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 Reproductive 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

REPRODUCTIVE AND/OR PERSONAL RISKS

Negative for pathogenic and likely pathogenic variants related to personal disease risks and X-linked conditions.

CARRIERSHIP(S) OF AUTOSOMAL RECESSIVE DISEASE(S)

The individual is heterozygous for *ABCB11* c.3148C>T, p.(Arg1050Cys), which is pathogenic.
The individual is heterozygous for *DYSF* c.3121C>T, p.(Arg1041Cys), which is likely pathogenic.
The individual is heterozygous for *CEP290* c.6277del, p.(Val2093Serfs*4), which is pathogenic.
The individual is heterozygous for *PMM2* c.422G>A, p.(Arg141His), which is pathogenic.
The individual is heterozygous for a deletion of exon 7 in the *SMN1* gene. This alteration is classified as pathogenic.

Please see further details below: CARRIERSHIP(S) OF AUTOSOMAL RECESSIVE DISEASE(S).

SEQUENCING PERFORMANCE METRICS

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 20X	MEDIAN COVERAGE	PERCENT > 20X
Comprehensive Reproductive Screen	460	8126	1516574	1514888	180	99.89

TARGET REGION AND GENE LIST

The Blueprint Genetics Comprehensive Reproductive Screen (version 1, Jun 10, 2023) Plus Analysis includes sequence analysis and copy number variation analysis of the following genes: *AAAS*, *AARS2*, *ABCA3*, *ABCB11*, *ABCC6**, *ABCC8*, *ABCD1**, *ACAD9*, *ACADM*, *ACADS*, *ACADSB*, *ACADVL*, *ACOX1*, *ACSF3*, *ADA*, *ADAMTS2*[#], *ADAMTSL4*, *ADGRG1*, *ADGRV1*, *AGA*, *AGL*, *AGPS*, *AGXT*, *AHI1*, *AIFM1*, *AIRE*, *ALDH3A2*, *ALDH7A1*, *ALDOB*, *ALG6*, *ALMS1**, *ALPL*, *AMT*, *ANO10*, *AP1S1*, *AP3B1*, *AR*, *ARG1*, *ARL6*, *ARSA*, *ARSB*, *ARX*[#], *ASL*, *ASNS**, *ASPA*, *ASS1*, *ATM*, *ATP13A2*, *ATP6V1B1*, *ATP7A*, *ATP7B*, *ATRX*, *AVPR2*, *B9D1*, *B9D2*, *BBS1*, *BBS10*, *BBS12*, *BBS2*, *BBS4*, *BBS5*, *BBS7*, *BBS9*, *BCKDHA*, *BCKDHB*, *BCS1L*, *BLM*, *BRAT1*, *BSND*, *BTD*, *C10ORF2*, *CANT1*, *CAPN3*, *CASQ2*, *CBS*, *CC2D2A*[#], *CCDC88C*, *CD40*, *CD40LG*, *CDH23*, *CECR1*, *CENPJ*, *CEP290**, *CERKL*, *CFTR*, *CHAT*, *CHM*[#], *CHRNE*, *CIITA*, *CLCN1*, *CLN3*, *CLN5*, *CLN6*, *CLN8*, *CLRN1*, *CNGA3*, *CNGB3*, *COL27A1*, *COL4A3*, *COL4A4*, *COL4A5*, *COL7A1*, *COLQ*, *CPS1*, *CPT1A*, *CPT2*, *CRB1*, *CTNS*, *CTSD*, *CTSF*, *CTSK*, *CYBA*, *CYBB*, *CYP11B1**, *CYP11B2**, *CYP17A1*, *CYP19A1*, *CYP1B1*, *CYP21A2**, *CYP27A1*, *CYP27B1*, *CYP7B1*, *DBT*, *DCLRE1C**, *DDX11**, *DGAT1*, *DHCR7*, *DHDDS*, *DKC1*, *DLD*, *DMD*, *DNAH5*, *DNAI1*, *DNAI2*, *DOK7*, *DYNC2H1*, *DYSF*, *EDA*, *EIF2AK4*, *EIF2B2*, *EIF2B5*, *EMD*, *EPG5*, *ERCC2*, *ERCC6**, *ERCC6L2*, *ERCC8*, *ESCO2*, *ETFA*,

ETFB, *ETFDH*, *ETHE1*, *EVC*, *EVC2*, *EXOSC3*, *EYS**, *F11*, *F9*, *FAH*, *FAM161A*, *FANCA*, *FANCC*, *FANCE*, *FANCG*, *FANCI*, *FANCL*, *FH*, *FHL1**, *FKRP*, *FKTN*, *FMO3*, *FRAS1*, *G6PC*, *GAA*, *GALC*, *GALE*, *GALK1*, *GALNS*, *GALNT3*, *GALT*, *GAMT*, *GBA**, *GBE1*, *GCDH*, *GCH1*, *GCSH*, *GFM1*, *GFPT1*, *GJB1*, *GJB2*, *GJB6*, *GLA*, *GLB1*, *GLDC*, *GLE1*, *GNE*, *GNPAT*, *GNPTAB*, *GNPTG*, *GNS*, *GORAB*, *GP9*, *GRHPR*, *GRIP1*, *HADH*, *HADHA*, *HADHB*, *HAMP*, *HAX1*, *HBA1**, *HBA2**,[#], *HBB*, *HEXA*, *HEXB*, *HFE2*, *HGD*, *HGSNAT*, *HLCS*, *HMGCL*, *HOGA1*, *HPRT1*, *HPS1**, *HPS3*, *HPS4*, *HPS5*, *HPS6*, *HSD17B3*, *HSD17B4*, *HSD3B2*, *HYLS1*, *IDS**, *IDUA*, *IKBKAP*, *IL2RG*, *IL7R*, *IQCB1*, *ISPD*, *IVD*, *JAK3*, *KCNJ1*, *KCTD7*, *KIAA0586*[#], *KIF14*, *L1CAM*, *LAMA2*, *LAMA3*, *LAMB3*, *LAMC2*, *LARGE*, *LCA5*, *LDLR*, *LHX3*, *LIFR*, *LIG4*, *LIPA*, *LIPH*, *LOXHD1*, *LRP2*, *LRPPRC*, *LYST*, *MAN2B1*, *MCCC1*, *MCCC2*, *MCOLN1*, *MCPH1*[#], *MED17*, *MEFV*, *MESP2*, *MFSD8*, *MID1**, *MKKS*, *MKS1*, *MLC1*, *MMAA*, *MMAB*, *MMACHC*, *MMADHC*, *MPI*, *MPL*, *MPV17*, *MRE11A*, *MTHFR*, *MTM1*, *MTTP*, *MUT*, *MVK*, *MYO15A*, *MYO7A*, *NAGLU*, *NAGS*, *NBN*, *NCF2*, *NDRG1*, *NDUFAF5*, *NDUFAF6*, *NDUFS4*, *NDUFS6*, *NEB**,[#], *NGLY1*, *NPC1*, *NPC2*, *NPHP1*, *NPHP3*, *NPHS1*, *NPHS2*, *NR0B1*, *NR2E3*, *NTRK1*, *OAT*, *OCA2*, *OPA3*, *ORC4*, *OTC*, *OTOF*, *PAH*, *PC*, *PCCA*, *PCCB*[#], *PCDH15*, *PCNT*, *PDHA1*, *PDHB*, *PEPD*, *PET100*, *PEX1*, *PEX10*, *PEX12*, *PEX2*, *PEX26*, *PEX6*, *PEX7*, *PFKM*, *PHGDH*, *PHKB*, *PHYH*, *PKHD1*, *PLP1*, *PMM2*, *PNPO*, *POLG*, *POMGNT1*, *POMT1*, *POMT2*, *POR*, *PPT1*, *PRF1*, *PROP1*, *PRPS1**, *PSAP*, *PTS*, *PUS1*, *PYGL*, *PYGM*, *QDPR*, *RAB23*, *RAG1*, *RAG2*, *RAPSN*, *RARS2*, *RDH12*, *RECQL4*, *RMRP*, *RNASEH2A*, *RNASEH2B*, *RNASEH2C*, *RPE65*, *RPGR*, *RPGRIP1*, *RPGRIP1L*[#], *RS1*, *RTEL1*, *SACS*, *SAMHD1*, *SCO2*, *SEPSECS*, *SGCA*, *SGCB*, *SGCD*, *SGCG*, *SGSH*, *SLC12A1*, *SLC12A3*, *SLC12A6*, *SLC17A5*, *SLC19A2*, *SLC19A3*, *SLC1A4*, *SLC22A5*, *SLC25A13*, *SLC25A15**, *SLC25A20*, *SLC26A2*, *SLC26A3*, *SLC26A4*, *SLC35A2*, *SLC35A3*, *SLC37A4*, *SLC39A4*, *SLC6A8**, *SLC7A7*, *SMARCAL1*, *SMN1**,[#], *SMPD1*, *SPG11*, *SPG7*, *SPINK5*, *SPR*, *SRD5A2*, *ST3GAL5*, *STAR*, *STS*, *STXBP2*, *SUMF1*, *SURF1*, *SYNE4*, *TANGO2*, *TAT*, *TBCE*, *TCIRG1*, *TCTN2*, *TECPR2*, *TECRL*, *TF*, *TFR2*, *TG*, *TGM1*, *TH*, *TMEM107*, *TMEM216*, *TMEM231*, *TMEM67*, *TMPRSS3*, *TPO*, *TPP1*, *TRAF3IP1*, *TREX1*, *TRIM37*, *TSEN2*[#], *TSEN34*, *TSEN54*, *TSFM*[#], *TTC37*, *TTC8*, *TTPA*, *TYMP*, *TYR**, *UBR1*, *UNC13D*, *USH1C*, *USH2A*, *VPS13A*, *VPS13B*, *VPS45*[#], *VPS53*, *VRK1*, *XPA*, *XPC*, *ZFYVE26*, *ZNF469* and *ZNHIT3*[#]. The following exons are not included in the panel as they are not covered with sufficient high quality sequence reads: ADAMTS2 (NM_021599:11), CC2D2A (NM_020785:7), CHM (NM_001145414:5), KIAA0586 (NM_001244189:6, 33), MCPH1 (NM_001322042:14), PCCB (NM_001178014:4), RPGRIP1L (NM_015272:23), TSEN2 (NM_001321278:12), TSFM (NM_001172696:5), VPS45 (NM_001279353:13) and ZNHIT3 (NM_001281432:5). 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

TEST INDICATION

This individual is a 40-year-old. Genetic testing with the Comprehensive Reproductive Screen Panel has been requested.

CLINICAL REPORT

Sequence and Del/Dup (CNV) analysis using the Blueprint Genetics (BpG) Comprehensive Reproductive Screen Panel detected one or more heterozygous likely pathogenic or pathogenic variants. Please see Carriership(s) of autosomal recessive disease(s) for further details.

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

CARRIERSHIP(S) OF AUTOSOMAL RECESSIVE DISEASE(S)

This table includes variants that indicate a heterozygous carrier status of a pathogenic or likely pathogenic variant related to autosomal recessive disorder.

Heterozygous variants related to autosomal recessive disorders are not sufficient to cause disease without a second disease-causing variant on the other parental allele in the same gene. For reproductive risk estimation, genetic testing of a partner is recommended. In addition, genetic counseling is recommended.

SEQUENCE ALTERATIONS

GENE	TRANSCRIPT	NOMENCLATURE	GENOTYPE	CONSEQUENCE	INHERITANCE	CLASSIFICATION
ABCB11	NM_003742.4	c.3148C>T, p.(Arg1050Cys)	HET	missense_variant	AD,AR	**Pathogenic**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
	rs72549398	GRCh37/hg19	2:169788952	G/A		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	3/241292	probably damaging	deleterious	disease causing	Cholestasis, Cholestasis, benign recurrent intrahepatic, 2	
GENE	**TRANSCRIPT**	**NOMENCLATURE**	**GENOTYPE**	**CONSEQUENCE**	**INHERITANCE**	**CLASSIFICATION**
DYSF	NM_001130978.2	c.3121C>T, p.(Arg1041Cys)	HET	missense_variant	AR	**Likely pathogenic**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
	rs144598063	GRCh37/hg19	2:71797818	C/T		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	51/281918	probably damaging	deleterious	disease causing	Miyoshi muscular dystrophy, Muscular dystrophy, limb-girdle, Myopathy, distal, with anterior tibial onset	
GENE	**TRANSCRIPT**	**NOMENCLATURE**	**GENOTYPE**	**CONSEQUENCE**	**INHERITANCE**	**CLASSIFICATION**
CEP290	NM_025114.4	c.6277del, p.(Val2093Serfs*4)	HET	frameshift_variant	AR	**Pathogenic**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
		GRCh37/hg19	12:88456548	AC/A		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	11/224022	N/A	N/A	N/A	Bardet-Biedl syndrome, Joubert syndrome, Leber congenital amaurosis, Meckel syndrome, Senior-Loken syndrome	
GENE	**TRANSCRIPT**	**NOMENCLATURE**	**GENOTYPE**	**CONSEQUENCE**	**INHERITANCE**	**CLASSIFICATION**
PMM2	NM_000303.3	c.422G>A, p.(Arg141His)	HET	missense_variant	AR	**Pathogenic**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
	rs28936415	GRCh37/hg19	16:8905010	G/A		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	891/224376	benign	deleterious	disease causing	Congenital disorder of glycosylation	

COPY NUMBER ABERRATIONS

GENE	EVENT	COPY NUMBER	GENOTYPE	IMPACT	LINKS	CLASSIFICATION
SMN1	COPY NUMBER LOSS	1	HET	SMN1	UCSC	**Pathogenic**
	OMIM	**PHENOTYPE**			**COMMENT**	
		Spinal muscular atrophy			-	

DETAILS ABOUT THESE FINDINGS

ABCB11 c.3148C>T, p.(Arg1050Cys)

This variant has been reported as heterozygous in a male patient with progressive familial intrahepatic cholestasis (PFIC) diagnosed at birth who also had two heterozygous variants (phase unknown) in *MDR3* gene. The variant was also reported in a female patient with benign recurrent intrahepatic cholestasis (BRIC) together with *ABCB11* p.(Glu135Lys) variant (phase unknown; PMID: 28733223). In addition, it has been reported as homozygous in two siblings and as compound heterozygous together with *ABCB11* IVS19+1G>A variant in two siblings with BRIC (PMID: 5300568). The variant was also reported as compound heterozygous with *ABCB11* c.3904G>T, p.(Glu1302*) variant in a baby girl with severe cholestasis (PMID: 31015375). Functional *in vitro* analysis of the variant has shown that ABCB11 p.(Arg1050Cys) variant result an intermediate level (~25% of WT) of expression at the plasma membrane and an immature protein formation (PMID: 17855769, 19101985). The variant has been detected by other laboratories in the context of clinical testing and submitted to ClinVar (variation ID 374098).

DYSF c.3121C>T, p.(Arg1041Cys)

DYSF c.3121C>T, p.(Arg1041Cys) has been identified in the homozygous state in two patients. One of them was diagnosed with Miyoshi myopathy (PMID: 15469449) and the other one had a diagnosis of autosomal recessive limb-girdle muscular dystrophy-2 (LGMDR2) (PMID: 33610434). Western blot analysis of that patient's muscle biopsy sample showed absent dysferlin protein. The variant has been submitted to ClinVar by other clinical testing laboratories (variation ID 286743).

CEP290 c.6277del, p.(Val2093Serfs*4)

This variant generates a premature stop codon in exon 46 (of 54 total exons) and is predicted to lead to loss of normal protein function, either through protein truncation or nonsense-mediated mRNA decay. This variant has been reported together with another disease-causing variant in *CEP290* in patients affected with Joubert syndrome or Joubert-syndrome-related disorder (PMID: 17564967, 21866095, 26092869) The variant has also been detected by other laboratories in the context of clinical testing and submitted to ClinVar (Variation ID: 217621).

PMM2 c.422G>A, p.(Arg141His)

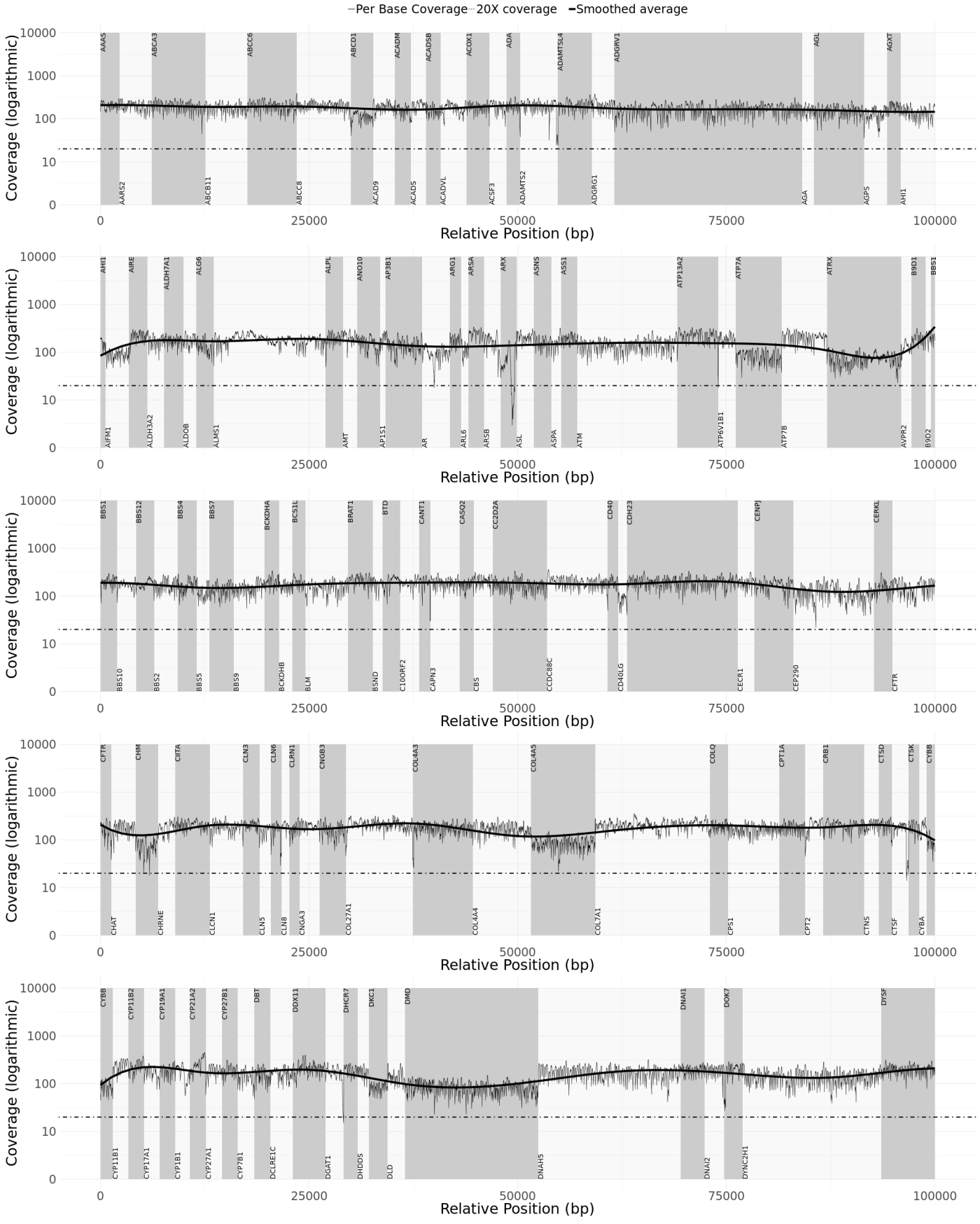
PMM2 c.422G>A, p.(Arg141His) is a well established pathogenic variant, and has been reported in compound heterozygous state in multiple individuals with *PMM2*-related congenital disorder of glycosylation (PMM2-CDG, or CDG1A) (PMID: 9140401, 11517108, 21541725, 24424124, 21228398, 23988505, 11589167, 19357119, 25192236, 30991241, 31981409, HGMD, ClinVar ID 7706). The variant is well documented as the most frequent pathogenic variant in the *PMM2* gene, present in up to 75% of PMM2-CDG patients of Caucasian origin. PMM2 p.(Arg141His) was first described by Matthijs et al. (1997) in a group of 33 unrelated individuals with confirmed phosphomannomutase deficiency (PMID: 9140401). A subsequent review of 249 patients with *PMM2* variants from six different studies and 23 different countries concluded that the p.(Arg141His) variant accounts for 37 percent of all disease-associated *PMM2* alleles (PMID: 11058895). Interestingly, while the carrier frequency of this variant can be as high as 1 in 60 in some populations, no patient homozygous for this variant has been reported, indicating that it may not be compatible with life in the homozygous state (PMID: 11530212, 10854097). Functional studies of the Arg141His mutant enzyme in bacteria demonstrated reduced enzyme stability and catalytic activity below detection limits (PMID: 21541725).

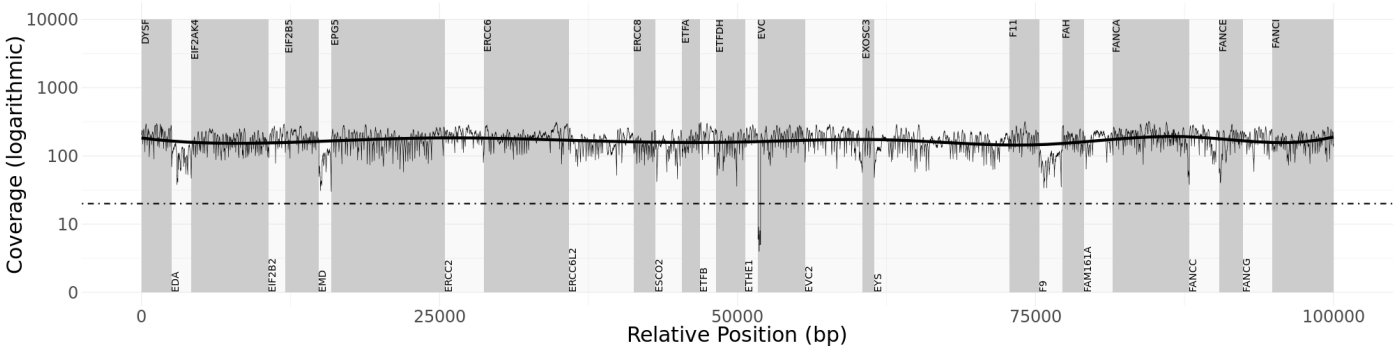
Deletion of exon 7 in the *SMN1* gene

This deletion includes at least exon 7 of the *SMN1* gene, and it is considered compatible to a deletion of whole *SMN1* gene. There is limited sensitivity to detect the exact break points of the deletion due to the complexity of the *SMN1* gene region. Homozygous loss of *SMN1,* caused by deletion causes spinal muscular atrophy (SMA; OMIM #253300); absence of exon 7 of *SMN1* has been identified in about 95% of patients with SMA (PMID: 9950358).

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.

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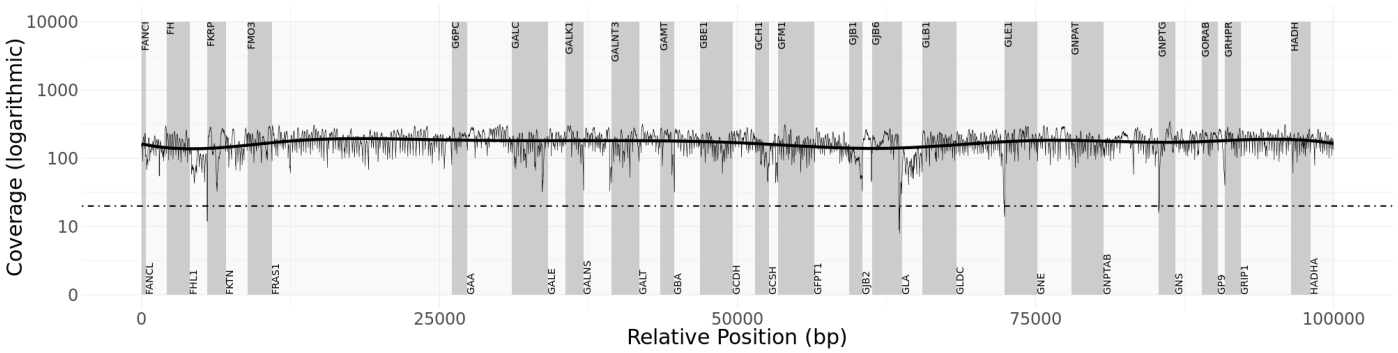
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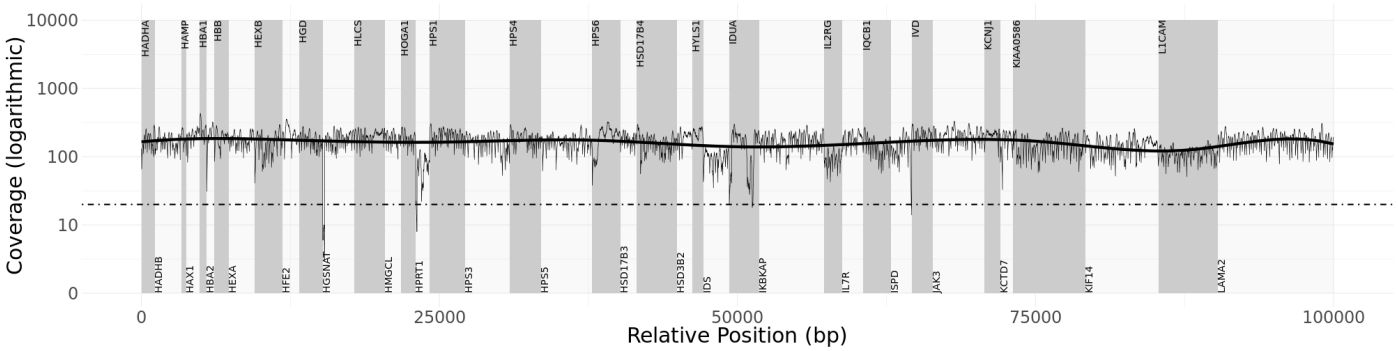
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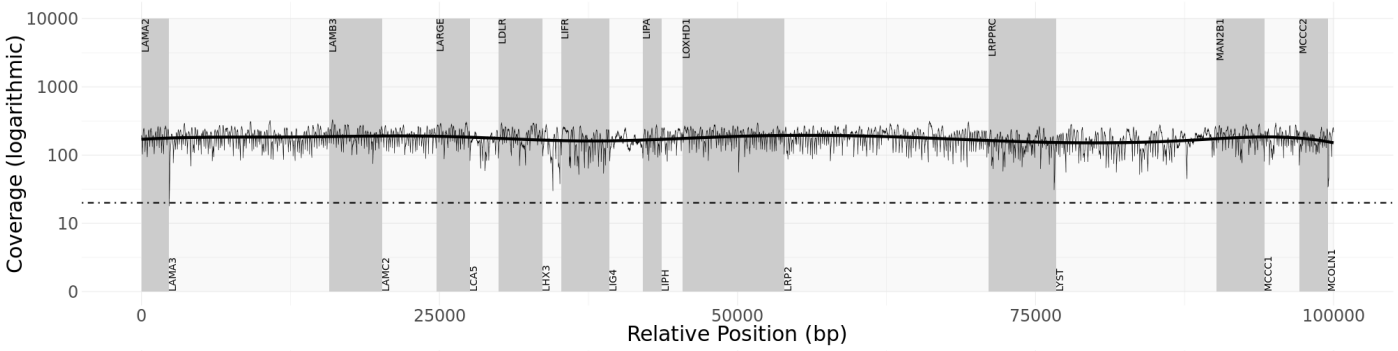
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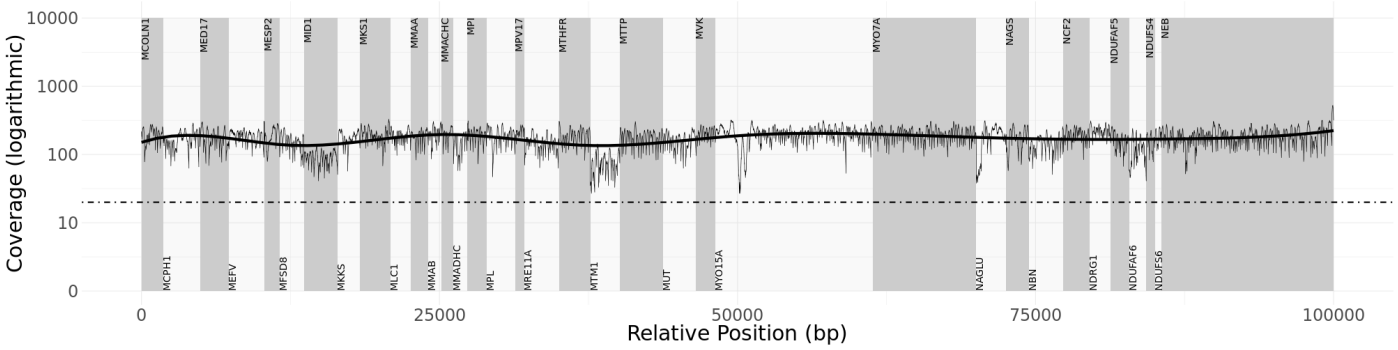
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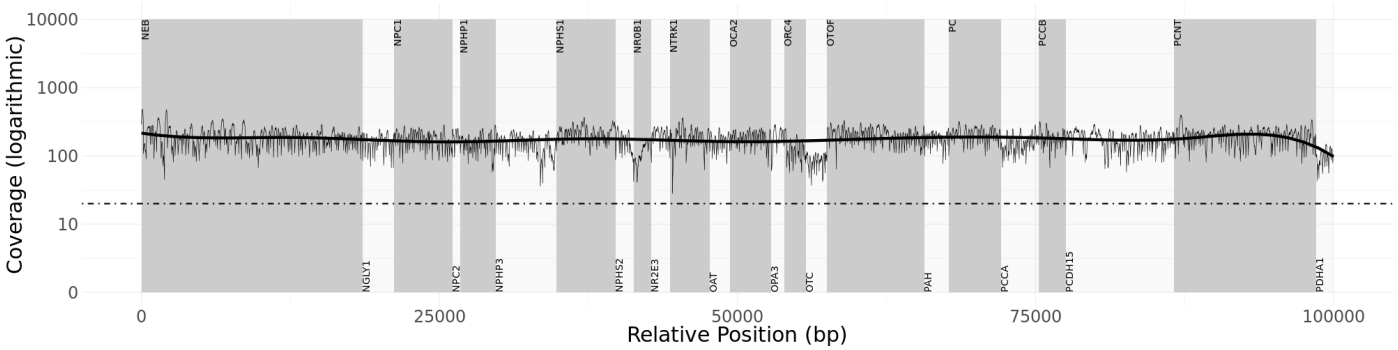
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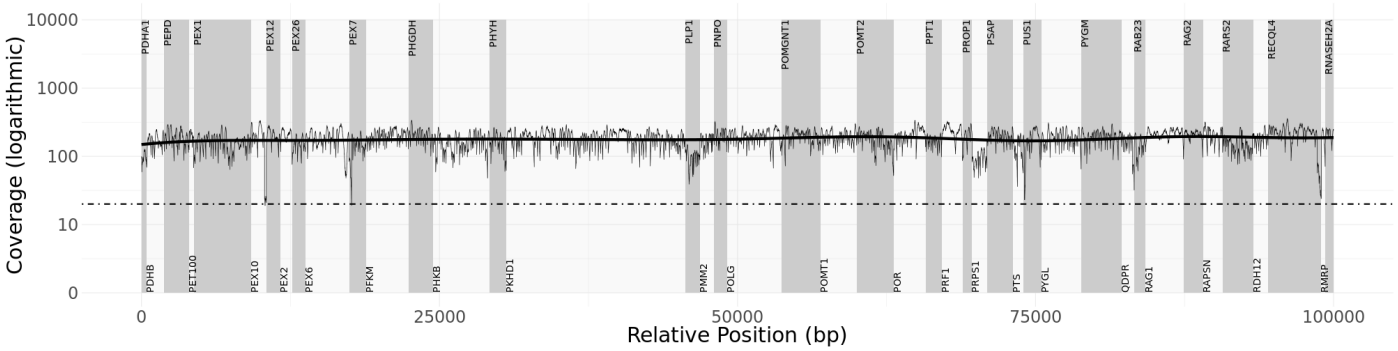
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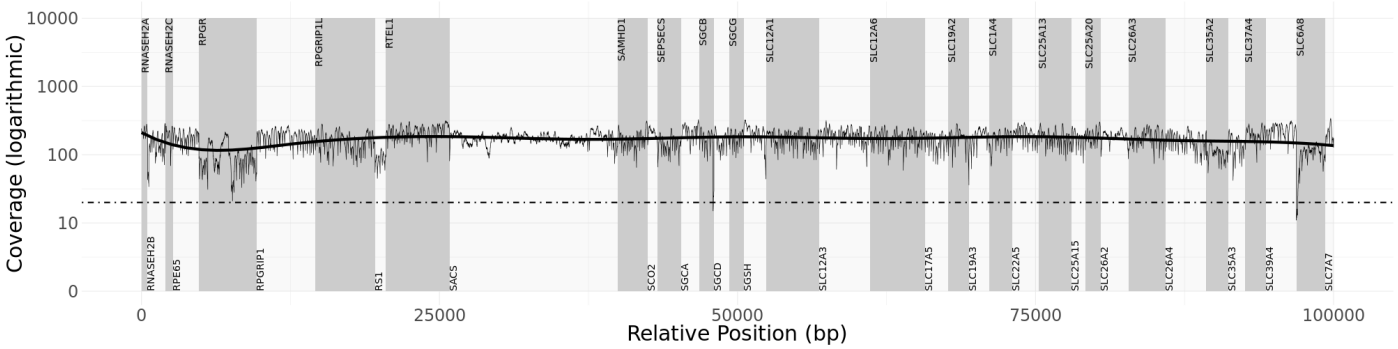
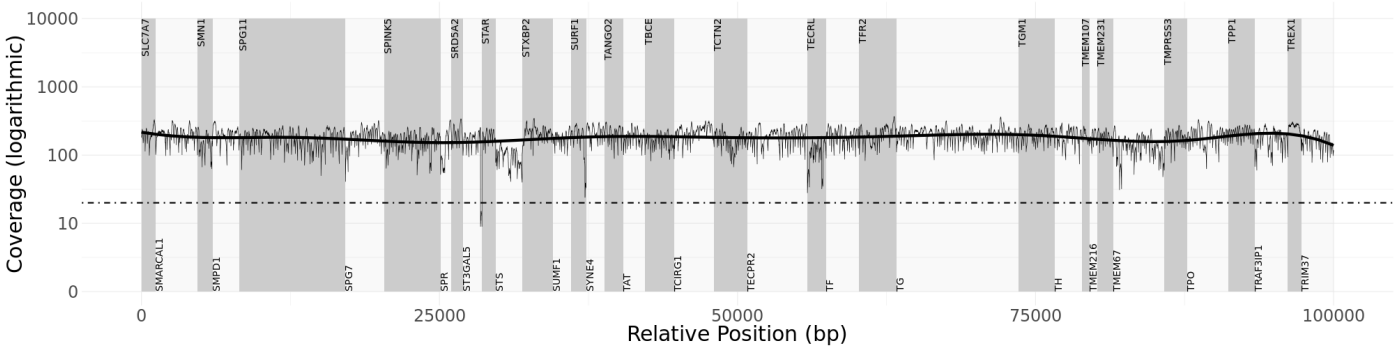
Relative Position (bp)


Coverage (logarithmic)
10000
1000
100
10
0
RNASEH2A
RNASEH2C
RPGR
RPGRIP1L
RTEL1
SAMHD1
SEPSECS
SGCB
SGCG
SLC12A1
SLC12A6
SLC19A2
SLC1A4
SLC25A13
SLC25A20
SLC26A3
SLC35A2
SLC37A4
SLC6A8
RNASEH2B
RPE65
RPGRIP1
RS1
SACS
SCO2
SGCA
SGCD
SGSH
SLC12A3
SLC17A5
SLC19A3
SLC22A5
SLC25A15
SLC26A2
SLC26A4
SLC35A3
SLC39A4
SLC7A7
0
25000
50000
75000
100000
Relative Position (bp)


Coverage (logarithmic)
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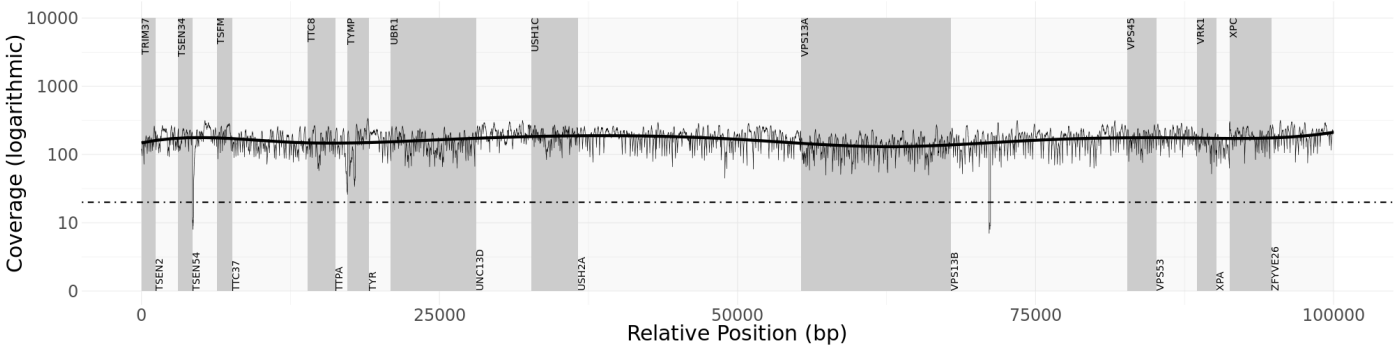
1000
100
10
0
SLC7A7
SMN1
SPG11
SPINK5
SRD5A2
STAR
STXBP2
SURF1
TANGO2
TBCE
TCTN2
TECRL
TFR2
TGM1
TMEM107
TMEM231
TMPRSS3
TPP1
TREX1
SMARCAL1
SMPD1
SPG7
SPR
ST3GAL5
STS
SUMF1
SYNE4
TAT
TCIRG1
TECPR2
TF
TG
TH
TMEM216
TMEM67
TPO
TRAF3IP1
TRIM37
0
25000
50000
75000
100000
Relative Position (bp)


Coverage (logarithmic)
10000
1000
100
10
0
TRIM37
TSEN34
TSFM
TTC8
TYMP
UBR1
USH1C
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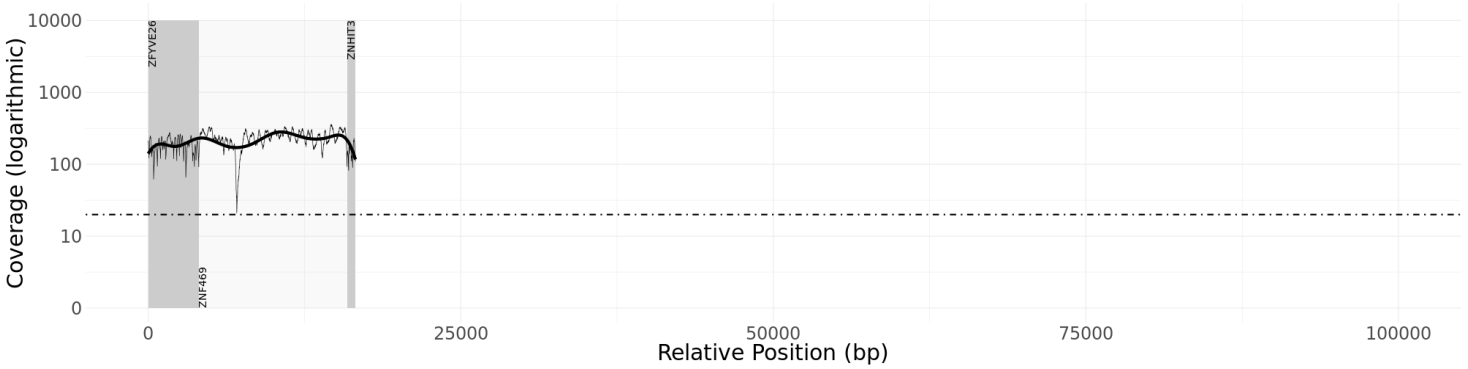
VPS45
VRK1
XPC
TSEN2
TSEN54
TTC37
TTPA
TYR
UNC13D
USH2A
VPS13B
VPS53
XPA
ZFYVE26
0
25000
50000
75000
100000
Relative Position (bp)


10000
1000
100
10
0
Coverage (logarithmic)
ZFYVE26
ZNF469
ZNHIT3
0
25000
50000
75000
100000
Relative Position (bp)

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_005957.4(*MTHFR*):c.1753-18G>A
NM_005957.4(*MTHFR*):c.-13-28_-13-27delCT
NM_000478.4(*ALPL*):c.-195C>T
NM_000478.4(*ALPL*):c.793-30_793-11delGGCATGTGCTGACACAGCCC
NM_024887.3(*DHDDS*):c.441-24A>G
NM_000310.3(*PPT1*):c.*526_*529delATCA
NM_000310.3(*PPT1*):c.125-15T>G
NM_013339.3(*ALG6*):c.347-13C>G
NM_000329.2(*RPE65*):c.246-11A>G
NM_000016.4(*ACADM*):c.388-19T>A
NM_000016.4(*ACADM*):c.600-18G>A
NM_000028.2(*AGL*):c.4260-12A>G

NM_001918.3(*DBT*):c.1018-550A>G
NM_213653.3(*HFE2*):c.-89-4dupT
NM_000396.3(*CTSK*):c.244-29A>G
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_002529.3(*NTRK1*):c.575-19G>A
NM_002529.3(*NTRK1*):c.851-33T>A
NM_002529.3(*NTRK1*):c.2206-12C>A
NM_002529.3(*NTRK1*):c.2206-11G>A
NM_014625.2(*NPHS2*):c.-164C>T
NM_000228.2(*LAMB3*):c.1133-22G>A
NM_000228.2(*LAMB3*):c.-38+1G>A
NM_206933.2(*USH2A*):c.14583-20C>G
NM_206933.2(*USH2A*):c.9959-4159A>G
NM_206933.2(*USH2A*):c.8845+628C>T
NM_206933.2(*USH2A*):c.7595-2144A>G
NM_206933.2(*USH2A*):c.5573-834A>G
NM_206933.2(*USH2A*):c.486-14G>A
NM_206933.2(*USH2A*):c.-259G>T
NM_000124.3(*ERCC6*):c.2599-26A>G
NM_001142763.1(*PCDH15*):c.-29+1G>C
NM_001042465.1(*PSAP*):c.778-26C>A
NM_199292.2(*TH*):c.1198-24T>A
NM_199292.2(*TH*):c.738-34G>C
NM_199292.2(*TH*):c.-69T>A
NM_199292.2(*TH*):c.-70G>A
NM_199292.2(*TH*):c.-71C>T
NM_000518.4(*HBB*):c.*132C>A/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>G/T
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>A/C
NM_000518.4(*HBB*):c.*110T>G
NM_000518.4(*HBB*):c.*108A>C/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.-29G>A/T
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.-50A>G/T
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
NM_000518.4(*HBB*):c.-77A>G/T
chr11:g.5248328-5248328
chr11:g.5248328-5248328
chr11:g.5248329-5248329
chr11:g.5248329-5248329
NM_000518.4(*HBB*):c.-78A>C/G
NM_000518.4(*HBB*):c.-79A>G
chr11:g.5248330-5248330
chr11:g.5248331-5248331
chr11:g.5248331-5248331
NM_000518.4(*HBB*):c.-80T>A/C
NM_000518.4(*HBB*):c.-81A>C/G
chr11:g.5248332-5248332
chr11:g.5248332-5248332
chr11:g.5248333-5248333
chr11:g.5248333-5248333
NM_000518.4(*HBB*):c.-82C>A/T
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
NM_000518.4(*HBB*):c.-136C>A/G/T
NM_000518.4(*HBB*):c.-137C>A/G/T
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.-138C>A/T
chr11:g.5248391-5248391
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_000543.4(*SMPD1*):c.1341-21_1341-18delAATG
NM_000391.3(*TPP1*):c.887-18A>G
NM_000352.3(*ABCC8*):c.4412-13G>A
NM_000352.3(*ABCC8*):c.3399+13G>A
NM_000352.3(*ABCC8*):c.2041-12C>A
NM_000352.3(*ABCC8*):c.2041-21G>A
NM_000352.3(*ABCC8*):c.2041-25G>A
NM_000352.3(*ABCC8*):c.1672-20A>G
NM_000352.3(*ABCC8*):c.1333-1013A>G
NM_000352.3(*ABCC8*):c.1177-53_1177-51delGTG
NM_000352.3(*ABCC8*):c.-190C>G
NM_000536.3(*RAG2*):c.-28G>C
NM_005055.4(*RAPSN*):c.193-15C>A
NM_005055.4(*RAPSN*):c.-199C>G
NM_005055.4(*RAPSN*):c.-210A>G
NM_005609.2(*PYGM*):c.661-601G>A
NM_005609.2(*PYGM*):c.425-26A>G
NM_024649.4(*BBS1*):c.951+58C>T
NM_000920.3(*PC*):c.1369-29A>G
NM_006019.3(*TCIRG1*):c.-5+1G>C/T
NM_006019.3(*TCIRG1*):c.-5+1G>C
NM_006019.3(*TCIRG1*):c.-5+1G>T
NM_006019.3(*TCIRG1*):c.1887+132T>C
NM_006019.3(*TCIRG1*):c.1887+142T>A
NM_006019.3(*TCIRG1*):c.1887+146G>A
NM_006019.3(*TCIRG1*):c.1887+149C>T
NM_000260.3(*MYO7A*):c.-48A>G
NM_000260.3(*MYO7A*):c.3109-21G>A
NM_000260.3(*MYO7A*):c.5327-14T>G
NM_000260.3(*MYO7A*):c.5327-11A>G
NM_000260.3(*MYO7A*):c.5857-27_5857-26insTTGAG
NM_000372.4(*TYR*):c.1037-18T>G
NM_001080463.1(*DYNC2H1*):c.2819-14A>G
NM_001080463.1(*DYNC2H1*):c.6478-16G>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_000317.2(*PTS*):c.84-323A>T
NM_000317.2(*PTS*):c.84-291A>G

NM_000317.2(*PTS*):c.164-716A>T
NM_000317.2(*PTS*):c.187-38dupG
NM_001166686.1(*PFKM*):c.1626-64A>G
NM_015665.5(*AAAS*):c.-405C>T
NM_025114.3(*CEP290*):c.6012-12T>A
NM_025114.3(*CEP290*):c.2991+1655A>G
NM_025114.3(*CEP290*):c.1910-11T>G
NM_025114.3(*CEP290*):c.103-18_103-13delGCTTTT
NM_024312.4(*GNPTAB*):c.1613-25delA
NM_000277.1(*PAH*):c.*144A>G
NM_000277.1(*PAH*):c.1199+20G>C
NM_000277.1(*PAH*):c.1199+17G>A
NM_000277.1(*PAH*):c.1066-11G>A
NM_000277.1(*PAH*):c.1066-12delT
NM_000277.1(*PAH*):c.1066-13T>G
NM_000277.1(*PAH*):c.1066-14C>G
NM_000277.1(*PAH*):c.1065+39G>T
NM_000277.1(*PAH*):c.509+15_509+18delCTTG
NM_000277.1(*PAH*):c.169-13T>G
NM_000431.2(*MVK*):c.769-7dupT
NM_004004.5(*GJB2*):c.-22-2A>C
NM_004004.5(*GJB2*):c.-23+2T>A
NM_004004.5(*GJB2*):c.-23+1G>A
NM_004004.5(*GJB2*):c.-23G>T
NM_004004.5(*GJB2*):c.-259C>T
NM_004004.5(*GJB2*):c.-260C>T
NM_000231.2(*SGCG*):c.-127_-121delACAGTTG
NM_000231.2(*SGCG*):c.-1+1G>T
NM_024570.3(*RNASEH2B*):c.65-13G>A
NM_024570.3(*RNASEH2B*):c.511-13G>A
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_000282.3(*PCCA*):c.1285-1416A>G
NM_020366.3(*RPGRIP1*):c.1468-263G>C
NM_020366.3(*RPGRIP1*):c.1611+27G>A
NM_020366.3(*RPGRIP1*):c.2367+23delG
NM_020366.3(*RPGRIP1*):c.2367+23delG
NM_020366.3(*RPGRIP1*):c.2711-13G>T
NM_000359.2(*TGM1*):c.509-329C>T
NM_000161.2(*GCH1*):c.-22C>T
NM_013382.5(*POMT2*):c.1333-14G>A
NM_000153.3(*GALC*):c.*12G>A
NM_000153.3(*GALC*):c.-66G>C
NM_000153.3(*GALC*):c.-67T>G
NM_001201402.1(*GALC*):c.-74T>A
NM_001201402.1(*GALC*):c.-128C>T
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_000070.2(*CAPN3*):c.380-13T>A
NM_000070.2(*CAPN3*):c.1746-20C>T
NM_173089.1(*CAPN3*):c.-188G>C
NM_000070.2(*CAPN3*):c.2184+21G>A
NM_000070.2(*CAPN3*):c.2185-16A>G
NM_174916.2(*UBR1*):c.1911+14C>G
NM_174916.2(*UBR1*):c.1094-12A>G
NM_174916.2(*UBR1*):c.1094-13A>G
NM_174916.2(*UBR1*):c.529-13G>A
NM_000338.2(*SLC12A1*):c.976-14C>G
NM_017882.2(*CLN6*):c.297+113G>C
NM_000520.4(*HEXA*):c.1146+18A>G
NM_033028.4(*BBS4*):c.77-216delA
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_032520.4(*GNPTG*):c.610-16_609+28del
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_000243.2(*MEFV*):c.-12C>G
NM_000243.2(*MEFV*):c.-382C>G
chr16:g.8891573-8891573
NM_000303.2(*PMM2*):c.179-25A>G
NM_000303.2(*PMM2*):c.640-15479C>T
NM_000303.2(*PMM2*):c.640-23A>G
NM_001171.5(*ABCC6*):c.4403+11C>G
NM_001171.5(*ABCC6*):c.3506+15G>A
NM_001171.5(*ABCC6*):c.1780-29T>A
NM_001171.5(*ABCC6*):c.1432-22C>A
NM_000086.2(*CLN3*):c.1056+34C>A
NM_000086.2(*CLN3*):c.461-13G>C
NM_000339.2(*SLC12A3*):c.602-16G>A
NM_000339.2(*SLC12A3*):c.1567+297T>G
NM_000339.2(*SLC12A3*):c.1670-191C>T
NM_000339.2(*SLC12A3*):c.2548+253C>T
NM_001145774.1(*ADGRG1*):c.-435_-421delCAACGGTTGCCAGGG
NM_001077416.2(*TMEM231*):c.824-11T>C
NM_000101.3(*CYBA*):c.288-15C>G
NM_000512.4(*GALNS*):c.899-167A>G
NM_000512.4(*GALNS*):c.245-11C>G
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
NM_000135.2(*FANCA*):c.893+920C>A
NM_004937.2(*CTNS*):c.-643_-642insT
NM_001031681.2(*CTNS*):c.-19-1G>A
NM_001031681.2(*CTNS*):c.141-24T>C
NM_001031681.2(*CTNS*):c.971-12G>A
NM_000080.3(*CHRNE*):c.501-16G>A
NM_000080.3(*CHRNE*):c.-94G>A
NM_000080.3(*CHRNE*):c.-95G>A
NM_000080.3(*CHRNE*):c.-96C>T
NM_000018.3(*ACADVL*):c.-144_-132delCCCAGCATGCCCCinsT
NM_001270447.1(*ACADVL*):c.822-27C>T
NM_001270447.1(*ACADVL*):c.822-11T>G
NM_001270447.1(*ACADVL*):c.1146+15C>T
NM_001270447.1(*ACADVL*):c.1252-15A>G
NM_001270447.1(*ACADVL*):c.1747+23C>T
NM_001031806.1(*ALDH3A2*):c.681-14T>A/G
NM_001031806.1(*ALDH3A2*):c.681-14T>A
NM_001031806.1(*ALDH3A2*):c.681-14T>G
NM_000151.3(*G6PC*):c.446+39G>A
NM_000151.3(*G6PC*):c.446+42G>A
NM_153006.2(*NAGS*):c.-3063C>A
NM_000023.2(*SGCA*):c.585-31_585-23delTCTGCTGAC
NM_000023.2(*SGCA*):c.585-31_585-24delTCTGCTGA
NM_000023.2(*SGCA*):c.748-12_748-11delCTinsAA
NM_015294.3(*TRIM37*):c.1949-12A>G
NM_000154.1(*GALK1*):c.-22T>C
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_138793.3(*CANT1*):c.-342+1G>A
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_000199.3(*SGSH*):c.249+27_249+28delGG
NM_000271.4(*NPC1*):c.1554-1009G>A
NM_000271.4(*NPC1*):c.882-28A>G/T
NM_000271.4(*NPC1*):c.882-28A>G
NM_000271.4(*NPC1*):c.882-28A>T
NM_198129.1(*LAMA3*):c.4584+22_4584+24delTCT
NM_138924.2(*GAMT*):c.391+15G>T
NM_006949.3(*STXBP2*):c.326-23_326-16delGCCCCACT
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.-138delT
NM_000527.4(*LDLR*):c.-139C>A/G
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.-22delC
NM_000527.4(*LDLR*):c.-23A>C
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_000159.3(*GCDH*):c.1244-11A>G
NM_000215.3(*JAK3*):c.2680+89G>A
NM_000215.3(*JAK3*):c.1915-11G>A
NM_021175.2(*HAMP*):c.-153C>T
NM_021175.2(*HAMP*):c.-28G>T

NM_021175.2(*HAMP*):c.-25G>A
NM_004646.3(*NPHS1*):c.1931-17C>A
NM_004646.3(*NPHS1*):c.1930+11C>A
NM_004646.3(*NPHS1*):c.-475_-468delGAGAGAGA
NM_000709.3(*BCKDHA*):c.*223T>A
chr19:g.44031407-44031407
NM_024301.4(*FKRP*):c.-272G>A
NM_000183.2(*HADHB*):c.442+614A>G
NM_000183.2(*HADHB*):c.442+663A>G
NM_000104.3(*CYP1B1*):c.-322A>C
NM_000104.3(*CYP1B1*):c.-337G>T
NM_001114636.1(*FANCL*):c.375-2033C>G
NM_003494.3(*DYSF*):c.3443-33A>G
NM_003494.3(*DYSF*):c.4410+13T>G
NM_003494.3(*DYSF*):c.4886+1249G>T
NM_003494.3(*DYSF*):c.5668-824C>T
NM_003494.3(*DYSF*):c.*107T>A
NM_003124.4(*SPR*):c.-13G>A
NM_001298.2(*CNGA3*):c.-37-1G>C
NM_001271208.1(*NEB*):c.24220-151C>A
NM_001271208.1(*NEB*):c.19429-381_19429-379delTTTinsA
NM_003742.2(*ABCB11*):c.77-19T>A
NM_152384.2(*BBS5*):c.619-27T>G
NM_001875.4(*CPS1*):c.4102-239A>G
NM_004328.4(*BCS1L*):c.-147A>G
NM_004328.4(*BCS1L*):c.-50+155T>A
NM_000092.4(*COL4A4*):c.4334-23A>G
NM_000091.4(*COL4A3*):c.2224-11C>T
NM_000091.4(*COL4A3*):c.4028-27A>G
NM_000091.4(*COL4A3*):c.4462+457C>G
NM_000091.4(*COL4A3*):c.4463-18dupA
NM_025243.3(*SLC19A3*):c.980-14A>G
NM_024120.4(*NDUFAF5*):c.223-907A>C
NM_000022.2(*ADA*):c.1079-15T>A
NM_000022.2(*ADA*):c.976-34G>A
NM_001178008.1(*CBS*):c.-86_-85+8delAGGTAGAAGA
NM_017424.2(*CECR1*):c.1082-1113delA
NM_015166.3(*MLC1*):c.895-226T>G
NM_015166.3(*MLC1*):c.-42C>T
NM_000487.5(*ARSA*):c.1108-12C>G
NM_000487.5(*ARSA*):c.1108-20A>G
NM_004628.4(*XPC*):c.*156G>A
NM_004628.4(*XPC*):c.413-24A>G
NM_000060.2(*BTD*):c.310-15delT
NM_000060.2(*BTD*):c.*159G>A
NM_000094.3(*COL7A1*):c.8620+26G>A
NM_000094.3(*COL7A1*):c.8305-12T>A
NM_000094.3(*COL7A1*):c.7929+11_7929+26delGATGGGGGCTGGGGGG
NM_000094.3(*COL7A1*):c.7759-18_7759-14delCATCTinsTTCA
NM_000094.3(*COL7A1*):c.5821-19A>G
NM_000094.3(*COL7A1*):c.5236-23A>G
NM_000094.3(*COL7A1*):c.2587+40G>A
NM_000094.3(*COL7A1*):c.977-15G>A
NM_000094.3(*COL7A1*):c.-187C>T
NM_000094.3(*COL7A1*):c.-188C>T

NM_000481.3(*AMT*):c.-55C>T
NM_000158.3(*GBE1*):c.2053-3358_2053-3350delGTGTGGTGGinsTGTTTTTTACATGACAGGT
NM_000187.3(*HGD*):c.650-17G>A
NM_000187.3(*HGD*):c.650-56G>A
NM_001178014.1(*PCCB*):c.714+462A>G
NM_032383.3(*HPS3*):c.2888-1612G>A
NM_001195794.1(*CLRN1*):c.254-649T>G
NM_003907.2(*EIF2B5*):c.685-13C>G
NM_153717.2(*EVC*):c.940-150T>G
NM_000320.2(*QDPR*):c.436+2552A>G
NM_000253.2(*MTTP*):c.619-5_619-2delTTTA
NM_000253.2(*MTTP*):c.1237-28A>G
NM_001184705.2(*HADH*):c.636+471G>T
NM_001184705.2(*HADH*):c.709+39C>G
NM_004453.2(*ETFDH*):c.-75A>G
NM_004453.2(*ETFDH*):c.176-636C>G
NM_000128.3(*F11*):c.-456G>A
NM_000128.3(*F11*):c.595+11A>G
NM_000128.3(*F11*):c.1304+12G>A
NM_002185.3(*IL7R*):c.379+288G>A
NM_000082.3(*ERCC8*):c.173+1119G>C
NM_000082.3(*ERCC8*):c.173+1046A>G
NM_022132.4(*MCCC2*):c.384-20A>G
NM_022132.4(*MCCC2*):c.1073-12C>G
NM_000521.3(*HEXB*):c.1243-17A>G
NM_000521.3(*HEXB*):c.1509-26G>A
NM_000521.3(*HEXB*):c.1613+15_1613+18dupAAGT
NM_000521.3(*HEXB*):c.1614-16_1615dupTTCATGTTATCTACAGAC
NM_000521.3(*HEXB*):c.1614-14C>A
NM_001199291.1(*HSD17B4*):c.1285-11C>G
NM_003060.3(*SLC22A5*):c.394-16T>A
NM_003060.3(*SLC22A5*):c.825-52G>A
NM_006846.3(*SPINK5*):c.283-12T>A
NM_006846.3(*SPINK5*):c.1431-12G>A
NM_006846.3(*SPINK5*):c.1820+53G>A
NM_000112.3(*SLC26A2*):c.-26+2T>C
NM_006261.4(*PROP1*):c.343-11C>G
NM_000500.7(*CYP21A2*):c.293-13C>G
NM_000287.3(*PEX6*):c.2301-15C>G
NM_000287.3(*PEX6*):c.2300+28G>A
NM_000255.3(*MUT*):c.-39-1G>A
NM_138694.3(*PKHD1*):c.8798-459C>A
NM_138694.3(*PKHD1*):c.7350+653A>G
NM_001142800.1(*EYS*):c.-448+5G>A
NM_020320.3(*RARS2*):c.613-3927C>T
NM_000426.3(*LAMA2*):c.3175-22G>A
NM_000426.3(*LAMA2*):c.3556-13T>A
NM_000426.3(*LAMA2*):c.5235-18G>A
NM_000426.3(*LAMA2*):c.8989-12C>G
NM_000045.3(*ARG1*):c.306-611T>C
NM_000288.3(*PEX7*):c.-45C>T
NM_000941.2(*POR*):c.-5+4A>G
NM_000441.1(*SLC26A4*):c.-103T>C
NM_000441.1(*SLC26A4*):c.-60A>G
NM_000441.1(*SLC26A4*):c.-4+1G>C

NM_000441.1(*SLC26A4*):c.-4+5G>A
NM_000441.1(*SLC26A4*):c.918+45_918+47delCAA
NM_000441.1(*SLC26A4*):c.1150-35_1150-28delTTTGTAGG
NM_000441.1(*SLC26A4*):c.1264-12T>A
NM_000441.1(*SLC26A4*):c.1438-7dupT
NM_000441.1(*SLC26A4*):c.1708-27_1708-11delTAAGTAACTTGACATTT
NM_000441.1(*SLC26A4*):c.2090-52_2090-49delCAAA
NM_000492.3(*CFTR*):c.-495C>T
chr7:g.117119797-117119797
NM_000492.3(*CFTR*):c.-249G>C
NM_000492.3(*CFTR*):c.-165G>A
NM_000492.3(*CFTR*):c.-85C>G
NM_000492.3(*CFTR*):c.-34C>T
NM_000492.3(*CFTR*):c.53+124T>C
NM_000492.3(*CFTR*):c.870-1113_870-1110delGAAT
NM_000492.3(*CFTR*):c.1117-26_1117-25delAT
NM_000492.3(*CFTR*):c.1393-18G>A
NM_000492.3(*CFTR*):c.1585-9412A>G
NM_000492.3(*CFTR*):c.1585-19T>C
NM_000492.3(*CFTR*):c.1679+34G>T
NM_000492.3(*CFTR*):c.1680-886A>G
NM_000492.3(*CFTR*):c.1680-883A>G
NM_000492.3(*CFTR*):c.1680-877G>T
NM_000492.3(*CFTR*):c.2908+19G>C
NM_000492.3(*CFTR*):c.2909-15T>G
NM_000492.3(*CFTR*):c.2988+33G>T
NM_000492.3(*CFTR*):c.3140-26A>G
NM_000492.3(*CFTR*):c.3140-16T>A
NM_000492.3(*CFTR*):c.3140-11A>G
NM_000492.3(*CFTR*):c.3469-1304C>G
NM_000492.3(*CFTR*):c.3717+40A>G
NM_000492.3(*CFTR*):c.3718-2477C>T
NM_000492.3(*CFTR*):c.3873+33A>G
NM_000492.3(*CFTR*):c.3874-4522A>G
NM_000492.3(*CFTR*):c.*1233T>A
NM_000083.2(*CLCN1*):c.-59C>A
NM_000083.2(*CLCN1*):c.1167-15_1167-14delCT
NM_001017420.2(*ESCO2*):c.1354-18G>A
NM_000349.2(*STAR*):c.466-11T>A
NM_152419.2(*HGSNAT*):c.821-28_821-10delTTGCTTATGCTTTGTACTT
NM_152416.3(*NDUFAF6*):c.298-768T>C
NM_152416.3(*NDUFAF6*):c.420+784C>T
NM_000497.3(*CYP11B1*):c.595+16G>T
NM_130849.3(*SLC39A4*):c.192+19G>A
NM_000155.3(*GALT*):c.-96T>G
NM_000155.3(*GALT*):c.83-11T>G
NM_000155.3(*GALT*):c.508-29delT
NM_000155.3(*GALT*):c.687+66T>A
NM_000155.3(*GALT*):c.820+13A>G
NM_000155.3(*GALT*):c.1059+56C>T
chr9:g.35658026-35658026
chr9:g.35658026-35658026
chr9:g.35658026-35658026
chr9:g.35658026-35658026
chr9:g.35658027-35658027

chr9:g.35658027-35658027
chr9:g.35658027-35658027
chr9:g.35658027-35658027
chr9:g.35658027-35658027
chr9:g.35658028-35658028
chr9:g.35658028-35658028
chr9:g.35658029-35658029
chr9:g.35658029-35658029
chr9:g.35658032-35658032
NM_016042.3(*EXOSC3*):c.475-12A>G
NM_000136.2(*FANCC*):c.-78-2A>G
NM_000136.2(*FANCC*):c.-79+1G>A
NM_000380.3(*XPA*):c.390-12A>G
NM_000035.3(*ALDOB*):c.*516T>A
NM_000035.3(*ALDOB*):c.-11+1G>C
chr9:g.104198194-104198194
NM_006731.2(*FKTN*):c.648-1243G>T
NM_000050.4(*ASS1*):c.-5-10C>G
NM_000050.4(*ASS1*):c.175-1119G>A
NM_000050.4(*ASS1*):c.773+49C>T
NM_007171.3(*POMT1*):c.-30-2A>G
NM_003172.3(*SURF1*):c.324-11T>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_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
NM_004006.2(*DMD*):c.531-16T>G
NM_004006.2(*DMD*):c.531-16T>A/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_000397.3(*CYBB*):c.-69A>C
chrX:g.37639262-37639262
NM_000397.3(*CYBB*):c.-67T>C
NM_000397.3(*CYBB*):c.-65C>T
NM_000397.3(*CYBB*):c.-64C>T
NM_000397.3(*CYBB*):c.46-14_46-11delTTCTinsGAA
NM_000397.3(*CYBB*):c.46-11T>G
NM_000397.3(*CYBB*):c.142-28_142-12delACTCTGCTCCCTTTCCC
NM_000397.3(*CYBB*):c.142-12delCinsACCTCTTCTAG
NM_000397.3(*CYBB*):c.483+978G>T
NM_000397.3(*CYBB*):c.674+1080A>G
NM_000397.3(*CYBB*):c.674+1337T>G
NM_000397.3(*CYBB*):c.675-1157A>G
NM_000397.3(*CYBB*):c.1152-11T>G
chrX:g.38128234-38128234
NM_001034853.1(*RPGR*):c.1059+363G>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_000044.3(*AR*):c.-547C>T
NM_000044.3(*AR*):c.1616+22072A>C
NM_000044.3(*AR*):c.1769-11T>A
NM_000044.3(*AR*):c.2450-118A>G
NM_000044.3(*AR*):c.2450-42G>A
NM_001399.4(*EDA*):c.707-13T>C/G
NM_001399.4(*EDA*):c.707-13T>C
NM_001399.4(*EDA*):c.707-13T>G
NM_000206.2(*IL2RG*):c.*307_*308delAA
NM_000206.2(*IL2RG*):c.*308A>G
NM_000206.2(*IL2RG*):c.270-15A>G
NM_000206.2(*IL2RG*):c.-105C>T
NM_001097642.2(*GJB1*):c.-16-576_-16-575insT
NM_001097642.2(*GJB1*):c.-16-524C>G
NM_001097642.2(*GJB1*):c.-16-513T>C/G
NM_001097642.2(*GJB1*):c.-16-513T>C

NM_001097642.2(*GJB1*):c.-16-513T>G
NM_001097642.2(*GJB1*):c.-16-511G>C
NM_000166.5(*GJB1*):c.-103C>T
NM_000166.5(*GJB1*):c.-17G>A
NM_000166.5(*GJB1*):c.-17+1G>T
NM_000166.5(*GJB1*):c.-17+2T>C
NM_001097642.2(*GJB1*):c.-16-3C>G
NM_001097642.2(*GJB1*):c.-16-2A>G
NM_001097642.2(*GJB1*):c.-16-1G>A
NM_001097642.2(*GJB1*):c.*15C>T
NM_000052.5(*ATP7A*):c.2916+2480T>G
NM_000052.5(*ATP7A*):c.3294+763C>G
NM_000390.2(*CHM*):c.315-1536A>G
NM_000390.2(*CHM*):c.315-4587T>A
chrX:g.85302626-85302626
chrX:g.85302634-85302634
chrX:g.85302634-85302634
chrX:g.85302644-85302644
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_000533.3(*PLP1*):c.4+78_4+85delGGGGGTTC
NM_000533.3(*PLP1*):c.453+28_453+46delTAACAAGGGGTGGGGGAAA
NM_000533.3(*PLP1*):c.454-322G>A
NM_000533.3(*PLP1*):c.454-314T>A/G
NM_000533.3(*PLP1*):c.454-314T>A
NM_000533.3(*PLP1*):c.454-314T>G
NM_033380.2(*COL4A5*):c.385-719G>A
NM_033380.2(*COL4A5*):c.466-12G>A
NM_033380.2(*COL4A5*):c.609+875G>T
NM_033380.2(*COL4A5*):c.646-12_646-11delTT
NM_033380.2(*COL4A5*):c.1423+57dupC
NM_033380.2(*COL4A5*):c.1424-20T>A
NM_033380.2(*COL4A5*):c.1948+894C>G
NM_033380.2(*COL4A5*):c.2042-18A>G
NM_033380.2(*COL4A5*):c.2245-40A>G
NM_033380.2(*COL4A5*):c.2245-14T>A
NM_033380.2(*COL4A5*):c.2395+2750A>G
NM_033380.2(*COL4A5*):c.3374-11C>A
NM_033380.2(*COL4A5*):c.4529-2300T>G
NM_033380.2(*COL4A5*):c.4529-345A>G
NM_033380.2(*COL4A5*):c.4821+121T>C
NM_033380.2(*COL4A5*):c.4822-152dupT
NM_033380.2(*COL4A5*):c.4822-151_4822-150insT
NM_004208.3(*AIFM1*):c.697-44T>G
NM_004208.3(*AIFM1*):c.-123G>C
NM_000194.2(*HPRT1*):c.27+47C>T
NM_000194.2(*HPRT1*):c.402+1229A>G
NM_000194.2(*HPRT1*):c.485+1202T>A
NM_000194.2(*HPRT1*):c.533-13T>G
NM_000074.2(*CD40LG*):c.289-32_289-25delAAAATGAC
NM_000074.2(*CD40LG*):c.289-15T>A
NM_000074.2(*CD40LG*):c.347-915A>T
NM_000133.3(*F9*):c.-55G>A/C/T

chrX:g.138612869-138612869
chrX:g.138612869-138612869
chrX:g.138612869-138612869
NM_000133.3(*F9*):c.-53A>G
chrX:g.138612871-138612871
NM_000133.3(*F9*):c.-52C>G/T
chrX:g.138612872-138612872
chrX:g.138612872-138612872
NM_000133.3(*F9*):c.-50T>C/G
chrX:g.138612874-138612874
chrX:g.138612874-138612874
NM_000133.3(*F9*):c.-49T>A/C
chrX:g.138612875-138612875
chrX:g.138612875-138612875
NM_000133.3(*F9*):c.-48G>C
NM_000133.3(*F9*):c.-38A>G
NM_000133.3(*F9*):c.-35G>A/C
chrX:g.138612889-138612889
chrX:g.138612889-138612889
NM_000133.3(*F9*):c.-34A>G/T
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>G/T
NM_000133.3(*F9*):c.-18A>T
NM_000133.3(*F9*):c.-18A>G
NM_000133.3(*F9*):c.-17A>C/G
NM_000133.3(*F9*):c.-17A>C
NM_000133.3(*F9*):c.-17A>G
NM_000133.3(*F9*):c.253-25A>G/T
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/T
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_000202.5(*IDS*):c.1181-15C>A
NM_006123.4(*IDS*):c.*57A>G
NM_000202.5(*IDS*):c.709-657G>A
NM_000252.2(*MTM1*):c.137-19_137-16delACTT
NM_000252.2(*MTM1*):c.137-11T>A
NM_000252.2(*MTM1*):c.232-26_232-23delGACT

NM_000252.2(*MTM1*):c.529-909A>G
NM_000252.2(*MTM1*):c.868-13T>A
NM_000425.4(*L1CAM*):c.3531-12G>A
NM_000425.4(*L1CAM*):c.2432-19A>C
NM_000425.4(*L1CAM*):c.1704-75G>T
NM_000425.4(*L1CAM*):c.1547-14delC
NM_000425.4(*L1CAM*):c.523+12C>T
NM_000117.2(*EMD*):c.266-27_266-10delTCTGCTACCGCTGCCCCC
NM_001363.3(*DKC1*):c.-142C>G
NM_001363.3(*DKC1*):c.-141C>G
NM_001363.3(*DKC1*):c.85-15T>C

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.