Blueprint Genetics

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

Comprehensive Epilepsy Panel

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

Negative

SEQUENCING PERFORMANCE METRICS - NUCLEAR GENOME

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 20X	MEDIAN COVERAGE	PERCENT > 20X
Comprehensive Epilepsy Panel	474	7161	1401467	1401002	164	99.97

SEQUENCING PERFORMANCE METRICS - MITOCHONDRIAL GENOME

PANEL	GENES	EXONS / REGIONS	BASES	BASES > 1000X	MEDIAN COVERAGE	PERCENT > 1000X
Mitochondrial genome	37	-	15358	15358	6606	100

TARGET REGION AND GENE LIST

The Blueprint Genetics Comprehensive Epilepsy Panel (version 8, Jul 09, 2025) Plus Analysis includes sequence analysis and copy number variation analysis of the following genes: *AARS*, *ABAT*, *ABCA2*, *ABCD1**, *ACTL6B*, *ACY1*, *ADAM22*, *ADAR*, *ADNP*, *ADPRHL2*, *ADSL*, *AFG3L2**, *AGA*, *AIFM1*, *AIMP1*, *ALDH3A2*, *ALDH5A1*[#], *ALDH7A1*, *ALG13*, *ALG6*, *ALKBH8*, *AMACR*, *AMT*, *ANKRD11**, *AP2M1*, *AP3B2*, *AP4B1*, *AP4E1*, *AP4M1*, *AP4S1**,[#], *APOPT1*, *ARG1*, *ARHGEF9*, *ARID1B*, *ARSA*, *ARV1*[#], *ARX*, *ASAH1*, *ASNS**, *ASPA*, *ASXL3*, *ATAD1**, *ATP13A2*, *ATP1A1*, *ATP1A2*, *ATP1A3*, *ATP6V1A*, *ATRX*, *BCKDK*, *BRAT1*, *BTD*, *C12ORF57*, *CACNA1A*, *CACNA1B*, *CACNA1D*, *CACNA1E*, *CACNA1G*, *CACNA1H*, *CACNA2D2*, *CACNB4*, *CAD*, *CAMK2B*, *CARS2*, *CASK*, *CASR*, *CC2D1A*, *CDK9*, *CDKL5*, *CERS1*, *CHD2*, *CHRNA2*, *CHRNA4*, *CHRNB2*, *CLCN2*, *CLCN4*, *CLN3*, *CLN5*, *CLN6*, *CLN8*, *CLTC*, *CNKSR2*, *CNPY3*, *CNTNAP2*, *COA7*, *COL4A1*, *COL4A2*, *COL4A3BP*, *COQ2*, *COQ4*, *COX15*, *COX6B1*, *CPLX1*, *CPT2*, *CSF1R*, *CSNK2B*, *CSTB*, *CTC1*, *CTSD*, *CTSF*, *CUL4B*, *CUX2*,

CYFIP2, *CYP27A1*, *D2HGDH*, *DARS*, *DARS2*, *DCX*, *DDC*, *DDX3X*, *DEAF1*[#], *DEGS1*[#], *DENND5A*, *DEPDC5*, *DHDDS*, *DHFR**, *DHPS*[#], *DIAPH1*, *DMXL2*, *DNAJC5*, *DNM1**, *DNM1L*, *DOCK7*, *DOLK*, *DPAGT1*, *DPM1*, *DPM2*, *DPYD*, *DPYS*, *DYNC1H1*, *DYRK1A*, *EARS2*, *ECHS1*, *ECM1*, *EEF1A2*, *EFHC1*, *EIF2B1*, *EIF2B2*, *EIF2B3*, *EIF2B4*, *EIF2B5*, *EIF3F*, *EML1*, *EPM2A*, *EPRS*, *ETFA*, *ETFB*, *ETFDH*, *ETHE1*, *FA2H*, *FAM126A*, *FAR1**, *FARS2*, *FDFT1*, *FDX1L*, *FGF12*, *FH*, *FKTN*, *FLNA*, *FOLR1*, *FOXG1*, *FOXRED1*, *FRRS1L*, *FUT8*, *GABBR2*, *GABRA1*,

GABRB1, *GABRB2*, *GABRB3*, *GABRG2*[#], *GALC*, *GAMT*, *GCDH*, *GCH1*, *GCSH*, *GFAP*, *GFM1*, *GFM2*[#], *GJC2*, *GLB1*, *GLDC*, *GLRB*, *GLS*, *GLUD1**, *GNAO1*, *GNB1*, *GNE*, *GOLGA2*, *GOSR2**, *GPAA1*, *GPHN*, *GRIA3*, *GRIA4*, *GRIK2*, *GRIN1*, *GRIN2A*, *GRIN2B*, *GRIN2D*, *GRN*, *GTPBP3*, *GUF1*, *HACE1*, *HCN1*, *HCN2**, *HECW2*, *HEPACAM*, *HIBCH*, *HNRNPU*, *HSD17B10*, *HSPD1**, *HTRA1*, *HTT*, *IBA57*, *ICK*, *IER3IP1*, *IFIH1*, *IQSEC2*, *IRF2BPL*, *ITPA*, *KCNA1*, *KCNA2*, *KCNB1*, *KCNC1*, *KCNH1*, *KCNJ10*, *KCNMA1*, *KCNQ2*, *KCNQ3*, *KCNQ5*, *KCNT1*, *KCNT2*,

KCTD3, *KCTD7*, *KDM5C*, *KIAA1715*[#], *KIAA2022*, *KIF1A*, *KIF5A*, *KIF5C*, *KMT2E*, *L2HGDH*, *LGI1*, *LIAS*, *LMNB1*, *LMNB2*, *LRPPRC*, *LYRM7*, *MACF1*, *MAGI2*, *MARS2*, *MBD5*, *MBOAT7*, *MDH2*, *MECP2*, *MED12*, *MED17*, *MEF2C*, *MFSD8*, *MIPEP**, *MLC1*, *MOCS1**, *MOCS2*, *MRPL44*, *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*, *MTFMT*, *MTHFR*, *MTOR*, *NACC1*, *NBEA**, *NDST1*, *NDUFAF3*, *NDUFAF5*, *NDUFAF6*, *NDUFS2*, *NDUFS4*, *NDUFS6*, *NDUFS7*, *NDUFS8*, *NDUFV1*, *NECAP1**, *NEU1*, *NEUROD2*, *NFU1*, *NHLRC1*,

NKX6-2, *NOTCH3*, *NPRL2*, *NPRL3*, *NR2F1*, *NRXN1*, *NSDHL*, *NT5C2*, *NTRK2*, *NUBPL*, *NUS1**, *OCLN**,[#], *OFD1*, *OPHN1*, *P4HTM*, *PACS1*, *PACS2*, *PAFAH1B1*, *PARS2*, *PCDH19*, *PEX1*, *PEX10*, *PEX12*, *PEX13*, *PEX14*, *PEX16*, *PEX19*, *PEX2*, *PEX26*, *PEX3*, *PEX5*, *PEX6*, *PGK1*, *PHACTR1*, *PHF6*, *PIGA**, *PIGB*, *PIGC**, *PIGG*, *PIGN**, *PIGO*, *PIGP*, *PIGQ*, *PIGS*, *PIGT*, *PIGV*, *PIGW*, *PITRM1*, *PLAA*, *PLCB1*, *PLP1*, *PNKP*, *PNPO*, *POLG*, *POLR3A*, *POLR3B*, *PPP2CA*, *PPP3CA*, *PPT1*, *PRICKLE1*, *PRICKLE2*, *PRIMA1*, *PRODH**, *PROSC*, *PRRT2*, *PRUNE*, *PSAP*, *PSAT1**, *PTPN23*, *PTS*, *PUM1*, *PURA*, *PYCR2*, *QARS*, *QDPR*, *RAB11A*, *RAB11B*, *RAB39B*, *RALA**, *RARS*, *RELN*, *RHOBTB2*, *RMND1**, *RNASEH2A*, *RNASEH2B*, *RNASEH2C*, *RNASET2*, *RNF13**, *RNF216**, *ROGDI*, *RORA*, *RORB*, *RUSC2*, *SAMHD1*, *SCARB2*, *SCN1A*, *SCN1B*, *SCN2A*, *SCN3A*, *SCN8A*, *SCN9A*, *SCO1*, *SDHAF1*, *SERAC1*, *SERPINI1*, *SETBP1*, *SETD1B*, *SGSH*, *SIK1*, *SLC12A5*, *SLC13A5*,

SLC19A3, *SLC1A2*, *SLC1A4*, *SLC25A1*, *SLC25A15**, *SLC25A22*, *SLC25A42*, *SLC2A1*, *SLC35A1*, *SLC35A2*, *SLC39A8*[#], *SLC46A1*, *SLC6A1*, *SLC6A5*, *SLC6A8**, *SLC9A6*, *SMARCA2*, *SMC1A*, *SMS*, *SNAP25*, *SNORD118*, *SOX10*, *SPATA5*, *SPTAN1*, *SPTBN4*, *SSR4*, *ST3GAL3*, *ST3GAL5*, *STRADA*, *STX1B*, *STXBP1*, *SUMF1*, *SUOX*, *SYN1*, *SYNGAP1*, *SYNJ1*, *SZT2*, *TAF1*, *TANGO2*, *TBC1D20*, *TBC1D24*,

TBCD, *TBCE*, *TBCK*, *TBL1XR1**, *TCF4*, *TK2*[#], *TPK1*, *TPP1*, *TRAK1*, *TREX1*, *TRIM8*, *TRIT1*, *TSC1*, *TSC2*, *TSFM*[#], *TTC19*, *TUBA1A**, *TUBB2A**, *TUBB2B**, *TUBB4A**, *UBA5**, *UBE2A*, *UBE3A**, *UBTF*, *UNC80*, *VAMP2*, *VARS*, *VPS13A*, *WARS2*, *WASF1*, *WDR26*, *WDR45*,

WWOX, *YWHAG*, *YY1*, *ZDHHC9*, *ZEB2**, *ZFYVE26*, *ZNHIT3*[#] and *ZSWIM6*. The following exons are not included in the panel as they are not covered with sufficient high quality sequence reads: ALDH5A1 (NM_170740.1:5), AP4S1 (NM_001254727.2:6), ARV1 (NM_001346992.2:4), DEAF1 (NM_001293634.1:9), DEGS1 (NM_001321541.2:3), DHPS (NM_001206974.2:1), GABRG2 (NM_198903.2:6), GFM2 (NM_001281302.2:2), KIAA1715 ((NM_001305009.1:10)), OCLN (NM_001205254.2:5-8), SLC39A8 (NM_001135148.2:1), TK2 (NM_001271934.2:3), TSFM (NM_001172696.2:5) and ZNHIT3 (NM_001281432.2:5).
*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 male with epilepsy.

CLINICAL REPORT

Sequence and Del/Dup (CNV) analysis using the Blueprint Genetics (BpG) Comprehensive Epilepsy Panel did not detect any known disease-causing or rare variants that could explain the patient's phenotype as described to the laboratory at the time of interpretation.

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.)

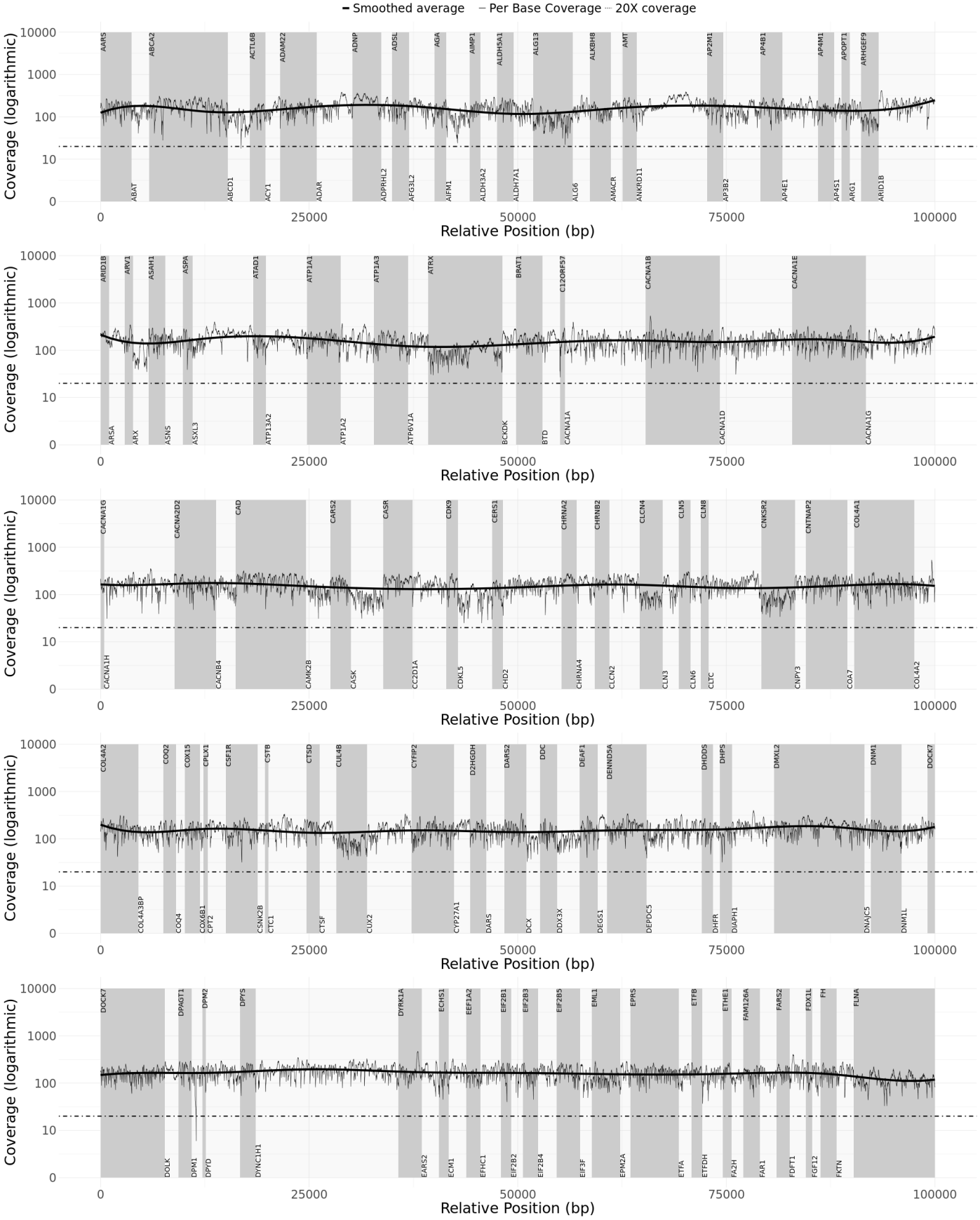
Signature

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)
AARS
ABCA2
ACTL6B
ADAM22
ADNP
ADSL
AGA
AIMP1
ALDH5A1
ALG13
ALKBH8
AMT
AP2M1
AP4B1
AP4M1
APOPT1
ARHGEF9
ABAT
ABCD1
ACY1
ADAR
ADPRHL2
AFG3L2
AIFM1
ALDH3A2
ALDH7A1
ALG6
AM4CR
ANKRD11
AP3B2
AP4E1
AP4S1
ARG1
ARID1B
ARID1B
ARV1
ASAH1
ASPA
ATAD1
ATP1A1
ATP1A3
ATRX
BRAT1
C12ORF57
CACNA1B
CACNA1E
ARSA
ARX
ASNS
ASXL3
ATP13A2
ATP1A2
ATP6V1A
BCKDK
BTD
CACNA1A
CACNA1D
CACNA1G
CACNA1G
CACNA2D2
CAD
CARS2
CASR
CDK9
CERS1
CHRNA2
CHRNB2
CLCN4
CLN5
CLN8
CNKSR2
CNTNAP2
COL4A1
CACNA1H
CACNB4
CAMK2B
CASK
CC2D1A
CDKL5
CHD2
CHRNA4
CLCN2
CLN3
CLN6
CLTC
CNPY3
COA7
COL4A2
COL4A2
COQ2
COX15
CPLX1
CSF1R
CSTB
CTSD
CUL4B
CYFIP2
D2HGDH
DARS2
DDC
DEAF1
DENND5A
DHDDS
DHPS
DMXL2
DNM1
DOCK7
COL4A3BP
COQ4
COX6B1
CPT2
CSNK2B
CTC1
CTSF
CUX2
CYP27A1
DARS
DCX
DDX3X
DEGS1
DEPDC5
DHFR
DIAPH1
DNAJC5
DNM1L
DOCK7
DPAGT1
DPM2
DPYS
DYRK1A
ECHS1
EEF1A2
EIF2B1
EIF2B3
EIF2B5
EML1
EPRS
ETFB
ETHE1
FAM126A
FARS2
FDXL
FH
FLNA
DOLK
DPM1
DPYD
DYNC1H1
EARS2
ECM1
EFHC1
EIF2B2
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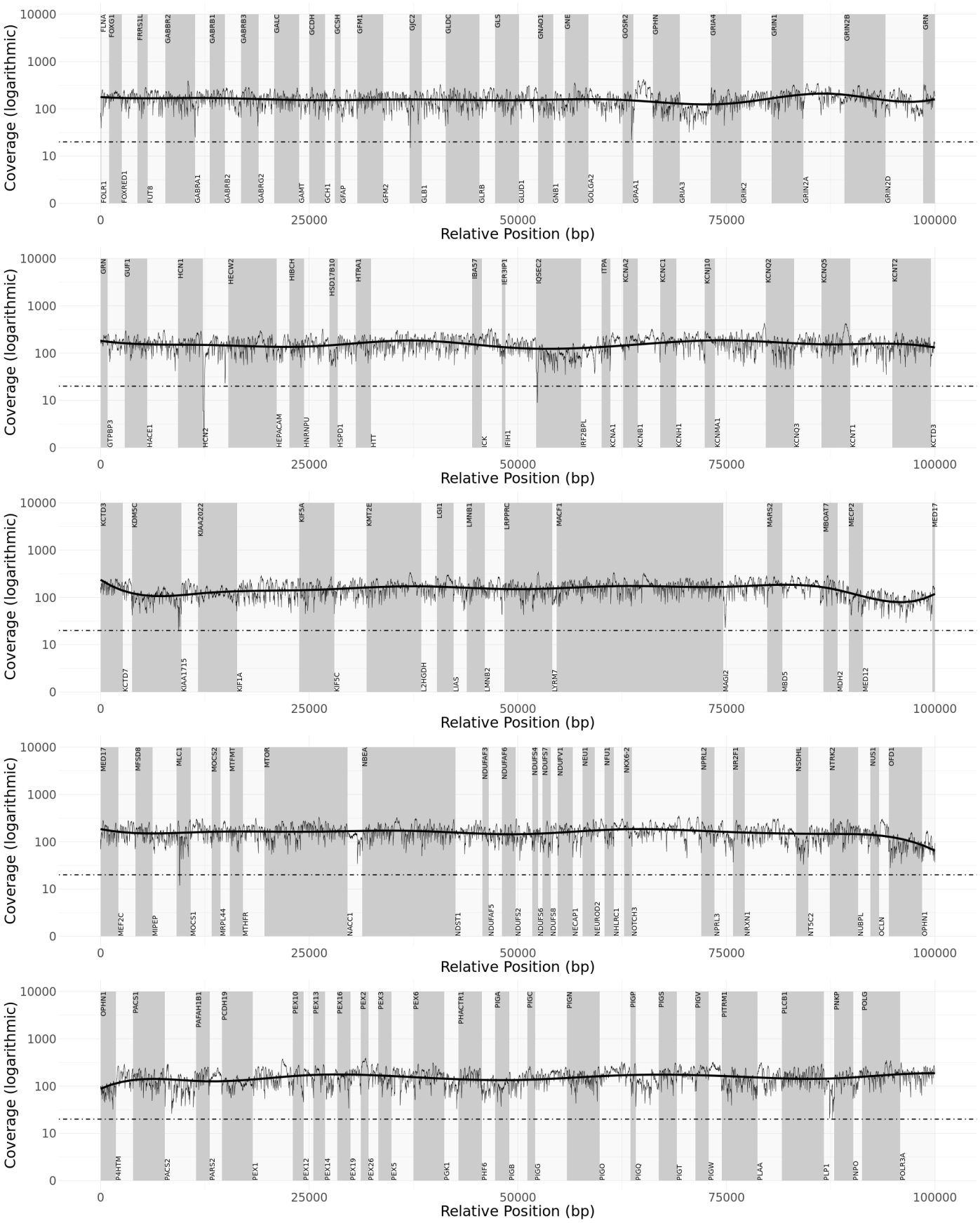
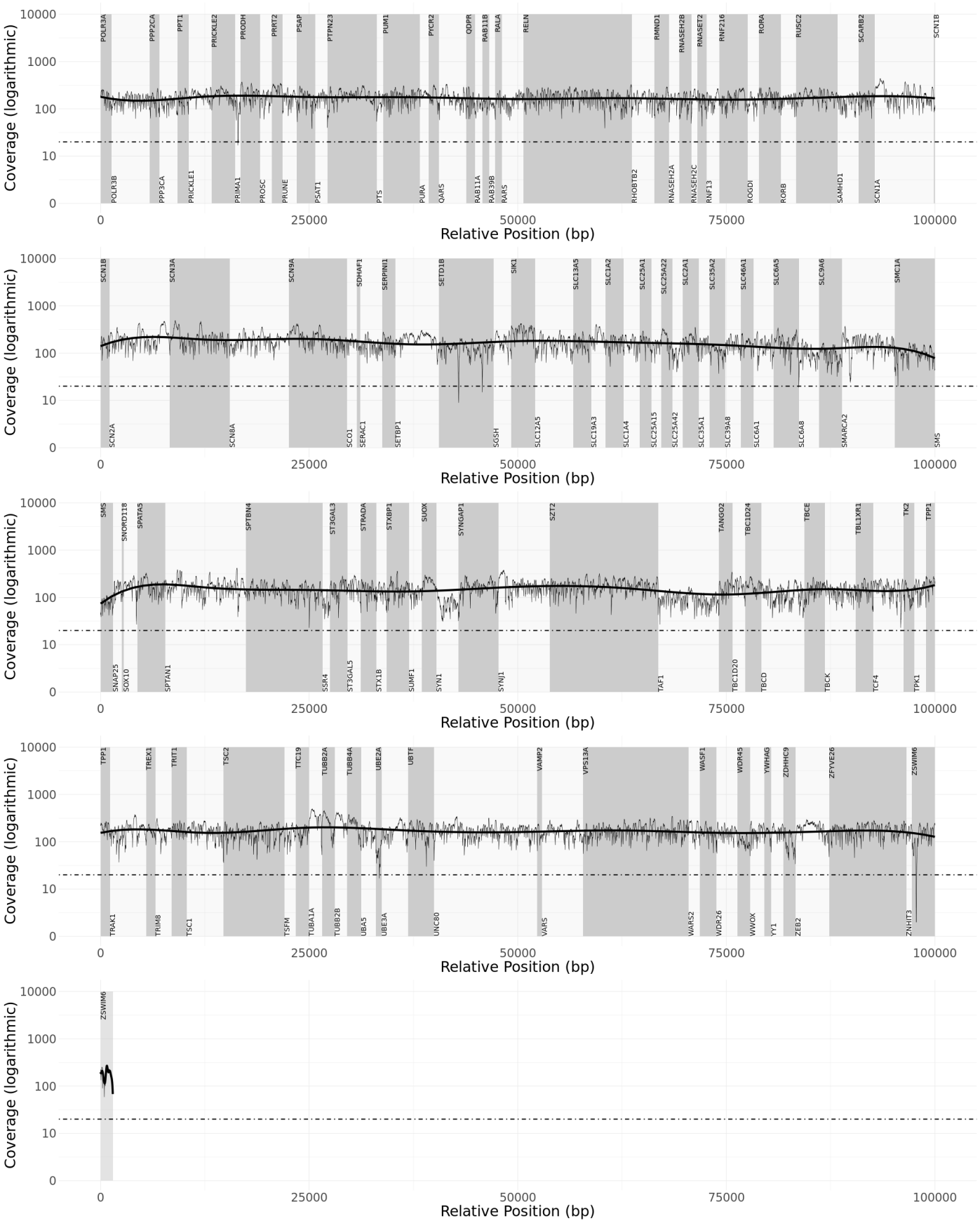
EIF3F
EPM2A
ETFA
ETFDH
FA2H
FAR1
FDFT1
FGF12
FKTN

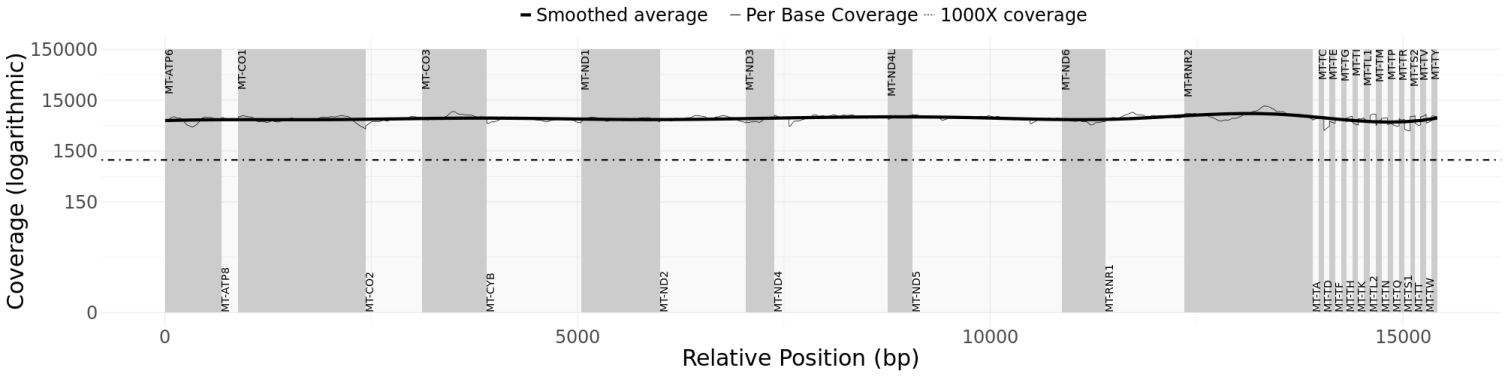

Coverage (logarithmic)
10000
1000
100
10
0
0
25000
50000
75000
100000
Relative Position (bp)
FLNA FOXG1 FRRS1L GABBR2 GABRB1 GABRB3 GALC GCDH GCSH GFM1 GJC2 GLDC GLS GNAO1 GNE GOSR2 GPHN GRIA4 GRIN1 GRIN2B GRN
FOLR1 FOXRED1 FUT8 GABRA1 GABRB2 GABRG2 GAMT GCH1 GFAP GFM2 GLB1 GLRB GLUD1 GNB1 GOLGA2 GPAA1 GRIA3 GRIK2 GRIN2A GRIN2D
GRN GUF1 HCN1 HECW2 HIBCH HSD17B10 HTRA1 IBA57 IER3IP1 IQSEC2 ITPA KCNA2 KCNC1 KCNJ10 KCNQ2 KCNQ5 KCNT2
GTPBP3 HACE1 HCN2 HEPACAM HNRNPU HSPD1 HTT ICK IFIH1 IRF2BPL KCNA1 KCNB1 KCNH1 KCNMA1 KCNQ3 KCNT1 KCTD3
KCTD3 KDM5C KIAA2022 KIF5A KMT2E LGI1 LMNB1 LRPPRC MACF1 MARS2 MBOAT7 MECP2 MED17
KCTD7 KIAA1715 KIF1A KIF5C L2HGDH LIAS LMNB2 LYRM7 MAGI2 MBD5 MDH2 MED12
MED17 MFSD8 MLC1 MOCS2 MTFMT MTOR NBEA NDUFAF3 NDUFAF6 NDUFS4 NDUFS7 NDUFV1 NEU1 NFU1 NKX6-2 NPRL2 NR2F1 NSDHL NTRK2 NUS1 OFD1
MEF2C MIPEP MOCS1 MRPL44 MTHFR NACC1 NDST1 NDUFAF5 NDUFS2 NDUFS6 NDUFS8 NECAP1 NEUROD2 NHLRC1 NOTCH3 NPRL3 NRXN1 NT5C2 NUBPL OCLN OPHN1
OPHN1 PACS1 PAFAH1B1 PCDH19 PEX10 PEX13 PEX16 PEX2 PEX3 PEX6 PHACTR1 PIGA PIGC PIGN PIGP PIGS PIGV PITRM1 PLCB1 PNKP POLG
P4HTM PACS2 PARS2 PEX1 PEX12 PEX14 PEX19 PEX26 PEX5 PGK1 PHF6 PIGB PIGG PIGO PIGQ PIGT PIGW PLAA PLP1 PNPO POLR3A



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_005957.4(*MTHFR*):c.1753-18G>A
NM_005957.4(*MTHFR*):c.-13-28_-13-27delCT
NM_024887.3(*DHDDS*):c.441-24A>G
NM_000310.3(*PPT1*):c.*526_*529delATCA
NM_000310.3(*PPT1*):c.125-15T>G
NM_006516.2(*SLC2A1*):c.680-11G>A
NM_006516.2(*SLC2A1*):c.-107G>A
NM_013339.3(*ALG6*):c.347-13C>G
NM_002241.4(*KCNJ10*):c.-1+1G>T
NM_018122.4(*DARS2*):c.228-22T>A

NM_018122.4(*DARS2*):c.228-22T>C
NM_018122.4(*DARS2*):c.228-21_228-20delTTinsC
NM_018122.4(*DARS2*):c.228-21_228-20delTTinsCC
NM_018122.4(*DARS2*):c.228-16C>A
NM_018122.4(*DARS2*):c.228-16C>G
NM_018122.4(*DARS2*):c.228-15C>G
NM_018122.4(*DARS2*):c.228-15C>A
NM_018122.4(*DARS2*):c.228-12C>A
NM_018122.4(*DARS2*):c.228-11C>G
NM_020435.3(*GJC2*):c.-170A>G
NM_020435.3(*GJC2*):c.-167A>G
NM_020435.3(*GJC2*):c.-20+1G>C
NM_001042465.1(*PSAP*):c.778-26C>A
NM_007055.3(*POLR3A*):c.*18C>T
NM_007055.3(*POLR3A*):c.3337-11T>C
NM_007055.3(*POLR3A*):c.1909+22G>A
NM_007055.3(*POLR3A*):c.1909+18G>A
NM_000391.3(*TPP1*):c.887-18A>G
NM_000317.2(*PTS*):c.84-323A>T
NM_000317.2(*PTS*):c.84-291A>G
NM_000317.2(*PTS*):c.187-38dupG
NM_018082.5(*POLR3B*):c.967-15A>G
NM_018082.5(*POLR3B*):c.1857-12A>G
NM_024570.3(*RNASEH2B*):c.65-13G>A
NM_024570.3(*RNASEH2B*):c.511-13G>A
NM_001845.4(*COL4A1*):c.*35C>A
NM_001845.4(*COL4A1*):c.*31G>T
NM_024884.2(*L2HGDH*):c.906+354G>A
NM_000161.2(*GCH1*):c.-22C>T
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_000814.5(*GABRB3*):c.-53G>T
NM_000814.5(*GABRB3*):c.-902A>T
NM_000814.5(*GABRB3*):c.-2204G>A
NM_000814.5(*GABRB3*):c.-2290T>C
NM_017882.2(*CLN6*):c.297+113G>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_024589.2(*ROGDI*):c.46-30_45+37delGGCGGGGC
NM_000086.2(*CLN3*):c.1056+34C>A
NM_000086.2(*CLN3*):c.461-13G>C
NM_001256443.1(*PRRT2*):c.*345G>A
chr17:g.8076761-8076761
chr17:g.8076761-8076761
chr17:g.8076762-8076762

NM_017775.3(*TTC19*):c.-42G>T
NM_001031806.1(*ALDH3A2*):c.681-14T>A
NM_001031806.1(*ALDH3A2*):c.681-14T>G
NM_002087.2(*GRN*):c.-9A>G
NM_002087.2(*GRN*):c.-8+3A>T
NM_002087.2(*GRN*):c.-8+3A>G
NM_002087.2(*GRN*):c.-8+5G>C
NM_000199.3(*SGSH*):c.249+27_249+28delGG
NM_005993.4(*TBCD*):c.1564-12C>G
NM_138924.2(*GAMT*):c.391+15G>T
NM_000159.3(*GCDH*):c.1244-11A>G
NM_001127221.1(*CACNA1A*):c.*1500_*1504dupCTTTT
NM_001127221.1(*CACNA1A*):c.5404-13G>A
chr19:g.13617793-13617793
NM_000435.2(*NOTCH3*):c.341-26_341-24delAAC
chr19:g.44031407-44031407
NM_007254.3(*PNKP*):c.1387-33_1386+49delCCTCCTCCCCTGACCCC
NM_014795.3(*ZEB2*):c.-69-1G>A
NM_014795.3(*ZEB2*):c.-69-2A>C
NM_006920.4(*SCN1A*):c.4820-14T>G
NM_006920.4(*SCN1A*):c.4306-14T>G
NM_006920.4(*SCN1A*):c.964+14T>G
NM_006920.4(*SCN1A*):c.474-13T>A
NM_025243.3(*SLC19A3*):c.980-14A>G
NM_152783.3(*D2HGDH*):c.293-23A>G
NM_024120.4(*NDUFAF5*):c.223-907A>C
NM_001242896.1(*DEPDC5*):c.-57G>C
NM_006941.3(*SOX10*):c.-31954C>T
NM_006941.3(*SOX10*):c.-32520C>G
NM_000026.2(*ADSL*):c.-49T>C
NM_015166.3(*MLC1*):c.-42C>T
NM_000487.5(*ARSA*):c.1108-12C>G
NM_000487.5(*ARSA*):c.1108-20A>G
NM_000060.2(*BTD*):c.310-15delT
NM_000481.3(*AMT*):c.-55C>T
NM_001178065.1(*CASR*):c.1378-19A>C
NM_003907.2(*EIF2B5*):c.685-13C>G
NM_021032.4(*FGF12*):c.*4722T>C
NM_000320.2(*QDPR*):c.436+2552A>G
NM_004453.2(*ETFDH*):c.-75A>G
NM_005211.3(*CSF1R*):c.1859-119G>A
NM_000806.5(*GABRA1*):c.-248+1G>T
NM_005943.5(*MOCS1*):c.*365_*366delAG
NM_005943.5(*MOCS1*):c.*7+6T>C
NM_005943.5(*MOCS1*):c.251-418delT
NM_000287.3(*PEX6*):c.2301-15C>G
NM_000287.3(*PEX6*):c.2300+28G>A
chr6:g.52284844-52284844
NM_000045.3(*ARG1*):c.306-611T>C
NM_032861.3(*SERAC1*):c.92-165C>T
NM_032861.3(*SERAC1*):c.92-239G>C

chr8:g.11660094-11660094
NM_004462.3(*FDFT1*):c.880-24_880-23delTCinsAG
NM_152416.3(*NDUFAF6*):c.298-768T>C
NM_152416.3(*NDUFAF6*):c.420+784C>T
NM_006731.2(*FKTN*):c.648-1243G>T
NM_001130438.2(*SPTAN1*):c.6690-17G>A
NM_000368.4(*TSC1*):c.363+668G>A
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
NM_003159.2(*CDKL5*):c.-162-2A>G
NM_007075.3(*WDR45*):c.236-18A>G
chrX:g.70749635-70749635
NM_000291.3(*PGK1*):c.1214-25T>G
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
NM_000533.3(*PLP1*):c.454-314T>G
NM_004208.3(*AIFM1*):c.697-44T>G
NM_004208.3(*AIFM1*):c.-123G>C
NM_015922.2(*NSDHL*):c.*129C>T
NM_001110556.1(*FLNA*):c.6023-27_6023-16delTGACTGACAGCC

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.