Blueprint Genetics

*Sample report as of June 14th, 2022. 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 Proactive Screen

POSITIVE

REFERRING HEALTHCARE PROFESSIONAL

NAME **HOSPITAL**

INDIVIDUAL

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

PRIMARY SAMPLE TYPE **SAMPLE COLLECTION DATE** **CUSTOMER SAMPLE ID**

SUMMARY OF RESULTS

PERSONAL RISKS

The individual is heterozygous for *PALB2* c.1592del, p.(Leu531Cysfs*30), which is pathogenic.

CARRIERSHIP(S) OF AUTOSOMAL RECESSIVE DISEASE(S)

Negative for pathogenic or likely pathogenic variants.

SEQUENCING PERFORMANCE METRICS

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 20X	MEDIAN COVERAGE	PERCENT > 20X
Comprehensive Proactive Screen	180	3555	749284	748067	293	99.84

TARGET REGION AND GENE LIST

The Blueprint Genetics Comprehensive Proactive Screen (version 1, Jun 10, 2023) Plus Analysis includes sequence analysis and copy number variation analysis of the following genes: *ACTA2*, *ACTC1*, *ACTN2*, *ACVRL1*, *AIP*, *ANKRD26*, *APC*, *APOB*, *ATM*, *ATP7B*, *AXIN2*, *BAG3*, *BAP1*, *BARD1*, *BMPR1A**, *BMPR2*, *BRCA1**, *BRCA2*, *BRIP1*, *BTD*, *CACNA1C**, *CACNA1S*, *CALM1**, *CALM2*, *CALM3*, *CASQ2*, *CASZ1*, *CAV1*, *CDC73*, *CDH1*, *CDK4*, *CDKN1B*, *CDKN2A*, *CEBPA*, *CHEK2**, *CHRM2*, *COL3A1*, *COL5A1*, *COL5A2*, *CSRP3*, *CTNNA1*, *CYLD*, *DDX41*, *DES*, *DICER1**, *DMD*, *DSC2*, *DSG2*, *DSP*, *EGFR*, *EMD*, *ENG*, *EPCAM*, *ERCC6L2*, *ETV6*, *EXT1*, *EXT2*, *F5*, *FBN1*, *FH*, *FHL1**, *FLCN*, *FLNC**, *GAA*, *GATA2*, *GATA4**, *GDF2*, *GLA*, *GREM1*, *HCN4*, *HFE*, *HNF1A*, *HOXB13*, *JUP*, *KCNE1*, *KCNE2*, *KCNH2*, *KCNJ2*, *KCNQ1*, *KIT*, *LAMP2*, *LDLR*, *LDLRAP1*, *LMNA*, *LMOD2*, *LZTR1*, *MAX*, *MEN1*, *MET*, *MLH1*, *MSH2*, *MSH6*, *MT-RNR1*, *MUTYH*, *MYBPC3*, *MYH11*, *MYH7*, *MYL2*, *MYL3*, *MYLK**, *NBN*, *NEXN*, *NF1**, *NF2*, *NKX2-5*, *NTHL1*, *OTC*, *PALB2*, *PCSK9*, *PDGFRA*#, *PHOX2B*, *PKP2**,#, *PLN*, *PMS2**, *POLD1*, *POLE*, *POT1*, *PRKAG2*, *PRKAR1A*, *PRKG1*, *PROC*, *PROS1**, *PTCH1*, *PTEN**, *RAD50*, *RAD51C*, *RAD51D*, *RAF1*, *RB1*, *RBM20*, *RECQL**, *RET*, *RHBDF2*, *RPE65*, *RUNX1*, *RYR1*, *RYR2*, *SCN5A*, *SCNN1B*, *SCNN1G*, *SDHA**, *SDHAF2*, *SDHB*, *SDHC*, *SDHD*#, *SERPINA1*, *SERPINC1*, *SGCD*, *SMAD3*, *SMAD4*, *SMAD9*, *SMARCA4*, *SMARCB1*, *STK11*, *SUFU*, *TAB2*, *TBX20**, *TCAP*, *TECRL*, *TERC*, *TERT*, *TGFB2*, *TGFB3*, *TGFBR1*, *TGFBR2*, *TINF2*, *TMEM127*, *TMEM43*, *TNNC1*, *TNNI3*, *TNNT2*, *TP53*, *TPM1*, *TRDN*, *TSC1*, *TSC2*, *TTN**, *TTR*, *VCL*, *VHL* and *WT1*. The following exons are not included in the panel as they are not covered with sufficient high quality sequence reads: PDGFRA (NM_001347828:2), PKP2 (NM_001254727:6) and SDHD (NM_001276506:4).

*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

TEST INDICATION

This individual is a 30-year-old. Genetic testing with the Comprehensive Proactive Screen Panel has been requested.

PERSONAL RISKS: SEQUENCE ALTERATIONS

GENE	TRANSCRIPT	NOMENCLATURE	GENOTYPE	CONSEQUENCE	INHERITANCE	CLASSIFICATION
PALB2	NM_024675.4	c.1592del, p.(Leu531Cysfs*30)	HET	frameshift_variant	AD,AR	**Pathogenic**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
	rs180177102	GRCh37/hg19	16:23646274	CA/C		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	49/282888	N/A	N/A	N/A	Breast cancer, Fanconi anemia, Pancreatic cancer	

CLINICAL REPORT

Sequence analysis using the Blueprint Genetics (BpG) Comprehensive Proactive Screen identified a heterozygous frameshift variant *PALB2* c.1592del, p.(Leu531Cysfs*30).

PALB2 c.1592del, p.(Leu531Cysfs*30)

There are 49 individuals heterozygous for this variant in gnomAD, a large reference population database (n>120,000 exomes and >15,000 genomes) which aims to exclude individuals with severe pediatric disease. This variant generates a frameshift in exon 4 (of a total of 13 exons) resulting in a premature stop codon. This is predicted to lead to a loss of normal protein function, either through protein truncation or nonsense-mediated mRNA decay. Loss of function is an established disease-mechanism in this gene (HGMD). Analysis of lymphoblastoid cell lines from heterozygous female variant carriers showed a truncated protein that is highly unstable and functionally abnormal (PMID: 24153426). The *PALB2* c.1592del, p.(Leu531Cysfs*30) variant has been reported in individuals and families affected with breast cancer (PMID: 17287723, 18628482, 19383810, 27595995) and detected several times by other laboratories in the context of clinical testing and submitted to ClinVar (variation ID 126609). It was found in 0.6 - 1% of Finnish unselected breast cancer cases and in 2% of Finnish familial breast cancer cases; the variant is considered as a founder mutation in Finland (PMID: 17287723, 19383810) and has been associated with an increased risk of breast cancer, with an estimated odds ratio (OR) 3.94 (95% CI 1.5-12.1) (PMID: 18628482, 27595995). Estimated breast cancer risk to age 70 years is 40% (95% CI, 17-77). Non-Finnish patients have also been reported.

PALB2

PALB2 (Partner and Localizer of BRCA2) participates in DNA damage response. It colocalizes with BRCA2 in nuclear foci, promotes its localization and stability in nuclear structures, and enables its recombinational repair and checkpoint functions (MIM *610355). PALB2 is crucial for key BRCA2 genome caretaker functions and has also been shown to interact with BRCA1 (PMID: 19268590).

Heterozygous pathogenic *PALB2* variants are associated with an increased risk for breast cancer. A meta-analysis has estimated an RR of 5.3 (PMID: 26014596). In women with *PALB2* pathogenic variant, breast cancer risk has been shown to increase with age and also with increasing number of relatives with breast cancer. Breast cancer risk by 70 years of age for those with no first-degree relatives with breast cancer was 33%, compared to 58% in those with two first degree relatives (PMID: 25099575). It was calculated that *PALB2* loss-of-function variants could account for approximately 2.4% of familial aggregation of breast cancer (PMID: 25099575). *PALB2* loss-of-function variants have in some studies associated with increased ovarian cancer, but all studies have not shown a significant increase (PMID: 24448499, 26720728).

PALB2 pathogenic variants have also been identified in families with multiple cases of pancreatic cancer, but the exact risk for pancreatic cancer is still unclear (PMID: 19264984, 20412113). Yang *et al*. (2020) analyzed data from 524 families with *PALB2* pathogenic variants (PVs) from 21 countries and found associations between *PALB2* PVs and risk of female breast cancer, ovarian cancer, pancreatic cancer, and male breast cancer (PMID: 31841383). There was no evidence for increased risks of prostate or colorectal cancer. The breast cancer RRs declined with age (*P* for trend = 2.0 × 10-3). After adjusting for family ascertainment, breast cancer risk estimates on the basis of multiple case families were similar to the estimates from families ascertained through population-based studies (*P* for difference = 0.41). On the basis of the combined data, the estimated risks to age 80 years were 53% (95% CI, 44% to 63%) for female breast cancer, 5% (95% CI, 2% to 10%) for ovarian cancer, 2%-3% (95% CI females, 1% to 4%; 95% CI males, 2% to 5%) for pancreatic cancer, and 1% (95% CI, 0.2% to 5%) for male breast cancer.

Tischkowitz *et al*. (2021) sought to outline management of *PALB2* heterozygotes based on current evidence (PMID: 33976419). They concluded that there is strong evidence that pathogenic or likely pathogenic (P/LP) *PALB2* variants confer moderate to high breast cancer risk and consequently, enhanced surveillance and the option of risk-reducing interventions are warranted. They also found that there is reasonable evidence that *PALB2* P/LP variants confer a small to moderately increased risk for ovarian cancer that may warrant risk-reducing interventions, albeit their clinical benefit is not sufficiently proven yet with respect to the efficacy of preventive measures to reduce morbidity and mortality. Likewise, there is reasonable evidence that such variants confer a small to moderately increased risk of pancreatic cancer, but the role of surveillance remains controversial.

Biallelic *PALB2* variants cause a subtype of Fanconi anemia, FA-N, and confer a high risk of childhood cancer (MIM # 610832). Reid *et al*. identified four individuals with biallelic truncating variants in *PALB2* and affected with Fanconi anemia and childhood embryonal tumors (PMID: 17200671). Additional three individuals with Fanconi anemia did not have samples available for analysis, however, their parents were shown to carry truncating variants in *PALB2*. The phenotype of these patients with Fanconi anemia of complementation group N (FANCN) closely resembled Fanconi anemia and these patients presented with growth retardation and variable congenital malformations. One of the patients manifested with microcephaly, growth retardation, hypoplastic thumb, and at the age of 2 years old he developed medulloblastoma. Another patient presented with microcephaly, growth retardation, ventricular and atrial septal defect, thumb and radial anomalies, skin hyperpigmentation, and also developed neuroblastoma in infancy and acute myeloblastic leukemia at the age of 2 years. All of the seven individuals with FANCN reported developed cancers in early childhood, including 5 medulloblastomas, 3 Wilms tumors, 2 cases of acute myeloblastic leukemia, and 1 case of neuroblastoma.

There are currently more than 500 variants in *PALB2* annotated as disease-causing (DM) in the HGMD Professional variant database (version 2023.2). Most of them are associated with different cancer types, mainly breast cancer, and 18 variants are associated with Fanconi anemia, N-type. Majority of the listed variants are nonsense and frameshift variants, small deletions and duplications, splicing variants, and gross deletions or insertions. To date, all reported variants associating with Fanconi anemia in *PALB2* are truncating variants (nonsense variants, frameshift variants, and variants affecting splicing) and gross deletions.

Mutation nomenclature is based on GenBank accession NM_024675.4 (*PALB2*) with nucleotide one being the first nucleotide of the translation initiation codon ATG.

CONCLUSION

PALB2 c.1592del, p.(Leu531Cysfs*30) is classified as pathogenic, based on currently available evidence supporting its disease-causing role. Disease caused by this variant is inherited in an autosomal dominant manner. Therefore, this individual is at risk of developing *PALB2*-related disease. Any offspring of this individual are at 50% risk of inheriting the variant. Genetic counseling and family member testing are recommended.

STEP	DATE
Order date	
Sample received	
Sample in analysis	
Reported	

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

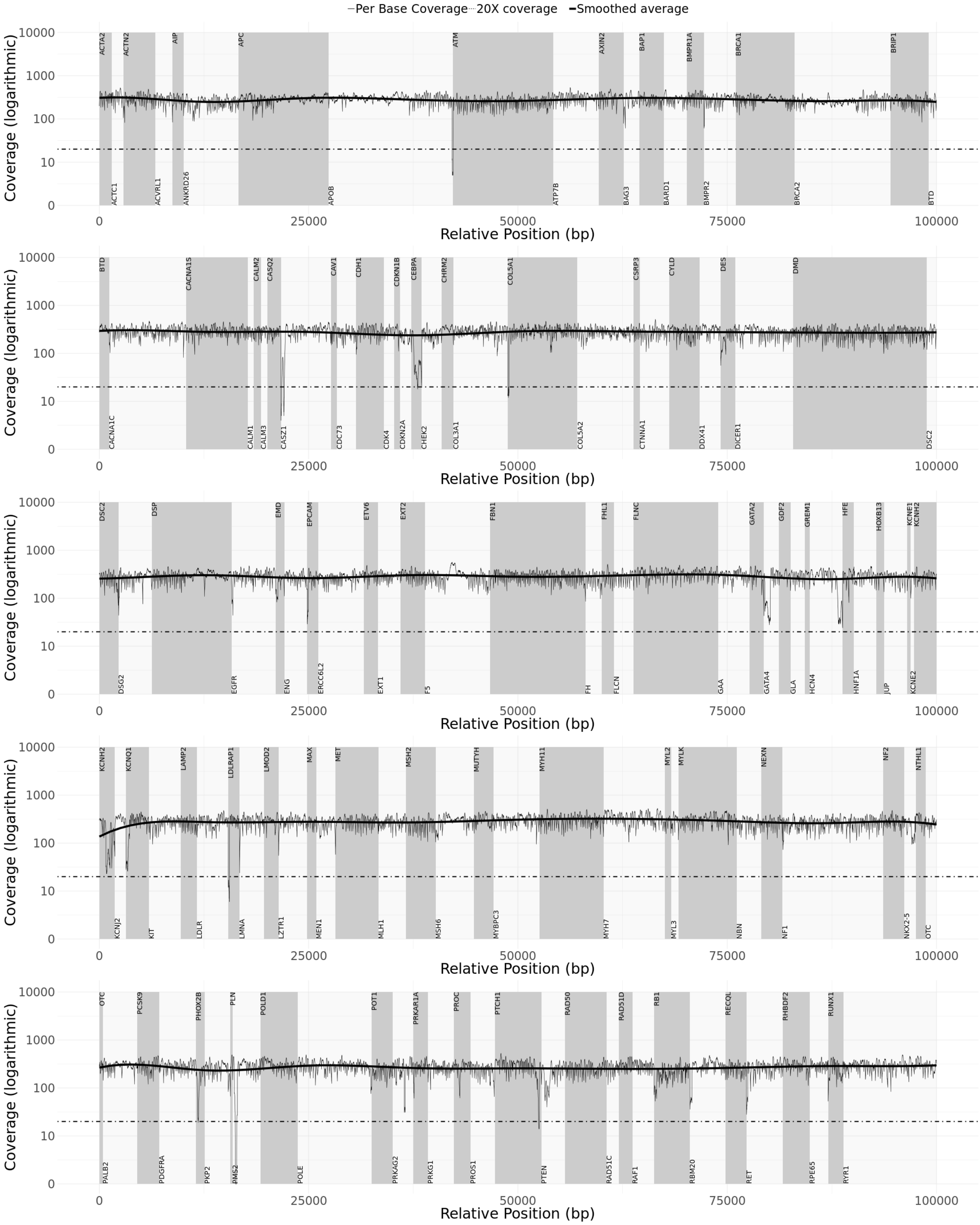
Signature

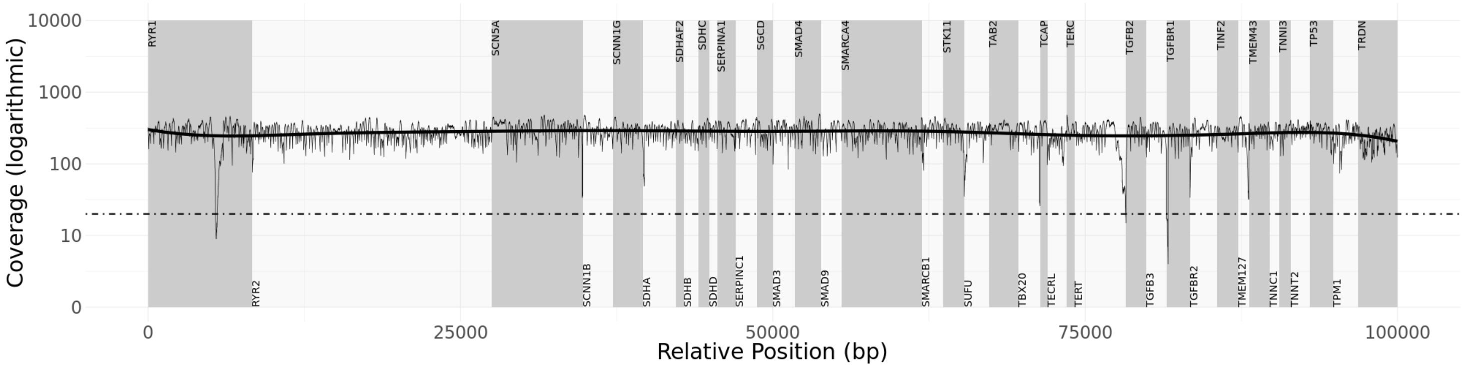
Name

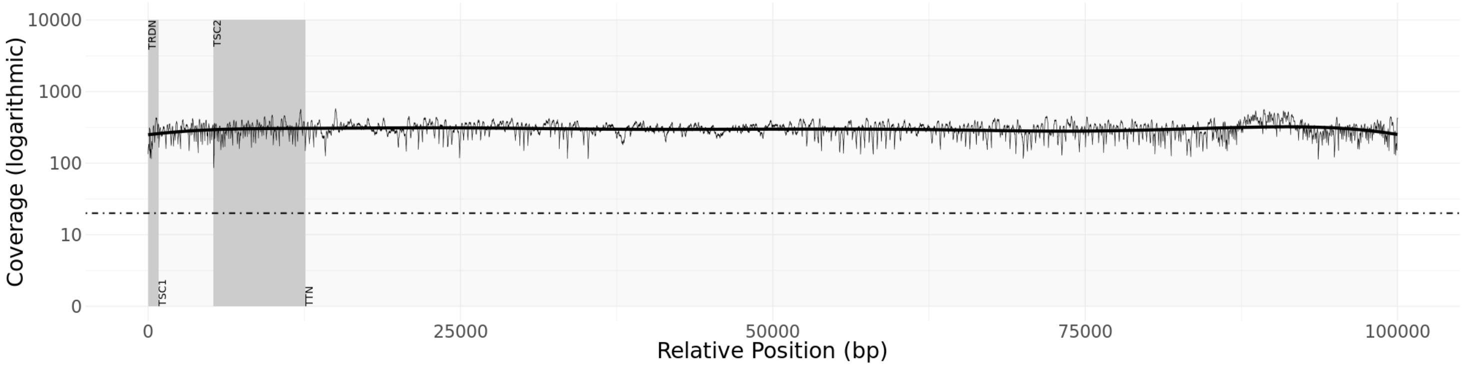
Title

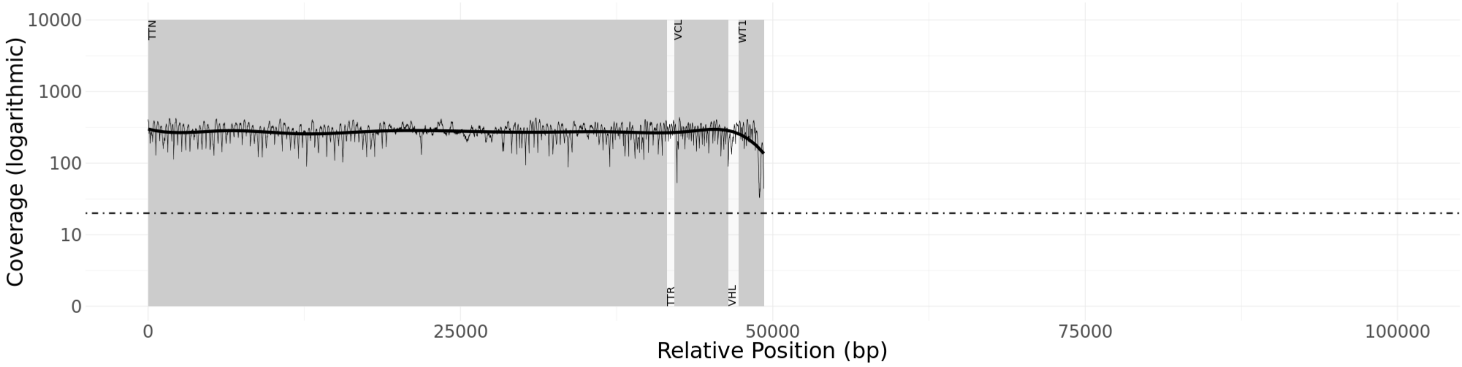
COVERAGE PLOT - NUCLEAR GENES

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.




Coverage (logarithmic)
Relative Position (bp)
RYR1
RYR2
SCN5A
SCN1B
SCNN1G
SDHA
SDHAF2
SDHB
SDHC
SDHD
SERPINA1
SERPINC1
SGCD
SMAD3
SMAD4
SMAD9
SMARCA4
SMARCB1
STK11
SUFU
TAB2
TBX20
TCAP
TECRL
TERC
TERT
TGFB2
TGFB3
TGFBR1
TGFBR2
TINF2
TMEM127
TMEM43
TNNC1
TNNI3
TNNT2
TP53
TPM1
TRDN


Coverage (logarithmic)
Relative Position (bp)
TRDN
TSC1
TSC2
TTN


Coverage (logarithmic)
Relative Position (bp)
TTN
TTR
VCL
VHL
WT1

SUMMARY OF THE TEST

Laboratory process: When required, the total genomic DNA was extracted from the biological sample using bead-based method. Quantity of DNA was assessed using fluorometric method. After assessment of DNA quantity, qualified genomic DNA sample was randomly fragmented using non-contact, isothermal sonochemistry processing. Sequencing library was prepared by ligating sequencing adapters to both ends of DNA fragments. Sequencing libraries were size-selected with 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 hybridization-based target capture method. The quality of the completed sequencing library was controlled by ensuring the correct template size and quantity and to eliminate the presence of leftover primers and adapter-adapter dimers. Ready sequencing libraries that passed the quality control were sequenced using the illumina's sequencing-by-synthesis method using paired-end sequencing (150 by 150 bases). Primary data analysis converting images into base calls and associated quality scores was carried out by the sequencing instrument using illumina's proprietary software, generating CBC files as the final output.

Bioinformatics and quality control: Base called raw sequencing data was transformed into FAST format using Illumina's software (bcl2fastq). Sequence reads of each sample were mapped to the human reference genome (GRCh37/hq19). 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 DNA. Variant data for was annotated using a collection of tools (VcfAnno and VP) 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 samples) 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: The clinical interpretation team assessed the pathogenicity of the identified variants by reviewing the relevant scientific literature and manually inspecting the sequencing data if needed. All available evidence of the identified variants was compared to classification criteria. Reporting was carried out using HGNC-approved gene nomenclature and mutation nomenclature following the HGVS guidelines. Benign variants, Likely benign variants, and Variants of uncertain significance (VUS) were not reported. Information about estimated residual risks after negative test result using Blueprint Genetics Reproductive Screen Panels is available on our website: https://blueprintgenetics.com/residual-risk-table/

Variant classification: Our variant classification follows the Blueprint Genetics Variant Classification Schemes modified from the ACMG guideline 2015. Minor modifications were made to increase reproducibility of the variant classification and improve the clinical validity of the report. The classification and interpretation of the variants) identified reflect the current state of Blueprint Genetics' understanding at the time of this report. 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. For questions regarding variant classification updates, please contact us at support@blueprintgenetics.com

Databases: The pathogenicity potential of the identified variants were assessed by considering the predicted consequence of the variant, the degree of evolutionary conservation as well as a number of reference population databases and mutation databases such as, but not limited to, the gnomAD, ClinVar, HGMD Professional and Alamut Visual. In addition, the clinical relevance of any identified CNVs was evaluated by reviewing the relevant literature and databases such as Database of Genomic Variants and DECIPHER. For interpretation of mDNA variants specific databases including e.g. Mitomap, HmtVar and 1000G were used.

Confirmation of variants: Reporting focuses on high-quality variants that meet our stringent NGS quality metrics for a true positive call but they were not confirmed with alternative methods. Ordering health care professional should consider further confirmation of the reported variants using a diagnostic test.

Analytic validation: The detection performance of this panel is expected to be in the same range as our high-quality, clinical grade NGS sequencing assay used to generate the panel data (nuclear DNA: sensitivity for SNVs 99.89%, indels 1-50 bps 99.2%, one-exon deletion 100% and five exons CNV 98.7%, and specificity >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% (10-100% heteroplasmy level), 94.7% (5-10% heteroplasmy level), 87.3% (<5% heteroplasmy level) and for gross deletions 100.0%. Specificity is >99.9% for all.

Test restrictions: A normal result does not rule out a pathogenic or likely pathogenic variant in the tested genes 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.

Technical limitations: This test does not detect the following: complex inversions, gene conversions, balanced translocations, repeat expansion disorders unless specifically mentioned, non-coding variants deeper than #20 base pairs from 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, single exon 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 above.

Regulation and accreditations: This test was developed and its performance characteristics determined by Blueprint Genetics (see Analytic validation). It has not been cleared or approved by the US Food and Drug Administration. This analysis has been performed in a CLIA-certified laboratory (#99D2092375), accredited by the College of American Pathologists (CAP #9257331) and by FINAS Finnish Accreditation Service, (laboratory no. T292), accreditation requirement SFS-EN ISO 15189:2013. All the tests are under the scope of the ISO 15189 accreditation.

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
- Interpretation

NON-CODING VARIANTS COVERED BY THE PANEL:

NM_015627.2(*LDLRAP1*):c.-17_-12dupGGCGGC
NM_015627.2(*LDLRAP1*):c.748-608G>A
NM_001128425.1(*MUTYH*):c.998-13T>G
NM_001128425.1(*MUTYH*):c.504+19_504+31delTAGGGGAAATAGG
NM_174936.3(*PCSK9*):c.-331C>A
NM_000329.2(*RPE65*):c.246-11A>G
NM_144573.3(*NEXN*):c.-52-78C>A
NM_170707.3(*LMNA*):c.513+45T>G
NM_170707.3(*LMNA*):c.937-11C>G
NM_170707.3(*LMNA*):c.1608+14G>A
NM_170707.3(*LMNA*):c.1609-12T>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_000488.3(*SERPINC1*):c.1154-14G>A
NM_000488.3(*SERPINC1*):c.42-18C>G
NM_000488.3(*SERPINC1*):c.-171C>G
NM_001035.2(*RYR2*):c.3423+32dupG
NM_014915.2(*ANKRD26*):c.-116C>G
NM_014915.2(*ANKRD26*):c.-118C>A
NM_014915.2(*ANKRD26*):c.-119C>A/G
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
chr10:g.89622883-89623482
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
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NM_000314.4(*PTEN*):c.-799G>C
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NM_000314.4(*PTEN*):c.254-21G>C
NM_000314.4(*PTEN*):c.*65T>A
NM_000314.4(*PTEN*):c.*75_*92delTAATGGCAATAGGACATTinsCTATGGCAATAGGACATTG
chr11:g.2484803-2484803
NM_000256.3(*MYBPC3*):c.*26+2T>C
NM_000256.3(*MYBPC3*):c.3628-12C>G
NM_000256.3(*MYBPC3*):c.2309-26A>G
NM_000256.3(*MYBPC3*):c.2149-80G>A
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NM_000256.3(*MYBPC3*):c.1224-19G>A
NM_000256.3(*MYBPC3*):c.1224-52G>A
NM_000256.3(*MYBPC3*):c.1091-575A>C
NM_000256.3(*MYBPC3*):c.1090+453C>T

NM_000256.3(*MYBPC3*):c.906-22G>A
NM_000256.3(*MYBPC3*):c.906-36G>A
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
chr11:g.67250360-67250360
NM_003977.2(*AIP*):c.-220G>A
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_004064.3(*CDKN1B*):c.-454_-451delTTCC
NM_000020.2(*ACVRL1*):c.1378-274C>G
NM_000020.2(*ACVRL1*):c.1378-216C>G
NM_000020.2(*ACVRL1*):c.1378-156_1378-155invCT
NM_000020.2(*ACVRL1*):c.1378-131C>G
NM_000020.2(*ACVRL1*):c.1378-78T>G
NM_000020.2(*ACVRL1*):c.1378-69C>A
NM_000545.5(*HNF1A*):c.-538G>C
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NM_000545.5(*HNF1A*):c.-258A>G
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NM_000545.5(*HNF1A*):c.-187C>A/T
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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
NM_000321.2(*RB1*):c.-193T>A/G
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>T
NM_000321.2(*RB1*):c.2490-26A>G
NM_000321.2(*RB1*):c.2490-26A>C/G/T
NM_000321.2(*RB1*):c.2490-26A>C
NM_000053.3(*ATP7B*):c.3061-12T>A
NM_000053.3(*ATP7B*):c.-78A>C
NM_000053.3(*ATP7B*):c.-123C>A
NM_000053.3(*ATP7B*):c.-128_-124delAGCCG
NM_000053.3(*ATP7B*):c.-133A>C
NM_000053.3(*ATP7B*):c.-210A>T
chr13:g.52585894-52585894
chr13:g.52585897-52585897
NM_000053.3(*ATP7B*):c.-442G>A
NM_003239.2(*TGFB3*):c.*495C>T
NM_003239.2(*TGFB3*):c.-30G>A
NM_000295.4(*SERPINA1*):c.-5+2dupT
NM_000295.4(*SERPINA1*):c.-5+1G>A
NM_177438.2(*DICER1*):c.5364+1187T>G
NM_005159.4(*ACTC1*):c.*1784T>C
NM_000138.4(*FBN1*):c.8051+375G>T
NM_000138.4(*FBN1*):c.6872-14A>G
NM_000138.4(*FBN1*):c.6872-961A>G
NM_000138.4(*FBN1*):c.5672-87A>G
NM_000138.4(*FBN1*):c.5672-88A>G
NM_000138.4(*FBN1*):c.4211-32_4211-13delGAAGAGTAACGTGTGTTTCT
NM_000138.4(*FBN1*):c.2678-15C>A
NM_000138.4(*FBN1*):c.1589-14A>G
NM_000138.4(*FBN1*):c.863-26C>T
NM_001018005.1(*TPM1*):c.241-12_241-11delCTinsTG
NM_000548.3(*TSC2*):c.-30+1G>C

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_024675.3(*PALB2*):c.109-12T>A
NM_015247.2(*CYLD*):c.1139-148A>G
NM_004360.3(*CDH1*):c.687+92T>A
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.3198-314G>A
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_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_000152.3(*GAA*):c.-32-13T>G
NM_000152.3(*GAA*):c.-32-13T>A
NM_000152.3(*GAA*):c.-32-3C>A/G
NM_000152.3(*GAA*):c.-32-2A>G
NM_000152.3(*GAA*):c.-32-1G>C
NM_000152.3(*GAA*):c.-17C>T
NM_000152.3(*GAA*):c.1076-22T>G
NM_000152.3(*GAA*):c.2190-345A>G
NM_000152.3(*GAA*):c.2647-20T>G
NM_024422.4(*DSC2*):c.-1445G>C
NM_000455.4(*STK11*):c.597+16_597+33delGGGGGGCCCTGGGGCGCCinsTG
NM_000455.4(*STK11*):c.598-32_597+31delGCCCCCTCCCGGGC
chr19:g.11199939-11199939
NM_000527.4(*LDLR*):c.-267A>G
NM_000527.4(*LDLR*):c.-228G>C
chr19:g.11200000-11200000
NM_000527.4(*LDLR*):c.-206C>T
chr19:g.11200031-11200031
chr19:g.11200032-11200032
chr19:g.11200032-11200032
NM_000527.4(*LDLR*):c.-191C>A
NM_000527.4(*LDLR*):c.-188C>T
NM_000527.4(*LDLR*):c.-185_-183delCTT
NM_000527.4(*LDLR*):c.-172G>A
NM_000527.4(*LDLR*):c.-168A>G
NM_000527.4(*LDLR*):c.-163T>C
NM_000527.4(*LDLR*):c.-161A>C
NM_000527.4(*LDLR*):c.-156C>T
NM_000527.4(*LDLR*):c.-155_-154delACinsTTCTGCAAACTCCT
NM_000527.4(*LDLR*):c.-155_-150delACCCCA
NM_000527.4(*LDLR*):c.-155_-154delACinsTTCTGCAAACTCCT
NM_000527.4(*LDLR*):c.-155_-150delACCCCAinsTT
NM_000527.4(*LDLR*):c.-154C>T

NM_000527.4(*LDLR*):c.-153C>T
NM_000527.4(*LDLR*):c.-152C>T
NM_000527.4(*LDLR*):c.-151C>G
NM_000527.4(*LDLR*):c.-150A>G
NM_000527.4(*LDLR*):c.-149C>A
NM_000527.4(*LDLR*):c.-146C>A
NM_000527.4(*LDLR*):c.-142C>G/T
NM_000527.4(*LDLR*):c.-139_-130delCTCCCCCTGC
NM_000527.4(*LDLR*):c.-140C>A/G/T
NM_000527.4(*LDLR*):c.-139C>A/G
NM_000527.4(*LDLR*):c.-138delT
NM_000527.4(*LDLR*):c.-138T>C
NM_000527.4(*LDLR*):c.-137C>T
NM_000527.4(*LDLR*):c.-136C>G/T
NM_000527.4(*LDLR*):c.-136C>G
NM_000527.4(*LDLR*):c.-136C>T
NM_000527.4(*LDLR*):c.-135C>G
NM_000527.4(*LDLR*):c.-134C>T
NM_000527.4(*LDLR*):c.-124dupA
NM_000527.4(*LDLR*):c.-120C>T
NM_000527.4(*LDLR*):c.-101T>C
NM_000527.4(*LDLR*):c.-99A>G
NM_000527.4(*LDLR*):c.-98C>T
NM_000527.4(*LDLR*):c.-23A>C
NM_000527.4(*LDLR*):c.-22delC
NM_000527.4(*LDLR*):c.-14C>A
NM_000527.4(*LDLR*):c.940+14delC
NM_000527.4(*LDLR*):c.941-13T>A
NM_000527.4(*LDLR*):c.1359-31_1359-23delGCGCTGATGinsCGGCT
NM_000527.4(*LDLR*):c.1359-25A>G
NM_000527.4(*LDLR*):c.1845+11C>G
NM_000527.4(*LDLR*):c.1845+15C>A
NM_000527.4(*LDLR*):c.2140+86C>G
NM_000527.4(*LDLR*):c.2140+103G>T
NM_000527.4(*LDLR*):c.*43G>A
NM_000540.2(*RYR1*):c.8692+131G>A
NM_000540.2(*RYR1*):c.14647-1449A>G
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_017849.3(*TMEM127*):c.-18C>T
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_000090.3(*COL3A1*):c.3256-43T>G
NM_000393.3(*COL5A2*):c.1924-11T>C
NM_001204.6(*BMPR2*):c.-947_-946delGCinsAT
NM_001204.6(*BMPR2*):c.-347C>T
NM_001204.6(*BMPR2*):c.-279C>A
NM_001204.6(*BMPR2*):c.-92C>A
NM_001204.6(*BMPR2*):c.968-12T>G
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_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_000060.2(*BTD*):c.310-15delT
NM_000060.2(*BTD*):c.*159G>A
NM_001024847.2(*TGFBR2*):c.-59C>T
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_198056.2(*SCN5A*):c.2024-11T>A
NM_198056.2(*SCN5A*):c.-53+1G>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
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_198253.2(*TERT*):c.2383-15C>T
NM_198253.2(*TERT*):c.-57A>C
chr5:g.112043009-112043595
NM_001127511.2(*APC*):c.-195A>C
NM_001127511.2(*APC*):c.-192A>G/T
NM_001127511.2(*APC*):c.-192A>G
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
chr5:g.112072710-112073585
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.172662741-172662741
chr5:g.172672291-172672291
chr5:g.172672303-172672303
NM_000410.3(*HFE*):c.-20G>A
NM_002667.4(*PLN*):c.-271A>G
NM_002667.4(*PLN*):c.-236C>G
NM_006073.3(*TRDN*):c.22+29A>G
NM_000535.5(*PMS2*):c.1145-31_1145-13delCTGACCCTCTTCTCCGTCC
NM_000535.5(*PMS2*):c.23+21_23+28delTCCGGTGT
NM_001077653.2(*TBX20*):c.-549G>A
NM_001753.4(*CAV1*):c.-88delC
NM_000238.3(*KCNH2*):c.2399-28A>G
NM_002052.3(*GATA4*):c.-989C>T
NM_002052.3(*GATA4*):c.-902G>T
chr8:g.11561399-11561399
NM_002052.3(*GATA4*):c.910-55T>C
NM_002052.3(*GATA4*):c.997+103G>T
NM_002052.3(*GATA4*):c.998-26G>A
NM_000077.4(*CDKN2A*):c.458-105A>G
NM_000077.4(*CDKN2A*):c.151-1104C>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_000264.3(*PTCH1*):c.2561-2057A>G
NM_001114753.2(*ENG*):c.1742-22T>C
NM_001114753.2(*ENG*):c.361-11T>A
NM_001114753.2(*ENG*):c.-58G>A
NM_001114753.2(*ENG*):c.-127C>T
NM_001114753.2(*ENG*):c.-142A>T
NM_000368.4(*TSC1*):c.363+668G>A
NM_000093.4(*COL5A1*):c.1720-11T>A
NM_000093.4(*COL5A1*):c.2647-12A>G
NM_000093.4(*COL5A1*):c.2701-25T>G
NM_000093.4(*COL5A1*):c.5137-11T>A

NM_004006.2(*DMD*):c.10554-18C>G
NM_004006.2(*DMD*):c.9974+175T>A
NM_004006.2(*DMD*):c.9564-30A>T
NM_004006.2(*DMD*):c.9564-427T>G
NM_004006.2(*DMD*):c.9563+1215A>G
NM_004006.2(*DMD*):c.9362-1215A>G
NM_004006.2(*DMD*):c.9361+117A>G
NM_004006.2(*DMD*):c.9225-160A>G
NM_004006.2(*DMD*):c.9225-285A>G
NM_004006.2(*DMD*):c.9225-287C>A
NM_004006.2(*DMD*):c.9225-647A>G
NM_004006.2(*DMD*):c.9225-648A>G
NM_004006.2(*DMD*):c.9224+9192C>A
NM_004006.2(*DMD*):c.9085-15519G>T
NM_004006.2(*DMD*):c.8217+32103G>T
NM_004006.2(*DMD*):c.8217+18052A>G
NM_004006.2(*DMD*):c.7661-11T>C
NM_004006.2(*DMD*):c.6913-4037T>G
NM_004006.2(*DMD*):c.6614+3310G>T
NM_004006.2(*DMD*):c.6290+30954C>T
NM_004006.2(*DMD*):c.6118-15A>G
NM_004006.2(*DMD*):c.5740-15G>T
NM_004006.2(*DMD*):c.5326-215T>G
NM_004006.2(*DMD*):c.5325+1743_5325+1760delTATTAAAAAATGGGTAGA
NM_004006.2(*DMD*):c.4675-11A>G
NM_004006.2(*DMD*):c.3787-843C>A
NM_004006.2(*DMD*):c.3603+2053G>C
NM_004006.2(*DMD*):c.3432+2240A>G
NM_004006.2(*DMD*):c.3432+2036A>G
NM_004006.2(*DMD*):c.961-5831C>T
NM_004006.2(*DMD*):c.961-5925A>C
NM_004006.2(*DMD*):c.832-15A>G
NM_004006.2(*DMD*):c.650-39498A>G
NM_004006.2(*DMD*):c.531-16T>A/G
NM_004006.2(*DMD*):c.531-16T>A
NM_004006.2(*DMD*):c.531-16T>G
NM_004006.2(*DMD*):c.265-463A>G
NM_004006.2(*DMD*):c.93+5590T>A
NM_004006.2(*DMD*):c.31+36947G>A
NM_004006.2(*DMD*):c.-54T>A
NM_000531.5(*OTC*):c.-9384G>T
chrX:g.38211584-38211584
NM_000531.5(*OTC*):c.-157T>G
NM_000531.5(*OTC*):c.-142G>A
NM_000531.5(*OTC*):c.-139A>G
NM_000531.5(*OTC*):c.-116C>T
NM_000531.5(*OTC*):c.-115C>T
NM_000531.5(*OTC*):c.-106C>A
NM_000531.5(*OTC*):c.540+265G>A
NM_000531.5(*OTC*):c.867+1126A>G
NM_000531.5(*OTC*):c.1005+1091C>G
NM_000169.2(*GLA*):c.640-11T>A
NM_000169.2(*GLA*):c.640-801G>A
NM_000169.2(*GLA*):c.640-859C>T
NM_000169.2(*GLA*):c.547+395G>C

NM_000117.2(*EMD*):c.266-27_266-10delTCTGCTACCGCTGCCCCC

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.