Blueprint Genetics

Regional differences may apply. For complete and up-to-date test methodology description, please see your report in Nucleus online portal. Accreditation and certification information available at ***blueprintgenetics.com/certifications***

Comprehensive Hematology and Hereditary Cancer Panel (Hematology) Plus

REFERRING HEALTHCARE PROFESSIONAL

NAME **HOSPITAL**

PATIENT

NAME **DOB** **AGE** **GENDER** **ORDER ID**

PRIMARY SAMPLE TYPE **SAMPLE COLLECTION DATE** **ORDER REFERENCE(S)**

SUMMARY OF RESULTS

The results presented in this report were obtained with a test in the scope of the accreditation. See Appendix 5 for details.

PRIMARY FINDINGS

The patient is heterozygous for *PIEZO1* c.76C>T, p.(Arg26Cys), which is a variant of uncertain significance (VUS).

PRIMARY FINDINGS: SEQUENCE ALTERATIONS

GENE	TRANSCRIPT	NOMENCLATURE	GENOTYPE	CONSEQUENCE	INHERITANCE	CLASSIFICATION
PIEZO1	NM_001142864.4	c.76C>T, p.(Arg26Cys)	HET	missense_variant	AD,AR	**Variant of uncertain significance (VUS)**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
		GRCh37/hg19	16:88815876	G/A		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	2/131798	probably damaging	deleterious	disease causing	Dehydrated hereditary stomatocytosis; Lympehedema, hereditary III	

SEQUENCING PERFORMANCE METRICS - NUCLEAR GENOME

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 20X	MEDIAN COVERAGE	PERCENT > 20X
Comprehensive Hematology and Hereditary Cancer Panel (Hematology)	369	5380	1056236	1055829	171	99.96

TARGET REGION AND GENE LIST

The Blueprint Genetics Comprehensive Hematology and Hereditary Cancer Panel (version 3, Jul 09, 2025) Plus Analysis includes sequence analysis and copy number variation analysis of the following genes: *ABCA3*, *ABCB7*, *ABCG5*, *ABCG8*, *ACD*, *ACTB**,

ACTN1, *ADAMTS13*, *AIP*, *AK1*, *AK2*, *ALAS2*, *ALK*, *AMN*, *ANK1*, *ANKRD26*, *AP3B1*, *AP3D1*, *APC*, *ARPC1B*, *ATM*, *ATR*, *ATRX*, *AXIN2*, *BAP1*, *BARD1*, *BLM*, *BLOC1S3*, *BLOC1S6*, *BMPR1A**, *BRAF**, *BRCA1**, *BRCA2*, *BRIP1*, *BUB1B*, *C15ORF41*, *C6ORF25*, *CBL*, *CD59*, *CD70*, *CDAN1*, *CDC42**, *CDC73*, *CDH1*, *CDK4*, *CDKN1B*, *CDKN1C*, *CDKN2A*, *CEBPA*, *CECR1*, *CEP57*, *CHEK2**,#, *CLCN7*, *CLPB*, *CSF2RA**,#, *CSF3R*, *CTC1*, *CTLA4*, *CTNNA1*, *CTSC*, *CUBN**, *CXCR4*, *CYB5R3*, *CYCS**, *CYLD*, *DDB2*, *DDX41*, *DHFR**, *DICER1**, *DIS3L2**, *DKC1*, *DNAJC21*, *DNASE2*, *DTNBP1*, *EFL1**, *EGFR*, *EGLN1**, *ELANE*, *EPAS1*, *EPB41*, *EPB42*, *EPCAM*, *EPOR*, *ERCC1*, *ERCC2*, *ERCC3*, *ERCC4*, *ERCC5*, *ERCC6L2*, *ETV6*, *EXO1*, *EXT1*, *EXT2*, *EZH2*, *F10*, *F11*, *F12*, *F13A1*, *F13B*, *F2*, *F5*, *F7*, *F8**, *F9*, *FADD*, *FAM111B**, *FANCA*, *FANCB*, *FANCC*, *FANCD2**, *FANCE*, *FANCF*, *FANCG*, *FANCI*, *FANCL*, *FANCM*, *FAS*, *FASLG*, *FGA*, *FGB*, *FGG*, *FH*, *FLCN*, *FLI1*, *FLNA*, *FYB*, *G6PC3*, *G6PD*, *GALNT12*, *GATA1*, *GATA2*, *GBA**, *GCLC*, *GFI1*, *GFI1B*, *GGCX*, *GINS1*, *GLRX5*, *GP1BA*, *GP1BB*, *GP9*, *GPC3*, *GPI*, *GPR101*, *GPR143*, *GREM1*, *GSS*, *HAVCR2*, *HAX1*, *HBA1**, *HBA2**,#, *HBB*, *HFE*, *HK1*#, *HMOX1*, *HNF1A*, *HOXA11*, *HOXB13*, *HPS1**, *HPS3**, *HPS4*, *HPS5*, *HPS6*, *HRAS*, *IFNGR2*, *IKZF1*, *ITGA2*, *ITGA2B*, *ITGB3*, *ITK*, *JAGN1*, *JAK2*, *KCNN4*, *KIF1B*, *KIF23*, *KIT*, *KITLG*, *KLF1*, *KRAS**, *LAMTOR2*, *LMAN1*, *LPIN2*, *LYST**, *LZTR1*, *MAGT1*, *MAP2K1*, *MAP2K2*, *MASTL*, *MAX*, *MCFD2*, *MECOM*, *MEN1*, *MET*, *MITF*, *MKL1*, *MLH1*, *MLH3*, *MPL*, *MRE11A*, *MSH2*, *MSH3*, *MSH6*, *MTHFD1*, *MTR*, *MUTYH*, *MYH9*, *MYO5A*, *NAF1*, *NBEAL2*, *NBN*, *NF1**, *NF2*, *NHP2*, *NOP10*, *NRAS*, *NSD1*, *NSUN2*, *NT5C3A*, *NTHL1*, *OBFC1*, *OCA2*, *P2RY12*, *PALB2*, *PARN**, *PAX5*, *PC*, *PDGFRA*#, *PDHA1*, *PDHX*, *PGK1*, *PGM3*#, *PHOX2B*, *PIEZO1*, *PKLR*, *PMS1*#, *PMS2**,#, *POLD1*, *POLE*, *POLH**, *POT1*, *PPM1D*, *PRF1*, *PRKACG*, *PRKAR1A*, *PROC*, *PROS1**, *PTCH1*, *PTEN**, *PTPN11*, *PUS1*, *RAB27A*, *RAC2*, *RAD50*, *RAD51C*, *RAD51D*, *RAF1*, *RASA2*, *RASGRP2*, *RB1*, *RBM8A**, *RECQL**, *RECQL4*, *REN*, *REST*, *RET*, *RHAG*, *RHBDF2*, *RIT1*, *RNF168*, *RPL11*, *RPL15**,#, *RPL26*, *RPL27*, *RPL31*, *RPL35A*, *RPL5*, *RPS10*, *RPS19*, *RPS20*, *RPS24*, *RPS26*, *RPS28*, *RPS29*, *RPS7*, *RRAS*, *RTEL1*, *RUNX1*, *SAMD9*, *SAMD9L*, *SBDS**, *SDHA**, *SDHAF2*, *SDHB*, *SDHC*, *SDHD*#, *SEC23B*, *SERPINC1*, *SERPINF2*, *SFTPB*, *SFTPC*, *SH2D1A*, *SHOC2*, *SLC11A2*, *SLC19A2*, *SLC25A38*, *SLC37A4*, *SLC45A2*, *SLC46A1*, *SLC4A1*, *SLFN14*, *SLX4*, *SMAD4*, *SMARCA4*, *SMARCB1*, *SMARCD2*, *SMARCE1*, *SOS1*, *SOS2*, *SPRED1*, *SPTA1*, *SPTB*, *SRC*, *SRP54*, *SRP72**, *STAT3*, *STK11*, *STX11*, *STXBP2*, *SUFU*, *TBXA2R*, *TBXAS1*, *TCN2*, *TERC*, *TERT*, *TF*, *THBD*, *THPO*, *TINF2*, *TMEM127*, *TMPRSS6*, *TP53*, *TPI1*, *TRIP13*, *TRNT1*, *TSC1*, *TSC2*, *TUBB1*, *TYR**, *TYRP1*, *UBE2T*, *UNC13D*, *USB1*, *VHL*, *VKORC1*#, *VPS13B*, *VPS45*#, *VWF**, *WAS*, *WDR1*, *WIPF1*, *WRAP53*, *WRN**, *WT1*, *XIAP**, *XPA*, *XPC*, *XRCC2*, *YARS2* and *ZCCHC8*. The following exons are not included in the panel as they are not covered with sufficient high quality sequence reads: CHEK2 (NM_001005735.2:3), CSF2RA (NM_001161530.2:11), HK1 (NM_001322365.2:5), PDGFRA (NM_001347828.2:2), PGM3 (NM_001199919.2:14), PMS1 (NM_001321051.2:5), PMS2 (NM_000535.7:13-15), RPL15 (NM_001253384.2:5), SDHD (NM_001276506.2:4), VKORC1 (NM_001311311.2:3) and VPS45 (NM_001279353.2:13). This panel targets protein coding exons, exon-intron boundaries (± 20 bps) and selected non-coding, deep intronic variants (listed in the SUMMARY OF THE TEST section). This panel should be used to detect single nucleotide variants and small insertions deletions (INDELs) and copy number variations defined as single exon or larger deletions and duplications. This panel should not be used for the detection of repeat expansion disorders or diseases caused by mitochondrial DNA (mtDNA) mutations. The test does not detect balanced translocations or complex rearrangements, and it may not detect low-level mosaicism.

*Some, or all, of the gene is duplicated in the genome. Read more: https://blueprintgenetics.com/pseudogene/

#The gene has suboptimal coverage when >90% of the gene's target nucleotides are not covered at >20x with a mapping quality score of MQ>20 reads.

The sensitivity to detect variants may be limited in genes marked with an asterisk (*) or number sign (#).

STATEMENT

CLINICAL HISTORY

Patient is a 25-year-old female with lymphedema.

CLINICAL REPORT

Sequence analysis using the Blueprint Genetics (BpG) Comprehensive Hematology and Hereditary Cancer Panel identified a heterozygous missense variant *PIEZO1* c.76C>T, p.(Arg26Cys).

***PIEZO1* c.76C>T, p.(Arg26Cys)**

There are 2 individuals heterozygous for this variant in gnomAD v2, a large reference population database (n>120,000 exomes and >15,000 genomes) which aims to exclude individuals with severe pediatric disease. The computational tool REVEL indicates this variant may be disease associated. To the best of our knowledge, this variant has not been reported in the medical literature or on disease-related variation databases.

PIEZO1

The protein encoded by *PIEZO1* gene (MIM *611184) is a mechanically-activated ion channel that links mechanical forces to biological signals. The encoded protein contains 36 transmembrane domains, functions as a homotetramer and is conserved among various species. 'Piezo' comes from the Greek word 'piesi,' meaning 'pressure' (Gene, MIM *611184). The protein is big (2521 aa), polymorphic and has a high tolerance for missense variants (PMID: 29191841).

Pathogenic heterozygous variants in *PIEZO1* cause autosomal dominant dehydrated hereditary stomatocytosis (DHS) with or without pseudohyperkalemia and/or perinatal edema (MIM #194380), and autosomal recessive variants cause hereditary lymphedema (MIM #616843). DHS, also known as hereditary xerocytosis, is an autosomal dominant hereditary hemolytic anemia characterized by erythrocyte dehydration due to loss of the cation content. DHS is a condition with defective red blood cell membrane properties that causes an imbalance in intracellular cation concentrations (PMID: 23695678). DHS red blood cells (RBCs) exhibit decreased intraerythrocytic K+ content and increased intraerythrocytic Na+ content, usually accompanied by increased mean corpuscolar hemoglobin (Hb concentration) (PMID: 23479567). Affected subjects exhibit highly variable clinical presentation, ranging from absence of clinical symptoms to lethal perinatal edema. Blood smears show variable numbers of stomatocytes and patients may also exhibit splenomegaly. The reticulocyte count is elevated, and red cell mean corpuscular volume (MCV) is slightly increased. Patients may present severe iron overload leading to hepatic transplantation, or life-threatening thromboembolic disease after splenectomy (PMID: 29191841). Several electrophysiology studies have demonstrated that the mutations cause a gain-of-function phenotype with delayed inactivation of the channel (PMID: 29191841). DHS is part of a pleiotropic syndrome that may also exhibit pseudohyperkalemia and perinatal edema (MIM #194380, 23479567). Familial pseudohyperkalemia (FP) is an asymptomatic condition that presents through blood tests as unexpectedly high plasma potassium levels, after storage at or below room temperature (Orphanet). In addition to mutations in *PIEZO1,* also pathogenic variants in *KCNN4* gene cause DHS (MIM #602754).

DHS is generally inherited in an autosomal dominant manner, however also biallelic variants in *PIEZO1* have been described in the patients with hereditary stomatocytosis. In one large family, the *PIEZO1* c.6674T>G, p.(Met2225Arg) variant was described in 22 heterozygous affected family members and in three homozygotes. Heterozygous individuals exhibited well-compensated hemolysis, elevated mean corpuscular hemoglobin concentration, and decreased osmotic fragility. Three homozygous patients had severe hemolytic anemia with reticulocytosis, shortened red blood cell survival, stomatocytosis, hyperbilirubinemia, and markedly altered erythrocyte sodium and potassium levels (PMID: 22529292).

Hereditary lymphedema III (MIM #616843) is a form of generalized lymphatic dysplasia (GLD), which is characterized by a uniform, widespread lymphedema affecting all segments of the body, with systemic involvement such as intestinal and/or pulmonary lymphangiectasia, pleural effusions, chylothoraces and/ or pericardial effusions caused by biallelic loss-of-function mutations in *PIEZO1*. There is a high incidence of nonimmune hydrops fetalis (NIHF) with either death or complete resolution of the neonatal edema, but also childhood onset of lymphedema with or without systemic involvement. Mild facial edema is often present (MIM #616843, PMID: 26333996).

Currently, HGMD Professional (2025.1) lists more than 80 disease mutations (class DM) in *PIEZO1* gene. Out of these 73% are missense, 8% nonsense, 8% splicing site, 4% small deletions and 6% small insertions.

Mutation nomenclature is based on GenBank accession NM_001142864.4 (*PIEZO1*) with nucleotide one being the first nucleotide of the translation initiation codon ATG.

CONCLUSION

PIEZO1 c.76C>T, p.(Arg26Cys) is classified as a variant of uncertain significance (VUS), as there is insufficient evidence to evaluate its clinical relevance. This variant should not be used for clinical decision-making or risk evaluation in family members. Management of the patient and family should be based on clinical evaluation and judgment. Genetic counseling is recommended.

STEP	DATE
Order date	MMM DD, YYYY
Sample received	MMM DD, YYYY
Sample in analysis	MMM DD, YYYY
Reported	MMM DD, YYYY

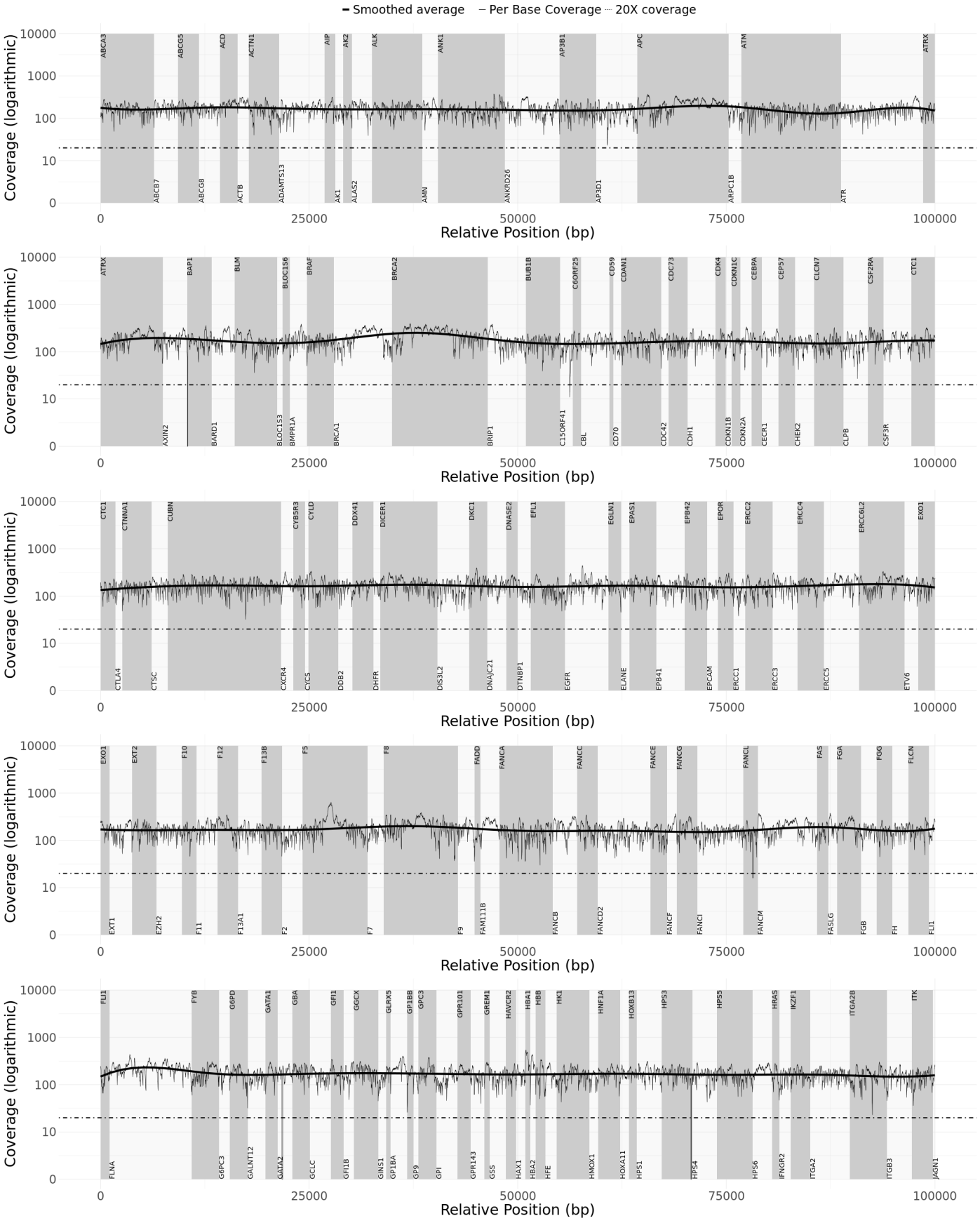
(This statement has been prepared by our geneticists and physicians, who have together evaluated the sequencing results.)

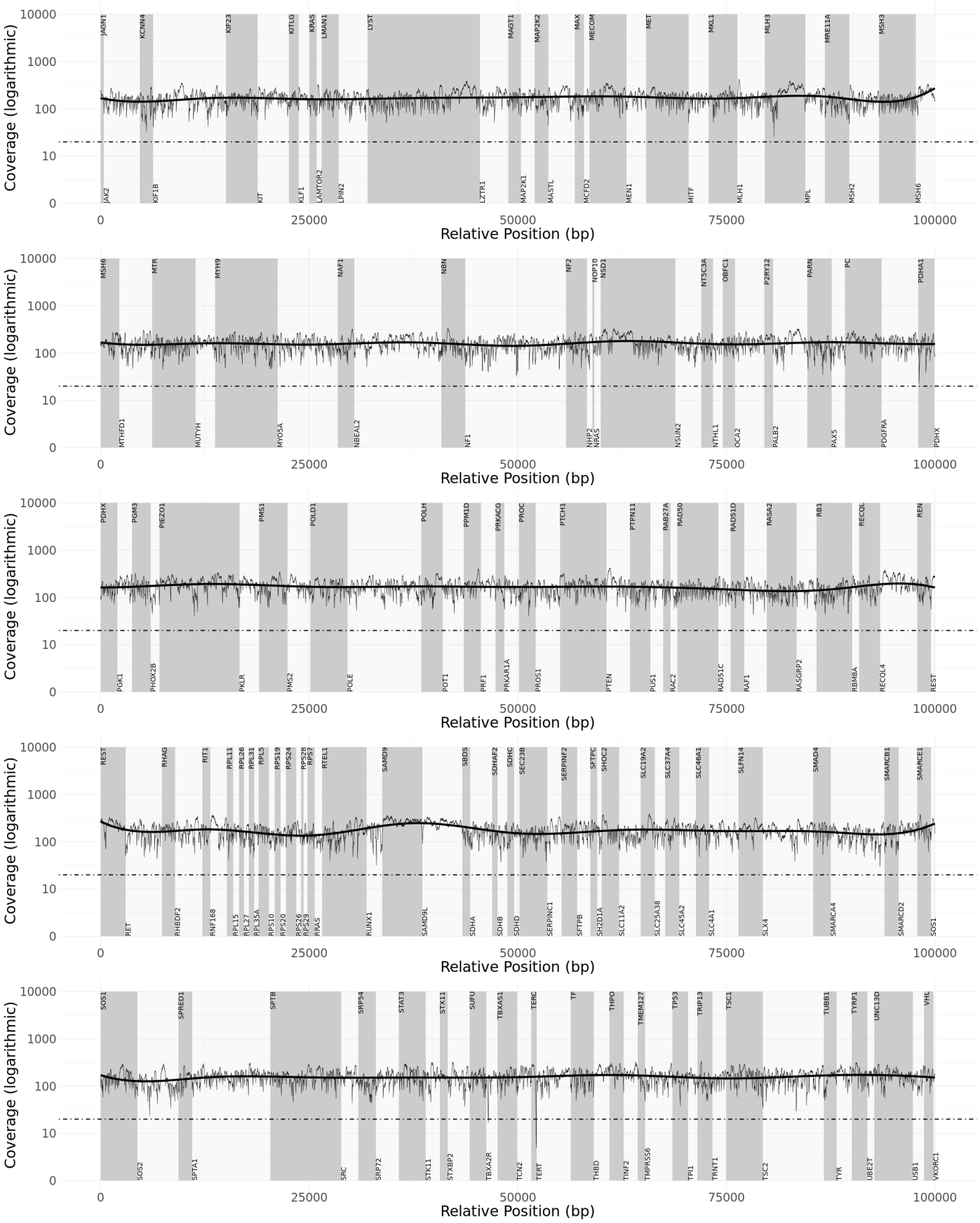
Signature

Name

COVERAGE PLOT - NUCLEAR GENES

Readability of the coverage plot may be hindered by faxing. A high quality coverage plot can be found with the full report on nucleus.blueprintgenetics.com.


Smoothed average
Per Base Coverage
20X coverage
Coverage (logarithmic)
Relative Position (bp)
ABCA3 ABCB7 ABCG5 ABCG8 ACD ACTB ACTN1 ADAMTS13 AIP AK1 AK2 ALAS2 ALK AMN ANK1 ANKRD26 AP3B1 AP3D1 APC ARPC1B ATM ATR ATRX
ATRX AXIN2 BAP1 BARD1 BLM BLOC1S3 BLOC1S6 BMPR1A BRAF BRCA1 BRCA2 BRIP1 BUB1B C15ORF41 C6ORF25 CBL CD59 CD70 CDAN1 CDC42 CDC73 CDH1 CDK4 CDKN1B CDKN1C CDKN2A CEBPA CECR1 CEP57 CHEK2 CLCN7 CLPB CSF2RA CSF3R CTC1
CTC1 CTLA4 CTNNA1 CTSC CUBN CXCR4 CYB5R3 CYCS CYLD DDB2 DDX41 DHFR DICER1 DIS3L2 DKC1 DNAJC21 DNASE2 DYNBP1 EFL1 EGFR EGLN1 ELANE EPAS1 EPB41 EPB42 EPCAM EPOR ERCC1 ERCC2 ERCC3 ERCC4 ERCC5 ERCC6L2 ETV6 EXO1
EXO1 EXT1 EXT2 EZH2 F10 F11 F12 F13A1 F13B F2 F5 F7 F8 F9 FADD FAM111B FANCA FANCB FANCC FANCD2 FANCE FANCF FANCG FANCI FANCL FANCM FAS FASLG FGA FGB FGG FH FLCN FLI1
FLI1 FLNA FYB G6PC3 G6PD GALNT12 GATA1 GATA2 GBA GCLC GFI1 GFI1B GGCX GINS1 GLRX5 GP1BA GP1BB GP9 GPC3 GPI GPR101 GPR143 GREM1 GSS HAVCR2 HAX1 HBA1 HBA2 HBB HFE HK1 HMOX1 HNF1A HOXA11 HOXB13 HPS1 HPS3 HPS4 HPS5 HPS6 HRAS IFNGR2 IKZF1 ITGA2 ITGA2B ITGB3 ITK JAGN1


Coverage (logarithmic)
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1000
100
10
0
JAGN1
KCNN4
KIF23
KITLG
KRAS
LMAN1
LYST
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MAX
MECOM
MET
MKL1
MLH3
MRE11A
MSH3
JAK2
KIF1B
KIT
KLF1
LAMTOR2
LPIN2
LZTR1
MAP2K1
MASTL
MCFD2
MEN1
MITF
MLH1
MPL
MSH2
MSH6
0
25000
50000
75000
100000
Relative Position (bp)
MSH6
MTR
MYH9
NAF1
NBN
NF2
NOP10
NSD1
NT5C3A
OBFC1
P2RY12
PARN
PC
PDHA1
MTHFD1
MUTYH
MYO5A
NBEAL2
NF1
NHP2
NRAS
NSUN2
NTHL1
OCA2
PALB2
PAX5
PDGFRA
PDHX
PDHX
PGM3
PIEZO1
PMS1
POLD1
POLH
PPM1D
PRKACG
PROC
PTCH1
PTPN11
RAB27A
RAD50
RAD51D
RASA2
RB1
RECQL
REN
PGK1
PHOX2B
PKLR
PMS2
POLE
POT1
PRF1
PRKAR1A
PROS1
PTEN
PUS1
RAC2
RAD51C
RAF1
RASGRP2
RBM8A
RECQL4
REST
REST
RET
RHAG
RHBDF2
RIT1
RNF168
RPL11
RPL15
RPL26
RPL27
RPL31
RPL35A
RPL5
RPS10
RPS19
RPS20
RPS24
RPS26
RPS28
RPS29
RPS7
RRAS
RTEL1
RUNX1
SAMD9
SAMD9L
SBDS
SDHA
SDHAF2
SDHB
SDHC
SDHD
SEC23B
SERPINC1
SERPINF2
SFTPB
SFTPC
SH2D1A
SHOC2
SLC11A2
SLC19A2
SLC25A38
SLC37A4
SLC45A2
SLC46A1
SLC4A1
SLFN14
SLX4
SMAD4
SMARCA4
SMARCB1
SMARCD2
SMARCE1
SOS1
SOS1
SOS2
SPRED1
SPTA1
SPTB
SRC
SRP54
SRP72
STAT3
STK11
STX11
STXBP2
SUFU
TBXA2R
TBXAS1
TCN2
TERC
TERT
TF
THBD
THPO
TINF2
TMEM127
TMPRSS6
TP53
TPI1
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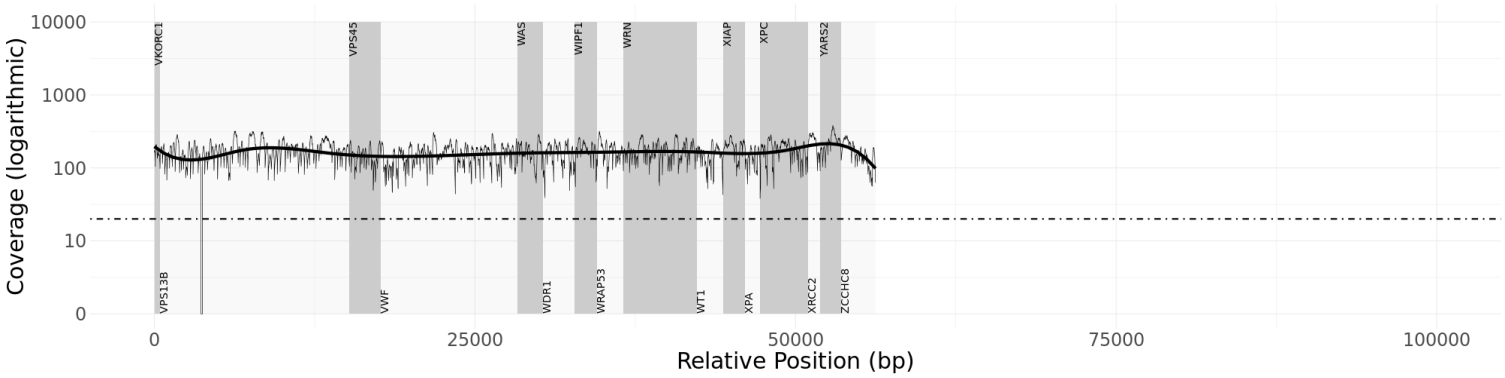
TYR
TYRP1
UBE2T
UNC13D
USB1
VHL
VKORC1


Coverage (logarithmic)
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1000
100
10
0
VKORC1
VPS45
WAS
WIPF1
WRN
XIAP
XPC
YARS2
VPS13B
VWF
WDR1
WRAP53
WT1
XPA
XRCC2
ZCCHC8
0
25000
50000
75000
100000
Relative Position (bp)

APPENDIX 5: SUMMARY OF THE TEST

PLUS ANALYSIS

Laboratory process: When required, the total genomic DNA was extracted from the specimen using a bead-based method. The quantity of DNA was measured using a fluorometric method. DNA was randomly fragmented using a noncontact, isothermal sonochemistry-based method. The sequencing library was prepared by ligating sequencing adapters to both ends of DNA fragments. Sequencing libraries were size-selected with a bead-based method to ensure optimal template size and amplified by polymerase chain reaction (PCR). Regions of interest (exons and intronic targets) were targeted using a hybridization-based target capture method. The quality of the completed sequencing library was controlled by ensuring the correct template size and quantity. Sequencing libraries that passed quality control were sequenced with Illumina's sequencing-by-synthesis method using paired-end sequencing (2x150 bases). Additional variant confirmation was performed using Sanger sequencing or digital PCR assay when needed.

Bioinformatics and quality control: Base called raw sequencing data were transformed into FASTQ format. Sequence reads of each sample were mapped to the human reference genome (GRCh37/hg19). Burrows-Wheeler Aligner (BWA-MEM) software was used for read alignment. Duplicate read marking, local realignment around indels, base quality score recalibration and variant calling were performed using GATK algorithms (Sentieon) for nDNA. Variant data was annotated using a collection of tools (VcfAnno and VEP) with a variety of public variant databases, including but not limited to gnomAD, ClinVar, and HGMD. The median sequencing depth and coverage across the target regions for the tested sample were calculated based on MQ0 aligned reads. The sequencing run included in-process reference sample(s) for quality control, which passed our thresholds for sensitivity and specificity. The patient's sample was subjected to thorough quality control measures, including assessments for contamination and sample mix-up. Copy number variations (CNVs), defined as single exon or larger deletions or duplications (Del/Dups), were detected from the sequence analysis data using a proprietary bioinformatics pipeline. The difference between observed and expected sequencing depth at the targeted genomic regions was calculated and regions were divided into segments with variable DNA copy number. The expected sequencing depth was obtained by using other samples processed in the same sequence analysis as a guiding reference. The sequence data was adjusted to account for the effects of varying guanine and cytosine content.

Interpretation and limitations of variant analysis: This test is indicated for germline testing. The clinical interpretation team assessed the pathogenicity of the identified variants by evaluating the information in the patient requisition and reviewing the relevant scientific literature, databases, and in silico predictions. The classification and interpretation of the variant(s) identified reflect the current state of Blueprint Genetics' understanding at the time of this report and are consistent with ACMG/AMP and ClinGen recommendations. Variant classification and interpretation are subject to professional judgment and may change for a variety of reasons, including, but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the health care provider's clinical evaluation. Inquiry regarding potential changes to the classification of the variant is strongly recommended prior to making any future clinical decision. For more information on the Blueprint Genetics variant classification process, please visit Blueprint Genetics | Global Genetic Tests and Genetic Diagnostics. For questions regarding variant classification updates, please contact Blueprint Genetics at: support@blueprintgenetics.com. Careful reconciliation of this molecular data with this individual's clinical presentation, family history, and other laboratory results is recommended. Genetic counseling is recommended for this individual and any family member undergoing genetic testing.

Reporting: The following variants were not reported: benign variants, likely benign variants and variants of uncertain significance (VUS) with limited evidence and/or not considered relevant for the patient’s molecular diagnosis.

Confirmation: An additional confirmation method was used when the variant quality did not meet the internal threshold for a true positive/negative call or when otherwise needed.

Analytic validation: This laboratory-developed test has been independently validated by Blueprint Genetics. The validated performance of this whole exome sequencing laboratory assay: sensitivity for SNVs 99.6%, and indels 2-50 bps 97.6%, one-exon deletion 100% and 1-10 exon duplications 82%. Specificity is >99.9% for most variant types. It does not detect very low level mosaicism as a variant with minor allele fraction of 14.6% can be detected in 90% of the cases. Detection performance for mtDNA variants (analytic and clinical validation): sensitivity for SNVs and INDELs 100.0% (5-100% heteroplasmy level), and for simulated gross deletions (500-5000 bp) >99.9%. Specificity is >99.9% for all.

Test restrictions: A normal result does not rule out the diagnosis of a genetic disorder, since some DNA abnormalities may be undetectable by the applied technology. Test results should always be interpreted in the context of clinical findings, family history, and other relevant data. Inaccurate, or incomplete information may lead to misinterpretation of the results.

Technical limitations: This test does not detect the following: complex inversions, gene conversions, balanced translocations, repeat expansion disorders unless specifically mentioned, and non-coding variants deeper than ±20 base pairs from the exon-intron boundary unless otherwise indicated (please see the list of non-coding variants covered by the test). Additionally, this test may not reliably detect the following: low-level mosaicism, stretches of mononucleotide repeats, indels larger than 50bp, small deletions or duplications, and variants within pseudogene regions/duplicated segments. The sensitivity of this test may be reduced if DNA is extracted by a laboratory other than Blueprint Genetics. Laboratory error is also possible. Please see the “Analytic validation” section, above.

Regulation and accreditations: The next-generation sequencing, Sanger and Digital PCR -based tests have been developed and validated by Blueprint Genetics (see the “Analytic validation” section). These tests have not been cleared or approved by the US Food and Drug Administration. Other examination methods performed by Blueprint Genetics are commercially available tests approved by or meeting the requirements of internationally recognized regulatory authorities and have been verified before being put into service. This analysis has been performed in a CLIA-certified laboratory (#99D2092375), accredited by the College of American Pathologists (CAP #9257331) and by the FINAS Finnish Accreditation Service, (laboratory no. T292), accreditation requirement SFS-EN ISO 15189:2022. The tests developed and validated by Blueprint Genetics are under the scope of the ISO 15189 accreditation. Detailed information about the scope is available on the FINAS website.

PERFORMING SITE:

BLUEPRINT GENETICS OY, KEILARANTA 16 A-B, 02150 ESPOO, FINLAND Laboratory Director: JUHA KOSKENVUO, MD, PHD, CLIA: 99D2092375

- DNA extraction and QC
- Next-generation sequencing
- Bioinformatic analysis
- Confirmation of sequence alterations
- Confirmation of copy number variants

NON-CODING VARIANTS COVERED BY THE PANEL:

NM_001128425.1(*MUTYH*):c.998-13T>G
NM_001128425.1(*MUTYH*):c.504+19_504+31delTAGGGGAAATAGG
NM_000969.3(*RPL5*):c.190-12_191dupCTCTTACTATAGAT
NM_005105.4(*RBM8A*):c.-21G>A
NM_005105.4(*RBM8A*):c.67+32G>C
NM_000157.3(*GBA*):c.1225-14_1225-11delTGTCinsAGT
NM_000157.3(*GBA*):c.589-12C>G
NM_000157.3(*GBA*):c.-150A>G
NM_000298.5(*PKLR*):c.1269+44C>T
NM_000298.5(*PKLR*):c.507+20C>A

NM_000298.5(*PKLR*):c.-72A>G
NM_000298.5(*PKLR*):c.-73G>C
NM_000298.5(*PKLR*):c.-83G>C
chr1:g.155271269-155271269
NM_014017.3(*LAMTOR2*):c.*23C>A
NM_003126.2(*SPTA1*):c.4339-99C>T
NM_003126.2(*SPTA1*):c.2806-13T>G
NM_000130.4(*F5*):c.5717-12T>A
NM_000130.4(*F5*):c.1296+268A>G
NM_000130.4(*F5*):c.1119-12C>G
NM_000639.1(*FASLG*):c.-261T>C
NM_000488.3(*SERPINC1*):c.1154-14G>A
NM_000488.3(*SERPINC1*):c.42-18C>G
NM_000488.3(*SERPINC1*):c.-171C>G
NM_000537.3(*REN*):c.374-12_374-11delTCinsAG
NM_000254.2(*MTR*):c.340-166A>G
NM_000254.2(*MTR*):c.609+1088G>A
NM_000254.2(*MTR*):c.3205-196A>G
NM_001081.3(*CUBN*):c.3330-439C>G
NM_014915.2(*ANKRD26*):c.-116C>G
NM_014915.2(*ANKRD26*):c.-118C>A
NM_014915.2(*ANKRD26*):c.-119C>A
NM_014915.2(*ANKRD26*):c.-121A>C
NM_014915.2(*ANKRD26*):c.-127_-126delAT
NM_014915.2(*ANKRD26*):c.-126T>C
NM_014915.2(*ANKRD26*):c.-126T>G
NM_014915.2(*ANKRD26*):c.-127A>G
NM_014915.2(*ANKRD26*):c.-127A>T
NM_014915.2(*ANKRD26*):c.-128G>T
NM_014915.2(*ANKRD26*):c.-128G>A
NM_014915.2(*ANKRD26*):c.-128G>C
NM_014915.2(*ANKRD26*):c.-134G>A
NM_020975.4(*RET*):c.-37G>C
NM_020975.4(*RET*):c.-27C>G
NM_020975.4(*RET*):c.73+9385_73+9395delAGCAACTGCCA
NM_020975.4(*RET*):c.1522+35C>T
NM_020975.4(*RET*):c.2284+13C>T
NM_020975.4(*RET*):c.2284+19C>T
NM_020975.4(*RET*):c.2392+19T>C
NM_033500.2(*HK1*):c.-390-3838G>C
NM_033500.2(*HK1*):c.-390-3818G>C
NM_033500.2(*HK1*):c.27+14901A>G
NM_000314.6(*PTEN*):c.-1239A>G
NM_000314.6(*PTEN*):c.-1178C>T
NM_000314.6(*PTEN*):c.-1171C>T
NM_000314.6(*PTEN*):c.-1111A>G
NM_000314.4(*PTEN*):c.-1001T>C
NM_000314.4(*PTEN*):c.-931G>A
NM_000314.4(*PTEN*):c.-921G>T
NM_000314.4(*PTEN*):c.-896T>C
NM_000314.4(*PTEN*):c.-862G>T

NM_000314.4(*PTEN*):c.-854C>G
NM_000314.4(*PTEN*):c.-835C>T
NM_000314.4(*PTEN*):c.-799G>C
NM_000314.4(*PTEN*):c.-765G>A
NM_000314.4(*PTEN*):c.210-8dupT
NM_000314.4(*PTEN*):c.254-21G>C
NM_000314.4(*PTEN*):c.*65T>A
NM_000314.4(*PTEN*):c.*75_*92delTAATGGCAATAGGACATTinsCTATGGCAATAGGACATTG
NM_000043.4(*FAS*):c.506-16A>G
NM_000076.2(*CDKN1C*):c.*5+20G>T
NM_000518.4(*HBB*):c.*132C>A
NM_000518.4(*HBB*):c.*132C>T
NM_000518.4(*HBB*):c.*129T>C
NM_000518.4(*HBB*):c.*115_*116delAA
NM_000518.4(*HBB*):c.*110_*114delTAAAA
NM_000518.4(*HBB*):c.*113A>G
NM_000518.4(*HBB*):c.*112A>T
NM_000518.4(*HBB*):c.*112A>G
NM_000518.4(*HBB*):c.*110_*111delTA
NM_000518.4(*HBB*):c.*111A>G
NM_000518.4(*HBB*):c.*110T>G
NM_000518.4(*HBB*):c.*108A>C
NM_000518.4(*HBB*):c.*108A>G
NM_000518.4(*HBB*):c.*93_*105delATCTGGATTCTGC
NM_000518.4(*HBB*):c.*96T>C
NM_000518.4(*HBB*):c.*74A>G
NM_000518.4(*HBB*):c.*47C>G
NM_000518.4(*HBB*):c.*32A>C
NM_000518.4(*HBB*):c.316-14T>G
NM_000518.4(*HBB*):c.316-90A>G
NM_000518.4(*HBB*):c.316-106C>G
NM_000518.4(*HBB*):c.316-146T>G
NM_000518.4(*HBB*):c.316-197C>T
NM_000518.4(*HBB*):c.316-260T>C
NM_000518.4(*HBB*):c.315+203_315+205delTCTinsCC
NM_000518.4(*HBB*):c.93-15T>G
NM_000518.4(*HBB*):c.93-21G>A
NM_000518.4(*HBB*):c.93-22delT
NM_000518.4(*HBB*):c.-12C>T
NM_000518.4(*HBB*):c.-18C>G
NM_000518.4(*HBB*):c.-21T>A
NM_000518.4(*HBB*):c.-31delC
NM_000518.4(*HBB*):c.-31C>T
NM_000518.4(*HBB*):c.-41delT
NM_000518.4(*HBB*):c.-43C>T
NM_000518.4(*HBB*):c.-50A>C
NM_000518.4(*HBB*):c.-75G>T
NM_000518.4(*HBB*):c.-75G>C
chr11:g.5248326-5248326
NM_000518.4(*HBB*):c.-76A>C
chr11:g.5248328-5248328

chr11:g.5248328-5248328
chr11:g.5248329-5248329
chr11:g.5248329-5248329
chr11:g.5248330-5248330
NM_000518.4(*HBB*):c.-79A>G
chr11:g.5248331-5248331
chr11:g.5248331-5248331
chr11:g.5248332-5248332
chr11:g.5248332-5248332
chr11:g.5248333-5248333
chr11:g.5248333-5248333
NM_000518.4(*HBB*):c.-91A>C
NM_000518.4(*HBB*):c.-92C>G
NM_000518.4(*HBB*):c.-100G>A
NM_000518.4(*HBB*):c.-121C>T
chr11:g.5248373-5248373
NM_000518.4(*HBB*):c.-123A>T
NM_000518.4(*HBB*):c.-126C>A
NM_000518.4(*HBB*):c.-127G>C
chr11:g.5248384-5248384
chr11:g.5248387-5248387
chr11:g.5248387-5248387
chr11:g.5248387-5248387
chr11:g.5248388-5248388
chr11:g.5248388-5248388
chr11:g.5248388-5248388
chr11:g.5248389-5248389
chr11:g.5248389-5248389
NM_000518.4(*HBB*):c.-142C>T
NM_000518.4(*HBB*):c.-143C>G
NM_000518.4(*HBB*):c.-151C>T
chr11:g.5248402-5248402
NM_000518.4(*HBB*):c.-152C>A
NM_000518.4(*HBB*):c.-240G>A
NM_000518.4(*HBB*):c.-273T>C
NM_003477.2(*PDHX*):c.816+11C>G
NM_000506.3(*F2*):c.241-25C>G
NM_000506.4(*F2*):c.*97G>A
NM_000506.3(*F2*):c.*106T>A
NM_000506.3(*F2*):c.*108C>T
NM_000244.3(*MEN1*):c.*412G>A
NM_000244.3(*MEN1*):c.670-15_670-14delTC
NM_000244.3(*MEN1*):c.-23-11_-22delTTGCCTTGCAGGC
NM_000244.3(*MEN1*):c.-23_-22insT
NM_000244.3(*MEN1*):c.-23-22C>A
NM_000920.3(*PC*):c.1369-29A>G
chr11:g.67250360-67250360
NM_003977.2(*AIP*):c.-220G>A
NM_001814.4(*CTSC*):c.-55C>A
NM_000372.4(*TYR*):c.1037-18T>G
NM_000051.3(*ATM*):c.-174A>G

NM_000051.3(*ATM*):c.-31+595G>A
NM_000051.3(*ATM*):c.-30-1G>T
NM_000051.3(*ATM*):c.2639-384A>G
NM_000051.3(*ATM*):c.2839-579_2839-576delAAGT
NM_000051.3(*ATM*):c.3403-12T>A
NM_000051.3(*ATM*):c.3994-159A>G
NM_000051.3(*ATM*):c.4612-12A>G
NM_000051.3(*ATM*):c.5763-1050A>G
NM_000051.3(*ATM*):c.8418+681A>G
NM_000552.3(*VWF*):c.6599-20A>T
NM_000552.3(*VWF*):c.5312-19A>G
NM_000552.3(*VWF*):c.3675-14G>A
NM_000552.3(*VWF*):c.-1+3A>C
NM_000552.3(*VWF*):c.-128G>A
NM_000552.3(*VWF*):c.-672C>T
NM_004064.3(*CDKN1B*):c.-454_-451delTTCC
NM_001029.3(*RPS26*):c.4-25_4-14delTAACAGTTTTCC
NM_002834.3(*PTPN11*):c.934-59T>A
NM_000545.5(*HNF1A*):c.-538G>C
NM_000545.5(*HNF1A*):c.-462G>A
NM_000545.5(*HNF1A*):c.-291T>C
NM_000545.5(*HNF1A*):c.-287G>A
chr12:g.121416285-121416285
NM_000545.5(*HNF1A*):c.-283A>C
NM_000545.5(*HNF1A*):c.-258A>G
NM_000545.5(*HNF1A*):c.-218T>C
chr12:g.121416385-121416385
chr12:g.121416385-121416385
chr12:g.121416391-121416391
chr12:g.121416437-121416437
chr12:g.121416446-121416446
NM_000545.5(*HNF1A*):c.-119G>A
NM_000545.5(*HNF1A*):c.-97T>G
chr12:g.121416508-121416508
NM_006231.2(*POLE*):c.1686+32C>G
NM_000059.3(*BRCA2*):c.-40+1G>A
NM_000059.3(*BRCA2*):c.-39-89delC
NM_000059.3(*BRCA2*):c.-39-1_-39delGA
NM_000059.3(*BRCA2*):c.-39-1G>A
NM_000059.3(*BRCA2*):c.426-12_426-8delGTTTT
NM_000059.3(*BRCA2*):c.8488-14A>G
NM_000059.3(*BRCA2*):c.8954-15T>G
NM_000059.3(*BRCA2*):c.9502-28A>G
NM_000059.3(*BRCA2*):c.9502-12T>G
chr13:g.48877814-48877814
chr13:g.48877836-48877836
NM_000321.2(*RB1*):c.-212G>A
NM_000321.2(*RB1*):c.-198G>A
NM_000321.2(*RB1*):c.-198G>T
NM_000321.2(*RB1*):c.-197G>A
chr13:g.48877853-48877853

chr13:g.48877856-48877856
chr13:g.48877856-48877856
NM_000321.2(*RB1*):c.-192G>A
NM_000321.2(*RB1*):c.-189G>T
NM_000321.2(*RB1*):c.-150G>C
NM_000321.2(*RB1*):c.-149G>T
NM_000321.2(*RB1*):c.501-15T>G
NM_000321.2(*RB1*):c.608-3418A>G
NM_000321.2(*RB1*):c.861+828T>G
NM_000321.2(*RB1*):c.1215+63T>G
NM_000321.2(*RB1*):c.1390-14A>G
NM_000321.2(*RB1*):c.1421+20_1421+33delTAAAAAATTTTTTT
NM_000321.2(*RB1*):c.1696-14C>T
NM_000321.2(*RB1*):c.1696-12T>G
NM_000321.2(*RB1*):c.1815-11A>G
NM_000321.2(*RB1*):c.2212-13T>A
NM_000321.2(*RB1*):c.2326-14T>C
NM_000321.2(*RB1*):c.2490-1398A>G
NM_000321.2(*RB1*):c.2490-28T>C
NM_000321.2(*RB1*):c.2490-26A>C
NM_000321.2(*RB1*):c.2490-26A>T
NM_000321.2(*RB1*):c.2490-26A>G
NM_000123.3(*ERCC5*):c.881-26T>G
NM_000131.4(*F7*):c.-96C>T
NM_000131.4(*F7*):c.-94C>G
NM_000131.4(*F7*):c.-65G>C
chr13:g.113760094-113760094
NM_000131.4(*F7*):c.-62C>T
NM_000131.4(*F7*):c.-61T>G
chr13:g.113760096-113760096
chr13:g.113760096-113760096
NM_000131.4(*F7*):c.-59T>G
NM_000131.4(*F7*):c.-57C>T
chr13:g.113760101-113760101
chr13:g.113760101-113760101
NM_000131.4(*F7*):c.-44T>C
NM_000131.4(*F7*):c.-39A>G
NM_000131.4(*F7*):c.-32A>C
NM_000131.4(*F7*):c.-30A>C
NM_000131.4(*F7*):c.131-11G>A
NM_000131.4(*F7*):c.291+1065delC
NM_000131.4(*F7*):c.571+78G>A
NM_000131.4(*F7*):c.572-12T>A
NM_177438.2(*DICER1*):c.5364+1187T>G
NM_030943.3(*AMN*):c.514-34G>A
NM_030943.3(*AMN*):c.1007-31_1006+34delCCTCGCCCCGCCGCG
NM_030943.3(*AMN*):c.1007-29_1006+36delTCGCCCCGCCGCGGG
NM_000275.2(*OCA2*):c.1117-11T>A
NM_000275.2(*OCA2*):c.1117-17T>C
NM_000275.2(*OCA2*):c.1045-15T>G
NM_000275.2(*OCA2*):c.574-19A>G

NM_001211.5(*BUB1B*):c.-44133G>A
NM_001211.5(*BUB1B*):c.2386-11A>G
NM_001113378.1(*FANCI*):c.1583+142C>T
NM_000517.4(*HBA2*):c.*47G>C
NM_000517.4(*HBA2*):c.*74_*89delCCTTCCTGGTCTTTGA
NM_000517.4(*HBA2*):c.*93_*94delAA
NM_000517.4(*HBA2*):c.*92A>G
NM_000517.4(*HBA2*):c.*94A>G
NM_000517.4(*HBA2*):c.*94A>C
NM_000517.4(*HBA2*):c.*104G>T
NM_000558.3(*HBA1*):c.*63_*65delCCT
NM_001287.5(*CLCN7*):c.916+57A>T
NM_001287.5(*CLCN7*):c.739-18G>A
NM_000548.3(*TSC2*):c.600-145C>T
NM_000548.3(*TSC2*):c.848+281C>T
NM_000548.3(*TSC2*):c.976-15G>A
NM_000548.3(*TSC2*):c.2838-122G>A
NM_000548.3(*TSC2*):c.5069-18A>G
NM_001089.2(*ABCA3*):c.3863-98C>T
NM_001089.2(*ABCA3*):c.1112-20G>A
NM_001089.2(*ABCA3*):c.-26-2A>G
NM_001134477.2(*PARN*):c.-165+2C>T
NM_024675.3(*PALB2*):c.109-12T>A
NM_015247.2(*CYLD*):c.1139-148A>G
NM_004360.3(*CDH1*):c.687+92T>A
NM_000135.3(*FANCA*):c.4261-19_4261-12delACCTGCTC
NM_000135.2(*FANCA*):c.3239+82T>G
NM_000135.2(*FANCA*):c.2982-192A>G
NM_000135.2(*FANCA*):c.2778+83C>G
NM_000135.2(*FANCA*):c.2504+134A>G
NM_000135.2(*FANCA*):c.2223-138A>G
NM_000135.2(*FANCA*):c.1567-20A>G
chr17:g.7571520-7571520
NM_000546.5(*TP53*):c.673-39G>A
NM_000546.5(*TP53*):c.97-11C>G
NM_000546.5(*TP53*):c.-29+1G>T
NM_001042492.2(*NF1*):c.-273A>C
NM_001042492.2(*NF1*):c.-272G>A
NM_001042492.2(*NF1*):c.60+9031_60+9035delAAGTT
NM_001042492.2(*NF1*):c.61-7486G>T
NM_001042492.2(*NF1*):c.288+2025T>G
NM_001042492.2(*NF1*):c.587-14T>A
NM_001042492.2(*NF1*):c.587-12T>A
NM_001042492.2(*NF1*):c.888+651T>A
NM_001042492.2(*NF1*):c.888+744A>G
NM_001042492.2(*NF1*):c.888+789A>G
NM_001042492.2(*NF1*):c.889-12T>A
NM_001042492.2(*NF1*):c.1260+1604A>G
NM_001042492.2(*NF1*):c.1261-19G>A
NM_001042492.2(*NF1*):c.1392+754T>G
NM_001042492.2(*NF1*):c.1393-592A>G

NM_001042492.2(*NF1*):c.1527+1159C>T
NM_001042492.2(*NF1*):c.1642-449A>G
NM_001128147.2(*NF1*):c.*481A>G
NM_001042492.2(*NF1*):c.2002-14C>G
NM_001042492.2(*NF1*):c.2252-11T>G
NM_001042492.2(*NF1*):c.2410-18C>G
NM_001042492.2(*NF1*):c.2410-16A>G
NM_001042492.2(*NF1*):c.2410-15A>G
NM_001042492.2(*NF1*):c.2410-12T>G
NM_001042492.2(*NF1*):c.2851-14_2851-13insA
NM_001042492.2(*NF1*):c.2991-11T>G
NM_001042492.2(*NF1*):c.3974+260T>G
NM_001042492.2(*NF1*):c.4110+945A>G
NM_001042492.2(*NF1*):c.4173+278A>G
NM_001042492.2(*NF1*):c.4578-20_4578-18delAAG
NM_001042492.2(*NF1*):c.4578-14T>G
NM_001042492.2(*NF1*):c.5269-38A>G
NM_001042492.2(*NF1*):c.5610-456G>T
NM_001042492.2(*NF1*):c.5812+332A>G
NM_001042492.2(*NF1*):c.5813-279A>G
NM_001042492.2(*NF1*):c.6428-11T>G
NM_001042492.2(*NF1*):c.6642+18A>G
NM_001042492.2(*NF1*):c.7190-12T>A
NM_001042492.2(*NF1*):c.7190-11_7190-10insGTTT
NM_001042492.2(*NF1*):c.7971-321C>G
NM_001042492.2(*NF1*):c.7971-17C>G
NM_001042492.2(*NF1*):c.8113+25A>T
NM_007294.3(*BRCA1*):c.*1340_*1342delTGT
NM_007294.3(*BRCA1*):c.*1271T>C
NM_007294.3(*BRCA1*):c.*528G>C
NM_007294.3(*BRCA1*):c.*103_*106delTGTC
NM_007294.3(*BRCA1*):c.*58C>T
NM_007294.3(*BRCA1*):c.5468-40T>A
NM_007294.3(*BRCA1*):c.5407-25T>A
NM_007294.3(*BRCA1*):c.5333-36_5333-22delTACTGCAGTGATTTT
NM_007294.3(*BRCA1*):c.5277+2916_5277+2946delAAATTCTAGTGCTTTGGATTTTTTCCTCCATinsGG
NM_007294.3(*BRCA1*):c.5194-12G>A
NM_007294.3(*BRCA1*):c.5075-27delA
NM_007294.3(*BRCA1*):c.442-22_442-13delTGTTCTTTAC
NM_007294.3(*BRCA1*):c.213-11T>G
NM_007294.3(*BRCA1*):c.213-12A>G
NM_007294.3(*BRCA1*):c.213-15A>G
NM_007294.3(*BRCA1*):c.-19-2A>G
NM_000342.3(*SLC4A1*):c.-62G>A
NM_000419.3(*ITGA2B*):c.*165T>C
NM_000419.3(*ITGA2B*):c.2095-19T>A
NM_000419.3(*ITGA2B*):c.1211-78A>G
NM_000419.3(*ITGA2B*):c.408+11C>A
NM_000419.3(*ITGA2B*):c.-4082G>A
NM_032043.2(*BRIP1*):c.1629-498A>T
NM_002734.4(*PRKAR1A*):c.-97G>A

NM_002734.4(*PRKAR1A*):c.-7G>A
NM_002734.4(*PRKAR1A*):c.-7+1G>A
NM_002734.4(*PRKAR1A*):c.550-17T>A
NM_002734.4(*PRKAR1A*):c.709-7_709-2delTTTTTA
NM_199242.2(*UNC13D*):c.2831-13G>A
NM_199242.2(*UNC13D*):c.2448-13G>A
NM_199242.2(*UNC13D*):c.118-307G>A
NM_199242.2(*UNC13D*):c.118-308C>T
NM_005570.3(*LMAN1*):c.822+34_822+35insGTTG
NM_000455.4(*STK11*):c.597+16_597+33delGGGGGGCCCTGGGGCGCCinsTG
NM_000455.4(*STK11*):c.598-32_597+31delGCCCCCTCCCGGGC
NM_006949.3(*STXBP2*):c.326-23_326-16delGCCCCACT
NM_006563.3(*KLF1*):c.-124T>C
chr19:g.12998102-12998102
NM_006563.3(*KLF1*):c.-154C>T
NM_001022.3(*RPS19*):c.-144_-141delTTTC
NM_202001.2(*ERCC1*):c.603-26G>A
NM_001011.3(*RPS7*):c.-19+1G>T
NM_001011.3(*RPS7*):c.-19+2T>C
NM_002354.2(*EPCAM*):c.556-14A>G
NM_000251.2(*MSH2*):c.-225G>C
NM_000251.2(*MSH2*):c.-181G>A
NM_000251.2(*MSH2*):c.-81dupA
NM_000251.2(*MSH2*):c.-78_-77delTG
NM_000251.2(*MSH2*):c.1662-17dupG
NM_000179.2(*MSH6*):c.457+33_457+34insGTGT
NM_000179.2(*MSH6*):c.3173-16_3173-5delCCCTCTCTTTTA
NM_000179.2(*MSH6*):c.*15A>C
NM_000179.2(*MSH6*):c.*49_*68dupTTCAGACAACATTATGATCT
NM_001114636.1(*FANCL*):c.375-2033C>G
NM_017849.3(*TMEM127*):c.-18C>T
NM_001098577.2(*RPL31*):c.-1+1G>A
NM_000312.3(*PROC*):c.-107A>G
NM_000312.3(*PROC*):c.-106A>G
NM_000312.3(*PROC*):c.-102T>A
chr2:g.128175991-128175991
NM_000312.3(*PROC*):c.-96T>G
NM_000312.3(*PROC*):c.-89T>C
NM_000312.3(*PROC*):c.-85C>T
NM_000312.3(*PROC*):c.-43A>C
NM_000312.3(*PROC*):c.-32G>A
NM_000312.3(*PROC*):c.237+15G>A
NM_000312.3(*PROC*):c.263-28T>G
NM_000312.3(*PROC*):c.401-18_401-3delGCCCTCCCCTGCCCGC
NM_000312.3(*PROC*):c.536-99C>G
NM_000312.3(*PROC*):c.*73C>T
NM_006363.4(*SEC23B*):c.-571A>G
NM_006363.4(*SEC23B*):c.-16A>G
NM_006363.4(*SEC23B*):c.221+31A>G
NM_006363.4(*SEC23B*):c.221+163A>G
NM_006363.4(*SEC23B*):c.222-78C>T

NM_006363.4(*SEC23B*):c.1743+168A>G
chr20:g.23030319-23030319
NM_000361.2(*THBD*):c.-302C>A
NM_021067.3(*GINS1*):c.-60A>G
NM_021067.3(*GINS1*):c.-48C>G
NM_000178.2(*GSS*):c.129+1663A>G
NM_000178.2(*GSS*):c.-9+5G>A
NM_017424.2(*CECR1*):c.1082-1113delA
NM_006767.3(*LZTR1*):c.-38T>A
NM_006767.3(*LZTR1*):c.2220-17C>A
NM_003073.3(*SMARCB1*):c.93+559A>G
NM_003073.3(*SMARCB1*):c.1119-12C>G
NM_003073.3(*SMARCB1*):c.*70C>T
NM_003073.3(*SMARCB1*):c.*82C>T
NM_000268.3(*NF2*):c.516+232G>A
NM_000355.3(*TCN2*):c.581-176A>T
NM_000355.3(*TCN2*):c.581-176A>G
NM_182916.2(*TRNT1*):c.609-26T>C
NM_033084.3(*FANCD2*):c.696-121C>G
NM_033084.3(*FANCD2*):c.1766+40T>G
NM_033084.3(*FANCD2*):c.1948-16T>G
NM_000551.3(*VHL*):c.-75_-55delCGCACGCAGCTCCGCCCCGCG
NM_000551.3(*VHL*):c.-54_-44dupTCCGACCCGCG
NM_000551.3(*VHL*):c.*70C>A
NM_000551.3(*VHL*):c.*70C>T
NM_004628.4(*XPC*):c.*156G>A
NM_004628.4(*XPC*):c.413-24A>G
NM_000249.3(*MLH1*):c.-413_-411delGAG
NM_000249.3(*MLH1*):c.-107C>G
NM_000249.3(*MLH1*):c.-63_-58delGTGATTinsCACGAGGCACGAGCACGA
NM_000249.3(*MLH1*):c.-42C>T
NM_000249.3(*MLH1*):c.-27C>A
NM_000249.3(*MLH1*):c.116+106G>A
NM_000249.3(*MLH1*):c.117-11T>A
NM_000249.3(*MLH1*):c.454-13A>G
NM_000249.3(*MLH1*):c.885-9_887dupTCCTGACAGTTT
NM_000249.3(*MLH1*):c.1558+13T>A
NM_004656.3(*BAP1*):c.*644delG
NM_000313.3(*PROS1*):c.1871-20_1871-13delCTAATATT
NM_000313.3(*PROS1*):c.1871-14T>G
NM_000313.3(*PROS1*):c.1493-17T>C
NM_000313.3(*PROS1*):c.1323+33A>G
NM_000313.3(*PROS1*):c.966-17C>G
NM_000313.3(*PROS1*):c.-168C>T
NM_000313.3(*PROS1*):c.-190C>G
NM_032638.4(*GATA2*):c.1017+572C>T
NM_032638.4(*GATA2*):c.1017+513_1017+540delGGAGTTTCCTATCCGGACATCTGCAGCC
NM_032638.4(*GATA2*):c.1017+532T>A
NM_032383.3(*HPS3*):c.2888-1612G>A
NR_001566.1(*TERC*):n.-22C>T
chr3:g.169482906-169482906

NR_001566.1(*TERC*):n.-100C>G
chr3:g.169483086-169483086
NM_006206.4(*PDGFRA*):c.*34G>A
NM_005612.4(*REST*):c.983-2247C>G
NM_005141.4(*FGB*):c.115-600A>G
NM_005141.4(*FGB*):c.958+13C>T
NM_021870.2(*FGG*):c.1129+632A>G
NM_021870.2(*FGG*):c.1129+66_1129+69delAATA
NM_021870.2(*FGG*):c.667-320A>T
NM_000128.3(*F11*):c.-456G>A
NM_000128.3(*F11*):c.595+11A>G
NM_000128.3(*F11*):c.1304+12G>A
NM_198253.2(*TERT*):c.2383-15C>T
NM_198253.2(*TERT*):c.-57A>C
NM_017755.5(*NSUN2*):c.538-11T>G
chr5:g.33985176-33985176
chr5:g.33985764-33985764
NM_001127511.2(*APC*):c.-192A>T
NM_001127511.2(*APC*):c.-191T>C
NM_001127511.2(*APC*):c.-190G>A
NM_001127511.2(*APC*):c.-125delA
NM_000038.5(*APC*):c.423-12A>G
NM_000038.5(*APC*):c.423-11A>G
NM_000038.5(*APC*):c.532-941G>A
NM_000038.5(*APC*):c.835-17A>G
NM_000038.5(*APC*):c.1408+731C>T
NM_000038.5(*APC*):c.1408+735A>T
chr5:g.176836590-176836590
NM_000129.3(*F13A1*):c.-19_-19+19delGGTAAGCCACCGACCCTGCA
NM_000410.3(*HFE*):c.-20G>A
NM_006502.2(*POLH*):c.-5+1G>C
NM_003764.3(*STX11*):c.*85_*86insT
NM_000535.5(*PMS2*):c.1145-31_1145-13delCTGACCCTCTTCTCCGTCC
NM_000535.5(*PMS2*):c.23+21_23+28delTCCGGTGT
NM_000553.4(*WRN*):c.2089-3024A>G
NM_000553.4(*WRN*):c.3234-160A>G
NM_001142446.1(*ANK1*):c.1900-17G>A
NM_001142446.1(*ANK1*):c.1900-18C>A
NM_020476.2(*ANK1*):c.-73_-72delTG
NM_000077.4(*CDKN2A*):c.458-105A>G
NM_000077.4(*CDKN2A*):c.150+1104C>A
NM_058197.4(*CDKN2A*):c.*73+2T>G
NM_000077.4(*CDKN2A*):c.-21C>T
NM_000077.4(*CDKN2A*):c.-49C>A
NM_000077.4(*CDKN2A*):c.-56G>T
NM_000077.4(*CDKN2A*):c.-93_-91delAGG
NM_000136.2(*FANCC*):c.-78-2A>G
NM_000136.2(*FANCC*):c.-79+1G>A
NM_000264.3(*PTCH1*):c.2561-2057A>G
NM_000380.3(*XPA*):c.390-12A>G
NM_000368.4(*TSC1*):c.363+668G>A

NM_000273.2(*GPR143*):c.885+748G>A
NM_000273.2(*GPR143*):c.659-131T>G
NM_001173454.1(*PDHA1*):c.533-17_533-14delTGTT
NM_001173454.1(*PDHA1*):c.625-30G>A
NM_001173454.1(*PDHA1*):c.873+26G>A
NM_001173454.1(*PDHA1*):c.*79_*90dupAGTCAATGAAAT
NM_001173454.1(*PDHA1*):c.*79_*90dupAGTCAATGAAAT
NM_000377.2(*WAS*):c.1339-19_1339-11delTGATCCCTGinsATCTGCAGACC
NM_002049.3(*GATA1*):c.-19-2A>G
NM_000032.4(*ALAS2*):c.-15-2186C>G
NM_000032.4(*ALAS2*):c.-15-2187T>C
NM_000032.4(*ALAS2*):c.-15-2188A>G
NM_000032.4(*ALAS2*):c.-34C>T
NM_000032.4(*ALAS2*):c.-258C>G
NM_000291.3(*PGK1*):c.1214-25T>G
NM_002351.4(*SH2D1A*):c.138-17_138-11delAGTTTAT
chrX:g.138612869-138612869
chrX:g.138612869-138612869
chrX:g.138612869-138612869
chrX:g.138612871-138612871
NM_000133.3(*F9*):c.-53A>G
chrX:g.138612872-138612872
chrX:g.138612872-138612872
chrX:g.138612874-138612874
chrX:g.138612874-138612874
chrX:g.138612875-138612875
chrX:g.138612875-138612875
NM_000133.3(*F9*):c.-48G>C
NM_000133.3(*F9*):c.-38A>G
chrX:g.138612889-138612889
chrX:g.138612889-138612889
chrX:g.138612890-138612890
chrX:g.138612890-138612890
NM_000133.3(*F9*):c.-22delT
NM_000133.3(*F9*):c.-24T>A
NM_000133.3(*F9*):c.-23T>C
NM_000133.3(*F9*):c.-22T>C
NM_000133.3(*F9*):c.-21C>G
NM_000133.3(*F9*):c.-19C>G
NM_000133.3(*F9*):c.-17delA
NM_000133.3(*F9*):c.-18A>T
NM_000133.3(*F9*):c.-18A>G
NM_000133.3(*F9*):c.-17A>C
NM_000133.3(*F9*):c.-17A>G
NM_000133.3(*F9*):c.253-25A>T
NM_000133.3(*F9*):c.253-25A>G
NM_000133.3(*F9*):c.253-19_253-16delCTTC
NM_000133.3(*F9*):c.253-16_253-12delCTTTT
NM_000133.3(*F9*):c.278-13A>G
NM_000133.3(*F9*):c.278-12C>G
NM_000133.3(*F9*):c.278-12C>T

NM_000133.3(*F9*):c.392-22_392-21delCT
NM_000133.3(*F9*):c.520+13A>G
NM_000133.3(*F9*):c.723+18T>A
NM_000133.3(*F9*):c.*1157A>G
NM_000133.3(*F9*):c.*1368A>G
NM_001110556.1(*FLNA*):c.6023-27_6023-16delTGACTGACAGCC
NM_001363.3(*DKC1*):c.-142C>G
NM_001363.3(*DKC1*):c.-141C>G
NM_001363.3(*DKC1*):c.85-15T>C
NM_000132.3(*F8*):c.6430-14A>G
NM_000132.3(*F8*):c.5999-11G>A
NM_000132.3(*F8*):c.5999-23_5999-22delCT
NM_000132.3(*F8*):c.5999-33_5999-22delGAAATAATTTCTinsATTC
NM_000132.3(*F8*):c.5999-33_5999-28delGAAATA
NM_000132.3(*F8*):c.5999-277G>A
NM_000132.3(*F8*):c.5999-798G>A
NM_000132.3(*F8*):c.5816-14_5816-13delGTinsTA
NM_000132.3(*F8*):c.5816-34A>T
NM_000132.3(*F8*):c.5587-93C>T
NM_000132.3(*F8*):c.5220-16_5220-15insA
NM_000132.3(*F8*):c.2113+12T>A
NM_000132.3(*F8*):c.1904-37G>A
NM_000132.3(*F8*):c.1538-18G>A
NM_000132.3(*F8*):c.1537+325A>G
NM_000132.3(*F8*):c.1444-15C>A
NM_000132.3(*F8*):c.788-14T>G
NM_000132.3(*F8*):c.671-11T>C
NM_000132.3(*F8*):c.602-11T>G
NM_000132.3(*F8*):c.602-32A>G
NM_000132.3(*F8*):c.601+1632G>A
NM_000132.3(*F8*):c.389-16T>G
NM_000132.3(*F8*):c.144-11T>G
NM_000132.3(*F8*):c.144-26A>T
NM_000132.3(*F8*):c.144-10758_144-10757insTATA
NM_000132.3(*F8*):c.143+1567A>G
NM_000132.3(*F8*):c.-112G>A
chrX:g.154251045-154251045
NM_000132.3(*F8*):c.-218T>C
NM_000132.3(*F8*):c.-219C>T
NM_000132.3(*F8*):c.-221T>A
NM_000132.3(*F8*):c.-255A>G
chrX:g.154251084-154251084
chrX:g.154251084-154251084
NM_000132.3(*F8*):c.-860A>G
NM_000132.3(*F8*):c.-966G>T

GLOSSARY OF USED ABBREVIATIONS:

AD = autosomal dominant
AF = allele fraction (proportion of reads with mutated DNA / all reads)
AR = autosomal recessive

CNV = Copy Number Variation e.g. one exon or multiexon deletion or duplication
gnomAD = genome Aggregation Database (reference population database; >138,600 individuals)
gnomAD AC/AN = allele count/allele number in the genome Aggregation Database (gnomAD)
HEM = hemizygous
HET = heterozygous
HOM = homozygous
ID = rsID in dbSNP
MT = Mitochondria
MutationTaster = *in silico* prediction tools used to evaluate the significance of identified amino acid changes.
Nomenclature = HGVS nomenclature for a variant in the nucleotide and the predicted effect of a variant in the protein level
OMIM = Online Mendelian Inheritance in Man®
PolyPhen = *in silico* prediction tool used to evaluate the significance of amino acid changes.
POS = genomic position of the variant in the format of chromosome:position
SIFT = *in silico* prediction tool used to evaluate the significance of amino acid changes.