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***

Retinal Dystrophy Panel Plus

REFERRING HEALTHCARE PROFESSIONAL

NAME **HOSPITAL**

PATIENT

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

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

SUMMARY OF RESULTS

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

PRIMARY FINDINGS

The patient is heterozygous for *PRPF3* c.1510A>G, p.(Met504Val), which is a variant of uncertain significance (VUS).

PRIMARY FINDINGS: SEQUENCE ALTERATIONS

GENE	TRANSCRIPT	NOMENCLATURE	GENOTYPE	CONSEQUENCE	INHERITANCE	CLASSIFICATION
PRPF3	NM_004698.4	c.1510A>G, p.(Met504Val)	HET	missense_variant	AD	**Variant of uncertain significance (VUS)**
	ID	**ASSEMBLY**	**POS**	**REF/ALT**		
		GRCh37/hg19	1:150316721	A/G		
	gnomAD AC/AN	**POLYPHEN**	**SIFT**	**MUTTASTER**	**PHENOTYPE**	
	1/251420	possibly damaging	deleterious	disease causing	Retinitis pigmentosa	

SEQUENCING PERFORMANCE METRICS - NUCLEAR GENOME

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 20X	MEDIAN COVERAGE	PERCENT > 20X
Retinal Dystrophy Panel	314	5012	990736	990479	155	99.97

SEQUENCING PERFORMANCE METRICS - MITOCHONDRIAL GENOME

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 1000X	MEDIAN COVERAGE	PERCENT > 1000X
Mitochondrial genome	37	-	15358	15358	6463	100

TARGET REGION AND GENE LIST

The Blueprint Genetics Retinal Dystrophy Panel (version 8, Jul 09, 2025) Plus Analysis includes sequence analysis and copy number variation analysis of the following genes: *ABCA4*, *ABCC6**, *ABCD1**, *ABHD12*, *ACO2*, *ADAM9*, *ADAMTS18*, *ADGRV1*, *ADIPOR1**, *AGBL5*, *AHI1*, *AIPL1*, *ALMS1**, *AMACR*, *ARHGEF18*, *ARL13B*, *ARL2BP*, *ARL3*, *ARL6*, *ARMC9*, *ARR3*, *ARSG*, *ATF6*, *ATOH7*, *B9D1*, *B9D2*, *BBIP1*, *BBS1*, *BBS10*, *BBS12*, *BBS2*, *BBS4*, *BBS5*, *BBS7*, *BBS9*, *BEST1*, *C1QTNF5*, *C21ORF2*, *C2ORF71*, *C5ORF42*, *C8ORF37*, *CA4*, *CABP4*, *CACNA1F*, *CACNA2D4*, *CAPN5*, *CC2D2A*, *CDH23*, *CDH3*, *CDHR1*, *CEP104*, *CEP120*, *CEP164*, *CEP19*,

CEP250, *CEP290**, *CEP41*, *CEP78*, *CEP83*, *CERKL*, *CHM*[#], *CIB2*, *CISD2**, *CLN3*, *CLN5*, *CLN6*, *CLN8*, *CLRN1*, *CNGA1*[#], *CNGA3*, *CNGB1*, *CNGB3*, *CNNM4*, *COL11A1*, *COL11A2*, *COL18A1*, *COL2A1*, *COL9A1*, *COL9A2*, *COL9A3*, *COQ2*, *CPE*, *CRB1*, *CRX*, *CSPP1*, *CTC1*, *CTNNA1*, *CTNNB1*, *CTSD*, *CWC27*, *CYP4V2*, *DFNB31*, *DHDDS*, *DHX38*, *DNAJC5*, *DRAM2*, *DTHD1*, *DYNC2H1*, *EFEMP1*, *ELOVL4*, *EMC1*, *ESPN**, *EXOSC2*, *EYS**, *FAM161A*, *FDXR*, *FLVCR1*, *FRMD7*, *FZD4*, *GNAT1*, *GNAT2*, *GNB3*, *GNPTG*, *GPR143*, *GPR179*, *GRK1*,

GRM6, *GUCA1A*, *GUCY2D*, *HARS**, *HGSNAT*, *HK1*[#], *HMX1*, *IDH3A*, *IDH3B*, *IFT140*, *IFT172*, *IFT27*, *IFT81*[#], *IMPDH1*, *IMPG1*, *IMPG2*,

INPP5E, *INVS*, *IQCB1*, *ISPD*, *JAG1*, *KCNJ13*, *KCNV2*, *KIAA0556*, *KIAA0586*[#], *KIAA0753*, *KIAA1549*, *KIF11*, *KIF7*, *KIZ*, *KLHL7*, *LCA5*, *LRAT*, *LRIT3*, *LRP2*, *LRP5**, *LZTFL1*, *MAK*, *MERTK*, *MFN2*, *MFRP*, *MFSD8*, *MKKS*, *MKS1*, *MMACHC*, *MT-ATP6*, *MT-ATP8*, *MT-CO1*, *MT-CO2*, *MT-CO3*, *MT-CYB*, *MT-ND1*, *MT-ND2*, *MT-ND3*, *MT-ND4*, *MT-ND4L*, *MT-ND5*, *MT-ND6*, *MT-RNR1*, *MT-RNR2*, *MT-TA*, *MT-TC*, *MT-TD*, *MT-TE*, *MT-TF*, *MT-TG*, *MT-TH*, *MT-TI*, *MT-TK*, *MT-TL1*, *MT-TL2*, *MT-TM*, *MT-TN*, *MT-TP*, *MT-TQ*, *MT-TR*, *MT-TS1*, *MT-TS2*, *MT-TT*,

MT-TV, *MT-TW*, *MT-TY*, *MTTP*, *MVK*, *MYO7A*, *NAGLU*, *NDP*, *NEK2*[#], *NMNAT1*, *NPHP1*, *NPHP3*, *NPHP4*, *NR2E3*, *NR2F1*, *NRL*, *NYX*, *OAT*, *OCA2*, *OFD1*, *OPA1*, *OPA3*, *OPN1SW*, *OTX2*, *P3H2*, *PANK2*, *PAX2*, *PCDH15*, *PCYT1A*, *PDE6A*, *PDE6B*, *PDE6C*, *PDE6D*, *PDE6G*,

PDE6H, *PDSS1*[#], *PDSS2*, *PDZD7*, *PEX1*, *PEX10*, *PEX11B*, *PEX12*, *PEX13*, *PEX14*, *PEX16*, *PEX19*, *PEX2*, *PEX26*, *PEX3*, *PEX5*, *PEX6*, *PEX7*, *PHYH*, *PISD*, *PITPNM3*, *PLA2G5*, *PLK4*, *PNPLA6*, *POC1B*, *POMGNT1*, *PPT1*, *PRCD*, *PRDM13*, *PROM1*, *PRPF3*, *PRPF31*, *PRPF4*, *PRPF6*, *PRPF8*, *PRPH2*, *PRPS1**, *RAB28*, *RAX2*, *RBP3*, *RBP4*, *RCBTB1*, *RD3*, *RDH11*, *RDH12*, *RDH5*, *REEP6*, *RGR*, *RGS9*, *RGS9BP*,

RHO, *RIMS1*, *RLBP1*, *ROM1*, *RP1*, *RP1L1*, *RP2*, *RPE65*, *RPGR*, *RPGRIP1*, *RPGRIP1L*[#], *RS1*, *RTN4IP1*, *SAG*, *SAMD11*, *SCAPER*, *SCLT1*[#],

SDCCAG8, *SEMA4A*, *SGSH*, *SLC24A1*, *SLC25A46*, *SLC45A2*, *SLC7A14*, *SNRNP200*, *SPATA7*, *SPP2*, *SRD5A3**, *TCTN1*[#], *TCTN2*, *TCTN3*, *TEAD1*, *TIMM8A**, *TIMP3*, *TMEM107*, *TMEM126A*, *TMEM138*, *TMEM216*, *TMEM231*, *TMEM237*, *TMEM67*, *TOPORS*, *TPP1*, *TRAF3IP1*, *TREX1*, *TRIM32*, *TRPM1*, *TSPAN12*, *TTC21B*, *TTC8*, *TTLL5*, *TTPA*, *TUB*, *TUBB4B*, *TUBGCP4*, *TUBGCP6*, *TULP1*, *TYR**, *TYRP1*, *USH1C*, *USH1G*, *USH2A*, *VCAN*, *VPS13B*, *WDPCP*, *WDR19*, *WFS1*, *YME1L1**, *ZNF408*, *ZNF423* and *ZNF513*. The following exons are not included in the panel as they are not covered with sufficient high quality sequence reads: CHM (NM_001145414.4:5), CNGA1 (NM_001142564.2:2), HK1 (NM_001322365.2:5), IFT81 (NM_031473.4:12), KIAA0586 (NM_001244189.2:6,33), NEK2 (NM_001204182.2:8), PDSS1 (NM_014317.5:2), RPGRIP1L (NM_015272.5:23), SCLT1 (NM_001300898.2:6) and TCTN1 (NM_001173976.2:2;NM_024549.6:6).
*Some, or all, of the gene is duplicated in the genome. Read more: https://blueprintgenetics.com/pseudogene/

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

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

STATEMENT

CLINICAL HISTORY

Patient is a 25-year-old female with retinitis pigmentosa.

CLINICAL REPORT

Sequence analysis using the Blueprint Genetics (BpG) Retinal Dystrophy Panel identified a heterozygous missense variant *PRPF3* c.1510A>G, p.(Met504Val).

***PRPF3* c.1510A>G, p.(Met504Val)**

There is 1 individual heterozygous for this variant in gnomAD v2, a large reference population database (n>120,000 exomes and >15,000 genomes) which aims to exclude individuals with severe pediatric disease. The computational tool REVEL indicates this variant may be disease associated. To the best of our knowledge, this variant has not been reported in the medical literature or on disease-related variation databases.

PRPF3

The *PRPF3* (MIM *607301) gene encodes a component of a spliceosome complex that catalyzes the splicing of pre-mRNA in the nucleus. Pathogenic variants in *PRPF3* have been associated with autosomal dominant retinitis pigmentosa 18 (RP18; MIM #601414).

Retinitis pigmentosa (RP; MIM #268000) refers to a group of inherited disorders in which abnormalities of the photoreceptors (rods and cones) or the retinal pigment epithelium of the retina lead to progressive visual loss. Affected individuals first experience defective dark adaptation or “night blindness”, followed by constriction of peripheral visual fields and, eventually, loss of central vision late in the course of the disease (GeneReviews NBK1417). RP can be inherited in an autosomal dominant, autosomal recessive, or X-linked manner which partly correlates with the severity of the disease with X-linked cases having the most severe course, autosomal recessive cases having intermediate severity, and autosomal dominant cases the most favorable course. Prevalence of RP is reported to be 1/3,000 to 1/5,000 (ORPHA791). The association between the *PRPF3* gene and RP18 was first shown by Chakarova *et al.* who identified two different missense variants in two adjacent codons, *PRPF3* c.1480A>G, p.(Thr494Met) and *PRPF3* c.1477C>T, p.(Pro493Ser), in exon 11 of the *PRPF3* gene (PMID: 11773002). Subsequently, a third variant in the same exon, *PRPF3* c.1466C>A, p.(Ala489Asp), and two additional missense variants in the adjacent exons [*PRPF3* c.1345C>G, p.(Arg449Gly) in exon 10 and *PRPF3* c.1532A>C, p.(His511Pro) in exon 12] have been identified (PMID: 18412284, 27886254). In the family with the *PRPF3* c.1480A>G, p.(Thr494Met) variant, the affected individuals showed early onset of night blindness at age 4 to 10 years, with subsequent loss of far peripheral vision after 20 years (PMID: 27886254). All variants observed thus far are clustered in the C-terminal conserved region (amino acids 195–683), which is important for protein-protein interactions. Of the identified substitutions, the *PRPF3* c.1480A>G, p.(Thr494Met) is the most frequent and it has been shown to reduce Prpf3 phosphorylation and impair its association with Prpf4 and U4/U6 snRNP (PMID: 17932117).

There are currently 7 variants in *PRPF3* annotated as disease-causing (DM) in the HGMD Professional variant database (version 2025.1), all of which are missense variants reported in association with retinitis pigmentosa.

Mutation nomenclature is based on GenBank accession NM_004698.4 (*PRPF3*) with nucleotide one being the first nucleotide of the translation initiation codon ATG.

CONCLUSION

PRPF3 c.1510A>G, p.(Met504Val) is classified as a variant of uncertain significance (VUS), as there is insufficient evidence to evaluate its clinical relevance. This variant should not be used for clinical decision-making or risk evaluation in family members. Management of the patient and family should be based on clinical evaluation and judgment. Genetic counseling is recommended.

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

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

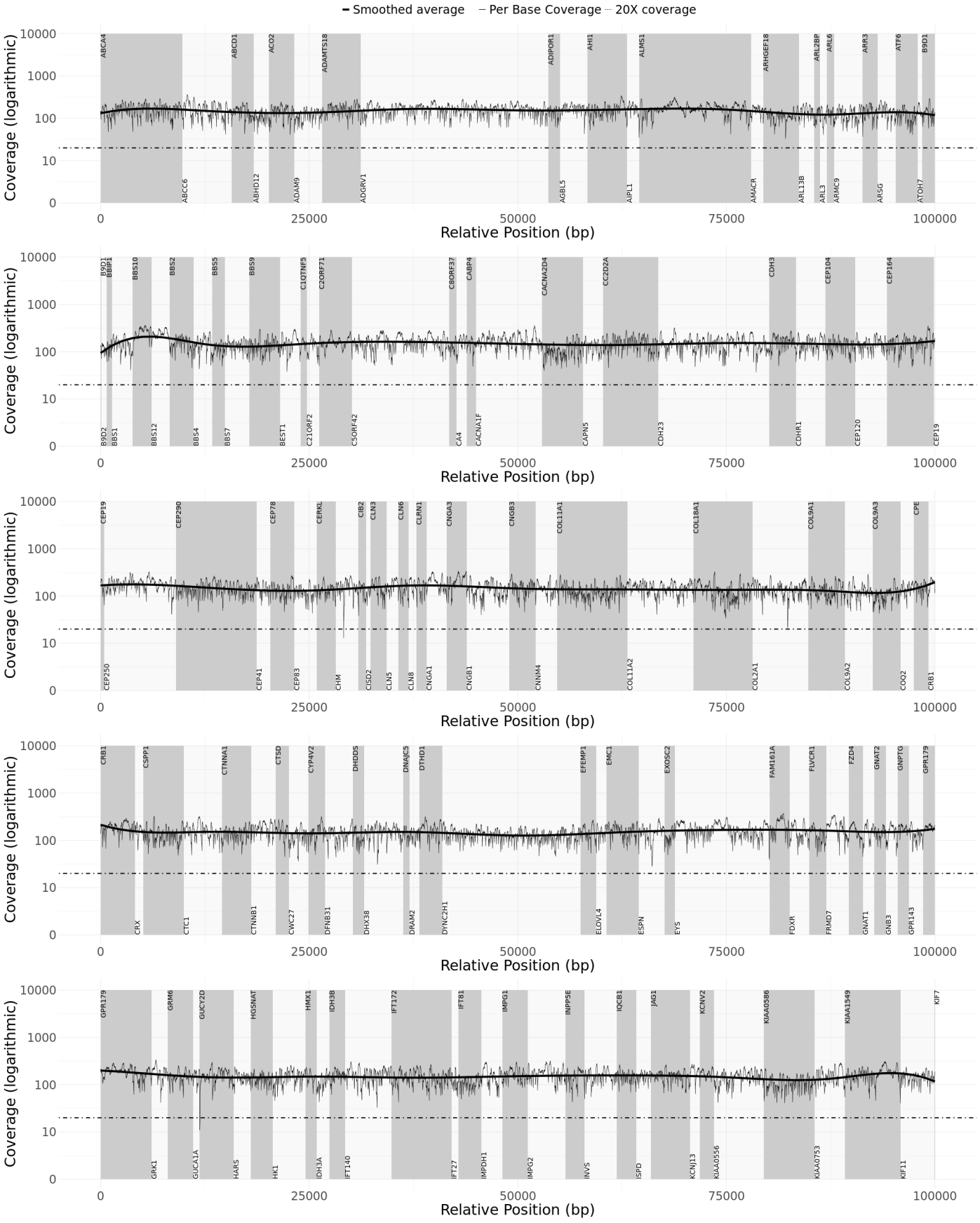
Signature

Name

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.


Smoothed average
Per Base Coverage
20X coverage
Coverage (logarithmic)
Relative Position (bp)
ABCA4
ABCC6
ABCD1
ABHD12
ACO2
ADAM9
ADAMTS18
ADGRV1
ADIPOR1
AGBL5
AHI1
AIPL1
ALMS1
AMACR
ARHGEF18
ARL13B
ARL2BP
ARL3
ARL6
ARMC9
ARR3
ARSG
ATF6
ATOH7
B9D1
B9D2
BBIP1
BBS1
BBS10
BBS12
BBS2
BBS4
BBS5
BBS7
BBS9
BEST1
C1QTNF5
C21ORF2
C2ORF71
C5ORF42
C8ORF37
CA4
CABP4
CACNA1F
CACNA2D4
CAPN5
CC2D2A
CDH23
CDH3
CDHR1
CEP104
CEP120
CEP164
CEP19
CEP250
CEP290
CEP41
CEP78
CEP83
CERKL
CHM
CIB2
CISD2
CLN3
CLN5
CLN6
CLN8
CLNN1
CNGA1
CNGA3
CNGB1
CNGB3
CNNM4
COL11A1
COL11A2
COL18A1
COL2A1
COL9A1
COL9A2
COL9A3
COQ2
CPE
CRB1
CRX
CSPP1
CTC1
CTNNA1
CTNNB1
CTSD
CWC27
CYP4V2
DFNB31
DHDDS
DHX38
DNAJC5
DRAM2
DTHD1
DYNC2H1
EFEMP1
ELOVL4
EMC1
ESPN
EXOSC2
EYS
FAM161A
FDXR
FLVCR1
FRMD7
FZD4
GNAT1
GNAT2
GNB3
GNPTG
GPR143
GPR179
GRK1
GRM6
GUCA1A
GUCY2D
HARS
HGSNAT
HK1
HMX1
IDH3A
IDH3B
IFT140
IFT172
IFT27
IFT81
IMPDH1
IMPG1
IMPG2
INPP5E
INVS
IQCB1
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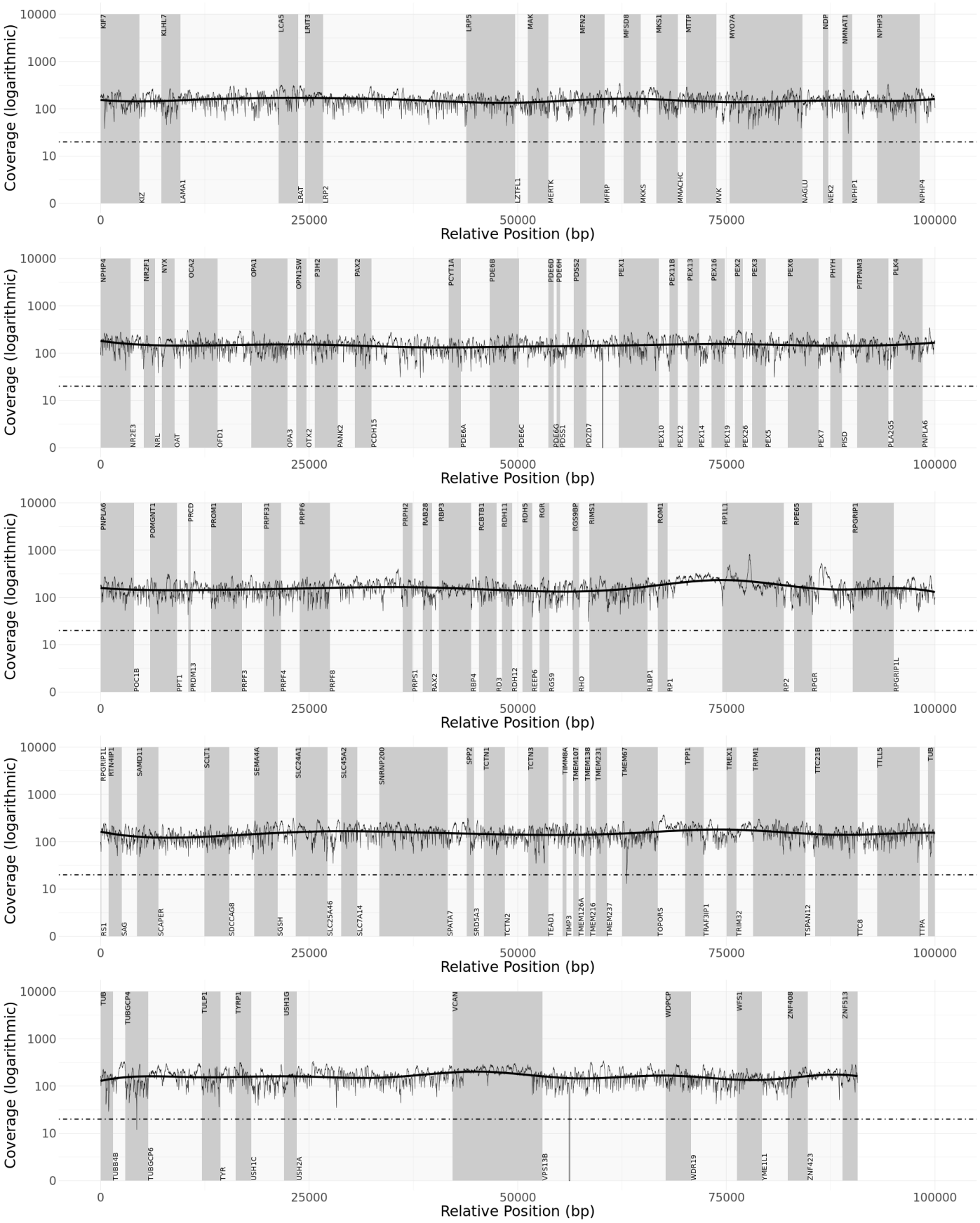
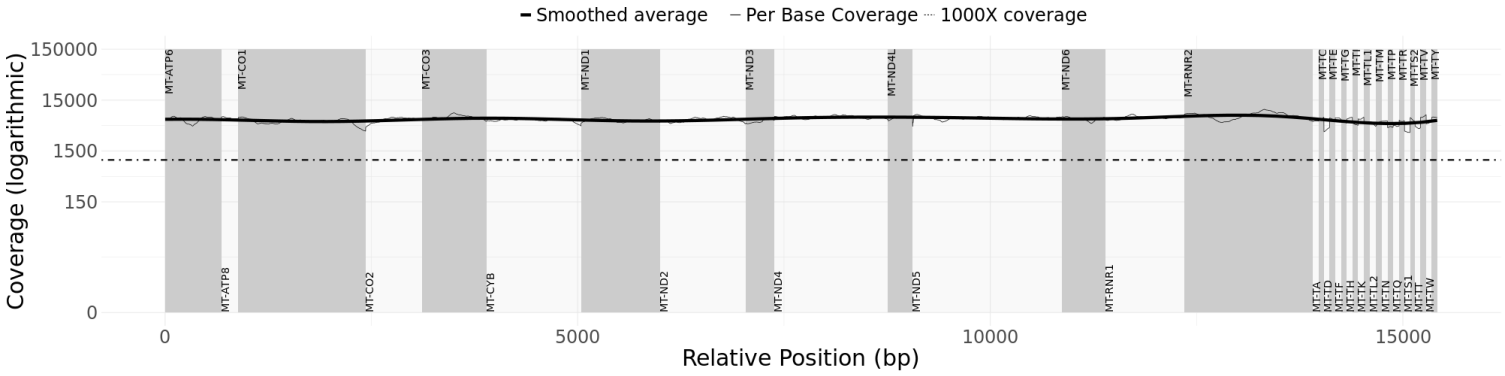
KIAA0586
KIAA0753
KIAA1549
KIF11
KIF7



COVERAGE PLOT- MITOCHONDRIAL 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.



APPENDIX 5: SUMMARY OF THE TEST

PLUS ANALYSIS

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

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

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

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

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

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

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

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

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

PERFORMING SITE:

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

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

NON-CODING VARIANTS COVERED BY THE PANEL:

NM_022787.3(*NMNAT1*):c.-70A>T
NM_022787.3(*NMNAT1*):c.-69C>T
NM_022787.3(*NMNAT1*):c.-57+7T>G
NM_024887.3(*DHDDS*):c.441-24A>G
NM_000310.3(*PPT1*):c.*526_*529delATCA
NM_000310.3(*PPT1*):c.125-15T>G
NM_000329.2(*RPE65*):c.246-11A>G
NM_000350.2(*ABCA4*):c.6730-19G>A
NM_000350.2(*ABCA4*):c.6148-471C>T
NM_000350.2(*ABCA4*):c.5197–557G>T

NM_000350.2(*ABCA4*):c.5196+1137G>A
NM_000350.2(*ABCA4*):c.5196+1137G>T
NM_000350.2(*ABCA4*):c.5196+1056A>G
NM_000350.2(*ABCA4*):c.4539+2065C>G
NM_000350.2(*ABCA4*):c.4539+2064C>T
NM_000350.2(*ABCA4*):c.4539+2028C>T
NM_000350.2(*ABCA4*):c.4539+2001G>A
NM_000350.2(*ABCA4*):c.4539+1928C>T
NM_000350.2(*ABCA4*):c.4539+1729G>T
NM_000350.2(*ABCA4*):c.4539 +1106C>T
NM_000350.2(*ABCA4*):c.4539+1100A>G
NM_000350.2(*ABCA4*):c.4253+43G>A
NM_000350.2(*ABCA4*):c.3191–11T>A
NM_000350.2(*ABCA4*):c.3051-16T>A
NM_000350.2(*ABCA4*):c.3050+370C>T
NM_000350.2(*ABCA4*):c.2919-383C>T
NM_000350.2(*ABCA4*):c.2160+584A>G
NM_000350.2(*ABCA4*):c.1938-619A>G
NM_000350.2(*ABCA4*):c.1937+435C>G
NM_000350.2(*ABCA4*):c.1937+13T>G
NM_000350.2(*ABCA4*):c.769–784C>T
NM_000350.2(*ABCA4*):c.768+3223C>T
NM_000350.2(*ABCA4*):c.570+1798A>G
NM_000350.2(*ABCA4*):c.302+68C>T
NM_000350.2(*ABCA4*):c.161–23T>G
NM_000350.2(*ABCA4*):c.67-16T>A
NM_080629.2(*COL11A1*):c.3744+437T>G
NM_080629.2(*COL11A1*):c.1027-24A>G
NM_080629.2(*COL11A1*):c.781-450T>G
NM_005272.3(*GNAT2*):c.461+24G>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_001142763.1(*PCDH15*):c.-29+1G>C
NM_033500.2(*HK1*):c.-390-3838G>C
NM_033500.2(*HK1*):c.-390-3818G>C
NM_033500.2(*HK1*):c.27+14901A>G
NM_006204.3(*PDE6C*):c.481-12T>A
NM_000391.3(*TPP1*):c.887-18A>G
NM_001139443.1(*BEST1*):c.-29+1G>T
NM_001139443.1(*BEST1*):c.-29+5G>A
NM_024649.4(*BBS1*):c.951+58C>T
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_002905.3(*RDH5*):c.-33+2dupT
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_000431.2(*MVK*):c.769-7dupT
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_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_017882.2(*CLN6*):c.297+113G>C
NM_033028.4(*BBS4*):c.77-216delA
NM_032520.4(*GNPTG*):c.610-16_609+28del
NM_014714.3(*IFT140*):c.2577+25G>A
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_001077416.2(*TMEM231*):c.824-11T>C
NM_000199.3(*SGSH*):c.249+27_249+28delGG
NM_015629.3(*PRPF31*):c.1073+20_1073+36delCGGTAGGCATGGGGGTC
NM_015629.3(*PRPF31*):c.1374+654C>G
NM_001298.2(*CNGA3*):c.-37-1G>C
NM_152384.2(*BBS5*):c.619-27T>G
NM_153638.2(*PANK2*):c.*40G>C
NM_000214.2(*JAG1*):c.1349-12T>G
NM_001271441.1(*C21ORF2*):c.1000-23A>T
NM_001195794.1(*CLRN1*):c.254-649T>G
NM_130837.2(*OPA1*):c.449-34dupA
NM_130837.2(*OPA1*):c.2179-40G>C
NM_006005.3(*WFS1*):c.-43G>T
NM_006017.2(*PROM1*):c.2077-521A>G
NM_000253.2(*MTTP*):c.619-5_619-2delTTTA
NM_000253.2(*MTTP*):c.1237-28A>G
NM_004744.3(*LRAT*):c.541-15T>G
chr5:g.33985176-33985176
chr5:g.33985764-33985764
NM_000287.3(*PEX6*):c.2301-15C>G
NM_000287.3(*PEX6*):c.2300+28G>A
NM_001142800.1(*EYS*):c.-448+5G>A
chr6:g.100040906-100040906
chr6:g.100040987-100040987

chr6:g.100041040-100041040
NM_021620.3(*PRDM13*):c.-8128A>C
NM_021620.3(*PRDM13*):c.-8107T>C
NM_000288.3(*PEX7*):c.-45C>T
NM_152419.2(*HGSNAT*):c.821-28_821-10delTTGCTTATGCTTTGTACTT
chr9:g.116037909-116037909
NM_000273.2(*GPR143*):c.885+748G>A
NM_000273.2(*GPR143*):c.659-131T>G
NM_003611.2(*OFD1*):c.935+706A>G
NM_003611.2(*OFD1*):c.1130-22_1130-19delAATT
NM_003611.2(*OFD1*):c.1130-20_1130-16delTTGGT
chrX:g.38128234-38128234
NM_001034853.1(*RPGR*):c.1059+363G>A
NM_000266.3(*NDP*):c.-207-1G>A
NM_000266.3(*NDP*):c.-208+5G>A
NM_000266.3(*NDP*):c.-208+2T>G
NM_000266.3(*NDP*):c.-208+1G>A
NM_000266.3(*NDP*):c.-343A>G
NM_000266.3(*NDP*):c.-391_-380delCTCTCTCTCCCTinsGTCTCTC
NM_000266.3(*NDP*):c.-396_-383delTCCCTCTCTCTCTC
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_004085.3(*TIMM8A*):c.133-23A>C
NM_194277.2(*FRMD7*):c.285-118C>T

GLOSSARY OF USED ABBREVIATIONS:

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