.2	6:30681424-30681494	3	518_587	173_196
MDC1	NM_014641.2	6:30681579-30681960	2	137_517	46_173
MDC1	NM_014641.2	6:30682816-30682952	1	1_136	1_46
MST1	NM_020998.3	3:49722444-49722522	13	1545_1622	515_541
MST1	NM_020998.3	3:49723292-49723395	9	1148_1250	383_417
MST1	NM_020998.3	3:49723745-49723914	7	848_1016	283_339
MST1	NM_020998.3	3:49724116-49724235	6	729_847	243_283
MST1	NM_020998.3	3:49726030-49726124	0	1_94	1_32
NCOR1	NM_006311.3	17:16068292-16068475	4	436_618	146_206
NOTCH2	NM_024408.3	1:120547951-120548211	2	156_415	52_139
NOTCH2	NM_024408.3	1:120572528-120572610	1	74_155	25_52
NOTCH2	NM_024408.3	1:120611947-120612020	0	1_73	1_25
NOTCH4	NM_004557.3	6:32163213-32163927	29	5299_6012	1767_2004
NOTCH4	NM_004557.3	6:32164100-32164198	28	5201_5298	1734_1766
NOTCH4	NM_004557.3	6:32164701-32164849	27	5053_5200	1685_1734

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
NOTCH4	NM_004557.3	6:32165075-32165371	26	4757_5052	1586_1684
NOTCH4	NM_004557.3	6:32166197-32166336	25	4618_4756	1540_1586
NOTCH4	NM_004557.3	6:32166425-32166507	24	4536_4617	1512_1539
NOTCH4	NM_004557.3	6:32166702-32166922	23	4316_4535	1439_1512
NOTCH4	NM_004557.3	6:32168607-32168783	22	4140_4315	1380_1439
NOTCH4	NM_004557.3	6:32168893-32169277	21	3756_4139	1252_1380
NOTCH4	NM_004557.3	6:32169852-32170376	20	3232_3755	1078_1252
NOTCH4	NM_004557.3	6:32171546-32171659	19	3119_3231	1040_1077
NOTCH4	NM_004557.3	6:32171913-32172166	18	2866_3118	956_1040
NOTCH4	NM_004557.3	6:32178528-32178713	17	2681_2865	894_955
NOTCH4	NM_004557.3	6:32180250-32180404	16	2527_2680	843_894
NOTCH4	NM_004557.3	6:32180600-32180688	15	2439_2526	813_842
NOTCH4	NM_004557.3	6:32180911-32181029	14	2321_2438	774_813
NOTCH4	NM_004557.3	6:32181464-32181617	13	2168_2320	723_774
NOTCH4	NM_004557.3	6:32181886-32182032	12	2022_2167	674_723
NOTCH4	NM_004557.3	6:32183002-32183162	11	1862_2021	621_674
NOTCH4	NM_004557.3	6:32184721-32184844	10	1739_1861	580_621
NOTCH4	NM_004557.3	6:32184929-32185043	9	1625_1738	542_580
NOTCH4	NM_004557.3	6:32185771-32185885	8	1511_1624	504_542
NOTCH4	NM_004557.3	6:32187368-32187563	7	1316_1510	439_504
NOTCH4	NM_004557.3	6:32187905-32188061	6	1160_1315	387_439
NOTCH4	NM_004557.3	6:32188181-32188418	5	923_1159	308_387
NOTCH4	NM_004557.3	6:32188532-32188655	4	800_922	267_308
NOTCH4	NM_004557.3	6:32188754-32189102	3	452_799	151_267
NOTCH4	NM_004557.3	6:32190287-32190583	2	156_451	52_151
NOTCH4	NM_004557.3	6:32190781-32190863	1	74_155	25_52
NOTCH4	NM_004557.3	6:32191632-32191705	0	1_73	1_25
PDPK1	NM_002613.4	16:2607703-2607964	1	25_285	9_95
PDPK1	NM_002613.4	16:2611480-2611523	2	286_328	96_110
PDPK1	NM_002613.4	16:2611771-2611909	3	329_466	110_156
PDPK1	NM_002613.4	16:2615553-2615698	4	467_611	156_204
PDPK1	NM_002613.4	16:2616356-2616454	5	612_709	204_237
PDPK1	NM_002613.4	16:2627425-2627501	6	710_785	237_262
PDPK1	NM_002613.4	16:2631295-2631364	7	786_854	262_285

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
PDPK1	NM_002613.4	16:2631607-2631704	8	855_951	285_317
PDPK1	NM_002613.4	16:2633412-2633586	9	952_1125	318_375
PIK3CA	NM_006218.2	3:178937736-178937840	12	1912_2015	638_672
PIK3CB	NM_006219.2	3:138461399-138461623	2	398_621	133_207
PIK3R2	NM_005027.3	19:18272088-18272305	5	599_815	200_272
PIK3R2	NM_005027.3	19:18279284-18279356	13	1737_1808	579_603
PMS2	NM_000535.5	7:6013029-6013173	14	2446_2589	816_863
PMS2	NM_000535.5	7:6017218-6017388	13	2276_2445	759_815
PMS2	NM_000535.5	7:6018226-6018327	12	2175_2275	725_759
PMS2	NM_000535.5	7:6022454-6022622	11	2007_2174	669_725
PPP6C	NM_002721.4	9:127933363-127933459	1	76_171	26_57
PTEN	NM_000314.6	10:89725043-89725229	8	1027_1212	343_404
PTPN11	NM_002834.3	12:112942498-112942568	14	1713_1782	571_594
PTPRS	NM_002850.3	19:5256118-5256130	8	707_718	236_240
PTPRT	NM_133170.3	20:40733208-40733358	25	3448_3597	1150_1199
RAC1	NM_018890.3	7:6431554-6431672	2	108_225	36_75
RAD51C	NM_058216.2	17:56801400-56801461	6	905_965	302_322
RAD51C	NM_058216.2	17:56811478-56811583	8	1027_1131	343_377
RASA1	NM_002890.2	5:86629083-86629154	3	829_899	277_300
RB1	NM_000321.2	13:48923091-48923159	5	540_607	180_203
RB1	NM_000321.2	13:48942662-48942740	10	1050_1127	350_376
RB1	NM_000321.2	13:48953729-48953786	13	1333_1389	445_463
RB1	NM_000321.2	13:49037866-49037971	20	2107_2211	703_737
RBM10	NM_001204468.1	X:47032526-47032596	4	628_697	210_233
RECQL	NM_032941.2	12:21623127-21623280	15	1798_1950	600_650
REL	NM_002908.3	2:61147517-61147613	8	923_1018	308_340
RYBP	NM_012234.6	3:72495646-72495774	0	1_128	1_43
SDHA	NM_004168.3	5:254507-254621	13	1795_1908	599_636
SDHC	NM_003001.3	1:161332118-161332223	5	406_510	136_170
SMARCA4	NM_003072.3	19:11144442-11144541	26	3775_3873	1259_1291
STAT5A	NM_003152.3	17:40451768-40451899	6	551_681	184_227
STAT5A	NM_003152.3	17:40452147-40452299	7	682_833	228_278
STAT5A	NM_003152.3	17:40452732-40452888	8	834_989	278_330
STAT5B	NM_012448.3	17:40370740-40370896	7	834_989	278_330

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
STAT5B	NM_012448.3	17:40371329-40371481	6	682_833	228_278
STAT5B	NM_012448.3	17:40371729-40371860	5	551_681	184_227
STK19	NM_004197.1	6:31939773-31939993	0	1_220	1_74
STK19	NM_004197.1	6:31940078-31940288	1	221_430	74_144
STK19	NM_004197.1	6:31940397-31940534	2	431_567	144_189
STK19	NM_004197.1	6:31946679-31946775	3	568_663	190_221
STK19	NM_004197.1	6:31947190-31947330	4	664_803	222_268
STK19	NM_004197.1	6:31948227-31948325	5	804_901	268_301
STK19	NM_004197.1	6:31948430-31948578	6	902_1049	301_350
STK19	NM_004197.1	6:31948780-31948826	7	1050_1095	350_365
SUZ12	NM_015355.2	17:30267304-30267351	1	275_321	92_107
SUZ12	NM_015355.2	17:30267440-30267505	2	322_386	108_129
SUZ12	NM_015355.2	17:30300164-30300250	5	506_591	169_197
SUZ12	NM_015355.2	17:30310017-30310123	8	918_1023	306_341
TAP1	NM_000593.5	6:32813355-32813562	10	2221_2427	741_809
TAP1	NM_000593.5	6:32814844-32814981	9	2084_2220	695_740
TAP1	NM_000593.5	6:32815289-32815452	8	1921_2083	641_695
TAP1	NM_000593.5	6:32815695-32815869	7	1747_1920	583_640
TAP1	NM_000593.5	6:32816428-32816617	6	1558_1746	520_582
TAP1	NM_000593.5	6:32816766-32816895	5	1429_1557	477_519
TAP1	NM_000593.5	6:32818096-32818294	4	1231_1428	411_476
TAP1	NM_000593.5	6:32818720-32818926	3	1025_1230	342_410
TAP1	NM_000593.5	6:32819885-32820016	2	894_1024	298_342
TAP1	NM_000593.5	6:32820164-32820279	1	779_893	260_298
TAP1	NM_000593.5	6:32820815-32821593	0	1_778	1_260
TAP2	NM_018833.2	6:32790065-32790095	11	1933_1962	645_654
TAP2	NM_018833.2	6:32797176-32797313	10	1796_1932	599_644
TAP2	NM_018833.2	6:32797706-32797866	9	1636_1795	546_599
TAP2	NM_018833.2	6:32798043-32798217	8	1462_1635	488_545
TAP2	NM_018833.2	6:32798394-32798583	7	1273_1461	425_487
TAP2	NM_018833.2	6:32800109-32800238	6	1144_1272	382_424
TAP2	NM_018833.2	6:32800403-32800601	5	946_1143	316_381
TAP2	NM_018833.2	6:32802930-32803136	4	740_945	247_315
TAP2	NM_018833.2	6:32803419-32803550	3	609_739	203_247

Gene	Transcript ID	Chromosome coordinates	Exon	cDNA	Amino Acid
TAP2	NM_018833.2	6:32805313-32805428	2	494_608	165_203
TAP2	NM_018833.2	6:32805517-32806010	1	1_493	1_165

Appendix 2: Mutations Represented in the Accuracy Summary Per Gene

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ABL1	3	exon11	SNV	p.E694K; p.R1103*; p.S1139L; p.A917V; p.R593Q
ABL1	1	exon6	SNV	p.Q365*
ABL1	1	exon9	SNV	Splicing
ACVR1	2	exon6	SNV	p.R206H; p.R202*
ACVR1	2	exon7	SNV	p.N253T; p.R240C
ACVR1	1	exon8	SNV	p.R307Q
AGO2	1	exon11	DEL	p.Q459Sfs*9
AGO2	1	exon13	SNV	p.K566N
AGO2	1	exon5	SNV	p.M213I
AKT1	1	exon12	SNV	p.D323G
AKT1	1	exon15	SNV	p.S475L
AKT1	7	exon4	SNV	Splicing; p.E17K
AKT1	1	exon7	SNV	p.E149K
AKT2	1	exon13	SNV	p.V430G
AKT2	1	exon3	SNV	p.E17K
AKT2	1	exon9	SNV	p.L266V
AKT3	2	exon10	SNV	p.R367*; p.R388H
AKT3	1	exon12	SNV	p.D431Y
AKT3	1	exon2	SNV	p.E40G
AKT3	1	exon5	SNV	p.R166Q
AKT3	1	exon7	SNV	p.L211F
AKT3	1	exon8	SNV	p.S244F
ALK	3	exon1	SNV	p.G30S; p.E172D; p.F58C
ALK	1	exon10	SNV	p.W615L
ALK	2	exon11	SNV	p.R658*; p.E668K
ALK	1	exon15	SNV	p.S865Y
ALK	1	exon16	SNV	p.G935R
ALK	1	exon20	SNV	p.S1086L
ALK	1	exon21	SNV	p.V1147A
ALK	1	exon23	SNV	p.G1202R
ALK	1	exon26	SNV	p.A1280V
ALK	1	exon27	SNV	p.W1320*; p.E1321K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ALK	2	exon29	SNV	p.E1419D; p.R1467T
ALK	1	exon3	SNV	p.R292C
ALK	1	exon4	SNV	p.L376H
ALOX12B	1	exon11	SNV	p.A504V
ALOX12B	2	exon12	SNV	p.Y512H; p.V527M
ALOX12B	1	exon14	SNV	p.M616I
ALOX12B	1	exon5	SNV	p.V212I
ALOX12B	1	exon6	SNV	p.S248Y
ANKRD11	2	exon10	DEL	p.Y761Qfs*20; p.K1245del
ANKRD11	15	exon10	SNV	p.A309T; p.V2490M; p.E1021Q; p.R942I; p.T1214P; p.A520V; p.V1748M; p.E1897K; p.T1920M; p.G1537D; p.E1656Q; p.E346K; p.R2232H; p.V420M; p.P1638H; p.E1291*; p.S1790L; p.D1379N; p.S774L; p.S929G; p.R1585S; p.P1974L
ANKRD11	1	exon12	SNV	p.E2533D
ANKRD11	1	exon4	SNV	p.M1?
APC	1	exon10	SNV	p.R283*
APC	2	exon11	SNV	p.S346Y; p.R402C
APC	2	exon13	SNV	p.Y500*; p.R499*
APC	2	exon15	SNV	p.R564*; p.R554*
APC	5	exon16	SNV	p.C607*; p.D605N; p.E582*; p.R653K; p.W593*
APC	22	exon17	DEL	p.E902Kfs*14; p.Q1406Rfs*9; p.N1455Ifs*18; p.S1421Rfs*52; p.E1322Afs*8; p.S1465Wfs*3; p.E1494Vfs*12; p.E1309Kfs*12; p.L1488Yfs*19; p.D1512Efs*11; p.I1287*; p.E941_T946del; p.Y796Wfs*2; p.V1452Yfs*21; p.G1312Cfs*12; p.D1498Wfs*15; p.Q886Dfs*25; p.I1580Mfs*69; p.S1495Vfs*12; p.S843Lfs*17; p.S1206Rfs*59; p.I1055Nfs*4; p.G977Sfs*7; p.E1309Dfs*4; p.I1401Qfs*6

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
APC	33	exon17	SNV	p.E1397*; p.I1008M; p.R923*; p.R1435*; p.K2412R; p.D1860N; p.E1306*; p.Q1549*; p.Q1367*; p.S2429Y; p.L1618I; p.S1272*; p.E1322*; p.R1114*; p.R1450*; p.E1668K; p.S1400*; p.Y1854*; p.Q1429*; p.E763*; p.R876*; p.S1327*; p.H712P; p.S2026Y; p.R1640Q; p.C1410*; p.E941*; p.E1295*; p.E1494*; p.P2435L; p.E771*; p.P2788L; p.D1974G; p.Q2829*
APC	11	exon17	INS	p.S1403Rfs*15; p.I1746Dfs*23; p.A1492Cfs*22; p.Q1447Kfs*29; p.T1301Ifs*3; p.P832Tfs*12; p.T1556Nfs*3; p.E1397Sfs*2
APC	1	exon5	SNV	p.S80N; p.E129*
APC	1	exon6	SNV	p.A146V
APC	1	exon7	INS	p.R213Tfs*39
APC	5	exon8	SNV	p.R232*; p.R216*
APC	1	exon9	DEL	p.Q278Ffs*8
APC	1	exon9	SNV	p.E268*
AR	5	exon1	SNV	p.T437A; p.P153S; p.M1?; p.Q147K; p.E442K; p.R20Q
AR	1	exon5	SNV	p.G744A
AR	1	exon6	SNV	p.F814V
AR	2	exon7	SNV	p.F827L; p.K837R
ARAF	1	exon13	SNV	p.R456Q
ARAF	1	exon2	SNV	p.V32M
ARAF	2	exon7	SNV	p.S214P; p.S214T
ARAF	1	exon9	SNV	p.D286N
ARID1A	1	exon1	SNV	p.S334*
ARID1A	1	exon1	INS	p.G277Rfs*123
ARID1A	4	exon1	DEL	p.S11Afs*91; p.P224Rfs*8; p.S265del
ARID1A	1	exon10	SNV	p.S993F
ARID1A	2	exon11	SNV	p.K1059N; p.G1063E
ARID1A	2	exon12	INS	p.K1129Qfs*64; p.A1102Sfs*63
ARID1A	1	exon13	DEL	p.S1140Cfs*20
ARID1A	1	exon13	INS	p.L1160Ffs*33
ARID1A	1	exon14	DEL	p.T1198Nfs*8
ARID1A	1	exon15	INS	p.F1245Lfs*14

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ARID1A	2	exon15	DEL	p.F1245Sfs*24; p.R1287Sfs*10
ARID1A	2	exon15	SNV	p.V1271A; p.V1271L
ARID1A	1	exon16	SNV	p.Y1324*
ARID1A	3	exon18	DEL	p.M1634*; p.P1546Lfs*19; p.L1405Cfs*76
ARID1A	3	exon18	SNV	p.G1515S; p.D1536H; p.H1524D
ARID1A	1	exon18	INS	p.N1571*
ARID1A	1	exon2	INS	p.S409Vfs*214
ARID1A	3	exon2	SNV	p.Q450*; p.G444S; p.Q393*
ARID1A	9	exon20	SNV	p.R1721*; p.Q2115*; p.R1989*; p.G2012D; p.I2067N; p.E1864K; p.R1833H; p.Q2219*; p.W2048*
ARID1A	1	exon20	INS	p.I2192Yfs*33
ARID1A	8	exon20	DEL	p.P2102Lfs*47; p.Q2273Rfs*4; p.G1737Vfs*33; p.R2143Afs*57; p.D1850Tfs*33; p.F2141Sfs*59; p.L1712Cfs*2
ARID1A	1	exon3	INS	p.Q553Tfs*70
ARID1A	1	exon3	DEL	p.Y551Tfs*68
ARID1A	7	exon3	SNV	Splicing; p.Q546*; p.Q467*; p.Q543*; p.Q502*; p.Q557*; p.Q528*
ARID1A	1	exon4	DEL	p.K622Nfs*14
ARID1A	1	exon4	SNV	p.S634*
ARID1A	1	exon6	SNV	p.R750*
ARID1A	1	exon6	DEL	p.P726Vfs*84
ARID1A	2	exon8	SNV	p.C884*; p.S862F
ARID1A	1	exon9	SNV	p.Q944*
ARID1B	4	exon1	SNV	p.A394T; p.E288K; p.E27D; p.Q211*; p.C295Y
ARID1B	1	exon1	DEL	p.R372Gfs*7
ARID1B	1	exon12	SNV	p.R1075*
ARID1B	1	exon13	DEL	p.K1174Sfs*37
ARID1B	1	exon15	SNV	p.D1238N
ARID1B	5	exon18	SNV	p.P1601T; p.P1573L; p.R1604K; p.R1505K; p.P1576S; p.G1442R
ARID1B	1	exon19	DEL	p.W1637Cfs*6
ARID1B	1	exon2	SNV	p.R525K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ARID1B	6	exon20	SNV	p.R1944*; p.R1990*; p.R2128*; p.D1727Y; p.E1748*; p.E1685*
ARID1B	1	exon7	SNV	p.Q763*
ARID2	1	exon10	SNV	p.K435R
ARID2	1	exon11	SNV	p.K466E
ARID2	1	exon13	SNV	p.R558C
ARID2	6	exon15	SNV	p.Q871*; p.R1414K; p.S742F; p.Q934*; p.Q1100*; p.A1547V
ARID2	1	exon15	INS	p.L1007Ffs*50
ARID2	1	exon16	SNV	p.Q1630H
ARID2	1	exon18	SNV	p.Q1702*
ARID2	1	exon19	SNV	p.S1725*
ARID2	1	exon19	INS	p.T1723Hfs*6
ARID2	1	exon2	DEL	p.I37Sfs*21
ARID2	1	exon2	SNV	p.G59*
ARID2	1	exon21	SNV	p.E1832Q
ARID2	1	exon7	SNV	p.R247C
ARID5B	1	exon1	SNV	p.N4D
ARID5B	5	exon10	SNV	p.D552Y; p.E1044V; p.E984D; p.V755I; p.L814S
ARID5B	2	exon2	SNV	p.K49Q; p.E80D
ASXL1	1	exon10	SNV	p.G360*
ASXL1	3	exon11	SNV	p.R413W; p.A433T; p.F481C
ASXL1	1	exon12	INS	p.G646Wfs*12
ASXL1	8	exon12	SNV	p.S689*; p.Q768L; p.L1314I; p.L691V; p.E1020*; p.K1412N; p.E997K; p.S689L; p.R606Q
ASXL1	2	exon12	DEL	p.L762Ffs*10; p.K580Nfs*121
ASXL2	2	exon11	SNV	p.R614*; p.E407K
ASXL2	1	exon12	SNV	p.A677T; p.E730*; p.R1252I; p.E804Q; p.D1148Y; p.E722Q; p.R1004T; p.R828T; p.D1257N
ASXL2	1	exon12	DEL	p.T709Sfs*3
ASXL2	1	exon6	SNV	p.C182Y
ASXL2	1	exon7	SNV	p.S228*
ASXL2	1	exon8	SNV	p.A267T
ASXL2	1	exon9	SNV	p.E330K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ATM	2	exon10	SNV	p.R447*; p.W488L
ATM	1	exon17	SNV	p.K828N
ATM	1	exon19	SNV	p.S974F
ATM	2	exon20	SNV	p.R981C
ATM	2	exon25	SNV	p.H1213N; p.F1247L; p.F1201S
ATM	1	exon31	INS	p.I1581Nfs*5
ATM	1	exon31	SNV	p.I1559V
ATM	1	exon33	SNV	p.V1649L
ATM	2	exon34	SNV	p.C1674R; p.E1724*
ATM	1	exon35	SNV	p.R1730*
ATM	1	exon37	SNV	p.W1858*
ATM	1	exon39	SNV	Splicing
ATM	1	exon39	DEL	p.F1928Lfs*9
ATM	2	exon42	SNV	p.W2042C; p.R2034*
ATM	1	exon47	SNV	p.P2271S
ATM	1	exon53	SNV	p.Q2615*
ATM	3	exon57	SNV	p.L2780I; p.T2773I; p.G2765S
ATM	2	exon58	SNV	p.R2832C; p.T2853M
ATM	1	exon6	DEL	p.S214Pfs*16
ATM	1	exon63	SNV	p.A3006P
ATM	3	exon7	SNV	p.E257*; p.R248L; p.R248*
ATM	2	exon8	SNV	p.L312*; p.R329T
ATR	1	exon12	SNV	p.S860*
ATR	2	exon22	SNV	p.A1363V; p.D1380Y
ATR	1	exon23	SNV	p.A1403T
ATR	1	exon24	SNV	p.R1445G
ATR	1	exon36	SNV	p.L2070I
ATR	1	exon38	SNV	p.D2111N
ATR	1	exon39	SNV	p.K2206E
ATR	1	exon4	SNV	p.Q151P
ATR	2	exon43	SNV	p.R2407C; p.L2428I
ATR	1	exon9	SNV	p.R635*
ATRX	1	exon11	SNV	p.E1279*
ATRX	2	exon16	SNV	p.T1534I; p.D1540G

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ATRX	1	exon20	DEL	p.S1732Lfs*4
ATRX	1	exon21	SNV	p.R1803C
ATRX	1	exon22	SNV	p.T1824K
ATRX	1	exon25	SNV	p.G1965C
ATRX	1	exon30	SNV	p.S2184P
ATRX	1	exon31	DEL	p.E2265del
ATRX	1	exon32	SNV	p.E2325D
ATRX	1	exon35	SNV	p.D2455G
ATRX	1	exon4	SNV	p.S67C
ATRX	1	exon8	SNV	Splicing
ATRX	4	exon9	SNV	p.V593A; p.K1182*; p.K1000T; p.D1051N
ATRX	5	exon9	DEL	p.V1002Lfs*30; p.Y341del; p.V725Gfs*7; p.K720Cfs*8; p.K329Ifs*3
ATRX	1	exon9	INS	p.L868Ffs*10
AURKA	2	exon9	SNV	p.I272N; p.L240*
AXIN1	1	exon10	SNV	p.V784I
AXIN1	3	exon2	SNV	p.T187S; p.Y207N; p.S100G
AXIN1	3	exon6	SNV	p.A441T; p.G514R; p.G507V
AXIN2	1	exon11	SNV	p.S809T
AXIN2	2	exon2	INS	p.V40Gfs*101; p.L99Pfs*42
AXIN2	1	exon2	SNV	p.A174T
AXIN2	1	exon2	DEL	p.T254Lfs*2
AXIN2	2	exon3	SNV	p.H284D; p.G286C
AXIN2	1	exon4	SNV	p.R339H; p.P353L
AXIN2	1	exon6	SNV	p.R414W
AXIN2	1	exon8	INS	p.N666Qfs*41
AXL	1	exon18	SNV	p.V686M
AXL	2	exon20	SNV	p.E787*; p.T806A
AXL	1	exon3	SNV	p.L118F
AXL	2	exon7	SNV	p.G274R; p.L299F
AXL	2	exon8	SNV	p.A370V; p.R357L
B2M	1	exon1	SNV	p.M1?
B2M	1	exon1	DEL	p.L15Ffs*41
B2M	2	exon2	SNV	p.N62S; p.S72*

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
BAP1	2	exon13	SNV	p.A514D; p.E450K
BAP1	1	exon14	DEL	p.M615Wfs*2
BAP1	1	exon15	SNV	p.E647*
BAP1	1	exon16	SNV	p.Q684*
BAP1	2	exon17	SNV	p.R718Q; p.N695H
BAP1	1	exon2	SNV	p.D21Y
BAP1	2	exon4	SNV	p.E54K; p.S63C
BAP1	2	exon5	SNV	p.V99M; p.A92E
BAP1	1	exon5	DEL	p.S105Rfs*17
BAP1	1	exon8	SNV	p.R208W
BARD1	1	exon11	SNV	p.V713G
BARD1	2	exon4	SNV	p.V248G; p.E268*
BARD1	1	exon4	DEL	p.E225del
BBC3	1	exon3	INS	p.Q183Dfs*48
BCL10	1	exon2	SNV	p.E54Q
BCL10	2	exon3	SNV	p.G199W; p.T229S
BCL2	1	exon2	SNV	p.S70*
BCL2L1	1	exon2	INS	p.S106_D107insG
BCL2L11	1	exon2	SNV	p.R17T
BCL2L11	1	exon3	SNV	p.Q55*
BCL2L11	1	exonN/A	DEL	p.P54Rfs*16
BCL2L11	1	exonN/A	SNV	p.E86K
BCL6	2	exon3	SNV	p.R26W; p.P4L
BCL6	3	exon5	SNV	p.R214W; p.K377E; p.S438F
BCL6	1	exon8	SNV	p.C579Y
BCOR	3	exon10	SNV	p.E1464Q; p.N1459S; p.N1461K
BCOR	1	exon12	SNV	p.F1578L
BCOR	1	exon14	SNV	p.S1655Y
BCOR	1	exon2	DEL	p.L7Cfs*9
BCOR	1	exon4	DEL	p.G81Tfs*23
BCOR	8	exon4	SNV	p.T940A; p.A199T; p.R748W; p.V973M; p.R861H; p.S214*; p.A165V; p.P572A; p.E485K
BCOR	2	exon8	SNV	p.R1215K; p.R1209C
BIRC3	1	exon3	DEL	p.M158Vfs*7

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
BIRC3	1	exon4	SNV	p.D307N
BIRC3	1	exon8	SNV	p.D497H
BLM	1	exon18	SNV	p.S1093*
BLM	1	exon23	SNV	p.S1368Y
BLM	1	exon8	SNV	p.D554N
BMPR1A	1	exon4	SNV	p.L59*
BMPR1A	1	exon6	INS	p.V142Cfs*7
BRAF	1	exon13	SNV	p.R509L
BRAF	16	exon15	SNV	p.N581S; p.V600E
BRAF	1	exon15	DEL	p.V600_K601delinsE
BRAF	1	exon2	SNV	p.S76L
BRCA1	6	exon10	SNV	p.N1059S; p.T557A; p.S1262P; p.G394S; p.K1264N; p.N635K
BRCA1	1	exon10	INS	p.Q1359Afs*9
BRCA1	1	exon12	SNV	p.E1413G
BRCA1	1	exon13	SNV	p.Q1467*
BRCA1	1	exon15	SNV	p.A1627T
BRCA1	1	exon23	SNV	p.E1829K
BRCA2	5	exon11	SNV	p.D1990N; p.N1100Y; p.R1160K; p.N1619H; p.S1461F
BRCA2	1	exon11	DEL	p.D1990Cfs*12
BRCA2	1	exon17	SNV	p.P2639A
BRCA2	1	exon18	SNV	p.D2692N
BRCA2	1	exon20	SNV	p.L2838*
BRCA2	1	exon23	SNV	p.R2991L
BRCA2	1	exon27	SNV	p.R3268T
BRCA2	1	exon3	SNV	p.K63Q
BRCA2	1	exon6	SNV	p.K169T
BRCA2	1	exon9	INS	Splicing
BRD4	3	exon10	SNV	p.E668Q; p.T663R; p.T598M
BRD4	1	exon11	SNV	p.D687N
BRD4	1	exon5	SNV	p.Q245E
BRIP1	1	exon14	SNV	p.S682F
BRIP1	1	exon5	SNV	p.R160I
BRIP1	1	exon7	SNV	p.V271F

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
BTK	1	exon12	SNV	p.H333Y
BTK	1	exon13	SNV	p.V377G
BTK	1	exon15	SNV	p.E455D
BTK	1	exon16	SNV	p.G541S
BTK	2	exon2	SNV	p.V4M; p.R28H
CALR	1	exon3	INS	p.G106Vfs*6
CARD11	1	exon10	SNV	p.D476N; p.D475N
CARD11	1	exon16	SNV	p.T681I
CARD11	1	exon18	SNV	p.D799N
CARD11	1	exon21	SNV	p.E907K
CARD11	1	exon23	SNV	p.A1047T
CARD11	1	exon5	SNV	p.D199E
CARD11	1	exon8	SNV	p.M365T
CARD11	1	exon9	SNV	p.V429I
CARM1	1	exon16	SNV	p.A594V
CARM1	1	exon2	SNV	p.E89*
CASP8	1	exon3	SNV	p.D18Y
CASP8	3	exon6	SNV	p.D209N; p.I174S; p.L204M; p.E212*
CASP8	2	exon8	SNV	p.R265L; p.E266D
CASP8	1	exon9	DEL	p.F357Sfs*4
CASP8	3	exon9	SNV	p.P432S; p.C326*; p.R449*
CBFB	1	exon1	DEL	p.F18Lfs*4
CBFB	1	exon2	DEL	p.I55Mfs*34
CBFB	1	exon2	INS	Splicing
CBFB	1	exon2	SNV	Splicing
CBFB	1	exon3	DEL	p.N63Ifs*26
CBFB	2	exon4	SNV	p.M101V
CBL	1	exon1	SNV	p.T50K
CBL	1	exon4	SNV	p.S204G
CBL	2	exon6	SNV	p.I314F; p.D311N
CBL	1	exon8	SNV	p.I393S
CBL	1	exon9	INS	p.D460_D456ins
CCND1	2	exon1	SNV	p.V42E; p.D19H
CCND1	1	exon5	INS	p.V290_R291insL

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
CCND1	2	exon5	SNV	p.V290E; p.D292H
CCND2	1	exon5	SNV	p.Q260*
CCND3	1	exon2	SNV	p.E115K
CCND3	1	exon2	DEL	p.C125Hfs*34
CCND3	1	exon5	DEL	p.P284Lfs*20
CCND3	2	exon5	INS	p.P278Rfs*27; p.T285Vfs*40
CCNE1	1	exon12	SNV	p.S399R
CCNE1	1	exon5	SNV	p.C68Y
CD274	1	exon2	SNV	p.M10I
CD276	1	exon8	SNV	p.A516S
CD79A	2	exon2	SNV	p.S118Y; p.A40V
CD79A	1	exon3	DEL	p.R131Gfs*61
CD79B	1	exon3	SNV	p.E103K
CDC73	1	exon14	SNV	p.V387I
CDC73	1	exon16	SNV	p.W492C; p.W492L
CDC73	1	exon6	SNV	p.A154P
CDC73	1	exon8	SNV	p.L253P
CDH1	1	exon1	SNV	p.S9W
CDH1	1	exon12	DEL	p.L582Cfs*2
CDH1	1	exon12	SNV	p.D616H
CDH1	1	exon15	SNV	p.V787I
CDH1	1	exon2	SNV	p.Q23*
CDH1	3	exon3	SNV	p.N93S; p.H122Y; p.R63*
CDH1	2	exon5	SNV	p.L214R; p.R224H
CDH1	1	exon6	DEL	p.S232Tfs*17
CDH1	2	exon6	SNV	p.P277L; p.E261K
CDH1	1	exon7	DEL	p.Y302Tfs*54
CDH1	1	exon8	DEL	p.S337Rfs*9
CDH1	1	exon9	INS	p.N405Kfs*14
CDH1	2	exon9	SNV	p.Q422*; p.E410V
CDK12	3	exon1	SNV	p.R322Q; p.S251*; p.Y285*
CDK12	1	exon1	INS	p.P324Rfs*27
CDK12	1	exon1	DEL	p.S209Kfs*127
CDK12	1	exon14	SNV	p.G1339R

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
CDK12	1	exon14	DEL	p.R1374Gfs*7
CDK12	1	exon4	SNV	p.R722C; p.K743T
CDK12	1	exon6	SNV	p.R858Q
CDK12	1	exon7	SNV	p.R882W
CDK12	1	exon8	SNV	p.R890H
CDK4	1	exon5	SNV	p.E206Q
CDK8	1	exon8	SNV	p.M273T
CDKN1A	1	exon3	SNV	p.F51L
CDKN1A	2	exon3	DEL	p.C34*; p.L71Qfs*14
CDKN1A	1	exon3	INS	p.F53Gfs*99
CDKN1B	1	exon1	SNV	p.S27*
CDKN2B	1	exon1	SNV	p.A23P
CDKN2B	1	exon2	SNV	p.V117L
CDKN2C	1	exon3	SNV	p.F92L
CEBPA	1	exon1	SNV	p.P101S
CHEK1	1	exon6	SNV	p.R160C
CHEK2	3	exon2	SNV	p.Q51H; p.V25I; p.A104V
CHEK2	1	exon3	SNV	p.F125V
CHEK2	2	exon4	SNV	p.I189M; p.S194R
CHEK2	1	exon7	SNV	p.H282Y
CIC	2	exon10	DEL	p.P509Hfs*14
CIC	3	exon10	SNV	p.N702D; p.P882H; p.P718H
CIC	1	exon11	DEL	p.S902Ffs*21
CIC	1	exon15	SNV	p.G1195D
CIC	1	exon3	SNV	p.G136*
CIC	2	exon4	SNV	p.I155M; p.P165L
CIC	1	exon7	DEL	p.Q371Rfs*35
CREBBP	1	exon13	SNV	p.S767P
CREBBP	1	exon15	SNV	p.A966V
CREBBP	1	exon16	DEL	p.I1084Sfs*15
CREBBP	1	exon16	SNV	p.R1081C
CREBBP	1	exon19	SNV	p.R1223H
CREBBP	2	exon2	SNV	p.P153L; p.G211E; p.V159M
CREBBP	1	exon21	SNV	p.N1272S

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
CREBBP	1	exon24	SNV	p.E1353*
CREBBP	1	exon25	SNV	p.D1406H
CREBBP	2	exon26	SNV	p.G1456R; p.L1464*
CREBBP	1	exon28	SNV	p.P1546L
CREBBP	1	exon30	INS	p.S1657Pfs*90
CREBBP	7	exon31	SNV	p.R1866H; p.R2344L; p.A2265E; p.C1783Y; p.C1826*; p.A1782T; p.R2151W; p.E1963K
CREBBP	1	exon4	INS	p.R370Lfs*57
CREBBP	1	exon4	SNV	p.H363Y
CREBBP	1	exon6	DEL	p.S469*
CREBBP	1	exon6	SNV	p.Q495H
CRKL	2	exon2	SNV	p.E179K; p.P231L
CSDE1	1	exon10	SNV	p.D303Y; p.D303V
CSDE1	2	exon11	SNV	p.R332C; p.T378A
CSDE1	1	exon12	SNV	p.P443S
CSDE1	1	exon17	SNV	p.G633V
CSDE1	1	exon21	SNV	p.R838H
CSDE1	1	exon4	SNV	p.G84*
CSDE1	1	exon9	DEL	p.I279Sfs*35
CSF1R	1	exon11	SNV	p.M523I
CSF1R	1	exon15	SNV	p.E680K
CSF1R	1	exon16	SNV	p.N734K
CSF1R	1	exon3	SNV	p.G52D
CSF1R	1	exon7	SNV	p.D344G
CSF3R	1	exon10	SNV	p.R428T
CSF3R	1	exon11	DEL	p.S469Afs*22
CSF3R	2	exon13	SNV	p.G555R; p.S528T
CSF3R	1	exon14	SNV	p.L614F
CSF3R	1	exon17	SNV	p.A859T
CSF3R	1	exon4	INS	p.R74Qfs*7
CSF3R	1	exon4	SNV	p.G120R
CTCF	1	exon12	SNV	p.E687K
CTCF	3	exon3	SNV	p.A88D; p.E104*; p.L98S
CTCF	1	exon4	SNV	p.R283H

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
CTCF	1	exon5	SNV	p.R342C
CTCF	1	exon6	DEL	p.L383_S385del
CTCF	1	exon6	SNV	p.P378Q
CTCF	2	exon7	SNV	p.D451G; p.C409S
CTCF	1	exon8	SNV	p.H460R
CTNNB1	1	exon10	SNV	p.H524D
CTNNB1	2	exon12	SNV	p.R612*; p.C619S
CTNNB1	2	exon15	SNV	p.E738K; p.D747N
CTNNB1	11	exon3	SNV	p.D32V; p.S33C; p.S33A; p.S45F; p.G34E; p.T41I; p.S45P
CTNNB1	1	exon3	DEL	p.I35_G38del
CTNNB1	1	exon4	SNV	p.A146V
CTNNB1	3	exon5	SNV	p.R212C; p.V241M; p.R200H
CTNNB1	1	exon6	SNV	p.F293L
CTNNB1	1	exon7	SNV	p.K335I
CTNNB1	2	exon8	SNV	p.L377V; p.N387K
CTNNB1	2	exon9	SNV	p.L405V; p.R486H
CTNNB1	1	exon9	INS	p.C429Wfs*3
CUL3	1	exon10	SNV	p.S480*
CUL3	1	exon11	DEL	p.P525Hfs*16
CUL3	1	exon12	SNV	p.R546Q
CUL3	3	exon4	SNV	p.R148L; p.R158G; p.V178I
CUL3	1	exon9	SNV	p.R439T
CXCR4	1	exon2	SNV	p.G258W
CYLD	1	exon10	SNV	p.R489C
CYLD	1	exon13	SNV	p.E626D
CYLD	1	exon17	SNV	p.F763L
CYLD	1	exon18	SNV	p.E798Q
CYLD	1	exon19	SNV	p.E860Q
CYLD	1	exon4	SNV	p.L101S
CYSLTR2	2	exon6	SNV	p.W51C; p.E310K
DDR2	1	exon10	SNV	p.D326Y
DDR2	2	exon14	SNV	p.D552Y; p.P509S
DDR2	2	exon16	SNV	p.S683N; p.D661Y
DDR2	1	exon17	DEL	p.I743_R746del

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
DDR2	1	exon17	SNV	p.R714Q
DDR2	1	exon5	SNV	p.R31G
DDR2	2	exon6	SNV	p.E113K; p.E113A; p.E67*
DDR2	1	exon8	SNV	p.S193C
DICER1	1	exon16	SNV	p.E824K
DICER1	1	exon17	DEL	p.I932Tfs*16
DICER1	1	exon2	SNV	p.A20T
DICER1	1	exon20	SNV	p.E1036*
DICER1	2	exon21	SNV	p.P1231S; p.F1332C
DICER1	1	exon22	SNV	p.V1387A; p.R1368H
DICER1	3	exon24	SNV	p.D1709N; p.Q1702R; p.P1729L
DIS3	1	exon4	SNV	p.E166K
DIS3	1	exon5	SNV	p.L202V
DNAJB1	2	exon2	SNV	p.H99R; p.P98H
DNAJB1	1	exon2	DEL	p.K248Efs*23
DNMT1	1	exon19	SNV	p.G487V
DNMT1	1	exon36	SNV	p.G1406R
DNMT1	1	exon37	SNV	p.V1460L
DNMT1	1	exon41	SNV	p.E1627*
DNMT1	1	exon8	SNV	p.R221H
DNMT3A	1	exon14	SNV	p.A525S
DNMT3A	1	exon17	SNV	p.Q692*
DNMT3A	2	exon19	SNV	p.R736C; p.R736H
DNMT3A	1	exon2	SNV	p.E18K
DNMT3A	2	exon6	SNV	p.E205*; p.P195S
DNMT3A	1	exon9	SNV	p.S349L
DNMT3B	1	exon14	SNV	p.R497Q
DNMT3B	1	exon2	SNV	p.R42C
DNMT3B	1	exon5	SNV	Splicing
DOT1L	2	exon14	SNV	p.Q439*; p.Q443*
DOT1L	1	exon17	SNV	p.E520Q
DOT1L	1	exon20	SNV	p.G715D
DOT1L	1	exon24	SNV	p.V1103M
DOT1L	1	exon25	SNV	p.D1161N

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
DOT1L	1	exon27	SNV	p.T1274A
DOT1L	1	exon3	SNV	p.D50Y
DROSHA	1	exon11	SNV	p.K559N
DROSHA	1	exon14	DEL	p.C641Vfs*2
DROSHA	1	exon20	SNV	p.G884R
DROSHA	1	exon21	SNV	p.H902R
DROSHA	2	exon29	SNV	p.E1147K; p.G1141D
DROSHA	1	exon31	SNV	p.D1231N
DROSHA	1	exon35	SNV	p.E1345K
DROSHA	1	exon4	SNV	p.R20Q
DROSHA	1	exon8	SNV	p.P465L
DUSP4	2	exonN/A	SNV	p.S99C; p.C69R
E2F3	2	exon5	SNV	p.D330G; p.R305*
EED	1	exon1	SNV	p.E3Q
EED	1	exon12	SNV	p.R439*
EGFL7	1	exon6	SNV	p.R85C
EGFL7	1	exon7	SNV	p.V118D
EGFR	1	exon1	SNV	p.S22I
EGFR	1	exon13	SNV	p.R527W
EGFR	1	exon15	SNV	p.G598V
EGFR	1	exon18	SNV	p.R705M
EGFR	1	exon18	DEL	p.F723Afs*24
EGFR	4	exon19	DEL	p.E746Ifs*16; p.L747_P753delinsS; p.E746_A750del
EGFR	1	exon20	SNV	p.R776C
EGFR	2	exon21	SNV	p.L858R
EGFR	1	exon24	SNV	p.I966S
EGFR	1	exon28	SNV	p.Q1159H
EGFR	1	exon3	SNV	p.R108K
EGFR	1	exon4	SNV	p.W164*; p.S177L
EGFR	1	exon7	SNV	p.A289V
EGFR	1	exon8	SNV	p.R324L
EGFR	1	exon9	SNV	p.T363A
EIF1AX	2	exon2	SNV	p.G8R; p.R13H
EIF4A2	1	exon1	SNV	p.G4A

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ELF3	1	exon2	DEL	p.P31Lfs*12
ELF3	1	exon3	DEL	p.K82_Y85delinsN
ELF3	1	exon5	INS	p.S190Lfs*6
ELF3	1	exon7	DEL	p.D234Afs*59
ELF3	1	exon8	DEL	p.Q313Kfs*8
ELF3	2	exon8	SNV	p.N323D; p.F303Y; p.F303L
ELF3	1	exon8	INS	p.K304Qfs*167
ELF3	2	exon9	SNV	p.S358*; p.R344L
ELF3	1	exon9	INS	p.R339Nfs*109
EP300	1	exon15	SNV	p.Q965*
EP300	1	exon2	SNV	p.Q240K
EP300	1	exon23	SNV	p.E1285K
EP300	1	exon24	SNV	p.P1294Q
EP300	1	exon27	SNV	p.A1437P
EP300	1	exon29	SNV	p.S1577C
EP300	1	exon30	DEL	p.S1642Lfs*67
EP300	3	exon30	SNV	p.F1595L; p.L1633M; p.L1631F
EP300	1	exon31	DEL	p.Q2311Sfs*42
EP300	5	exon31	SNV	p.R1830S; p.S2366T; p.Q1912E; p.F2364S; p.G2394*; p.H1820Y; p.T1856A
EP300	1	exon4	SNV	p.K336*
EP300	1	exon6	SNV	p.I429V
EPAS1	1	exon13	SNV	p.R710*
EPAS1	1	exon16	SNV	p.E838K
EPCAM	1	exon3	DEL	p.D96_D98del
EPCAM	1	exon4	SNV	p.R153I
EPCAM	1	exon5	SNV	p.T182M
EPHA3	1	exon10	SNV	p.S622F
EPHA3	1	exon13	SNV	p.S768*
EPHA3	1	exon14	SNV	p.G784V
EPHA3	1	exon16	SNV	p.I931T
EPHA3	1	exon17	SNV	p.V983M
EPHA3	1	exon3	INS	p.F204Ifs*24
EPHA3	1	exon4	SNV	p.D316G

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
EPHA3	1	exon5	SNV	p.E338K
EPHA5	2	exon1	SNV	p.Y32*; p.P25T
EPHA5	1	exon14	SNV	p.L801R
EPHA5	1	exon15	SNV	p.G838E
EPHA5	1	exon16	SNV	p.R919Q
EPHA5	2	exon18	SNV	p.V1035M; p.E1025*
EPHA5	2	exon3	SNV	p.E297D; p.D216N
EPHA5	2	exon4	SNV	p.K340N; p.A332V
EPHA5	1	exon5	SNV	p.R413S
EPHA5	1	exon6	SNV	p.P492T
EPHA5	1	exon7	SNV	p.R541*
EPHA7	1	exon10	SNV	p.K601T
EPHA7	1	exon13	SNV	p.E784Q
EPHA7	2	exon14	SNV	p.A807T; p.Y832N
EPHA7	1	exon15	SNV	p.I846T
EPHA7	1	exon3	SNV	p.P278S
EPHA7	1	exon4	SNV	p.Y316C
EPHA7	1	exon5	SNV	p.G387A
EPHA7	1	exon6	SNV	p.K451N; p.E452*
EPHB1	1	exon14	SNV	p.R842Q
EPHB1	1	exon14	DEL	p.L878*
EPHB1	1	exon15	SNV	p.D909Y
EPHB1	1	exon2	SNV	p.T27M
EPHB1	4	exon3	SNV	p.R56L; p.R170Q; p.R79G; p.L186F
EPHB1	1	exon5	SNV	p.G350W
EPHB1	1	exon6	SNV	p.E468*
EPHB1	1	exon7	SNV	p.H476R
EPHB1	1	exon9	SNV	p.E572*
ERBB2	1	exon11	SNV	p.T246R
ERBB2	5	exon12	SNV	p.S280F; p.S280Y
ERBB2	1	exon19	SNV	p.P564A
ERBB2	2	exon22	SNV	p.G697A; p.V667L
ERBB2	4	exon23	SNV	p.D739Y; p.L725S; p.L725V
ERBB2	2	exon24	INS	p.A745_A741ins

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ERBB2	2	exon24	SNV	p.Q798H; p.G746V
ERBB2	1	exon25	SNV	p.V812I
ERBB2	1	exon28	SNV	p.R955H
ERBB2	1	exon29	SNV	p.R976H
ERBB2	4	exon31	SNV	p.K1208E; p.G1158V; p.K1147N; p.D1153N
ERBB2	1	exonN/A	DEL	Splicing
ERBB2	1	exonN/A	SNV	Splicing
ERBB3	1	exon20	SNV	p.G804E; p.G804R
ERBB3	2	exon28	SNV	p.S1172F; p.P1250S
ERBB3	2	exon9	SNV	p.E332V; p.T355I
ERBB4	2	exon12	SNV	p.C467F; p.S454N
ERBB4	1	exon14	SNV	p.C559Y
ERBB4	1	exon16	SNV	p.T639M
ERBB4	2	exon17	SNV	p.A655D; p.A685D
ERBB4	1	exon18	SNV	p.R711C
ERBB4	1	exon2	SNV	p.L49F
ERBB4	1	exon20	SNV	p.H811Q
ERBB4	1	exon22	SNV	p.S905G
ERBB4	1	exon25	SNV	p.S1001N; p.D1000G
ERBB4	2	exon27	SNV	p.A1130T; p.P1084T
ERBB4	1	exon28	SNV	p.G1194D
ERBB4	1	exon3	SNV	p.R103C
ERBB4	1	exon5	SNV	p.R196C
ERBB4	1	exon7	SNV	p.F279V
ERCC2	1	exon10	SNV	p.R282Q
ERCC2	1	exon12	SNV	p.R378C
ERCC2	1	exon15	SNV	p.T484M
ERCC2	1	exon23	SNV	p.K751Q
ERCC2	1	exon4	SNV	p.T76I
ERCC2	1	exon9	SNV	p.E264K
ERCC3	1	exon2	SNV	p.E27Q
ERCC3	1	exon3	SNV	p.S144I
ERCC3	1	exon6	SNV	p.E259D
ERCC3	1	exon8	SNV	p.G414S

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
ERCC4	1	exon11	SNV	p.E725A
ERCC4	1	exon3	SNV	p.W193C
ERCC4	1	exon4	DEL	p.H215Yfs*27
ERCC4	1	exon5	SNV	p.T305I
ERCC4	1	exon7	DEL	p.K396del
ERCC4	2	exon8	SNV	p.L433F; p.R490W
ERCC5	1	exon13	SNV	p.E898D
ERCC5	2	exon8	SNV	p.V451A; p.D469N; p.E418*
ERF	1	exon2	SNV	p.Q28L
ERF	2	exon3	SNV	p.Y104F; p.L94V
ERF	1	exon4	DEL	p.G299Efs*12
ERF	1	exon4	INS	p.Q263Sfs*46
ERF	3	exon4	SNV	p.R546Q; p.T538M; p.F303V
ERG	1	exon12	DEL	p.P411Rfs*25
ERG	1	exon4	SNV	p.R57H
ERG	1	exon5	SNV	p.K118N
ERG	1	exon6	SNV	p.P138H
ERG	1	exon8	SNV	p.T248M
ERRFI1	1	exon4	DEL	p.C146Vfs*29
ESR1	7	exon10	SNV	p.Y537S; p.Y537D; p.D538G; p.Y537N
ESR1	2	exon3	SNV	p.E135D; p.Q75H
ESR1	1	exon4	SNV	p.D190Y
ESR1	1	exon6	SNV	p.R256Q
ESR1	1	exon7	SNV	p.E380Q
ETV1	1	exon10	SNV	p.D295N
ETV1	1	exon11	DEL	p.L323Wfs*2
ETV1	1	exon11	SNV	p.E352Q
ETV1	1	exon7	DEL	p.T121Hfs*116
ETV1	1	exon7	SNV	p.T125M
ETV1	4	exon8	SNV	p.T222I; p.A227V; p.T184M
ETV6	1	exon5	SNV	p.T187M
ETV6	1	exon8	SNV	p.F419I
EZH1	1	exon12	SNV	p.E439Q
EZH1	1	exon3	SNV	p.L29R

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
EZH2	1	exon10	SNV	p.S410L
EZH2	2	exon14	DEL	p.H530Ifs*13; p.D529Efs*7
EZH2	1	exon17	SNV	p.R659K
EZH2	1	exon6	SNV	p.E173G
EZH2	1	exon8	SNV	p.R298H
EZH2	1	exon9	SNV	p.E317K
FAM46C	1	exon2	SNV	p.T147M
FAM58A	2	exon2	SNV	p.L58I; p.R65C
FANCA	1	exon11	SNV	p.G300R
FANCA	2	exon13	SNV	p.E370*; p.R388T
FANCA	1	exon14	SNV	p.A430V
FANCA	1	exon21	SNV	p.A610T
FANCA	1	exon22	SNV	p.G658E
FANCA	1	exon23	SNV	p.Q703E
FANCA	1	exon29	SNV	p.S947L
FANCA	1	exon5	SNV	p.Q174*
FANCA	1	exon6	SNV	p.A181V
FANCA	1	exon7	SNV	p.A228V
FANCC	1	exon12	SNV	p.E380G
FANCC	1	exon4	SNV	p.W113C
FAT1	2	exon10	DEL	p.L2389Rfs*8; p.Q2286_T2291del
FAT1	2	exon10	INS	p.V2069Gfs*8; p.V1878Cfs*8
FAT1	5	exon10	SNV	p.V2689F; p.Q2074*; p.Q2784*; p.E2055D; p.A1709D
FAT1	1	exon13	SNV	p.F3125L
FAT1	1	exon16	SNV	p.E3401Q
FAT1	1	exon17	SNV	p.V3439I
FAT1	1	exon18	SNV	Splicing
FAT1	1	exon2	INS	p.I895Dfs*2
FAT1	1	exon2	DEL	p.N126Ifs*20
FAT1	5	exon2	SNV	Splicing; p.M1?; p.D1066H; p.E435Q; p.A49S; p.R443T; p.G60A; p.A1063T
FAT1	1	exon22	SNV	p.G3884E
FAT1	1	exon25	SNV	p.H4138Y
FAT1	2	exon27	SNV	p.S4461F; p.Q4458*

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
FAT1	1	exon3	SNV	p.E1150A; p.K1177T
FAT1	1	exon5	SNV	p.K1299R
FAT1	3	exon9	SNV	p.S1574*; p.R1570W; p.S1603L
FBXW7	6	exon10	SNV	p.R505G; p.R505L; p.G477S; p.R505C; p.R479Q
FBXW7	2	exon11	SNV	p.W606L; p.S582L
FBXW7	1	exon4	SNV	p.W237*
FBXW7	2	exon5	SNV	p.Q275*; p.S282*
FBXW7	1	exon7	SNV	p.Q358*
FBXW7	1	exon8	INS	p.T385Dfs*9
FBXW7	1	exon9	DEL	p.G437Dfs*61
FBXW7	3	exon9	SNV	p.S462F; p.G437V; p.R465H
FGF19	1	exon1	SNV	p.W17*
FGF19	1	exon3	SNV	p.H164N
FGF3	1	exon1	SNV	p.W16*
FGF3	2	exon3	SNV	p.S232C; p.R162W
FGF4	1	exon3	SNV	p.R192*
FGFR1	1	exon10	SNV	p.R475L
FGFR1	2	exon14	SNV	p.K656E; p.R622*
FGFR1	1	exon18	SNV	p.F780L
FGFR1	2	exon3	SNV	p.A74V; p.S107L
FGFR1	1	exon5	SNV	p.E196Q
FGFR2	2	exon12	SNV	p.N550K; p.N550D
FGFR2	1	exon4	SNV	p.D133N
FGFR2	2	exon5	SNV	p.G195R; p.A172T
FGFR2	2	exon7	SNV	p.S252W
FGFR2	3	exon9	SNV	p.Y376C; p.C383R
FGFR2	1	exonN/A	SNV	Splicing
FGFR3	1	exon10	SNV	p.A441T
FGFR3	1	exon12	SNV	p.N540K
FGFR3	1	exon18	SNV	p.P796L
FGFR3	3	exon3	SNV	p.H101Q; p.R93Q
FGFR3	1	exon4	SNV	p.E135K
FGFR3	3	exon7	SNV	p.S249C
FGFR4	1	exon12	SNV	p.N535K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
FGFR4	1	exon15	SNV	p.D663H
FGFR4	1	exon16	SNV	p.S688F
FGFR4	1	exon17	SNV	p.P734H
FH	1	exon1	SNV	p.Y2*
FH	2	exon2	SNV	p.D55G; p.E53*
FH	1	exon9	SNV	p.G424R
FLCN	2	exon4	SNV	p.L19F; p.N2H
FLCN	1	exon8	SNV	p.E280*
FLT1	1	exon10	SNV	p.W466C
FLT1	1	exon11	SNV	p.R508H
FLT1	1	exon19	SNV	p.A869D
FLT1	1	exon2	SNV	p.D31Y
FLT1	1	exon20	SNV	p.E910Q
FLT1	1	exon28	SNV	p.R1224K
FLT1	1	exon4	SNV	p.E144K
FLT1	1	exon5	SNV	p.R183C
FLT1	1	exon8	SNV	p.Q341E
FLT1	1	exon9	SNV	Splicing
FLT3	1	exon14	SNV	p.E608*
FLT3	1	exon16	SNV	p.G679W
FLT3	1	exon18	SNV	p.I745V
FLT3	1	exon2	SNV	p.G48E
FLT3	1	exon20	SNV	p.T820I
FLT3	1	exon22	SNV	p.Q910*
FLT3	1	exon23	SNV	p.C925S
FLT3	1	exon24	DEL	p.P986Afs*27
FLT3	1	exon6	SNV	p.C232*
FLT4	1	exon13	SNV	p.G603W
FLT4	1	exon14	SNV	p.W711R
FLT4	1	exon15	SNV	p.V750M
FLT4	1	exon2	SNV	p.T35M
FLT4	1	exon21	SNV	p.E989Q
FLT4	4	exon3	SNV	p.A103V; p.A125T; p.E121*; p.A89V
FLT4	3	exon30	SNV	p.P1343S; p.A1353T; p.G1328*

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
FLT4	2	exon6	SNV	p.E228Q; p.R237T; p.R237S
FOXA1	6	exon2	SNV	p.D158N; p.D249V; p.M41I; p.N135K; p.F266S; p.K414T
FOXA1	2	exon2	DEL	p.K267_S280del; p.M253_N256del
FOXA1	1	exon2	INS	p.L379Tfs*11
FOXL2	7	exon1	SNV	p.C176Y; p.D184H; p.D184N; p.C134W; p.P157A; p.L62P; p.D50N
FOXO1	4	exon1	SNV	p.N73K; p.E3Q; p.V94A; p.E174G
FOXO1	4	exon2	SNV	p.W295*; p.E334K; p.T648M; p.V635A
FOXO1	1	exon2	INS	p.A271Gfs*35
FOXP1	1	exon12	SNV	p.E295K
FOXP1	1	exon14	SNV	p.Q381*
FOXP1	3	exon15	SNV	p.V417I; p.S396L; p.P434L
FOXP1	1	exon19	DEL	p.L569Qfs*10
FOXP1	1	exon21	SNV	p.L649M
FOXP1	1	exon6	SNV	p.R33W
FOXP1	1	exon9	DEL	p.K149Nfs*57
FUBP1	1	exon12	SNV	p.R331Q
FUBP1	1	exon17	SNV	p.A538V
FYN	1	exon10	SNV	p.P308S
GATA1	2	exon3	SNV	p.A184V; p.F172L
GATA1	1	exon4	SNV	p.H232D
GATA1	2	exon6	SNV	p.G368C
GATA2	1	exon5	SNV	p.A302S
GATA2	1	exon6	SNV	p.R343K
GATA3	1	exon2	SNV	p.V60I
GATA3	3	exon6	SNV	p.M401V; p.S430C; p.Q363*
GATA3	4	exon6	INS	p.P409Afs*99; p.M401Nfs*107
GLI1	3	exon12	SNV	p.D677G; p.E778*; p.P566S
GLI1	1	exon4	SNV	p.R100H
GLI1	1	exon5	SNV	p.Q178H
GLI1	1	exon7	SNV	p.G241V
GNA11	1	exon2	SNV	p.A89V
GNA11	1	exon3	SNV	p.D155N
GNA11	1	exon5	SNV	p.V240L

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
GNAS	3	exon8	SNV	p.R202C; p.R202H
GPS2	2	exon11	SNV	p.Q320*; p.R312W
GPS2	1	exon7	SNV	p.S183C
GRIN2A	1	exon10	SNV	p.L665P
GRIN2A	1	exon11	SNV	p.N696K
GRIN2A	1	exon13	SNV	p.E803K
GRIN2A	11	exon14	SNV	p.K1116T; p.L930F; p.T1064M; p.Q926*; p.F1344S; p.V1453M; p.E1205K; p.K1078N; p.L975F; p.S906N; p.R1285K; p.E962K; p.R1288H
GRIN2A	5	exon3	SNV	p.A131T; p.A33V; p.L82I; p.A33T; p.D45N
GRIN2A	1	exon4	SNV	p.I204M
GRIN2A	1	exon5	SNV	p.D347N
GRIN2A	1	exon6	SNV	p.T426P
GRIN2A	1	exon8	SNV	p.V521M
GRIN2A	1	exon9	SNV	Splicing
GSK3B	1	exon10	SNV	p.R321*
GSK3B	1	exon3	SNV	p.L104V
H3F3A	1	exon2	SNV	p.G35R
H3F3B	1	exon2	SNV	p.S32F
H3F3C	2	exon1	SNV	p.A8S; p.Y41*
HGF	1	exon1	SNV	p.L7M
HGF	1	exon10	SNV	p.L403F
HGF	1	exon11	SNV	p.R441Q
HGF	1	exon12	SNV	p.T476S
HGF	1	exon13	SNV	p.R494Q
HGF	1	exon14	SNV	p.W528C
HGF	1	exon17	SNV	p.L634I
HGF	1	exon18	SNV	p.G674D
HGF	1	exon4	SNV	p.S141Y
HGF	2	exon5	SNV	p.R181*; p.E183G
HGF	1	exon6	SNV	p.C232Y
HGF	1	exon8	SNV	p.D297Y
HGF	1	exon9	SNV	Splicing
HIST1H1C	3	exon1	SNV	p.T167I; p.E53K; p.P161S

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
HIST1H3A	2	exon1	SNV	p.T4S; p.E51K
HIST1H3B	2	exon1	SNV	p.A30T; p.E51D
HIST1H3C	2	exon1	SNV	p.H40Y; p.R130H
HIST1H3D	1	exon2	SNV	p.E98K
HIST1H3H	1	exon1	SNV	p.S58C
HIST1H3I	2	exon1	SNV	p.A128S; p.M121I
HIST1H3J	1	exon1	DEL	p.L49Sfs*22
HIST1H3J	2	exon1	SNV	p.I113V; p.I52F
HNF1A	2	exon1	SNV	p.L38Q; p.G51D
HNF1A	2	exon2	SNV	p.A174V; p.T156R
HNF1A	2	exon4	DEL	p.P291Qfs*51
HNF1A	1	exon5	SNV	p.G355S
HNF1A	1	exon6	SNV	p.H387P
HNF1A	1	exon7	SNV	p.P499H
HOXB13	3	exon1	SNV	p.T73K; p.E119Q; p.R25Q
HOXB13	1	exon2	SNV	p.S224N
HRAS	1	exon2	SNV	p.M1?
HRAS	2	exon3	SNV	p.Q61R; p.Q61K
HRAS	1	exon4	SNV	p.V114M; p.Q99*
ICOSLG	1	exon4	SNV	p.G148*
ICOSLG	1	exon7	SNV	p.V217I
ICOSLG	1	exonN/A	SNV	Splicing
ID3	3	exon1	SNV	p.E35Q; p.C16S; p.P95L
IDH1	3	exon4	SNV	p.R132H; p.R132C
IFNGR1	1	exon7	SNV	p.P367L
IGF1R	1	exon1	SNV	p.S24W
IGF1R	1	exon12	SNV	p.G870R
IGF1R	1	exon2	SNV	p.E193K
IGF1R	1	exon21	SNV	p.M1254I
IGF1R	1	exon3	SNV	p.A223V
IGF2	1	exon2	SNV	p.E14K
IGF2	3	exon3	SNV	p.R83H; p.R104C; p.A75T
IKBKE	1	exon10	SNV	p.A371T
IKBKE	1	exon13	SNV	p.R456Q

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
IKBKE	1	exon16	SNV	p.L547S
IKBKE	1	exon21	SNV	p.E688K
IKBKE	1	exon6	SNV	p.E177Q
IKBKE	1	exon9	SNV	p.A300V
IKZF1	1	exon2	SNV	p.D4N
IKZF1	2	exon3	SNV	p.D25Y; p.S41L
IL10	1	exon3	SNV	Splicing
IL10	1	exon5	SNV	p.F164L
IL7R	1	exon2	SNV	p.H53N
IL7R	1	exon5	SNV	p.E230D
IL7R	1	exon6	SNV	p.W264*
IL7R	1	exon8	SNV	p.S420F
INHA	1	exon1	SNV	p.E23Q
INHA	1	exon2	SNV	p.P192L
INHBA	3	exon3	SNV	p.R166T; p.R309Q; p.E351D
INPP4A	1	exon15	SNV	p.A416T
INPP4A	1	exon17	SNV	p.E557D
INPP4A	1	exon20	DEL	p.L697_G701del
INPP4A	1	exon22	SNV	Splicing
INPP4A	1	exon23	SNV	p.T816A
INPP4A	1	exon26	SNV	p.A952V
INPP4A	1	exon5	SNV	p.S53N
INPP4A	1	exon9	DEL	p.L220Afs*19
INPP4B	1	exon20	SNV	p.T621M
INPP4B	1	exon22	SNV	p.M682T
INPP4B	1	exon25	SNV	p.K816N
INPP4B	1	exon26	SNV	p.S850W
INPPL1	1	exon12	DEL	p.N441Tfs*2
INPPL1	1	exon14	SNV	p.K541Q
INPPL1	1	exon15	SNV	p.L597V
INPPL1	1	exon2	SNV	p.E76A
INPPL1	2	exon21	SNV	p.S780N; p.Q786*
INPPL1	1	exon23	SNV	p.G835E
INPPL1	2	exon25	SNV	p.R960K; p.A924D

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
INPPL1	1	exon26	SNV	p.D1180N
INPPL1	2	exon26	DEL	p.R1156Gfs*46; p.G1157Dfs*45
INPPL1	1	exon4	DEL	p.P140Rfs*39
INPPL1	1	exon9	SNV	p.E339Q
INSR	1	exon10	SNV	p.F732S
INSR	1	exon12	SNV	p.N769S
INSR	1	exon2	SNV	p.E71K
INSR	1	exon20	SNV	p.G1179*
IRF4	1	exon4	SNV	p.E142D
IRF4	1	exon5	SNV	p.P183L
IRF4	2	exon6	SNV	p.E227A; p.E243K
IRF4	1	exon7	INS	p.Y325*
IRF4	1	exon7	SNV	p.Q363H
IRF4	1	exon9	SNV	p.Y415H
IRS1	1	exon1	SNV	p.R213C; p.E895D
IRS1	3	exon1	DEL	p.H598Tfs*38; p.Y765del
IRS1	1	exon1	INS	p.S686_S680ins
JAK1	2	exon10	SNV	p.E456K; p.D471N
JAK1	1	exon14	SNV	p.D660N
JAK1	2	exon15	SNV	p.V685A; p.R681W
JAK1	1	exon16	SNV	p.R724H
JAK1	3	exon19	DEL	p.K860Nfs*16
JAK1	1	exon22	SNV	p.R997Q
JAK1	1	exon24	SNV	p.G1097D
JAK1	1	exon25	SNV	p.R1139W
JAK1	1	exon3	SNV	p.A57T
JAK1	1	exon7	INS	p.V331Kfs*6
JAK2	1	exon11	INS	p.N457Kfs*2
JAK2	1	exon13	SNV	p.R564*
JAK2	1	exon20	SNV	p.D894V
JAK2	1	exon7	SNV	p.K269N
JAK3	1	exon10	SNV	p.R451Q
JAK3	1	exon12	SNV	p.G546W
JAK3	1	exon16	SNV	p.M724I

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
JAK3	1	exon20	SNV	p.R899W
JAK3	1	exon21	SNV	p.G987D
JAK3	1	exon4	SNV	p.K122N
JAK3	1	exon9	SNV	p.R403H
JUN	5	exon1	SNV	p.L98M; p.P229L; p.I33F; p.K56N; p.R276G
KDM5A	1	exon10	SNV	p.G427R
KDM5A	1	exon11	SNV	p.S479C
KDM5A	1	exon14	SNV	p.E614D
KDM5A	1	exon15	SNV	p.S686P
KDM5A	1	exon22	DEL	p.V1093*
KDM5A	4	exon23	SNV	p.A1272S; p.S1196F; p.E1293K; p.L1236F
KDM5A	1	exon28	SNV	p.E1651*
KDM5A	1	exon4	SNV	p.E131Q
KDM5A	1	exon7	SNV	p.A274T
KDM5C	1	exon12	SNV	p.P573L
KDM5C	1	exon14	DEL	p.S643Pfs*16
KDM5C	1	exon14	SNV	p.A652T
KDM5C	1	exon16	SNV	p.L781P
KDM5C	1	exon19	DEL	p.R888_S893del
KDM5C	1	exon20	SNV	p.R1030Q
KDM5C	1	exon21	SNV	p.A1078V
KDM5C	1	exon23	SNV	p.R1165H
KDM5C	1	exon3	DEL	p.S99Qfs*23
KDM5C	1	exon6	SNV	p.R258W
KDM5C	1	exon7	SNV	p.M308I
KDM6A	1	exon12	SNV	p.D370N
KDM6A	1	exon13	SNV	p.A429T
KDM6A	1	exon18	INS	p.S944Tfs*15
KDM6A	1	exon18	DEL	p.G698Afs*9
KDM6A	1	exon19	SNV	Splicing
KDM6A	1	exon24	SNV	p.V1165I
KDM6A	1	exon24	DEL	p.L1171del
KDM6A	1	exon25	SNV	p.I1209K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
KDM6A	1	exon27	SNV	p.I1333T
KDM6A	2	exon29	INS	p.G1419Efs*11; p.D1412Gfs*29
KDM6A	1	exon9	SNV	p.V241I
KDR	1	exon10	SNV	p.Q461L
KDR	1	exon13	SNV	p.R549K
KDR	2	exon14	SNV	p.D703Y; p.D703H
KDR	1	exon15	SNV	p.E732K
KDR	1	exon21	SNV	p.S974R
KDR	2	exon23	SNV	p.S1037L; p.R1027S
KDR	1	exon25	SNV	p.M1125L
KDR	1	exon29	SNV	p.L1264P
KDR	1	exon7	SNV	p.K271R
KDR	1	exon8	SNV	p.E326G
KEAP1	5	exon2	SNV	p.D180N; p.M209T; p.A63V; p.D87V; p.D78G
KEAP1	4	exon3	SNV	p.V271M; p.R415H; p.A344T; p.C249F; p.L281Q
KEAP1	2	exon4	SNV	p.R460G; p.L484F
KEAP1	1	exon5	DEL	p.E542Dfs*31
KIT	1	exon11	SNV	p.V560D
KIT	2	exon11	DEL	p.W557_V559delinsC; p.Y568_L576delinsF
KIT	1	exon12	SNV	p.T594A
KIT	1	exon20	SNV	p.I920S
KIT	3	exon3	SNV	p.R181W; p.S197L; p.L156V
KIT	1	exon9	INS	p.Y503_A502ins
KLF4	1	exon3	SNV	p.G174D
KLF4	1	exon4	SNV	p.E378D
KMT2A	1	exon20	SNV	p.R1858Q
KMT2A	1	exon22	SNV	p.R1963*
KMT2A	3	exon27	SNV	p.G2633R; p.Q3454*; p.G2959R
KMT2A	1	exon27	DEL	p.S2693Lfs*64
KMT2A	6	exon3	SNV	p.A707T; p.S479C; p.D368Y; p.P603A; p.P561S; p.S316*; p.Q491E; p.H753Y; p.R613Q; p.S783C; p.P553S
KMT2A	1	exon3	DEL	p.P773Rfs*8

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
KMT2A	1	exon36	SNV	p.I3921S
KMT2A	1	exon4	SNV	p.R1081*
KMT2A	1	exon5	SNV	p.N1183I
KMT2A	1	exon6	SNV	Splicing
KMT2A	1	exon7	SNV	p.E1269K
KMT2B	1	exon11	SNV	p.P1197S
KMT2B	1	exon14	SNV	p.C1297Y; p.A1299V
KMT2B	1	exon23	SNV	p.C1666F
KMT2B	1	exon26	SNV	p.R1762C
KMT2B	3	exon27	DEL	p.G1879Vfs*16
KMT2B	6	exon28	SNV	p.N2195I; p.S2307*; p.P2049S; p.P2226L; p.E1981D; p.E2059K
KMT2B	5	exon3	SNV	p.P597T; p.G723R; p.A228V; p.G273S; p.S423C
KMT2B	1	exon31	SNV	p.E2429D
KMT2B	2	exon37	SNV	p.C2655Y; p.G2706D
KMT2B	1	exon4	SNV	p.R843W
KMT2B	1	exon8	SNV	p.A1076P
KMT2C	1	exon11	SNV	p.E495K
KMT2C	3	exon14	SNV	p.R833I; p.E746Q; p.P828Q
KMT2C	1	exon17	SNV	p.E936*
KMT2C	3	exon20	SNV	p.A1079T; p.T1076R; p.R1092*
KMT2C	1	exon27	DEL	p.P1406Lfs*3
KMT2C	1	exon28	SNV	p.D1433N
KMT2C	1	exon29	DEL	p.N1477Rfs*6
KMT2C	1	exon33	INS	p.K1665Pfs*12
KMT2C	1	exon34	SNV	p.A1685S
KMT2C	1	exon35	SNV	p.R1705C
KMT2C	3	exon36	DEL	p.R2240Sfs*29; p.L2067Cfs*11; p.P2356Qfs*3
KMT2C	5	exon36	SNV	p.R1890Q; p.M2373V; p.Q1780H; p.R2382K; p.D2270N
KMT2C	5	exon38	SNV	p.S2638Y; p.G3014E; p.E3065Q; p.S2691F; p.A2660G; p.E2844*; p.E2866Q
KMT2C	1	exon38	DEL	p.S2658*
KMT2C	2	exon38	INS	p.E2798Gfs*11; p.S2984Ffs*18

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
KMT2C	2	exon4	SNV	p.I155M; p.R190Q
KMT2C	1	exon42	SNV	p.E3239*
KMT2C	3	exon43	SNV	p.L3780F; p.R3507*; p.E3662*
KMT2C	2	exon43	DEL	p.F3501Hfs*4; p.P3766Lfs*12
KMT2C	1	exon46	SNV	p.G3965C
KMT2C	1	exon48	SNV	p.E4092K
KMT2C	1	exon51	SNV	p.R4225*
KMT2C	3	exon52	SNV	p.H4512D; p.A4488T; p.C4503W
KMT2C	1	exon52	DEL	p.K4351Nfs*5
KMT2C	1	exon54	SNV	p.R4693Q
KMT2C	1	exon59	SNV	p.F4890S
KMT2D	5	exon10	SNV	p.E731K; p.Q928*; p.L720P; p.P439S; p.Q800*; p.P763L; p.P478T
KMT2D	2	exon10	DEL	p.P528Hfs*402; p.L725Rfs*205
KMT2D	2	exon11	SNV	p.P1084S; p.L935I
KMT2D	1	exon12	SNV	p.P1314L; p.P1314S
KMT2D	1	exon14	SNV	p.Q1402*
KMT2D	1	exon2	SNV	p.S48N
KMT2D	1	exon20	SNV	p.Y1692H
KMT2D	1	exon23	SNV	p.S1812*
KMT2D	1	exon24	INS	p.T1843Yfs*5
KMT2D	1	exon27	DEL	p.L1953Gfs*3
KMT2D	1	exon27	SNV	p.F1952L
KMT2D	1	exon28	SNV	p.D1970N
KMT2D	1	exon29	SNV	p.S2039*
KMT2D	2	exon3	SNV	Splicing; p.W91C
KMT2D	2	exon31	DEL	p.P2230Hfs*34; p.P2354Lfs*30
KMT2D	4	exon31	SNV	p.E2270*; p.E2544*; p.P2390A; p.G2490W
KMT2D	2	exon32	SNV	p.R2697H; p.R2685*
KMT2D	2	exon34	DEL	p.K3140Rfs*2; p.V3089Wfs*30
KMT2D	1	exon34	INS	p.L3367Ffs*56
KMT2D	3	exon34	SNV	p.L3150P; p.E3181K; p.P2987S
KMT2D	5	exon39	SNV	p.S3713L; p.N3590K; p.T4193M; p.Q4464*; p.R4162W

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
KMT2D	2	exon39	DEL	p.K3586del; p.N3597Tfs*61
KMT2D	1	exon4	SNV	p.E156*
KMT2D	1	exon41	SNV	p.S4574N
KMT2D	1	exon42	SNV	p.E4646K
KMT2D	1	exon44	SNV	p.S4701F
KMT2D	1	exon47	SNV	p.G4847A
KMT2D	3	exon48	SNV	p.R5097Q; p.G5189R; p.R5031C
KMT2D	2	exon51	SNV	p.E5425K; p.E5394K
KMT2D	2	exon6	SNV	p.W268L; p.W268*
KMT2D	1	exon7	INS	p.C302Lfs*40
KRAS	66	exon2	SNV	p.G13D; p.G12D; p.G12C; p.G13C; p.G12A; p.G12V; p.G12R; p.G12S; p.G13R; p.D33E; p.L6I
KRAS	7	exon3	SNV	p.Q61R; p.Q61H
KRAS	2	exon4	SNV	p.A146T; p.A146V
KRAS	1	exon5	SNV	p.E174Q
LATS1	1	exon5	SNV	p.K738N
LATS1	1	exon8	SNV	p.P1044L
LATS2	4	exon4	SNV	p.P350L; p.Q618H; p.S425Y; p.R558C
LATS2	1	exon5	SNV	p.R700M
LATS2	1	exon7	SNV	p.Q910*
LATS2	2	exon8	SNV	p.V1088M; p.G1060R
LMO1	1	exon3	SNV	p.R121T
LMO1	1	exon4	SNV	p.E142*
LYN	1	exon13	SNV	p.S495R
LYN	1	exon8	SNV	p.R244W
LYN	1	exon8	DEL	p.I247Wfs*12
MALT1	1	exon10	SNV	p.M351I
MAP2K2	1	exon11	SNV	p.S372P
MAP2K2	1	exon3	SNV	p.I107M
MAP2K2	1	exon9	SNV	p.P330H
MAP2K4	1	exon1	SNV	p.S16G
MAP2K4	1	exon4	SNV	p.V141F
MAP3K1	1	exon1	SNV	p.P113L
MAP3K1	1	exon10	SNV	p.A634V

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
MAP3K1	1	exon13	SNV	p.W751*
MAP3K1	1	exon13	DEL	p.R790Gfs*9
MAP3K1	2	exon14	SNV	p.G1068C; p.L1122*
MAP3K1	1	exon14	DEL	p.I1219del
MAP3K1	1	exon16	DEL	p.Y1276Cfs*7
MAP3K1	2	exon4	SNV	p.I322M; p.R288*
MAP3K13	4	exon12	SNV	p.P761T; p.G571R; p.G716D; p.R592Q
MAP3K13	5	exon3	SNV	p.S12F; p.V105M; p.L131F; p.T23N; p.Q99*
MAP3K13	3	exon5	SNV	p.Y269C; p.D279Y; p.I232T
MAP3K13	2	exon6	SNV	p.F313I; p.E323Q
MAP3K14	1	exon10	SNV	p.L557P
MAP3K14	1	exon12	SNV	p.G701C
MAP3K14	1	exon16	SNV	p.R918Q
MAP3K14	1	exon2	SNV	p.A77V
MAP3K14	1	exon3	SNV	p.R99P
MAP3K14	1	exon5	SNV	p.M328I
MAP3K14	1	exon7	SNV	p.G432S
MAPK1	1	exon2	SNV	p.K55R
MAPK1	1	exon3	SNV	p.N158S
MAPK3	1	exon3	SNV	p.L163V
MAPK3	1	exon6	SNV	p.R278*
MAPKAP1	1	exon12	SNV	p.T509M
MAPKAP1	1	exon2	SNV	p.M46V
MAPKAP1	1	exon7	SNV	p.Q316H
MAPKAP1	1	exon9	SNV	p.H381P
MAX	1	exon3	SNV	p.D37V
MAX	1	exon4	DEL	p.H81Pfs*5
MAX	5	exon4	SNV	p.E69V; p.G99W; Splicing; p.R76T
MCL1	2	exonN/A	SNV	Splicing
MDM2	2	exon11	DEL	p.T371Lfs*2; p.L321Ffs*52
MDM2	1	exon11	SNV	p.E316V
MDM2	1	exon8	SNV	p.T224M
MDM4	2	exon11	SNV	p.T454M; p.S367L
MDM4	1	exon4	SNV	p.S89N

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
MED12	2	exon10	SNV	p.R470H; p.E495K
MED12	1	exon11	SNV	p.R521H
MED12	1	exon13	SNV	p.F593L
MED12	1	exon16	SNV	p.E743*
MED12	1	exon21	SNV	p.S994I
MED12	1	exon23	SNV	p.I1074F
MED12	1	exon26	SNV	p.A1222S
MED12	1	exon27	SNV	p.G1237S; p.G1237V
MED12	1	exon3	SNV	p.R82C
MED12	1	exon30	SNV	Splicing
MED12	1	exon31	SNV	p.E1450K
MED12	1	exon37	SNV	p.A1736T
MED12	1	exon40	SNV	p.S1934C
MED12	1	exon5	SNV	p.Y193*
MED12	1	exon6	SNV	p.G268V
MED12	1	exon8	SNV	p.L371M
MEF2B	1	exon9	SNV	p.R345W
MEN1	1	exon10	SNV	p.R489Q
MEN1	1	exon2	DEL	p.S15del
MEN1	4	exon2	SNV	p.R98*; p.M1?; p.I140M; p.W126*
MEN1	1	exon3	SNV	p.G161S
MET	1	exon11	SNV	p.D842Y
MET	1	exon12	SNV	p.K894Q
MET	1	exon13	SNV	p.V937F
MET	2	exon14	SNV	p.T1011I; p.R1005Q; p.R988P
MET	1	exon19	SNV	p.K1237N
MET	5	exon2	SNV	p.V176M; p.Q232*; p.K306N; p.S353F; p.Y321F
MET	1	exon6	SNV	p.S609N
MGA	1	exon11	SNV	p.R1246*
MGA	1	exon13	SNV	p.S1320F
MGA	2	exon15	SNV	p.A1596V; p.V1582F
MGA	4	exon17	SNV	p.E1971*; p.K2108T; p.V2214A; p.Q1967*
MGA	1	exon17	DEL	p.L2167Qfs*26

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
MGA	2	exon2	SNV	p.H159Y; p.G39V
MGA	3	exon20	SNV	p.R2425C; p.R2436H
MGA	1	exon20	DEL	Splicing
MGA	1	exon24	SNV	p.T2874A
MGA	1	exon3	SNV	p.E392K
MGA	1	exon3	DEL	p.K542Nfs*5
MGA	1	exon6	SNV	p.D744H
MGA	1	exon8	SNV	p.R896*
MITF	1	exon5	SNV	p.I155F
MITF	1	exon6	SNV	p.S194C
MITF	1	exon9	SNV	p.E322Q
MITF	2	exonN/A	SNV	p.V92M; p.P91L
MLH1	1	exon1	SNV	p.G22E
MLH1	1	exon10	DEL	p.N287Tfs*10
MLH1	2	exon12	SNV	Splicing; p.P435S
MLH1	1	exon13	SNV	p.E512*
MLH1	1	exon16	SNV	p.W597*
MLH1	1	exon17	SNV	p.P649R
MLH1	1	exon19	SNV	p.I719V
MPL	1	exon12	SNV	p.S566N
MPL	1	exon5	SNV	p.G262V
MPL	1	exon7	SNV	p.A388D
MRE11A	1	exon12	SNV	p.N413I
MRE11A	1	exon13	SNV	p.E460*
MRE11A	1	exon3	DEL	p.D9del
MRE11A	1	exon5	SNV	p.V122L
MSH2	2	exon1	SNV	p.Q24*
MSH2	1	exon11	SNV	p.E580*
MSH2	1	exon12	SNV	p.Q601*
MSH2	1	exon15	SNV	p.E852K
MSH2	1	exon3	SNV	p.V200G
MSH2	1	exon4	INS	p.A230Sfs*2
MSH2	1	exon6	SNV	p.Q337*
MSH3	1	exon21	SNV	p.M953L

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
MSH3	1	exon3	DEL	p.K136Nfs*2
MSH3	2	exon6	SNV	p.R328W; p.R322K
MSH3	2	exon8	SNV	p.A409V; p.G398D
MSH6	1	exon3	SNV	p.Q177L
MSH6	3	exon4	SNV	p.R1035Q; p.R482Q; p.A680V; p.L798I
MSH6	1	exon5	DEL	p.F1088Sfs*2
MSH6	4	exon5	SNV	p.P1082T; Splicing; p.R1095C; p.P1082Q; p.T1142M
MSH6	2	exon8	SNV	p.V1232A; p.E1234*
MSH6	2	exon9	SNV	p.R1331Q; p.S1329P
MSI1	1	exon11	SNV	p.R247G
MSI1	1	exon9	SNV	p.A196D
MSI2	2	exon6	SNV	p.R108T; p.T107R
MST1	1	exon2	SNV	p.P59L; p.P59S
MST1	1	exon5	INS	p.C171Lfs*3
MST1R	1	exon14	SNV	p.L1029V
MST1R	1	exon19	SNV	p.S1315Y
MST1R	1	exon6	SNV	p.H667Y
MST1R	1	exon7	SNV	p.V689E
MTOR	2	exon13	SNV	p.L718P; p.E681K
MTOR	1	exon25	DEL	p.T1252Qfs*4
MTOR	1	exon32	SNV	p.R1538Q
MTOR	1	exon35	SNV	p.T1652I
MTOR	2	exon4	SNV	p.E124G; p.L115F
MTOR	2	exon5	SNV	p.R173H; p.Q186*
MTOR	1	exon6	SNV	Splicing
MTOR	4	exon7	SNV	p.E288*; p.D371H; p.S348F; p.L330W
MTOR	1	exon8	SNV	p.R381S
MUTYH	2	exon12	SNV	p.N357S; p.G337*
MUTYH	1	exon13	SNV	p.H438D
MUTYH	1	exon16	SNV	p.N531T
MUTYH	1	exon3	SNV	p.A55T
MYC	2	exon2	SNV	p.S235P; p.F153Y
MYC	1	exon3	SNV	p.V376L
MYCN	1	exon2	SNV	p.P60S

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
MYCN	3	exon3	SNV	p.R286C; p.R302H; p.K349N
MYD88	2	exon1	SNV	p.D113H; p.A6V
MYD88	2	exon5	SNV	p.A313T; p.L273P
MYOD1	2	exon1	SNV	p.P181T; p.A174S
MYOD1	1	exon3	SNV	p.K242N
NBN	1	exon1	SNV	p.W2*
NBN	1	exon10	DEL	p.R466Gfs*18
NBN	1	exon11	INS	p.R551Kfs*5
NBN	1	exon14	SNV	p.D707N
NBN	1	exon3	SNV	p.T67I
NBN	1	exon4	SNV	p.I132M
NCOA3	1	exon10	SNV	Splicing
NCOA3	3	exon11	SNV	p.R485H; p.I480M; p.S402N
NCOA3	2	exon12	SNV	p.K732R; p.S660F
NCOA3	1	exon18	SNV	p.Q1145R
NCOA3	1	exon4	SNV	p.E47*
NCOA3	1	exon9	SNV	p.Y319N
NCOR1	1	exon10	SNV	p.Q345R
NCOR1	1	exon10	DEL	p.E354Tfs*21
NCOR1	1	exon12	SNV	p.D433N
NCOR1	1	exon15	INS	p.T525Nfs*11
NCOR1	1	exon19	SNV	p.R688Q
NCOR1	3	exon20	SNV	p.E735K; p.S771R; p.E854G; p.D812G
NCOR1	1	exon21	SNV	p.S917*
NCOR1	1	exon23	SNV	p.L1036V; p.R1028G; p.P1032T
NCOR1	2	exon24	SNV	p.L1088V; p.Q1071*
NCOR1	1	exon3	SNV	p.E74Q
NCOR1	1	exon30	SNV	p.R1326Q
NCOR1	2	exon31	SNV	p.G1416E; p.D1504N
NCOR1	1	exon37	SNV	p.R1922T
NCOR1	1	exon4	SNV	p.K107R
NEGR1	1	exon1	SNV	p.M3I
NEGR1	1	exon3	SNV	p.V156I
NEGR1	1	exon4	SNV	p.E183D

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
NF1	1	exon1	SNV	p.M1?
NF1	1	exon12	DEL	p.I460Gfs*7
NF1	2	exon12	SNV	p.R440*
NF1	1	exon18	INS	p.N736*
NF1	2	exon21	SNV	p.G848E; p.E836*
NF1	1	exon29	SNV	p.R1306*
NF1	1	exon30	SNV	Splicing
NF1	1	exon35	SNV	p.W1559*
NF1	1	exon39	SNV	p.T1905I
NF1	1	exon40	DEL	p.M1981Ifs*31
NF1	1	exon42	SNV	p.Y2102F
NF1	2	exon48	SNV	p.N2385I; p.P2366S
NF1	1	exon53	SNV	p.E2607K
NF1	1	exon9	SNV	p.Q347*
NF2	2	exon12	SNV	p.T391A; p.R424H
NF2	1	exon2	SNV	p.R52W
NF2	1	exon3	SNV	p.E89K
NFE2L2	1	exon2	SNV	p.G81S; p.G81D
NFE2L2	3	exon5	SNV	p.S310F; p.S399Y; p.S323*; p.Y286*
NFKBIA	2	exon1	SNV	p.Q50*; p.E14G
NFKBIA	1	exon5	INS	p.T247Yfs*38
NKX2-1	1	exon2	SNV	p.A96T
NKX2-1	1	exon3	INS	p.S311Kfs*128
NKX2-1	1	exon3	SNV	p.G339S
NKX3-1	1	exon3	SNV	p.Q71H
NKX3-1	1	exonN/A	SNV	Splicing
NOTCH1	1	exon1	SNV	p.R20Q
NOTCH1	1	exon18	SNV	p.N918Y
NOTCH1	1	exon19	SNV	p.G1034S
NOTCH1	1	exon20	SNV	p.D1095Y
NOTCH1	1	exon25	SNV	p.W1497*
NOTCH1	1	exon26	SNV	p.E1636K
NOTCH1	1	exon30	SNV	p.A1857T
NOTCH1	1	exon31	SNV	p.D1925N

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
NOTCH1	1	exon34	DEL	p.S2486Rfs*103
NOTCH1	3	exon34	SNV	p.G2345V; p.S2215L; p.Q2404*
NOTCH1	1	exon4	SNV	p.L246R
NOTCH1	1	exon5	DEL	p.R282Afs*19
NOTCH1	1	exon7	SNV	p.S385F
NOTCH1	3	exon8	SNV	p.E424K; p.P422S; p.G427D
NOTCH1	2	exon9	SNV	p.E483K; p.K508N
NOTCH2	1	exon16	SNV	p.E843G
NOTCH2	1	exon21	SNV	p.C1137F
NOTCH2	1	exon22	SNV	p.F1209L
NOTCH2	1	exon24	SNV	p.M1311I
NOTCH2	1	exon25	SNV	p.P1371S
NOTCH2	1	exon33	SNV	p.T1993N
NOTCH2	8	exon34	SNV	p.R2053C; p.S2145*; p.K2109N; p.H2096Q; p.Q2452*; p.R2298W; p.P2087L
NOTCH2	1	exon5	SNV	Splicing
NOTCH2	1	exon6	SNV	p.W330C
NOTCH2	3	exon7	SNV	p.T392I; p.L375V; p.P383S
NOTCH3	1	exon1	SNV	p.V24L
NOTCH3	1	exon10	SNV	p.E510K
NOTCH3	2	exon18	SNV	p.G975C; p.G937E
NOTCH3	1	exon22	SNV	p.V1183M
NOTCH3	2	exon25	SNV	p.D1481N; p.R1571Q
NOTCH3	1	exon26	SNV	p.Q1593*
NOTCH3	3	exon33	SNV	p.G2112S; p.P2101H; p.P2169L
NOTCH3	2	exon33	DEL	p.E2250Nfs*79; p.G2035Vfs*50
NOTCH3	1	exon5	SNV	p.V252L
NPM1	1	exon9	SNV	p.Q252E
NRAS	3	exon2	SNV	p.G13D; p.G12D
NRAS	5	exon3	SNV	p.Q61R; p.D92N; p.Q61H; p.Q61L; p.Q61K
NSD1	1	exon10	SNV	p.R1473Q
NSD1	1	exon10	DEL	p.V1486*
NSD1	1	exon11	DEL	p.M1531Cfs*43

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
NSD1	1	exon16	DEL	p.K1786Efs*3
NSD1	1	exon16	SNV	p.E1817*
NSD1	1	exon18	DEL	p.K1964Rfs*5
NSD1	2	exon18	SNV	p.I1884T; p.R1952Q
NSD1	5	exon2	SNV	p.K147N; p.E171K; p.S80C; p.D64H; p.L43R
NSD1	5	exon23	SNV	p.G2642E; p.Q2163*; p.R2187*; p.L2236P; p.A2659T
NSD1	8	exon5	SNV	p.E1147*; p.I447M; p.G596C; p.S552C; p.G989D; p.L1256F; p.R1176T; p.D659G
NSD1	1	exon7	SNV	p.S1334Y
NTHL1	1	exon3	SNV	p.T147M
NTHL1	1	exon5	SNV	p.R263H
NTRK1	1	exon12	SNV	p.N493S
NTRK1	2	exon15	SNV	p.A636V; p.V630M
NTRK1	1	exon4	SNV	Splicing
NTRK1	2	exon5	SNV	Splicing; p.S146L
NTRK1	1	exon7	SNV	p.V282I
NTRK2	1	exon10	SNV	p.D258N
NTRK2	1	exon11	SNV	p.K364N
NTRK2	1	exon16	SNV	p.P530S
NTRK2	1	exon19	SNV	p.G679D
NTRK3	2	exon16	SNV	p.L574M; p.A573T
NTRK3	2	exon17	SNV	p.Q673H; p.P644S
NTRK3	3	exon19	SNV	p.L732F; p.F756L; p.E750Q
NTRK3	1	exon20	SNV	p.I823M
NTRK3	2	exon4	SNV	p.V97M; p.A96S
NTRK3	2	exon8	SNV	p.H216Q; p.V221I
NUF2	1	exon5	SNV	p.L110V
NUF2	1	exon8	SNV	p.E178*
NUP93	2	exon2	SNV	p.G40V; p.Q15*
NUP93	1	exon3	SNV	p.Q99*
PAK1	1	exon13	SNV	p.R438Q
PAK1	1	exon14	SNV	p.L490M
PAK1	2	exon7	SNV	p.P246L; p.R203W

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PAK7	1	exon10	DEL	p.L649Sfs*15
PAK7	1	exon4	SNV	p.D26H
PAK7	2	exon5	SNV	p.M326V; p.E248K
PAK7	1	exon6	SNV	p.A434V
PAK7	1	exon8	SNV	p.E542K
PAK7	1	exon9	SNV	p.T606N
PALB2	2	exon12	SNV	p.T1099M; p.S1087I
PALB2	1	exon13	SNV	p.G1166R
PALB2	1	exon4	SNV	p.K379N
PALB2	1	exon5	DEL	p.T696Pfs*17
PALB2	3	exon5	SNV	p.T720A; p.E673Q; p.T773A
PALB2	1	exon7	SNV	p.G881A
PARK2	1	exon3	SNV	p.Q71H
PARK2	1	exon4	SNV	p.V164A
PARP1	1	exon1	SNV	p.K22N
PARP1	1	exon11	SNV	p.P535S
PARP1	1	exon12	SNV	p.V560M
PARP1	1	exon13	SNV	p.M604I
PARP1	1	exon3	SNV	p.S120P
PARP1	1	exon4	SNV	p.D145H
PARP1	2	exon7	SNV	p.P294L; p.P294S; p.M287I
PAX5	1	exon1	SNV	p.P10H
PBRM1	2	exon12	SNV	p.M426I; p.P385A
PBRM1	1	exon14	SNV	p.E486*
PBRM1	2	exon17	SNV	p.E692Q; p.S648Y
PBRM1	1	exon18	SNV	p.T895I
PBRM1	1	exon19	SNV	p.A971V
PBRM1	1	exon21	DEL	p.C1034Nfs*99
PBRM1	1	exon23	SNV	p.E1197K
PBRM1	1	exon24	DEL	p.C1232Tfs*8
PBRM1	1	exon26	SNV	p.A1320V
PBRM1	1	exon30	SNV	p.R1563*
PBRM1	1	exon4	SNV	p.A114T
PBRM1	1	exon5	SNV	p.D168N

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PBRM1	1	exon7	SNV	p.Y217F
PBRM1	1	exon9	DEL	p.I279Yfs*4
PDCD1	1	exon2	DEL	p.T36Pfs*9
PDCD1	2	exon2	SNV	p.V64M; p.S60L
PDCD1	1	exon3	SNV	p.E150K
PDCD1	3	exon5	SNV	p.R264H; p.P271S; p.R231*
PDCD1LG2	1	exon4	SNV	p.V160I
PDGFRA	2	exon10	SNV	p.A491T; p.R479Q
PDGFRA	2	exon18	SNV	p.M844I; p.S851L
PDGFRA	1	exon20	SNV	p.A921S
PDGFRA	2	exon22	SNV	p.F969S; p.R981H
PDGFRA	1	exon23	SNV	p.G1077S
PDGFRA	1	exon5	SNV	p.L245V
PDGFRA	2	exon7	SNV	p.P345H; p.R340W
PDGFRB	2	exon14	SNV	p.V665I; p.E651K
PDGFRB	1	exon17	SNV	p.Q809K
PDGFRB	1	exon20	SNV	p.P908S
PDGFRB	2	exon21	SNV	p.E942K; p.P950S
PDGFRB	1	exon22	SNV	p.N1015S
PDGFRB	2	exon23	SNV	p.P1074T; p.S1092L
PDGFRB	2	exon7	SNV	p.F351L; p.G314S
PDPK1	1	exon12	SNV	p.D464N
PDPK1	1	exon14	SNV	p.G530R
PGR	6	exon1	SNV	p.P69A; p.P69T; p.E395Q; p.E532*; p.A180T; p.V448F
PGR	2	exon1	DEL	p.F275Sfs*102; p.L161_S162delinsF
PGR	1	exon4	SNV	p.R637Q
PGR	1	exon6	SNV	p.Q803H
PGR	1	exon8	SNV	p.V912D
PHOX2B	2	exon3	SNV	p.V294I; p.R149H
PIK3C2G	1	exon13	SNV	p.A387T
PIK3C2G	2	exon2	SNV	p.E218A; p.M1?
PIK3C2G	1	exon24	SNV	p.H877Q
PIK3C2G	2	exon32	SNV	p.L1188I; p.P1204L
PIK3C2G	1	exon5	SNV	p.G116D

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PIK3C3	2	exon11	SNV	p.Q423*; p.D391Y
PIK3C3	2	exon13	SNV	p.Y492C; p.D474Y
PIK3C3	1	exon4	SNV	p.R174H
PIK3CA	25	exon10	SNV	p.E542A; p.Q546E; p.E542K; p.E545D; p.E545G; p.E545A; p.P539R; p.E545K
PIK3CA	1	exon11	SNV	p.I561T
PIK3CA	1	exon12	SNV	p.G613C
PIK3CA	10	exon2	SNV	p.P104L; p.R88*; p.R38H; p.R4Q; p.K111N; p.E81K; p.K111E; p.R88Q
PIK3CA	1	exon2	DEL	p.V105_G106del
PIK3CA	23	exon21	SNV	p.H1047R; p.Q981H; p.H1047L; p.Y1021C; p.M1043V
PIK3CA	2	exon5	SNV	p.N345D; p.N345I
PIK3CB	1	exon1	SNV	p.D16H
PIK3CB	1	exon10	SNV	p.I512F
PIK3CB	1	exon12	SNV	p.L591I
PIK3CB	1	exon15	SNV	p.V713F
PIK3CB	1	exon16	SNV	p.N791D
PIK3CB	1	exon17	SNV	p.R821G
PIK3CB	2	exon18	SNV	p.D879A; p.A880T
PIK3CB	1	exon21	SNV	p.R994*
PIK3CD	2	exon4	SNV	p.E85K; p.M61I
PIK3CD	3	exon5	SNV	p.R184Q; p.S174W; p.Q156K
PIK3CD	1	exon8	SNV	p.R338W
PIK3CG	1	exon11	DEL	p.K1066Sfs*42
PIK3CG	1	exon11	SNV	p.L1069I
PIK3CG	7	exon2	SNV	p.E108K; p.R477H; p.R476C; p.V162I; p.T128M; p.D654N; p.V83M
PIK3CG	1	exon2	DEL	p.E573Dfs*24
PIK3CG	1	exon4	SNV	p.M728L
PIK3CG	1	exon7	SNV	p.R849*
PIK3R1	2	exon10	DEL	p.L420_K430del; p.Y426Sfs*15
PIK3R1	1	exon10	SNV	p.H407R
PIK3R1	1	exon11	SNV	Splicing
PIK3R1	2	exon11	DEL	p.K448_L449del; p.D440_I442delinsV
PIK3R1	1	exon12	SNV	p.R503W

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PIK3R1	1	exon13	INS	p.R577Efs*25
PIK3R1	3	exon13	SNV	p.N564D; p.K567E; p.D548H
PIK3R1	4	exon13	DEL	p.R574_K575del; p.Q579Lfs*21; p.T576del; p.R562_K567del
PIK3R1	1	exon14	SNV	p.K593*
PIK3R1	1	exon15	DEL	p.E621del
PIK3R1	1	exon16	SNV	p.R724L
PIK3R1	1	exon16	DEL	p.Y685_N686del
PIK3R1	1	exon2	SNV	p.P85S
PIK3R2	1	exon10	SNV	p.K416M
PIK3R2	1	exon11	SNV	p.S457N
PIK3R2	2	exon2	SNV	p.A11T; p.E45*
PIK3R3	3	exon3	SNV	p.R72K; p.G52A; p.D69N
PIK3R3	1	exon4	SNV	p.K78R
PIK3R3	1	exon8	DEL	p.M295*
PIM1	1	exon5	SNV	p.E246K
PIM1	1	exon6	SNV	p.L271F
PLCG2	1	exon19	SNV	p.R674Q
PLCG2	3	exon27	SNV	p.T1016M; p.R1001H; p.G1006S
PLCG2	1	exon30	SNV	p.D1126Y
PLCG2	1	exon6	SNV	p.K184E
PLK2	1	exon12	SNV	p.P473L
PLK2	1	exon15	SNV	p.E625K
PLK2	1	exon7	SNV	p.G253S
PMS1	1	exon10	DEL	p.M745del
PMS1	1	exon5	SNV	p.A145V
PMS1	1	exon9	SNV	p.S507L
PMS2	2	exon11	SNV	p.T485M; p.H633R
PNRC1	1	exon1	SNV	p.L57I
POLD1	1	exon11	SNV	p.R443W
POLD1	1	exon13	SNV	p.R525Q
POLD1	1	exon17	SNV	p.A692V
POLD1	1	exon19	SNV	p.R776W
POLD1	1	exon2	SNV	p.E36Q
POLD1	1	exon23	SNV	p.G975S

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
POLD1	1	exon24	DEL	Splicing
POLD1	2	exon5	SNV	p.G178R; p.R177C
POLD1	1	exon7	SNV	p.W265C
POLE	1	exon1	SNV	p.A15G
POLE	1	exon11	SNV	p.W347L
POLE	1	exon13	SNV	p.V411L
POLE	1	exon19	SNV	p.P696L
POLE	1	exon20	SNV	p.R742C
POLE	1	exon24	SNV	p.K954N
POLE	1	exon25	SNV	p.F993V
POLE	3	exon34	SNV	p.R1436W; p.L1441V; p.R1471C
POLE	1	exon37	SNV	p.M1632I
POLE	1	exon43	SNV	p.L1997F
POLE	1	exon44	SNV	p.P2020L; p.P2020S
POLE	3	exon45	SNV	p.C2109Y; p.A2056T
POLE	1	exon6	SNV	p.D181H
POLE	2	exon9	SNV	p.S297F; p.P286R
PPARG	1	exon5	DEL	p.S158Vfs*47
PPARG	1	exon7	SNV	p.Q286E
PPARG	2	exon8	SNV	p.E460K; p.T447M
PPM1D	1	exon1	SNV	p.S85W
PPM1D	1	exon6	DEL	p.Q462*
PPM1D	3	exon6	SNV	p.E525*; p.S575C; p.H596Y; p.R441H
PPP2R1A	1	exon10	SNV	p.R381L
PPP2R1A	1	exon10	INS	p.L429Hfs*12
PPP2R1A	1	exon12	SNV	p.H497R
PPP2R1A	2	exon4	SNV	p.R134Q; p.E125D
PPP2R1A	3	exon5	SNV	p.R183Q; p.R183W
PPP2R1A	3	exon6	SNV	p.S256F; p.W257C
PPP4R2	1	exon8	SNV	p.G274R
PRDM1	6	exon5	SNV	p.R286C; p.R554K; p.T334A; p.A581T; p.T334I; p.S227I
PRDM1	1	exon7	SNV	p.L791F
PRDM14	2	exon2	SNV	p.P70L; p.R194W
PRDM14	1	exon7	SNV	p.Y493H

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PREX2	1	exon1	SNV	p.R6H
PREX2	1	exon15	SNV	p.N542D
PREX2	2	exon23	SNV	p.S872Y; p.V876L
PREX2	1	exon24	SNV	p.S976F
PREX2	1	exon25	SNV	p.I1025M
PREX2	1	exon26	SNV	Splicing
PREX2	1	exon28	SNV	p.R1149G
PREX2	2	exon30	SNV	p.E1205*; p.P1211S
PREX2	1	exon32	SNV	p.E1297D
PREX2	1	exon34	SNV	p.Y1380C
PREX2	1	exon35	SNV	p.F1429C
PREX2	1	exon37	SNV	p.G1509R
PRKAR1A	1	exon11	SNV	p.L330M
PRKAR1A	1	exon3	SNV	p.I100M
PRKAR1A	2	exon4	SNV	p.A127T; p.D121E
PRKCI	1	exon16	SNV	p.N524D
PRKCI	1	exon2	SNV	p.R55Q
PRKCI	1	exon3	SNV	p.T79A
PRKCI	1	exon5	SNV	p.R126H
PRKD1	1	exon10	SNV	p.E493K
PRKD1	1	exon15	SNV	Splicing
PRKD1	1	exon16	SNV	p.H790R
PRKD1	1	exon3	SNV	p.R172C
PRKD1	1	exon4	SNV	p.V207I
PRKD1	1	exon8	SNV	p.N402S
PTCH1	1	exon14	SNV	p.D717E
PTCH1	3	exon15	SNV	p.F795L; p.Q816*; p.K809N
PTCH1	1	exon16	SNV	p.A858V
PTCH1	1	exon18	SNV	p.V1042M
PTCH1	1	exon22	SNV	p.A1187T
PTCH1	1	exon23	SNV	p.T1372M
PTCH1	1	exon3	SNV	p.Q156E
PTCH1	1	exon6	SNV	p.P299A
PTEN	1	exon1	INS	p.D24Rfs*20

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PTEN	1	exon1	SNV	p.L25*
PTEN	1	exon2	SNV	p.N31D
PTEN	1	exon4	DEL	p.K144Ifs*35
PTEN	8	exon4	SNV	p.K164N; p.R130P; p.D92E; p.A126T; p.R130*; p.R130G; p.E150Q
PTEN	1	exon4	INS	p.A148Gfs*32
PTEN	1	exon6	SNV	Splicing
PTEN	4	exon7	SNV	p.R36*; p.P49L; p.V20F; p.E59*
PTEN	1	exon7	INS	p.P51Tfs*5
PTEN	3	exon8	DEL	p.V93*; p.N126Mfs*21; p.T122*
PTEN	2	exon8	SNV	p.D113G; p.Q101E
PTEN	1	exon8	INS	p.N126Kfs*2
PTEN	1	exonN/A	SNV	Splicing
PTP4A1	1	exon2	SNV	p.E35K
PTPN11	1	exon2	SNV	p.V45L
PTPRD	2	exon15	SNV	p.G173V; p.R174H
PTPRD	1	exon20	SNV	p.E246K
PTPRD	5	exon21	SNV	p.G499R; p.G450*; p.R427*; p.T410I; p.A420D
PTPRD	1	exon25	SNV	p.S727F
PTPRD	2	exon28	SNV	p.F860C; p.H997Y
PTPRD	1	exon29	SNV	p.N1023K
PTPRD	1	exon34	SNV	p.P1329L
PTPRD	2	exon37	SNV	p.D1438N; p.S1431F
PTPRD	1	exon44	SNV	p.E1818*
PTPRD	2	exon46	SNV	p.R1898C
PTPRS	1	exon10	SNV	p.V297M
PTPRS	1	exon11	SNV	p.R420S
PTPRS	1	exon13	SNV	p.E582Q
PTPRS	1	exon14	SNV	p.L682M
PTPRS	1	exon15	SNV	p.A729V
PTPRS	1	exon17	SNV	p.T828A
PTPRS	1	exon18	DEL	p.V1025Sfs*18
PTPRS	1	exon18	SNV	p.E925K
PTPRS	1	exon19	SNV	p.L1051M

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
PTPRS	1	exon20	SNV	p.V1128I
PTPRS	1	exon31	SNV	p.T1565M
PTPRS	2	exon32	SNV	p.V1601I; p.T1634M
PTPRS	1	exon33	SNV	p.R1710C
PTPRS	1	exon37	SNV	p.V1896M
PTPRS	1	exon5	SNV	p.R188*
PTPRS	1	exon7	SNV	p.P196S; p.P196L
PTPRS	1	exon8	SNV	p.S221N
PTPRT	3	exon10	SNV	p.S587L; p.G559E; p.R546Q
PTPRT	1	exon17	SNV	p.D836Y
PTPRT	1	exon20	SNV	p.D956N
PTPRT	1	exon25	SNV	p.R1136H
PTPRT	1	exon28	SNV	p.T1275I
PTPRT	2	exon29	SNV	p.D1330Y; p.F1322L
PTPRT	3	exon3	SNV	p.G132E; p.S139F; p.C102S
PTPRT	3	exon30	SNV	p.E1390D; p.R1363G; p.R1349C
PTPRT	2	exon4	SNV	p.V179M; p.A178T
PTPRT	1	exon6	SNV	p.R270H
PTPRT	1	exon8	SNV	p.R450Q
RAB35	1	exon3	SNV	p.V50M
RAB35	1	exon5	SNV	p.A151T
RAC2	1	exon6	SNV	p.D170N
RAD21	1	exon2	INS	Splicing
RAD21	1	exon9	SNV	p.E365Q
RAD50	1	exon13	SNV	p.E696*
RAD50	1	exon19	SNV	p.E995*
RAD50	1	exon4	SNV	p.F156L
RAD50	2	exon5	SNV	p.Q197*; p.A232D
RAD51	1	exon3	SNV	p.D37N
RAD51	1	exon9	INS	p.P286Tfs*37
RAD51	1	exon9	SNV	p.N290D
RAD51B	1	exon7	DEL	p.G203Efs*5
RAD51C	1	exon4	SNV	p.E218K
RAD54L	1	exon12	SNV	p.C391Y

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
RAD54L	1	exon19	SNV	p.A704T
RAD54L	1	exon6	SNV	p.P124S
RAF1	1	exon14	SNV	p.T506I
RAF1	1	exon15	SNV	p.D521H
RAF1	1	exon2	SNV	p.Q34K
RASA1	2	exon1	INS	p.G88Dfs*2; p.G68Wfs*6
RASA1	1	exon16	INS	p.I717Nfs*9
RASA1	1	exon2	SNV	p.R211L
RASA1	1	exon20	SNV	p.R894T
RASA1	1	exon24	INS	p.P978Tfs*2
RASA1	1	exon3	SNV	p.E266V
RASA1	1	exon8	SNV	p.P383A
RB1	2	exon1	INS	p.A10Gfs*21; p.A22Gfs*9
RB1	1	exon10	SNV	p.K327*
RB1	1	exon13	SNV	p.E428D
RB1	1	exon16	DEL	p.M495Ifs*2
RB1	1	exon17	SNV	p.E545*
RB1	1	exon18	SNV	p.S567*
RB1	1	exon19	SNV	p.S634*
RB1	2	exon19	DEL	p.G617Vfs*6
RB1	1	exon19	INS	p.V654Sfs*14
RB1	1	exon2	INS	p.A74Efs*4
RB1	1	exon20	DEL	p.L660Pfs*3
RB1	1	exon20	SNV	p.R698W
RB1	1	exon22	SNV	p.I752V
RB1	1	exon25	INS	p.N854*
RB1	1	exon25	SNV	p.R876H
RB1	1	exon3	SNV	p.E97*
RB1	1	exon4	SNV	p.E137*
RB1	2	exon8	SNV	p.R255Q; p.R251*
RB1	1	exon8	DEL	p.L277Pfs*9
RBM10	2	exon10	SNV	p.R332S; p.R332L
RBM10	1	exon13	SNV	p.P431Q
RBM10	1	exon18	SNV	p.W658G

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
RBM10	2	exon22	SNV	p.A818V; p.R817H
RBM10	1	exon3	SNV	p.R15L
RBM10	1	exon9	SNV	p.Q255*
RECQL	1	exon11	SNV	p.R384M
RECQL	1	exon12	SNV	Splicing
RECQL	1	exon15	SNV	p.S560N
RECQL	1	exon7	SNV	p.H169Y
RECQL4	1	exon1	SNV	p.E2K
RECQL4	1	exon11	SNV	p.D605N
RECQL4	1	exon15	SNV	p.D782H
RECQL4	2	exon22	SNV	p.H1193N; p.R1181*
RECQL4	1	exon5	INS	p.S325Ifs*5
RECQL4	1	exon5	DEL	p.K141Nfs*39
RECQL4	1	exon5	SNV	p.R375H
RECQL4	2	exon6	SNV	p.Q381H; p.K385N
REL	1	exon3	SNV	p.R85K
RET	1	exon19	SNV	p.T1022I
RET	2	exon20	SNV	p.D1093Y; p.S1065T
RET	1	exon7	SNV	p.A432V
RET	1	exon9	SNV	p.R587W
RFWD2	1	exon1	SNV	p.V74I
RFWD2	1	exon11	SNV	p.D382G
RFWD2	1	exon8	SNV	p.D309Y
RHOA	1	exon2	SNV	p.G17A
RHOA	1	exon4	SNV	p.P108S
RICTOR	1	exon22	SNV	p.L685H; p.R709I
RICTOR	1	exon30	SNV	p.S1017C
RICTOR	2	exon31	SNV	p.T1295M; p.V1241L
RICTOR	1	exon35	SNV	p.E1538K
RICTOR	1	exon37	SNV	p.R1633H
RICTOR	1	exon9	SNV	p.P266L
RIT1	1	exon3	SNV	p.P52A
RIT1	2	exon5	SNV	p.E110K; p.M90V
RIT1	3	exon6	SNV	p.R168H; p.R154Q; p.E155*

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
RNF43	2	exon2	DEL	p.S41*; p.L65Wfs*11
RNF43	1	exon3	INS	p.L88Afs*8
RNF43	1	exon3	SNV	p.P87L
RNF43	1	exon4	DEL	p.F139*
RNF43	2	exon5	SNV	p.E174K; p.P191S
RNF43	1	exon7	INS	p.A273Vfs*147
RNF43	1	exon7	SNV	p.W238*; p.A239T
RNF43	1	exon8	SNV	p.W302*
RNF43	3	exon9	DEL	p.G659Vfs*41; p.S646Afs*54
RNF43	4	exon9	SNV	p.R584L; p.R529L; p.S449C; p.A406E
ROS1	2	exon19	SNV	p.D976H; p.S941F
ROS1	1	exon20	SNV	p.R1035Q
ROS1	1	exon23	SNV	p.H1210R
ROS1	3	exon24	SNV	p.L1283I; p.P1263S; p.A1241S
ROS1	1	exon25	SNV	p.E1348D
ROS1	1	exon28	SNV	p.M1527I
ROS1	1	exon29	SNV	p.E1589A
ROS1	1	exon30	SNV	p.L1636F
ROS1	1	exon35	SNV	p.K1893T
ROS1	1	exon39	SNV	p.L2059M
ROS1	2	exon5	SNV	p.F124S; p.W133*
ROS1	1	exon9	SNV	p.R317W
RPS6KA4	1	exon4	SNV	p.R139H
RPS6KA4	1	exon6	SNV	p.G199D
RPS6KA4	1	exon8	SNV	p.P263S; p.P263L
RPS6KB2	1	exon14	SNV	p.Y389C
RPS6KB2	1	exon15	SNV	p.R482C
RPTOR	1	exon10	SNV	p.W382*
RPTOR	2	exon16	SNV	p.V596M
RPTOR	2	exon22	SNV	p.V850D; p.S855C
RPTOR	2	exon23	SNV	p.S884R; p.P879A
RPTOR	1	exon24	SNV	p.T957M
RPTOR	1	exon25	SNV	p.D979N
RPTOR	1	exon34	SNV	p.V1334L

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
RRAGC	1	exon2	SNV	p.V110M
RRAGC	1	exon7	SNV	p.D353Y
RRAS	1	exon6	SNV	p.K192E
RRAS2	1	exon3	SNV	p.Q37L
RRAS2	2	exonN/A	SNV	Splicing
RTEL1	1	exon24	SNV	p.D728E
RTEL1	1	exon32	SNV	p.A1083T
RTEL1	1	exon34	SNV	p.K1197R
RUNX1	1	exon8	SNV	p.T312A
RUNX1	1	exon9	INS	p.A435Qfs*166
RUNX1	3	exon9	SNV	p.A470T; p.M347I; p.S388L; p.T379A
RUNX1	2	exon9	DEL	p.T464Pfs*130; p.P357Rfs*237
RXRA	1	exon10	DEL	p.F439del
RXRA	1	exon10	SNV	p.A416P
RYBP	1	exon2	DEL	p.K71del
RYBP	1	exon4	SNV	p.R168S
SDHA	1	exon10	SNV	p.S456A
SDHA	1	exon13	SNV	p.R585Q
SDHA	1	exon15	SNV	p.E651K
SDHAF2	1	exon4	SNV	p.Q151P
SESN1	1	exon8	SNV	p.S458L
SESN2	2	exon5	DEL	p.P241Qfs*6
SESN3	1	exon10	SNV	p.M476I
SETD2	1	exon12	SNV	p.E1907D; p.L2012F
SETD2	1	exon14	INS	p.D2064Efs*84
SETD2	1	exon14	SNV	p.Q2070*
SETD2	1	exon17	SNV	p.R2399*
SETD2	8	exon3	SNV	p.G966A; p.R1089W; p.R1455S; p.R1065H; p.I793L; p.K1405*; p.E1435D; p.E1339D
SETD2	1	exon3	DEL	p.R1407Gfs*5
SETD2	2	exon3	INS	p.S917Kfs*18; p.N1392Kfs*6
SETD2	1	exon6	SNV	p.V1583G
SETD2	1	exon7	SNV	p.H1629D
SETD2	1	exon8	DEL	p.F1664Ifs*41

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
SETD2	1	exon9	DEL	p.G1687Vfs*18
SETD2	1	exon9	SNV	p.A1675V
SF3B1	1	exon18	SNV	p.A861V
SF3B1	1	exon6	SNV	p.R166Q
SF3B1	1	exon7	SNV	p.R270Q
SF3B1	1	exon8	SNV	p.M365I
SH2B3	3	exon2	SNV	p.E208K; p.R149H; p.A206V
SH2B3	1	exon3	SNV	p.W262*
SH2B3	1	exon5	SNV	p.S330F
SH2B3	1	exon6	SNV	p.G387E
SH2D1A	1	exon1	SNV	p.G39V
SHQ1	1	exon11	SNV	p.K556E
SHQ1	1	exon4	SNV	p.H147R
SHQ1	1	exon7	SNV	p.L279F
SLX4	1	exon12	DEL	p.A1221Rfs*67
SLX4	8	exon12	SNV	p.P1292S; p.R1372W; p.P1503L; p.P1076L; p.S1254L; p.P1414S; p.Q925*; p.S1270F
SLX4	1	exon13	SNV	p.P1562L
SLX4	1	exon6	SNV	p.A426V
SMAD2	1	exon10	SNV	p.S435N
SMAD2	1	exon2	SNV	p.K53N
SMAD2	2	exon3	SNV	p.D106Y; p.R103C
SMAD2	2	exon7	SNV	p.R291Q; p.S287F
SMAD2	1	exonN/A	SNV	Splicing
SMAD3	1	exon1	DEL	p.E28del
SMAD3	1	exon2	INS	p.H85Pfs*27
SMAD3	1	exon2	SNV	p.G73S
SMAD3	2	exon6	SNV	p.S266L; p.T261I
SMAD3	1	exon7	SNV	p.S317C
SMAD3	1	exon8	SNV	Splicing
SMAD4	3	exon10	SNV	p.F408V; p.L414*; p.I383K
SMAD4	2	exon12	SNV	p.L536P; p.G508D
SMAD4	1	exon12	DEL	p.E538del
SMAD4	3	exon3	SNV	p.A118V; p.Q116*; p.R100K

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
SMAD4	1	exon5	SNV	p.Q183L
SMAD4	1	exon6	SNV	p.Q245*
SMAD4	1	exon7	SNV	p.G270E
SMAD4	1	exon7	DEL	p.H287Lfs*43
SMAD4	2	exon9	INS	p.T349Nfs*3; p.E337Rfs*4
SMAD4	6	exon9	SNV	p.R361C; p.R361H; p.C363Y; p.D351H; p.E330K
SMARCA4	1	exon12	SNV	p.V619M
SMARCA4	2	exon16	SNV	p.V766L; p.S813L
SMARCA4	1	exon19	SNV	p.T912A
SMARCA4	1	exon2	SNV	p.G15D
SMARCA4	1	exon24	SNV	p.F1102L
SMARCA4	1	exon26	DEL	p.F1234*
SMARCA4	3	exon26	SNV	p.K1219R; p.D1235Y; p.R1243W
SMARCA4	2	exon29	SNV	p.E1342K; p.K1349N
SMARCA4	2	exon31	SNV	p.E1431*; p.R1443W
SMARCA4	1	exon34	SNV	p.E1614K; p.E1580*
SMARCA4	1	exon35	SNV	p.R1645W
SMARCA4	2	exon4	SNV	p.R208Q; p.N173K
SMARCA4	1	exon6	SNV	p.P324L
SMARCB1	1	exon4	SNV	p.R153G
SMARCD1	1	exon1	SNV	p.R39Q
SMARCD1	1	exon4	SNV	p.R138H
SMARCD1	1	exon6	SNV	p.F233L
SMO	1	exon10	SNV	p.E559A
SMO	2	exon12	SNV	p.K672E; p.R762C
SMYD3	1	exon5	SNV	p.L112V
SMYD3	2	exon9	SNV	p.E229Q; p.K232N
SOCS1	1	exon2	SNV	p.E91D
SOS1	1	exon10	SNV	p.I525M
SOS1	2	exon23	SNV	p.L1326V; p.G1239D
SOS1	1	exon3	SNV	p.I110V
SOS1	1	exon5	SNV	p.N233Y
SOX17	3	exon1	SNV	p.D12G; p.A56D; p.R69Q; p.D12H
SOX17	1	exon1	INS	p.W32Lfs*49

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
SOX17	4	exon2	SNV	p.G308A; p.P197L; p.A166T
SOX17	2	exon2	DEL	p.R273Dfs*114; p.D401Afs*45
SOX17	1	exon2	INS	p.G331Rfs*34
SOX2	3	exon1	SNV	p.A236G; p.S209Y; p.G129S
SOX9	1	exon1	DEL	p.V80_K82del
SOX9	3	exon2	SNV	p.P170L; p.H169Y; p.A206V
SOX9	2	exon3	SNV	p.K398N; p.V502I
SOX9	2	exon3	DEL	p.E388Sfs*15; p.P346Rfs*37
SOX9	1	exon3	INS	p.Q496Pfs*82
SPEN	1	exon11	DEL	p.V3289del
SPEN	11	exon11	SNV	p.G2649D; p.E1891K; p.K1308T; p.R3193Q; p.D1198H; p.L3270F; p.G2674D; p.P2963L; p.T2981M; p.E2125*; p.V1840I; p.Q3304*; p.L1136V; p.R630C
SPEN	1	exon11	INS	p.V1022Cfs*4
SPEN	1	exon12	SNV	p.P3428L
SPEN	3	exon3	SNV	p.D166G; p.S137L; p.A259G
SPEN	1	exon9	SNV	p.E563Q
SPOP	1	exon11	SNV	p.L297R
SPOP	3	exon5	SNV	p.E47K; p.E50K
SPOP	3	exon6	SNV	p.E112Q; p.S80R; p.Y87S
SPOP	2	exon7	SNV	p.F133V; p.D140G
SPOP	3	exon9	SNV	p.P223L; p.S226I; p.E232K
SPRED1	1	exon2	SNV	p.K46R
SPRED1	1	exon3	DEL	p.D79Tfs*42
SPRED1	1	exon3	SNV	p.R117*
SPRED1	1	exon7	INS	p.L410Ffs*22
SPRED1	1	exon7	SNV	p.S392L
SRC	1	exon11	SNV	p.V367L
SRC	1	exon13	SNV	p.E457*
SRC	1	exon4	SNV	p.T72I
SRSF2	1	exon2	SNV	p.S159L
STAG2	1	exon12	DEL	p.V343Dfs*35
STAG2	1	exon23	SNV	p.T749S
STAG2	1	exon30	SNV	p.R1066Q

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
STAG2	1	exon34	SNV	p.S1256*
STAG2	1	exon4	SNV	p.K33N
STAG2	1	exon5	SNV	p.N81S
STAG2	1	exon6	SNV	p.Q123K
STAG2	1	exon9	SNV	p.Q247*
STAT3	1	exon15	SNV	p.E435*
STAT3	1	exon18	SNV	p.W546R
STAT3	1	exon2	SNV	p.E39Q
STAT3	1	exon3	SNV	p.Q91R
STAT3	1	exon7	SNV	p.R214L
STAT5A	1	exon10	DEL	p.Q368Rfs*2
STAT5A	1	exon15	SNV	p.E579*; p.M578I
STAT5A	2	exon20	SNV	p.V764M; p.R769H
STAT5B	1	exon16	SNV	p.V669M
STAT5B	1	exon19	SNV	p.R775W
STAT5B	1	exon2	SNV	p.E29D
STAT5B	1	exon4	SNV	p.E122K
STK11	1	exon1	SNV	p.R28H
STK11	2	exon1	DEL	p.S59Lfs*103; p.K44Sfs*7
STK11	2	exon4	SNV	p.E165*; p.I177M
STK11	1	exon4	INS	p.D162*
STK11	3	exon5	SNV	p.Q214*; p.G242R; p.S216F
STK11	1	exon6	SNV	p.L263F
STK11	2	exon7	SNV	p.L290R; p.R304W
STK40	1	exon12	SNV	p.E387K
STK40	1	exon4	SNV	p.D60H
STK40	1	exon6	SNV	p.R116S
SUFU	1	exon1	INS	p.A25Gfs*23
SUFU	1	exon2	SNV	p.D66Y
SUZ12	1	exon10	SNV	p.E347K
SUZ12	1	exon14	SNV	p.S541F
SUZ12	1	exon8	SNV	p.E289V
SYK	2	exon2	SNV	p.R59T; p.R68W
SYK	1	exon4	SNV	p.R217H; p.H209Y

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
TBX3	1	exon1	SNV	p.P48L
TBX3	1	exon1	DEL	p.P57Afs*30
TBX3	1	exon1	INS	p.K110*
TBX3	1	exon2	SNV	p.P134S
TBX3	1	exon4	INS	p.F278Ifs*10
TBX3	1	exon5	DEL	p.G309Efs*12
TBX3	3	exon7	SNV	p.F513Y; p.R447G; p.A431V
TBX3	1	exon7	INS	p.I395Dfs*12
TBX3	1	exon7	DEL	p.H492Rfs*134
TBX3	2	exon8	DEL	p.P602Lfs*30; p.G730Rfs*154
TBX3	1	exon8	SNV	p.S616Y
TCF3	1	exon12	SNV	p.S320C
TCF3	1	exon14	SNV	p.R371Q
TCF3	1	exon17	SNV	p.R499Q
TCF3	1	exon2	SNV	p.K14N
TCF3	1	exon7	SNV	p.S152C
TCF3	1	exon9	SNV	p.P201S
TCF3	1	exonN/A	SNV	Splicing
TCF7L2	1	exon1	DEL	p.D40Ifs*2
TCF7L2	1	exon11	SNV	p.G401E
TCF7L2	1	exon14	SNV	p.R465H
TCF7L2	1	exon14	INS	p.C463Vfs*8
TCF7L2	1	exon4	SNV	p.P155L
TCF7L2	1	exon9	SNV	p.F334L
TCF7L2	1	exonN/A	SNV	Splicing
TEK	1	exon12	SNV	p.S599L
TEK	2	exon13	SNV	p.E705K; p.T706K
TEK	1	exon17	SNV	p.H943Y
TEK	1	exon2	SNV	p.H52L
TEK	1	exon5	SNV	Splicing
TEK	1	exon8	SNV	p.S363N
TERT	1	exon1	SNV	p.V39M
TERT	1	exon2	SNV	p.R515W
TET1	1	exon11	SNV	p.A1772V

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
TET1	4	exon12	SNV	p.D2115Y; p.K2074*; p.P1916A; p.E2069K
TET1	3	exon2	SNV	p.P470L; p.E187K; p.K60T
TET1	3	exon4	SNV	p.R757I; p.H721D; p.S1412C
TET1	1	exon8	SNV	p.S1580L
TET2	1	exon10	SNV	p.E1492*
TET2	3	exon11	SNV	p.P1528S; p.Q1699*; p.M1570I
TET2	8	exon3	SNV	p.E433D; p.H850D; p.E433K; p.T212A; p.Y899C; p.R369W; p.Q80K; p.Q726K
TET2	1	exon6	SNV	p.R1262W
TET2	2	exon8	SNV	Splicing; p.S1330F
TGFBR1	2	exon2	SNV	p.V56I; p.S69N
TGFBR1	1	exon3	SNV	p.R168C
TGFBR1	1	exon4	SNV	p.E209*
TGFBR1	2	exon5	SNV	p.G325R; p.E300Q
TGFBR1	1	exon7	SNV	p.R413*
TGFBR2	1	exon3	DEL	p.D80Efs*6
TGFBR2	1	exon5	SNV	p.P183S
TGFBR2	3	exon6	SNV	p.D471N; p.G445E; p.V478E
TGFBR2	1	exon7	DEL	p.D514Efs*26
TGFBR2	5	exon8	SNV	p.R553H; p.R553C; p.E544*; p.W546*
TMEM127	1	exon4	SNV	p.E218K
TMPRSS2	1	exon5	SNV	p.G141S
TMPRSS2	1	exon9	SNV	p.W290C
TNFAIP3	1	exon3	SNV	p.E154Q
TNFAIP3	1	exon5	SNV	p.R216K
TNFAIP3	1	exon6	SNV	p.T321R
TNFAIP3	4	exon7	SNV	p.K413N; p.N584K; p.G493R; p.E352G
TNFAIP3	1	exon7	DEL	p.C627Ffs*44
TNFAIP3	1	exon9	SNV	p.E751K
TNFRSF14	1	exon4	SNV	p.V118M
TNFRSF14	1	exon5	SNV	p.P172S
TOP1	2	exon12	SNV	p.R376*; p.R362C
TOP1	1	exon2	SNV	p.N19S
TP53	5	exon11	SNV	p.R205C; p.R210*; p.K219N

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
TP53	3	exon11	DEL	p.E204_R205del; p.R210Efs*3; p.M208_E211del; p.R210Qfs*3
TP53	1	exon12	SNV	p.Q243*
TP53	1	exon12	DEL	p.D261Tfs*29
TP53	1	exon3	SNV	p.E28D
TP53	2	exon3	DEL	p.N29Tfs*15; p.L32Cfs*12
TP53	9	exon4	SNV	p.T125S; Splicing; p.W53*; p.L111P; p.W91*; p.R110C; p.Q100*; p.T125A; p.T125K; p.S96Y; p.R110P
TP53	1	exon4	INS	p.P36Afs*7
TP53	3	exon4	DEL	p.V122Dfs*26; p.Q104Wfs*12; p.V73Wfs*50
TP53	1	exon5	INS	p.P153Afs*28
TP53	41	exon5	SNV	p.F134C; p.R181H; p.F134L; p.S127F; p.R175C; p.C135Y; p.M160I; p.V157F; p.L130V; p.R175H; p.H179Y; p.V173L; p.C135F; p.E171*; p.P152S; Splicing; p.G154V; p.A159V; p.V143E; p.R158G; p.R158H; p.Y163C; p.R156P; p.Q144*; p.A161T; p.K132N; p.Q136*; p.K132R; p.Y163H; p.P177R; p.P151S
TP53	8	exon5	DEL	p.H178Tfs*69; p.G154Afs*12; p.P152Rfs*18; p.V157Pfs*23; p.P128Lfs*42; p.P177_C182del; p.M133_K139del
TP53	31	exon6	SNV	p.V218E; Splicing; p.Y205F; p.V216L; p.R196Q; p.V216M; p.L194R; p.R196P; p.R213Q; p.H193Y; p.P190L; p.H193R; p.I195T; p.I195F; p.Q192*; p.R213*; p.Y220C; p.R196*
TP53	4	exon6	DEL	p.V217Afs*2; p.V218del; p.P219Lfs*27; p.H214Qfs*7
TP53	1	exon6	INS	p.Y205Lfs*4
TP53	35	exon7	SNV	p.R249S; p.P250L; p.R248Q; p.R249T; p.R248W; p.C242F; p.I232F; p.C238S; p.G245D; p.G245C; p.Y234C; p.Y236C; p.R249G; p.G245S; p.R248L; p.D259G; p.I232T; p.E258K; p.G245V; p.M246V; p.S241F
TP53	4	exon7	DEL	p.N235del; p.I232*; p.M237Vfs*2; p.S241Lfs*22
TP53	3	exon7	INS	p.L252Vfs*13; p.N235Qfs*5; p.S240Kfs*24
TP53	1	exon8	DEL	p.E286Kfs*65

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
TP53	38	exon8	SNV	p.E271*; p.C275F; p.C277F; p.E286K; p.R306*; p.R273P; p.R273L; p.E271K; p.E285K; p.E285*; p.E294*; p.E286Q; p.R273H; Splicing; p.R273C; p.C275Y; p.G266R
TP53	2	exon9	SNV	Splicing; p.Q317*
TP53BP1	4	exon12	SNV	p.L504F; p.S532N; p.M684I; p.K680*; p.Q626H
TP53BP1	1	exon15	SNV	Splicing
TP53BP1	1	exon17	SNV	p.M1157I
TP53BP1	1	exon19	SNV	p.R1355*
TP53BP1	1	exon21	SNV	p.D1531H
TP53BP1	1	exon26	SNV	p.A1806V
TP53BP1	1	exon28	SNV	p.D1968N
TP53BP1	1	exon4	SNV	p.S95Y
TP63	1	exon12	SNV	p.L519I
TP63	2	exon14	SNV	p.E609K; p.Q648H
TP63	1	exon4	SNV	p.S167F
TP63	1	exon6	SNV	p.R266*
TP63	1	exon7	SNV	p.T326A
TRAF2	1	exon2	SNV	p.A30T
TRAF2	1	exon5	SNV	p.R144C
TRAF2	1	exon8	SNV	p.R305W
TRAF7	1	exon10	SNV	p.D327N
TRAF7	1	exon12	SNV	p.G378D
TRAF7	1	exon19	SNV	p.A604V
TRAF7	2	exon8	SNV	p.A176V; p.E204Q
TSC1	1	exon15	SNV	p.P589S
TSC1	1	exon17	SNV	p.M736I
TSC1	1	exon18	SNV	p.T773I
TSC1	1	exon23	SNV	p.R1062Q
TSC1	1	exon4	SNV	p.T52A
TSC1	1	exon6	SNV	p.Q145*
TSC1	1	exon7	SNV	p.E211*
TSC1	1	exon7	INS	p.V178Gfs*40
TSC1	2	exon9	SNV	p.S289*

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
TSC2	1	exon11	INS	p.A328_A328insL
TSC2	1	exon15	SNV	p.R505*
TSC2	2	exon17	SNV	p.R611Q
TSC2	1	exon2	SNV	p.E13D
TSC2	1	exon22	SNV	p.R799C
TSC2	1	exon27	SNV	p.V1034I
TSC2	1	exon3	SNV	p.R59Q
TSC2	1	exon30	SNV	p.P1188L
TSC2	1	exon34	SNV	p.S1449F
TSC2	1	exon37	INS	p.L1581Tfs*22
TSC2	1	exon37	DEL	p.L1581Qfs*18
TSC2	1	exon4	SNV	p.L88V
TSC2	1	exon42	SNV	p.V1807M
TSC2	1	exon5	SNV	p.R115H
TSC2	1	exon9	SNV	p.I273V
TSHR	5	exon10	SNV	p.S671T; p.C408S; p.S314Y; p.G545S; p.I654V
TSHR	1	exon5	SNV	p.K146N
UPF1	1	exon15	SNV	p.I669M
UPF1	1	exon2	SNV	p.N86S
UPF1	1	exon4	SNV	p.R162G
VEGFA	1	exon6	DEL	p.K322Nfs*32
VEGFA	1	exon6	SNV	p.K336E
VHL	1	exon1	SNV	p.W88*
VHL	1	exon1	DEL	p.V83Afs*76
VHL	1	exon2	DEL	p.H125Afs*4
VHL	1	exon2	SNV	p.L128R
VHL	1	exon3	SNV	p.Q195R
VTCN1	1	exon3	SNV	p.D66N
WHSC1	1	exon12	SNV	p.G756S
WHSC1	1	exon14	SNV	p.D882A
WHSC1	1	exon18	SNV	p.E1099K
WHSC1	2	exon22	DEL	p.P1343Qfs*49
WHSC1	1	exon4	SNV	p.T303M
WHSC1	1	exon6	SNV	p.R501L

Gene (n=417)	#Samples (n=401)	Exon (n=1835)	Type	Mutations Assessed
WT1	1	exon8	SNV	p.D215N
WT1	3	exonN/A	SNV	p.G38S; p.Q68*; p.T7M
WWTR1	1	exon3	SNV	p.S89W
WWTR1	1	exon8	SNV	p.P395S
XIAP	1	exon2	SNV	p.L121Q
XPO1	1	exon13	SNV	p.D449Y
XPO1	2	exon15	SNV	p.E571K
XPO1	1	exon17	SNV	p.Q644E
XPO1	1	exon25	SNV	p.R1043W
XRCC2	1	exon3	DEL	p.F270Lfs*27
YAP1	1	exon1	DEL	p.A50Pfs*24
YES1	1	exon10	SNV	p.R398W
YES1	1	exon12	SNV	p.R487Q
ZFHX3	2	exon10	SNV	p.A3584T; p.F3307L
ZFHX3	4	exon2	SNV	p.S497N; p.P895A; p.A896T; p.Q541*
ZFHX3	2	exon2	DEL	p.E763Sfs*61; p.V777del
ZFHX3	1	exon6	SNV	p.D1188N
ZFHX3	1	exon7	SNV	p.T1288M
ZFHX3	14	exon9	SNV	p.E3025K; p.E2628K; p.R1712L; p.Q2639E; p.Q1390H; p.A2749V; p.A2709T; p.P1392S; p.N1602I; p.A2487V; p.A1896T; p.S2541*; p.P2106L
ZFHX3	2	exon9	DEL	p.P2208Lfs*47; p.V2159Lfs*6