## Patient Details

| | | | | | |
|---|---|---|---|---|---|
| **Name** | : MADHURI BHASKAR | **Sex / Age** | : FEMALE / 32 Years | **Case ID** | : 21107401264 |
| **Ref By** | : Dr. Deepti Saxena | **Dis.Loc.** | : | **PT. ID** | : 2022903396 |
| **Test Name** | : ORION (WES-Whole Exome Sequencing) | **Bill. Loc.** | : SGPGI HOSPITAL, LUCKNOW | **PT. Loc.** | : |

## Sample Details

| | | | | | |
|---|---|---|---|---|---|
| **Registration Date & Time** | : 2022-11-15 16:19:33 | **Sample Type** | : Whole Blood EDTA | **Ph # Re** | : |
| **Sample Date & Time** | : 2022-11-15 16:19:00 | **Ref ID 1.** | : | **Acc. Remarks** | : |
| **Report Date & Time** | : 2022-11-30 10:34:51 AM | **Ref ID 2** | : | **PT. Loc.** | : |

## Clinical History

Consanguinity: Absent
Clinical symptoms: features of weakness and episodes of syncopal attacks 3-4 times within a period of 4 months 1 year back. H/o difficulty in night vision with no history of chest pain or diabetes or hypertension.
Investigations done:
Serum triglycerides- 786 mg/dl, serum cholesterol- 144 mg/dl, HDL- 36 mg/dl, LDL- 58.7 mg/dl, VLDL- 157.2 mg/dl, RBS- 97mg/dl and hba1c- 5.6 %.
Family history: Proband's mother passed away at 46years of age due to myocardial infarction
Clinical suspicion: Familial hypertriglyceridemia.

## Test Results and Interpretation

**HETEROZYGOUS VARIANT OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED: CLINICAL CORRELATION RECOMMENDED**

## Summary of Variants

| Gene and Transcript | Exon/Intron Number | Variant Nomenclature | Zygosity | Classification | Disease | Inheritance |
|---|---|---|---|---|---|---|
| APOA5 (NM_001371904.1) | Exon 3 | c.853C>T p.Leu285Phe [Depth-56X] | Heterozygous | Uncertain significance | {Hypertriglyceridemia, susceptibility to} | Autosomal dominant |

## Variant Details

APOA5





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| | |
|---|---|
| **Variant Nomenclature** | c.853C>T (p.Leu285Phe) |
| **Genomic Nomenclature** | chr11:g.116661092G>A |
| **Zygosity** | Heterozygous |

The missense c.853C>T (p.Leu285Phe) variant in APOA5 gene has not been reported previously as a pathogenic variant nor as a benign variant, to our knowledge. The p.Leu285Phe variant is novel (not in any individuals) in gnomAD Exomes and 1000 Genomes. This variant has not been reported to the ClinVar database. The amino acid change p.Leu285Phe in APOA5 is predicted as conserved by GERP++ and PhyloP across 100 vertebrates. The amino acid Leu at position 285 is changed to a Phe changing protein sequence and it might alter its composition and physico-chemical properties. For these reasons, this variant has been classified as Variant of Uncertain Significance (VUS).

## Disease

**HYPERTRIGLYCERIDEMIA, FAMILIAL**

Mutations in the APOA5 gene, leading to truncated apolipoprotein A-V devoid of lipid-binding domains located in the carboxy-terminal end of the protein, if present in the homozygous state, are expected to cause severe type V hyperlipidemia If present in the heterozygous state, these mutations predispose to hypertriglyceridemia in combination with other genetic factors or pathological conditions. **Mutations in APO5 gene are also known to cause Hyperchylomicronemia, late-onset**

## References

1. Calandra S et al. APOA5 and triglyceride metabolism, lesson from human APOA5 deficiency. Curr Opin Lipidol. 2006 Apr;17(2):122-7.

## Special Recommendations

**Parental testing for the above variant is recommended**

## Recommendations

1. **Please correlate clinically.**
2. **Genetic counseling for accurat e int erpretation of t est results is recommended.**
3. **The observed variation has not been Sanger confirmed. Hence Sanger sequencing is recommended.**
4. **Segregation analysis of the above variants (testing of multiple affected as well as unaffected members) is recommended after Sanger confirmation in proband. Variant classification may be modified after segregation analysis.**
5. **If the above results do not correlate completely with patient phenotype, additional testing is advised based on clinician's discretion.**

## Technical Notes

**Methodology: Massively Parallel Sequencing (Next Generation Sequencing)** Genomic DNA from the submitted specimen was enriched for the complete coding regions and splice site junctions of genes listed below using a custom bait- capture system. Paired End Sequencing was performed with 2x100/2x150 chemistry, on an Illumina platform. Reads were assembled and were aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. Data was filtered and analyzed to identify variants of interest and interpreted in the context of a single most damaging, clinically relevant



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transcript for the purpose of the report, indicated as a part of variant details. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 5-10bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions thought to be significant are interrogated on request by Sanger backfill. Deletion and duplication analysis is performed in cases when indicated but detected variations need to be confirmed by an alternate methodology. Sequence and copy number variants are reported according to the Human Genome Variation Society (HGVS).

**Tools and Databases employed for analysis:**Clinvar, OMIM, HGMD, UCSC genome browser, Uniprot, Ensembl, dbSNP, gnomAD, ExAC, Pubmed, Dgap, icgc, Kaviar, various bioinformatics analysis, predictive tools and disease specific databases used as available and appropriate. Such tools/databases would be mentioned wherever used.

**Bioinformatics pipeline version: 5.0.0**.

## Gene Coverage

Indication Based Analysis:

| Gene | Coverage | Gene | Coverage | Gene | Coverage | Gene | Coverage |
|---|---|---|---|---|---|---|---|
| ABCA1 | 100% | ABCA4 | 100% | ABCB6 | 100% | ABCC6 | 100% |
| ABCC8 | 100% | ABCC9 | 100% | ABCG5 | 100% | ABCG8 | 100% |
| ABHD5 | 100% | ACAT1 | 100% | ACTA2 | 100% | ACTN4 | 100% |
| ACVRL1 | 100% | ADA2 | 100% | ADAMTSL4 | 100% | ADAR | 100% |
| AEBP1 | 100% | AGBL5 | 100% | AGL | 100% | AGPAT2 | 100% |
| AHI1 | 100% | AHR | 100% | AIP | 100% | AIRE | 100% |
| AKAP10 | 100% | AKAP9 | 100% | AKR1B1 | 100% | AKT2 | 100% |
| ALG10B | 100% | ALPK3 | 100% | ALX4 | 100% | AMACR | 100% |
| ANGPTL6 | 99.3% | ANK2 | 100% | APC | 100% | APOA1 | 100% |
| APOA5 | 100% | APOB | 100% | APOC2 | 100% | APOE | 100% |
| APPL1 | 100% | AR | 100% | AREG | 100% | ARHGAP31 | 100% |
| ARHGEF18 | 100% | ARL2BP | 100% | ARL3 | 100% | ARL4D | 100% |
| ARL6 | 100% | ARMC5 | 100% | ARNT2 | 100% | ARVCF | 100% |
| ATM | 100% | AURKAIP1 | 100% | B2M | 100% | BANF1 | 100% |
| BAZ1B | 100% | BBIP1 | 100% | BBS1 | 100% | BBS10 | 100% |
| BBS12 | 100% | BBS2 | 100% | BBS4 | 100% | BBS5 | 100% |
| BBS7 | 100% | BBS9 | 100% | BEST1 | 100% | BLK | 100% |
| BLM | 100% | BMP2 | 100% | BMPR2 | 100% | BNC2 | 100% |
| BRCA1 | 100% | BRCA2 | 100% | BSCL2 | 100% | BTNL2 | 100% |




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| | | | | | | | |
|---|---|---|---|---|---|---|---|
| BVES | 100% | C1QA | 100% | C1R | 100% | C3 | 100% |
| CA4 | 100% | CABP4 | 100% | CACNA1C | 100% | CACNA1D | 100% |
| CACNA1F | 100% | CACNA1H | 98.7% | CACNA2D1 | 100% | CACNB2 | 100% |
| CALM1 | 100% | CALM2 | 100% | CALM3 | 100% | CALR | 100% |
| CASQ2 | 100% | CASR | 100% | CAT | 100% | CAV1 | 100% |
| CAV3 | 100% | CAVIN1 | 100% | CBS | 100% | CCDC28B | 100% |
| CCN2 | 100% | CCND1 | 100% | CCR6 | 100% | CD2AP | 100% |
| CD46 | 100% | CDH23 | 98.6% | CDHR1 | 100% | CDKN2A | 100% |
| CEL | 100% | CEP164 | 100% | CEP19 | 100% | CEP290 | 100% |
| CERKL | 100% | CFB | 100% | CFH | 100% | CFHR1 | 100% |
| CFHR3 | 100% | CFI | 100% | CFTR | 100% | CIDEC | 100% |
| CISD2 | 100% | CLCN2 | 100% | CLCNKB | 100% | CLIP2 | 99.9% |
| CLRN1 | 100% | CNBP | 100% | CNGB1 | 100% | CNOT1 | 100% |
| COL1A1 | 100% | COL3A1 | 100% | COL4A3 | 100% | COL4A4 | 100% |
| COL4A5 | 100% | COL5A1 | 98.6% | COL5A2 | 100% | COMT | 100% |
| COQ2 | 100% | COQ7 | 100% | CORIN | 100% | CP | 100% |
| CPA1 | 100% | CPOX | 100% | CRAT | 100% | CRB1 | 100% |
| CRELD1 | 100% | CRX | 100% | CTC1 | 100% | CTLA4 | 100% |
| CTNNB1 | 100% | CTNS | 100% | CTRC | 100% | CTSH | 100% |
| CUL3 | 100% | CXXC1 | 100% | CYP11B1 | 100% | CYP11B2 | 100% |
| CYP17A1 | 100% | CYP19A1 | 100% | CYP21A2 | 100% | CYP7A1 | 100% |
| DBH | 100% | DCAF17 | 100% | DDR2 | 100% | DEAF1 | 98.9% |
| DHDDS | 100% | DHX38 | 100% | DIS3L2 | 100% | DKC1 | 100% |
| DLEC1 | 100% | DLL1 | 100% | DLST | 100% | DNAJB11 | 100% |
| DNAJC21 | 88.6% | DNAJC3 | 100% | DNM1L | 100% | DPM3 | 100% |
| DSC2 | 100% | DSC3 | 100% | DSP | 100% | DSPP | 100% |
| DXO | 100% | DYRK1B | 100% | E2F1 | 83% | ECE1 | 100% |
| EDA | 100% | EDA2R | 100% | EFL1 | 100% | EFNA1 | 100% |





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|---|---|---|---|---|---|---|---|
| EGFR | 100% | EIF2AK3 | 99.9% | EIF2S3 | 100% | ELMO2 | 100% |
| ELN | 100% | ELP1 | 100% | EMD | 100% | ENG | 100% |
| ENPP1 | 97.1% | EPAS1 | 100% | ERCC4 | 100% | ERCC6 | 100% |
| ERCC8 | 100% | ERGIC1 | 100% | EXT2 | 100% | EYS | 100% |
| FAM161A | 100% | FBN1 | 100% | FECH | 100% | FGA | 100% |
| FGF8 | 100% | FGFR1 | 100% | FGFR2 | 100% | FH | 100% |
| FHL1 | 100% | FIG4 | 100% | FLII | 100% | FLT1 | 100% |
| FMO3 | 100% | FMR1 | 100% | FN1 | 100% | FOS | 100% |
| FOXC2 | 100% | FOXE3 | 82.3% | FOXF1 | 100% | FOXH1 | 100% |
| FOXP1 | 100% | FOXP3 | 100% | FOXRED1 | 100% | FSCN2 | 100% |
| FUZ | 100% | FXN | 100% | GANAB | 100% | GAS1 | 99.5% |
| GATA3 | 100% | GATA4 | 97.3% | GATA5 | 100% | GATA6 | 100% |
| GBA | 100% | GCGR | 100% | GCH1 | 100% | GCK | 100% |
| GDNF | 100% | GJA1 | 100% | GJB3 | 100% | GJB4 | 100% |
| GK | 100% | GLA | 100% | GLIS3 | 100% | GLRX5 | 100% |
| GLYAT | 100% | GNAS | 100% | GNAT1 | 100% | GP1BB | 86.4% |
| GPC3 | 100% | GPD1 | 100% | GPD1L | 100% | GPD2 | 100% |
| GPR101 | 100% | GPR179 | 100% | GPR35 | 100% | GRK1 | 100% |
| GRM6 | 93.3% | GTF2I | 100% | GTF2IRD1 | 100% | GUCA1B | 100% |
| GUCY1A1 | 100% | HAMP | 100% | HAVCR2 | 100% | HBB | 100% |
| HCN4 | 100% | HCRT | 100% | HERC2 | 100% | HESX1 | 100% |
| HFE | 100% | HGD | 100% | HGSNAT | 93.3% | HIRA | 100% |
| HJV | 100% | HLA-B | 100% | HMBS | 100% | HMGA1 | 100% |
| HMGA2 | 100% | HNF1A | 100% | HNF1B | 100% | HNF4A | 100% |
| HPSE2 | 100% | HSD11B2 | 83.9% | IDH3A | 100% | IDH3B | 100% |
| IDUA | 100% | IER3IP1 | 100% | IFIH1 | 100% | IFT140 | 100% |
| IFT172 | 100% | IFT27 | 100% | IFT88 | 100% | IGF1R | 100% |
| IGF2BP2 | 100% | IL12B | 100% | IL2RA | 100% | IL6 | 100% |



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|---|---|---|---|---|---|---|---|
| IMPDH1 | 100% | IMPG2 | 100% | INS | 100% | INSR | 99.7% |
| INVS | 100% | IQCB1 | 100% | IQSEC2 | 100% | IRAK1 | 100% |
| IRF5 | 100% | IRS1 | 100% | IRS2 | 95.2% | ITCH | 100% |
| ITGA8 | 100% | ITPR3 | 100% | IVNS1ABP | 100% | JAG1 | 100% |
| JAK2 | 100% | JMJD1C | 100% | JUP | 100% | KCNE1 | 100% |
| KCNE2 | 100% | KCNE3 | 100% | KCNE5 | 100% | KCNH2 | 100% |
| KCNJ11 | 100% | KCNJ2 | 100% | KCNJ5 | 100% | KCNJ8 | 100% |
| KCNQ1 | 97.8% | KCTD1 | 100% | KDSR | 100% | KIAA1549 | 96.7% |
| KIF1B | 100% | KIZ | 100% | KLF11 | 97.2% | KLHL3 | 100% |
| KLHL7 | 100% | KLRG1 | 100% | KRAS | 100% | KRT18 | 100% |
| KRT8 | 100% | LARS2 | 100% | LCAT | 100% | LCP1 | 100% |
| LDLR | 100% | LDLRAP1 | 100% | LEMD3 | 100% | LEP | 100% |
| LEPR | 100% | LEPROT | 100% | LHX1 | 100% | LIG4 | 100% |
| LIMK1 | 100% | LIPA | 100% | LIPC | 100% | LIPE | 100% |
| LMNA | 100% | LMNB2 | 96.7% | LMX1B | 100% | LOX | 100% |
| LPL | 100% | LRAT | 100% | LRIG2 | 100% | LRIT3 | 100% |
| LRP6 | 100% | LYZ | 100% | LZTFL1 | 100% | MAFA | 95.2% |
| MAFB | 100% | MAGEL2 | 100% | MAK | 100% | MAP4K2 | 100% |
| MAPK8IP1 | 100% | MAPRE2 | 100% | MAT2A | 100% | MAX | 100% |
| MC4R | 100% | MDH2 | 100% | MED12 | 100% | MEF2A | 100% |
| MEFV | 100% | MEN1 | 100% | MERTK | 100% | MFAP5 | 100% |
| MGP | 100% | MKKS | 100% | MKRN3 | 100% | MKS1 | 100% |
| MLX | 100% | MLXIPL | 100% | MMP14 | 100% | MMP2 | 100% |
| MOG | 100% | MPL | 100% | MST1 | 100% | MTNR1B | 100% |
| MTRR | 100% | MUC1 | 100% | MYBPC3 | 100% | MYH11 | 100% |
| MYH7 | 100% | MYL2 | 100% | MYLK | 100% | MYMK | 100% |
| MYO6 | 100% | NANOS3 | 100% | NAT8 | 100% | NDN | 100% |
| NDP | 100% | NDUFA1 | 100% | NDUFA11 | 100% | NDUFA6 | 100% |



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|---|---|---|---|---|---|---|---|
| NDUFAF1 | 100% | NDUFAF2 | 100% | NDUFAF3 | 100% | NDUFAF4 | 100% |
| NDUFAF5 | 100% | NDUFB10 | 100% | NDUFB11 | 100% | NDUFB3 | 100% |
| NDUFB9 | 100% | NDUFS1 | 100% | NDUFS3 | 100% | NDUFS4 | 100% |
| NDUFS6 | 100% | NDUFS7 | 100% | NDUFS8 | 100% | NDUFV1 | 100% |
| NDUFV2 | 100% | NEK2 | 100% | NEUROD1 | 100% | NEUROG3 | 100% |
| NF1 | 100% | NF2 | 100% | NFIX | 97.5% | NFU1 | 100% |
| NHP2 | 100% | NOD2 | 100% | NODAL | 100% | NOP10 | 100% |
| NOS1AP | 100% | NOS3 | 100% | NOTCH1 | 99.1% | NOTCH2 | 100% |
| NOTCH3 | 98.7% | NPAP1 | 100% | NPHP1 | 100% | NPHP3 | 100% |
| NPHP4 | 100% | NPM1 | 100% | NR2E3 | 99.9% | NR2F2 | 100% |
| NR3C1 | 100% | NR3C2 | 100% | NRL | 100% | NSMCE2 | 100% |
| NUBPL | 100% | NUDT19 | 92.5% | OFD1 | 100% | OPA1 | 100% |
| OSGEP | 100% | OTX2 | 100% | P2RY11 | 100% | PALB2 | 100% |
| PALLD | 100% | PAM16 | 100% | PARN | 100% | PAX4 | 100% |
| PCARE | 100% | PCNT | 100% | PCSK9 | 100% | PDE11A | 100% |
| PDE3A | 100% | PDE4D | 100% | PDE6A | 100% | PDE6B | 100% |
| PDE6G | 100% | PDE8B | 100% | PDGFB | 100% | PDHX | 100% |
| PDX1 | 100% | PEX1 | 100% | PEX10 | 100% | PEX6 | 100% |
| PHF21A | 100% | PHKA2 | 100% | PHKG2 | 100% | PIGH | 100% |
| PIGT | 100% | PIK3R1 | 100% | PKD1 | 98.4% | PKD2 | 95.3% |
| PKHD1 | 100% | PKP2 | 100% | PLAGL1 | 100% | PLCD1 | 100% |
| PLIN1 | 100% | PLN | 100% | PLVAP | 100% | PNPLA2 | 100% |
| PNPLA6 | 100% | POC1A | 100% | POLA1 | 100% | POLD1 | 100% |
| POLG | 100% | POLG2 | 100% | POLR3A | 100% | POMGNT1 | 100% |
| POR | 100% | POU3F4 | 100% | POU6F2 | 100% | PPARG | 100% |
| PPP1R15B | 100% | PPP1R3A | 100% | PRCD | 100% | PRF1 | 100% |
| PRKACA | 100% | PRKAR1A | 100% | PRKCA | 100% | PRKD2 | 100% |
| PRKG1 | 100% | PROC | 100% | PROK2 | 100% | PROKR2 | 100% |





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|---|---|---|---|---|---|---|---|
| PROM1 | 100% | PRPF3 | 100% | PRPF31 | 100% | PRPF4 | 100% |
| PRPF6 | 100% | PRPF8 | 100% | PRPH2 | 100% | PRSS1 | 100% |
| PRSS58 | 100% | PRTN3 | 100% | PSEN1 | 100% | PSEN2 | 100% |
| PSMB4 | 100% | PSMB8 | 100% | PSMB9 | 100% | PSTPIP1 | 100% |
| PTCH1 | 100% | PTF1A | 94.6% | PTGS1 | 100% | PTGS2 | 100% |
| PTPN1 | 100% | PTPN22 | 100% | PTRH2 | 100% | PYGL | 100% |
| RAC1 | 100% | RAI1 | 100% | RANGRF | 100% | RBP3 | 100% |
| RDH12 | 100% | RDH5 | 100% | REEP6 | 100% | REST | 100% |
| RET | 100% | RETN | 100% | RFC2 | 100% | RGR | 100% |
| RHO | 100% | RLBP1 | 100% | RNASE1 | 100% | RNASE12 | 100% |
| RNASEH2A | 100% | RNASEH2B | 100% | RNASEH2C | 100% | RNF125 | 100% |
| RNF6 | 100% | RNF7 | 100% | ROM1 | 100% | RP1 | 100% |
| RP2 | 100% | RP9 | 100% | RPE65 | 100% | RPGR | 100% |
| RPGRIP1L | 99.3% | RREB1 | 100% | RRM2B | 100% | RSPO1 | 100% |
| RTEL1 | 100% | RYR2 | 100% | SAG | 100% | SAMHD1 | 100% |
| SARS2 | 100% | SBDS | 100% | SCAPER | 100% | SCGB1D1 | 100% |
| SCN10A | 100% | SCN1B | 100% | SCN2B | 100% | SCN3B | 100% |
| SCN4A | 100% | SCN4B | 100% | SCN5A | 100% | SCNN1A | 100% |
| SCNN1B | 100% | SCNN1G | 100% | SDCCAG8 | 100% | SDHA | 100% |
| SDHAF2 | 100% | SDHB | 100% | SDHC | 100% | SDHD | 100% |
| SEC24C | 100% | SEMA4A | 100% | SERPINA6 | 100% | SGPL1 | 100% |
| SH2B3 | 100% | SHH | 100% | SIX3 | 100% | SLC12A3 | 100% |
| SLC16A2 | 100% | SLC19A2 | 100% | SLC24A1 | 96.3% | SLC25A11 | 100% |
| SLC25A13 | 100% | SLC25A4 | 100% | SLC29A3 | 100% | SLC2A10 | 100% |
| SLC2A2 | 100% | SLC30A8 | 100% | SLC37A4 | 100% | SLC52A2 | 100% |
| SLC52A3 | 100% | SLC7A14 | 100% | SMAD3 | 100% | SMAD4 | 100% |
| SMAD6 | 100% | SMARCAL1 | 100% | SMARCB1 | 100% | SMIM6 | 100% |
| SMPD1 | 100% | SNRNP200 | 100% | SNRPN | 100% | SNTA1 | 98.2% |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| SOAT1 | 100% | SOX2 | 100% | SOX3 | 100% | SPATA7 | 100% |
| SPP1 | 100% | SPRY2 | 100% | SRP54 | 100% | STAT1 | 100% |
| STAT3 | 100% | STAT4 | 100% | STK19 | 100% | STK4 | 100% |
| STOX1 | 95.2% | STUB1 | 100% | STX11 | 100% | STX16 | 100% |
| STXBP2 | 100% | SUFU | 100% | SUGCT | 100% | SYNE1 | 100% |
| SYNE2 | 100% | TADA2A | 100% | TBL2 | 100% | TCF4 | 100% |
| TCF7L2 | 100% | TDGF1 | 100% | TECRL | 100% | TERT | 100% |
| TET2 | 100% | TGFB2 | 100% | TGFB3 | 100% | TGFBR1 | 94.1% |
| TGFBR2 | 100% | TGFBR3 | 100% | TGIF1 | 100% | THBD | 100% |
| THPO | 100% | TIMMDC1 | 100% | TINF2 | 100% | TKT | 100% |
| TMEM126B | 100% | TMEM127 | 100% | TMEM237 | 100% | TMEM43 | 100% |
| TMEM67 | 100% | TMEM70 | 100% | TNFRSF11A | 95.4% | TNFRSF11B | 100% |
| TNFRSF1A | 100% | TNFSF4 | 100% | TNNC1 | 100% | TNNI3 | 100% |
| TOPORS | 100% | TP53 | 100% | TRAF3IP1 | 100% | TRAPPC9 | 100% |
| TRDN | 100% | TREX1 | 100% | TRIM28 | 95.5% | TRIM32 | 100% |
| TRIP13 | 100% | TRMT10A | 100% | TRPC6 | 100% | TRPM1 | 100% |
| TRPM4 | 100% | TSC1 | 100% | TSC2 | 100% | TTC7A | 100% |
| TTC8 | 100% | TTPA | 100% | TUB | 100% | TULP1 | 100% |
| TWNK | 100% | UBR1 | 100% | UFD1 | 100% | UNC13D | 100% |
| USB1 | 100% | USH2A | 100% | USP8 | 100% | VAC14 | 100% |
| VANGL1 | 100% | VDAC2 | 100% | VHL | 100% | WAS | 100% |
| WDPCP | 100% | WDR19 | 100% | WDR35 | 100% | WFS1 | 100% |
| WNK1 | 100% | WNK4 | 100% | WRAP53 | 100% | WRN | 100% |
| WT1 | 100% | WWOX | 100% | XIAP | 100% | XPNPEP3 | 100% |
| XRCC4 | 100% | XYLT1 | 93.4% | XYLT2 | 100% | YY1AP1 | 100% |
| ZBTB20 | 100% | ZFP57 | 100% | ZFPM1 | 86.5% | ZFYVE26 | 100% |
| ZIC2 | 100% | ZMPSTE24 | 100% | ZNF260 | 100% | ZNF365 | 100% |
| ZNF395 | 100% | ZNF408 | 100% | ZNF513 | 100% | | |



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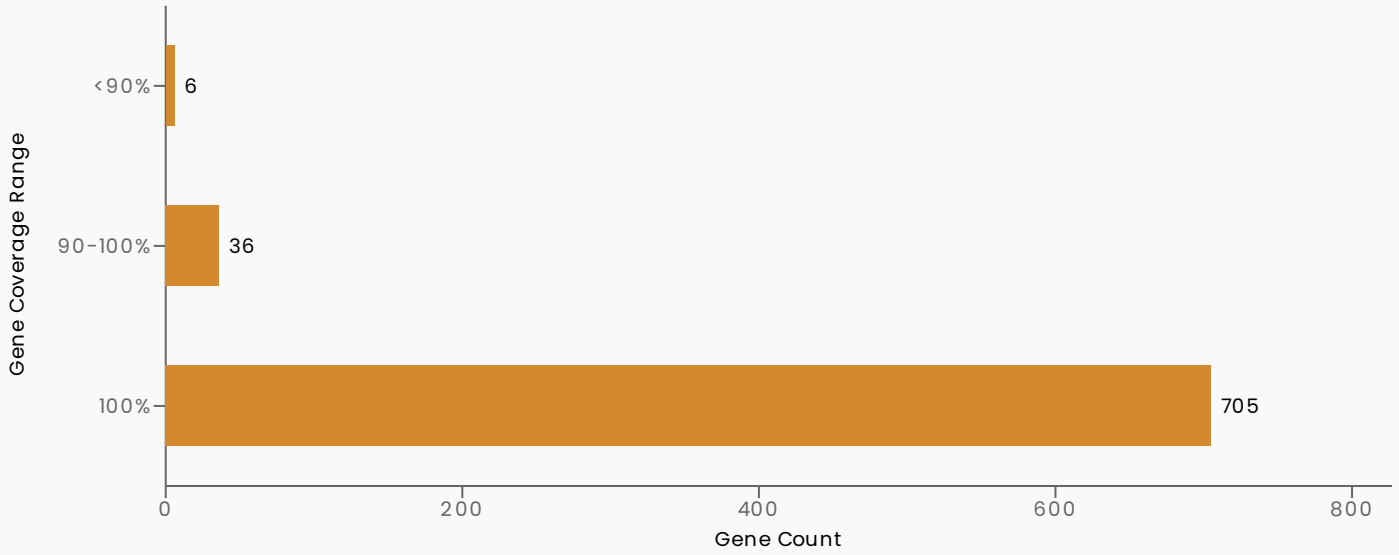
## Gene Coverage Distribution




## QC Metrics

| | |
|---|---|
| **Total Reads** | 76.58 (M) |
| **Total Aligned Reads** | 99.92 % |
| **Total data generated** | 12.00 (Gb) |
| **Total reads which passed mapping quality cut-off** | 10.93 (Gb) |

## Test Limitations

- Testing has been performed assuming that the sample received belongs to the above named individual(s) and any stated relationships between individuals are accepted as true. It is also assumed that consent for the same was provided after pre-test counseling at the point of collection/referral.
- The current results are based on analysis of coding regions (exons) as well as certain intron padding regions on patient's genomic DNA with respect to patient phenotype as defined in the target regions (link available below). However, due to inherent technology limitations, coverage is not uniform across all regions. Hence pathogenic variants present in areas of insufficient coverage as well as those variants which currently do not co-relate with the provided phenotype may not be analyzed/ reported. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity.
- The reported variants have not been Sanger confirmed. Sanger confirmation is recommended for the same.
- The test methodology currently does not detect large deletions/duplications, triplet repeat expansions and epigenetic changes. The test also does not include analysis of predictors for multifactorial, polygenic and/or complex diseases. Novel synonymous changes as well as intronic mutations (excluding those affecting invariant splice nucleotides) are not routinely reported.
- Phenotype variability may be due to modifying genetic/non-genetic factors and is not a part of the current analysis.

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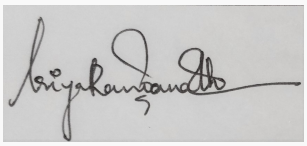
- The classification and interpretation of all the variants in this assay reflects the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific information comes to light. We recommend re- analysis of this report yearly, in order to take advantage of any new scientific data that may become available. Please contact laboratory in case re-analysis of the report is desired. It is the lab's policy to perform re-analysis once on a complimentary basis. However, this re-analysis is performed only when requested.

## Reviewed By

**Bijal Vyas PhD**
Analyzed by
Senior Genome Analyst



**Priya Ranganath**
Reviewed by
Clinical Geneticist

**Dr. Udhaya Kotecha**
Approved by
Head of Division-Inherited Genomics-NGS



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