PATIENT INFORMATION

Name	:	[redacted]	Referred by	:	Dr. D A SAWANT
Age	:	38 Years	Sample ID	:	510100258
Gender	:	Female	Sample Collected	:	09-10-2025
Specimen	:	Peripheral Blood	Report Generated	:	07-11-2025

NEXT GENERATION SEQUENCING TEST

Nx GEN WHOLE EXOME SEQUENCING

CLINICAL DETAILS

[redacted] is a 38 Years old female. She is asymptomatic and undergoing broad genomic screening to assess risk for inherited conditions.
Comprehensive evaluation for all Pathogenic/Likely Pathogenic variants across the Exome including cardiac, neurological, metabolic, endocrine, hematological, and hereditary cancer-related predisposition.
Reporting of Carrier status for autosomal recessive and X-linked disorders.
Late-onset or multifactorial disease predispositions, where clinically relevant.

FAMILY HISTORY AND PEDIGREE

There is no mention of any significant history of any genetic conditions in the family.

RESULT SUMMARY

A broad genomic screening has not detected any variants as below.

INCIDENTAL VARIANT DETAILS (alterations of significance unrelated to the clinical phenotype)

GENE/ REFSEQ	COORDINATE (GRCh38)	VARIANT* (x)	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
			NONE				

CARRIER SCREENING FINDINGS

GENE/ REFSEQ	COORDINATE (GRCh38)	VARIANT* (89x; 96x; 47x)	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
HBA1 NM_000558.5	Chr16: 177308	c.326C>A p.Thr109Asn	Exon 3	Missense	Heterozygous AR	Thalassemias, alpha- (OMIM# 604131)	Likely Pathogenic
GUF1 NM_021927.3	Chr4: 44685998	c.709C>T p.Gln237*	Exon 7	Nonsense	Heterozygous AR	?Developmental and epileptic encephalopathy 40 (OMIM# 617065)	Likely Pathogenic
CFAP52 NM_145054.5	Chr17: 9638652	c.1516C>T p.Gln506*	Exon 12	Nonsense	Heterozygous AR	Heterotaxy, visceral, 10, autosomal, with male infertility (OMIM# 619607)	Likely Pathogenic

Heterozygous likely pathogenic variants in HBA1, GUF1 and CFAP52 genes (Autosomal recessive condition) may not have clinical impact for the individual but can be relevant in the reproductive setting. Partner testing may be considered if clinically indicated for reproductive risk assessment. Genetic counseling can be offered to the individual/ family.

RECOMMENDATION:

HBA1: While most carriers may remain clinically asymptomatic, there is evidence that heterozygous individuals can manifest hematological changes such as mild anemia, reticulocytosis, or features of hemoglobin instability. Correlation with clinical findings and a detailed hematological evaluation (complete blood counts, red cell indices, peripheral smear, hemoglobin electrophoresis/HPLC, and screening for Heinz bodies) is advised.

ANALYSIS STATISTICS

Exome Coverage at ≥20x	**98.98 %**	Exome Coverage at ≥50x	**90.36 %**
Target genes/regions Coverage at 20x	**99.56** %	Target genes/regions Coverage at 50x	**93.24 %**
Total Reads generated (millions)	**58**	Total Reads Aligned	**99.33** %

Exome coverage refers to the percentage of protein coding genes covered by the sequencing at a depth of 20x and 50x. Target genes/regions coverage refers to the percentage of the defined target region of the gene relevant to the given phenotype where the read depth was at least 20x to permit high quality exome variant base calling, annotation and evaluation. Average Quality threshold may range from 90 to 95% of the targeted region, indicating a small portion of the target region may not be covered with optimal depth or quality to call the variant positions confidently. Variant Read Depth refers to the count of reads covering the base called. Variant Allele Frequency refers to the percentage of sequence reads observed matching the sample specific DNA variant divided by the overall coverage at that locus, including the Reference bases.

SECONDARY FINDINGS#

Negative for secondary findings according to the ACMG (v3.2).

GENE	COORDINATE (GRCh38)	VARIANT*	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
			NONE				

[#]Secondary findings are genetic test results that provide information about changes (variants) in a gene unrelated to the primary clinical finding of the patient. As per recommendations by the American College of Medical Genetics and Genomics (ACMG, 2013 and 2020) secondary findings, in addition to any variants that are found related to the primary purpose of the testing are to be reported. The ACMG proposed list of 83 genes, which are associated with a variety of conditions, from cancer to heart disease, and are associated with conditions that have a definable set of clinical features, the possibility of early diagnosis, a reliable clinical genetic test, and effective intervention or treatment, are reported. Variants of unknown significance, whose involvement in disease at the current time is unclear, are reported based on the strength of Phenotype-Genotype correlation.

GENERAL RECOMMENDATIONS

- Genetic Counselling is strongly recommended to discuss the implications of this test result.
- Test results must be interpreted in the context of this individual's clinical history by a Qualified Medical Practitioner.

TEST METHODOLOGY

Genomic DNA was extracted from the sample submitted and libraries were prepared using Twist exome 2.0 kit. Target exonic regions (GRCh38) were captured using standard hybridization-based target enrichment protocol. The libraries were sequenced at a mean coverage of >90x on the Illumina NovaSeqX Plus sequencing platform. Variant calling was performed using DRAGEN (Dynamic Read Analysis for GENomics) pipeline, version v4.3.13.

*VARIANT CLASSIFICATION (BASED ON ACMG RECOMMENDATIONS)

Pathogenic	A genetic variant that causes, increases or contributes to an individual's disease or disorder.
Likely pathogenic	A genetic variant is most likely responsible for causing disease or disorder, but need additional scientific evidence to be certain.
Variant of uncertain significance (VUS)	A variant that has unknown effect in the development of disease or disorder and not be enough scientific evidence to confirm or refute a disease association or the study may be inconsistent.
Likely benign	A variant is not responsible, expected, or probable to major cause disease, but need additional scientific evidence to be certain.
Benign	A variant is not a cause / responsible for a disease or disorder.

VARIANT CALLING AND PRIORITIZATION

All disease-causing variants reported in OMIM and ClinVar, as well as all variants with minor allele frequency (MAF) below 0.05 in ExAC, 1000 GENOMES and gnomAD database are considered. The investigation for relevant variants is focused on coding exons, splice sites (up to 10bp flanking regions) and UTR regions as well. All potential modes of inheritance patterns are considered. The effect of the variants on the proteins is calculated using the *In silico* prediction parameters using multiple algorithms such as REVEL, MetaLR, BLOSUM, PolyPhen-2, SIFT, MutationTaster, Mutation Assessor, and many others. In addition, provided family history and clinical information may be used to evaluate identified variants with respect to their pathogenicity and causality, and are categorized into classes 1 – 5 according to ACMG guidelines. All variants related to the

phenotype of the patient, except benign or likely benign variants, are reported.

TEST INFORMATION/LIMITATIONS

Exome sequencing is a targeted sequencing approach that is restricted to the protein-coding regions of genomes. The human exome is estimated to encompass approximately 1% of the genome, yet contains approximately 85% of disease-causing mutations. For genetic researchers trying to identify the genes implicated in over 6,800 rare diseases, exome sequencing enables rapid, cost-effective identification of common single nucleotide variants (SNVs) and small insertions or deletions (indels), as well as rare de novo mutations that may explain the heritability of Mendelian and complex disorders. The results are interpreted in the context of the provided clinical findings, family history, and other laboratory data. Only variants in genes potentially related to the proband's medical condition are reported. The test should not be used for detection of complex genetic events such as copy number variations (CNVs), inversions, translocations and for analysis of sequence repeats or for diagnosis of disorders caused by mutations in the mitochondrial DNA. In addition, due to technology limitations, certain regions may be either not or poorly covered. In these regions variants cannot be confidently detected. Extremely low coverage calls (homo/hemizygous or heterozygous calls with less than three or four reads, respectively) are expected to be artifacts based on our extensive validations and consequently are not considered during the analysis. Misinterpretation of results may occur if the provided information is inaccurate and/or incomplete. If the obtained genetic results do not concur with the clinical findings, additional testing should be considered.

GENES OF INTEREST

Gene	Coverage1X	Coverage20X	Coverage50X	*Gene*	Coverage1X	Coverage20X	Coverage50X
AATF	100	100	95.61	*MCOLN1*	100	100	84.91
ABCA13	100	100	98.81	*MECP2*	100	99.83	66.95
ABCA2	100	99.13	81.27	*MEGF6*	100	100	82.94
ABCA8	100	100	100	*MFSD9*	100	100	84.43
ABCC11	100	100	97.43	*MGAT5B*	100	100	88.94
ABCC8	100	100	86.06	*MIB2*	100	96.95	56.54
ACAD10	100	100	93.53	*MICALL1*	100	99.14	78.52
ACAD9	100	100	98.36	*MICOS13*	100	100	57.58
ACIN1	100	100	93.76	*MID1*	100	100	98.83
ACOT11	100	100	91.6	*MISP*	100	100	63.63
ACOT9	100	100	97.15	*MKKS*	100	100	98.07
ACSS2	100	100	94.81	*MKRN1*	100	98.99	75.06
ACTR8	100	100	94.83	*MLH3*	100	100	99.11
ACTRT2	100	100	68.61	*MMRN1*	100	100	99.92
ADAMTS12	100	100	98.33	*MRGPRE*	100	100	66.13
ADAMTS9	100	100	98.06	*MRPS9*	100	100	96.36
ADGRE5	100	100	88.53	*MST1R*	100	100	79.25
AGL	100	100	99.87	*MT1M*	100	100	100
AHNAK2	100	100	95.22	*MTHFD1L*	100	94.04	89.29
AKAP13	100	100	95.93	*MTMR4*	100	100	93.67
ALKBH8	100	100	100	*MUC1*	100	98.92	88.65
ANKK1	100	100	91.84	*MUC16*	100	100	97.87
ANKRD20A1	100	94.68	87.77	*MUC2*	100	99.61	87.65
ANKRD36C	100	99.74	98.63	*MUC3A*	100	100	98.84
AOPEP	100	97.51	88.88	*MUC5B*	100	100	94.08
APP	100	99.83	99.5	*MUC6*	100	100	90.38
AR	100	99.91	82.11	*MYO16*	100	100	84.49
ARAP2	100	100	99.62	*MYOM2*	100	100	97.11
ARFGAP2	100	100	92.03	*MYOM3*	100	100	91.24
ARHGEF4	100	96.69	74.72	*N4BP2*	100	99.55	92.39
ARID1B	98.21	95.07	74.8	*NAGK*	100	100	99.83
ARID4B	100	100	99.56	*NANOG*	100	100	100
ARMH1	100	100	83.08	*NBPF10*	100	100	99.57

ARPP21	100	100	96.18	*NBPF20*	100	99.83	97.82
ASB15	100	100	98.73	*NBPF26*	98.44	98.44	95.76
ASGR2	100	100	91.65	*NBPF8*	100	100	99.12
ASTN1	100	100	87.58	*NDUFV3*	100	100	94.55
ASXL2	100	100	98.74	*NEB*	100	100	99.3
ATOH8	100	100	58.01	*NEDD4*	100	100	98.39
ATP10A	100	97.49	87.54	*NEMP1*	100	100	94.65
ATP1A4	100	99.67	86.47	*NFASC*	100	99.98	91.79
ATP6V0D1	100	100	86.5	*NFRKB*	100	100	92.88
ATXN3	100	96.66	95	*NHSL1*	100	99.82	87.12
B4GALT2	100	100	90.8	*NIBAN1*	100	100	96.67
BAG4	100	100	92.99	*NINL*	100	100	91.64
BAHCC1	100	97.94	76.61	*NISCH*	100	100	88.89
BAMBI	100	100	90.29	*NME1*	100	100	100
BCL7C	100	100	66.01	*NOP16*	100	100	92.71
BCORL1	100	100	93.57	*NOTCH4*	100	99.68	89.32
BOD1L1	100	98.99	97.38	*NPIPB13*	100	99.58	97.19
BRPF1	100	100	89.21	*NR3C2*	100	100	95.83
BYSL	100	100	91.64	*NRG1*	100	94.6	82.31
C10orf62	100	100	80.06	*NRIP1*	100	100	99.83
CACNB2	100	100	98.5	*NTSR1*	100	100	76.92
CAND2	100	100	91.12	*NUP98*	100	100	92.77
CASP10	100	100	94.61	*NUTM2D*	100	100	100
CATSPER4	100	100	90.98	*NWD1*	100	100	77.97
CCDC124	100	100	75.6	*OBSCN*	100	99.31	79.96
CCDC170	100	100	92.09	*OPRM1*	100	100	98.93
CCDC187	100	99.8	81.72	*OR10Z1*	100	100	87.28
CCDC197	100	100	71.26	*OR1A1*	100	100	100
CCDC78	100	100	87.17	*OR2L5*	100	100	100
CCDC85C	100	88.57	46.22	*OR2T2*	100	100	100
CCSER1	100	100	98.57	*OR2T6*	100	100	100
CCT6B	100	100	100	*OR4A5*	100	100	100
CD48	100	100	93.05	*OR4F5*	100	100	100
CDC14A	100	100	94.2	*OR4K15*	100	100	100
CDC42BPG	100	100	85.02	*OR51A2*	100	100	97.24
CDK14	100	100	97.85	*OR51B5*	100	100	92.54
CEP126	100	100	95.52	*OR51M1*	100	100	100
CEP131	100	100	87.66	*OR7D4*	100	100	100
CEP192	100	100	99.09	*OR8B2*	100	100	100
CEP85L	100	100	99.82	*OR9K2*	100	100	100
CEP89	100	100	98.6	*OVOL1*	100	100	63.06
CFAP52	100	100	97.24	*PACC1*	100	100	82.89
CFAP54	100	100	98.72	*PADI6*	100	100	90.02
CFAP65	100	100	82.24	*PAG1*	100	100	82.76
CGREF1	100	100	94.5	*PAN2*	100	100	87.53
CHRNA1	100	100	97.97	*PAOX*	100	100	69.18
CKAP5	100	100	98.52	*PAPPA2*	100	100	92.8
CNTLN	100	100	99.5	*PAX6*	100	100	90.79
CNTN2	100	100	91.91	*PCLO*	100	100	99.21
CNTNAP3B	100	100	99.44	*PCNX3*	100	100	88.23
COL11A2	100	100	84.32	*PDGFRB*	100	100	91.75
COL18A1	100	100	82.91	*PDZD2*	100	100	85.82
COL6A2	100	100	89.36	*PDZD4*	100	100	81.18
CPEB2	100	93.95	65.76	*PEX1*	100	100	99.49
CRYBB3	100	100	99.38	*PGAM1*	100	100	100
CRYBG2	100	98.25	80.67	*PHLPP2*	100	100	90.24
CRYBG3	100	100	99.91	*PIDD1*	100	100	77.62
CSAG2	100	100	100	*PKHD1*	100	100	95.39
CSF2RB	100	100	79.61	*PKM*	100	100	85.99
CSMD2	100	98.46	92.52	*PLCG2*	100	100	96.15
CT45A6	100	100	100	*PODN*	100	100	84.08
CT47A7	89.97	46.25	32.99	*POLK*	100	100	100
CXADR	100	99.49	84.32	*POLR3B*	100	100	96.31
CYB5R2	100	100	99.27	*POTEG*	100	100	100
CYP1A2	100	100	75.77	*PPP1R14D*	100	100	84.08
CYP2A13	100	100	99.53	*PPWD1*	100	100	97.67

DCAF8L2	100	100	100	*PRAG1*	100	99.09	81.12
DCBLD1	100	95.94	87.49	*PRAMEF7*	100	100	99.65
DDHD1	100	96.22	85.65	*PRELID3B*	100	100	100
DEGS1	100	100	98.69	*PRR29*	100	100	91.11
DENND2B	100	100	86.43	*PRR5L*	100	100	91.79
DGAT1	100	94.8	55.13	*PRSS21*	100	100	83.85
DHCR24	100	100	83.26	*PSD4*	100	100	89.66
DHX37	100	100	84.78	*PSTK*	100	100	93.58
DKK3	100	100	79.11	*PYGM*	100	100	86.34
DLEU7	100	100	72.75	*QSER1*	100	97.05	96.09
DLG5	100	100	90.05	*RANGRF*	100	100	98.77
DNAH17	100	100	87.15	*RASA4B*	100	100	96.05
DNAH3	100	100	94.76	*RB1*	100	99.89	96.97
DNAH7	100	100	99.43	*RBBP8*	100	100	99.89
DNAH8	100	100	99	*RBM26*	100	100	98.9
DNHD1	100	99.97	85.76	*RCCD1*	100	100	96.37
DNMBP	100	100	98.07	*REPS2*	100	91.61	87.11
DNTT	100	100	99.28	*RESF1*	100	100	100
DOC2B	100	88.91	67.47	*RETSAT*	100	100	86.84
DOCK5	100	100	97.76	*REV1*	100	100	99.25
DOCK6	100	100	88.99	*RGS11*	100	96.59	90.59
DOK1	100	100	88.75	*RGS22*	100	100	98.42
DOK3	100	100	79.98	*RHCE*	100	100	93.53
DPY19L4	100	100	90.67	*RHOBTB3*	100	100	95.54
DQX1	100	100	82.65	*RIC8B*	100	100	96.71
DST	100	99.93	98.12	*RIMBP2*	100	100	95.66
DZANK1	100	99.96	95	*RIMBP3B*	100	100	91.79
EARS2	100	100	95.38	*RIMBP3C*	100	100	92.99
EFCAB9	100	100	100	*RNF10*	100	100	91
EFTUD2	100	100	94.69	*RNF19A*	100	100	100
EIF2AK3	100	100	93.27	*RNF220*	100	100	93.23
ELMOD3	100	100	88.93	*RO60*	100	100	91.9
ELP4	100	100	98.03	*ROBO1*	100	100	99.1
EME1	100	100	96.58	*RPGRIP1*	100	100	91.19
ENDOU	100	100	98.22	*RPL29*	100	100	93.57
EPHB2	99.79	98.15	94.34	*RPS6KA1*	100	100	91.96
EPPK1	100	99.65	93.19	*RSC1A1*	100	100	98.98
EPRS1	100	100	98.62	*RSF1*	100	100	98.34
EPX	100	100	89.34	*RTL3*	100	100	83.26
ERAL1	100	98.76	76.56	*RTRAF*	100	100	100
ESPL1	100	100	86.01	*RTTN*	100	100	99.71
EXD1	100	100	97.03	*RYR1*	99.31	98.21	81.13
EXOC6	100	100	100	*SAMD4A*	100	100	90.82
EXTL1	100	100	81.41	*SBK2*	100	100	92.28
F11R	100	100	96.49	*SCARF1*	100	98.65	63.72
FAM117B	97.46	87.85	71.81	*SCN4A*	100	100	91.3
FAM120B	100	100	90.29	*SEC23IP*	100	100	97.44
FAM136A	100	100	95.93	*SEC24C*	100	100	89.38
FAM149B1	100	100	99.85	*SERPINA10*	100	100	98.69
FANCA	100	97.7	88.51	*SERPINH1*	100	100	88.24
FBXW12	100	100	96.99	*SERTAD1*	100	100	61.18
FBXW8	100	100	83.04	*SH3TC1*	100	100	76.45
FCGBP	100	100	89.4	*SH3TC2*	100	100	92.06
FCHSD1	100	100	88.13	*SHROOM2*	100	98.7	79.96
FER1L6	100	100	94.69	*SHROOM3*	100	100	83.83
FGF21	100	100	84.92	*SIGLEC1*	100	99.98	77.1
FMN2	100	100	91.78	*SIRPB1*	86.67	72.42	56.78
FOXB2	100	98.61	77.52	*SLC18A3*	100	100	57.16
FOXC1	100	72.63	38.42	*SLC22A5*	100	100	89.22
FRMPD2	100	100	96.24	*SLC24A3*	100	98.4	84.91
FUT1	100	100	87.8	*SLC25A11*	100	100	88.63
GABARAP	100	100	98.07	*SLC25A31*	100	100	96
GASK1B	100	100	98.7	*SLC26A1*	100	100	68.49
GATA4	100	98.01	72.25	*SLC26A9*	100	100	91.27
GDAP2	100	100	100	*SLC27A3*	100	100	74.06
GEMIN4	100	100	71.12	*SLC2A7*	100	100	86.01

GFAP	100	100	86.81	*SLC37A4*	100	100	85.9
GIT1	100	97.93	86.02	*SLC4A11*	100	100	88.82
GNLY	100	100	99.76	*SLC5A6*	100	100	96.47
GOLGA6L9	100	100	100	*SLC7A3*	100	100	94.52
GOLGA8B	100	100	99.58	*SLC9A3*	99.62	89.97	73.19
GOLGA8N	100	100	100	*SLFN5*	100	100	97.31
GOLGA8R	100	100	100	*SLX4IP*	100	100	100
GP1BA	100	100	85.25	*SMC5*	100	100	99.2
GPR150	100	85.75	39.31	*SMCO3*	100	100	100
GPR157	100	97.32	76.29	*SORBS3*	100	100	86.43
GPR20	100	100	71.03	*SPARCL1*	100	100	100
GPR42	100	100	100	*SPATA9*	100	100	99.33
GPRC5B	100	100	79.36	*SPDYE6*	100	100	100
GRAMD1A	100	99.74	80.12	*SPEM2*	100	100	86.19
GTDC1	100	100	100	*SPOCK3*	100	100	98.83
GTPBP6	50	45.78	41.97	*SPTA1*	100	100	98.13
GUCA1B	100	100	95.79	*SPTBN4*	100	99.96	72.02
GUF1	100	100	100	*SPTBN5*	100	99.91	85
HAVCR2	100	100	97.9	*SRCAP*	100	100	86.84
HBA1	100	100	100	*SREBF2*	100	100	85.51
HBA2	100	100	100	*SRRM1*	100	100	85.96
HBQ1	100	100	61.31	*SSC5D*	100	99.98	84.46
HIVEP3	100	100	88.78	*STAB1*	100	100	86.65
HLA-DRB1	100	100	100	*STARD13*	100	100	91.69
HLA-E	100	100	100	*STON2*	100	100	98.85
HOXD9	100	93.86	54.86	*SUGP1*	100	100	87.35
HPF1	100	100	98.54	*SUMO3*	100	95.98	88.7
HRC	100	100	98.3	*SUOX*	100	100	97.02
HRNR	100	100	100	*SVIL*	100	100	94.38
HSF1	100	100	88.62	*SYNC*	100	96.57	50.26
HSF5	100	100	77.65	*SYNE2*	100	100	97.75
HSPA12A	100	100	97.08	*SYP*	100	100	83.88
HSPA14	100	100	99.21	*TAAR8*	100	100	100
HYDIN	100	99.78	92.62	*TAF11L3*	100	100	100
IDUA	100	100	71.32	*TAF11L4*	100	100	100
IFI35	100	100	87.89	*TAF11L9*	100	100	100
IFNL1	100	100	85.41	*TAOK2*	100	100	71.64
IGSF5	100	100	95.26	*TBC1D3F*	100	98.93	90
IKBKG	100	98.28	63.56	*TCAF1*	100	100	100
IKZF4	100	100	89.59	*TCF20*	100	100	96.76
IL16	100	100	97.3	*TENM2*	100	100	90.65
IL9R	55.09	55.09	47.35	*TENT4A*	99.29	80.51	67.45
IPO9	100	100	93.16	*TGFB2*	100	99.79	97.85
IQGAP3	100	100	91.04	*TGIF1*	100	100	93.03
IQSEC3	100	98.18	78.26	*TIMELESS*	100	100	93.8
IST1	100	100	88.22	*TLCD1*	100	100	75.41
ITGA1	100	100	99.15	*TMEM164*	100	100	96.04
ITGA2	100	100	99.59	*TMEM200C*	100	91.16	52.52
ITGA7	100	100	86.12	*TMEM30B*	100	100	70.55
ITGAE	100	100	84.18	*TMPRSS11A*	100	100	98.5
ITPA	100	100	100	*TMPRSS6*	100	100	87.24
IZUMO1	100	100	83.66	*TNFRSF21*	100	100	96.19
IZUMO4	100	100	94.01	*TNRC6C*	100	100	91.02
JMJD7	100	100	95.06	*TNS4*	100	100	77.28
KAT2A	100	100	91.39	*TNXB*	100	99.92	88.72
KAT6A	100	100	98.24	*TOGARAM1*	100	100	99.49
KCNJ18	100	100	100	*TPP2*	100	100	98.03
KDM4B	100	100	87.48	*TPSG1*	100	100	89.5
KDM4C	100	100	97.01	*TRAK1*	100	100	92.36
KDM6B	100	100	81.54	*TRAPPC14*	100	100	75.27
KIAA1328	100	100	96.02	*TRIM49D1*	100	100	100
KIF13A	100	99.96	97.93	*TRIM49D2*	100	100	100
KIF21B	100	100	83.04	*TRIM77*	100	100	100
KIF3B	100	100	98.66	*TSC22D1*	100	100	94.47
KISS1	100	100	100	*TSHB*	100	100	100
KLC4	100	100	87.99	*TTC28*	100	98.52	92.96

KLHL30	100	100	68.28	*TTI2*	100	100	98.49
KLHL40	100	100	78.13	*TTN*	100	100	99.21
KMT2D	100	99.98	83.3	*TUBGCP6*	100	100	82.91
KRAS	100	100	77.79	*TXNRD3*	100	98.29	89.12
KRCC1	100	100	99.74	*TYRP1*	100	100	97.97
KRT26	100	100	97.44	*UAP1*	100	100	100
KRT84	100	100	92.35	*UBXN11*	100	100	91.33
KRTAP10-6	100	100	100	*UGT2B28*	100	100	100
KRTAP9-6	83.64	76.81	63.35	*UNC13B*	100	100	97.34
KRTAP9-7	100	95.88	82.55	*UQCRC1*	100	100	92.99
LAMA1	100	100	96.93	*URB2*	100	100	93.84
LAMA2	100	100	98.95	*USF3*	100	100	100
LAMP5	100	100	94.66	*USH2A*	100	99.97	98.55
LAP3	100	100	91.86	*USP17L17*	100	99.57	97.16
LCORL	100	100	99.79	*USP17L21*	99.26	85.78	66.69
LETMD1	100	100	96.27	*USP17L29*	100	100	100
LHX3	100	100	77.51	*USP17L30*	100	100	100
LIFR	100	100	99.28	*USP34*	100	100	99.45
LIG3	100	100	97.6	*USP40*	100	99.51	94.38
LILRA6	100	100	100	*UTP18*	100	100	93.77
LIMA1	100	100	95.97	*UXS1*	100	99.67	92.73
LIMK2	100	100	94.12	*VNN2*	100	100	99.75
LIPF	100	100	97.96	*VPS13D*	100	100	98.11
LMBR1L	100	100	94.21	*VPS16*	100	100	80.6
LMCD1	100	100	93.89	*WASHC1*	100	100	68.62
LMO7	100	100	99.12	*WASHC2C*	100	100	99.28
LMTK2	100	99.49	86.88	*WDR1*	100	100	87.57
LPCAT1	100	99.68	89.64	*WDR19*	100	100	99.31
LRIF1	100	100	99.44	*WDR89*	100	100	100
LRP12	100	100	96.98	*WDR91*	100	100	87.78
LRP2	100	100	98.06	*XRCC1*	100	100	95.27
LRP4	100	99	85.54	*YJEFN3*	100	100	57.52
LRRFIP1	100	100	93.31	*ZAN*	100	100	81.59
LTBP4	100	99.47	73.63	*ZC3H11A*	100	100	99.15
LUM	100	100	100	*ZDHHC11B*	100	99.05	59.14
LURAP1L-AS1	100	100	100	*ZFPM1*	98.09	73.58	51.15
LYPD2	100	100	92.06	*ZNF208*	100	100	94.78
LYRM9	100	100	89.45	*ZNF233*	100	100	99.17
MACF1	100	99.9	96.08	*ZNF284*	100	100	98.95
MAMDC4	100	100	89.26	*ZNF385C*	100	100	86.03
MAOB	100	100	99.1	*ZNF395*	100	100	84.69
MAP1A	100	100	78.37	*ZNF420*	100	100	99.58
MAP3K15	100	97.47	86.92	*ZNF618*	100	98.86	67.54
MAP3K7	100	100	99.01	*ZNF662*	100	100	98.84
MARS1	100	100	88.62	*ZNF729*	100	100	100
MAT1A	100	100	93.21	*ZNF775*	100	87.11	62.76
MCM9	100	100	97.27				

DISCLAIMER

Any preparation and processing of a sample from patient material provided to LAB by a physician, clinical institute or a laboratory (by a "Partner") and the requested genetic and/or biochemical testing itself is based on the highest and most current scientific and analytical standards. However, in very few cases genetic or biochemical tests may not show the correct result, e.g. because of the quality of the material provided by a Partner to LABS or in cases where any test provided by LAB fails for unforeseeable or unknown reasons that cannot be influenced by LAB in advance. In such cases, LAB shall not be responsible and/or liable for the incomplete, potentially misleading or even wrong result of any testing if such issue could not be recognized by LAB in advance.

This report provides information about the patient's mutations that may aid the physician's decision making

process, but this test should not be the sole source of information for making decisions on patient care and treatment. These tests should be interpreted in the context of standard clinical, laboratory, and pathological findings. Identification of a mutation in one or more of these genes does not guarantee activity of the drug in a given indication. Insertions and deletions greater than 20bp in size may not be detected by this assay. Mutations in the intronic regions and CNVs in the complex, repeats and high GC rich region have not been included in this report.

The information provided in this report was collected from various sources that we believe to be reliable and quality control procedures have been put in place to ensure the information provided is as accurate, comprehensive, and current as possible. The information provided should only be utilized as a guide or aid and the decision to select any therapy option based on the information reported here resides solely with the discretion of the treating physician. Patient care and treatment decisions should only be made by the physician after taking into account all relevant information available including but not limited to the patient's condition, family history, findings upon examination, results of other diagnostic tests, and the current standards of care. This report should only be used as an aid and the physician should employ clinical judgment in arriving at any decision for patient care or treatment.

REFERENCES

1. Genes, Global. "Rare diseases." RARE facts. Global Genes https://globalgenes.org/learn/rare-disease-facts/ (2019).
2. US department of Health and Human services. "Office of Rare diseases Research of the National center for advancing translational research-Rare Diseases Clinical Research Network (RDCRN) consisting of Rare Diseases Clinical Research Consortia (RDCRC) and a Data Management and Coordinating Center (DMCC) https://rarediseases.info.nih.gov/." Last accessed Jan 18 (2013).
3. Yang Y, Muzny DM, Reid JG, et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. N Engl J Med. 2013;369(16):1502-1511.
4. Marth AR, et al. The functional spectrum of low-frequency coding variation. Genome Biol 2011; 12(9):R84.
5. Amberger JS, Bocchini CA, Schiettecatte F, Scott AF, Hamosh A. OMIM.org: Online Mendelian Inheritance in Man (OMIM®), an online catalog of human genes and genetic disorders. Nucleic Acids Res. 2015;43(Database issue):D789-D798. https://www.omim.org/
6. Landrum MJ, Chitipiralla S, Brown GR, et al. ClinVar: improvements to accessing data. Nucleic Acids Res. 2020;48(D1):D835-D844. (https://www.ncbi.nlm.nih.gov/clinvar/)
7. Richards S, et al. Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17(5):405-24.
8. Rehder C, Bean LJH, Bick D, et al. Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021;23(8):1399-1415.
9. Miller DT, et al. ACMG Secondary Findings Working Group. ACMG SF v3.1 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2022 Jul;24(7):1407-1414.
10. Library Preparation EF 2.0 with Enzymatic Fragmentation and Twist Universal Adapter System: https://www.twistbioscience.com/resources/protocol/library-preparation-ef-20-enzymatic-fragmentation-and-twist-universal-adapter
11. Twist Target Enrichment Standard Hybridization v2 Protocol: https://www.twistbioscience.com/resources/protocol/twist-target-enrichment-standard-hybridization-v2-protocol
12. Behera, S., Catreux, S., Rossi, M. et al., Comprehensive genome analysis and variant detection at scale using DRAGEN, Nat Biotechnol (2024). https://doi.org/10.1038/s41587-024-02382-1.

*This test has been outsourced to our collaborative lab.

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