December 23, 2022

ACT Genomics
% David Kern
Principal and Founder
K2 Regulatory Consulting, LLC
479 Cumberland Drive
Burlingame, California 94010

Re: K210017
Trade/Device Name: ACTOnco, ACTOnco IVD
Regulation Number: 21 CFR 866.6080
Regulation Name: Next generation sequencing based tumor profiling test
Regulatory Class: Class II
Product Code: PZM
Dated: September 19, 2022
Received: September 26, 2022

Dear David Kern:

We have reviewed your Section 510(k) premarket notification of intent to market the device referenced above and have determined the device is substantially equivalent (for the indications for use stated in the enclosure) to legally marketed predicate devices marketed in interstate commerce prior to May 28, 1976, the enactment date of the Medical Device Amendments, or to devices that have been reclassified in accordance with the provisions of the Federal Food, Drug, and Cosmetic Act (Act) that do not require approval of a premarket approval application (PMA). You may, therefore, market the device, subject to the general controls provisions of the Act. Although this letter refers to your product as a device, please be aware that some cleared products may instead be combination products. The 510(k) Premarket Notification Database located at https://www.accessdata.fda.gov/scripts/cdrh/cfdocs/cfpmn/pmn.cfm identifies combination product submissions. The general controls provisions of the Act include requirements for annual registration, listing of devices, good manufacturing practice, labeling, and prohibitions against misbranding and adulteration. Please note: CDRH does not evaluate information related to contract liability warranties. We remind you, however, that device labeling must be truthful and not misleading.

If your device is classified (see above) into either class II (Special Controls) or class III (PMA), it may be subject to additional controls. Existing major regulations affecting your device can be found in the Code of Federal Regulations, Title 21, Parts 800 to 898. In addition, FDA may publish further announcements concerning your device in the Federal Register.

Please be advised that FDA's issuance of a substantial equivalence determination does not mean that FDA has made a determination that your device complies with other requirements of the Act or any Federal statutes and regulations administered by other Federal agencies. You must comply with all the Act's

requirements, including, but not limited to: registration and listing (21 CFR Part 807); labeling (21 CFR Part 801 and Part 809); medical device reporting (reporting of medical device-related adverse events) (21 CFR 803) for devices or postmarketing safety reporting (21 CFR 4, Subpart B) for combination products (see https://www.fda.gov/combination-products/guidance-regulatory-information/postmarketing-safety-reporting-combination-products); good manufacturing practice requirements as set forth in the quality systems (QS) regulation (21 CFR Part 820) for devices or current good manufacturing practices (21 CFR 4, Subpart A) for combination products; and, if applicable, the electronic product radiation control provisions (Sections 531-542 of the Act); 21 CFR 1000-1050.

Also, please note the regulation entitled, "Misbranding by reference to premarket notification" (21 CFR Part 807.97). For questions regarding the reporting of adverse events under the MDR regulation (21 CFR Part 803), please go to https://www.fda.gov/medical-devices/medical-device-safety/medical-device-reporting-mdr-how-report-medical-device-problems.

For comprehensive regulatory information about medical devices and radiation-emitting products, including information about labeling regulations, please see Device Advice (https://www.fda.gov/medical-devices/device-advice-comprehensive-regulatory-assistance) and CDRH Learn (https://www.fda.gov/training-and-continuing-education/cdrh-learn). Additionally, you may contact the Division of Industry and Consumer Education (DICE) to ask a question about a specific regulatory topic. See the DICE website (https://www.fda.gov/medical-devices/device-advice-comprehensive-regulatory-assistance/contact-us-division-industry-and-consumer-education-dice) for more information or contact DICE by email (DICE@fda.hhs.gov) or phone (1-800-638-2041 or 301-796-7100).

Sincerely,

Zivana Tezak-fragale -S

Zivana Tezak, Ph.D.
Branch Chief
Division of Molecular Genetics
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OHT7: Office of In Vitro Diagnostics
Office of Product Evaluation and Quality
Center for Devices and Radiological Health

Enclosure

DEPARTMENT OF HEALTH AND HUMAN SERVICES
Food and Drug Administration

Indications for Use

Form Approved: OMB No. 0910-0120
Expiration Date: 06/30/2023
See PRA Statement below.

510(k) Number *(if known)*
K210017

Device Name
ACTOnco IVD

Indications for Use *(Describe)*
The ACTOnco IVD assay is an in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed, paraffin-embedded tumor tissue from patients with solid malignant neoplasms to detect genetic alterations in a broad multi gene panel. The test is intended to provide information on point mutations, small insertions and deletions, ERBB2 gene amplification, and tumor mutational burden 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. ACTOnco IVD is a single-site assay performed at ACT Genomics.

Type of Use *(Select one or both, as applicable)*

☒ Prescription Use (Part 21 CFR 801 Subpart D) ☐ Over-The-Counter Use (21 CFR 801 Subpart C)

CONTINUE ON A SEPARATE PAGE IF NEEDED.

510(k) Summary for ACTOnco IVD

1 Submitter

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2 Submission Correspondent - US

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3 Device Identification

Name of Device:	ACTOnco IVD
Common or Usual Name	ACTOnco
Classification Name:	Next generation sequencing based tumor profiling test
Classification Regulation:	866.6080
Product Code:	PZM
Device Class:	Class II
Classification Panel:	Pathology

4 Legally Marketed Predicate Device

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

DEN170058

5 Device Description

The ACTOnco IVD assay is an in-vitro diagnostic assay intended to provide information 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.

The assay is a custom targeted sequencing platform, utilizing amplicon-based sequencing, to detect point mutations (single nucleotide variants, or SNVs), small insertions and deletions (Indels), ERBB2 gene amplification, and tumor mutational burden (TMB) in tumor specimens. The assay uses custom DNA primers corresponding to all exons and selected introns of oncogenes, tumor suppressor genes, drug metabolism genes, and immune-related genes. Primers are synthesized by a secondary manufacturer (Thermo Fisher Scientific). An overlapping amplicon approach is utilized in which tiled primers are designed to generate multiple overlapping amplicons of the same region to avoid allele dropout. In total, the primers target approximately 1.8Mb of the human genome. Genomic DNA is extracted from FFPE tissue samples.

Sequence libraries are prepared through a multiplex polymerase chain reaction (PCR) amplification step to enrich target sequences. Target sequences are tagged with index oligonucleotide to identify individual sample and adaptor oligonucleotide to anchor the amplicon to complimentary oligonucleotides embedded on the surface of the sequencing bead. Target sequences on the sequencing beads are amplified using emulsion PCR before sequencing. Multiple barcoded sequence libraries (from different patients) are pooled and then sequenced on a Thermo Fisher Ion GeneStudio™ S5 Prime System. Sequence reads are then aligned to the reference human genome. By comparing the identity of bases from the tumor DNA and the reference human genome, variant alterations are identified in the tumor.

The assay system includes a sequencing instrument, reagents (DNA extraction, library preparation and sequencing), software (operation of the sequencing instrument and variant calling), and standard operating procedures (SOPs) for the use of the system. ACT Genomics takes the responsibilities in monitoring the instrument; reagents and consumable materials which will be used in the assay process.

Multiple software components will be used in the assay. The NGS raw read analysis will be done using Thermo Fisher software. Variant calling for SNVs, insertions and deletions will be done using Thermo Fisher software. Mutation and variant annotation will be done using software from ACT Genomics, the Cunningham Lab and Golden Helix software. ERBB2 gene amplification will be done using software from Boeva Lab. Tumor Purity and Zygosity will be done using software from Halgamuge Lab (Kaushalya Amarasinghe). Calculations for tumor mutational burden will be done using ACT Genomics software.

6 Indication for Use Statement

The ACTOnco IVD assay is an in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed, paraffin-embedded tumor tissue from patients with solid malignant neoplasms to detect genetic alterations in a broad multi gene panel. The test is intended to provide information on point mutations, small insertions and deletions, ERBB2 gene amplification, and tumor mutational burden 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. ACTOnco IVD is a single-site assay performed at ACT Genomics.

Special conditions for use statement

- *For prescription use only*
- *For in vitro diagnostic use*

Special instrument requirements

- *Thermo Fisher Ion GeneStudio™ S5 Prime System (qualified by ACT Genomics).*

7 Comparison of Technological Characteristics with The Predicate Device

The ACTOnco IVD test has technological characteristics that are substantially equivalent to the predicate device as described in the tables below. Both the subject device and the predicate device use targeted, high throughput parallel sequencing for the detection genetic alterations. The subject device uses an amplicon-based target enrichment approach and Ion Torrent's semiconductor sequencing technology, whereas the predicate device used a hybrid capture target enrichment approach and Illumina's sequencing by synthesis (SBS) technology. Both sequencing platforms have been used in IVD devices that have been either cleared or approved by the FDA.

Both devices use FFPE tumor tissue samples collected from patients with solid malignant neoplasms. The predicate device also uses matched normal samples, which are not required for the ACTOnco IVD test. The ACTOnco IVD test has established a baseline using pooled normal samples, which tumor samples are then compared.

Table 7.1 Similarities between the predicate and subject devices

Characteristic	Predicate MSK-IMPACT (DEN170058)	Subject Device ACTOnco IVD
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 ACTOnco IVD assay is an in vitro diagnostic test that uses targeted next generation sequencing of formalin-fixed, paraffin-embedded tumor tissue from patients with solid malignant neoplasms to detect genetic alterations in a broad multi gene panel. The test is intended to provide information on point mutations, small insertions and deletions, ERBB2 amplification, and tumor mutational burden 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. ACTOnco IVD is a single-site assay performed at ACT Genomics.
Specimen Types	Formalin-fixed, paraffin-embedded (FFPE) tumor tissue with matched normal specimens from patients with solid malignant neoplasms	Formalin-fixed, paraffin-embedded (FFPE) tumor tissue from patients with solid malignant neoplasms
Target Population	Patients with solid malignant neoplasms	Same
Assay cut-off	MSK-IMPACT does not report mutations below 2% for known hotspot mutations and 5% for non-hotspot mutations.	Same
Laboratory	Single-site laboratory	Single-site laboratory

Table 7.2 Differences between the predicate and subject devices

Characteristic	Predicate MSK-IMPACT (DEN170058)	Subject Device ACTOnco IVD
Differences		
Sequencing Instrument	Illumina HiSeq™ 2500 Sequencer	Thermo Fisher Ion GeneStudio™ S5 Prime System
Target Enrichment Technology	Hybrid Capture	Amplicon
Genes on Panel	468	440
Black List	73 exons	650 amplicons within 199 genes excluded from reporting SNV/ Indels due to pseudo gene or consistently low coverage (≤ 35x).
Variant Type	Intended to provide information on somatic mutations (point mutations and small insertions and deletions), and microsatellite instability	ACTOnco provides information on somatic mutations as indicated and also includes copy number alteration and provides information on tumor mutational burden (TMB). No microsatellite instability.
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 ≥85% target regions; • hotspot mutation calling threshold (mutation coverage (DP) ≥ 35, mutant reads (AD) ≥ 5, strand bias (SB) < 0.9, mutation frequency (VF) ≥ 2%), and non-hotspot mutation threshold (DP ≥ 35, AD ≥ 10, SB < 0.9, VF ≥ 5%).
Controls	• Matched normal • Positive control • Negative control • No template control (NTC)	• Positive control • Negative control • No template control (NTC)
Clinical Evidence Curation Oncopanel results are reported under one of these two categories:	Uses OncoKB, knowledge base that includes biologic, clinical and therapeutic information curated from professional guidelines and recommendations, therapeutic labeling, disease specific expert and advocacy group	A variant interpretation summary is generated, which includes biologic impact, variant specific effect and therapeutic relevance curated from professional guidelines and recommendations, therapeutic labeling, disease

Characteristic	Predicate MSK-IMPACT (DEN170058)	Subject Device ACTOnco IVD
Differences		
• "Cancer Mutations with Evidence of Clinical Significance" or • "Cancer Mutations with Potential Clinical Significance."	recommendations, and medical literature. Classification criteria were developed by MSK to communicate the level of clinical evidence available for individual mutations in the test report. • OncoKB undergoes periodic updates through the review of new information by a panel of experts	specific expert and advocacy group recommendations, and medical literature. Classification criteria were developed by ACT Genomics with the reference of AMP guideline, to communicate the level of clinical evidence available for individual mutations in the test report. • ACT Genomics undergoes periodic updates through the review of new information by medical informatics scientists and scientific content management team

8 Performance Testing

8.1 Precision / Reproducibility

The objective of this study was to assess within lab precision by evaluating sources of variability (reagent lots, operators, sequencing instruments, and days).

A set of 20 unique samples comprising ten cancer types containing different variants were evaluated. Twelve (12) of the samples across ten cancer types were single clinical samples (FFPE), and eight (8) of them across four cancer types were pooled DNA from multiple FFPE blocks within the same cancer type.

For SNVs, MNVs, insertions, deletions, and copy number variants, all 20 samples were evaluated. For TMB, the 12 unique single FFPE samples were evaluated.

The study design utilized three reagent lots, two instruments, and two operators across twelve (12) non-consecutive days. Duplicate observations for each sample were run. This resulted in 48 observations per level (3 reagent lots x 2 operators x 2 instruments x 2 start days per instrument/operator/reagent lot combination x 2 replicates). The study design resulted in 24 degrees of freedom for the replicate variability evaluation. Prior to conducting the variance component analysis, all data were visually inspected.

8.1.1 SNVs, Insertions and Deletions

Seven hundred fifty-one (751) variants were found within the 20 samples with 638 unique variants and 247 genes.

The qualitative analysis of the data was done using the correct call rate. A correct call was defined as the same mutational variant call within each of the observations of a sample with that variant. Correct calls may be positive (mutation present) or negative (wild type present).

The ACTOnco system evaluated 751 variants in the assay. There were 48 observations of each variant; this resulted in 36,048 total data points (751 x 48). Of the 36,048 observations, 32,496 (677 x 48) were from SNVs, 1,008 (21 x 48) were from MNVs, 528 (11 x 48) were from insertions, and 2,016 (42 x 48) were from deletions. Since this assay has a final QC check at the variant level, there were 142 variant observations removed from the data set due to QC failing at the variant level, resulting in 35,906 variants remaining for the study analysis (36048 – 142).

Across all the mutational variant data, the call rate was 98.33% (35,308/ 35,906). The breakout for each variant type was as follows:

SNV: 98.33% 31,837 / 32,377 MNV: 97.18% 963 / 991
INS: 96.97% 512 / 528 DEL: 99.30% 1,996 / 2,010

There were 15,076 WT calls across all the samples, and given 48 observations, this resulted in 723,648 (15,076 x 48) WT calls in the data set. Across all the WT data there were 99.997% correct calls (723,628/723,648). See Table 8-1.

Table 8-1 Correct Calls for SNVs, Insertions and Deletions

Mutational Variant Type	Operator	Instrument	Reagent lot	Days	Number Correct	Number Attempted	Call rate (95%CI)
All	All	All	All	All	35308	35906	98.33 (0.982-0.985)
Deletion	All	All	All	All	1996	2010	99.30 (0.988-0.996)
Insertion	All	All	All	All	512	528	96.97 (0.951-0.981)
MNV	All	All	All	All	963	991	97.18 (0.959-0.980)
SNV	All	All	All	All	31837	32377	98.33 (0.982-0.985)
Negative (WT)	All	All	All	All	723628	723648	99.997 (0.99996-0.99998)

These data were also evaluated at the gene level with information on the gene, the mutation, the normalized coverage range, statistics on the MAF, and the positive call rates with a 2 sided 95% confidence interval. Table 8-2 (single samples) and Table 8-3 (mixed samples) from the study are presented below.

Table 8-2 Correct Calls for SNVs, Insertions and Deletions by Gene (Single Samples)

Tissue type	Mutation type	Gene/Exon	cDNA change	Amino acid change	Mutation frequency	FDA level
colon adenocarcinoma	SNV	BRAF exon15	140453136_A>T	p.V600E	29.06%	2
colon adenocarcinoma	SNV	GRIN2A exon4	10032000_C>A	p.G275*	50.66%	3
colon adenocarcinoma	DEL	KDM5C exon23	53223787_CCA>C	p.C1190fs	58.95%	3
colon adenocarcinoma	INS	KMT2D exon34	49432235_C>CG	p.S2969fs	24.81%	3
colon adenocarcinoma	DEL	MAP3K1 exon15	56179358_CAG>C	p.E1225fs	27.16%	3
colon adenocarcinoma	SNV	RECQL4 exon5	145741926_G>A	p.R193*	19.53%	3
colon adenocarcinoma	DEL	RNF43 exon9	56435160_AC>A	p.G659fs	70.63%	3
colon adenocarcinoma	DEL	SPOP exon8	47685274_TA>T	p.F225fs	28.11%	3
colon adenocarcinoma	SNV	TP53 exon11	7572929_A>G	p.*394Rext*9	28.98%	3
colon adenocarcinoma	SNV	TP53 exon5	7578554_A>G	p.Y126H	19.00%	3
kidney cancer	SNV	NSD1 exon19	176709523_C>T	p.R1984*	8.28%	3
kidney cancer	SNV	SETD2 exon3	47162711_T>A	p.K1139*	19.24%	3
tumor of exocrine pancreas	SNV	IDH1 exon4	209113112_C>G	p.R132P	36.71%	3
tumor of exocrine pancreas	SNV	KRAS exon2	25398284_C>T	p.G12D	32.13%	3
tumor of exocrine pancreas	SNV	TP53 exon7	7577539_G>A	p.R248W	53.06%	3
liver cancer	SNV	ARID2 exon5	46211626_A>T	p.K198*	30.76%	3
liver cancer	DEL	ARID2 exon15	46245542_TC>T	p.P1213fs	16.36%	3
liver cancer	SNV	TP53 exon6	7578224_T>A	p.R209*	40.45%	3
cholangiocarcinoma	SNV	KRAS exon2	25398284_C>T	p.G12D	20.98%	3
cholangiocarcinoma	SNV	TP53 exon6	7578190_T>C	p.Y220C	31.86%	3
colon adenocarcinoma	DEL	B2M exon2	45007740_GGA>G	p.R65fs	17.84%	3
colon adenocarcinoma	SNV	CTNNB1 exon3	41266137_C>T	p.S45F	35.38%	3
colon adenocarcinoma	SNV	ERBB2 exon19	37880220_T>C	p.L755S	18.69%	3
colon adenocarcinoma	SNV	GNAS exon6	57480483_C>T	p.R160C	17.13%	3
colon adenocarcinoma	SNV	KRAS exon2	25398285_C>T	p.G12S	17.30%	2
colon adenocarcinoma	SNV	MLH1 exon4	37045935_C>T	p.T117M	66.43%	3
colon adenocarcinoma	DEL	NBN exon10	90967511_CT>C	p.R466fs	32.29%	3
colon adenocarcinoma	DEL	PIK3R1 exon3	67569265_GC>G	p.P129fs	16.39%	3
colon adenocarcinoma	SNV	RAF1 exon7	12645694_A>T	p.S259T	17.54%	3
endometrial cancer	SNV	AKT1 exon4	105243045_A>G	p.W80R	25.22%	3
endometrial cancer	DEL	EZH2 exon10	148515024_TC>T	p.G395fs	25.03%	3
endometrial cancer	SNV	KRAS exon2	25398284_C>T	p.G12D	40.10%	3
endometrial cancer	DEL	MAP2K4 exon4	11998922_CA>C	p.K143fs	24.62%	3

Tissue type	Mutation type	Gene/Exon	cDNA change	Amino acid change	Mutation frequency	FDA level
endometrial cancer	SNV	PAX5 exon2	37020768_A>C	p.V26G	27.27%	3
endometrial cancer	SNV	PIK3CA exon2	178916876_G>A	p.R88Q	25.20%	3
endometrial cancer	SNV	PIK3CA exon8	178928079_G>A	p.E453K	28.15%	3
endometrial cancer	DEL	TP53 exon8	7577143_CAGT>C	p.L265del	27.08%	3
urinary system cancer	SNV	ERBB2 exon8	37868208_C>A	p.S310Y	21.48%	3
endometrial cancer	SNV	ARID1A exon18	27101135_C>T	p.Q1473*	44.33%	3
endometrial cancer	DEL	KDM6A exon17	44928975_AG>A	p.A694fs	43.77%	3
endometrial cancer	SNV	PIK3CA exon2	178916890_C>T	p.R93W	44.63%	3
endometrial cancer	SNV	PIK3CA exon10	178936092_A>G	p.E545G	45.45%	3
endometrial cancer	SNV	PTEN exon5	89692905_G>A	p.R130Q	88.18%	3
endometrial cancer	DEL	RNF43 exon9	56435160_AC>A	p.G659fs	48.78%	3
Lung cancer	DEL	CDKN2A exon2	21971147_TGGGCTCCGCGCCGTGGA>T	p.L65fs	18.53%	3
Lung cancer	DEL	CDKN2A exon2	21971176_TCCGCCACTCGGGCG>T	p.S56fs	6.41%	3
Lung cancer	SNV	PIK3CA exon21	178952085_A>G	p.H1047R	19.21%	3
Lung cancer	INS	TP53 exon6	7578186_C>CT	p.P222fs	26.73%	3
Skin cancer	SNV	BRAF exon15	140453136_A>T	p.V600E	52.96%	2
Skin cancer	SNV	CDKN2A exon2	21971179_G>C	p.A60G	46.91%	3
Skin cancer	SNV	ERBB4 exon14	212537975_G>A	p.R544W	24.89%	3
Skin cancer	SNV	PIK3R1 exon13	67591106_A>G	p.K567E	16.22%	3
Skin cancer	SNV	RAC1 exon2	6426893_C>T	p.P29L	34.31%	3
Skin cancer	SNV	SMARCA4 exon26	11141519_C>T	p.Q1166*	27.56%	3

Table 8-3 Correct Calls for SNVs, Insertions and Deletions by Gene (Mixed Samples)

Tissue type	Mutation type	Gene/Exon	cDNA change	Amino Acid change	Mutation frequency	FDA level
Lung cancer	SNV	CTNNB1 exon3	41266101_C>T	p.S33F	11.72%	3
Lung cancer	SNV	EGFR exon18	55241708_G>C	p.G719A	10.32%	2
Lung cancer	SNV	EGFR exon20	55249071_C>T	p.T790M	11.41%	2
Lung cancer	SNV	EGFR exon21	55259515_T>G	p.L858R	11.17%	2
Lung cancer	SNV	SMARCA4 exon32	11169037_A>T	p.K1511*	13.94%	3
Lung cancer	SNV	TP53 exon8	7577106_G>C	p.P278A	11.51%	3
Lung cancer	SNV	B2M exon1	45003746_T>C	p.M1?	7.48%	3

Tissue type	Mutation type	Gene/Exon	cDNA change	Amino Acid change	Mutation frequency	FDA level
Lung cancer	INS	B2M exon2	45007890_G>GT	p.K114*	7.43%	3
Lung cancer	SNV	BRAF exon15	140453136_A>T	p.V600E	12.41%	2
Lung cancer	SNV	EGFR exon18	55241708_G>C	p.G719A	5.41%	2
Lung cancer	SNV	EGFR exon20	55249071_C>T	p.T790M	6.04%	2
Lung cancer	SNV	EGFR exon21	55259469_G>A	p.V843I	6.40%	2
Lung cancer	SNV	EGFR exon21	55259515_T>G	p.L858R	6.37%	2
Lung cancer	SNV	SMARCA4 exon32	11169037_A>T	p.K1511*	6.99%	3
Lung cancer	SNV	TP53 exon8	7577106_G>C	p.P278A	5.43%	3
Breast cancer	SNV	PIK3CA exon21	178952085_A>G	p.H1047R	16.57%	2
Breast cancer	SNV	TP53 exon7	7577539_G>A	p.R248W	17.03%	3
Breast cancer	SNV	PIK3CA exon21	178952085_A>G	p.H1047R	11.32%	2
Breast cancer	SNV	TP53 exon7	7577539_G>A	p.R248W	10.29%	3
Breast cancer	SNV	TP53 exon7	7577556_C>T	p.C242Y	33.94%	3
Skin cancer	SNV	ARID1B exon3	157222648_C>T	p.Q626*	5.96%	3
Skin cancer	SNV	BRAF exon15	140453136_A>T	p.V600E	7.95%	2
Skin cancer	SNV	BRAF exon11	140481397_C>A	p.V471F	20.06%	3
Skin cancer	SNV	CARD11 exon6	2979559_C>T	p.D230N	19.56%	3
Skin cancer	SNV	FANCA exon27	89833576_G>C	p.S858R	7.10%	3
Skin cancer	SNV	NF1 exon12	29533315_C>T	p.R440*	23.72%	3
Skin cancer	SNV	NOTCH4 exon4	32188899_G>A	p.Q219*	5.58%	3
Skin cancer	SNV	NRAS exon3	115256529_T>C	p.Q61R	8.71%	2
Skin cancer	DEL	TP53 exon4	7579546_CG>C	p.P47fs	6.82%	3
Skin cancer	SNV	BRAF exon15	140453136_A>T	p.V600E	2.02%	2
Skin cancer	SNV	BRAF exon11	140481397_C>A	p.V471F	5.56%	3
Skin cancer	MNV	DNMT3A exon16	25466800_GG>AA	p.R635W	26.56%	3
Skin cancer	SNV	EZH2 exon16	148508728_A>T	p.Y646N	24.10%	3
Skin cancer	SNV	NF1 exon12	29533315_C>T	p.R440*	7.87%	3
Skin cancer	SNV	NRAS exon3	115256529_T>A	p.Q61L	28.18%	2
Skin cancer	SNV	NRAS exon3	115256529_T>C	p.Q61R	2.31%	2
Skin cancer	SNV	TP53 exon8	7577099_C>T	p.R280K	36.85%	3
urinary system cancer	INS	ARID1A exon9	27092804_A>AC	p.Q944fs	12.90%	3
urinary system cancer	SNV	RXRA exon10	137328351_C>T	p.S427F	9.84%	3
urinary system cancer	INS	ARID1A exon9	27092804_A>AC	p.Q944fs	8.05%	3

Tissue type	Mutation type	Gene/Exon	cDNA change	Amino Acid change	Mutation frequency	FDA level
urinary system cancer	SNV	RXRA exon10	137328351_C>T	p.S427F	6.52%	3

The positive and negative call rates for sequence mutations (SNVs, MNVs, insertions and deletions) in each sample are summarized in Table 8-4 to Table 8-7. Call rates based on the total number of mutations along with the 2-sides 95% confidence interval were calculated. Table 8-4 and Table 8-5 summarize the results of positive call rate in per clinical sample (single and mixed). Table 8-6 and Table 8-7 summarize the results of negative call rate in per clinical sample (single and mixed).

Table 8-4 Positive call rate per sample (Single sample)

Sample ID	Total No unique mutations detected across all 48 replicates	Positive Call Rate per Mutation	Positive call rate (two-sided 95% CI)
Sample 1	66	28/48 for 1 45/48 for 1 46/46 for 1 48/48 for 63	3143/3166 99.27% (98.91%, 99.51%)
Sample 2	34	48/48 for 34	1632/1632 100% (99.76%, 100%)
Sample 3	14	48/48 for 14	672/672 100% (99.43%, 100%)
Sample 4	52	48/48 for 52	2496/2496 100% (99.84%, 100%)
Sample 5	17	48/48 for 17	816/816 100% (99.53%, 100%)
Sample 6	45	46/46 for 1 46/47 for 1 48/48 for 43	2156/2157 99.95% (99.73%, 99.99%)
Sample 7	35	47/47 for 1 48/48 for 34	1679/1679 100% (99.77%, 100%)
Sample 8	13	5/47 for 1 48/48 for 12	581/623 93.25% (91.01%, 94.97%)
Sample 9	49	24/48 for 1 32/48 for 1 33/48 for 1 46/48 for 2 48/48 for 44	2293/2352 97.49% (96.77%, 98.05%)
Sample 10	11	48/48 for 11	528/528 100% (99.27%, 100%)
Sample 11	16	34/46 for 1 48/48 for 15	754/766 98.43% (97.28%, 99.1%)

Sample ID	Total No unique mutations detected across all 48 replicates	Positive Call Rate per Mutation	Positive call rate (two-sided 95% CI)
Sample 12	34	20/20 for 1 24/24 for 1 48/48 for 32	1580/1580 100% (99.75%, 100%)

* MAF = mutational allele frequency, CO = cut-off

Table 8-5 Positive call rate per sample (Mixed Sample)

Sample ID	Total No unique mutations detected across all 48 replicates	Positive Call Rate per Mutation	Positive call rate (two-sided 95% CI)
Sample 13	30	43/48 for 1 48/48 for 29	1435/1440 99.65% (99.19%, 99.85%)
Sample 14	52	21/48 for 1 26/48 for 1 29/48 for 1 36/48 for 1 39/48 for 1 42/48 for 1 43/48 for 2 44/48 for 1 45/48 for 1 46/48 for 3 47/48 for 3 48/48 for 36	2375/2496 95.15% (94.24%, 95.93%)
Sample 15	22	48/48 for 22	1056/1056 100% (99.64%,100%)
Sample 16	32	46/48 for 1 47/48 for 1 48/48 for 30	1533/1536 99.8% (99.43%, 99.93%)
Sample 17	78	26/48 for 2 31/48 for 1 37/47 for 1 37/48 for 1 38/48 for 1 39/48 for 1 44/44 for 1 46/48 for 4 47/47 for 4 47/48 for 3 48/48 for 59	3623/3735 97% (96.4%, 97.5%)

Sample ID	Total No unique mutations detected across all 48 replicates	Positive Call Rate per Mutation	Positive call rate (two-sided 95% CI)
Sample 18	81	23/48 for 1 36/48 for 1 37/48 for 1 39/48 for 1 40/48 for 2 41/48 for 1 43/47 for 1 45/45 for 1 45/48 for 1 46/47 for 1 46/48 for 2 47/48 for 5 48/48 for 63	3786/3883 97.5% (96.96%, 97.95%)
Sample 19	32	39/45 for 1 42/42 for 1 47/48 for 1 31/31 for 1 48/48 for 28	1503/1510 99.54% (99.05%, 99.78%)
Sample 20	38	3/40 for 1 17/17 for 1 21/48 for 1 33/48 for 1 39/48 for 1 42/48 for 1 43/48 for 2 44/48 for 1 45/48 for 1 46/46 for 1 46/48 for 3 47/48 for 1 48/48 for 24	1667/1783 93.49% (92.25%, 94.55%)

* MAF = mutational allele frequency, CO = cut-off

Table 8-6 Negative call rate per sample (Single Sample)

Sample ID	Total No unique mutations detected across all 48 replicates	Negative Call Rate per Mutation	Negative call rate (two-sided 95% CI)
Sample 1	754	48/48 for 754	36192/36192 100% (99.99%, 100%)

Sample ID	Total No unique mutations detected across all 48 replicates	Negative Call Rate per Mutation	Negative call rate (two-sided 95% CI)
Sample 2	755	47/48 for 1 48/48 for 754	36239/36240 99.99% (99.98%, 99.99%)
Sample 3	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 4	755	46/48 for 1 47/48 for 8 48/48 for 746	36230/36240 99.97% (99.95%, 99.99%)
Sample 5	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 6	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 7	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 8	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 9	753	47/48 for 4 48/48 for 749	36140/36144 99.99% (99.97%, 99.99%)
Sample 10	755	448/48 for 755	36240/36240 100% (99.99%, 100%)
Sample 11	754	48/48 for 754	36192/36192 100% (99.99%, 100%)
Sample 12	754	48/48 for 754	36192/36192 100% (99.99%, 100%)

* CI = interval confidence

Table 8-7 Negative call rate per sample (Mixed Sample)

Sample ID	Total No unique mutations detected across all 48 replicates	Negative Call Rate per Mutation	Negative call rate (two-sided 95% CI)
Sample 13	752	47/48 for 3 48/48 for 749	36093/36096 99.99% (99.98%,99.99%)
Sample 14	751	47/48 for 1 48/48 for 750	36047/36048 99.99% (99.98%,99.99%)
Sample 15	754	48/48 for 754	36192/36192 100% (99.99%,100%)
Sample 16	754	48/48 for 754	36192/36192 100% (99.99%,100%)
Sample 17	753	48/48 for 753	36144/36144 100% (99.99%,100%)
Sample 18	752	48/48 for 752	36096/36096 100% (99.99%,100%)

Sample ID	Total No unique mutations detected across all 48 replicates	Negative Call Rate per Mutation	Negative call rate (two-sided 95% CI)
Sample 19	755	47/48 for 1 48/48 for 754	36239/36240 99.99% (99.98%,99.99%)
Sample 20	755	48/48 for 755	36240/36240 100% (99.99%,100%)

* CI = interval confidence

8.1.2 ERBB2 Gene Amplification

The same 20 samples with 12 single- FFPE and 8 mixed samples used for SNV and Indel analysis were used for ERBB2 amplification analysis as well. Within the 20 samples, there were 3 samples with ERBB2 amplification. The rest of the samples were ERBB2 no amplification. Within the assay, ERBB2 amplification is reported when the observed copy number for the gene is greater than or equal to 4 copies of the observed copy number determined by the assay.

The qualitative evaluation of the data was done by determining the call rates for the amplification and no amplification samples. There were 48 observations of the ERBB2 gene in each sample, resulting in 960 total observations in this study (20 samples x 48 observations per sample). None of the observations were removed in this study. Amongst 960 observations, 144 (3 x 48) observations were amplification, and the other 816 (17 x 48) observations were from no amplification. The call rate was 100% for both amplification and no amplification groups. The summarized data can be found in Table 8-8

Table 8-8 Summary of the ERBB2 gene amplification performance

Sample ID	Sample Type	Standardized Cancer Type	Gene	N	CNV Status	Mean Observed CNV	Min Observed CNV	Max Observed CNV	No. Positive Calls	Positive Call Rate	Lower Bound 95% CI	Upper Bound 95% CI
Sample 3	Single	Pancreas Adenocarcinoma	ERBB2	48	Amp	8.03	7.5	8.5	48	100.00%	0.926	1
Sample 18	Mixed	Breast Invasive ductal carcinoma	ERBB2	48	Amp	14.31	13	16	48	100.00%	0.926	1
Sample 19	Mixed	Breast Invasive ductal carcinoma	ERBB2	48	Amp	31.16	28.5	32.5	48	100.00%	0.926	1

8.1.3 Tumor Mutational Burden

The same twelve (12) single clinical samples were used for TMB (see Table 8-9). These samples presented with TMB scores that ranged from 0.7 to 45.8.

Specimens spanned the range of TMB scores and included 10 different tumor types. The tumor purity for these samples ranged from 32.5% to 72.7%. The mean TMB score, SD, percent CV and mean target region coverage for each sample are shown in Table 8-9 below

Table 8-9 TMB Precision Data

Sample ID	Tumor Type	Mean Tumor Purity	Mean TMB Value	# of Valid Results	SD	Percent CV	Mean Average Target Coverage *
Sample 1	Colon adenocarcinoma	72.7	39.25	48	1.629	4.151	768.771

Sample ID	Tumor Type	Mean Tumor Purity	Mean TMB Value	# of Valid Results	SD	Percent CV	Mean Average Target Coverage *
Sample 2	Kidney cancer	33.0	10.7	47	0.870	8.131	833.511
Sample 3	Tumor of exocrine pancreas	51.8	1.93	48	0.895	46.352	731.438
Sample 4	Liver cancer	37.8	26.42	48	2.380	9.008	835.354
Sample 5	Cholangiocarcinoma	44.5	2.81	48	1.100	39.178	765.250
Sample 6	Colon adenocarcinoma	34.6	28.71	48	1.317	4.587	796.667
Sample 7	Endometrial cancer	60.0	18.81	48	1.194	6.347	854.021
Sample 8	Urinary system cancer	52.1	2.42	48	0.999	41.321	869.063
Sample 9	Endometrial cancer	66.8	18.33	48	3.186	17.381	728.229
Sample 10	Breast cancer	32.5	1.16	37	0.686	59.144	840.054
Sample 11	Lung cancer	39.0	4.47	39	0.678	15.166	838.769
Sample 12	Skin cancer	52.9	13.03	48	1.014	7.782	881.792

* SD = Standard Deviation, CV = Coefficient of Variation

8.2 Analytical Sensitivity/ Limit of Detection

A study for determining the LoD of the ACTOnco assay for SNVs and INDELs was done by evaluating 10 different cancer specimens with 15 SNVs, 5 insertions and 11 deletions with 5 dilution levels and 5 replicate observations per level. The call rate was determined for each variant and the LOD/C95 was approximated between the call rate that was below 95% and the highest call rate (100%). Table 8-10 summarizes the data within mutational types and

Table 8-11 summarizes the estimation of the LoD range for each variant within the subjects evaluated in the study. The call rate here requires the use of the assay cutoff and as such we refer to this as the LOD/C95. Since the concentrations for each level from the underlying internal continuous response values (MAF) were available these are also presented in the tables.

Table 8-10. Analytical Sensitivity (LoD MAF) for Representative SNVs and INDELs

Variant	Established MAF Range	No. of Variants in Clinical Cases in the Established Range
Hotspot SNVs (cutoff of 2%)	1.5%-6.6%	7
Non-hotspot SNVs (cutoff of 5%)	2.4%-15.1%	8
Insertions	1.1%-45.4%	5
Deletions	1.9%-22.0%	11

Table 8-11 Estimation of the LoD range for the representative variants

FDA Level	Variant Type	Variant ID	Gene Exon	Subject ID	RGT Lot	Mean MAF Range	Dilution Levels	Call Rates (n/N)	LoD Approx (MAF)	Assay cutoff
2	SNV	EGFR_chr7_55259515_T>G	EGFR 21	Sample1	A	0.027 to 0.211	Level 4 Level 5	100% (5/5) 100% (5/5)	0.027 to 0.041	0.02
2	SNV	EGFR_chr7_55259515_T>G	EGFR 21	Sample1	B	0.022 to 0.201	Level 4 Level 5	100% (5/5) 60% (3/5)	0.022 to 0.045	0.02
2	SNV	BRCA1_chr17_41244716_A>T	BRCA1 10	Sample2	A	0.021 to 0.332	Level 3 Level 4	100% (5/5) 0% (0/5)	0.041 to 0.079	0.05
2	SNV	BRCA1_chr17_41244716_A>T	BRCA1 10	Sample2	B	0.019 to 0.315	Level 3 Level 4	100% (5/5) 0% (0/5)	0.037 to 0.087	0.05
2	SNV	BRAF_chr7_140453136_A>T	BRAF 15	Sample3	A	0.013 to 0.259	Level 3 Level 4	100% (5/5) 60% (3/5)	0.022 to 0.043	0.02
2	SNV	BRAF_chr7_140453136_A>T	BRAF 15	Sample3	B	0.011 to 0.272	Level 3 Level 4	100% (5/5) 40% (2/5)	0.019 to 0.044	0.02
2	SNV	NRAS_chr1_115256530_G>T	NRAS 3	Sample5	A	0.035 to 0.395	Level 4 Level 5	100% (5/5) 100% (5/5)	0.035 to 0.066	0.02
2	SNV	NRAS_chr1_115256530_G>T	NRAS 3	Sample5	B	0.039 to 0.415	Level 4 Level 5	100% (5/5) 100% (5/5)	0.039 to 0.047	0.02
2	SNV	PIK3CA_chr3_178952085_A>G	PIK3CA 21	Sample7	A	0.006 to 0.209	Level 2 Level 3	100% (5/5) 20% (1/5)	0.016 to 0.036	0.02
2	SNV	PIK3CA_chr3_178952085_A>G	PIK3CA 21	Sample7	B	0.005 to 0.201	Level 2 Level 3	100% (5/5) 0% (0/5)	0.015 to 0.040	0.02
2	SNV	EGFR_chr7_55249071_C>T	EGFR 20	Sample8	A	0.015 to 0.240	Level 2 Level 3	100% (5/5) 40% (2/5)	0.018 to 0.044	0.02
2	SNV	EGFR_chr7_55249071_C>T	EGFR 20	Sample8	B	0.012 to 0.237	Level 2 Level 3	100% (5/5) 33% (1/3)	0.019 to 0.062	0.02
3	SNV	KRAS_chr12_25398284_C>T	KRAS 2	Sample4	A	0.026 to 0.326	Level 4 Level 5	100% (5/5) 100% (5/5)	0.026 to 0.048	0.02
3	SNV	KRAS_chr12_25398284_C>T	KRAS 2	Sample4	B	0.030 to 0.337	Level 4 Level 5	100% (5/5) 100% (5/5)	0.030 to 0.039	0.02
2	INS	KIT_chr4_55593640_T>TTTACATAGACCC	KIT 11	Sample6	A	0.005 to 0.283	Level 1 Level 2	100% (5/5) 80% (4/5)	0.066 to 0.283	0.05
2	INS	KIT_chr4_55593640_T>TTTACATAGACCC	KIT 11	Sample6	B	0.004 to 0.321	Level 2 Level 3	100% (5/5) 0% (0/5)	0.030 to 0.063	0.05
2	INS	ERBB2_chr17_37880981_A>AGCATACGTGATG	ERBB2 20	Sample7	A	0.005 to 0.177	Level 1 Level 2	100% (5/5) 60% (3/5)	0.029 to 0.177	0.02
2	INS	ERBB2_chr17_37880981_A>AGCATACGTGATG	ERBB2 20	Sample7	B	0.005 to 0.174	Level 2 Level 3	100% (5/5) 20% (1/5)	0.011 to 0.027	0.02
3	INS	KMT2D_chr12_49432655_G>GA	KMT2D 34	Sample10	A	0.008 to 0.375	Level 2 Level 3	100% (5/5) 0% (0/5)	0.027 to 0.064	0.05

FDA Level	Variant Type	Variant ID	Gene Exon	Subject ID	RGT Lot	Mean MAF Range	Dilution Levels	Call Rates (n/N)	LoD Approx (MAF)	Assay cutoff
3	INS	KMT2D_chr12_49432655_G>GA	KMT2D 34	Sample10	B	0.009 to 0.356	Level 2 Level 3	100% (5/5) 0% (0/4)	0.028 to 0.071	0.05
-	INS	MUC16_chr19_9019605_C>CTGG	MUC16 22	Sample8	A	0.009 to 0.446	Level 2 Level 3	100% (5/5) 0% (0/5)	0.022 to 0.061	0.05
-	INS	MUC16_chr19_9019605_C>CTGG	MUC16 22	Sample8	B	0.007 to 0.454	Level 1 Level 2	100% (5/5) 40% (5/2)	0.052 to 0.454	0.05
2	DEL	EGFR_chr7_55242464_AGGAATTAAGAGAAGC>A	EGFR 19	Sample8	A	0.005 to 0.420	Level 2 Level 3	100% (5/5) 80% (4/5)	0.028 to 0.066	0.02
2	DEL	EGFR_chr7_55242464_AGGAATTAAGAGAAGC>A	EGFR 19	Sample8	B	0.007 to 0.411	Level 2 Level 3	100% (5/5) 40% (2/5)	0.023 to 0.073	0.02
2	DEL	BRCA1_chr17_41222948_TTCTTCTGGGGTCAGGCCAG>T	BRCA1 15	Sample9	A	0.046 to 0.209	Level 2 Level 3	100% (4/4) 0% (0/2)	0.046 to 0.118	0.05
2	DEL	BRCA1_chr17_41222948_TTCTTCTGGGGTCAGGCCAG>T	BRCA1 15	Sample9	B	0.072 to 0.298	Level 3 Level 4	100% (1/1) 100% (1/1)	0.072 to 0.087	0.05
3	DEL	KMT2D_chr12_49434491_AG>A	KMT2D 31	Sample2	A	0.012 to 0.644	Level 3 Level 4	100% (5/5) 20% (1/5)	0.042 to 0.112	0.05
3	DEL	KMT2D_chr12_49434491_AG>A	KMT2D 31	Sample2	B	0.017 to 0.390	Level 3 Level 4	100% (5/5) 80% (4/5)	0.055 to 0.099	0.05
3	DEL	PTEN_chr10_89692782_CT>C	PTEN 5	Sample3	A	0.014 to 0.284	Level 2 Level 3	100% (5/5) 0% (0/5)	0.039 to 0.107	0.05
3	DEL	PTEN_chr10_89692782_CT>C	PTEN 5	Sample3	B	0.021 to 0.258	Level 2 Level 3	100% (5/5) 20% (1/5)	0.046 to 0.105	0.05
3	SNV	TP53_chr17_7578524_G>A	TP53 5	Sample1	A	0.034 to 0.260	Level 4 Level 5	100% (5/5) 0% (0/5)	0.034 to 0.067	0.05
3	SNV	TP53_chr17_7578524_G>A	TP53 5	Sample1	B	0.024 to 0.284	Level 3 Level 4	100% (5/5) 60% (3/5)	0.057 to 0.090	0.05
3	SNV	PTEN_chr10_89692904_C>G	PTEN 5	Sample4	A	0.051 to 0.680	Level 4 Level 5	100% (5/5) 100% (5/5)	0.051 to 0.102	0.05
3	SNV	PTEN_chr10_89692904_C>G	PTEN 5	Sample4	B	0.051 to 0.672	Level 4 Level 5	100% (5/5) 100% (5/5)	0.051 to 0.088	0.05
3	SNV	KRAS_chr12_25398285_C>G	KRAS 2	Sample9	A	0.008 to 0.085	Level 2 Level 3	100% (5/5) 20% (1/5)	0.017 to 0.032	0.02
3	SNV	KRAS_chr12_25398285_C>G	KRAS 2	Sample9	B	0.006 to 0.086	Level 2 Level 3	100% (5/5) 0% (0/5)	0.015 to 0.034	0.02
3	SNV	RB1_chr13_48953788_T>C	RB1 -	Sample9	A	0.013 to 0.222	Level 2 Level 3	100% (5/5) 40% (2/5)	0.049 to 0.107	0.05
3	SNV	RB1_chr13_48953788_T>C	RB1 -	Sample9	B	0.016 to 0.237	Level 2 Level 3	100% (5/5) 20% (1/5)	0.046 to 0.102	0.05
3	SNV	TP53_chr17_7577568_C>T	TP53 7	Sample10	A	0.006 to 0.524	Level 2 Level 3	100% (5/5) 0% (0/5)	0.025 to 0.067	0.05

FDA Level	Variant Type	Variant ID	Gene Exon	Subject ID	RGT Lot	Mean MAF Range	Dilution Levels	Call Rates (n/N)	LoD Approx (MAF)	Assay cutoff
3	SNV	TP53_chr17_7577568_C>T	TP53 7	Sample10	B	0.007 to 0.527	Level 2 Level 3	100% (5/5) 0% (0/5)	0.024 to 0.065	0.05
-	SNV	BRCA2_chr13_32910983_G>A	BRCA2 11	Sample3	A	0.024 to 0.252	Level 2 Level 3	100% (5/5) 40% (2/5)	0.043 to 0.092	0.05
-	SNV	BRCA2_chr13_32910983_G>A	BRCA2 11	Sample3	B	0.015 to 0.259	Level 2 Level 3	100% (5/5) 40% (2/5)	0.050 to 0.093	0.05
-	SNV	BRCA2_chr13_32930651_G>A	BRCA2 15	Sample6	A	0.012 to 0.518	Level 3 Level 4	100% (5/5) 0% (0/5)	0.026 to 0.058	0.05
-	SNV	BRCA2_chr13_32930651_G>A	BRCA2 15	Sample6	B	0.015 to 0.508	Level 2 Level 3	100% (5/5) 80% (4/5)	0.056 to 0.147	0.05
-	SNV	TP53_chr17_7577108_C>T	TP53 8	Sample8	A	0.026 to 0.137	Level 1 Level 2	100% (5/5) 50% (1/2)	0.051 to 0.137	0.05
-	SNV	TP53_chr17_7577108_C>T	TP53 8	Sample8	B	0.151	Level 1	100% (5/5)	0.151	0.05
3	INS	SETD2_chr3_47058705_T>TA	SETD2 21	Sample8	A	0.012 to 0.259	Level 1 Level 2	100% (5/5) 0% (0/5)	0.028 to 0.259	0.05
3	INS	SETD2_chr3_47058705_T>TA	SETD2 21	Sample8	B	0.010 to 0.249	Level 1 Level 2	100% (5/5) 0% (0/5)	0.027 to 0.249	0.05
3	DEL	RNF43_chr17_565435160_AC>A	RNF43 9	Sample3	A	0.051 to 0.606	Level 3 Level 4	100% (5/5) 60% (3/5)	0.051 to 0.118	0.05
3	DEL	RNF43_chr17_565435160_AC>A	RNF43 9	Sample3	B	0.038 to 0.656	Level 3 Level 4	100% (5/5) 75% (3/4)	0.071 to 0.097	0.05
-	DEL	CDK4_chr12_58145274_CCTT>C	CDK4 -	Sample3	A	0.012 to 0.278	Level 2 Level 3	100% (5/5) 20% (1/5)	0.044 to 0.111	0.05
-	DEL	CDK4_chr12_58145274_CCTT>C	CDK4 -	Sample3	B	0.017 to 0.286	Level 2 Level 3	100% (5/5) 20% (1/5)	0.041 to 0.094	0.05
-	DEL	HIST1H1E_chr6_26156677_TGAA>T	HIST1H1E 1	Sample3	A	0.034 to 0.512	Level 3 Level 4	100% (3/3) 0% (0/2)	0.034 to 0.083	0.05
-	DEL	HIST1H1E_chr6_26156677_TGAA>T	HIST1H1E 1	Sample3	B	0.070 to 0.529	Level 2 Level 3	100% (5/5) 100% (5/5)	0.070 to 0.096	0.05
-	DEL	NOTCH4_chr6_32181941_TC>T	NOTCH4 13	Sample3	A	0.038 to 0.575	Level 3 Level 4	100% (5/5) 33% (1/3)	0.047 to 0.096	0.05
-	DEL	NOTCH4_chr6_32181941_TC>T	NOTCH4 13	Sample3	B	0.033 to 0.544	Level 2 Level 3	100% (5/5) 67% (2/3)	0.092 to 0.220	0.05
-	DEL	PRKN_chr6_161771210_TG>T	PRKN 12	Sample3	A	0.011 to 0.271	Level 2 Level 3	100% (5/5) 0% (0/5)	0.038 to 0.111	0.05
-	DEL	PRKN_chr6_161771210_TG>T	PRKN 12	Sample3	B	0.009 to 0.290	Level 2 Level 3	100% (5/5) 0% (0/5)	0.042 to 0.101	0.05
-	DEL	MAP2K2_chr19_4095411_CG>C	MAP2K2 9	Sample4	A	0.038 to 0.348	Level 4 Level 5	100% (5/5) 20% (1/5)	0.038 to 0.065	0.05
-	DEL	MAP2K2_chr19_4095411_CG>C	MAP2K2 9	Sample4	B	0.034 to 0.328	Level 3 Level 4	100% (5/5) 80% (4/5)	0.061 to 0.127	0.05
-	DEL	MUC16_chr19_9064309_AG>A	MUC16 3	Sample4	A	0.024 to 0.282	Level 2 Level 3	100% (4/4) 0% (0/1)	0.019 to 0.171	0.05
-	DEL	MUC16_chr19_9064309_AG>A	MUC16 3	Sample4	B	0.049 to 0.372	Level 2 Level 3	100% (4/4) 50% (2/4)	0.049 to 0.152	0.05

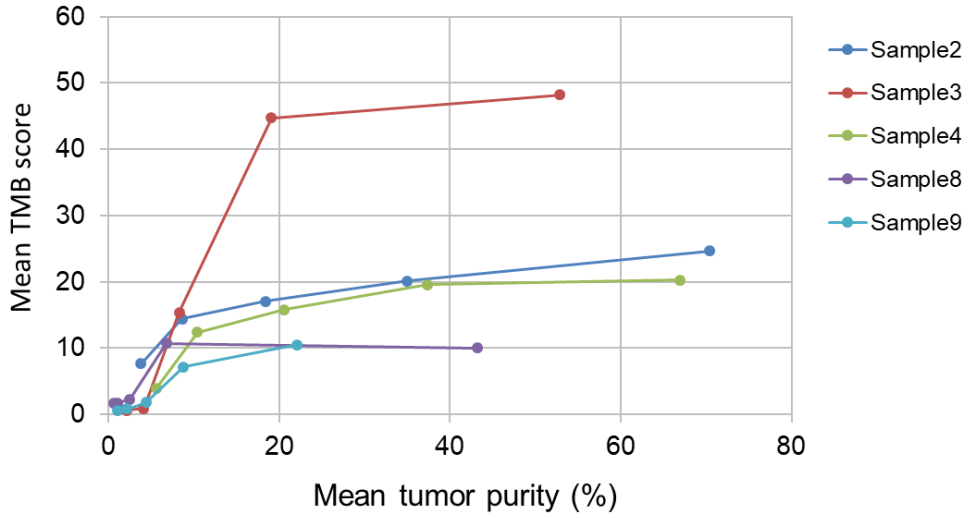
LoD - Tumor Mutation Burden (TMB) and DNA input

To evaluate the LoD for TMB reporting, five (5) FFPE tumor specimens with TMB ≥10 from the LoD SNV supplement study were evaluated. Each sample was serially diluted across 5 levels at 10 replicates per level. The DNA extracted from the 5 FFPE specimens from the LoD supplement study were serially diluted with matched normal DNA across 5 dilution levels with 5 replicates per level per reagent lot (2 were used) resulting in 10 observations per level. The cancer types, tumor purity, mean TMB score, SD, percent CV and average mean exon coverage of all valid replicates are shown below in Table 8-12. Figure 8-1 further visualizes the TMB scores with respective tumor purities. These data show ACTOnco assay has acceptable TMB performance across tumor purities at or above 20% with the percent CV < 16%. ACT Genomics is taking a conservative approach and limiting the tumor purity at greater than 30%.

Table 8-12: TMB Data from Supplemental LoD Study

Tumor Type	Subject ID	Dilution level	Tumor Purity (%)	Mean TMB Score	# of Valid Results	SD	%CV	Average Mean Base Coverage Depth
Endometrial cancer	Sample 2	1	70.4	24.63	10	2.99	12.16	894.1
Endometrial cancer		2	35	20.12	10	4.28	21.25	907.8
Endometrial cancer		3	18.4	17.08	10	2.59	15.16	1043.4
Endometrial cancer		4	8.6	14.43	10	6.12	42.43	1102.8
Endometrial cancer		5	3.8	7.63	10	5.83	76.42	1240.8
Colon adenocarcinoma	Sample 3	1	52.9	48.24	10	1.72	3.57	844.8
Colon adenocarcinoma		2	19.1	44.73	10	2.29	5.12	775.6
Colon adenocarcinoma		3	8.3	15.37	10	3.74	24.32	859.8
Colon adenocarcinoma		4	4.1	0.93	10	1.07	115.04	1026.4
Colon adenocarcinoma		5	2.1	0.62	10	1.08	174.51	1045.4
Endometrial cancer	Sample 4	1	66.9	20.24	10	4.42	21.83	831.4
Endometrial cancer		2	37.4	19.56	10	5.28	26.98	965.4
Endometrial cancer		3	20.6	15.84	10	2.27	14.36	818.4
Endometrial cancer		4	10.4	12.4	10	2.94	23.74	833.2
Endometrial cancer		5	5.6	3.92	10	2.73	69.61	1011.7
Lung cancer	Sample 8	1	43.2	10.03	10	3.21	31.96	747.5
Lung cancer		2	6.8	10.73	10	1.31	12.25	721.8
Lung cancer		3	2.5	2.31	10	0.95	41.04	811.5
Lung cancer		4	1.1	1.75	10	1.06	60.61	904.7
Lung cancer		5	0.6	1.64	10	1.77	108.09	976.9
Ovarian cancer	Sample 9	1	22.1	10.54	10	1.38	13.09	818.7
Ovarian cancer		2	8.8	7.2	10	1.21	16.78	742.7
Ovarian cancer		3	4.4	1.82	10	1.27	69.73	814.4
Ovarian cancer		4	2.2	0.82	10	0.62	75.57	938.4
Ovarian cancer		5	1.1	0.62	10	0.55	88.60	1030.7

Figure 8-1: Linearity of TMB score with respective tumor purity calculated by the ACTOnco assay.



The same ten (10) samples used for the SNV/INDEL and ERBB2 copy number analyses were also evaluated for the performance of TMB reporting across the three (3) DNA input levels. Five replicates at each DNA input level for each sample were run. This resulted in 50 observations per DNA level (10 samples x 5 replicates). To evaluate the DNA input level and the tumor purity of ACTOnco TMB assay 3 of the 10 samples tested were at the required tumor purity for the assay and seven (7) of the ten (10) samples were purposely selected for their low tumor purity (15% – 30%). Table 8-13 summarizes the sample accounting and TMB performance, including the standard deviation (SD) and coefficient of variation (CV) for all 5 replicates for each DNA input level per sample.

Table 8-13: Summary of the TMB Results for All 5 Replicates for Each DNA Input Level per Sample

Sample ID	Cancer Type	Tumor Purity	DNA Input	N	Mean TMB	Median TMB	SD	CV%	TMB Range
Sample 1	Stomach cancer	30%	20ng	5	4.7	4.4	0.720	15.4%	3.8 - 5.7
			40ng	5	5.1	5.7	0.902	17.8%	3.8 - 5.7
			80ng	5	4.5	4.4	0.921	20.3%	3.2 - 5.7
Sample 2	Urinary system cancer	37%	20ng	5	9.5	10	1.038	11.0%	8.1 - 10.6
			40ng	5	9.7	10	1.558	16.0%	8.1 - 11.2
			80ng	5	9.6	9.4	0.329	3.4%	9.4 - 10.0
Sample 3	Lung cancer	30%	20ng	5	4.2	3.8	1.285	30.8%	3.2 - 6.3
			40ng	5	2.7	2.6	0.686	25.4%	1.9 - 3.8
			80ng	5	3.2	3.8	1.461	46.2%	1.3 - 4.4
Sample 4	Colon adenocarcinoma	33%	20ng	5	30.6	31	1.285	4.2%	28.5 - 31.6
			40ng	5	29.1	29.1	1.689	5.8%	27.3 - 31.6

Sample ID	Cancer Type	Tumor Purity	DNA Input	N	Mean TMB	Median TMB	SD	CV%	TMB Range
			80ng	5	29.5	29.8	0.733	2.5%	28.5 - 30.4
Sample 5	Ovarian cancer	73%	20ng	5	4.8	5	0.716	15.0%	3.8 - 5.7
			40ng	5	5.0	5	0.997	19.8%	3.8 - 6.3
			80ng	5	4.9	4.4	1.352	27.5%	3.8 - 6.9
Sample 6	Endo gland cancer	20%	20ng	5	1.2	0.7	0.825	68.7%	0.7 - 2.6
			40ng	5	3.9	4.4	1.338	34.3%	1.9 - 5.0
			80ng	5	4.2	5.7	3.002	71.8%	0.1 - 6.9
Sample 7	Lung cancer	15%	20ng	5	0.1	0.1	0.000	0.0%	0.1 - 0.1
			40ng	5	0.2	0.1	0.268	122.0%	0.1 - 0.7
			80ng	5	0.5	0.1	0.537	116.7%	0.1 - 1.3
Sample 8	Colon adenocarcinoma	25%	20ng	5	40.5	40.9	0.950	2.3%	39.0 - 41.5
			40ng	5	41.0	40.9	1.154	2.8%	39.7 - 42.7
			80ng	5	41.0	40.9	0.890	2.2%	39.7 - 42.1
Sample 9	Endometrial cancer	30%	20ng	5	13.9	13.7	1.686	12.1%	11.8 - 16.2
			40ng	5	13.9	14.3	0.910	6.5%	12.5 - 14.9
			80ng	5	12.2	11.8	1.550	12.7%	11.2 - 14.9
Sample 10	Breast cancer	30%	20ng	5	15.2	15.5	0.836	5.5%	14.3 - 16.2
			40ng	5	14.5	15.5	1.607	11.1%	11.8 - 15.5
			80ng	5	14.8	14.9	0.782	5.3%	13.7 - 15.5

LoD - ERBB2 copy number amplification

In this study, one (1) sample with ERBB2 amplification was evaluated with 5 levels of copy number alteration. Each dilution level was run with 5 replicates per reagent lot, and 2 reagent lots were used. This results in a total of 10 replicates per level, and 50 aliquots per sample. The cutoff for ERBB2 detection is an observed copy number ≥ 4. The data showed 100% call rate for samples with tumor purity over 10% and two lots of reagents presented with very similar results.

Table 8-12 Detection of ERBB2 Amplification from the ACTOnco LoD SNV Supplement Study

Gene	CNV Status	Dilution Level	Tumor Type	Subject ID	RGT Lot	Mean Tumor purity	Mean Observed CN	Observed CN range	Positive call rate	n/N
ERBB2	Amplification	1	Breast Cancer	Sample 10	B	60.0%	20.9	20.5-21.5	100%	5/5
ERBB2	Amplification	1	Breast Cancer		A	58.2%	19.9	19.5-20.0	100%	5/5
ERBB2	Amplification	2	Breast Cancer		A	10.4%	5.2	5.0-5.5	100%	5/5
ERBB2	Amplification	2	Breast Cancer		B	10.4%	5.2	5.0-5.5	100%	5/5
ERBB2	No amplification	3	Breast Cancer		A	3.8%	2.5	2.5-2.5	0%	0/5
ERBB2	No amplification	3	Breast Cancer		B	3.8%	2.5	2.5-2.5	0%	0/5
ERBB2	No amplification	4	Breast Cancer		B	1.6%	2.5	2.5-2.5	0%	0/5
ERBB2	No amplification	4	Breast Cancer		A	1.6%	2.3	2.0-2.5	0%	0/5
ERBB2	No amplification	5	Breast Cancer		B	0.8%	2.0	2.0-2.0	0%	0/5
ERBB2	No amplification	5	Breast Cancer		A	0.8%	2.0	2.0-2.0	0%	0/5

These findings were further supported in the DNA input study done at ACT Genomics. In that study, two (2) FFPE tumor specimen with low tumor purity and ERBB2 amplification near the cutoff was evaluated in 5 replicates. The data support the consistent repeatability of the ACTOnco ERBB2 amplification assay with a tumor purity at 30% (Table 8-13).

Table 8-13 Tumor Purity and DNA Input on ERBB2 Amplification

Gene	Tumor Type	Sample ID	DNA Level	Mean Reported Tumor purity	Mean Observed CN	Observed CN range	Positive call rate	n/N
ERBB2	Stomach cancer	Sample 1	80ng	30%	7.1	6.5-7.5	100%	5/5
			40ng	30%	7.3	7-7.5	100%	5/5
			20ng	30%	7.3	7-7.5	100%	5/5
ERBB2	Breast cancer	Sample 2	80ng	30%	6.2	6-6.5	100%	5/5
			40ng	30%	6.3	6-6.5	100%	5/5
			20ng	30%	6.3	6-6.5	100%	5/5

8.3 Analytical Specificity / Interference

The objective of the study was to evaluate the potential impact of endogenous and exogenous interfering substances on the performance of the ACTOnco assay.

Two study designs were implemented. The spike-in approach was used to evaluate interfering substances which were hemoglobin, melanin, and wash buffer that could be spiked into an aliquot of a sample. For a potential interferent that could not be "spiked in" such as necrotic tissue, a compare-to-similar approach was used.

In the spike-in design, all substances were tested using six FFPE specimens representative of six tumor types (skin, breast, colorectal, endometrial, lung cancers and kidney cancer).

469 specimens representing 21 cancer types which enrolled in the Method Comparison study were used in the compare to similar approach and divided into test group (necrotic tissue ≥ 10%); and control (necrotic tissue < 10% and matched for mutational variant and level) for analysis. Both the control and test group samples were compared to an appropriate comparator method to determine if the necrotic tissue would alter the agreement of the ACTOnco assay with the comparator.

The specimens included representative variant types in single nucleotide variants (SNV), insertions and deletions (Indels), ERBB2 gene amplification, and tumor mutational burden (TMB).

8.3.1 Spike-in study Arm

SNVs and Indels

Each sample was run as a control (no interferent) and a test (spiked interferent) with 4 replicates in each of the 6 study groups, control (3), hemoglobin (1), melanin (1), wash buffer (1) which resulted in 24 observations (Table 8-14).

Table 8-14 Interfering Substances Tested

Substance	Amount
Hemoglobin	4 mg/mL
Melanin	1ng/μL
Wash buffer	5% wash buffer

One hundred thirty seven (137) variants (126 SNVs, 2 insertions, and 9 deletions) within 104 genes were detected across the 6 samples. A total of 3288 expected positive calls in both controls and test interfering substances were detected which resulted in 100% positive

corrective calls with the lower bound of a 95% CI across all variants at 99.88% (Wilson Score, Table 8-15).

Since the same specimens can be wild type for some locations, force calls were made to the hotspot locations within the assay for each sample for the analysis for negative calls. Out of the 122,664 expected wild type observations, 122,656 were detected which resulted in 99.99% negative corrective calls with the lower bound of a 95%CI across all WTs at 99.99% (Wilson Score, Table 8-15).

Table 8-15 Summary of Correct Calls SNV and Indels

Study Group	Variant/WT	Correct % (n/N)	95% CI
Control	All Variants	100 (1644/1644)	99.77 – 100.00
Hemoglobin	All Variants	100 (548/548)	99.30 – 100.00
Melanin	All Variants	100 (548/548)	99.30 – 100.00
Wash buffer	All Variants	100 (548/548)	99.30 – 100.00
All Conditions	All Variants & Subjects	100 (3288/3288)	99.88 – 100.00
Control	All WT	100.00 (61332/61332)	99.99 – 100.00
Hemoglobin	All WT	99.97 (20438/20444)	99.94 – 99.99
Melanin	All WT	99.99 (20442/20444)	99.96 – 100.00
Wash buffer	All WT	100.00 (20444/20444)	99.98 – 100.00
All Conditions	All WT& Subjects	99.99 (122656/122664)	99.99 – 100.00

* Wilson Score Method

Copy Number Alterations

Copy number alteration was limited to a qualitative reporting of ERBB2 amplifications. The positive and negative percent agreement were generated along with a 2-sided 95% Confidence Interval (Wilson Score method). All 6 samples were evaluated for ERBB2 gene copy number alteration. There was one sample with ERBB2 gene amplification. All 24 calls for ERBB2 gene amplification outcomes were detected which resulted in 100% correct calls (

Table 8-16).

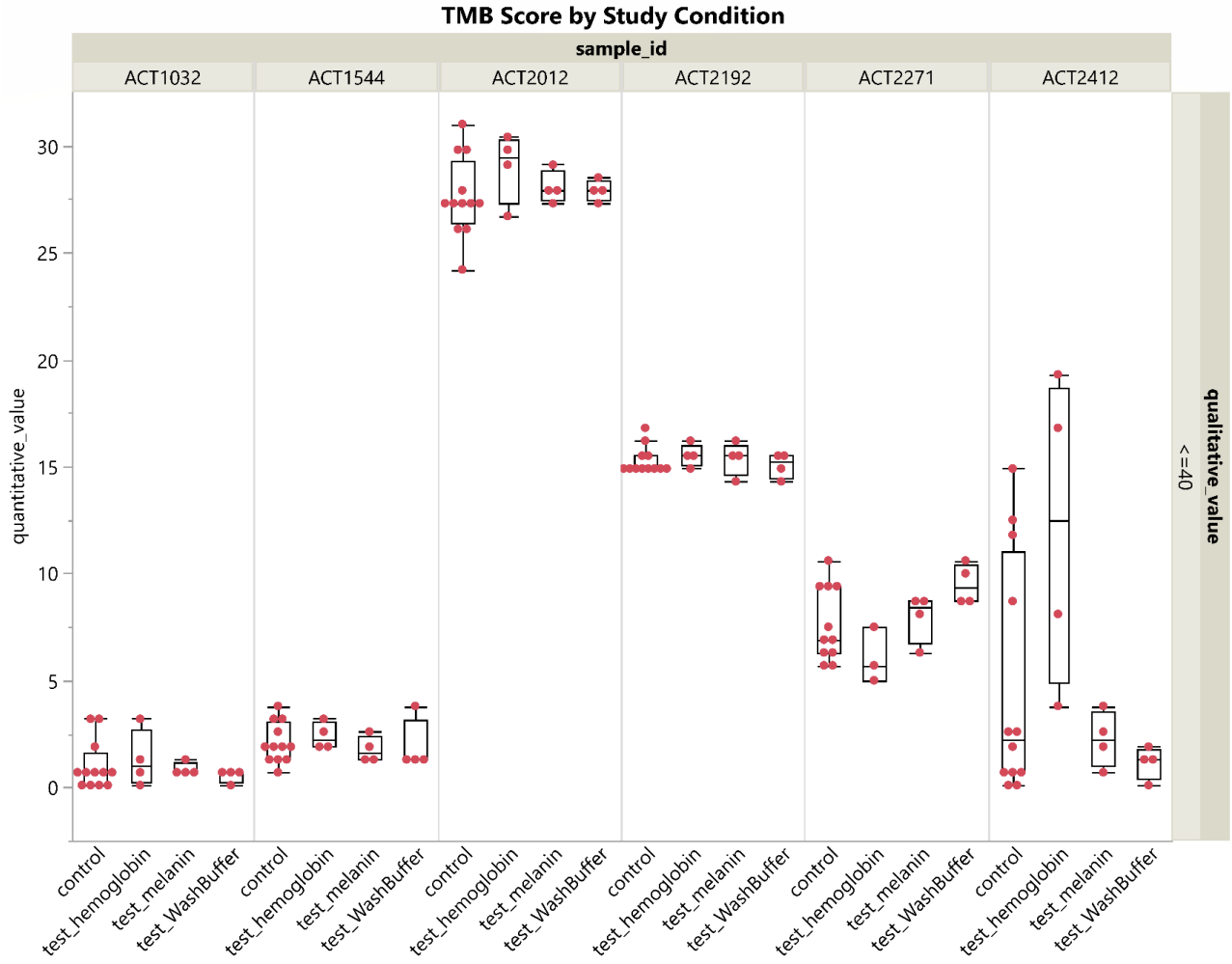
Table 8-16 Summary of Corrective Call Rate for ERBB2 gene copy number alteration

ERBB2 Gene	Study Group	Number Attempted	Number Correct	Correct Percent (%)	95% CI
Amplifications	Control	12	12	100.00	75.75 - 100.0
	Test Hemoglobin	4	4	100.00	51.01 - 100.0
	Test Melanin	4	4	100.00	51.01 - 100.0
	Wash Buffer	4	4	100.00	51.01 - 100.0
	All Conditions	24	24	100.00	86.20 - 100.0
No Amplification	Control	60	60	100.00	93.98 - 100.0
	Test Hemoglobin	20	20	100.00	83.89 - 100.0
	Test Melanin	20	20	100.00	83.89 - 100.0
	Wash Buffer	20	20	100.00	83.89 - 100.0
	All Conditions	120	120	100.00	96.90 - 100.0

Tumor Mutational Burden

The TMB analysis used same specimens that were assessed for SNV/Indels, with tumor purity >30%. The data are presented in Figure 8-2.

Figure 8-2. Visualization of TMB Score Data (Spike-In Study Arm)



All samples tested in the spike-in arm of the study were considered as non-interfering, when compared to control (no interferent).

8.3.2 Compare to Similar Arm

SNV and Indels

For the SNVs and Indels, there were 388 samples were evaluated by the comparator and ACTOnco. Table 8-17 summarizes the correct calls for SNVs and Indels.

Table 8-17 High Level Summary of Correct Calls SNV and Indels

Study Group	Variant/WT	Reported by Comparator	Reported by ACTG	Agreement (%)	95% CI*
Control (<10% Necrosis)	All Variants	1065	1041	97.75	96.67 - 98.48
	All WT	422998	422998	100.00	100.00 – 100.00
Test (10-50% Necrosis)	All Variants	189	186	98.41	95.44 - 99.46
	All WT	69121	69121	100.00	99.99 – 100.00

For the qualitative analysis, the control group had a correct call of 95.97% (999/1041), and the test group, a correct call at 98.39% (183/186). Based on these results, the assay will process samples with up to 50% necrotic tissue.

Copy Number Alterations

For ERBB2 gene amplification, a total of 246 samples were evaluated by the comparator and ACTOnco. 215 samples were in the control (<10% necrotic tissue) and 31 samples were in the test (necrotic range: 10 to 40%). All ten (10) of the ERBB2 gene amplifications in the control group (necrotic tissue < 10%) were detected positive by ACTOnco assay, resulting in 100% correct call rate. All three (3) of the ERBB2 gene amplifications in the test group (necrotic tissue 10-40%) were detected positive by ACTOnco assay, resulting in 100% correct call rate (Table 8-18).

Table 8-18 ERBB2 gene amplification compared with Comparator

Necrotic Tissue Level (Percentage)	ERBB2 Status	Correct Call % (n/N)	95% CI
Control (< 10%)	Amp	100 (10/10)	72.25 - 100.0
Test (10 – 40%)	Amp	100 (3/3)	43.85 - 100.0
Control (< 10%)	Non Amp	99.51 (204/205)	97.29 – 99.91
Test (10 – 40%)	Non Amp	100 (28/28)	87.94 – 100.0

A total of 126 samples were evaluated by both DISH and ACTOnco, there were 3 samples with discordant call which were removed from the analysis. There were one hundred seven (107) samples were in the control (<10% necrotic tissue) and sixteen (16) samples in the test group (necrotic range: 10 to 40%). The call agreement for ERBB2 gene amplification was 100% (35/35) in the control group and 100% (7/7) in the test group (Table 8-19). The call agreement for ERBB2 gene no amplification was 100% (72/72) in the control group and 100% (9/9) in the test group. Since in both cases for either the ERBB2 gene amplification or ERBB2 gene no amplification had 100% agreement with DISH, these data suggest that up to 40% necrotic tissues does not act as an interferent for the ACTOnco assay.

Table 8-19 ERBB2 gene amplification compared with DISH

Necrotic Tissue Level (Percentage)	Correct Percent (n/N)	95% CI
Control (< 10%)	100 (35/35)	90.11-100.00
Test (10 – 40%)	100 (7/7)	64.57-100.00

Tumor Mutational Burden

Fifty (50) samples were used to evaluate the TMB and only those samples that had tumor purity above 30% were used for the study analysis the final n was forty-five (45). For TMB, there were forty one (41) samples in the control group (<10% necrotic tissue) and four (4) samples in the test group (10-15% necrotic tissue). The samples in the test group did not show performance different from the control group.

Based on the results shown in the study, the results showed minimal risk to assay performance from interfering substances and necrosis.

8.4 Cross Contamination

Cross-contamination of DNA samples during the library preparation (contamination from one sample to another within the same sequencing run) was assessed using the positive and negative process control samples for the ACTOnco IVD assay.

The study was conducted by evaluating 15 known variants - consisting of SNVs and deletions - in the positive process control sample. These 15 known variants are considered as positive variants. The same 15 genomic locations were evaluated in the assay negative process control, and they are known to be WT in the negative process control sample.

The study was done with a designated pattern of 12 positive control samples alternating with 12 negative control samples within 96-well plate.

All library construction batch and sequencing runs were completed without QC failure or deviation. No positive variants were detected in the negative control samples, and all these 15 positive variants were detected in every positive control samples. Therefore, no cross-contamination was detected.

8.5 DNA Input

The objective of this study was to evaluate DNA input levels (20, 40, 80 ng) on the performance of the ACTOnco Assay for the reporting on SNVs, INDELs, ERBB2 copy number amplification, and TMB. A set of 10 FFPE samples, representative of 8 cancer types, were sequenced to report SNVs, INDELs, ERBB2 copy number amplification, and TMBs. Five replicates at each DNA input level for each sample were run. This resulted in 50

observations per DNA level (10 samples x 5 replicates) and 150 sample observations for the study (10 samples x 5 replicates x 3 levels).

A total of 4230 variant observations were generated. Data was evaluated between the DNA input levels by looking at the QC outcome of the sample and variants at each input level and evaluating the call rates for each variant at each DNA input level. All of the sample level QC metrics passed without issue for all of the input levels. At the variant level, focusing on variants at 5% with MAF levels between 5% to 10% with variant read counts between 1-15, the data summarized in Table 8-20 indicated that the lower input level of 20ng had 13 observations below the variant read count of 10 with 3 of these at MAF levels above the 5% cutoff as compared to 40ng (15 with 4 above the 5% cutoff) and 80ng (14 with 1 above the 5% cutoff). Due to this, the 80ng level was considered as it would allow more template in the sample processing.

Table 8-20 Outcome of the Variant Level QC Results

DNA Input Level (ng)	N	Base Quality Pass	Variant Total Depth Pass (≥35)	Strand Bias Pass (<0.9)	Assay CO 2% Pass (VRC ≥ 5)	Assay CO 5% Pass (VRC ≥ 10)	Obs. that failed VRC ≥ 10 but above the 5% CO
20	1400	1400/1400	1400/1400	1386/1400	40/40	1307/1360	3
40	1400	1400/1400	1400/1400	1381/1400	40/40	1305/1360	4
80	1400	1400/1400	1399/1400	1379/1400	40/40	1306/1360	1

The variant calls (positive and negative) for the 10 samples at the DNA input of 20, 40, and 80 ng were evaluated by determining the correct calls. The negative process control force calls 755 hotspot variants on all samples and replicates for evaluating variability of negative variant callings. There were a total of 37710 negative calls in the dataset.

Table 8-21 and Table 8-22 summarized the call rates for positive and negative variant callings, respectively for each DNA input level. Furthermore, the call rates for positive and negative variant callings for each DNA input level per sample are shown in Table 8-23 and Table 8-24.

Table 8-21 Call Rate of positive Variant Reported at Each DNA Input Level

DNA Input	Variant Calls	# of Unique Variants	# of Variant Obs. Reported (n)	# of Variant Obs. Attempted (N)	Call Rate (n/N)
20ng	Positive	267	1315	1347	97.6%
40ng	Positive	268	1309	1342	97.5%
80ng	Positive	267	1305	1342	97.2%

Table 8-22 Call Rate of Negative Variant Reported at Each DNA Input Level

DNA Input	Variant Calls	# of Unique Variants	# of Sample	# of Variant Obs. Reported (n)	# of Variant Obs. Attempted (N)	Call Rate (n/N)
20ng	Negative	755	4	15100	15100	100.0%
40ng	Negative	755	4	15100	15100	100.0%
80ng	Negative	755	4	15100	15100	100.0%
20ng	Negative	754	4	15080	15080	100.0%
40ng	Negative	754	4	15080	15080	100.0%
80ng	Negative	754	4	15080	15080	100.0%
20ng	Negative	753	2	7530	7530	100.0%
40ng	Negative	753	2	7530	7530	100.0%
80ng	Negative	753	2	7530	7530	100.0%

Table 8-23 Call Rate of Positive Variant Reported at Each DNA Input Level per Sample

Sample ID	Cancer Type	DNA Input	Variant calls	# of Variants	# of Variant Obs. Reported (n)	# of Variant Obs. Attempted (N)	Call Rate (n/N)
Sample 1	Stomach cancer	20ng	Positive	23	115	115	100.0%
		40ng	Positive	23	114	115	99.1%
		80ng	Positive	23	114	115	99.1%
Sample 2	Urinary system cancer	20ng	Positive	29	140	145	96.6%
		40ng	Positive	29	139	145	95.9%
		80ng	Positive	28	137	144	95.1%
Sample 3	Lung cancer	20ng	Positive	12	60	60	100.0%
		40ng	Positive	12	60	60	100.0%
		80ng	Positive	12	59	59	100.0%
Sample 4	Colon adenocarcinoma	20ng	Positive	49	244	245	99.6%
		40ng	Positive	49	244	244	100.0%
		80ng	Positive	49	243	243	100.0%
Sample 5	Ovarian cancer	20ng	Positive	13	65	65	100.0%
		40ng	Positive	13	65	65	100.0%
		80ng	Positive	13	65	65	100.0%
Sample 6	Endo gland cancer	20ng	Positive	9	45	45	100.0%
		40ng	Positive	9	45	45	100.0%
		80ng	Positive	9	45	45	100.0%
Sample 7	Lung cancer	20ng	Positive	8	40	40	100.0%
		40ng	Positive	8	40	40	100.0%
		80ng	Positive	8	40	40	100.0%
Sample 8	Colon adenocarcinoma	20ng	Positive	60	297	305	97.4%
		40ng	Positive	60	297	302	98.3%
		80ng	Positive	60	298	303	98.3%
Sample 9	Endometrial cancer	20ng	Positive	31	142	159	89.3%
		40ng	Positive	32	144	160	90.0%
		80ng	Positive	32	144	160	90.0%
Sample 10	Breast cancer	20ng	Positive	34	167	168	99.4%
		40ng	Positive	34	161	166	97.0%
		80ng	Positive	34	160	168	95.2%

Table 8-24 Call Rate of Negative Variant Reported at Each DNA Input Level per Sample

Sample ID	Cancer Type	DNA Input	Variant calls	# of variants	# of Variant Obs. Reported (n)	# of Variant Obs. Attempted (N)	Call Rate (n/N)
Sample 1	Stomach cancer	20ng	Negative	755	3775	3775	100.0%
		40ng	Negative	755	3775	3775	100.0%
		80ng	Negative	755	3775	3775	100.0%
Sample 2	Urinary system cancer	20ng	Negative	755	3775	3775	100.0%
		40ng	Negative	755	3775	3775	100.0%
		80ng	Negative	755	3775	3775	100.0%
Sample 3	Lung cancer	20ng	Negative	753	3765	3765	100.0%
		40ng	Negative	753	3765	3765	100.0%
		80ng	Negative	753	3765	3765	100.0%
Sample 4	Colon adenocarcinoma	20ng	Negative	754	3770	3770	100.0%
		40ng	Negative	754	3770	3770	100.0%
		80ng	Negative	754	3770	3770	100.0%
Sample 5	Ovarian cancer	20ng	Negative	754	3770	3770	100.0%
		40ng	Negative	754	3770	3770	100.0%
		80ng	Negative	754	3770	3770	100.0%
Sample 6	Endo gland cancer	20ng	Negative	754	3770	3770	100.0%
		40ng	Negative	754	3770	3770	100.0%
		80ng	Negative	754	3770	3770	100.0%
Sample 7	Lung cancer	20ng	Negative	754	3770	3770	100.0%
		40ng	Negative	754	3770	3770	100.0%
		80ng	Negative	754	3770	3770	100.0%
Sample 8	Colon adenocarcinoma	20ng	Negative	753	3765	3765	100.0%
		40ng	Negative	753	3765	3765	100.0%
		80ng	Negative	753	3765	3765	100.0%
Sample 9	Endometrial cancer	20ng	Negative	755	3775	3775	100.0%
		40ng	Negative	755	3775	3775	100.0%
		80ng	Negative	755	3775	3775	100.0%
Sample 10	Breast cancer	20ng	Negative	755	3775	3775	100.0%
		40ng	Negative	755	3775	3775	100.0%
		80ng	Negative	755	3775	3775	100.0%

These data indicate the amount of DNA added to the assay behaves the same across all DNA input levels tested with respect to the call rates observed but that 80ng DNA input would be better for variants that are near their respective cutoffs.

8.6 DNA Extraction

DNA extraction method was validated based on the invalid rates across multiple tumor types obtained from historical data. The data demonstrated that the DNA extraction has been optimized across tumor types to reasonably conclude that the analytical performance presented in representative across FFPE tumor types. Table 8-25 below shows the historical data for invalid rates from a retrospective review of 1526 specimens tested with ACTOnco. The range of the overall (Pre and Post-run) invalid rates was ranged from 0% to 26%. The data shows that interference effects from different specimens are not significant across different tumor types where the amount of tumor does not increase the chance that the sample cannot be processed to a final result due to either low tumor purity or insufficient DNA yield.

Table 8-25 Specimen Invalid Rates for 27 FFPE Tumor Types

			Pre-Run Invalids		Pre-Run Invalids		Pre-Run Invalids		Pre-Run Invalids (All QC criteria)	Number of Samples pass Pre-run QC	Post-Run Invalids		Overall Invalids (Pre+Post)	
			Insufficient Tumor (< 10%)		Insufficient DNA Integrity (DNA CE < 15%)		Insufficient DNA Yield (< 80ng)				Sequencing Failure (Target base coverage at 100x <85%)			
Cancer Type	Type of Specimen	Number of sample received	Number of sample failed	Percentage	Number of sample failed	Percentage	Number of sample failed	Percentage			Number of sample failed	Percentage	Number of sample failed	Percentage
Adenocarcinoma	FFPE	13	1	7.69%	0	0.00%	1	7.69%	15.38%	11	0	0.00%	2	15.38%
Brain cancer	FFPE	29	0	0.00%	0	0.00%	3	10.34%	10.34%	26	0	0.00%	3	10.34%
Breast cancer	FFPE	160	10	6.25%	7	4.38%	9	5.63%	16.25%	134	0	0.00%	26	16.25%
Cervical cancer	FFPE	22	0	0.00%	3	13.64%	0	0.00%	13.64%	19	0	0.00%	3	13.64%
Cholangiocarcinoma	FFPE	65	6	9.23%	0	0.00%	1	1.54%	10.77%	58	0	0.00%	7	10.77%
Colon cancer	FFPE	158	19	12.03%	6	3.80%	2	1.27%	17.09%	131	0	0.00%	27	17.09%
Esophageal cancer	FFPE	31	1	3.23%	1	3.23%	0	0.00%	6.45%	29	0	0.00%	2	6.45%
Gastric cancer	FFPE	44	5	11.36%	1	2.27%	1	2.27%	15.91%	37	0	0.00%	7	15.91%
Gastrointestinal stromal tumor	FFPE	9	0	0.00%	2	22.22%	0	0.00%	22.22%	7	0	0.00%	2	22.22%
Glioblastoma Multiforme	FFPE	39	0	0.00%	0	0.00%	1	2.56%	2.56%	38	0	0.00%	1	2.56%
Head and Neck Cancer	FFPE	20	0	0.00%	0	0.00%	1	5.00%	5.00%	19	0	0.00%	1	5.00%
Hepatocellular carcinoma	FFPE	31	1	3.23%	3	9.68%	1	3.23%	16.13%	26	0	0.00%	5	16.13%
Kidney cancer	FFPE	16	1	6.25%	0	0.00%	1	6.25%	12.50%	14	0	0.00%	2	12.50%
Lung cancer	FFPE	303	36	11.88%	5	1.65%	28	9.24%	22.77%	234	0	0.00%	69	22.77%
Melanoma	FFPE	9	0	0.00%	0	0.00%	0	0.00%	0.00%	9	0	0.00%	0	0.00%
Neuroendocrine carcinoma	FFPE	18	1	5.56%	1	5.56%	0	0.00%	11.11%	16	0	0.00%	2	11.11%
Oral cancer	FFPE	20	1	5.00%	0	0.00%	0	0.00%	5.00%	19	0	0.00%	1	5.00%
Ovarian cancer	FFPE	140	15	10.71%	6	4.29%	0	0.00%	15.00%	119	0	0.00%	21	15.00%
Pancreatic cancer	FFPE	142	23	16.20%	2	1.41%	12	8.45%	26.06%	105	0	0.00%	37	26.06%

			Pre-Run Invalids		Pre-Run Invalids		Pre-Run Invalids				Post-Run Invalids		Overall Invalids (Pre+Post)	
			Insufficient Tumor (< 10%)		Insufficient DNA Integrity (DNA CE < 15%)		Insufficient DNA Yield (< 80ng)		Pre-Run Invalids (All QC criteria)	Number of Samples pass Pre-run QC	Sequencing Failure (Target base coverage at 100x <85%)			
Cancer Type	Type of Specimen	Number of sample received	Number of sample failed	Percentage	Number of sample failed	Percentage	Number of sample failed	Percentage			Number of sample failed	Percentage	Number of sample failed	Percentage
Peritoneal cancer	FFPE	10	0	0.00%	0	0.00%	1	10.00%	10.00%	9	0	0.00%	1	10.00%
Prostate cancer	FFPE	21	0	0.00%	0	0.00%	2	9.52%	9.52%	19	0	0.00%	2	9.52%
Sarcoma	FFPE	80	4	5.00%	3	3.75%	2	2.50%	11.25%	71	0	0.00%	9	11.25%
Small bowel cancer	FFPE	9	0	0.00%	0	0.00%	2	22.22%	22.22%	7	0	0.00%	2	22.22%
Thymic cancer	FFPE	13	2	15.38%	1	7.69%	0	0.00%	23.08%	10	0	0.00%	3	23.08%
Thyroid cancer	FFPE	9	0	0.00%	1	11.11%	0	0.00%	11.11%	8	0	0.00%	1	11.11%
Unknown primary	FFPE	25	5	20.00%	0	0.00%	0	0.00%	20.00%	20	0	0.00%	5	20.00%
Urothelial cancer	FFPE	25	3	12.00%	1	4.00%	0	0.00%	16.00%	21	0	0.00%	4	16.00%
Uterine cancer	FFPE	65	2	3.08%	2	3.08%	2	3.08%	9.23%	59	2	3.08%	8	12.31%
Total	**FFPE**	**1526**	**136**	**8.91%**	**45**	**2.95%**	**70**	**4.59%**	**16.45%**	**1275**	**2**	**0.13%**	**253**	**16.58%**

8.7 Method Comparison Data

The method comparison study assesses the concordance between the results of ACTOnco assay and results of an appropriate comparator.

For single nucleotide variants (SNVs), multi-nucleotide variants (MNVs), insertions and deletions (indels), the comparator was a well-validated NGS based assay. For ERBB2 gene amplification, the comparator was HER2 Dual ISH DNA Probe Cocktail test (DISH). For TMB, the comparator was whole exome sequencing (WES, next generation sequencing).

Qualitative data for SNVs, indels, and ERBB2 gene amplification was defined as positive calls and negative calls, comparing to their respective comparators in 2 x 2 tables. These values were then calculated as positive percent agreement (PPA) and negative percent agreement (NPA) with 2 sided 95% confidence intervals (Wilson Score).

SNV/Indels

Four hundred thirty eight (438) FFPE samples spanning 21 cancer types were evaluated for SNVs, MNVs, insertions, and deletions between ACTOnco and a well-validated NGS based assay. Across all variant types, the comparator assay detected 1254 mutational variants and of these 1175 were unique observed in 59 exons over 106 genes. Of these, ACTOnco assay detected 1227, resulting in a PPA of 97.85% (95% CI: 96.89 to 98.52).

Within the same samples, the comparator assay reported 457,446 wild type. Of these, 457,319 were also reported as wild type by the ACTOnco assay, resulting in an NPA of 99.97 (95% CI: 99.97 to 99.98).

Table 8-26: Performance Summary of Mutational Variants (Across All SNV, Insertions and Deletions)

ACTOnco	**Comparator**		
	Positive	**Negative**	**Total**
Positive	1227	127	1354
Negative	27	457319	457346
Total	1254	457446	458700

	Performance Summary	
Agreement Statistic	**Point Estimate (Percentage)**	**95% CI Wilson Score**
PPA	1227/1254 = 97.85	96.89 – 98.52
NPA	457319/457446 = 99.97	99.97 – 99.98

Data assess with the following tumor types: brain, breast, cervical, cholangiocarcinoma, colon adenocarcinoma, endo gland, endometrial, esophageal, gallbladder, GIST, kidney, liver, lung, mesenchymal cell neoplasm, oral cavity, ovarian, prostate, skin, stomach, tumor of exocrine pancreas, urinary system. The agreement at mutational level are listed in Table 8-27 below.

Table 8-27 Performance summary of percent agreement at mutational-level

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
AKT1_chr14_105236728_G>C	AKT1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AKT1_chr14_105239823_G>A	AKT1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
AKT1_chr14_105243045_A>G	AKT1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
AKT3_chr1_243716188_T>C	AKT3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ALK_chr2_29436850_C>A	ALK	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ALK_chr2_29436880_T>A	ALK	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARAF_chrX_47426093_GC>G	ARAF	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A_chr1_27023860_C>CG	ARID1A	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A_chr1_27056262_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27056277_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27057732_GCAGCAGC>G	ARID1A	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27057788_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ARID1A_chr1_27057801_G>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27057976_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27059176_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ARID1A_chr1_27059227_A>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27087485_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27087500_A>G	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27087503_C>T	ARID1A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
ARID1A_chr1_27087891_TCGGCCACCCAG>T	ARID1A	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
ARID1A_chr1_27087961_C>CG	ARID1A	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27088760_A>C	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27088782_T>TC	ARID1A	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27089709_G>C	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27089758_C>G	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27092804_A>AC	ARID1A	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A_chr1_27092809_C>T	ARID1A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
ARID1A_chr1_27092986_G>A	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27094392_A>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27097621_CA>C	ARID1A	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
ARID1A_chr1_27097689_A>T	ARID1A	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
ARID1A_chr1_27097691_A>G	ARID1A	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
ARID1A_chr1_27097721_G>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27099881_G>A	ARID1A	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
ARID1A_chr1_27099947_C>T	ARID1A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
ARID1A_chr1_27099963_A>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27100837_G>A	ARID1A	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A_chr1_27100976_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101078_CA>C	ARID1A	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101082_T>G	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101116_AC>A	ARID1A	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
ARID1A_chr1_27101135_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101219_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101312_ATGC>A	ARID1A	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101435_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101511_G>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27101519_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27102103_A>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
ARID1A_chr1_27102138_A>AG	ARID1A	INS	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
ARID1A_chr1_27105809_G>GT	ARID1A	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27105853_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27105880_C>G	ARID1A	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ARID1A_chr1_27105892_C>T	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27105965_TC>T	ARID1A	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27106168_G>C	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27106180_G>C	ARID1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ARID1A_chr1_27106263_A>AC	ARID1A	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A_chr1_27106333_G>A	ARID1A	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ARID1A_chr1_27106648_G>A	ARID1A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
AR_chrX_66765778_CG>C	AR	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
AR_chrX_66766120_C>T	AR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108098563_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ATM_chr11_108100002_C>A	ATM	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATM_chr11_108100039_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108106435_ATC>G	ATM	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATM_chr11_108115601_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108119732_T>TACAG	ATM	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108121593_CAA>C	ATM	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
ATM_chr11_108121756_G>C	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108122680_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108122697_T>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108122700_T>C	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108123567_A>G	ATM	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATM_chr11_108123579_T>C	ATM	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	434	435
ATM_chr11_108123587_A>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108127050_T>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
ATM_chr11_108128319_A>C	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108142070_A>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108143287_T>C	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108153430_C>T	ATM	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATM_chr11_108155018_A>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108155132_G>A	ATM	SNV	100	51.01	100	NaN	NaN	NaN	100	51.01	100	NaN	NaN	NaN	4	0	0	0	4
ATM_chr11_108158382_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108158393_C>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108159742_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108160467_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108170491_A>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108170587_C>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108175459_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108186610_G>A	ATM	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATM_chr11_108190711_A>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108196825_C>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108199815_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ATM_chr11_108200946_C>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108200967_T>C	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108201008_C>T	ATM	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATM_chr11_108202261_G>T	ATM	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
ATM_chr11_108202715_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108203493_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108205756_C>T	ATM	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
ATM_chr11_108205766_G>A	ATM	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATM_chr11_108205805_C>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108206666_A>T	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108216524_C>CA	ATM	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
ATM_chr11_108225556_G>A	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATM_chr11_108235879_C>T	ATM	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
ATM_chr11_108236141_T>G	ATM	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76776337_G>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76777843_T>C	ATRX	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATRX_chrX_76812977_A>G	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76845343_C>T	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76855010_GCTA>G	ATRX	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76888736_C>T	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76889105_C>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76890112_G>T	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76891445_T>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76907620_C>T	ATRX	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATRX_chrX_76907714_T>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76918931_CT>C	ATRX	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76919018_C>T	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76937855_C>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76937942_C>G	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76938493_G>GT	ATRX	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76938741_T>G	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76938982_T>G	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76939180_G>A	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ATRX_chrX_76939300_T>G	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATRX_chrX_76939947_A>C	ATRX	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142168300_T>C	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142177859_G>A	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142188337_A>C	ATR	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATR_chr3_142204016_C>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
ATR_chr3_142204115_C>T	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142212159_A>G	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142215345_A>G	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142215882_C>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142215947_G>GAGAATC	ATR	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142216021_A>G	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142226816_G>A	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142242920_T>TC	ATR	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142254985_T>C	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142259869_T>C	ATR	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATR_chr3_142261533_T>C	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142266649_T>A	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142268484_G>A	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142272581_A>G	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142272780_C>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142274712_C>T	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142274770_T>C	ATR	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ATR_chr3_142274957_G>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142275334_T>C	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142275360_A>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142278201_T>C	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142281483_T>C	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ATR_chr3_142281705_C>T	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142281879_T>C	ATR	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ATR_chr3_142281930_G>A	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ATR_chr3_142285065_T>C	ATR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AXL_chr19_41725307_C>T	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
AXL_chr19_41737174_A>G	AXL	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
AXL_chr19_41743958_G>A	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AXL_chr19_41744483_G>A	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AXL_chr19_41745214_G>A	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AXL_chr19_41749577_G>A	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
AXL_chr19_41762376_G>A	AXL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52436815_CTACCT>C	BAP1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52436856_G>A	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52437777_G>T	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52437893_G>T	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52438503_C>G	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52438553_C>T	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52438554_G>A	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52439921_C>CT	BAP1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52440372_C>G	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52440889_CT>C	BAP1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BAP1_chr3_52442077_C>T	BAP1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRAF_chr7_140453131_A>G	BRAF	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
BRAF_chr7_140481397_C>A	BRAF	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41197711_G>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41199671_T>C	BRCA1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA1_chr17_41226390_G>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41234451_G>A	BRCA1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA1_chr17_41243479_CTTGA>C	BRCA1	DEL	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
BRCA1_chr17_41243509_T>C	BRCA1	SNV	100	43.85	100	NaN	NaN	NaN	100	43.85	100	NaN	NaN	NaN	3	0	0	0	3
BRCA1_chr17_41243736_C>T	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA1_chr17_41243886_T>G	BRCA1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA1_chr17_41243948_C>G	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
BRCA1_chr17_41244421_TG>T	BRCA1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41244699_G>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41244716_A>T	BRCA1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA1_chr17_41244822_T>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41244826_C>A	BRCA1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA1_chr17_41245197_G>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41245201_T>C	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA1_chr17_41245668_A>C	BRCA1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA1_chr17_41245699_TAGA>T	BRCA1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41245861_G>A	BRCA1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA1_chr17_41246037_C>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41246095_C>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41246164_C>T	BRCA1	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
BRCA1_chr17_41246484_T>C	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA1_chr17_41246512_G>A	BRCA1	SNV	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
BRCA1_chr17_41246724_C>T	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41246815_C>A	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41246856_G>A	BRCA1	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	434	435
BRCA1_chr17_41251842_C>T	BRCA1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA1_chr17_41251876_G>A	BRCA1	SNV	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
BRCA1_chr17_41256153_C>T	BRCA1	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
BRCA1_chr17_41256155_G>T	BRCA1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA2_chr13_32893271_A>G	BRCA2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA2_chr13_32893361_A>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA2_chr13_32893376_C>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32905123_T>C	BRCA2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	436	437
BRCA2_chr13_32905123_TGACA>T	BRCA2	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA2_chr13_32905125_A>G	BRCA2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
BRCA2_chr13_32906558_T>A	BRCA2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA2_chr13_32906739_C>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32906781_C>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32906817_C>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32907183_A>G	BRCA2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA2_chr13_32910614_T>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32911330_T>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32911748_A>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32911937_A>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32911943_A>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32912416_A>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32912855_G>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32912864_C>T	BRCA2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA2_chr13_32912982_T>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32913226_A>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32913242_G>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32913320_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA2_chr13_32913723_G>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32913983_A>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914004_G>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914114_T>G	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914184_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914474_A>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914509_G>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914592_C>T	BRCA2	SNV	100	43.85	100	NaN	NaN	NaN	100	43.85	100	NaN	NaN	NaN	3	0	0	0	3
BRCA2_chr13_32914733_G>C	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32914788_G>C	BRCA2	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
BRCA2_chr13_32914814_C>T	BRCA2	SNV	100	64.57	100	100	99.11	100	100	64.57	100	100	99.11	100	7	0	0	430	437

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
BRCA2_chr13_32915102_C>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32929042_C>G	BRCA2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA2_chr13_32929092_T>G	BRCA2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
BRCA2_chr13_32930651_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32937429_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32937488_G>T	BRCA2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
BRCA2_chr13_32937491_A>T	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32937521_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
BRCA2_chr13_32950876_G>A	BRCA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BRCA2_chr13_32968861_T>C	BRCA2	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
BTK_chrX_100617192_T>A	BTK	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
BTK_chrX_100617213_C>T	BTK	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CCND1_chr11_69457805_G>A	CCND1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND1_chr11_69458649_T>C	CCND1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND1_chr11_69458720_C>T	CCND1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CCND1_chr11_69465976_G>T	CCND1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND1_chr11_69466027_G>T	CCND1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND2_chr12_4383372_C>T	CCND2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND2_chr12_4388016_C>T	CCND2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND2_chr12_4388082_A>T	CCND2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND2_chr12_4398101_C>T	CCND2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND2_chr12_4398154_G>A	CCND2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CCND2_chr12_4409106_C>G	CCND2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCND3_chr6_41903745_CG>C	CCND3	DEL	NaN	NaN	NaN	99.56	97.54	99.92	0	0	79.35	100	98.32	100	0	1	0	225	226
CCNE1_chr19_30308425_A>G	CCNE1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCNE1_chr19_30312636_A>G	CCNE1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCNE1_chr19_30312722_A>T	CCNE1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CCNE1_chr19_30313451_G>C	CCNE1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
CCNE1_chr19_30313515_GTGC>G	CCNE1	DEL	NaN	NaN	NaN	96.49	94.08	97.94	0	0	22.81	100	98.94	100	0	13	0	357	370
CDK12_chr17_37618612_C>A	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37627472_G>A	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37627563_A>C	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37649077_G>A	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37649093_T>G	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37649108_A>G	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37650845_C>T	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37686934_C>G	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37686985_G>A	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37687088_G>C	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK12_chr17_37687514_G>A	CDK12	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK2_chr12_56360801_C>A	CDK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK2_chr12_56364912_G>A	CDK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
CDK4_chr12_58142334_G>A	CDK4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK4_chr12_58144849_C>A	CDK4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDK6_chr7_92404051_C>T	CDK6	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
CDKN1B_chr12_12870809_C>A	CDKN1B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN1B_chr12_12870927_A>C	CDKN1B	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CDKN1B_chr12_12871056_C>G	CDKN1B	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CDKN1B_chr12_12871129_T>C	CDKN1B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN1B_chr12_12871876_C>G	CDKN1B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2A_chr9_21970971_G>C	CDKN2A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
CDKN2A_chr9_21970971_G>T	CDKN2A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
CDKN2A_chr9_21970989_A>T	CDKN2A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2A_chr9_21971029_C>T	CDKN2A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
CDKN2A_chr9_21971060_C>A	CDKN2A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2A_chr9_21971096_C>A	CDKN2A	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
CDKN2A_chr9_21974751_C>A	CDKN2A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2A_chr9_21974777_GC>G	CDKN2A	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CDKN2B_chr9_22006077_C>T	CDKN2B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2B_chr9_22006212_A>C	CDKN2B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2B_chr9_22008850_C>A	CDKN2B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CDKN2B_chr9_22008850_C>T	CDKN2B	SNV	100	20.65	100	100	99.11	100	100	20.65	100	100	99.11	100	1	0	0	429	430
CDKN2B_chr9_22008892_C>T	CDKN2B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CHEK1_chr11_125507368_A>G	CHEK1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CHEK1_chr11_125507417_G>A	CHEK1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CHEK1_chr11_125513701_C>T	CHEK1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
CHEK1_chr11_125514461_G>A	CHEK1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CHEK2_chr22_29091710_A>G	CHEK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3778098_T>TG	CREBBP	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CREBBP_chr16_3778321_C>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3778390_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3778424_T>G	CREBBP	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CREBBP_chr16_3778439_T>TTGC	CREBBP	INS	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
CREBBP_chr16_3778821_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3778906_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3779076_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
CREBBP_chr16_3779094_C>T	CREBBP	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CREBBP_chr16_3779401_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3779496_C>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3779748_G>C	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3779814_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3781832_T>C	CREBBP	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CREBBP_chr16_3786177_T>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3789619_C>T	CREBBP	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
CREBBP_chr16_3801783_G>GA	CREBBP	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3807319_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3807814_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3807956_C>T	CREBBP	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CREBBP_chr16_3808034_C>T	CREBBP	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CREBBP_chr16_3817742_G>C	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3817820_T>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3819206_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3819284_T>C	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3820773_G>A	CREBBP	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
CREBBP_chr16_3820816_G>A	CREBBP	SNV	100	34.24	100	100	99.12	100	100	34.24	100	100	99.12	100	2	0	0	435	437
CREBBP_chr16_3820825_C>T	CREBBP	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
CREBBP_chr16_3820881_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3820912_G>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3823777_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3824614_T>C	CREBBP	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CREBBP_chr16_3827646_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3830736_T>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3841994_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3843507_T>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3860674_C>T	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3900704_G>A	CREBBP	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP_chr16_3900803_C>A	CREBBP	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CSF1R_chr5_149447798_G>C	CSF1R	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CSF1R_chr5_149447825_C>T	CSF1R	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
DDR2_chr1_162745588_G>A	DDR2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
DDR2_chr1_162745627_C>T	DDR2	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
EGFR_chr7_55233051_GG>AA	EGFR	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
EGFR_chr7_55233087_G>A	EGFR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
EGFR_chr7_55242505_A>T	EGFR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
EGFR_chr7_55249058_G>A	EGFR	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
EGFR_chr7_55259469_G>A	EGFR	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
EGFR_chr7_55260498_A>G	EGFR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB2_chr17_37879601_T>A	ERBB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB2_chr17_37881111_C>T	ERBB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB2_chr17_37881351_C>T	ERBB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB2_chr17_37881625_T>C	ERBB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB2_chr17_37882874_C>T	ERBB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB3_chr12_56481627_C>T	ERBB3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB3_chr12_56481632_C>T	ERBB3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB3_chr12_56481672_C>G	ERBB3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB3_chr12_56492581_C>A	ERBB3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERBB4_chr2_212488691_T>A	ERBB4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERCC2_chr19_45860548_G>A	ERCC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERCC2_chr19_45867709_C>T	ERCC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERCC2_chr19_45871995_C>T	ERCC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
ESR1_chr6_152163865_C>A	ESR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ESR1_chr6_152265380_G>T	ESR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ESR1_chr6_152419956_G>A	ESR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ESR1_chr6_152419988_T>A	ESR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
EZH2_chr7_148506408_C>T	EZH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89805041_C>T	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89805083_C>A	FANCA	SNV	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
FANCA_chr16_89805301_G>C	FANCA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCA_chr16_89805325_G>A	FANCA	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
FANCA_chr16_89809272_A>G	FANCA	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
FANCA_chr16_89811443_G>A	FANCA	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
FANCA_chr16_89813029_C>G	FANCA	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
FANCA_chr16_89813078_G>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89816219_C>A	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89825107_G>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89831344_CAG>C	FANCA	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
FANCA_chr16_89833576_G>C	FANCA	SNV	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	432	437
FANCA_chr16_89833595_G>A	FANCA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCA_chr16_89836249_G>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89836317_C>T	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89836374_C>T	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89838097_G>A	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89838136_T>C	FANCA	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
FANCA_chr16_89838187_G>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89839791_A>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89842176_C>G	FANCA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCA_chr16_89845355_C>T	FANCA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCA_chr16_89846290_C>T	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89846358_C>A	FANCA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCA_chr16_89849286_G>A	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89851311_A>G	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89857830_G>A	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89862403_G>A	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89865579_C>CCT	FANCA	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89865610_T>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89874739_C>T	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA_chr16_89877407_G>C	FANCA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10070387_C>A	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
FANCD2_chr3_10081439_T>C	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10083327_C>G	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10094093_A>T	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10094159_A>G	FANCD2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FANCD2_chr3_10105512_G>A	FANCD2	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
FANCD2_chr3_10105514_GC>AT	FANCD2	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10107113_G>A	FANCD2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
FANCD2_chr3_10108929_A>T	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10114976_C>G	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10116301_A>C	FANCD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCD2_chr3_10140447_ATGAGAG>A	FANCD2	DEL	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FBXW7_chr4_153244167_C>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153245501_G>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153247184_G>GC	FBXW7	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153249366_T>C	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153249463_T>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153249483_T>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153250906_G>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153251885_T>C	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153253749_CTCT>C	FBXW7	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153253802_A>G	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153332483_G>A	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FBXW7_chr4_153332708_C>T	FBXW7	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
FGF19_chr11_69514140_C>T	FGF19	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGF19_chr11_69514296_C>T	FGF19	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGF3_chr11_69625110_G>A	FGF3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGF3_chr11_69625158_C>A	FGF3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
FGF3_chr11_69625326_G>A	FGF3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGF3_chr11_69633536_G>T	FGF3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR1_chr8_38274929_C>T	FGFR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR1_chr8_38279354_C>T	FGFR1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	436	437
FGFR1_chr8_38285948_G>A	FGFR1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR2_chr10_123279660_G>A	FGFR2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR3_chr4_1806113_A>G	FGFR3	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
FGFR3_chr4_1806188_A>G	FGFR3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR4_chr5_176516647_C>G	FGFR4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR4_chr5_176517553_G>A	FGFR4	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
FGFR4_chr5_176518070_G>A	FGFR4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR4_chr5_176518092_T>C	FGFR4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
FGFR4_chr5_176519396_G>A	FGFR4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR4_chr5_176522560_G>A	FGFR4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR4_chr5_176523282_C>T	FGFR4	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
FLT3_chr13_28608075_C>T	FLT3	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
FLT3_chr13_28636080_G>T	FLT3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
GATA2_chr3_128199899_T>C	GATA2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
HNF1A_chr12_121431496_G>T	HNF1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
HRAS_chr11_533569_C>T	HRAS	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
IGF1R_chr15_99456467_G>A	IGF1R	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	434	437
IGF1R_chr15_99472828_G>A	IGF1R	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
IGF1R_chr15_99478548_C>T	IGF1R	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KDR_chr4_55955916_G>T	KDR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KDR_chr4_55955918_A>G	KDR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KIT_chr4_55594191_A>G	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
KIT_chr4_55594215_G>A	KIT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KIT_chr4_55595504_C>A	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
KIT_chr4_55595531_G>A	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
KIT_chr4_55595534_A>G	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
KIT_chr4_55595537_G>A	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
KIT_chr4_55595546_T>C	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
KIT_chr4_55595557_A>G	KIT	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	436	437
KIT_chr4_55597583_T>C	KIT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KIT_chr4_55598090_G>T	KIT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KIT_chr4_55603442_A>G	KIT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
KRAS_chr12_25398211_T>C	KRAS	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
KRAS_chr12_25398251_A>C	KRAS	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MAP2K1_chr15_66727562_T>C	MAP2K1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
MAP2K1_chr15_66782894_C>A	MAP2K1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MAP2K4_chr17_12011150_A>G	MAP2K4	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
MDM2_chr12_69233372_A>G	MDM2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MDM4_chr1_204518431_C>T	MDM4	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
MDM4_chr1_204518457_A>C	MDM4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MET_chr7_116339881_A>T	MET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MET_chr7_116340039_A>G	MET	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
MET_chr7_116340177_G>A	MET	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MET_chr7_116414961_G>A	MET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MET_chr7_116418842_T>C	MET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MET_chr7_116423365_G>A	MET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MLH1_chr3_37035097_C>T	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MLH1_chr3_37038192_G>C	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MLH1_chr3_37045935_C>T	MLH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37053505_G>T	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MLH1_chr3_37053562_C>T	MLH1	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
MLH1_chr3_37061886_G>A	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
MLH1_chr3_37067192_C>T	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MLH1_chr3_37070280_G>A	MLH1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MLH1_chr3_37083821_C>T	MLH1	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
MLH1_chr3_37089122_TGAA>T	MLH1	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37089130_AA>GC	MLH1	MNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37090075_T>C	MLH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37090464_C>T	MLH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37090471_A>G	MLH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MLH1_chr3_37092092_T>C	MLH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MLH1_chr3_37092136_A>G	MLH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH2_chr2_47630394_T>C	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MSH2_chr2_47637371_A>G	MSH2	SNV	100	51.01	100	NaN	NaN	NaN	100	51.01	100	NaN	NaN	NaN	4	0	0	0	4
MSH2_chr2_47639658_G>A	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MSH2_chr2_47643469_T>A	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47643504_G>A	MSH2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH2_chr2_47643537_C>T	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47690263_T>C	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47698159_G>A	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MSH2_chr2_47698172_T>C	MSH2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH2_chr2_47702337_C>T	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47702364_G>A	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47705488_C>T	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47705625_G>A	MSH2	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
MSH2_chr2_47705647_A>G	MSH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH2_chr2_47707892_A>G	MSH2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH2_chr2_47709997_C>G	MSH2	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH6_chr2_48010479_C>T	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MSH6_chr2_48018236_G>T	MSH6	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
MSH6_chr2_48023077_G>A	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48023174_C>T	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48025766_T>G	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48025988_GC>AA	MSH6	MNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH6_chr2_48026251_A>G	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
MSH6_chr2_48026596_A>G	MSH6	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH6_chr2_48026792_G>A	MSH6	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
MSH6_chr2_48026938_A>G	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027164_T>C	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027381_T>TA	MSH6	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027422_C>G	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027531_C>CA	MSH6	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027683_A>T	MSH6	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
MSH6_chr2_48027815_C>G	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48027853_C>T	MSH6	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH6_chr2_48028223_G>A	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48030630_C>T	MSH6	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MSH6_chr2_48030646_C>A	MSH6	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
MSH6_chr2_48032098_A>T	MSH6	SNV	100	60.97	100	NaN	NaN	NaN	100	60.97	100	NaN	NaN	NaN	6	0	0	0	6
MSH6_chr2_48032799_T>C	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48033352_C>T	MSH6	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
MSH6_chr2_48033417_T>C	MSH6	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MSH6_chr2_48033998_A>AGACT	MSH6	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MTOR_chr1_11182098_T>G	MTOR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MTOR_chr1_11190824_G>T	MTOR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MTOR_chr1_11217242_C>T	MTOR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MYCL_chr1_40366961_G>A	MYCL	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MYC_chr8_128750528_TCAC>T	MYC	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
MYC_chr8_128752745_C>A	MYC	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN_chr8_90955525_G>A	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN_chr8_90967511_CT>C	NBN	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
NBN_chr8_90971041_C>T	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NBN_chr8_90983460_G>A	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN_chr8_90993059_A>G	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN_chr8_90994990_T>C	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN_chr8_90995017_A>G	NBN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29508741_GGGT>G	NF1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29527612_A>G	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NF1_chr17_29528490_G>C	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29533315_C>T	NF1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NF1_chr17_29541542_ATAAG>A	NF1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29553483_C>T	NF1	SNV	0	0	65.76	100	99.13	100	NaN	NaN	NaN	99.54	98.35	99.87	0	0	2	436	438
NF1_chr17_29553493_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29554278_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29554590_T>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29554622_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29556157_G>A	NF1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
NF1_chr17_29556190_C>T	NF1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
NF1_chr17_29556978_G>A	NF1	SNV	NaN	NaN	NaN	99.77	98.71	99.96	0	0	79.35	100	99.12	100	0	1	0	435	436
NF1_chr17_29559765_A>G	NF1	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	434	437
NF1_chr17_29559887_A>G	NF1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NF1_chr17_29560064_G>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29560217_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NF1_chr17_29562656_CTGTT>C	NF1	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NF1_chr17_29562668_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29576037_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
NF1_chr17_29585419_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29652890_A>G	NF1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NF1_chr17_29653256_A>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29654553_C>T	NF1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NF1_chr17_29654658_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29654685_A>G	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29654691_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29654736_C>T	NF1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NF1_chr17_29663431_G>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29663483_T>G	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29663491_G>A	NF1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NF1_chr17_29663492_G>A	NF1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NF1_chr17_29664512_G>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29664852_A>G	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29665113_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29665751_CACTT>C	NF1	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NF1_chr17_29665813_T>C	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29667647_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29670042_T>C	NF1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NF1_chr17_29679313_G>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29679348_G>C	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29679436_A>G	NF1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NF1_chr17_29684022_A>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29684314_G>A	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29684326_C>T	NF1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NF1_chr17_29685599_A>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF1_chr17_29685610_C>T	NF1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF2_chr22_30000094_A>G	NF2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
NF2_chr22_30054192_T>C	NF2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF2_chr22_30057243_A>G	NF2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF2_chr22_30067870_C>T	NF2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NF2_chr22_30069355_A>T	NF2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139390941_G>C	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139391015_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH1_chr9_139391163_T>C	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139391522_G>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139391778_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139391826_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139391833_G>A	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139393680_TC>AA	NOTCH1	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139393696_G>A	NOTCH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NOTCH1_chr9_139395132_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139395252_C>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139396209_G>GC	NOTCH1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139396278_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139396532_T>C	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139396850_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139399386_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139399847_TG>AT	NOTCH1	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139400180_G>T	NOTCH1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH1_chr9_139401260_T>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139401317_T>TA	NOTCH1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139401358_C>A	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139401422_A>G	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139401425_C>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139401830_G>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
NOTCH1_chr9_139402695_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139402834_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139403333_G>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139403372_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139404282_C>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139405126_C>T	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139405649_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139407553_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH1_chr9_139407846_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139409076_T>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139409089_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139409816_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139409943_CC>TT	NOTCH1	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139409974_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139410085_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139410139_T>C	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139411829_C>T	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH1_chr9_139412341_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139412639_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH1_chr9_139412673_G>A	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH1_chr9_139412697_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139413084_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139413118_C>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139413214_C>T	NOTCH1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NOTCH1_chr9_139413222_T>C	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139413921_T>C	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139417343_C>T	NOTCH1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH1_chr9_139417353_C>T	NOTCH1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
NOTCH1_chr9_139417378_G>T	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH1_chr9_139417398_C>G	NOTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120458111_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120458122_A>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120458267_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120458318_G>A	NOTCH2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH2_chr1_120458978_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120459083_TG>T	NOTCH2	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH2_chr1_120459122_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120459148_T>C	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120461995_G>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120462092_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120462164_GCATCTT>G	NOTCH2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120462974_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120465262_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH2_chr1_120466307_C>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120466326_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120467986_T>C	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH2_chr1_120468127_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120468184_TG>T	NOTCH2	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH2_chr1_120468327_GG>AA	NOTCH2	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120471614_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120478125_A>C	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120484306_T>C	NOTCH2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH2_chr1_120484323_A>G	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH2_chr1_120484332_A>G	NOTCH2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH2_chr1_120496301_G>C	NOTCH2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
NOTCH2_chr1_120497840_A>T	NOTCH2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH2_chr1_120506317_A>C	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120506424_G>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120508138_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120510190_A>C	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120510194_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120510202_G>A	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120512160_GAGA>G	NOTCH2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120512221_C>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH2_chr1_120539879_G>T	NOTCH2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15272377_A>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15272447_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15272455_A>G	NOTCH3	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
NOTCH3_chr19_15276193_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15280905_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15281240_G>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15281535_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15289850_T>A	NOTCH3	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
NOTCH3_chr19_15289872_C>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15290180_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15290911_C>T	NOTCH3	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
NOTCH3_chr19_15291056_C>G	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15291593_C>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15295165_G>A	NOTCH3	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH3_chr19_15295197_G>C	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15295828_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15296155_C>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
NOTCH3_chr19_15298083_C>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15298126_G>A	NOTCH3	SNV	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	432	437
NOTCH3_chr19_15299124_C>T	NOTCH3	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
NOTCH3_chr19_15299817_G>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15300219_C>T	NOTCH3	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
NOTCH3_chr19_15302575_C>A	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NOTCH3_chr19_15302649_C>T	NOTCH3	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NOTCH3_chr19_15302941_T>C	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
NOTCH3_chr19_15302995_C>T	NOTCH3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK1_chr1_156838353_G>A	NTRK1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK1_chr1_156838381_G>A	NTRK1	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
NTRK1_chr1_156843643_G>A	NTRK1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK1_chr1_156843688_G>T	NTRK1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NTRK1_chr1_156846256_T>C	NTRK1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
NTRK1_chr1_156851382_G>A	NTRK1	SNV	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	433	437
NTRK2_chr9_87285719_G>A	NTRK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK2_chr9_87366934_G>T	NTRK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK2_chr9_87482239_C>T	NTRK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK2_chr9_87482250_G>A	NTRK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK2_chr9_87563430_G>T	NTRK2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK3_chr15_88669604_C>T	NTRK3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK3_chr15_88678343_T>A	NTRK3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NTRK3_chr15_88678361_T>C	NTRK3	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23614881_C>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23614974_C>G	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23632730_C>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PALB2_chr16_23637566_GT>AC	PALB2	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23640966_C>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
PALB2_chr16_23641001_C>G	PALB2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
PALB2_chr16_23641115_G>A	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23641146_C>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PALB2_chr16_23646323_T>C	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PALB2_chr16_23646594_C>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23646617_G>T	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23646636_T>A	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23646654_G>C	PALB2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
PALB2_chr16_23646807_AT>A	PALB2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23647569_G>A	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23652460_T>A	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PALB2_chr16_23652468_G>A	PALB2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PDGFRA_chr4_55144078_G>A	PDGFRA	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
PDGFRA_chr4_55153663_G>A	PDGFRA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PDGFRB_chr5_149499620_C>T	PDGFRB	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PDGFRB_chr5_149512406_G>A	PDGFRB	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PDGFRB_chr5_149515139_G>A	PDGFRB	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PDGFRB_chr5_149515148_C>T	PDGFRB	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3CA_chr3_178916933_A>C	PIK3CA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3CA_chr3_178921472_G>A	PIK3CA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3CA_chr3_178921513_G>T	PIK3CA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3CA_chr3_178938917_A>T	PIK3CA	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67569265_GC>G	PIK3R1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67575453_A>G	PIK3R1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67588151_C>CT	PIK3R1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67588930_G>T	PIK3R1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67588974_TTTGGTA>T	PIK3R1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
PIK3R1_chr5_67589010_T>TTA	PIK3R1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67589168_C>T	PIK3R1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
PIK3R1_chr5_67589590_A>ATATAACACTCAG	PIK3R1	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67589598_CTCAGTT>C	PIK3R1	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PIK3R1_chr5_67589619_G>A	PIK3R1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PIK3R1_chr5_67589622_AATATGATAGATT>A	PIK3R1	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PIK3R1_chr5_67589640_A>G	PIK3R1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PIK3R1_chr5_67591106_A>G	PIK3R1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PIK3R1_chr5_67591128_G>GA	PIK3R1	INS	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	434	435
PIK3R1_chr5_67591251_G>A	PIK3R1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67592024_G>A	PIK3R1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PIK3R1_chr5_67592140_TA>T	PIK3R1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PIK3R1_chr5_67592151_G>A	PIK3R1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PIK3R1_chr5_67593346_T>G	PIK3R1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PMS2_chr7_6026444_T>C	PMS2	SNV	0	0	65.76	100	99.13	100	NaN	NaN	NaN	99.54	98.35	99.87	0	0	2	436	438
PMS2_chr7_6026468_T>C	PMS2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PMS2_chr7_6026531_A>C	PMS2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PMS2_chr7_6026688_T>C	PMS2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PMS2_chr7_6026707_TC>CT	PMS2	MNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
PMS2_chr7_6026709_G>A	PMS2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PMS2_chr7_6035185_G>A	PMS2	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
PMS2_chr7_6035189_G>GT	PMS2	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PMS2_chr7_6048640_G>C	PMS2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133201522_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133201570_T>C	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
POLE_chr12_133202740_C>T	POLE	SNV	100	70.09	100	100	99.11	100	100	70.09	100	100	99.11	100	9	0	0	429	438
POLE_chr12_133202768_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133209037_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133209253_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133212519_C>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133212564_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133218865_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133219152_C>T	POLE	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
POLE_chr12_133219287_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133219820_A>G	POLE	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
POLE_chr12_133219838_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133219871_T>C	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133220130_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
POLE_chr12_133220131_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
POLE_chr12_133220529_T>C	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133226340_C>T	POLE	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
POLE_chr12_133226406_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133233948_G>A	POLE	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
POLE_chr12_133236062_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133237569_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133237641_C>T	POLE	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
POLE_chr12_133237644_C>G	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133240613_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133241958_G>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133241962_C>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133241973_T>C	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133245026_G>C	POLE	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
POLE_chr12_133245046_A>G	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
POLE_chr12_133245062_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133249218_T>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133249289_TCA>T	POLE	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133249847_G>A	POLE	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
POLE_chr12_133250174_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133252349_C>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133253180_A>T	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133256793_T>C	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133257819_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
POLE_chr12_133257834_G>A	POLE	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PPARG_chr3_12393134_G>A	PPARG	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PPP2R1A_chr19_52716331_G>A	PPP2R1A	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98209387_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98211371_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
PTCH1_chr9_98211496_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98211581_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98220372_ACAG>A	PTCH1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98221903_T>C	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98229521_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98231067_TG>AA	PTCH1	MNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98231110_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98238318_G>A	PTCH1	SNV	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
PTCH1_chr9_98240378_C>T	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98240437_G>C	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98240446_T>C	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1_chr9_98242862_G>A	PTCH1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89624271_A>AT	PTEN	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89624291_A>C	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
PTEN_chr10_89624297_A>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89624301_G>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89624302_A>C	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89653785_T>A	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89653797_T>G	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89653817_G>A	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89685270_GTT>G	PTEN	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PTEN_chr10_89685289_A>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89685308_A>G	PTEN	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PTEN_chr10_89692769_GT>G	PTEN	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89692782_CT>C	PTEN	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
PTEN_chr10_89692884_A>G	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89692887_G>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89692928_T>C	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89692948_AT>A	PTEN	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	436	437
PTEN_chr10_89692979_T>A	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711882_CTA>C	PTEN	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711887_C>G	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711894_A>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711938_CTGGATTATAGACCAG>C	PTEN	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711945_A>C	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89711946_TAG>T	PTEN	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89717672_C>T	PTEN	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PTEN_chr10_89717684_A>T	PTEN	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTEN_chr10_89717715_T>TA	PTEN	INS	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
PTEN_chr10_89717741_G>T	PTEN	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
PTEN_chr10_89717769_TA>T	PTEN	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
PTEN_chr10_89720798_GTACT>G	PTEN	DEL	100	43.85	100	NaN	NaN	NaN	100	43.85	100	NaN	NaN	NaN	3	0	0	0	3
PTEN_chr10_89720811_C>CA	PTEN	INS	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
PTEN_chr10_89720811_CA>C	PTEN	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
PTEN_chr10_89720857_C>CT	PTEN	INS	NaN	NaN	NaN	99.76	98.68	99.96	0	0	79.35	100	99.1	100	0	1	0	424	425
RAD50_chr5_131915153_G>T	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131924402_C>T	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131924538_A>G	RAD50	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
RAD50_chr5_131925417_C>G	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131930584_A>G	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
RAD50_chr5_131930698_G>A	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
RAD50_chr5_131939072_G>A	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131940600_A>T	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131945038_A>T	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131945057_G>A	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131973895_C>T	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD50_chr5_131976416_A>G	RAD50	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51B_chr14_68301789_T>C	RAD51B	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RAD51B_chr14_68353784_G>T	RAD51B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51B_chr14_68758679_G>A	RAD51B	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51B_chr14_69061217_C>CTG	RAD51B	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51C_chr17_56770089_T>C	RAD51C	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RAD51C_chr17_56770138_A>G	RAD51C	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51C_chr17_56772379_C>T	RAD51C	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51C_chr17_56774155_T>C	RAD51C	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51C_chr17_56787304_G>A	RAD51C	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51C_chr17_56798128_A>G	RAD51C	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
RAD51D_chr17_33428027_A>T	RAD51D	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
RAD51D_chr17_33428357_C>T	RAD51D	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
RAD51D_chr17_33430520_G>A	RAD51D	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51D_chr17_33433488_G>A	RAD51D	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51D_chr17_33434074_T>C	RAD51D	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51D_chr17_33434458_T>TTA	RAD51D	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51D_chr17_33446605_G>A	RAD51D	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51_chr15_41023374_T>G	RAD51	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAF1_chr3_12645694_A>T	RAF1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_48881488_C>CAG	RB1	INS	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
RB1_chr13_48919246_A>T	RB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_48936995_C>T	RB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_48941639_CT>C	RB1	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RB1_chr13_48941648_C>G	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_48953731_G>A	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_48953760_C>T	RB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_48955471_C>A	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_48955492_CAA>C	RB1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_48955550_C>T	RB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_49030378_C>G	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_49030408_CAG>C	RB1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_49030444_A>G	RB1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RB1_chr13_49033829_C>T	RB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
RB1_chr13_49033940_G>T	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_49039164_G>T	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_49039187_C>A	RB1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
RB1_chr13_49039246_G>A	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1_chr13_49039470_C>G	RB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RET_chr10_43613833_C>T	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
RET_chr10_43615015_G>A	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
RET_chr10_43615142_C>G	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RET_chr10_43615166_G>T	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RET_chr10_43617457_A>T	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RET_chr10_43620417_T>C	RET	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RICTOR_chr5_38955743_G>A	RICTOR	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56434883_C>T	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56435011_C>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56435027_G>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56435160_AC>A	RNF43	DEL	NaN	NaN	NaN	0	0	35.43	0	0	35.43	NaN	NaN	NaN	0	7	0	0	7
RNF43_chr17_56435173_T>C	RNF43	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RNF43_chr17_56435462_T>G	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56435723_G>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56436128_G>A	RNF43	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
RNF43_chr17_56438227_G>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56439918_CG>C	RNF43	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
RNF43_chr17_56439996_A>C	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56440713_C>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56448297_CG>C	RNF43	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
RNF43_chr17_56492854_C>A	RNF43	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43_chr17_56492923_G>A	RNF43	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ROS1_chr6_117630009_A>T	ROS1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
ROS1_chr6_117630072_A>G	ROS1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ROS1_chr6_117631413_C>T	ROS1	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
ROS1_chr6_117639361_G>T	ROS1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47058705_T>TA	SETD2	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47058740_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47084145_AG>A	SETD2	DEL	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
SETD2_chr3_47098528_T>C	SETD2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
SETD2_chr3_47108552_A>G	SETD2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SETD2_chr3_47125716_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47125725_ATGTATTCTCACTAGAATACC>GGTACAT	SETD2	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SETD2_chr3_47125729_ATTCT>A	SETD2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47125831_AT>A	SETD2	DEL	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SETD2_chr3_47127765_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47129604_T>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47129725_G>T	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47139495_T>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47158131_C>T	SETD2	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	435	436
SETD2_chr3_47161982_C>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47162454_GT>G	SETD2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47162711_T>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47162974_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163223_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163526_A>G	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163643_T>C	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
SETD2_chr3_47163824_C>G	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163843_C>T	SETD2	SNV	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
SETD2_chr3_47163847_A>G	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163971_T>C	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47163981_C>T	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47164003_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
SETD2_chr3_47164351_G>T	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47164516_A>C	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47164829_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47164963_C>T	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
SETD2_chr3_47165188_T>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47165420_T>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47165557_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SETD2_chr3_47165674_A>G	SETD2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SETD2_chr3_47165728_G>A	SETD2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
SETD2_chr3_47165816_G>T	SETD2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
SMAD4_chr18_48591925_G>A	SMAD4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMAD4_chr18_48604749_G>T	SMAD4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11094828_A>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11094880_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097038_C>G	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097101_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097194_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097236_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097613_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11097620_G>GC	SMARCA4	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SMARCA4_chr19_11097654_A>AC	SMARCA4	INS	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
SMARCA4_chr19_11098570_C>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11100052_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11100054_G>A	SMARCA4	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
SMARCA4_chr19_11107218_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11113756_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11118586_G>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11121085_T>C	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11123632_GT>G	SMARCA4	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11123784_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11132427_C>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11132428_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
SMARCA4_chr19_11132533_A>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11132591_G>T	SMARCA4	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
SMARCA4_chr19_11136115_G>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11141473_C>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11144122_G>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11145677_A>G	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11152055_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11152091_A>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11168980_A>C	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11169491_A>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11170729_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
SMARCA4_chr19_11170753_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCA4_chr19_11170813_C>T	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
SMARCA4_chr19_11170828_G>A	SMARCA4	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCB1_chr22_24134024_G>A	SMARCB1	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
SMARCB1_chr22_24143240_C>T	SMARCB1	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
SMARCB1_chr22_24167553_T>C	SMARCB1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMARCB1_chr22_24175835_ACT>A	SMARCB1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMO_chr7_128845128_G>A	SMO	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMO_chr7_128845502_C>T	SMO	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMO_chr7_128846005_T>C	SMO	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SMO_chr7_128850376_C>T	SMO	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SPOP_chr17_47696435_C>G	SPOP	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
STK11_chr19_1207162_A>T	STK11	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
STK11_chr19_1219375_G>A	STK11	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	436	437
STK11_chr19_1220649_G>T	STK11	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
STK11_chr19_1220691_G>T	STK11	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
STK11_chr19_1221243_G>T	STK11	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
STK11_chr19_1221979_C>A	STK11	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
STK11_chr19_1226479_C>A	STK11	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TERT_chr5_1253929_G>A	TERT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TERT_chr5_1280274_C>T	TERT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TERT_chr5_1294124_G>A	TERT	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7572929_A>G	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7573991_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7574003_G>A	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
TP53_chr17_7574010_CT>C	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7574012_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7574017_C>G	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7574018_G>A	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
TP53_chr17_7576857_AG>A	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
TP53_chr17_7576905_G>A	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7576908_C>T	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7576914_TTG>T	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7577046_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577057_TC>T	TP53	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577067_T>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577090_C>G	TP53	SNV	NaN	NaN	NaN	0	0	79.35	0	0	79.35	NaN	NaN	NaN	0	1	0	0	1
TP53_chr17_7577102_C>T	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
TP53_chr17_7577108_C>T	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577118_C>G	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577130_A>C	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577131_GC>G	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7577139_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577153_C>A	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2

			PPA			NPA			PPV			NPV							
Variant ID	Gene	Variant Type	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	Point Est. (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
TP53_chr17_7577530_TG>T	TP53	DEL	NaN	NaN	NaN	0	0	65.76	0	0	65.76	NaN	NaN	NaN	0	2	0	0	2
TP53_chr17_7577541_T>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7577586_A>G	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
TP53_chr17_7577592_G>GTA	TP53	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7577594_ACAGT>A	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7577599_C>CA	TP53	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7578186_C>CT	TP53	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7578205_C>T	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578211_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578211_C>T	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578217_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578217_GT>G	TP53	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578221_TTC>T	TP53	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578224_T>A	TP53	SNV	100	34.24	100	NaN	NaN	NaN	100	34.24	100	NaN	NaN	NaN	2	0	0	0	2
TP53_chr17_7578242_CA>AC	TP53	MNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
TP53_chr17_7578262_C>G	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578263_G>A	TP53	SNV	100	56.55	100	NaN	NaN	NaN	100	56.55	100	NaN	NaN	NaN	5	0	0	0	5
TP53_chr17_7578275_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578373_TC>T	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7578389_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578392_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578416_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578427_T>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578432_T>TG	TP53	INS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7578469_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578484_GAATC>G	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7578503_C>T	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578524_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
TP53_chr17_7578532_A>G	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7578554_A>G	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7579313_G>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579314_T>G	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7579329_T>C	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579343_TGCAAGAA>T	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7579362_AACCGT>A	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7579378_G>T	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579400_G>A	TP53	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
TP53_chr17_7579428_G>A	TP53	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	436	437
TP53_chr17_7579462_A>AG	TP53	INS	NaN	NaN	NaN	99.72	98.44	99.95	0	0	79.35	100	98.94	100	0	1	0	359	360
TP53_chr17_7579504_AT>A	TP53	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TP53_chr17_7579536_C>A	TP53	SNV	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579546_CG>C	TP53	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579589_G>C	TP53	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
TP53_chr17_7579715_AG>A	TP53	DEL	100	20.65	100	NaN	NaN	NaN	100	20.65	100	NaN	NaN	NaN	1	0	0	0	1
TP53_chr17_7579717_G>A	TP53	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135772014_C>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135772093_A>C	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135772927_G>C	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135777005_C>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135780974_A>G	TSC1	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
TSC1_chr9_135781312_GGCTT>G	TSC1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135781415_C>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135782187_T>G	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135782736_T>A	TSC1	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
TSC1_chr9_135785977_G>A	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
TSC1_chr9_135787737_G>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135787825_G>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135797257_AC>A	TSC1	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135797315_T>C	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135797337_C>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135798765_G>A	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135798813_G>A	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC1_chr9_135802616_A>T	TSC1	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2098636_A>G	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2100464_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2103373_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2103392_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2103442_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2106200_G>C	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2106225_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2107112_C>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2107145_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2110746_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2111923_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2111929_G>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2112514_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2112558_G>A	TSC2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
TSC2_chr16_2112983_C>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2114370_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2115550_G>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2115629_T>C	TSC2	SNV	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
TSC2_chr16_2120487_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2120559_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438

			PPA			NPA			PPV			NPV							
Variant ID	**Gene**	**Variant Type**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**Point Est. (%)**	**95% CI Lower Bound**	**95% CI Upper Bound**	**TP**	**FP**	**FN**	**TN**	**Total Count**
TSC2_chr16_2120572_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
TSC2_chr16_2121610_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
TSC2_chr16_2122282_T>C	TSC2	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
TSC2_chr16_2122925_G>A	TSC2	SNV	0	0	79.35	100	99.13	100	NaN	NaN	NaN	99.77	98.72	99.96	0	0	1	437	438
TSC2_chr16_2124203_CG>C	TSC2	DEL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2126573_G>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2126582_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2129119_T>C	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2130210_C>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2130253_C>T	TSC2	SNV	100	20.65	100	100	99.12	100	100	20.65	100	100	99.12	100	1	0	0	433	434
TSC2_chr16_2130352_C>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2134508_G>T	TSC2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
TSC2_chr16_2134656_A>G	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2134692_A>G	TSC2	SNV	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
TSC2_chr16_2136369_A>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2136784_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
TSC2_chr16_2136843_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
TSC2_chr16_2138295_G>A	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	436	437
TSC2_chr16_2138499_C>T	TSC2	SNV	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
SUM_ALL	NaN	NaN	97.85	96.89	98.52	99.97	99.97	99.98	90.62	88.95	92.06	99.99	99.99	100	1227	127	27	457319	458700

The agreement at gene-level are listed in Table 8-28 below. Both methods agreed on most results (93/108 genes), 86.11% (93/108 genes) have a PPA of 100% and, 63.89% (69/108 genes) have a NPA of 100%. There is no gene with no mutational calls across 438 samples

Table 8-28 Performance summary of percent agreement at gene-level

	PPA			NPA			PPV			NPV							
Gene	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
AKT1	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
AKT3	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ALK	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
AR	100	20.65	100	99.77	98.72	99.96	50	9.45	90.55	100	99.13	100	1	1	0	436	438
ARAF	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
ARID1A	95.45	84.87	98.74	98.98	97.42	99.6	91.3	79.68	96.57	99.49	98.16	99.86	42	4	2	390	438
ATM	97.67	87.94	99.59	98.73	97.07	99.46	89.36	77.41	95.37	99.74	98.57	99.95	42	5	1	390	438
ATR	100	84.54	100	98.08	96.26	99.02	72.41	54.28	85.3	100	99.07	100	21	8	0	409	438
ATRX	100	84.54	100	99.76	98.65	99.96	95.45	78.2	99.19	100	99.09	100	21	1	0	416	438
AXL	100	67.56	100	100	99.11	100	100	67.56	100	100	99.11	100	8	0	0	430	438
BAP1	100	74.12	100	100	99.11	100	100	74.12	100	100	99.11	100	11	0	0	427	438
BRAF	50	9.45	90.55	100	99.13	100	100	20.65	100	99.77	98.72	99.96	1	0	1	436	438
BRCA1	100	90.82	100	99.75	98.6	99.96	97.44	86.82	99.55	100	99.05	100	38	1	0	399	438
BRCA2	100	91.97	100	99.49	98.17	99.86	95.65	85.47	98.8	100	99.03	100	44	2	0	392	438
BTK	100	20.65	100	99.77	98.72	99.96	50	9.45	90.55	100	99.13	100	1	1	0	436	438
CCND1	100	51.01	100	99.77	98.71	99.96	80	37.55	96.38	100	99.12	100	4	1	0	433	438
CCND2	100	64.57	100	100	99.12	100	100	64.57	100	100	99.12	100	7	0	0	431	438
CCND3	NaN	NaN	NaN	99.77	98.72	99.96	0	0	79.35	100	99.13	100	0	1	0	437	438
CCNE1	100	51.01	100	97.24	95.23	98.41	25	10.18	49.5	100	99.1	100	4	12	0	422	438
CDK12	100	74.12	100	100	99.11	100	100	74.12	100	100	99.11	100	11	0	0	427	438
CDK2	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CDK4	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
CDK6	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
CDKN1B	100	64.57	100	100	99.12	100	100	64.57	100	100	99.12	100	7	0	0	431	438
CDKN2A	100	64.57	100	99.77	98.7	99.96	87.5	52.91	97.76	100	99.11	100	7	1	0	430	438
CDKN2B	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438

	PPA			NPA			PPV			NPV							
Gene	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
CHEK1	100	20.65	100	99.31	98	99.77	25	4.56	69.94	100	99.12	100	1	3	0	434	438
CHEK2	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
CREBBP	100	92.13	100	99.49	98.16	99.86	95.74	85.75	98.83	100	99.03	100	45	2	0	391	438
CSF1R	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
DDR2	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
EGFR	100	60.97	100	100	99.12	100	100	60.97	100	100	99.12	100	6	0	0	432	438
ERBB2	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
ERBB3	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
ERBB4	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
ERCC2	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
ESR1	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
EZH2	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FANCA	100	92.59	100	99.74	98.56	99.95	97.96	89.31	99.64	100	99.02	100	48	1	0	389	438
FANCD2	100	75.75	100	99.77	98.68	99.96	92.31	66.69	98.63	100	99.1	100	12	1	0	425	438
FBXW7	100	75.75	100	100	99.11	100	100	75.75	100	100	99.11	100	12	0	0	426	438
FGF19	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
FGF3	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
FGFR1	66.67	20.77	93.85	100	99.12	100	100	34.24	100	99.77	98.71	99.96	2	0	1	435	438
FGFR2	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
FGFR3	100	20.65	100	99.77	98.72	99.96	50	9.45	90.55	100	99.13	100	1	1	0	436	438
FGFR4	100	56.55	100	99.54	98.33	99.87	71.43	35.89	91.78	100	99.12	100	5	2	0	431	438
FLT3	100	20.65	100	99.77	98.72	99.96	50	9.45	90.55	100	99.13	100	1	1	0	436	438
GATA2	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
HNF1A	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
HRAS	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
IGF1R	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
KDR	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
KIT	100	51.01	100	98.39	96.71	99.22	36.36	15.17	64.62	100	99.11	100	4	7	0	427	438

	PPA			NPA			PPV			NPV							
Gene	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
KRAS	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MAP2K1	66.67	20.77	93.85	100	99.12	100	100	34.24	100	99.77	98.71	99.96	2	0	1	435	438
MAP2K4	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MDM2	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
MDM4	100	20.65	100	99.77	98.72	99.96	50	9.45	90.55	100	99.13	100	1	1	0	436	438
MET	100	70.09	100	100	99.11	100	100	70.09	100	100	99.11	100	9	0	0	429	438
MLH1	100	82.41	100	100	99.09	100	100	82.41	100	100	99.09	100	18	0	0	420	438
MSH2	100	84.54	100	100	99.09	100	100	84.54	100	100	99.09	100	21	0	0	417	438
MSH6	100	88.97	100	100	99.06	100	100	88.97	100	100	99.06	100	31	0	0	407	438
MTOR	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
MYC	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
MYCL	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
NBN	100	60.97	100	99.77	98.7	99.96	85.71	48.69	97.43	100	99.12	100	6	1	0	431	438
NF1	95.12	83.86	98.65	99.5	98.18	99.86	95.12	83.86	98.65	99.5	98.18	99.86	39	2	2	395	438
NF2	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
NOTCH1	100	91.97	100	98.48	96.72	99.3	88	76.2	94.38	100	99.02	100	44	6	0	388	438
NOTCH2	100	89.28	100	99.26	97.85	99.75	91.43	77.62	97.04	100	99.06	100	32	3	0	403	438
NOTCH3	100	90.11	100	100	99.06	100	100	90.11	100	100	99.06	100	35	0	0	403	438
NTRK1	100	78.47	100	100	99.1	100	100	78.47	100	100	99.1	100	14	0	0	424	438
NTRK2	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
NTRK3	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
PALB2	100	82.41	100	100	99.09	100	100	82.41	100	100	99.09	100	18	0	0	420	438
PDGFRA	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
PDGFRB	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
PIK3CA	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
PIK3R1	100	75.75	100	99.3	97.95	99.76	80	54.81	92.95	100	99.1	100	12	3	0	423	438
PMS2	72.73	43.44	90.25	99.77	98.69	99.96	88.89	56.5	98.01	99.3	97.96	99.76	8	1	3	426	438
POLE	100	92.73	100	100	99.02	100	100	92.73	100	100	99.02	100	49	0	0	389	438

	PPA			NPA			PPV			NPV							
Gene	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	Point Estmate (%)	95% CI Lower Bound	95% CI Upper Bound	TP	FP	FN	TN	Total Count
PPARG	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PPP2R1A	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
PTCH1	100	77.19	100	99.76	98.68	99.96	92.86	68.53	98.73	100	99.1	100	13	1	0	424	438
PTEN	100	84.54	100	98.8	97.22	99.49	80.77	62.12	91.49	100	99.08	100	21	5	0	412	438
RAD50	100	77.19	100	100	99.1	100	100	77.19	100	100	99.1	100	13	0	0	425	438
RAD51	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RAD51B	100	43.85	100	99.77	98.71	99.96	75	30.06	95.44	100	99.12	100	3	1	0	434	438
RAD51C	100	60.97	100	99.77	98.7	99.96	85.71	48.69	97.43	100	99.12	100	6	1	0	431	438
RAD51D	100	67.56	100	100	99.11	100	100	67.56	100	100	99.11	100	8	0	0	430	438
RAF1	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RB1	93.75	71.67	98.89	99.76	98.67	99.96	93.75	71.67	98.89	99.76	98.67	99.96	15	1	1	421	438
RET	100	60.97	100	100	99.12	100	100	60.97	100	100	99.12	100	6	0	0	432	438
RICTOR	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
RNF43	100	70.09	100	97.9	96.06	98.89	50	29.03	70.97	100	99.09	100	9	9	0	420	438
ROS1	100	56.55	100	100	99.12	100	100	56.55	100	100	99.12	100	5	0	0	433	438
SETD2	100	88.97	100	99.26	97.86	99.75	91.18	77.04	96.95	100	99.06	100	31	3	0	404	438
SMAD4	100	34.24	100	100	99.13	100	100	34.24	100	100	99.13	100	2	0	0	436	438
SMARCA4	92.59	76.63	97.94	99.51	98.24	99.87	92.59	76.63	97.94	99.51	98.24	99.87	25	2	2	409	438
SMARCB1	75	30.06	95.44	100	99.12	100	100	43.85	100	99.77	98.71	99.96	3	0	1	434	438
SMO	100	51.01	100	100	99.12	100	100	51.01	100	100	99.12	100	4	0	0	434	438
SPOP	100	20.65	100	100	99.13	100	100	20.65	100	100	99.13	100	1	0	0	437	438
STK11	85.71	48.69	97.43	100	99.12	100	100	60.97	100	99.77	98.7	99.96	6	0	1	431	438
TERT	100	43.85	100	100	99.12	100	100	43.85	100	100	99.12	100	3	0	0	435	438
TP53	98.57	92.34	99.75	98.37	96.49	99.25	92	83.63	96.28	99.72	98.46	99.95	69	6	1	362	438
TSC1	100	82.41	100	99.76	98.66	99.96	94.74	75.36	99.06	100	99.09	100	18	1	0	419	438
TSC2	97.14	85.47	99.49	99.75	98.61	99.96	97.14	85.47	99.49	99.75	98.61	99.96	34	1	1	402	438
ALL_SUM	98.45	97.56	99.02	99.77	99.72	99.81	91.57	89.9	92.99	99.96	99.94	99.98	1141	105	18	46040	47304

The agreements per gene and broken down by SNVs, insertion and deletions are listed in Table 8-29 to

Table 8-31.

Table 8-29 Percent Agreement for SNV/MNVs by gene

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
AKT1	3	3	3	3	3	3	3/3	100.0%	43.85%, 100.0%
AKT3	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ALK	1	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
AR	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ARID1A	14	42	37	37	42	40	40/42	95.24%	84.21%, 98.68%
ATM	29	45	39	41	45	44	44/45	97.78%	88.43%, 99.61%
ATR	15	22	19	21	22	22	22/22	100.0%	85.13%, 100.0%
ATRX	11	18	17	18	18	18	18/18	100.0%	82.41%, 100.0%
AXL	7	8	7	8	8	8	8/8	100.0%	67.56%, 100.0%
BAP1	5	8	8	8	8	8	8/8	100.0%	67.56%, 100.0%
BRAF	2	2	2	2	2	1	1/2	50.0%	9.45%, 90.55%
BRCA1	7	37	27	35	37	37	37/37	100.0%	90.59%, 100.0%
BRCA2	8	55	41	44	55	54	54/55	98.18%	90.39%, 99.68%
BTK	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
CCND1	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
CCND2	4	7	6	7	7	7	7/7	100.0%	64.57%, 100.0%
CCNE1	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
CDK12	5	11	11	11	11	11	11/11	100.0%	74.12%, 100.0%
CDK2	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
CDK4	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
CDK6	1	3	1	3	3	3	3/3	100.0%	43.85%, 100.0%
CDKN1B	2	7	5	7	7	7	7/7	100.0%	64.57%, 100.0%
CDKN2A	2	7	7	7	7	7	7/7	100.0%	64.57%, 100.0%
CDKN2B	2	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
CHEK1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
CHEK2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
CREBBP	17	45	34	44	45	45	45/45	100.0%	92.13%, 100.0%
CSF1R	1	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
DDR2	1	5	2	5	5	5	5/5	100.0%	56.55%, 100.0%
EGFR	6	6	6	6	6	6	6/6	100.0%	60.97%, 100.0%
ERBB2	5	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%
ERBB3	2	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
ERBB4	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ERCC2	3	3	3	3	3	3	3/3	100.0%	43.85%, 100.0%
ESR1	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
EZH2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
FANCA	20	50	29	46	50	50	50/50	100.0%	92.87%, 100.0%
FANCD2	9	11	10	10	11	10	10/11	90.91%	62.26%, 98.38%
FBXW7	7	10	10	10	10	10	10/10	100.0%	72.25%, 100.0%
FGF19	1	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
FGF3	2	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
FGFR1	3	3	3	3	3	2	2/3	66.67%	20.77%, 93.85%
FGFR2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
FGFR3	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
FGFR4	4	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%
FLT3	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
GATA2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
HNF1A	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
HRAS	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
IGF1R	3	5	3	5	5	5	5/5	100.0%	56.55%, 100.0%
KDR	1	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
KIT	4	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
KRAS	1	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
MAP2K1	2	3	2	3	3	2	2/3	66.67%	20.77%, 93.85%
MAP2K4	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MDM2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MDM4	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MET	4	9	6	9	9	9	9/9	100.0%	70.09%, 100.0%
MLH1	12	19	15	19	19	19	19/19	100.0%	83.18%, 100.0%
MSH2	10	22	16	21	22	22	22/22	100.0%	85.13%, 100.0%
MSH6	9	31	22	30	31	31	31/31	100.0%	88.97%, 100.0%
MTOR	3	3	3	3	3	3	3/3	100.0%	43.85%, 100.0%
MYC	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MYCL	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
NBN	5	6	6	6	6	6	6/6	100.0%	60.97%, 100.0%
NF1	24	42	37	38	42	38	38/42	90.48%	77.93%, 96.23%
NF2	5	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%
NOTCH1	27	47	45	44	47	47	47/47	100.0%	92.44%, 100.0%
NOTCH2	17	32	29	31	32	32	32/32	100.0%	89.28%, 100.0%
NOTCH3	18	36	25	35	36	36	36/36	100.0%	90.36%, 100.0%
NTRK1	4	14	6	14	14	14	14/14	100.0%	78.47%, 100.0%
NTRK2	4	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%
NTRK3	2	3	3	3	3	3	3/3	100.0%	43.85%, 100.0%
PALB2	6	18	16	17	18	18	18/18	100.0%	82.41%, 100.0%
PDGFRA	2	3	2	3	3	3	3/3	100.0%	43.85%, 100.0%
PDGFRB	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
PIK3CA	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
PIK3R1	7	7	7	6	7	6	6/7	85.71%	48.69%, 97.43%
PMS2	4	11	8	11	11	8	8/11	72.73%	43.44%, 90.25%
POLE	24	55	39	48	55	55	55/55	100.0%	93.47%, 100.0%

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
PPARG	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
PPP2R1A	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
PTCH1	8	12	12	12	12	12	12/12	100.0%	75.75%, 100.0%
PTEN	6	19	19	17	19	18	18/19	94.74%	75.36%, 99.06%
RAD50	9	13	12	13	13	13	13/13	100.0%	77.19%, 100.0%
RAD51	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
RAD51B	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
RAD51C	5	6	5	6	6	6	6/6	100.0%	60.97%, 100.0%
RAD51D	6	7	6	7	7	7	7/7	100.0%	64.57%, 100.0%
RAF1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
RB1	9	14	14	13	14	13	13/14	92.86%	68.53%, 98.73%
RET	4	6	6	6	6	6	6/6	100.0%	60.97%, 100.0%
RICTOR	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
RNF43	5	9	9	9	9	9	9/9	100.0%	70.09%, 100.0%
ROS1	3	5	4	5	5	5	5/5	100.0%	56.55%, 100.0%
SETD2	8	33	29	29	33	33	33/33	100.0%	89.57%, 100.0%
SMAD4	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
SMARCA4	19	31	31	27	31	29	29/31	93.55%	79.28%, 98.21%
SMARCB1	3	3	3	3	3	2	2/3	66.67%	20.77%, 93.85%
SMO	4	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
SPOP	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
STK11	6	7	7	7	7	6	6/7	85.71%	48.69%, 97.43%
TERT	3	3	3	3	3	3	3/3	100.0%	43.85%, 100.0%
TP53	9	53	43	51	53	51	51/53	96.23%	87.25%, 98.96%
TSC1	11	16	15	16	16	16	16/16	100.0%	80.64%, 100.0%
TSC2	23	40	37	34	40	38	38/40	95.0%	83.5%, 98.62%
ALL_SUM	585	1154	975	1086	1154	1128	1128/1154	97.75%	96.72%, 98.46%

Table 8-30 Percent Agreement for Insertions by gene

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
ARID1A	4	4	4	4	4	3	3/4	75.0%	30.06%, 95.44%
ATM	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ATR	0	0	0	0	0	0	0/0	nan%	nan%, nan%
ATRX	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
BAP1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
CREBBP	2	4	2	4	4	4	4/4	100.0%	51.01%, 100.0%
FANCA	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
FBXW7	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MSH6	2	3	3	2	3	3	3/3	100.0%	43.85%, 100.0%
NOTCH1	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
PIK3R1	4	4	4	3	4	4	4/4	100.0%	51.01%, 100.0%
PMS2	0	0	0	0	0	0	0/0	nan%	nan%, nan%
PTEN	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
RAD51B	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
RAD51D	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
RB1	1	3	1	3	3	3	3/3	100.0%	43.85%, 100.0%
SETD2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
SMARCA4	0	0	0	0	0	0	0/0	nan%	nan%, nan%
TP53	3	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
ALL_SUM	28	34	30	32	34	33	33/34	97.06%	85.08%, 99.48%

Table 8-31 Percent Agreement for Deletions by gene

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
AR	0	0	0	0	0	0	0/0	nan%	nan%, nan%
ARAF	0	0	0	0	0	0	0/0	nan%	nan%, nan%
ARID1A	4	5	5	5	5	5	5/5	100.0%	56.55%, 100.0%
ATM	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ATRX	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
BAP1	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
BRCA1	1	4	3	4	4	4	4/4	100.0%	51.01%, 100.0%
BRCA2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
CCND3	0	0	0	0	0	0	0/0	nan%	nan%, nan%
CCNE1	0	0	0	0	0	0	0/0	nan%	nan%, nan%
CDKN2A	0	0	0	0	0	0	0/0	nan%	nan%, nan%
FANCA	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
FANCD2	1	2	1	2	2	2	2/2	100.0%	34.24%, 100.0%
FBXW7	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MLH1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
MYC	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
NBN	0	0	0	0	0	0	0/0	nan%	nan%, nan%
NF1	4	4	4	4	4	4	4/4	100.0%	51.01%, 100.0%
NOTCH2	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
PALB2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
PIK3R1	4	5	5	4	5	5	5/5	100.0%	56.55%, 100.0%
POLE	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
PTCH1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
PTEN	3	7	5	6	7	7	7/7	100.0%	64.57%, 100.0%
RB1	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
RNF43	0	0	0	0	0	0	0/0	nan%	nan%, nan%

Gene	Number of unique exons by Comparator	Number of mutations by Comparator	Number of unique mutations by Comparator	Number of samples by Comparator	Total mutations called by Comparator (N)	Correctly called by ACT (n)	PPA (n/N)	PPA %	95% CI
SETD2	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
SMARCA4	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
SMARCB1	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
TP53	8	15	15	15	15	15	15/15	100.0%	79.61%, 100.0%
TSC1	2	2	2	2	2	2	2/2	100.0%	34.24%, 100.0%
TSC2	1	1	1	1	1	1	1/1	100.0%	20.65%, 100.0%
ALL_SUM	49	66	62	64	66	66	66/66	100.0%	94.5%, 100.0%

ERBB2 Amplification

One hundred twenty nine (129) FFPE samples spanning 21 different cancer types were evaluated for ERRBB2 gene amplification between ACTOnco and Ventana HER2 Dual ISH DNA Probe Cocktail test (DISH). The DISH assay detected 48 ERRBB2 gene amplification within the samples of which the ACTOnco assay detected 44 resulting in a PPA of 91.67% (95% CI: 80.45% and 96.71%). For the remaining 81 samples detected negative by DISH, ACTOnco detected 81 resulting in a NPA of 100%, (95% CI: 95.47% and 100%).

There were samples that presented with DNA input levels lower than 80ng which was the recommended input DNA in study protocol. When these were removed from the data set (total data set n= 126), there were 45 were positive by Dual ISH; of these, 42 were positive by the ACTOnco assay (PPA = 93.33% [82.14%, 97.71%]). The negative sample analysis remained unchanged.

A 2 × 2 table of the mutational results with positive percent agreement (PPA) and negative percent agreement (NPA) statistics provided in Table 8-32 to Table 8-33.

Table 8-32 PPA and NPA of ERBB2 copy number alteration results (All samples)

Category	Total Cases	PPA (95%CI)	NPA (95%CI)	PPV (95%CI)	NPV (95%CI)	TP	FP	TN	FN
All Cases	129	91.67% (80.45%, 96.71%)	100.00% (95.47%, 100.00%)	100.00% (91.97%, 100.00%)	95.29% (88.52%, 98.15%)	44	0	81	4
Excluding DISH 1.8-2.2	126	93.62% (82.84%, 97.81%)	100.00% (95.36%, 100.00%)	100.00% (91.97%, 100.00%)	96.34% (89.79%, 98.75%)	44	0	79	3
Only DISH 1.8-2.2	3	0.00% (0.00%, 79.35%)	100.00% (34.24%, 100.00%)	NA	66.67% (20.77%, 93.85%)	0	0	2	1
Excluding DISH 1.5-2.5	97	95.35% (84.54%, 98.72%)	100.0% (93.36%, 100.0%)	100.0% (91.43%, 100.0%)	96.43% (87.88%, 99.02%)	41	0	54	2
Only DISH 1.5-2.5	32	60.0% (23.07%, 88.24%)	100.0% (87.54%, 100.0%)	100.0% (43.85%, 100.0%)	93.1% (78.04%, 98.09%)	3	0	27	2

Table 8-33 PPA and NPA of ERBB2 copy number alteration results (3 Low DNA Samples Removed)

Category	Total Cases	PPA (95%CI)	NPA (95%CI)	PPV (95%CI)	NPV (95%CI)	TP	FP	TN	FN
All Cases	126	93.33% (82.14%, 97.71%)	100.0% (95.47%, 100.0%)	100.0% (91.62%, 100.0%)	96.43% (90.02%, 98.78%)	42	0	81	3
Excluding DISH 1.8-2.2	123	95.45% (84.87%, 98.74%)	100.0% (95.36%, 100.0%)	100.0% (91.62%, 100.0%)	97.53% (91.44%, 99.32%)	42	0	79	2
Only DISH 1.8-2.2	3	0.0% (0.0%, 79.35%)	100.0% (34.24%, 100.0%)	NA	66.67% (20.77%, 93.85%)	0	0	2	1
Excluding DISH 1.5-2.5	96	95.24% (84.21%, 98.68%)	100.0% (93.36%, 100.0%)	100.0% (91.24%, 100.0%)	96.43% (87.88%, 99.02%)	40	0	54	2
Only DISH 1.5-2.5	30	66.67% (20.77%, 93.85%)	100.0% (87.54%, 100.0%)	100.0% (34.24%, 100.0%)	96.43% (82.29%, 99.37%)	2	0	27	1

TMB

The ACTOnco IVD assay reports a TMB score using the sequenced regions of interest to estimate the number of somatic synonymous and nonsynonymous mutations per megabase of all protein-coding genes. The ability of ACTOnco assay to accurately identify TMB in multiple solid tissue FFPE tumor types was assessed by comparing the variant calls by ACTOnco and WES. This was done on the 45 clinical samples (FFPE) processed in the lab at ACT Genomics. Only mutations that 1) occur in the same sequencing covered region between ACTOnco and WES, and 2) passed QC for both ACTOnco and WES are tallied as mutation counts were evaluated.

Across 14 cancer types (breast, colon adenocarcinoma, endo gland, endometrial, kidney, liver, lung, oral cavity, ovarian, prostate, skin, stomach, pancreas, and urinary), 45 cases were enrolled covering a dynamic range of 0.1-63.2 Muts/Mb. There were 560 mutations for ACTOnco and 554 for WES in the regions that overlapped. Of these ACTOnco and WES matched for 426 of the variants, suggesting that estimated TMB would be overlapping the same variants. The TMB scores calculated from WES and ACTOnco were plotted and a model was fit to the data (Figure 8-3).

Additionally, the Spearman rank coefficient was calculated to determine if the rank ordering of TMB values would be changed between ACTOnco and WES within the data sets evaluated. Assessment of all 45 cases resulted in a Spearman correlation coefficient of 0.885.

Figure 8-3 Regression of TMB Results from ACTOnco and WES Analysis for the FFPE samples