PATIENT INFORMATION

Name	:	[redacted]	Referred by	:	Dr. D A SAWANT
Age	:	31 Years	Sample ID	:	510100257
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 31 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* (60x; 36x; 30x)	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
HBB NM_000518.5	Chr11: 5226925	c.92+5G>C	Exon 1	Splice site	Heterozygous AR	Thalassemia, beta (OMIM# 613985)	Likely Pathogenic
CUBN NM_001081.4	Chr10: 16990333	c.4350+1G>T	Exon 29	Splice site	Heterozygous AR	[Proteinuria, chronic benign] (OMIM# 618884)	Likely Pathogenic
SFXN4 NM_213649.2	Chr10: 119165632	c.16G>T p.Glu6**	Exon 1	Nonsense	Heterozygous AR	Combined oxidative phosphorylation deficiency 18 (OMIM# 615578)	Likely Pathogenic

Heterozygous likely pathogenic variants in HBB, CUBN and SFXN4 genes (Autosomal recessive condition) may not have clinical impact for the patient 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:

HBB: While most carriers of Thalassemia trait may remain clinically asymptomatic, there is evidence that heterozygous individuals can manifest hematological changes such as mild anemia, erythrocytosis, 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	**98.99 %**	Exome Coverage at ≥50x	**86.12 %**
Target genes/regions Coverage at 20x	**99.52** %	Target genes/regions Coverage at 50x	**89.07 %**
Total Reads generated (millions)	**52**	Total Reads Aligned	**99.35** %

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
A3GALT2	100	100	43.79	*MUC1*	100	98.99	82.88
AAGAB	100	100	99.45	*MUC12*	100	99.82	99.03
ABCA4	100	99.97	85.86	*MUC16*	100	100	95.41
ABCC12	100	100	91.98	*MUC19*	100	100	93.75
ACACB	100	100	77.65	*MUC2*	100	99.78	83.9
ACAP3	100	100	64.55	*MUC5AC*	100	100	92.81
ACSM5	100	100	91.56	*MUC6*	100	100	77.03
ADAM9	100	100	98.25	*MYBPC2*	100	100	87.51
ADAMDEC1	100	100	99.45	*MYH14*	100	99.74	79.55
ADGRG4	100	100	99.22	*MYO18B*	100	100	81.32
ADH1C	100	100	99.65	*MYO19*	100	100	76.55
ADRM1	100	100	70.64	*MYO1A*	100	100	95.9
AEBP1	100	99.57	66.96	*MYO1D*	100	100	96.58
AFAP1L1	100	100	93.52	*MYO6*	100	100	98.52
AGAP2	100	99.09	71.97	*MYO7A*	100	99.95	84.7
AGAP4	100	100	100	*MYO9B*	100	100	73.71
AHNAK2	96.64	91.92	84.7	*NAALADL2*	100	100	99.4
AIRE	100	100	85.38	*NACAD*	100	100	73.13
AKAP9	100	100	97.84	*NAT16*	100	100	47.59
AKR7A2	100	100	71.2	*NBEAL2*	100	99.38	65.11
ALG5	100	100	91.9	*NBPF10*	100	100	99.41
ALMS1	100	100	99.32	*NBPF20*	100	99.34	96.69
ALPK2	100	100	91.89	*NBPF8*	100	100	100
AMER3	100	100	39.87	*NDE1*	100	99.75	75.57
AMZ1	100	100	69.39	*NDRG1*	100	100	82.69
ANGEL1	100	100	72.02	*NDUFAF7*	100	100	99.4
ANK2	100	100	96.24	*NEB*	100	100	97.69
ANKFY1	100	100	86.52	*NECTIN1*	100	99.62	77.99
ANKRD18B	100	100	99.58	*NEK6*	100	100	97.7
ANKRD30B	100	100	97.64	*NELL2*	100	100	99.15
ANKRD36	100	100	100	*NEU4*	100	100	66.25
ANKRD9	100	76.73	0.21	*NF1*	100	100	96.75
ANXA1	100	100	100	*NHSL1*	100	99.63	81.84

AP4B1	100	100	90.39	*NINL*	100	100	79.77
APAF1	100	100	98.15	*NIPAL1*	100	100	95.91
APOB	100	99.75	96.84	*NLRP13*	100	100	95.12
AQR	100	100	98.6	*NOS3*	100	97.9	63.31
ARFGAP1	100	99.3	76.17	*NOTCH2NLB*	100	100	74.96
ARHGEF19	100	99.62	70.72	*NOTCH4*	100	98.79	71.93
ASAH2	100	100	99.14	*NPAS2*	100	100	93.65
ASB15	100	100	96.49	*NPIPB13*	100	99.8	97.28
ASB2	100	100	65.62	*NPIPB3*	99.97	98.65	96.96
ASCC3	100	100	99.31	*NPIPB4*	100	100	100
ASXL3	100	100	94.79	*NPY4R*	100	100	99.38
ATN1	100	99.38	66.41	*NRAP*	100	100	97.1
ATP12A	100	100	89.29	*NRXN2*	100	91.32	62.92
ATP2C2	100	100	75.6	*NT5DC2*	100	90.66	70.33
ATP4B	100	100	77.63	*NUAK2*	100	100	81.62
ATP6V0D2	100	100	95.16	*NUMA1*	100	99.89	80.17
ATRNL1	100	100	95	*NUP155*	100	100	99.15
B4GALT2	100	100	82.84	*NUP188*	100	100	82.73
BABAM2	100	98.72	82.81	*NUTM2A*	100	100	95.81
BAZ2B	100	100	96.84	*NUTM2D*	100	100	100
BCO1	100	100	96.03	*NWD1*	100	99.54	72.13
BHLHE40	100	100	91.28	*NXPE4*	100	100	96.85
BICRA	100	98.36	38.86	*OBSCN*	100	99.49	71.28
BIVM-ERCC5	100	100	57.14	*OCLN*	100	100	83.02
BMS1	100	99.33	86.58	*OLFML2B*	100	100	88.9
BPIFB3	100	100	95.74	*OPN1LW*	100	100	91.23
BRD1	100	100	80.25	*OR11L1*	100	100	78.84
BRPF3	100	100	83.57	*OR2L8*	100	100	100
BTBD10	100	100	97.04	*OR4C3*	100	100	100
BTBD3	100	97.72	81.83	*OR51B6*	100	100	99.25
BTNL2	100	97.67	86.41	*OR52W1*	100	100	87.54
C1QTNF5	100	100	90.35	*OR5M10*	100	100	100
C2CD3	100	100	90.08	*OR6Q1*	100	100	95.39
C4B	100	100	96.22	*ORAI2*	100	100	85.36
C6orf132	100	99.66	67.51	*OSBPL11*	100	100	96.84
CABLES1	96.31	77.23	59.41	*OSBPL5*	100	100	86.29
CABP4	100	99.9	76.41	*OXCT1*	100	100	98.72
CAMK4	100	100	97.54	*PARP6*	100	100	93.82
CAMSAP3	100	98.07	58.4	*PAX6*	100	100	86.78
CAPN14	100	100	84.62	*PBXIP1*	100	100	71.82
CARMIL1	100	100	95.64	*PCDH17*	100	100	84.23
CCDC17	100	100	83.1	*PCED1A*	100	100	76.78
CCDC170	100	100	94.16	*PCSK9*	100	100	75.03
CCDC171	100	100	98.53	*PCYOX1*	100	100	84.06
CCDC187	100	100	86.98	*PFN3*	100	100	55.8
CCDC93	100	100	94.57	*PIF1*	100	98.12	60.9
CCKBR	100	100	83.25	*PIK3R6*	100	100	91.89
CCNB3	100	100	97.58	*PIN4*	100	100	90.26
CD44	100	100	94.65	*PKD1*	99.48	97.77	80.01
CDC42BPA	100	100	98.42	*PKD1L1*	100	100	92.62
CDH9	100	100	99.28	*PKN1*	100	98.21	78.64
CDKL5	100	100	91.97	*PLA2G2C*	100	100	93.09
CELSR1	100	96.05	68.33	*PLCG2*	100	100	91.97
CENPF	100	100	99.13	*PLEKHG4*	100	99.78	73.21
CEP135	100	100	99.28	*PLEKHH3*	100	96.98	66.99
CEP85	100	100	92.73	*PLK1*	100	100	79.28
CERK	100	97.51	67.05	*PLSCR5*	100	100	99.15
CFAP221	100	100	91.15	*PLXNB1*	100	99.94	74.36
CFHR4	100	100	100	*PMF1*	100	100	88.63
CHCHD10	100	100	80.27	*PMS2*	100	100	92.97
CHEK1	100	100	85.23	*PMVK*	100	100	97.78
CHPF2	100	100	61.1	*POGLUT3*	100	98.95	88.07
CIZ1	100	100	78.72	*POTEF*	100	100	99.94
CLCN2	100	99.93	85.14	*PPP1R1B*	100	100	72.2
CLCN6	100	100	82.57	*PRAMEF7*	100	100	93.33
CLEC4D	100	100	100	*PRKN*	100	100	88.83

CLIP1	100	100	75.94	*PRPF8*	100	100	83.45
CLK1	100	100	97.25	*PRRT4*	100	94.07	62.59
CMYA5	100	100	97.57	*PRUNE2*	100	100	95.23
CNTN2	100	100	86.02	*PRXL2A*	100	100	91.91
CNTN5	100	100	99.62	*PTGDR2*	100	100	20.79
CNTNAP3B	100	98.47	85.15	*PTPDC1*	100	100	94.62
COBL	100	100	77.52	*PTPN21*	100	100	87.73
COL6A1	100	100	74.47	*PTPRA*	100	100	92.19
COL6A2	100	100	71.34	*PTPRM*	100	100	99.01
COL9A1	100	100	97.41	*PTRH1*	100	100	94.49
COLGALT1	100	93.72	73.4	*PUS1*	100	100	69.04
COQ5	100	100	93.24	*PWWP4*	100	100	87.13
CPED1	100	100	88.72	*PYCR3*	100	100	92.93
CPSF2	100	100	95.66	*QRICH2*	100	100	84.84
CREG2	100	91.58	55.18	*RAB11FIP2*	100	100	95.06
CROCC	100	98.33	80.5	*RAPGEF4*	100	100	93.5
CRPPA	100	98.4	90.7	*RARRES1*	100	100	70.48
CRYZ	100	100	97.37	*RASA4B*	100	100	86.84
CSAG2	100	100	100	*RBFOX2*	100	100	86.04
CSK	100	100	82.64	*RBM20*	100	99.92	82.94
CSTL1	100	100	88.81	*REST*	100	100	98.35
CT45A5	100	100	99.12	*REV3L*	100	100	97.16
CT45A6	100	100	100	*REXO1*	100	100	73.82
CT47A7	92.96	56.29	23.41	*RHBDL2*	100	100	91.7
CUBN	100	100	95.39	*RHCE*	100	100	93.32
CYP11B1	100	100	88.01	*RHEX*	100	100	84.78
CYP2D6	100	100	97.83	*RIMBP3B*	100	98.56	76.63
CYP2D7	100	100	100	*RIMBP3C*	100	100	92.6
CYP4F11	100	100	98.03	*RNLS*	100	100	98.57
DEFB116	100	100	100	*RP1*	100	100	99.26
DEPDC1	100	100	98.18	*RPL3L*	100	100	94.51
DIAPH3	100	100	99.79	*RRAGD*	100	100	91.83
DISP1	100	100	94.94	*RRP12*	100	100	75.79
DLST	100	100	92.15	*RTKN2*	100	100	100
DMGDH	100	100	93.17	*RUBCNL*	100	100	97.01
DNAAF1	100	100	95.19	*RYR1*	100	98.2	76.28
DNAAF2	100	99.72	73.46	*SCARF2*	100	94.19	48.58
DNAH10	100	99.86	89.47	*SCNN1G*	100	100	88.6
DNAH12	100	100	97.02	*SCYGR7*	100	100	100
DNAH17	100	100	80.2	*SDR39U1*	100	100	95.12
DNAH6	100	100	97.4	*SEC24A*	100	100	96.81
DNAJC13	100	100	99.69	*SEC24D*	100	99.67	94.63
DNAJC16	100	100	86.69	*SEMA5B*	100	98.55	80.49
DPY19L4	100	100	95.21	*SEPTIN1*	100	100	82.96
DZIP1L	100	100	89.45	*SEPTIN9*	100	99.9	71.37
ECPAS	100	97.24	90.68	*SFXN4*	100	100	91.63
ECT2L	100	100	97.83	*SHROOM4*	100	100	88.6
EFHC1	100	100	97.66	*SIM2*	98.67	82.93	72.49
EHD3	100	100	95.15	*SIRPB1*	87.6	71.26	57.9
EHMT1	99.64	99.59	79.36	*SLC16A7*	100	100	86.57
EIF3CL	100	100	99.75	*SLC22A31*	100	94.27	72.32
EMC7	100	100	99.45	*SLC25A31*	100	100	93.7
EMID1	100	100	67.4	*SLC25A36*	100	100	97.98
ENOSF1	100	100	95.69	*SLC26A11*	100	100	79.15
ENTPD5	100	100	96.14	*SLC2A7*	100	100	76.01
EPHA6	100	100	97.3	*SLC34A1*	100	100	88.04
EPOR	100	100	78.02	*SLC66A3*	100	100	64.27
EPPK1	100	99.65	85.08	*SLC7A11*	100	100	95.33
ERCC5	100	100	95.97	*SLCO1A2*	100	100	99.42
FA2H	100	100	73.51	*SMARCA4*	100	99.93	76.3
FAM149A	100	100	95.5	*SMARCD3*	100	100	80.17
FAM187A	100	100	72.14	*SMC3*	100	100	99.18
FAM53A	100	100	70.68	*SMG1*	100	100	92.36
FAXDC2	100	100	97	*SMG5*	100	100	80.63
FBXO33	100	90.41	73.62	*SMPD4*	100	99.66	82.7
FCGBP	100	100	87.86	*SMPDL3B*	100	100	64.26

FES	100	99.89	78.07	*SMYD3*	100	100	88.72
FGFR1	100	99.74	82.58	*SNTA1*	100	87.32	67.58
FNDC3B	100	100	95.94	*SOAT2*	100	100	89.31
FOXRED2	100	100	80.54	*SPATA31A7*	100	100	95.43
FRMPD2	100	100	90.51	*SPATA31D3*	100	100	98.33
FSD1L	100	100	97.35	*SPEN*	100	99.66	87.03
FTCD	100	99.85	84.82	*SPHKAP*	100	100	86.63
FUT9	100	100	100	*SPINT1*	100	99.21	65.58
GABBR1	100	100	86.17	*SRARP*	100	100	78.04
GALNT5	100	100	95.39	*SRFBP1*	100	100	99.64
GAPVD1	100	100	92.84	*SRL*	100	100	78.39
GCG	100	100	93.64	*SRMS*	100	100	83.78
GLCE	100	100	99.31	*SRP72*	100	99.75	92.71
GMPS	100	100	99.06	*SSBP2*	100	100	100
GOLGA8B	100	100	99.38	*ST6GALNAC6*	100	100	83.59
GOLGA8N	100	100	100	*STARD13*	100	100	81.26
GOLGA8R	100	100	100	*STIMATE*	100	99.59	88.3
GOLGA8T	100	91.38	78.81	*STIMATE-MUSTN1*	100	100	100
GPATCH1	100	100	92.49	*STX18*	100	100	97.73
GPR137B	100	100	92.11	*STYK1*	100	100	89.57
GPR158	100	100	86.24	*SUPT20HL2*	100	100	99.05
GPR42	100	100	97.98	*SVOPL*	100	100	85.91
GPX4	100	89.07	49.63	*SYT14*	100	100	98.74
GRIN2D	96.53	69.61	49.96	*TACC2*	100	100	84.76
GRIP2	100	100	79.34	*TAF11L3*	100	100	100
GTPBP4	100	100	98.48	*TAF11L4*	100	100	100
H2AL3	100	100	47.43	*TAF11L9*	100	100	100
HAO2	100	100	97.89	*TBC1D3E*	100	96.3	86.79
HBB	100	100	99.16	*TBC1D9B*	100	99.71	77.03
HEYL	100	100	69.5	*TCAF1*	100	100	88.45
HLA-DQB2	100	100	82.3	*TEC*	100	100	97.2
HNRNPC	100	100	100	*TENM4*	100	99.06	77.84
HOXB3	100	97.97	73.43	*TENT4A*	99.12	81.44	69.47
HPS3	100	100	98.02	*TENT5A*	100	100	89.9
HR	100	99.64	65.89	*TEX14*	100	100	90.35
HRG	100	100	98.38	*TG*	100	100	91.16
HRNR	100	100	100	*TIAM2*	100	100	80.23
HSF1	100	100	61.35	*TMCO5A*	100	100	100
HSPA9	100	100	88.46	*TMEM126B*	100	100	96.73
HSPG2	100	99.24	72.42	*TMEM260*	100	100	98.45
IFT74	100	100	97.97	*TMPRSS15*	100	100	99.53
IGSF8	100	100	61.46	*TNS1*	100	100	80.14
IL23R	100	100	99.47	*TOP2A*	100	100	96.85
IL6ST	100	100	96.09	*TOP3B*	100	99.86	64.89
IMPG1	100	100	92.78	*TOPORS*	100	100	98.63
INPP5F	100	99.14	92.03	*TOX*	100	98.92	78.37
INSR	100	98.43	82.7	*TPSAB1*	100	100	97.17
INTS1	100	99.26	71.55	*TRAF2*	100	98.39	62.85
IPO5	100	99.63	92	*TRAK1*	100	100	75.88
IQGAP3	100	100	84	*TRAPPC8*	100	100	96.25
IRF5	100	100	75.65	*TRAPPC9*	100	99.57	87.9
IRX4	100	100	58.53	*TREM1*	100	98.94	90.77
JAKMIP2	100	100	97.55	*TRIM36*	100	100	96.38
KCNJ18	100	100	100	*TRIOBP*	100	99.13	77.07
KDM6B	100	99.76	64.84	*TRPC4*	100	100	97.66
KDR	100	100	95.88	*TRPM4*	100	99.89	83.81
KIAA1614	100	99.46	64.41	*TSC2*	100	100	75.65
KIF7	100	95.54	63.01	*TSEN2*	100	100	86.87
KLF1	100	100	63.74	*TSHZ1*	100	98.77	74.28
KLHL11	100	100	70.71	*TSPAN4*	100	100	66.56
KLHL31	100	100	96.85	*TSPAN8*	100	100	100
KLHL38	100	100	71.59	*TTC31*	100	98.72	73.83
KLK14	100	98.63	54.73	*TTC6*	100	100	94.6
KLRF2	100	100	100	*TTN*	100	100	97.57
KRT15	100	100	94.72	*TXLNA*	100	100	73.97
KRTAP10-6	100	100	100	*TYW1*	100	100	97.36

KRTAP10-7	100	100	100	*TYW3*	100	100	95.64
KRTAP2-2	100	100	100	*U2AF1L4*	100	100	49.64
L2HGDH	100	100	93.65	*UBASH3B*	100	100	83.13
LAGE3	100	100	98.85	*UBN2*	100	94.89	88.46
LAMA4	100	99.51	92.96	*UBQLN3*	100	100	84.04
LAMA5	100	98.69	83.19	*UBXN11*	100	100	90.38
LILRA6	100	100	100	*UFL1*	100	100	100
LIM2	100	100	59.3	*UGT2B4*	100	100	99.09
LONRF3	100	100	78.64	*URB2*	100	100	75.69
LPIN2	100	99.93	92.42	*USH2A*	100	99.99	96.41
LRPPRC	100	100	99.82	*USP17L17*	100	100	100
LRRC4B	100	98.37	81.56	*USP17L21*	100	100	98.95
LRRC56	100	100	82.34	*USP17L30*	100	100	100
LRRFIP2	100	100	98.32	*USP36*	100	100	80.86
LRRN1	100	100	94.73	*VCPIP1*	100	100	91.5
LSG1	100	100	95.83	*WASHC1*	100	100	47.67
LTA4H	100	100	97.57	*WDR70*	100	100	97.84
LTBP2	100	99.84	80.53	*WWC1*	100	100	86.5
LTBP4	100	98.97	70.22	*WWP2*	100	100	92.7
MACF1	100	100	92.49	*XKR5*	100	100	75.28
MAGEC1	100	100	93.44	*XKR8*	100	97.49	50.16
MALSU1	100	100	90.35	*ZC3H11B*	100	100	100
MAMDC4	100	100	79.35	*ZFAND3*	100	100	85.44
MBD3L5	100	100	100	*ZFHX3*	100	100	84.51
MCF2L	100	100	80.78	*ZIM2*	100	100	85.32
MCM6	100	100	91.09	*ZNF280D*	100	100	97.98
MCM9	100	100	93.63	*ZNF319*	100	100	30.02
MCTP1	100	100	97.96	*ZNF335*	100	100	81.7
MCTP2	100	100	95.64	*ZNF33A*	100	100	98.63
MEIOSIN	100	100	69.93	*ZNF347*	100	100	99.6
MEP1B	100	100	97.96	*ZNF395*	100	100	68
MMP7	100	100	100	*ZNF418*	100	100	100
MORC2	100	100	89.38	*ZNF503*	100	100	70.63
MRC2	100	100	72.27	*ZNF518B*	100	100	99.78
MROH2A	100	100	92.55	*ZNF565*	100	100	91.42
MRPL38	100	100	76.38	*ZNF777*	100	97.3	72.45
MSANTD5	100	100	91.74	*ZNF778*	100	100	89.49
MTMR7	100	100	97.17	*ZSWIM5*	99.78	96.52	80.33

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.

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*This test has been outsourced to our collaborative lab.

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