PATIENT INFORMATION

Name	:	[redacted]	Referred by	:	Dr. D A SAWANT
Age	:	43 Years	Sample ID	:	510100254
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 43 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 detected the variant as below.

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

GENE/ REFSEQ	COORDINATE (GRCh38)	VARIANT* (36x)	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
HSF4 NM_001374675.1	chr16: 67167479	c.737del p.Pro246Glnfs Ter73	Exon 8	Frameshift	Heterozygous AD	Cataract 5, multiple types (OMIM#116800)	Likely Pathogenic

RECOMMENDATION:

Heterozygous likely pathogenic variant in the *HSF4* gene is detected. Variant in *HSF4* is associated with congenital cataract which has variable penetrance As [redacted] is currently asymptomatic, the result is predictive and should be carefully correlated with any clinical finding.

OMIM - Gene and disease association

***HSF4* (Heat Shock Transcription Factor 4) Gene:**

HSF4 (Heat Shock Transcription Factor 4) is a Protein Coding gene. Diseases associated with *HSF4* include Cataract 5, Multiple Types and Early-Onset Lamellar Cataract. Among its related pathways is 10q11.21q11.23 copy number variation syndrome. Gene Ontology (GO) annotations related to this gene include DNA-binding transcription factor activity and transcription corepressor activity. An important paralog of this gene is *HSF1*. https://www.genecards.org/cgi-bin/carddisp.pl?gene=HSF4#diseases

Cataract 5, multiple types (OMIM#116800):

Congenital cataracts cause 10 to 30% of all blindness in children, with one-third of cases estimated to have a genetic cause (summary by Bu et al., 2002). Mutations in the HSF4 gene have been found to cause multiple types of cataract, which have been described as infantile, lamellar, zonular, nuclear, anterior polar, stellate, and Marner-type. https://www.omim.org/entry/116800?search=HSF4&highlight=HSF4

VARIANT SUMMARY - INTERPRETATION

***HSF4* NM_001374675.1: c.737del; p.Pro246GlnfsTer73– Likely Pathogenic**

A heterozygous frameshift variant was detected in the *HSF4* gene (c.737del; p.Pro246GlnfsTer73).
It is very rare in gnomAD database and has not been previously reported in ClinVar database.
This variant is classified as Likely Pathogenic according to ACMG classification.
Variant coverage statistics: Ref allele coverage- TC=19; Alt allele coverage- T=17

CARRIER SCREENING FINDINGS

GENE/ REFSEQ	COORDINATE (GRCh38)	VARIANT* (62x; 50x; 63x; 97x)	EXON/ INTRON	VARIANT TYPE	ZYGOSITY/ INHERITANCE	OMIM/ PHENOTYPE	CLASSIFICATION* ACMG/AMP
SGCB NM_000232 .5	chr4: 52029836	c.271C>T p.Arg91Cys	Exon 3	Missense	Heterozygous AR	Muscular dystrophy, limb-girdle, autosomal recessive 4 (OMIM#604286)	Pathogenic
ITGA2B NM_000419 .5	chr17: 44384316	c.886G>A p.Gly296Arg	Exon 9	Missense	Heterozygous AD/AR	Glanzmann thrombasthenia 1 (OMIM#273800)	Likely Pathogenic
BCS1L NM_001079 866.2	chr2: 218661192	c.205C>T p.Arg69Cys	Exon 2	Missense	Heterozygous AR	Mitochondrial complex III deficiency, nuclear type 1 (OMIM#124000)	Pathogenic
LBR NM_002296 .4	chr1: 225403395	c.1756C>T p.Arg586Cys	Exon 14	Missense	Heterozygous/ AD/AR	Greenberg skeletal dysplasia (OMIM#215140) Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly (OMIM #618019)	Likely Pathogenic

Heterozygous pathogenic/likely pathogenic variants in SGCB, BCS1L, ITGA2B and LBR 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:

ITGA2B and *LBR:* Autosomal dominant condition has also been reported in OMIM, with individuals being asymptomatic to showing mild phenotype. *ITGA2B* gene is linked to platelet function disorder with variable bleeding risk. Hematology consultation is advised for evaluation of bleeding risk. *LBR* gene is associated with benign nuclear morphology anomaly which is usually asymptomatic.

ANALYSIS STATISTICS

Exome Coverage at 20x	**99.11 %**	Exome Coverage at 50x	**94.0 %**
Target genes/regions Coverage at 20x	**99.52 %**	Target genes/regions Coverage at 50x	**94.35 %**
Total Reads Aligned	**99.28 %**	Total Reads generated (millions)	**63**

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	Coverage 1X	Coverage 20X	Coverage 50X	*Gene*	Coverage 1X	Coverage 20X	Coverage 50X
ABCA1	100	100	97.92	*MYO15A*	100	98.15	83.66
ABCB7	100	100	98.45	*MYO18A*	100	100	89.65
ABTB1	100	100	97.37	*MYOF*	100	100	97.44
ACOT2	100	100	100	*MYOM2*	100	100	98.07
ACSS1	100	98.66	89.89	*NAT2*	100	100	100
ADAM30	100	100	100	*NAV2*	100	100	95.72
ADAMTS7	100	100	95.52	*NBPF10*	100	100	99.7
ADAMTS9	100	100	99.57	*NBPF20*	100	99.92	97.81
ADGRE3	100	100	95.82	*NBPF26*	98.44	98.44	98.31
ADGRV1	100	100	99.67	*NCAPD2*	100	100	95.79
ADRM1	100	100	95.82	*NCKIPSD*	100	100	94.06
AGGF1	100	100	95.17	*NCOR1*	100	100	98.33
AHNAK2	100	100	96.7	*NCR3LG1*	100	100	98.68
AK5	100	100	99.65	*NDUFB6*	100	100	92.87
AKAP8L	100	100	91.82	*NEMF*	100	100	100
ALKAL1	100	100	100	*NEU2*	100	100	94.14
ALMS1	100	100	99.13	*NHS*	100	98.32	92.34
AMELX	100	100	100	*NIPAL2*	100	95.52	82.86
ANKHD1	100	100	98.02	*NKTR*	100	100	99.66
ANKHD1-EIF4EBP3	100	100	100	*NLRP4*	100	100	96.02
ANKRD11	100	100	90.21	*NPIPB13*	100	99.75	96.8
ANKRD28	100	100	98.6	*NPIPB3*	100	99.09	96.55
ANKRD30BL	100	98.76	83.31	*NPNT*	100	100	93.33
ANKRD50	100	100	99.77	*NPY4R*	100	100	100
AOPEP	100	97.66	94.68	*NRCAM*	100	100	99.4
ARHGAP21	100	100	99.29	*NRP2*	100	100	91.97
ARHGEF18	100	99.31	85.96	*NUFIP2*	100	100	98.04
ARHGEF37	100	100	95.92	*NUTM2A*	100	100	100
ARHGEF40	100	100	98.16	*NUTM2D*	100	100	100
ARMC5	100	100	89.51	*ODR4*	100	100	100
ARRDC5	100	100	93	*OPN1LW*	100	100	98.17
ATN1	100	100	89.03	*OR13A1*	100	100	100
ATP10D	100	100	99.12	*OR1I1*	100	100	90.82
ATP1A4	100	100	94.99	*OXNAD1*	100	100	100
BAZ2A	100	100	96.46	*PAMR1*	100	100	98.37
BAZ2B	100	100	99.66	*PARP12*	100	90.41	83.4
BCL9	100	100	95.59	*PBXIP1*	100	100	92.26
BCS1L	100	100	94.06	*PCDHB11*	100	100	100
BDP1	100	100	99.71	*PDE11A*	100	100	100
BLK	100	100	92.33	*PDE6A*	100	100	96.05
BNC1	99.53	96.88	94.7	*PECR*	100	100	99.34
BPIFB3	100	100	97	*PERCC1*	100	100	84.83
BRD3	100	100	93.71	*PFKFB1*	100	100	99.2
BRF2	100	100	95.18	*PGM1*	100	99.55	95.01
BRIP1	100	100	99.86	*PGM2*	100	100	99.3
BYSL	100	100	93.45	*PHLDB1*	100	100	93.25
C16orf90	100	100	96.82	*PIEZO1*	100	100	85.83
C1QTNF5	100	100	90.21	*PIEZO2*	100	100	97.6
C2CD3	100	100	98.7	*PLD1*	100	100	100
C4B	100	100	99.47	*PLSCR5*	100	100	100
C8A	100	100	98.59	*PMFBP1*	100	100	98.82
CACHD1	100	100	98.3	*PNMA8A*	100	100	98.66
CACNA1A	100	100	94.41	*PNPLA6*	100	100	88.98

CACNA1F	100	100	98.06	*PODNL1*	100	100	86.09
CACNA1H	100	98.86	90.47	*PODXL2*	100	95.54	71.51
CADM1	100	95.65	95.65	*POLQ*	100	100	97.47
CAMTA1	100	100	94.66	*POM121C*	100	100	97.24
CARS2	100	97.47	92.41	*POTEG*	100	100	100
CCDC168	100	100	100	*POU5F1B*	100	100	100
CCDC171	100	100	99.71	*PPAN*	100	100	82.09
CCDC180	100	100	98.94	*PPARGC1B*	100	100	92.84
CCDC187	100	100	94.83	*PPP1R9A*	100	100	99.84
CCDC40	100	100	88.54	*PRAMEF20*	100	100	100
CCDC88C	100	100	87.27	*PRDM8*	100	97.68	67.9
CCNT2	100	100	98.44	*PROCR*	100	100	91.13
CCSER1	100	100	100	*PRODH2*	100	100	95.12
CD160	100	100	99.72	*PROM1*	100	100	99.82
CDCP2	100	100	95.27	*PRPSAP1*	100	100	96.11
CDH23	100	100	94.11	*PRR14L*	100	100	95.84
CDH8	100	100	100	*PRSS55*	100	100	98.55
CDON	100	99.9	97.68	*PSD2*	100	100	91.86
CEP152	100	99.98	99.91	*PSD4*	100	100	91.82
CEP164	100	100	93.13	*PTPRD*	100	100	98.5
CEP78	100	100	98.02	*RAB11FIP1*	100	100	90.07
CFAP54	100	100	99.82	*RAB11FIP5*	100	100	82.52
CFAP58	100	100	98.54	*RAB27A*	100	100	99.3
CFAP77	100	100	99.56	*RAB3GAP2*	100	100	99.35
CHCHD2	100	100	100	*RAD21L1*	100	100	100
CHD1	100	100	99.81	*RALGAPB*	100	100	99.46
CHRDL2	100	100	92.51	*RASA1*	100	100	99.97
CHST15	100	100	96.43	*RASA4B*	100	100	95.76
CHTF18	99.97	92.18	83.91	*RASAL3*	100	94.32	74.38
CILP	100	100	92.78	*RASL12*	100	100	94.12
CIROP	100	100	96.21	*RBM6*	100	100	96.39
CNTNAP3B	100	99.47	94	*RBP2*	100	100	100
COG1	100	99.93	90.99	*RCC1L*	100	99.26	77.23
COL16A1	100	100	89.58	*RCN3*	100	100	84.43
COL1A1	100	100	83.2	*RDX*	100	99.44	96.57
COL4A3	100	99.94	99.25	*RERGL*	100	100	100
COL4A6	100	100	96.88	*REXO1*	100	100	93.52
COL5A1	100	98.09	90.58	*REXO4*	100	100	96.38
COL9A1	100	100	98.83	*RFX7*	100	100	100
COMT	100	100	96.8	*RHBDD1*	100	100	100
COQ7	100	100	89.99	*RHCE*	100	100	94.41
COQ9	100	100	99.9	*RIMBP3B*	100	100	89.35
CORO1C	100	100	100	*RIMBP3C*	100	100	97.32
CPLX4	100	100	100	*RIPOR3*	100	100	94.16
CRB2	100	99.78	77.77	*RNASE10*	100	100	100
CRTAC1	100	100	99.68	*RNF10*	100	100	97.47
CRYL1	100	100	96.77	*RNF175*	100	100	94.05
CSE1L	100	100	100	*RNF25*	100	100	89.2
CT45A6	100	100	100	*ROBO1*	100	100	99.47
CT47A7	79.47	61.59	36.22	*RP1L1*	100	100	90.89
CTSC	100	100	100	*RTN1*	100	100	87.27
CUL9	100	100	92.01	*RUSC2*	100	100	90.71
CYLD	100	100	99.68	*RXFP4*	100	100	95.11
CYP2A13	100	100	99.73	*RYR3*	100	100	98.78
CYP2C9	100	100	100	*SAP25*	100	100	96.31
DAPK2	100	100	93.03	*SCNN1D*	100	100	90.54
DCAF8L2	100	100	100	*SCO1*	100	100	99.89
DCHS2	100	100	96.4	*SCYGR3*	100	100	100
DCLRE1A	100	100	100	*SDAD1*	100	100	100
DCUN1D4	100	100	100	*SDSL*	100	100	87.74
DDX60L	100	100	100	*SEC14L1*	100	100	96.72
DEFB126	100	100	98.21	*SELENOO*	98.01	86.12	63.43
DGKI	100	92.59	87.28	*SERPINB11*	100	100	100
DISC1	100	100	98.86	*SETX*	100	100	98.09
DLG4	100	99.51	88.65	*SF3A1*	100	100	89.08
DLG5	100	100	94.98	*SFT2D3*	100	50.46	12.81

DMPK	100	100	94.57	*SGCB*	100	96.17	93.99
DMWD	100	96.76	70.85	*SGCG*	100	100	100
DNAH14	100	100	99.62	*SH2B2*	100	92.93	61.63
DNAH9	100	100	95.4	*SH3RF2*	100	100	89.9
DPP4	100	100	100	*SHC3*	99.36	94.09	76.94
DSG2	100	100	98.28	*SHKBP1*	100	100	91.16
ECPAS	99.49	97.63	92.49	*SIGLEC16*	100	100	100
EEPD1	100	100	96.55	*SIN3A*	100	100	95.5
EFCAB13	100	100	100	*SIPA1L2*	100	100	98.36
EGF	100	100	99	*SIRPB1*	88.26	72.5	64.35
EHBP1	100	100	99.87	*SIX5*	100	94.23	59.14
EIF2S3B	100	100	100	*SKOR1*	100	97.19	75.3
EIF3CL	100	100	100	*SLA*	100	100	98.54
EMILIN1	100	100	90.7	*SLC12A2*	100	100	93.9
ENPP2	100	100	99.93	*SLC15A3*	100	91.95	74.02
EPHA6	100	100	99.81	*SLC22A4*	100	100	97.98
EPPK1	100	99.65	96.19	*SLC24A2*	100	100	99.35
ERCC6L2	100	100	98.79	*SLC24A3*	100	98.86	90.96
ERG	100	100	97.96	*SLC24A4*	100	100	99.49
ERICH2	100	100	86.97	*SLC27A2*	100	100	99.73
ETAA1	100	100	99.57	*SLC2A6*	100	100	93.84
EVPL	100	99.79	85.73	*SLC36A4*	100	100	100
EXD3	100	100	93.64	*SLC37A4*	100	100	88.3
FAM149A	100	100	95.92	*SLC6A15*	100	100	100
FAM186A	100	100	99.7	*SLC8A3*	100	100	98.94
FAM83C	100	98.84	82.89	*SLCO1C1*	100	100	100
FAT3	100	100	98.87	*SLFN5*	100	100	99.83
FAXDC2	100	100	100	*SMC4*	100	100	100
FBN3	100	100	91.91	*SMIM24*	100	100	100
FCGBP	100	100	94.53	*SMTN*	100	100	91.49
FCRL3	100	100	97.28	*SNX14*	100	100	100
FOXD2	100	91.4	40.86	*SNX2*	100	100	98.91
FOXM1	100	100	95.75	*SORCS2*	98.11	91.32	84.53
FREM1	100	100	98.2	*SOST*	100	100	68.44
FRMD4A	100	100	89.22	*SPAST*	100	95.82	89.44
FRMPD3	100	100	95.55	*SPATA31A3*	100	100	100
FSTL4	100	100	95.06	*SPDYE6*	100	100	100
GABBR1	100	100	95.48	*SPEG*	100	99.68	87.16
GARS1	100	100	97.67	*SPHKAP*	100	100	99.63
GLI2	100	97.26	77.53	*SPOCK1*	100	99.92	96.52
GLP1R	100	100	93.81	*SPRED1*	100	100	99.45
GNAI2	100	100	89.72	*SPRTN*	100	100	99.08
GOLGA8B	100	100	95.53	*SPTA1*	100	100	99.38
GOLGA8N	100	100	100	*SRSF12*	100	100	100
GOLGA8R	100	100	100	*SSH2*	100	100	98.44
GOLPH3	100	89.52	70.44	*ST18*	100	100	96.66
GPC5	100	100	100	*STAB1*	100	100	92.07
GPR174	100	100	99.1	*STAG2*	100	100	100
GRK7	100	100	98.01	*STAM*	100	100	100
GSS	100	100	99.53	*STK36*	100	100	91.94
GTPBP8	100	100	100	*STRA6*	100	100	98.87
HBA1	100	100	100	*SUOX*	100	100	93.8
HBA2	93.43	53.11	19.9	*SYNE2*	100	100	99.02
HELZ2	100	99.7	86.47	*SYT8*	100	100	94.35
HGS	100	100	90.39	*TAF11L3*	100	100	100
HHLA1	100	100	97.77	*TAF11L4*	100	100	100
HIVEP1	100	100	97.04	*TAF11L9*	100	100	100
HJURP	100	100	97.91	*TALDO1*	100	100	96.38
HLA-A	100	100	100	*TAS2R60*	100	100	98.43
HLA-DQB1	100	100	84.44	*TBC1D3E*	99.64	92.18	86.73
HLA-DRB1	100	97.01	75.69	*TBC1D3F*	100	100	98.05
HMCN2	100	99.98	90.59	*TCAF1*	100	100	100
HPSE2	100	100	97.94	*TCOF1*	100	100	90.1
HRCT1	100	100	74.71	*TDRD15*	100	100	100
HRNR	100	100	100	*TENM2*	100	100	92.54
HSF4	100	100	94.36	*TENT5A*	100	100	97.49

HTT	100	99.22	95.94	*TJP1*	100	99.54	95.41
ICMT	100	81.45	76.57	*TLCD5*	100	100	88.68
ID1	100	100	51.42	*TMEM212*	100	100	99.49
IGF2BP3	100	100	91.53	*TMEM216*	100	100	98.74
IGHMBP2	100	100	93.66	*TMEM219*	100	100	97.44
INSR	100	98.5	90.72	*TMEM39B*	100	100	97.54
INSYN1	100	100	93.42	*TMPRSS7*	100	100	99.01
INTU	100	100	99.9	*TNFRSF8*	100	100	92.11
IPO11	100	100	99.34	*TNRC18*	100	98.7	84.14
ITGA11	100	100	96.33	*TNRC6A*	100	100	97.92
ITGA2B	100	100	91.71	*TOMM40L*	100	100	100
ITSN2	100	100	98.86	*TRIM68*	100	100	95.28
KCNH2	99.95	99.34	83.81	*TSPAN12*	100	100	98.97
KCNT1	100	96.48	83.8	*TTC17*	100	100	99.92
KDM6B	100	99.76	89.84	*TTC19*	100	97.17	91.68
KIAA0825	100	100	99.75	*TTC31*	100	100	90.51
KIAA1217	100	100	98.43	*TTN*	100	100	99.37
KIF12	100	100	89.15	*TXK*	100	100	100
KIF1A	100	100	86.01	*UBFD1*	100	98.09	78.92
KLHDC7B	100	98.3	75.76	*UGT2B28*	100	100	98.3
KLHL38	100	100	95.82	*UMAD1*	100	100	77.27
KLHL9	100	100	100	*UMODL1*	100	100	86.54
KMT2D	100	100	90.13	*UNG*	100	100	95.46
KRT78	100	100	89.12	*USF3*	100	100	99.96
KRT79	100	100	96.67	*USP17L17*	100	100	100
KRTAP10-6	100	100	100	*USP17L21*	100	100	98.95
KRTAP10-8	100	100	100	*USP17L29*	100	100	100
KRTAP9-6	83.23	69.98	62.53	*USP17L30*	100	100	100
KRTAP9-7	100	100	88.04	*USP44*	100	100	99.18
LBR	100	100	99.48	*USP45*	100	100	100
LCN12	100	100	88.04	*USP8*	100	100	100
LEKR1	100	100	98.17	*UTRN*	100	100	99.33
LRP10	100	100	88.57	*VHL*	100	100	78.29
LRRC55	100	100	75.44	*VPS13B*	100	99.95	98.05
LSM11	100	92.15	77.38	*VPS13C*	100	100	99.69
LTBP1	100	97.72	85.05	*VPS37C*	100	100	91.46
LTK	100	100	88.81	*VPS39*	100	100	98.2
LUZP4	100	100	100	*VWC2*	100	100	94.75
LZTS2	100	100	88.61	*VWDE*	100	100	99.15
MAP3K15	100	97.6	90.83	*WASHC1*	100	100	100
MAP3K20	100	100	98.67	*WDCP*	100	100	99.23
MAP3K3	100	100	95.54	*XKR5*	100	100	91.6
MAP4	100	100	98.95	*XPOT*	100	100	97.82
MAPK8IP2	100	94.63	71.83	*XRCC1*	100	100	98.58
MCC	100	100	98.93	*ZBED4*	100	100	89.29
MED26	100	100	84.6	*ZC3H11B*	100	100	100
METTL13	100	100	94.09	*ZC3H13*	100	100	99.94
MFSD10	100	100	97.41	*ZFP1*	100	100	99.47
MFSD2A	100	100	95.53	*ZFP36*	100	100	94.8
MIXL1	100	76.07	45.64	*ZNF14*	100	100	100
MKRN2OS	100	100	97.47	*ZNF142*	100	100	85.82
MMP28	100	100	94.92	*ZNF20*	100	100	99.69
MOCS2	100	100	99.87	*ZNF280D*	100	100	99.37
MRGPRX3	100	100	100	*ZNF418*	100	100	99.41
MROH2A	100	100	95.83	*ZNF469*	100	100	90.04
MSH2	100	100	96.32	*ZNF516*	100	96.79	81.01
MTCL1	100	100	93.52	*ZNF556*	100	100	100
MTG1	100	100	99.5	*ZNF644*	100	100	100
MTUS2	100	100	91.66	*ZNFX1*	100	100	97.2
MUC1	100	99.22	96.15	*ZP2*	100	100	98.16
MUC2	100	99.81	91.87	*MUC6*	100	100	93.03
MUC5AC	100	100	96.91	*MYD88*	100	100	92.36

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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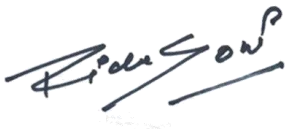
*This test has been outsourced to our collaborative lab.

Dr. Vamshi Krishna Thamtam
MCI-I7-25915
MBBS, MD, DipRCPath, UK (Molecular Genetics)
Post Doctoral Fellowship, Tata Medical Center
Technical Director - Genomics & Clinical Cytogenomics
National Reference Laboratory



Dr. Richa Soni
DMC 89643
MD Pediatrics
DM Medical Genetics
Senior Clinical Geneticist
Department of Genomics