

| | | |
|------------|-----------------------|-------------------------|
| Case ID | Sample Type | : EDTA PERIPHERAL BLOOD |
| Name | Date & Time Collected | : 10-May-2023 11:00 AM |
| Sex/Age | Date & Time Received | : 12-May-2023 03:29 PM |
| Bill. Loc. | Date & Time Reported | : 01-Jun-2023 07:01 PM |
| Ref. By | Report Version | : 1 |
| Indication | | |

Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platform

Clinical Indication:

A 10-month female child with the history of global developmental delayed, dysmorphism, hepatosplenomegaly, hypotonia. Consanguinity first cousin.

Pathogenic variant detected related to the clinical Phenotype

Key Findings:

| Gene & Transcript | Location | Variant | Zygoty/Inheritance | OMIM Phenotype | Clinical Significance |
|------------------------|----------|---------------------------------|---------------------------------------|-------------------------------|-----------------------|
| GLB1(-) NM_000404.4 | Exon 15 | c.1577dupG (p.Trp527Leufs*5) | Homozygous /Autosomal Recessive | GM1-gangliosidosis, type I | Pathogenic |

**Genetic test results are reported based on the recommendations of American College of Medical Genetics*

Variant Interpretation & Clinical correlation:

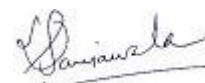
GLB1: c.1577dupG:

The submitted sample shows Homozygous variant in Exon 15 of gene GLB1 (chr3: g.33014212_33014213insC). A frameshift duplication "G" detected at nucleotide position 1577. This variant is predicted to cause loss of normal protein function through protein truncation caused a frameshift mutation. The frame shifted sequence continues 5 residues until a stop codon is reached. **In silico predictions:** The p.Trp527Leufs*5 variant is a loss of function variant in the gene GLB1, which is intolerant of Loss of Function variant. **Population frequency and Internal database:** This variant occurs in no individuals in a homozygous genotype state. The p.Trp527Leufs*5 variant is novel (not in any individuals) in 1kG All. This variant is absent in our internal database.


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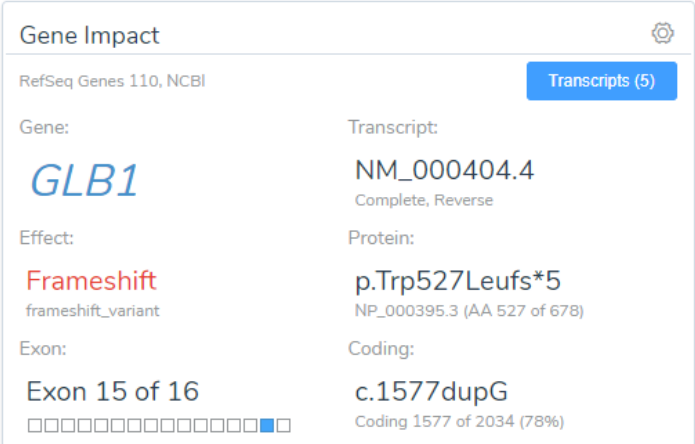
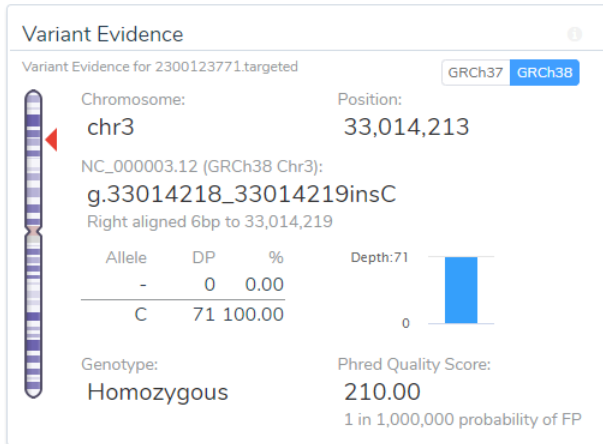
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Based on above evidence, this variant (GLB1: c.1577dupG) is classified as Pathogenic variant.

OMIM Phenotype:

GM1-gangliosidosis is an autosomal recessive lysosomal storage disease characterized by accumulation of ganglioside substrates in lysosomes. Clinically, patients show variable degrees of neurodegeneration and skeletal abnormalities. There are 3 main clinical variants categorized by severity and variable residual beta-galactosidase activity. Type I, or infantile form, shows rapid psychomotor deterioration beginning within 6 months of birth, generalized central nervous system involvement, hepatosplenomegaly, facial dysmorphism, macular cherry-red spots, skeletal dysplasia, and early death.

Case Specific Recommendations:

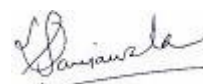
1. Segregation analysis of the variant by Sanger sequencing is recommended in affected and unaffected members of the family.
2. **Targeted mutation analysis of reported variant in parents.**

Recommendations

- Genetic counseling is advised for interpretation on the consequences of the variant(s).
- We recommend confirming the presence of these variants by Sanger Sequencing.
- Segregation analysis of the variant by Sanger sequencing is recommended in affected member and unaffected member of the family.
- The significance/classification of the variants might change based on parental and family members


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genetic testing.

- For questions about this report, or for assistance in locating nearby genetic counseling services, please contact the Laboratory: contact@sngenelab.com.
- If results obtained do not match the clinical findings, additional testing should be considered as per referring clinician's recommendation.

Methodology:

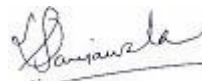
- Next Generation Sequencing: DNA extracted from Blood, Saliva, Amniotic fluid, CVS or any other standard source is used for targeted capture-based Library preparation. Targeted capture provides an efficient and sensitive means for sequencing specific genomic regions in a high-throughput manner. The libraries were sequenced to mean >85-100x coverage on Illumina Novaseq 6000 sequencing platform with Paired End 2x150 chemistry.
- We follow the GATK best practices framework for identification of variants in the sample. The sequences obtained are assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build (GRCh38).
- Haplotype caller has been used to identify variants which are relevant to the clinical indication.
- In addition to SNVs and small Indels, copy number variants (CNVs) are detected from targeted sequence data using the commercially available algorithm. This algorithm detects rare CNVs based on comparison of the read-depths of the test data with the matched aggregate reference dataset.
- Clinically relevant mutations were annotated using published variants in literature, Commercial datasets and a set of diseases databases. Common variants are filtered based on allele frequency in 1000Genome, ExAC, gnomAD, dbSNP & SN Genelab's internal database.
- Non-synonymous variants effect is calculated using multiple algorithms such as PolyPhen-2, SIFT, MutationTaster2.
- Based on annotations data, ACMG rules-based classification performed for classification of variants identified through next generation sequencing study.

Limitations/Disclaimer:

- It should be noted that this test does not sequence all bases in a human genome, not all variants have been identified or interpreted, and this report is limited only to variants with evidence for causing or contributing to disease/clinical details provided to SN Gene Lab Pvt Ltd.
- Testing has been performed assuming that the sample received belongs to the above named individual and any stated relationships between individuals are accepted as true.
- Test results are interpreted in the context of clinical findings, family history and other laboratory data. Only variations in genes potentially related to the proband's medical condition are reported. Rare

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polymorphisms may lead to false negative or positive results. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. The results should be interpreted in the context of the patient's medical evaluation, family history and racial/ethnic background. Please note that variant classification and/or interpretation may change over time if more information available. Re- interpretation of multi gene next generation sequencing data is recommended on an annual basis and may be requested by a medical provider.

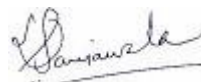
- The sensitivity of this assay to detect large deletions/duplications of more than 10 bp or copy number variations (CNV) is 70-75%. The CNVs detected with this assay have to be confirmed by alternate method such as MLPA & Microarray.
- Due to inherent technology limitations, coverage is not uniform 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.
- Triplet repeat expansions, translocations, large deletion or duplications and chromosomal rearrangements events are currently not reliably detected by next generation sequencing.
- This assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, other gene rearrangements like inversion or translocation and does not detect single or multi-exon deletions or duplications.
- Incidental or secondary findings (if any) that meet the ACMG guidelines can be given upon request.
- Genes with pseudogenes, paralog genes and genes with low complexity may have decreased sensitivity and specificity of variant detection and interpretation due to inability of the data and analysis tools to unambiguously determine the origin of the sequence data in such regions.
- Sequence and copy number variants are reported according to the Human Genome Variation Society (HGVS).
- The transcript used for clinical reporting generally represents the canonical transcript, which is usually the longest coding transcript with strong/multiple supporting evidence. However, clinically relevant variants annotated in alternate complete coding transcripts could also be reported.

Variant classification as per ACMG guidelines:

| | |
|---------|---|
| Variant | A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign). |
| Benign | A variant which is known not to be responsible for disease has been detected. Generally, no further action is warranted on such variants when detected. |

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|-----------------------------------|--|
| Likely Benign | A variant which is very unlikely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of Pathogenicity. |
| Pathogenic | A disease-causing variation in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed. |
| Likely Pathogenic | A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity. |
| Variant of Uncertain Significance | A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence. |

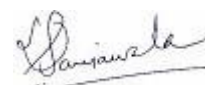
References:

1. Richards S, Aziz N, Bale 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, Genetics in Medicine, 2015 May;17(5):405-24
2. Kalia S.S. et al., Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update(ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genet Med., 19(2):249-255, 2017.
3. Meyer, L.R., et al., The UCSC Genome Browser database: extensions and updates 2013. Nucleic Acids Res, 2013. 41(D1): p.D64-9.
4. McKenna, A., et al., The Genome Analysis Toolkit: a MapReduce framework for analyzing next-generation DNA sequencing data. Genome Res, 2010. 20(9): p. 1297-303

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5. 1000 Genomes Project Consortium et al., A global reference for human genetic variation. *Nature*, 526(7571): 68-74, 2015.

6. Lek M. et al., Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, 536(7616):285-91, 2016.

7. McLaren, W., et al., Deriving the consequences of genomic variants with the Ensembl API and SNP Effect Predictor. *Bioinformatics*, 2010. 26(16): p. 2069-70.

Appendix 1: SAMPLE DATA AND STATISTICS

| Title | Data |
|---------------------------|--------|
| Total data generated (Gb) | 7.82 |
| Total Reads (%) | 100.00 |
| Total reads aligned (%) | 99.95 |
| Data >Q30 (%) | 94.24 |

Appendix 2: COVERAGE SUMMARY

| Mean Depth | Percentage target base pairs covered | | |
|------------|--------------------------------------|--------|--------|
| | 1x | 5x | 20x |
| 97.11x | 98.91% | 98.73% | 98.23% |

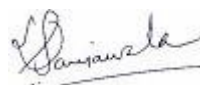
Appendix 3: COVERAGE OF ANALYZED GENES

| Gene | Region covered | Gene | Region covered | Gene | Region covered |
|----------|----------------|----------|----------------|----------|----------------|
| ABCC9 | 100.00% | ACAN | 100.00% | ACP5 | 100.00% |
| ACVR1 | 100.00% | ADAMTS10 | 100.00% | ADAMTS17 | 100.00% |
| ADAMTSL2 | 100.00% | AGA | 100.00% | AGPS | 100.00% |
| ALG12 | 100.00% | ALG3 | 100.00% | ALG9 | 100.00% |
| ALPL | 100.00% | ALX1 | 100.00% | ALX3 | 100.00% |
| ALX4 | 100.00% | AMER1 | 100.00% | ANKH | 100.00% |
| ANKRD11 | 100.00% | ANO5 | 100.00% | ANTXR2 | 100.00% |
| ARHGAP31 | 100.00% | ARL6 | 100.00% | ARSB | 100.00% |
| ASXL1 | 100.00% | ASXL2 | 100.00% | ATP6V0A2 | 100.00% |
| ATP7A | 100.00% | B3GALT6 | 100.00% | B3GAT3 | 100.00% |
| B3GLCT | 100.00% | B4GALT7 | 100.00% | BBS1 | 100.00% |
| BBS10 | 100.00% | BBS12 | 100.00% | BBS2 | 100.00% |
| BBS4 | 100.00% | BBS5 | 100.00% | BBS7 | 100.00% |

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|---------|---------|----------|---------|---------|---------|
| BBS9 | 100.00% | BHLHA9 | 100.00% | BMP1 | 100.00% |
| BMP2 | 100.00% | BMPER | 100.00% | BMPR1B | 100.00% |
| C2CD3 | 100.00% | CA2 | 100.00% | CANT1 | 100.00% |
| CASR | 100.00% | CC2D2A | 100.00% | CCDC8 | 100.00% |
| CDC45 | 100.00% | CDH3 | 100.00% | CDKN1C | 100.00% |
| CDT1 | 100.00% | CEP120 | 100.00% | CEP290 | 100.00% |
| CHST14 | 100.00% | CHST3 | 100.00% | CHSY1 | 100.00% |
| CLCN5 | 100.00% | CLCN7 | 100.00% | COG1 | 100.00% |
| COL10A1 | 100.00% | COL11A1 | 100.00% | COL11A2 | 100.00% |
| COL1A1 | 100.00% | COL1A2 | 100.00% | COL2A1 | 100.00% |
| COL9A1 | 100.00% | COL9A2 | 100.00% | COL9A3 | 100.00% |
| COLEC11 | 100.00% | COMP | 100.00% | CREB3L1 | 100.00% |
| CREBBP | 100.00% | CRTAP | 100.00% | CSPP1 | 100.00% |
| CTSA | 100.00% | CTSC | 100.00% | CTSK | 100.00% |
| CUL7 | 100.00% | CYP27B1 | 100.00% | CYP2R1 | 100.00% |
| DDR2 | 100.00% | DHCR24 | 100.00% | DHCR7 | 100.00% |
| DHODH | 100.00% | DIS3L2 | 100.00% | DLL3 | 100.00% |
| DLL4 | 100.00% | DLX3 | 100.00% | DLX5 | 100.00% |
| DMP1 | 100.00% | DNMT3A | 100.00% | DOCK6 | 100.00% |
| DPAGT1 | 100.00% | DPM1 | 100.00% | DSPP | 100.00% |
| DVL1 | 100.00% | DVL3 | 100.00% | DYM | 100.00% |
| DYNC2H1 | 100.00% | DYNC2LI1 | 100.00% | EBP | 100.00% |
| EED | 100.00% | EFTUD2 | 100.00% | EIF2AK3 | 100.00% |
| ENPP1 | 100.00% | EOGT | 100.00% | ERF | 100.00% |
| ESCO2 | 100.00% | EVC | 100.00% | EVC2 | 100.00% |
| EXT1 | 100.00% | EXT2 | 100.00% | EXTL3 | 100.00% |
| EZH2 | 100.00% | FAM111A | 100.00% | FAM20C | 100.00% |
| FBN1 | 100.00% | FBN2 | 100.00% | FERMT3 | 100.00% |
| FGF10 | 100.00% | FGF16 | 100.00% | FGF23 | 100.00% |
| FGFR1 | 100.00% | FGFR2 | 100.00% | FGFR3 | 100.00% |
| FIG4 | 100.00% | FKBP10 | 100.00% | FLNA | 100.00% |
| FLNB | 100.00% | FN1 | 100.00% | FUCA1 | 100.00% |
| FZD2 | 100.00% | GALNS | 100.00% | GALNT3 | 100.00% |
| GDF5 | 100.00% | GDF6 | 100.00% | GHR | 100.00% |
| GJA1 | 100.00% | GLB1 | 100.00% | GLI3 | 100.00% |
| GNAS | 100.00% | GNPAT | 100.00% | GNPTAB | 100.00% |
| GNPTG | 100.00% | GNS | 100.00% | GORAB | 100.00% |
| GPC6 | 100.00% | GSC | 100.00% | GUSB | 100.00% |

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| HDAC8 | 100.00% | HES7 | 100.00% | HGSNAT | 100.00% |
| HOXA13 | 100.00% | HOXD13 | 100.00% | HPGD | 100.00% |
| HSPG2 | 100.00% | IDH1 | 100.00% | IDS | 100.00% |
| IDUA | 100.00% | IFIH1 | 100.00% | IFITM5 | 100.00% |
| IFT122 | 100.00% | IFT140 | 100.00% | IFT172 | 100.00% |
| IFT43 | 100.00% | IFT52 | 100.00% | IFT80 | 100.00% |
| IFT81 | 100.00% | IHH | 100.00% | IKBKG | 100.00% |
| IL11RA | 100.00% | IL1RN | 100.00% | INPPL1 | 100.00% |
| KAT6B | 100.00% | KIAA0753 | 100.00% | KIF22 | 100.00% |
| KIF7 | 100.00% | KMT2D | 100.00% | LBR | 100.00% |
| LEMD3 | 100.00% | LIFR | 100.00% | LMBR1 | 100.00% |
| LMNA | 100.00% | LMX1B | 100.00% | LONP1 | 100.00% |
| LPIN2 | 100.00% | LRP4 | 100.00% | LRP5 | 100.00% |
| LTBP3 | 100.00% | MAFB | 100.00% | MAN2B1 | 100.00% |
| MAP3K7 | 100.00% | MASP1 | 100.00% | MATN3 | 100.00% |
| MEGF8 | 100.00% | MEOX1 | 100.00% | MESP2 | 100.00% |
| MGP | 100.00% | MKKS | 100.00% | MKS1 | 100.00% |
| MMP13 | 100.00% | MMP2 | 100.00% | MNX1 | 100.00% |
| MPDU1 | 100.00% | MSX2 | 100.00% | MYCN | 100.00% |
| NAGLU | 100.00% | NANS | 100.00% | NBAS | 100.00% |
| NEK1 | 100.00% | NEU1 | 100.00% | NF1 | 100.00% |
| NFIX | 100.00% | NIPBL | 100.00% | NKX3-2 | 100.00% |
| NLRP3 | 100.00% | NOG | 100.00% | NOTCH1 | 100.00% |
| NOTCH2 | 100.00% | NPR2 | 100.00% | NSD1 | 100.00% |
| NSDHL | 100.00% | OBSL1 | 100.00% | OFD1 | 100.00% |
| ORC1 | 100.00% | ORC4 | 100.00% | ORC6 | 100.00% |
| OSTM1 | 100.00% | P3H1 | 100.00% | P4HB | 100.00% |
| PAPSS2 | 100.00% | PAX3 | 100.00% | PCNT | 100.00% |
| PCYT1A | 100.00% | PDE3A | 100.00% | PDE4D | 100.00% |
| PEX5 | 100.00% | PEX7 | 100.00% | PGM3 | 100.00% |
| PHEX | 100.00% | PHGDH | 100.00% | PIGT | 100.00% |
| PIGV | 100.00% | PIK3C2A | 100.00% | PIK3R1 | 100.00% |
| PITX1 | 100.00% | PLOD2 | 100.00% | PLS3 | 100.00% |
| POC1A | 100.00% | POLR1A | 100.00% | POLR1C | 100.00% |
| POLR1D | 100.00% | POP1 | 100.00% | POR | 100.00% |
| PIIB | 100.00% | PRKAR1A | 100.00% | PRMT7 | 100.00% |
| PSAT1 | 100.00% | PSPH | 100.00% | PTDSS1 | 100.00% |
| PTH1R | 100.00% | PTHLH | 100.00% | PTPN11 | 100.00% |

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|-----------|---------|-----------|---------|------------|---------|
| PUF60 | 100.00% | PYCR1 | 100.00% | RAB23 | 100.00% |
| RAB33B | 100.00% | RASGRP2 | 100.00% | RBM8A | 100.00% |
| RBPJ | 100.00% | RECQL4 | 100.00% | RFT1 | 100.00% |
| RMRP | 100.00% | RNU4ATAC | 100.00% | ROR2 | 100.00% |
| RPGRIP1L | 96.50% | RPL13 | 72.64% | RUNX2 | 100.00% |
| SALL1 | 100.00% | SALL4 | 100.00% | SBDS | 100.00% |
| SCARF2 | 100.00% | SEC24D | 100.00% | SERPINF1 | 100.00% |
| SERPINH1 | 100.00% | SETD2 | 100.00% | SF3B4 | 100.00% |
| SFRP4 | 100.00% | SGSH | 100.00% | SH3BP2 | 100.00% |
| SH3PXD2B | 100.00% | SHOX | 200.00% | SKI | 100.00% |
| SLC10A7 | 100.00% | SLC17A5 | 100.00% | SLC26A2 | 100.00% |
| SLC29A3 | 100.00% | SLC34A1 | 100.00% | SLC34A3 | 100.00% |
| SLC35C1 | 100.00% | SLC35D1 | 100.00% | SLC39A13 | 100.00% |
| SLCO2A1 | 100.00% | SMAD3 | 100.00% | SMAD4 | 100.00% |
| SMARCAL1 | 100.00% | SMC1A | 100.00% | SMC3 | 100.00% |
| SMOC1 | 100.00% | SNRPB | 100.00% | SNX10 | 100.00% |
| SOST | 100.00% | SOX9 | 100.00% | SP7 | 100.00% |
| SPARC | 100.00% | SUMF1 | 100.00% | TALDO1 | 100.00% |
| TAPT1 | 100.00% | TBCE | 100.00% | TBX15 | 100.00% |
| TBX3 | 100.00% | TBX4 | 100.00% | TBX5 | 100.00% |
| TBX6 | 100.00% | TBXAS1 | 100.00% | TCIRG1 | 100.00% |
| TCOF1 | 100.00% | TCTN2 | 100.00% | TCTN3 | 100.00% |
| TERT | 100.00% | TGFB1 | 100.00% | TGFB2 | 100.00% |
| TGFBR2 | 100.00% | TMCO1 | 100.00% | TMEM165 | 100.00% |
| TMEM216 | 100.00% | TMEM231 | 100.00% | TMEM38B | 100.00% |
| TNFRSF11A | 100.00% | TNFRSF11B | 100.00% | TNFSF11 | 100.00% |
| TP63 | 100.00% | TRAPPC2 | 100.00% | TREM2 | 100.00% |
| TRIP11 | 100.00% | TRPS1 | 100.00% | TRPV4 | 100.00% |
| TRPV6 | 100.00% | TTC21B | 100.00% | TTC8 | 100.00% |
| TWIST1 | 100.00% | TYROBP | 100.00% | VDR | 100.00% |
| WDPCP | 100.00% | WDR19 | 100.00% | WDR35 | 100.00% |
| WNT1 | 100.00% | WNT10B | 100.00% | WNT5A | 100.00% |
| WNT7A | 100.00% | XRCC4 | 100.00% | XYLT1 | 100.00% |
| XYLT2 | 100.00% | YY1 | 100.00% | ZMPSTE24 | 100.00% |
| ZSWIM6 | 100.00% | ABL1 | 100.00% | ANAPC1 | 92.00% |
| B9D1 | 100.00% | COG4 | 100.00% | CSGALNACT1 | 100.00% |
| DCC | 100.00% | FBLN1 | 100.00% | GNPNAT1 | 100.00% |
| GPX4 | 100.00% | GZF1 | 100.00% | HDAC4 | 100.00% |

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Indication

| | | | | | |
|----------|---------|---------|---------|---------|---------|
| HNRNPK | 100.00% | HS2ST1 | 100.00% | KDELR2 | 100.00% |
| KIAA1217 | 100.00% | LTBP1 | 100.00% | MANBA | 100.00% |
| MBTPS1 | 100.00% | MIR17HG | 100.00% | MMP9 | 100.00% |
| MTX2 | 100.00% | NPR3 | 100.00% | NXN | 100.00% |
| PAM16 | 100.00% | PFN1 | 100.00% | PISD | 100.00% |
| PKDCC | 100.00% | PLEKHM1 | 100.00% | POLR1B | 100.00% |
| PRKG2 | 100.00% | RAD21 | 100.00% | RINT1 | 100.00% |
| SCUBE3 | 100.00% | SMAD6 | 100.00% | TONSL | 100.00% |
| UBA2 | 100.00% | UFSP2 | 100.00% | WBP11 | 93.04% |
| ACVR2B | 100.00% | ADGRV1 | 100.00% | ADI1 | 100.00% |
| AFF3 | 100.00% | AHI1 | 100.00% | AIPL1 | 100.00% |
| AKT1 | 100.00% | ARID1A | 100.00% | ARID1B | 100.00% |
| ARL13B | 100.00% | ATXN10 | 100.00% | B9D2 | 100.00% |
| BANF1 | 100.00% | CCDC28B | 100.00% | CCDC39 | 100.00% |
| CCDC40 | 100.00% | CD96 | 100.00% | CDC6 | 100.00% |
| CDH23 | 100.00% | CEP164 | 100.00% | CEP41 | 100.00% |
| CFTR | 100.00% | CKAP2L | 100.00% | CLRN1 | 100.00% |
| COL12A1 | 100.00% | COL5A1 | 100.00% | COLEC10 | 100.00% |
| CRB1 | 100.00% | CRELD1 | 100.00% | CRX | 100.00% |
| CYP26B1 | 100.00% | DACT1 | 100.00% | DLX6 | 89.68% |
| DNAAF1 | 100.00% | DNAAF2 | 100.00% | DNAAF3 | 100.00% |
| DNAH11 | 100.00% | DNAH5 | 100.00% | DNAI1 | 100.00% |
| DNAI2 | 100.00% | DNAL1 | 100.00% | DOLPP1 | 100.00% |
| DPM2 | 100.00% | DPM3 | 100.00% | EP300 | 100.00% |
| ESR1 | 100.00% | ETF1 | 100.00% | FBLIM1 | 56.77% |
| FBXW4 | 97.01% | FGF8 | 100.00% | FGF9 | 100.00% |
| FMN1 | 100.00% | FOXC1 | 100.00% | FOXH1 | 100.00% |
| GDF1 | 100.00% | GDF3 | 100.00% | GLIS2 | 100.00% |
| GREM1 | 100.00% | GUCY2D | 100.00% | HDAC5 | 100.00% |
| HOXA11 | 100.00% | HOXD11 | 100.00% | HYLS1 | 100.00% |
| IDH2 | 100.00% | IFT88 | 100.00% | IMPDH1 | 100.00% |
| INVS | 100.00% | IQCB1 | 100.00% | KCNJ13 | 100.00% |
| LCA5 | 100.00% | LEFTY2 | 98.82% | LFNG | 100.00% |
| LOXL3 | 100.00% | LRAT | 100.00% | LRP6 | 100.00% |
| LTBP2 | 100.00% | MAN2C1 | 100.00% | MCM5 | 100.00% |
| MIA3 | 100.00% | MMP14 | 100.00% | MTAP | 100.00% |
| MYO7A | 100.00% | NEK8 | 100.00% | NIN | 100.00% |
| NKX2-5 | 100.00% | NME8 | 100.00% | NODAL | 100.00% |

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|---------|---------|----------|---------|---------|---------|
| NPHP1 | 100.00% | NPHP3 | 100.00% | NPHP4 | 100.00% |
| NPPC | 100.00% | OAT | 100.00% | PCDH15 | 100.00% |
| PHF6 | 100.00% | PIK3CA | 100.00% | PIN1 | 100.00% |
| PIR | 100.00% | PKD2 | 100.00% | PKHD1 | 100.00% |
| PLCB3 | 100.00% | PLK4 | 100.00% | PLOD1 | 100.00% |
| PTPRQ | 100.00% | RAB3GAP2 | 100.00% | RD3 | 100.00% |
| RDH12 | 100.00% | RIPPLY2 | 100.00% | RPE65 | 100.00% |
| RPGR | 100.00% | RPGRIPI | 100.00% | RSPH4A | 100.00% |
| RSPH9 | 100.00% | SCNN1A | 100.00% | SCNN1B | 100.00% |
| SCNN1G | 100.00% | SDCCAG8 | 100.00% | SEM1 | 100.00% |
| SHH | 100.00% | SLCO5A1 | 100.00% | SMARCA2 | 100.00% |
| SMARCA4 | 100.00% | SMARCB1 | 100.00% | SMARCE1 | 100.00% |
| SOX11 | 100.00% | SPATA7 | 100.00% | SPECC1L | 100.00% |
| SULF1 | 100.00% | TCTN1 | 100.00% | TDP2 | 100.00% |
| TGDS | 100.00% | THPO | 100.00% | TMEM138 | 100.00% |
| TMEM237 | 100.00% | TMEM67 | 100.00% | TNXB | 100.00% |
| TOPORS | 100.00% | TRIM32 | 100.00% | TRMT10A | 100.00% |
| TSC1 | 100.00% | TSC2 | 100.00% | TULP1 | 100.00% |
| UMOD | 100.00% | USH1C | 100.00% | USH1G | 100.00% |
| USH2A | 100.00% | USP9X | 100.00% | VAC14 | 100.00% |
| VHL | 100.00% | WHRN | 100.00% | WNT3 | 100.00% |
| WRN | 100.00% | XPNPEP3 | 100.00% | ZBTB16 | 100.00% |
| ZIC3 | 100.00% | ZNF423 | 100.00% | ARCN1 | 100.00% |
| EFNB1 | 100.00% | EN1 | 100.00% | HHAT | 100.00% |
| IGF1R | 100.00% | KAT6A | 100.00% | MYO18B | 100.00% |
| NEPRO | 100.00% | NMNAT1 | 100.00% | TCF12 | 100.00% |
| TGFBR1 | 100.00% | TMEM251 | 100.00% | TWIST2 | 100.00% |
| ZIC1 | 100.00% | HUWE1 | 100.00% | CTCF | 100.00% |
| RAI1 | 100.00% | CHD2 | 100.00% | WDR45 | 100.00% |
| CTNNB1 | 100.00% | VPS13B | 100.00% | DDX3X | 100.00% |
| DYNC1H1 | 100.00% | DYRK1A | 100.00% | FOXG1 | 100.00% |
| MYT1L | 100.00% | IQSEC2 | 100.00% | POGZ | 100.00% |
| SATB2 | 100.00% | MED13L | 100.00% | ADNP | 100.00% |
| MTOR | 100.00% | EBF3 | 100.00% | GABRA1 | 100.00% |
| GABRB3 | 100.00% | NALCN | 100.00% | AUTS2 | 100.00% |
| FOXP1 | 100.00% | AHDC1 | 100.00% | GNAO1 | 100.00% |
| CHAMP1 | 100.00% | KANSL1 | 100.00% | GRIN2B | 100.00% |
| HNRNPU | 100.00% | ITPR1 | 100.00% | KCNB1 | 100.00% |

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|---------|---------|---------|---------|----------|---------|
| KCNQ2 | 100.00% | MECP2 | 100.00% | MEF2C | 100.00% |
| KMT2A | 100.00% | ATP1A3 | 100.00% | OPHN1 | 100.00% |
| WAC | 100.00% | ATRX | 100.00% | KIF1A | 100.00% |
| SETD5 | 100.00% | PPP2R5D | 100.00% | CHD7 | 100.00% |
| PACS1 | 100.00% | ASH1L | 100.00% | ZC4H2 | 100.00% |
| PTEN | 100.00% | CHD8 | 100.00% | PURA | 100.00% |
| ACTB | 100.00% | RIT1 | 100.00% | RPS6KA3 | 100.00% |
| SCN1A | 100.00% | SCN2A | 100.00% | SCN8A | 100.00% |
| SLC2A1 | 100.00% | SLC6A1 | 100.00% | SPTAN1 | 100.00% |
| BRAF | 100.00% | CDKL5 | 100.00% | STXBP1 | 100.00% |
| TCF4 | 100.00% | NR2F1 | 100.00% | TRIO | 100.00% |
| UBE3A | 100.00% | KDM6A | 100.00% | CACNA1A | 100.00% |
| BRPF1 | 100.00% | TBL1XR1 | 100.00% | EHMT1 | 100.00% |
| ASXL3 | 100.00% | KDM5C | 100.00% | NAA10 | 100.00% |
| SHANK3 | 100.00% | CASK | 100.00% | CDK13 | 100.00% |
| SYNGAP1 | 100.00% | ZEB2 | 100.00% | MED12 | 100.00% |
| AAAS | 100.00% | AARS2 | 100.00% | AASS | 100.00% |
| ABAT | 100.00% | ABCA1 | 100.00% | ABCB11 | 100.00% |
| ABCB4 | 100.00% | ABCB7 | 100.00% | ABCD1 | 100.00% |
| ABCD4 | 100.00% | ABCG5 | 100.00% | ABCG8 | 100.00% |
| ABHD12 | 100.00% | ABHD5 | 100.00% | ACAD8 | 100.00% |
| ACAD9 | 100.00% | ACADM | 100.00% | ACADS | 100.00% |
| ACADSB | 100.00% | ACADVL | 100.00% | ACAT1 | 100.00% |
| ACO2 | 100.00% | ACOX1 | 100.00% | ACSF3 | 100.00% |
| ACSL4 | 100.00% | ACTA1 | 100.00% | ACTG1 | 100.00% |
| ACTL6A | 100.00% | ACTL6B | 100.00% | ACTN2 | 100.00% |
| ACY1 | 100.00% | ADA | 100.00% | ADAR | 100.00% |
| ADAT3 | 100.00% | ADGRG1 | 100.00% | ADK | 100.00% |
| ADSL | 100.00% | AFF2 | 100.00% | AFF4 | 100.00% |
| AFG3L2 | 100.00% | AGK | 100.00% | AGL | 100.00% |
| AGRN | 100.00% | AGXT | 100.00% | AIFM1 | 100.00% |
| AIMP1 | 100.00% | AKR1D1 | 100.00% | AKT3 | 100.00% |
| ALAD | 100.00% | ALAS2 | 100.00% | ALDH18A1 | 100.00% |
| ALDH3A2 | 100.00% | ALDH4A1 | 100.00% | ALDH5A1 | 100.00% |
| ALDH6A1 | 100.00% | ALDH7A1 | 100.00% | ALDOA | 100.00% |
| ALDOB | 100.00% | ALG1 | 100.00% | ALG11 | 100.00% |
| ALG13 | 100.00% | ALG14 | 100.00% | ALG2 | 100.00% |
| ALG6 | 100.00% | ALG8 | 100.00% | ALKBH8 | 100.00% |

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|----------|---------|----------|---------|----------|---------|
| ALMS1 | 100.00% | AMACR | 100.00% | AMN | 100.00% |
| AMPD2 | 100.00% | AMT | 100.00% | ANO10 | 100.00% |
| AP1S1 | 100.00% | AP1S2 | 100.00% | AP2M1 | 99.92% |
| AP3B1 | 100.00% | AP3B2 | 100.00% | AP4B1 | 100.00% |
| AP4E1 | 100.00% | AP4M1 | 100.00% | AP4S1 | 100.00% |
| APC2 | 100.00% | APOA1 | 100.00% | APOA5 | 100.00% |
| APOB | 100.00% | APOC2 | 100.00% | APOE | 100.00% |
| APRT | 100.00% | APTX | 100.00% | AR | 100.00% |
| ARFGEF2 | 100.00% | ARG1 | 100.00% | ARHGEF9 | 100.00% |
| ARID2 | 100.00% | ARMC9 | 100.00% | ARSA | 100.00% |
| ARV1 | 100.00% | ARX | 100.00% | ASAH1 | 100.00% |
| ASL | 100.00% | ASNS | 100.00% | ASPA | 100.00% |
| ASPM | 100.00% | ASS1 | 100.00% | ATAD3A | 100.00% |
| ATIC | 100.00% | ATL1 | 100.00% | ATM | 100.00% |
| ATN1 | 100.00% | ATP13A2 | 100.00% | ATP1A1 | 100.00% |
| ATP6AP1 | 100.00% | ATP6AP2 | 100.00% | ATP6V1A | 100.00% |
| ATP6V1B2 | 100.00% | ATP7B | 100.00% | ATP8A2 | 100.00% |
| ATP8B1 | 100.00% | ATPAF2 | 100.00% | ATR | 100.00% |
| AUH | 100.00% | B3GALNT2 | 100.00% | B4GALNT1 | 100.00% |
| B4GALT1 | 100.00% | B4GAT1 | 100.00% | BAAT | 100.00% |
| BAG3 | 100.00% | BCAP31 | 100.00% | BCAT2 | 100.00% |
| BCKDHA | 100.00% | BCKDHB | 100.00% | BCKDK | 100.00% |
| BCL11A | 100.00% | BCL11B | 100.00% | BCOR | 100.00% |
| BCS1L | 100.00% | BICD2 | 100.00% | BIN1 | 100.00% |
| BLM | 100.00% | BMP4 | 100.00% | BOLA3 | 100.00% |
| BPTF | 100.00% | BRAT1 | 100.00% | BRD4 | 100.00% |
| BRF1 | 100.00% | BRSK2 | 100.00% | BRWD3 | 100.00% |
| BSCL2 | 100.00% | BTD | 100.00% | BUB1B | 100.00% |
| C12orf4 | 100.00% | C12orf57 | 100.00% | C19orf12 | 100.00% |
| C1QBP | 100.00% | CA5A | 100.00% | CA8 | 100.00% |
| CACNA1B | 99.94% | CACNA1C | 100.00% | CACNA1D | 100.00% |
| CACNA1E | 100.00% | CACNA1G | 100.00% | CACNA1S | 100.00% |
| CAD | 100.00% | CAMK2A | 100.00% | CAMK2B | 100.00% |
| CAMTA1 | 100.00% | CAPN3 | 100.00% | CARS2 | 100.00% |
| CAT | 100.00% | CAV3 | 100.00% | CBL | 100.00% |
| CBS | 100.00% | CC2D1A | 100.00% | CCBE1 | 100.00% |
| CCDC115 | 100.00% | CCDC22 | 100.00% | CCDC47 | 100.00% |
| CCDC78 | 100.00% | CCDC88C | 100.00% | CCND2 | 100.00% |

Dr. Pratap N. Mukhopadhyaya
Ph.D(Sc.)

Dr. Nirmal A. Vaniawala
MD (Path. & Bact.)

Dr. Salil Vaniawala
M.Sc Ph.D.(Human Genetics)
Consulting Geneticist

Case ID

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|---------|---------|----------|---------|--------|---------|
| CDC42 | 100.00% | CDH11 | 100.00% | CDH15 | 100.00% |
| CDK10 | 100.00% | CDK5RAP2 | 100.00% | CDK8 | 100.00% |
| CDON | 100.00% | CENPF | 100.00% | CENPJ | 100.00% |
| CEP135 | 100.00% | CEP152 | 100.00% | CEP57 | 100.00% |
| CEP83 | 100.00% | CFL2 | 100.00% | CHAT | 100.00% |
| CHCHD10 | 100.00% | CHD3 | 100.00% | CHD4 | 100.00% |
| CHKB | 100.00% | CHMP1A | 100.00% | CHRNA1 | 100.00% |
| CHRN1 | 100.00% | CHRN2 | 100.00% | CHRNE | 100.00% |
| CHRNA1 | 100.00% | CHRNA2 | 100.00% | CHRNE | 100.00% |
| CHRNA2 | 100.00% | CHRNA3 | 100.00% | CHRNE | 100.00% |
| CHRNA3 | 100.00% | CHRNA4 | 100.00% | CHRNE | 100.00% |
| CHRNA4 | 100.00% | CHRNA5 | 100.00% | CHRNE | 100.00% |
| CHRNA5 | 100.00% | CHRNA6 | 100.00% | CHRNE | 100.00% |
| CHRNA6 | 100.00% | CHRNA7 | 100.00% | CHRNE | 100.00% |
| CHRNA7 | 100.00% | CHRNA8 | 100.00% | CHRNE | 100.00% |
| CHRNA8 | 100.00% | CHRNA9 | 100.00% | CHRNE | 100.00% |
| CHRNA9 | 100.00% | CHRNA10 | 100.00% | CHRNE | 100.00% |
| CHRNA10 | 100.00% | CHRNA11 | 100.00% | CHRNE | 100.00% |
| CHRNA11 | 100.00% | CHRNA12 | 100.00% | CHRNE | 100.00% |
| CHRNA12 | 100.00% | CHRNA13 | 100.00% | CHRNE | 100.00% |
| CHRNA13 | 100.00% | CHRNA14 | 100.00% | CHRNE | 100.00% |
| CHRNA14 | 100.00% | CHRNA15 | 100.00% | CHRNE | 100.00% |
| CHRNA15 | 100.00% | CHRNA16 | 100.00% | CHRNE | 100.00% |
| CHRNA16 | 100.00% | CHRNA17 | 100.00% | CHRNE | 100.00% |
| CHRNA17 | 100.00% | CHRNA18 | 100.00% | CHRNE | 100.00% |
| CHRNA18 | 100.00% | CHRNA19 | 100.00% | CHRNE | 100.00% |
| CHRNA19 | 100.00% | CHRNA20 | 100.00% | CHRNE | 100.00% |
| CHRNA20 | 100.00% | CHRNA21 | 100.00% | CHRNE | 100.00% |
| CHRNA21 | 100.00% | CHRNA22 | 100.00% | CHRNE | 100.00% |
| CHRNA22 | 100.00% | CHRNA23 | 100.00% | CHRNE | 100.00% |
| CHRNA23 | 100.00% | CHRNA24 | 100.00% | CHRNE | 100.00% |
| CHRNA24 | 100.00% | CHRNA25 | 100.00% | CHRNE | 100.00% |
| CHRNA25 | 100.00% | CHRNA26 | 100.00% | CHRNE | 100.00% |
| CHRNA26 | 100.00% | CHRNA27 | 100.00% | CHRNE | 100.00% |
| CHRNA27 | 100.00% | CHRNA28 | 100.00% | CHRNE | 100.00% |
| CHRNA28 | 100.00% | CHRNA29 | 100.00% | CHRNE | 100.00% |
| CHRNA29 | 100.00% | CHRNA30 | 100.00% | CHRNE | 100.00% |
| CHRNA30 | 100.00% | CHRNA31 | 100.00% | CHRNE | 100.00% |
| CHRNA31 | 100.00% | CHRNA32 | 100.00% | CHRNE | 100.00% |
| CHRNA32 | 100.00% | CHRNA33 | 100.00% | CHRNE | 100.00% |
| CHRNA33 | 100.00% | CHRNA34 | 100.00% | CHRNE | 100.00% |
| CHRNA34 | 100.00% | CHRNA35 | 100.00% | CHRNE | 100.00% |
| CHRNA35 | 100.00% | CHRNA36 | 100.00% | CHRNE | 100.00% |
| CHRNA36 | 100.00% | CHRNA37 | 100.00% | CHRNE | 100.00% |
| CHRNA37 | 100.00% | CHRNA38 | 100.00% | CHRNE | 100.00% |
| CHRNA38 | 100.00% | CHRNA39 | 100.00% | CHRNE | 100.00% |
| CHRNA39 | 100.00% | CHRNA40 | 100.00% | CHRNE | 100.00% |
| CHRNA40 | 100.00% | CHRNA41 | 100.00% | CHRNE | 100.00% |
| CHRNA41 | 100.00% | CHRNA42 | 100.00% | CHRNE | 100.00% |
| CHRNA42 | 100.00% | CHRNA43 | 100.00% | CHRNE | 100.00% |
| CHRNA43 | 100.00% | CHRNA44 | 100.00% | CHRNE | 100.00% |
| CHRNA44 | 100.00% | CHRNA45 | 100.00% | CHRNE | 100.00% |
| CHRNA45 | 100.00% | CHRNA46 | 100.00% | CHRNE | 100.00% |
| CHRNA46 | 100.00% | CHRNA47 | 100.00% | CHRNE | 100.00% |
| CHRNA47 | 100.00% | CHRNA48 | 100.00% | CHRNE | 100.00% |
| CHRNA48 | 100.00% | CHRNA49 | 100.00% | CHRNE | 100.00% |
| CHRNA49 | 100.00% | CHRNA50 | 100.00% | CHRNE | 100.00% |
| CHRNA50 | 100.00% | CHRNA51 | 100.00% | CHRNE | 100.00% |
| CHRNA51 | 100.00% | CHRNA52 | 100.00% | CHRNE | 100.00% |
| CHRNA52 | 100.00% | CHRNA53 | 100.00% | CHRNE | 100.00% |
| CHRNA53 | 100.00% | CHRNA54 | 100.00% | CHRNE | 100.00% |
| CHRNA54 | 100.00% | CHRNA55 | 100.00% | CHRNE | 100.00% |
| CHRNA55 | 100.00% | CHRNA56 | 100.00% | CHRNE | 100.00% |
| CHRNA56 | 100.00% | CHRNA57 | 100.00% | CHRNE | 100.00% |
| CHRNA57 | 100.00% | CHRNA58 | 100.00% | CHRNE | 100.00% |
| CHRNA58 | 100.00% | CHRNA59 | 100.00% | CHRNE | 100.00% |
| CHRNA59 | 100.00% | CHRNA60 | 100.00% | CHRNE | 100.00% |
| CHRNA60 | 100.00% | CHRNA61 | 100.00% | CHRNE | 100.00% |
| CHRNA61 | 100.00% | CHRNA62 | 100.00% | CHRNE | 100.00% |
| CHRNA62 | 100.00% | CHRNA63 | 100.00% | CHRNE | 100.00% |
| CHRNA63 | 100.00% | CHRNA64 | 100.00% | CHRNE | 100.00% |
| CHRNA64 | 100.00% | CHRNA65 | 100.00% | CHRNE | 100.00% |
| CHRNA65 | 100.00% | CHRNA66 | 100.00% | CHRNE | 100.00% |
| CHRNA66 | 100.00% | CHRNA67 | 100.00% | CHRNE | 100.00% |
| CHRNA67 | 100.00% | CHRNA68 | 100.00% | CHRNE | 100.00% |
| CHRNA68 | 100.00% | CHRNA69 | 100.00% | CHRNE | 100.00% |
| CHRNA69 | 100.00% | CHRNA70 | 100.00% | CHRNE | 100.00% |
| CHRNA70 | 100.00% | CHRNA71 | 100.00% | CHRNE | 100.00% |
| CHRNA71 | 100.00% | CHRNA72 | 100.00% | CHRNE | 100.00% |
| CHRNA72 | 100.00% | CHRNA73 | 100.00% | CHRNE | 100.00% |
| CHRNA73 | 100.00% | CHRNA74 | 100.00% | CHRNE | 100.00% |
| CHRNA74 | 100.00% | CHRNA75 | 100.00% | CHRNE | 100.00% |
| CHRNA75 | 100.00% | CHRNA76 | 100.00% | CHRNE | 100.00% |
| CHRNA76 | 100.00% | CHRNA77 | 100.00% | CHRNE | 100.00% |
| CHRNA77 | 100.00% | CHRNA78 | 100.00% | CHRNE | 100.00% |
| CHRNA78 | 100.00% | CHRNA79 | 100.00% | CHRNE | 100.00% |
| CHRNA79 | 100.00% | CHRNA80 | 100.00% | CHRNE | 100.00% |
| CHRNA80 | 100.00% | CHRNA81 | 100.00% | CHRNE | 100.00% |
| CHRNA81 | 100.00% | CHRNA82 | 100.00% | CHRNE | 100.00% |
| CHRNA82 | 100.00% | CHRNA83 | 100.00% | CHRNE | 100.00% |
| CHRNA83 | 100.00% | CHRNA84 | 100.00% | CHRNE | 100.00% |
| CHRNA84 | 100.00% | CHRNA85 | 100.00% | CHRNE | 100.00% |
| CHRNA85 | 100.00% | CHRNA86 | 100.00% | CHRNE | 100.00% |
| CHRNA86 | 100.00% | CHRNA87 | 100.00% | CHRNE | 100.00% |
| CHRNA87 | 100.00% | CHRNA88 | 100.00% | CHRNE | 100.00% |
| CHRNA88 | 100.00% | CHRNA89 | 100.00% | CHRNE | 100.00% |
| CHRNA89 | 100.00% | CHRNA90 | 100.00% | CHRNE | 100.00% |
| CHRNA90 | 100.00% | CHRNA91 | 100.00% | CHRNE | 100.00% |
| CHRNA91 | 100.00% | CHRNA92 | 100.00% | CHRNE | 100.00% |
| CHRNA92 | 100.00% | CHRNA93 | 100.00% | CHRNE | 100.00% |
| CHRNA93 | 100.00% | CHRNA94 | 100.00% | CHRNE | 100.00% |
| CHRNA94 | 100.00% | CHRNA95 | 100.00% | CHRNE | 100.00% |
| CHRNA95 | 100.00% | CHRNA96 | 100.00% | CHRNE | 100.00% |
| CHRNA96 | 100.00% | CHRNA97 | 100.00% | CHRNE | 100.00% |
| CHRNA97 | 100.00% | CHRNA98 | 100.00% | CHRNE | 100.00% |
| CHRNA98 | 100.00% | CHRNA99 | 100.00% | CHRNE | 100.00% |
| CHRNA99 | 100.00% | CHRNA100 | 100.00% | CHRNE | 100.00% |

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Dr. Nirmal A. Vaniawala
MD (Path. & Bact.)


Dr. Salil Vaniawala
M.Sc Ph.D.(Human Genetics)
Consulting Geneticist

Dr. Pratap N. Mukhopadhyaya
Ph.D(Sc.)

Case ID

Sample Type : EDTA PERIPHERAL BLOOD

Name

Date & Time Collected : 10-May-2023 11:00 AM

Sex/Age

Date & Time Received : 12-May-2023 03:29 PM

Bill. Loc.

Date & Time Reported : 01-Jun-2023 07:01 PM

Ref. By

Report Version : 1

Indication

| | | | | | |
|---------|---------|---------|---------|---------|---------|
| DCX | 100.00% | DCXR | 100.00% | DDC | 100.00% |
| DDHD2 | 100.00% | DDOST | 100.00% | DDX11 | 100.00% |
| DDX59 | 100.00% | DDX6 | 100.00% | DEAF1 | 100.00% |
| DEGS1 | 100.00% | DEPDC5 | 100.00% | DES | 100.00% |
| DGUOK | 100.00% | DHDDS | 100.00% | DHFR | 100.00% |
| DHPS | 100.00% | DHTKD1 | 100.00% | DHX30 | 100.00% |
| DIAPH1 | 100.00% | DKC1 | 100.00% | DLAT | 100.00% |
| DLG | 100.00% | DLG3 | 100.00% | DLG4 | 100.00% |
| DMD | 100.00% | DMXL2 | 100.00% | DNA2 | 100.00% |
| DNAJB6 | 100.00% | DNAJC12 | 100.00% | DNAJC19 | 100.00% |
| DNAJC5 | 100.00% | DNM1 | 100.00% | DNM1L | 100.00% |
| DNM2 | 100.00% | DNMT3B | 100.00% | DOCK3 | 100.00% |
| DOCK7 | 100.00% | DOCK8 | 100.00% | DOK7 | 100.00% |
| DOLK | 100.00% | DPF2 | 100.00% | DPH1 | 100.00% |
| DPP6 | 100.00% | DPYD | 100.00% | DPYS | 100.00% |
| DYSF | 100.00% | EARS2 | 100.00% | ECEL1 | 100.00% |
| ECHS1 | 100.00% | EEF1A2 | 100.00% | EIF2S3 | 100.00% |
| EIF3F | 59.68% | EIF4A3 | 100.00% | ELAC2 | 100.00% |
| ELOVL4 | 100.00% | ELP2 | 100.00% | EMC1 | 100.00% |
| EMD | 100.00% | EML1 | 100.00% | EMX2 | 100.00% |
| ENO3 | 100.00% | EPG5 | 100.00% | EPM2A | 100.00% |
| ERCC1 | 100.00% | ERCC2 | 100.00% | ERCC3 | 100.00% |
| ERCC5 | 100.00% | ERCC6 | 100.00% | ERCC6L2 | 100.00% |
| ERCC8 | 100.00% | ERLIN2 | 100.00% | ETFA | 100.00% |
| ETFB | 100.00% | ETFDH | 100.00% | ETHE1 | 100.00% |
| EXOSC3 | 100.00% | FA2H | 100.00% | FAH | 100.00% |
| FAM126A | 100.00% | FAR1 | 100.00% | FARS2 | 100.00% |
| FASTKD2 | 100.00% | FAT4 | 100.00% | FBP1 | 100.00% |
| FBXL3 | 100.00% | FBXL4 | 100.00% | FBXO11 | 100.00% |
| FBXW11 | 100.00% | FDX2 | 100.00% | FDXR | 100.00% |
| FECH | 100.00% | FGD1 | 100.00% | FGF12 | 100.00% |
| FH | 100.00% | FHL1 | 100.00% | FKBP14 | 100.00% |
| FKRP | 100.00% | FKTN | 100.00% | FLAD1 | 100.00% |
| FLNC | 100.00% | FLVCR1 | 100.00% | FLVCR2 | 100.00% |
| FMN2 | 100.00% | FMO3 | 100.00% | FMR1 | 100.00% |
| FOLR1 | 100.00% | FOXP2 | 100.00% | FOXRED1 | 100.00% |
| FRMPD4 | 100.00% | FTCD | 100.00% | FTSJ1 | 100.00% |
| FUT8 | 100.00% | FXN | 100.00% | FXR1 | 96.30% |

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|---------|---------|----------|---------|---------|---------|
| G6PC3 | 100.00% | GAA | 100.00% | GABBR2 | 100.00% |
| GABRA2 | 100.00% | GABRA5 | 100.00% | GABRB2 | 100.00% |
| GABRG2 | 100.00% | GALC | 100.00% | GALE | 100.00% |
| GALK1 | 100.00% | GALT | 100.00% | GAMT | 100.00% |
| GATAD2B | 100.00% | GATM | 100.00% | GBA | 100.00% |
| GBE1 | 100.00% | GCDH | 100.00% | GCH1 | 100.00% |
| GCLC | 100.00% | GDAP1 | 100.00% | GDI1 | 100.00% |
| GFAP | 100.00% | GFER | 100.00% | GFM1 | 100.00% |
| GFM2 | 100.00% | GFPT1 | 100.00% | GJC2 | 100.00% |
| GK | 100.00% | GLA | 100.00% | GLDC | 100.00% |
| GLI2 | 100.00% | GLIS3 | 100.00% | GLRA1 | 100.00% |
| GLRX5 | 100.00% | GLS | 100.00% | GLUD1 | 100.00% |
| GLUL | 96.08% | GLYCTK | 100.00% | GM2A | 100.00% |
| GMPPA | 100.00% | GMPPB | 100.00% | GNAI1 | 100.00% |
| GNB1 | 100.00% | GNB5 | 100.00% | GNE | 100.00% |
| GNMT | 100.00% | GPAA1 | 100.00% | GPC3 | 100.00% |
| GPD1 | 100.00% | GPHN | 100.00% | GPSM2 | 100.00% |
| GPT2 | 100.00% | GRHPR | 100.00% | GRIA2 | 100.00% |
| GRIA3 | 100.00% | GRIA4 | 100.00% | GRID2 | 100.00% |
| GRIK2 | 100.00% | GRIN1 | 100.00% | GRIN2A | 100.00% |
| GRIN2D | 100.00% | GRM1 | 100.00% | GSS | 100.00% |
| GTF2H5 | 100.00% | GTPBP2 | 100.00% | GTPBP3 | 100.00% |
| GYG1 | 100.00% | GYS1 | 100.00% | GYS2 | 100.00% |
| HAAO | 100.00% | HACE1 | 100.00% | HADH | 100.00% |
| HADHA | 100.00% | HADHB | 100.00% | HAMP | 100.00% |
| HARS2 | 100.00% | HCCS | 100.00% | HCFC1 | 100.00% |
| HCN1 | 100.00% | HECW2 | 100.00% | HEPACAM | 100.00% |
| HERC1 | 100.00% | HESX1 | 100.00% | HEXA | 100.00% |
| HEXB | 100.00% | HFE | 100.00% | HGD | 100.00% |
| HIBCH | 100.00% | HIVEP2 | 100.00% | HK1 | 100.00% |
| HLCS | 100.00% | HMBS | 100.00% | HMGCL | 100.00% |
| HMGCS2 | 100.00% | HNRNPDL | 100.00% | HNRNPH2 | 100.00% |
| HNRNPR | 100.00% | HOGA1 | 100.00% | HOXA1 | 100.00% |
| HPD | 100.00% | HPRT1 | 100.00% | HPS1 | 100.00% |
| HRAS | 100.00% | HSD17B10 | 100.00% | HSD17B4 | 100.00% |
| HSD3B7 | 100.00% | HSPB1 | 100.00% | HSPB8 | 100.00% |
| HSPD1 | 100.00% | HTRA2 | 100.00% | HYAL1 | 100.00% |
| IARS2 | 100.00% | IBA57 | 100.00% | IER3IP1 | 100.00% |

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|-----------|---------|----------|---------|----------|---------|
| IGF1 | 100.00% | IGHMBP2 | 100.00% | IL1RAPL1 | 100.00% |
| INPP5E | 100.00% | INPP5K | 100.00% | INTS1 | 100.00% |
| IRF2BPL | 100.00% | IRX5 | 100.00% | ISCA1 | 96.41% |
| ISCA2 | 100.00% | ISCU | 100.00% | ITGA7 | 100.00% |
| ITPA | 100.00% | IVD | 100.00% | JAM3 | 100.00% |
| KBTBD13 | 100.00% | KCNA2 | 100.00% | KCNC1 | 100.00% |
| KCNC3 | 100.00% | KCNH1 | 100.00% | KCNJ10 | 100.00% |
| KCNJ11 | 100.00% | KCNJ6 | 100.00% | KCNK9 | 100.00% |
| KCNQ3 | 100.00% | KCNQ5 | 100.00% | KCNT1 | 100.00% |
| KCNT2 | 100.00% | KCTD3 | 100.00% | KCTD7 | 100.00% |
| KDM1A | 100.00% | KDM5B | 100.00% | KIAA0586 | 95.79% |
| KIDINS220 | 100.00% | KIF11 | 100.00% | KIF14 | 100.00% |
| KIF2A | 100.00% | KIF5A | 100.00% | KIF5C | 100.00% |
| KLHL40 | 100.00% | KLHL41 | 100.00% | KLHL7 | 100.00% |
| KMT2C | 100.00% | KMT2E | 100.00% | KMT5B | 100.00% |
| KNL1 | 100.00% | KPTN | 100.00% | KRAS | 100.00% |
| KYNU | 100.00% | L1CAM | 100.00% | L2HGDH | 100.00% |
| LAMA1 | 100.00% | LAMA2 | 100.00% | LAMB1 | 100.00% |
| LAMC3 | 100.00% | LAMP2 | 100.00% | LARGE1 | 100.00% |
| LARP7 | 100.00% | LARS2 | 100.00% | LCAT | 100.00% |
| LCT | 100.00% | LDB3 | 100.00% | LDHA | 100.00% |
| LDLR | 100.00% | LDLRAP1 | 100.00% | LGI4 | 100.00% |
| LIAS | 100.00% | LIG4 | 100.00% | LINS1 | 100.00% |
| LIPA | 100.00% | LIPT1 | 100.00% | LIPT2 | 100.00% |
| LMBRD1 | 100.00% | LMOD3 | 100.00% | LPIN1 | 100.00% |
| LPL | 100.00% | LRP2 | 100.00% | LRPPRC | 100.00% |
| LYRM7 | 100.00% | LZTR1 | 100.00% | MAB21L1 | 100.00% |
| MAB21L2 | 100.00% | MACF1 | 100.00% | MAF | 100.00% |
| MAGEL2 | 100.00% | MAGT1 | 100.00% | MAN1B1 | 100.00% |
| MAOA | 100.00% | MAP2K1 | 100.00% | MAP2K2 | 100.00% |
| MAP3K20 | 100.00% | MAPK8IP3 | 100.00% | MARS2 | 100.00% |
| MAST1 | 100.00% | MAT1A | 100.00% | MATR3 | 100.00% |
| MBD5 | 100.00% | MBOAT7 | 100.00% | MBTPS2 | 100.00% |
| MCCC1 | 100.00% | MCCC2 | 100.00% | MCEE | 100.00% |
| MCM3AP | 100.00% | MCOLN1 | 100.00% | MCPH1 | 100.00% |
| MDH2 | 100.00% | MECR | 100.00% | MED17 | 100.00% |
| MED23 | 100.00% | MED25 | 100.00% | MEGF10 | 100.00% |
| MEIS2 | 100.00% | METTL23 | 100.00% | MFF | 100.00% |

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| | | | | | |
|---------|---------|---------|---------|---------|---------|
| MFN2 | 100.00% | MFSD8 | 100.00% | MGAT2 | 100.00% |
| MGME1 | 100.00% | MICU1 | 100.00% | MID1 | 100.00% |
| MIPEP | 100.00% | MLC1 | 100.00% | MLYCD | 100.00% |
| MMAA | 100.00% | MMAB | 100.00% | MMACHC | 100.00% |
| MMADHC | 100.00% | MOCS1 | 100.00% | MOCS2 | 100.00% |
| MOGS | 100.00% | MPC1 | 100.00% | MPI | 100.00% |
| MPLKIP | 100.00% | MPV17 | 100.00% | MRPL3 | 100.00% |
| MRPL44 | 100.00% | MRPS2 | 100.00% | MRPS22 | 100.00% |
| MRPS34 | 100.00% | MSL3 | 98.34% | MSMO1 | 100.00% |
| MSTO1 | 100.00% | MT-ATP6 | 100.00% | MT-ATP8 | 100.00% |
| MT-CO1 | 100.00% | MT-CO2 | 100.00% | MT-CO3 | 100.00% |
| MT-CYB | 100.00% | MTFMT | 100.00% | MTHFR | 100.00% |
| MTM1 | 100.00% | MT-ND1 | 100.00% | MT-ND2 | 100.00% |
| MT-ND3 | 100.00% | MT-ND4 | 100.00% | MT-ND4L | 100.00% |
| MT-ND5 | 100.00% | MT-ND6 | 100.00% | MTO1 | 100.00% |
| MTPAP | 100.00% | MTR | 100.00% | MT-RNR1 | 100.00% |
| MTRR | 100.00% | MT-TA | 100.00% | MT-TC | 100.00% |
| MT-TD | 100.00% | MT-TE | 100.00% | MT-TF | 100.00% |
| MT-TG | 100.00% | MT-TH | 100.00% | MT-TI | 100.00% |
| MT-TK | 100.00% | MT-TL1 | 100.00% | MT-TL2 | 100.00% |
| MT-TM | 100.00% | MT-TN | 100.00% | MTTP | 100.00% |
| MT-TP | 100.00% | MT-TQ | 100.00% | MT-TR | 100.00% |
| MT-TS1 | 100.00% | MT-TS2 | 100.00% | MT-TV | 100.00% |
| MT-TW | 100.00% | MT-TY | 100.00% | MUSK | 100.00% |
| MVK | 100.00% | MYBPC1 | 100.00% | MYH2 | 100.00% |
| MYH3 | 100.00% | MYH7 | 100.00% | MYH8 | 100.00% |
| MYL1 | 100.00% | MYMK | 100.00% | MYO5A | 100.00% |
| MYO9A | 100.00% | MYOT | 100.00% | MYPN | 100.00% |
| NAA15 | 100.00% | NACC1 | 100.00% | NADK2 | 100.00% |
| NAGA | 100.00% | NAGS | 100.00% | NARS2 | 100.00% |
| NAXE | 100.00% | NBEA | 99.80% | NDE1 | 100.00% |
| NDP | 100.00% | NDST1 | 100.00% | NDUFA1 | 100.00% |
| NDUFA10 | 100.00% | NDUFA11 | 100.00% | NDUFA2 | 100.00% |
| NDUFA4 | 100.00% | NDUFA6 | 100.00% | NDUFA9 | 100.00% |
| NDUFAF1 | 100.00% | NDUFAF2 | 100.00% | NDUFAF3 | 100.00% |
| NDUFAF4 | 100.00% | NDUFAF5 | 100.00% | NDUFAF6 | 100.00% |
| NDUFAF8 | 100.00% | NDUFB11 | 100.00% | NDUFB3 | 100.00% |
| NDUFB8 | 100.00% | NDUFS1 | 100.00% | NDUFS2 | 100.00% |

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|--------|---------|----------|---------|---------|---------|
| NDUFS3 | 100.00% | NDUFS4 | 100.00% | NDUFS6 | 100.00% |
| NDUFS7 | 100.00% | NDUFS8 | 100.00% | NDUFV1 | 100.00% |
| NDUFV2 | 100.00% | NEB | 100.00% | NEXMIF | 100.00% |
| NFASC | 100.00% | NFIA | 100.00% | NFU1 | 100.00% |
| NGLY1 | 100.00% | NHLRC1 | 100.00% | NHS | 100.00% |
| NKAP | 100.00% | NKX2-1 | 100.00% | NLGN3 | 100.00% |
| NNT | 100.00% | NONO | 100.00% | NPC1 | 100.00% |
| NPC2 | 100.00% | NRAS | 100.00% | NRXN1 | 100.00% |
| NSD2 | 100.00% | NSUN2 | 100.00% | NT5C2 | 100.00% |
| NT5C3A | 100.00% | NTNG2 | 100.00% | NTRK1 | 100.00% |
| NTRK2 | 100.00% | NUBPL | 100.00% | OCLN | 100.00% |
| OCRL | 100.00% | ODC1 | 100.00% | OGT | 100.00% |
| OPA1 | 100.00% | OPA3 | 100.00% | ORA1 | 100.00% |
| OSGEP | 100.00% | OTC | 100.00% | OTUD6B | 100.00% |
| OTX2 | 100.00% | OXCT1 | 100.00% | P4HTM | 100.00% |
| PACS2 | 100.00% | PAFAH1B1 | 100.00% | PAH | 100.00% |
| PAK1 | 100.00% | PAK3 | 100.00% | PANK2 | 100.00% |
| PARN | 100.00% | PARS2 | 100.00% | PAX6 | 100.00% |
| PAX7 | 100.00% | PAX8 | 100.00% | PBX1 | 100.00% |
| PC | 100.00% | PCBD1 | 100.00% | PCCA | 100.00% |
| PCCB | 100.00% | PCDH12 | 100.00% | PCDH19 | 100.00% |
| PCGF2 | 100.00% | PCK1 | 100.00% | PCSK9 | 100.00% |
| PCYT2 | 100.00% | PDGFRB | 100.00% | PDHA1 | 100.00% |
| PDHB | 100.00% | PDHX | 100.00% | PDP1 | 100.00% |
| PDSS1 | 97.36% | PDSS2 | 100.00% | PEPD | 100.00% |
| PET100 | 100.00% | PEX1 | 100.00% | PEX10 | 100.00% |
| PEX11B | 100.00% | PEX12 | 100.00% | PEX13 | 100.00% |
| PEX14 | 100.00% | PEX16 | 100.00% | PEX19 | 100.00% |
| PEX2 | 100.00% | PEX26 | 100.00% | PEX3 | 100.00% |
| PEX6 | 100.00% | PFKM | 100.00% | PGAM2 | 100.00% |
| PGAP1 | 100.00% | PGAP2 | 100.00% | PGAP3 | 100.00% |
| PGK1 | 99.84% | PGM1 | 100.00% | PHACTR1 | 100.00% |
| PHF21A | 100.00% | PHF8 | 100.00% | PHIP | 100.00% |
| PHKA1 | 100.00% | PHKA2 | 100.00% | PHKB | 100.00% |
| PHKG2 | 100.00% | PHYH | 100.00% | PIEZO2 | 100.00% |
| PIGA | 100.00% | PIGB | 100.00% | PIGG | 100.00% |
| PIGL | 100.00% | PIGN | 100.00% | PIGO | 100.00% |
| PIGU | 92.51% | PIGW | 100.00% | PIK3R2 | 100.00% |

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|----------|---------|----------|---------|----------|---------|
| PINK1 | 100.00% | PITRM1 | 100.00% | PLA2G6 | 100.00% |
| PLAA | 100.00% | PLCB1 | 100.00% | PLEC | 100.00% |
| PLP1 | 100.00% | PLPBP | 100.00% | PMM2 | 100.00% |
| PMPCA | 100.00% | PMPCB | 100.00% | PNKP | 100.00% |
| PNP | 100.00% | PNPLA6 | 100.00% | PNPLA8 | 100.00% |
| PNPO | 100.00% | PNPT1 | 100.00% | POLA1 | 100.00% |
| POLG | 100.00% | POLG2 | 100.00% | POLR2A | 100.00% |
| POLR3A | 100.00% | POLR3B | 100.00% | POMGNT1 | 100.00% |
| POMGNT2 | 100.00% | POMK | 100.00% | POMT1 | 100.00% |
| POMT2 | 100.00% | PORCN | 100.00% | POU3F3 | 100.00% |
| PPA2 | 100.00% | PPM1D | 100.00% | PPOX | 100.00% |
| PPP1CB | 100.00% | PPP1R15B | 100.00% | PPP1R21 | 100.00% |
| PPP2CA | 100.00% | PPP2R1A | 100.00% | PPP3CA | 100.00% |
| PPT1 | 100.00% | PQBP1 | 100.00% | PRKAG2 | 100.00% |
| PRKD1 | 100.00% | PRODH | 99.83% | PRPS1 | 100.00% |
| PRR12 | 100.00% | PRSS12 | 100.00% | PRUNE1 | 100.00% |
| PSAP | 100.00% | PSMD12 | 100.00% | PTCH1 | 100.00% |
| PTCHD1 | 100.00% | PTF1A | 100.00% | PTPN23 | 100.00% |
| PTS | 100.00% | PUS1 | 100.00% | PUS3 | 100.00% |
| PUS7 | 100.00% | PYCR2 | 100.00% | PYGL | 100.00% |
| PYGM | 100.00% | PYROXD1 | 96.67% | QDPR | 100.00% |
| QRICH1 | 100.00% | QRSL1 | 97.42% | RAB11B | 100.00% |
| RAB18 | 100.00% | RAB39B | 100.00% | RAB3GAP1 | 100.00% |
| RAC1 | 100.00% | RAC3 | 100.00% | RAF1 | 100.00% |
| RALA | 100.00% | RAPSN | 100.00% | RARB | 100.00% |
| RARS2 | 100.00% | RBBP8 | 100.00% | RBCK1 | 100.00% |
| RBM10 | 100.00% | RBP4 | 100.00% | RELN | 100.00% |
| RERE | 100.00% | RHOBTB2 | 100.00% | RLIM | 100.00% |
| RMND1 | 100.00% | RNASEH1 | 100.00% | RNASEH2A | 100.00% |
| RNASEH2B | 100.00% | RNASEH2C | 100.00% | RNASET2 | 100.00% |
| RNF125 | 100.00% | ROGDI | 100.00% | RORA | 100.00% |
| RPIA | 100.00% | RPL10 | 100.00% | RRM2B | 100.00% |
| RTEL1 | 100.00% | RTN4IP1 | 100.00% | RTTN | 100.00% |
| RYR1 | 100.00% | RYR3 | 100.00% | SACS | 100.00% |
| SAMD9 | 100.00% | SAMHD1 | 100.00% | SAR1B | 100.00% |
| SARS2 | 100.00% | SC5D | 100.00% | SCAPER | 100.00% |
| SCN3A | 100.00% | SCN4A | 100.00% | SCO1 | 100.00% |
| SCO2 | 100.00% | SCP2 | 100.00% | SDHA | 100.00% |

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Report Version : 1

Indication

| | | | | | |
|----------|---------|----------|---------|----------|---------|
| SDHAF1 | 100.00% | SDHB | 100.00% | SDHD | 100.00% |
| SEC23B | 100.00% | SELENON | 94.25% | SEPSECS | 100.00% |
| SERAC1 | 100.00% | SET | 100.00% | SETBP1 | 100.00% |
| SETD1B | 100.00% | SETX | 100.00% | SFXN4 | 100.00% |
| SGCA | 100.00% | SGCB | 100.00% | SGCD | 100.00% |
| SGCG | 100.00% | SGPL1 | 100.00% | SHANK2 | 100.00% |
| SHOC2 | 100.00% | SI | 100.00% | SIK1 | 100.00% |
| SIL1 | 100.00% | SIN3A | 100.00% | SIX3 | 100.00% |
| SKIV2L | 100.00% | SLC12A3 | 100.00% | SLC12A5 | 100.00% |
| SLC12A6 | 100.00% | SLC13A5 | 100.00% | SLC16A1 | 100.00% |
| SLC16A2 | 100.00% | SLC18A2 | 100.00% | SLC18A3 | 100.00% |
| SLC19A2 | 100.00% | SLC19A3 | 100.00% | SLC1A2 | 100.00% |
| SLC1A4 | 100.00% | SLC22A5 | 100.00% | SLC25A1 | 100.00% |
| SLC25A12 | 100.00% | SLC25A13 | 100.00% | SLC25A15 | 100.00% |
| SLC25A19 | 100.00% | SLC25A20 | 100.00% | SLC25A22 | 100.00% |
| SLC25A26 | 100.00% | SLC25A3 | 100.00% | SLC25A32 | 100.00% |
| SLC25A38 | 100.00% | SLC25A4 | 100.00% | SLC25A42 | 100.00% |
| SLC25A46 | 100.00% | SLC2A2 | 100.00% | SLC30A10 | 100.00% |
| SLC33A1 | 100.00% | SLC35A1 | 100.00% | SLC35A2 | 100.00% |
| SLC37A4 | 100.00% | SLC39A14 | 100.00% | SLC39A4 | 100.00% |
| SLC39A8 | 100.00% | SLC3A1 | 100.00% | SLC40A1 | 100.00% |
| SLC46A1 | 100.00% | SLC4A4 | 100.00% | SLC52A2 | 100.00% |
| SLC52A3 | 100.00% | SLC5A1 | 100.00% | SLC5A7 | 100.00% |
| SLC6A17 | 100.00% | SLC6A19 | 100.00% | SLC6A20 | 100.00% |
| SLC6A3 | 100.00% | SLC6A8 | 100.00% | SLC6A9 | 100.00% |
| SLC7A7 | 100.00% | SLC7A9 | 100.00% | SLC9A6 | 100.00% |
| SLX4 | 100.00% | SMARCC2 | 100.00% | SMARCD1 | 100.00% |
| SMN1 | 100.00% | SMPD1 | 100.00% | SMPD4 | 68.52% |
| SMS | 100.00% | SNAP25 | 100.00% | SNAP29 | 100.00% |
| SNX14 | 100.00% | SON | 100.00% | SOS1 | 100.00% |
| SOS2 | 100.00% | SOX10 | 100.00% | SOX2 | 100.00% |
| SOX3 | 100.00% | SOX4 | 100.00% | SOX5 | 100.00% |
| SPART | 100.00% | SPATA5 | 100.00% | SPEG | 100.00% |
| SPG11 | 100.00% | SPG7 | 100.00% | SPR | 100.00% |
| SPRED1 | 100.00% | SPTBN2 | 100.00% | SPTLC1 | 100.00% |
| SPTLC2 | 100.00% | SQSTM1 | 100.00% | SRCAP | 100.00% |
| SRD5A3 | 100.00% | SSBP1 | 100.00% | SSR4 | 100.00% |
| ST3GAL3 | 100.00% | ST3GAL5 | 100.00% | STAC3 | 100.00% |

Dr. Pratap N. Mukhopadhyaya
Ph.D(Sc.)

Dr. Nirmal A. Vaniawala
MD (Path. & Bact.)

Dr. Salil Vaniawala
M.Sc Ph.D.(Human Genetics)
Consulting Geneticist

Case ID

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Indication

| | | | | | |
|----------|---------|----------|---------|----------|---------|
| STAG1 | 100.00% | STAG2 | 100.00% | STAMP | 100.00% |
| STIL | 100.00% | STIM1 | 100.00% | STRA6 | 100.00% |
| STRADA | 100.00% | STS | 100.00% | STX1B | 100.00% |
| SUCLA2 | 100.00% | SUCLG1 | 100.00% | SUOX | 100.00% |
| SURF1 | 100.00% | SVBP | 100.00% | SYN1 | 100.00% |
| SYNE1 | 100.00% | SYNE2 | 100.00% | SYNJ1 | 100.00% |
| SYP | 100.00% | SYT1 | 100.00% | SYT2 | 100.00% |
| SZT2 | 100.00% | TACO1 | 100.00% | TAF1 | 100.00% |
| TAF6 | 100.00% | TANGO2 | 100.00% | TAOK1 | 100.00% |
| TAT | 100.00% | TBC1D20 | 100.00% | TBC1D23 | 100.00% |
| TBC1D24 | 100.00% | TBCD | 100.00% | TBCK | 100.00% |
| TBR1 | 100.00% | TCAP | 100.00% | TCF20 | 100.00% |
| TCN2 | 100.00% | TECPR2 | 100.00% | TELO2 | 100.00% |
| TFR2 | 100.00% | TGIF1 | 100.00% | TH | 100.00% |
| THOC2 | 100.00% | THOC6 | 100.00% | THRA | 100.00% |
| TIA1 | 100.00% | TIMM50 | 100.00% | TIMM8A | 100.00% |
| TK2 | 100.00% | TLK2 | 99.91% | TMEM126B | 100.00% |
| TMEM199 | 100.00% | TMEM240 | 100.00% | TMEM70 | 100.00% |
| TMEM94 | 100.00% | TMTC3 | 100.00% | TMX2 | 98.65% |
| TNNI2 | 100.00% | TNNT1 | 100.00% | TNNT3 | 100.00% |
| TNPO3 | 100.00% | TOE1 | 100.00% | TOP3A | 100.00% |
| TPK1 | 100.00% | TPM2 | 100.00% | TPM3 | 100.00% |
| TPP1 | 100.00% | TRAF7 | 100.00% | TRAIP | 100.00% |
| TRAP1 | 100.00% | TRAPPC11 | 100.00% | TRAPPC12 | 100.00% |
| TRAPPC6B | 100.00% | TRAPPC9 | 100.00% | TREX1 | 100.00% |
| TRIM37 | 100.00% | TRIM8 | 100.00% | TRIP12 | 100.00% |
| TRIP4 | 100.00% | TRIT1 | 100.00% | TRMT1 | 100.00% |
| TRMT10C | 100.00% | TRMT5 | 100.00% | TRMU | 100.00% |
| TRNT1 | 100.00% | TRPM6 | 100.00% | TRRAP | 100.00% |
| TSEN2 | 100.00% | TSEN34 | 100.00% | TSEN54 | 100.00% |
| TSFM | 93.95% | TSHB | 100.00% | TSPAN7 | 100.00% |
| TTC19 | 100.00% | TTC37 | 100.00% | TTI2 | 100.00% |
| TTN | 100.00% | TTPA | 100.00% | TUBA1A | 100.00% |
| TUBB | 100.00% | TUBB2A | 100.00% | TUBB2B | 100.00% |
| TUBB3 | 100.00% | TUBB4A | 100.00% | TUBG1 | 100.00% |
| TUBGCP6 | 100.00% | TUFM | 100.00% | TUSC3 | 100.00% |
| TWNK | 100.00% | TYMP | 100.00% | UBA1 | 100.00% |
| UBA5 | 100.00% | UBE2A | 100.00% | UBE3B | 100.00% |

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|----------|---------|----------|---------|---------|---------|
| UBR1 | 100.00% | UBTF | 100.00% | UFM1 | 100.00% |
| UGT1A1 | 100.00% | UMPS | 100.00% | UNC80 | 100.00% |
| UPB1 | 100.00% | UPF3B | 100.00% | UQCC2 | 100.00% |
| UQCRB | 100.00% | UROC1 | 100.00% | UROD | 100.00% |
| UROS | 100.00% | VAMP1 | 100.00% | VAMP2 | 100.00% |
| VAR2 | 100.00% | VCP | 100.00% | VIPAS39 | 100.00% |
| VKORC1 | 100.00% | VLDLR | 100.00% | VMA21 | 100.00% |
| VPS33B | 100.00% | VPS53 | 100.00% | VRK1 | 100.00% |
| WARS2 | 100.00% | WASF1 | 100.00% | WDR26 | 100.00% |
| WDR37 | 100.00% | WDR45B | 100.00% | WDR62 | 100.00% |
| WDR73 | 100.00% | WDR81 | 100.00% | WFS1 | 100.00% |
| WVOX | 100.00% | XDH | 100.00% | YARS2 | 100.00% |
| YWHAG | 100.00% | ZBTB18 | 100.00% | ZBTB20 | 100.00% |
| ZBTB24 | 100.00% | ZDHHC9 | 100.00% | ZFYVE26 | 100.00% |
| ZIC2 | 100.00% | ZMIZ1 | 100.00% | ZMYND11 | 100.00% |
| ZNF142 | 100.00% | ZNF462 | 100.00% | ZNF711 | 100.00% |
| ABCA2 | 100.00% | ACTA2 | 100.00% | ADAM22 | 100.00% |
| ADARB1 | 100.00% | ADD3 | 100.00% | AGAP1 | 84.19% |
| AGMO | 100.00% | AGO1 | 100.00% | AGO2 | 100.00% |
| AHCY | 100.00% | AIMP2 | 100.00% | AIRE | 100.00% |
| AK2 | 100.00% | ALS2 | 100.00% | ANKRD17 | 100.00% |
| ANTXR1 | 100.00% | AP1B1 | 100.00% | AP2S1 | 100.00% |
| ARHGAP35 | 100.00% | ARL14EP | 100.00% | ASCC3 | 100.00% |
| ASTN1 | 100.00% | ATAD1 | 100.00% | ATP1A2 | 100.00% |
| ATP6V0A1 | 100.00% | ATXN2L | 95.69% | BCAS3 | 100.00% |
| BCORL1 | 100.00% | BICRA | 100.00% | BSND | 100.00% |
| CACNA2D2 | 100.00% | CACNB4 | 100.00% | CAMK2G | 100.00% |
| CAPN15 | 100.00% | CAPZA2 | 100.00% | CASQ1 | 100.00% |
| CAVIN1 | 100.00% | CCDC186 | 100.00% | CCDC32 | 100.00% |
| CCDC88A | 100.00% | CDC42BPB | 100.00% | CDH2 | 100.00% |
| CDK16 | 100.00% | CDK19 | 100.00% | CEP104 | 100.00% |
| CEP55 | 100.00% | CEP63 | 100.00% | CEP85L | 100.00% |
| CHD1 | 100.00% | CHD5 | 100.00% | CHL1 | 100.00% |
| CLCN2 | 100.00% | CNPY3 | 100.00% | CNTN1 | 100.00% |
| CNTNAP1 | 100.00% | COA3 | 100.00% | COPB1 | 100.00% |
| COX4I2 | 100.00% | CPE | 100.00% | CRBN | 100.00% |
| CSNK1G1 | 100.00% | CTNND1 | 100.00% | CTU2 | 100.00% |
| CUL3 | 100.00% | CYCS | 100.00% | DDB1 | 100.00% |

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|-----------|---------|----------|---------|---------|---------|
| DDX23 | 92.94% | DDX53 | 100.00% | DENND5A | 100.00% |
| DHX16 | 100.00% | DHX32 | 100.00% | DHX37 | 100.00% |
| DIP2B | 100.00% | DLG1 | 100.00% | DLG2 | 100.00% |
| DLL1 | 100.00% | DONSON | 100.00% | DPH2 | 100.00% |
| DPYSL5 | 100.00% | DYNC1I2 | 100.00% | EEF1B2 | 99.88% |
| EIF2AK2 | 100.00% | EIF5A | 100.00% | EMC10 | 100.00% |
| EMG1 | 100.00% | EPB41L1 | 100.00% | EPHA7 | 100.00% |
| ERAL1 | 100.00% | ERBB4 | 100.00% | ERGIC3 | 100.00% |
| ERMARD | 100.00% | EXOC2 | 100.00% | EXOC7 | 100.00% |
| EXOSC8 | 100.00% | FAAH2 | 100.00% | FAM120C | 100.00% |
| FAM50A | 100.00% | FANCA | 100.00% | FANCC | 100.00% |
| FANCD2 | 100.00% | FANCE | 100.00% | FANCF | 100.00% |
| FANCG | 100.00% | FANCI | 100.00% | FARSB | 100.00% |
| FBRSL1 | 100.00% | FBXO31 | 100.00% | FDFT1 | 100.00% |
| FGF13 | 100.00% | FGF14 | 100.00% | FIBP | 100.00% |
| FOXP4 | 100.00% | FRAS1 | 100.00% | FREM2 | 100.00% |
| FRRS1L | 100.00% | FRY | 100.00% | FTO | 100.00% |
| GAD1 | 100.00% | GALM | 100.00% | GALNT2 | 100.00% |
| GATA6 | 100.00% | GATC | 100.00% | GBA2 | 100.00% |
| GEMIN4 | 100.00% | GEMIN5 | 100.00% | GIGYF1 | 100.00% |
| GJB1 | 100.00% | GJB2 | 100.00% | GJB3 | 100.00% |
| GMNN | 100.00% | GNAI2 | 100.00% | GNB2 | 100.00% |
| GOLGA2 | 82.49% | GOT2 | 100.00% | GPC4 | 100.00% |
| GRIA1 | 100.00% | GSX2 | 100.00% | GTF2E2 | 100.00% |
| GTF3C3 | 100.00% | HACD1 | 100.00% | HAX1 | 100.00% |
| HEATR5B | 100.00% | HERC2 | 99.25% | HINT1 | 100.00% |
| HIRA | 100.00% | HNF1B | 100.00% | HNMT | 100.00% |
| HNRNPA2B1 | 100.00% | HNRNPD | 100.00% | HNRNPH1 | 100.00% |
| HPDL | 100.00% | HSPA9 | 99.80% | HTT | 100.00% |
| IFT27 | 100.00% | IL1RAPL2 | 100.00% | INPP4A | 100.00% |
| IQSEC1 | 100.00% | IREB2 | 100.00% | ITFG2 | 100.00% |
| JAG2 | 100.00% | JARID2 | 100.00% | JMJD1C | 100.00% |
| KAT5 | 100.00% | KAT8 | 100.00% | KATNB1 | 100.00% |
| KCNK4 | 100.00% | KCNMA1 | 100.00% | KCNN2 | 100.00% |
| KCNN3 | 100.00% | KDM3B | 100.00% | KDM4B | 100.00% |
| KDM6B | 100.00% | KIF21B | 100.00% | KLF7 | 100.00% |
| KLHL15 | 100.00% | KMT2B | 100.00% | KY | 100.00% |
| LAMB2 | 100.00% | LAS1L | 100.00% | LINGO1 | 100.00% |

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|---------|---------|----------|---------|----------|---------|
| LIPC | 100.00% | LMAN2L | 100.00% | LMBRD2 | 100.00% |
| LMNB1 | 100.00% | LMNB2 | 100.00% | LNPB | 100.00% |
| LRIF1 | 100.00% | LSS | 100.00% | LYRM4 | 100.00% |
| LZTFL1 | 100.00% | MADD | 100.00% | MAP1B | 100.00% |
| MAPK1 | 100.00% | MAPK10 | 100.00% | MAPKAPK5 | 100.00% |
| MAPRE2 | 100.00% | MED12L | 100.00% | MED13 | 100.00% |
| MED27 | 72.97% | METTL5 | 100.00% | MFSD2A | 100.00% |
| MINPP1 | 100.00% | MMGT1 | 100.00% | MN1 | 100.00% |
| MORC2 | 100.00% | MRM2 | 100.00% | MRPS14 | 100.00% |
| MRPS16 | 100.00% | MRPS7 | 100.00% | MSL2 | 100.00% |
| MTHFS | 100.00% | MTMR14 | 100.00% | MT-RNR2 | 100.00% |
| MT-TT | 100.00% | MYBPC3 | 100.00% | MYF5 | 100.00% |
| MYL2 | 100.00% | MYOD1 | 100.00% | NAA20 | 100.00% |
| NAXD | 100.00% | NBN | 100.00% | NCAPD2 | 99.10% |
| NCAPG2 | 100.00% | NCDN | 100.00% | NCKAP1 | 100.00% |
| NDUFA12 | 100.00% | NDUFB7 | 100.00% | NDUFB9 | 100.00% |
| NDUFC2 | 100.00% | NECAP1 | 100.00% | NEDD4L | 100.00% |
| NEFL | 100.00% | NEMF | 100.00% | NEUROD2 | 93.12% |
| NFIB | 100.00% | NHP2 | 100.00% | NOVA2 | 100.00% |
| NR4A2 | 100.00% | NRROS | 100.00% | NSUN3 | 100.00% |
| NUDT2 | 100.00% | NUP107 | 100.00% | NUP188 | 100.00% |
| NUP214 | 100.00% | NUP62 | 100.00% | NUS1 | 100.00% |
| NYX | 100.00% | OPLAH | 100.00% | OTUD5 | 100.00% |
| OXR1 | 91.34% | PAX1 | 100.00% | PCDHGC4 | 100.00% |
| PDCD6IP | 100.00% | PDE10A | 100.00% | PDE6D | 100.00% |
| PDK3 | 100.00% | PET117 | 100.00% | PGM2L1 | 100.00% |
| PIBF1 | 100.00% | PIGC | 100.00% | PIGH | 100.00% |
| PIGK | 100.00% | PIGM | 100.00% | PIGP | 100.00% |
| PIGQ | 100.00% | PIGS | 100.00% | PJA1 | 100.00% |
| PLEKHG2 | 100.00% | POGLUT1 | 100.00% | POU1F1 | 100.00% |
| PPIL1 | 100.00% | PPP1R12A | 100.00% | PRKACB | 100.00% |
| PRKAR1B | 100.00% | PRRT2 | 100.00% | PSMB8 | 100.00% |
| PSMC5 | 100.00% | PTCD3 | 100.00% | PTPN4 | 100.00% |
| PTRH2 | 100.00% | PTRHD1 | 100.00% | PUM1 | 100.00% |
| RAB11A | 100.00% | RAB14 | 100.00% | RALGAPA1 | 44.89% |
| RANBP2 | 100.00% | RAP1B | 100.00% | RAX | 100.00% |
| REEP1 | 100.00% | RHEB | 100.00% | RIC1 | 100.00% |
| RNF113A | 100.00% | RNF13 | 100.00% | RNU7-1 | 100.00% |

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|----------|---------|----------|---------|---------|---------|
| RPS23 | 94.21% | RSPRY1 | 100.00% | RSRC1 | 100.00% |
| RUSC2 | 100.00% | SATB1 | 100.00% | SBF1 | 99.21% |
| SCAF4 | 100.00% | SCAMP5 | 100.00% | SCN1B | 100.00% |
| SCYL1 | 100.00% | SDHAF2 | 100.00% | SDHC | 100.00% |
| SEC31A | 100.00% | SETD1A | 100.00% | SHMT2 | 100.00% |
| SHROOM4 | 100.00% | SIAH1 | 100.00% | SIN3B | 100.00% |
| SLC12A2 | 100.00% | SLC1A1 | 100.00% | SLC27A4 | 100.00% |
| SLC35A3 | 100.00% | SLC45A1 | 100.00% | SLC4A1 | 100.00% |
| SLC4A11 | 100.00% | SLC5A5 | 100.00% | SLC5A6 | 100.00% |
| SLC9A7 | 95.14% | SMARCA5 | 100.00% | SMARCD2 | 100.00% |
| SMCHD1 | 100.00% | SMG8 | 100.00% | SMG9 | 100.00% |
| SNORD118 | 100.00% | SNX27 | 100.00% | SOX6 | 100.00% |
| SPEN | 100.00% | SPOP | 100.00% | SPTBN1 | 100.00% |
| SPTBN4 | 100.00% | SRGAP3 | 100.00% | SRP54 | 100.00% |
| SRRM2 | 100.00% | SSR3 | 100.00% | STAT2 | 100.00% |
| STT3A | 100.00% | SUFU | 100.00% | SUPT16H | 99.87% |
| SUZ12 | 100.00% | SVIL | 100.00% | TAF13 | 100.00% |
| TAF1C | 100.00% | TANC2 | 100.00% | TARS2 | 100.00% |
| TASP1 | 94.54% | TBC1D2B | 100.00% | TBC1D7 | 100.00% |
| TBX1 | 100.00% | TCF7L2 | 100.00% | TENM3 | 100.00% |
| TET3 | 100.00% | TFAM | 100.00% | TFE3 | 100.00% |
| THRB | 100.00% | TIMMDC1 | 100.00% | TKFC | 100.00% |
| TKT | 100.00% | TMEM106B | 100.00% | TMEM222 | 100.00% |
| TMEM65 | 100.00% | TNIK | 100.00% | TNNC2 | 100.00% |
| TNR | 100.00% | TNRC6B | 100.00% | TOMM70 | 100.00% |
| TOR1A | 100.00% | TOR1AIP1 | 100.00% | TRAK1 | 100.00% |
| TRAPPC2L | 100.00% | TRAPPC4 | 100.00% | TRDN | 100.00% |
| TRPM3 | 100.00% | TSEN15 | 100.00% | TTC5 | 100.00% |
| TUBGCP2 | 100.00% | TUBGCP4 | 100.00% | U2AF2 | 100.00% |
| UBE4A | 100.00% | UBR7 | 100.00% | UFC1 | 100.00% |
| UGDH | 100.00% | UGP2 | 100.00% | UNC45B | 100.00% |
| UPF1 | 100.00% | UQCC3 | 100.00% | UQCRC2 | 100.00% |
| UQCRQ | 100.00% | USP27X | 100.00% | USP7 | 100.00% |
| VAPB | 100.00% | VPS11 | 100.00% | VPS41 | 100.00% |
| VPS4A | 100.00% | VPS51 | 84.76% | WASHC4 | 100.00% |
| WASHC5 | 100.00% | WDFY3 | 100.00% | WDR4 | 100.00% |
| XPA | 100.00% | YIF1B | 100.00% | YIPF5 | 100.00% |
| YME1L1 | 100.00% | ZBTB11 | 100.00% | ZC3H14 | 100.00% |

Case ID

Sample Type : EDTA PERIPHERAL BLOOD

Name

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Sex/Age

Date & Time Received : 12-May-2023 03:29 PM

Bill. Loc.

Date & Time Reported : 01-Jun-2023 07:01 PM

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Indication

| | | | | | |
|-----------|---------|----------|---------|----------|---------|
| ZFHX4 | 100.00% | ZFP57 | 100.00% | ZMYM2 | 100.00% |
| ZNF148 | 100.00% | ZNF292 | 100.00% | ZNF335 | 100.00% |
| ZNF407 | 100.00% | ZNF526 | 100.00% | A2ML1 | 100.00% |
| ABCC6 | 100.00% | ABCC8 | 100.00% | ABCG2 | 100.00% |
| ACBD6 | 100.00% | ACE2 | 100.00% | ACIN1 | 100.00% |
| ACOT9 | 100.00% | ACOX2 | 100.00% | ADCY5 | 100.00% |
| ADGRG4 | 100.00% | ADGRG6 | 100.00% | ADRA2B | 100.00% |
| AFP | 100.00% | AGPAT2 | 100.00% | AGT | 100.00% |
| AGTR2 | 100.00% | AK1 | 100.00% | AKAP17A | 100.00% |
| AKAP4 | 100.00% | AKAP6 | 100.00% | AKR1C2 | 100.00% |
| ALDH1A3 | 100.00% | ALDH1B1 | 100.00% | AMPD1 | 100.00% |
| ANK3 | 100.00% | ANO3 | 100.00% | AOX1 | 100.00% |
| AP5Z1 | 100.00% | AQP7 | 79.30% | ARHGAP36 | 100.00% |
| ARHGAP6 | 100.00% | ARHGEF2 | 100.00% | ARHGEF4 | 100.00% |
| ARHGEF6 | 100.00% | ARIH1 | 98.81% | ARSF | 100.00% |
| ARSG | 100.00% | ASB12 | 100.00% | ASCL1 | 100.00% |
| ASMT | 92.51% | ASMTL | 100.00% | ASPH | 100.00% |
| ATAD3B | 100.00% | ATCAY | 100.00% | ATP2A1 | 100.00% |
| ATP2A2 | 100.00% | ATP2B3 | 100.00% | ATP2C2 | 100.00% |
| ATP6V1B1 | 100.00% | ATXN1 | 100.00% | ATXN2 | 100.00% |
| ATXN3 | 100.00% | ATXN3L | 100.00% | ATXN7 | 100.00% |
| AVP | 100.00% | AVPR2 | 100.00% | AWAT2 | 100.00% |
| BCAT1 | 100.00% | BDP1 | 100.00% | BEAN1 | 100.00% |
| BET1 | 100.00% | BFSP2 | 100.00% | BGN | 100.00% |
| BMP15 | 100.00% | BOLA1 | 100.00% | BOLA2 | 24.14% |
| BPIFB6 | 100.00% | BRCA1 | 100.00% | BRCA2 | 100.00% |
| BRIP1 | 100.00% | BTK | 100.00% | BVES | 100.00% |
| C1GALT1C1 | 100.00% | C9orf72 | 100.00% | CACNA1F | 100.00% |
| CACNA1H | 100.00% | CACNA2D3 | 100.00% | CACNG2 | 100.00% |
| CAP1 | 100.00% | CAPN10 | 100.00% | CAPRIN1 | 100.00% |
| CASP2 | 100.00% | CCDC103 | 100.00% | CCDC174 | 100.00% |
| CCDC65 | 100.00% | CCNA2 | 100.00% | CCNB3 | 100.00% |
| CCNO | 100.00% | CCT5 | 100.00% | CD320 | 100.00% |
| CD99 | 100.00% | CDC40 | 100.00% | CDK5R1 | 100.00% |
| CEP89 | 97.79% | CETP | 100.00% | CFAP47 | 100.00% |
| CFP | 100.00% | CHM | 100.00% | CHRD1 | 100.00% |
| CHRNA2 | 100.00% | CHRNA4 | 100.00% | CHRN2 | 100.00% |
| CHUK | 100.00% | CIB2 | 100.00% | CLCN1 | 100.00% |

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|---------|---------|---------|---------|---------|---------|
| CLCNKA | | | 00.00% | CLIC2 | 100.00% |
| CLPS | 100.00% | CMC4 | 100.00% | CNDP1 | 100.00% |
| CNKSR1 | 100.00% | CNTN3 | 100.00% | CNTN4 | 100.00% |
| COA1 | 100.00% | COA5 | 100.00% | COG2 | 100.00% |
| COL18A1 | 100.00% | COL25A1 | 100.00% | COL4A3 | 100.00% |
| COL4A4 | 100.00% | COL4A6 | 100.00% | COQ5 | 100.00% |
| COX4I1 | 100.00% | COX5A | 100.00% | COX5B | 100.00% |
| COX6C | 100.00% | COX7A1 | 100.00% | COX7A2 | 100.00% |
| COX7B2 | 100.00% | COX7C | 100.00% | COX8A | 100.00% |
| CPA6 | 100.00% | CPD | 100.00% | CPT1B | 100.00% |
| CPXCR1 | 100.00% | CRLF2 | 200.00% | CRYAA | 100.00% |
| CRYBA1 | 100.00% | CRYBA4 | 100.00% | CRYBB1 | 100.00% |
| CRYBB2 | 100.00% | CRYBB3 | 100.00% | CRYGC | 100.00% |
| CRYGD | 100.00% | CSF1R | 100.00% | CSF2RA | 200.00% |
| CSTF2 | 100.00% | CTNND2 | 100.00% | CTPS2 | 100.00% |
| CTSF | 100.00% | CTTNBP2 | 100.00% | CXorf58 | 100.00% |
| CYP1B1 | 100.00% | CYP2C8 | 100.00% | CYP7A1 | 100.00% |
| DAB1 | 100.00% | DCHS2 | 100.00% | DCTN1 | 100.00% |
| DDB2 | 100.00% | DDHD1 | 100.00% | DDX58 | 100.00% |
| DECR1 | 100.00% | DGKH | 100.00% | DHFR2 | 47.87% |
| DHRX | 100.00% | DIAPH2 | 100.00% | DLGAP2 | 100.00% |
| DLST | 100.00% | DMGDH | 100.00% | DMPK | 100.00% |
| DNAAF4 | 100.00% | DNAJB2 | 100.00% | DNAJC3 | 100.00% |
| DNMT1 | 99.02% | DOCK11 | 96.88% | DPEP1 | 100.00% |
| DPF1 | 100.00% | DPF3 | 100.00% | DRD2 | 100.00% |
| DSCAM | 100.00% | DST | 100.00% | DSTYK | 100.00% |
| DTD1 | 100.00% | DUX4 | 100.00% | ECSIT | 100.00% |
| EDA | 100.00% | EDNRA | 100.00% | EDNRB | 100.00% |
| EFHC1 | 100.00% | EGF | 100.00% | EGR2 | 100.00% |
| EIF2A | 100.00% | EIF2AK1 | 100.00% | EIF4G1 | 100.00% |
| ELK1 | 95.49% | ELN | 100.00% | ELOVL5 | 100.00% |
| EN2 | 100.00% | ENOX2 | 100.00% | ENTPD1 | 100.00% |
| EOMES | 100.00% | EPPK1 | 100.00% | ERCC4 | 100.00% |
| ESX1 | 100.00% | EYA1 | 100.00% | F5 | 100.00% |
| FAM111B | 100.00% | FAM161A | 100.00% | FAM20A | 100.00% |
| FAM47B | 78.43% | FANCB | 100.00% | FARSA | 100.00% |
| FASN | 100.00% | FBLN5 | 100.00% | FBP2 | 100.00% |
| FBXO25 | 69.27% | FBXO38 | 100.00% | FBXO7 | 100.00% |

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|---------|---------|---------|---------|---------|---------|
| FBX08 | 100.00% | FEM1B | 100.00% | FGD4 | 100.00% |
| FGF3 | 100.00% | FKBP6 | 79.37% | FKBPL | 100.00% |
| FLT4 | 100.00% | FOLR2 | 100.00% | FOLR3 | 100.00% |
| FOXC2 | 100.00% | FOX1 | 100.00% | FOX3 | 100.00% |
| FOXF1 | 100.00% | FOXN1 | 100.00% | FOXP3 | 100.00% |
| FREM1 | 100.00% | FRMD7 | 100.00% | FTL | 100.00% |
| FXYD2 | 100.00% | FYCO1 | 100.00% | FZD3 | 100.00% |
| FZD6 | 100.00% | GAB3 | 100.00% | GABRG3 | 100.00% |
| GABRQ | 100.00% | GALNT12 | 100.00% | GAN | 100.00% |
| GAS8 | 100.00% | GATA2 | 100.00% | GATA4 | 100.00% |
| GATB | 100.00% | GCK | 100.00% | GCSH | 100.00% |
| GGT1 | 100.00% | GIGYF2 | 100.00% | GJA3 | 100.00% |
| GJA8 | 100.00% | GLE1 | 100.00% | GLMN | 100.00% |
| GLRA2 | 100.00% | GNAI3 | 100.00% | GNAL | 100.00% |
| GON4L | 90.96% | GOSR2 | 100.00% | GPR179 | 100.00% |
| GPRASP1 | 100.00% | GRB14 | 100.00% | GRHL3 | 100.00% |
| GRIP1 | 100.00% | GRM6 | 100.00% | GRN | 100.00% |
| GSPT2 | 100.00% | GTPBP8 | 100.00% | GUCY2C | 100.00% |
| HAL | 100.00% | HAUS7 | 100.00% | HDAC6 | 100.00% |
| HMGB3 | 99.50% | HNF4A | 100.00% | HNRNPA1 | 37.59% |
| HOXC13 | 100.00% | HOXD10 | 100.00% | HPSE2 | 100.00% |
| HR | 100.00% | HS6ST2 | 100.00% | HSF4 | 100.00% |
| HSPB3 | 100.00% | HYDIN | 98.11% | HYKK | 100.00% |
| IDH3B | 100.00% | IFNAR2 | 100.00% | IGBP1 | 94.80% |
| IGF2 | 100.00% | IGSF1 | 100.00% | IL3RA | 100.00% |
| ILF2 | 100.00% | INF2 | 100.00% | INSR | 100.00% |
| INTS6 | 65.02% | INTS6L | 100.00% | INTS8 | 100.00% |
| IQSEC3 | 95.04% | IRAK1 | 100.00% | IRF6 | 100.00% |
| ITCH | 100.00% | ITGA3 | 100.00% | ITGA4 | 100.00% |
| ITGB6 | 100.00% | ITIH6 | 100.00% | JAG1 | 100.00% |
| JAGN1 | 100.00% | JAK3 | 100.00% | JPH3 | 100.00% |
| KANK1 | 100.00% | KATNAL2 | 100.00% | KCNA1 | 100.00% |
| KCND1 | 100.00% | KCND3 | 100.00% | KCNE1 | 100.00% |
| KCNH5 | 100.00% | KCNK12 | 100.00% | KCNQ1 | 100.00% |
| KCTD1 | 100.00% | KDM5A | 100.00% | KHK | 100.00% |
| KIF1B | 100.00% | KIF1C | 100.00% | KIF21A | 100.00% |
| KIF26B | 100.00% | KIF4A | 100.00% | KIRREL3 | 100.00% |
| KIT | 100.00% | KLF1 | 100.00% | KLF8 | 100.00% |

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|---------|---------|---------|---------|---------|---------|
| KLHL21 | 100.00% | KLHL34 | 100.00% | KLHL4 | 100.00% |
| KLHL9 | 100.00% | KRIT1 | 100.00% | LACTB | 100.00% |
| LAMA5 | 100.00% | LETM1 | 99.59% | LG11 | 100.00% |
| LHFPL3 | 100.00% | LHX3 | 100.00% | LHX4 | 100.00% |
| LIMK1 | 100.00% | LIMS2 | 100.00% | LIPI | 100.00% |
| LITAF | 100.00% | LOXHD1 | 100.00% | LRP1 | 100.00% |
| LRRC32 | 100.00% | LRRK1 | 100.00% | LRRK2 | 100.00% |
| LYST | 100.00% | MACC1 | 100.00% | MAGEA11 | 100.00% |
| MAGEB1 | 100.00% | MAGEB10 | 100.00% | MAGEB2 | 100.00% |
| MAGEC1 | 100.00% | MAGEC3 | 100.00% | MAGED1 | 100.00% |
| MAGEE2 | 100.00% | MAGI2 | 100.00% | MAGIX | 100.00% |
| MAOB | 100.00% | MAP3K1 | 100.00% | MAP3K15 | 100.00% |
| MAP7D3 | 100.00% | MAPT | 100.00% | MBNL3 | 100.00% |
| MC2R | 100.00% | MCM9 | 100.00% | MEG3 | 100.00% |
| MET | 100.00% | METAP1 | 100.00% | MFRP | 100.00% |
| MGAT5B | 100.00% | MIB1 | 100.00% | MITF | 100.00% |
| MLH1 | 100.00% | MMP21 | 100.00% | MORC4 | 100.00% |
| MPDZ | 100.00% | MPZ | 100.00% | MRAP | 100.00% |
| MRE11 | 100.00% | MRPL12 | 100.00% | MRPL40 | 100.00% |
| MRPS23 | 100.00% | MSX1 | 100.00% | MTF1 | 100.00% |
| MTHFD1 | 100.00% | MTMR1 | 100.00% | MTMR2 | 100.00% |
| MTMR8 | 100.00% | MXRA5 | 100.00% | MYF6 | 100.00% |
| MYH10 | 100.00% | MYH14 | 100.00% | MYH6 | 100.00% |
| MYH9 | 100.00% | MYO1D | 100.00% | MYO1G | 100.00% |
| MYO5B | 99.84% | MYT1 | 94.59% | NAT8L | 100.00% |
| NCAPH | 100.00% | NDN | 100.00% | NDRG1 | 100.00% |
| NDUFA13 | 100.00% | NDUFA3 | 100.00% | NDUFA5 | 100.00% |
| NDUFA7 | 100.00% | NDUFA8 | 100.00% | NDUFAB1 | 100.00% |
| NDUFB1 | 100.00% | NDUFB10 | 100.00% | NDUFB2 | 100.00% |
| NDUFB4 | 34.62% | NDUFB5 | 100.00% | NDUFB6 | 100.00% |
| NDUFC1 | 100.00% | NDUFS5 | 100.00% | NDUFV3 | 100.00% |
| NECAB2 | 100.00% | NECTIN1 | 100.00% | NFS1 | 100.00% |
| NGF | 100.00% | NHEJ1 | 100.00% | NIPA1 | 100.00% |
| NLGN4X | 100.00% | NOP56 | 99.22% | NPHS1 | 100.00% |
| NPHS2 | 100.00% | NR1I3 | 100.00% | NR2F2 | 100.00% |
| NR5A1 | 100.00% | NRK | 100.00% | NRXN2 | 100.00% |
| NRXN3 | 100.00% | NSF | 99.06% | NT5C | 100.00% |
| NTM | 100.00% | NTNG1 | 100.00% | NXF4 | 100.00% |

Dr. Pratap N. Mukhopadhyaya
Ph.D.(Sc.)

Dr. Nirmal A. Vaniawala
MD (Path. & Bact.)

Dr. Salil Vaniawala
M.Sc Ph.D.(Human Genetics)
Consulting Geneticist

Case ID

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|----------|---------|----------|---------|----------|---------|
| NXF5 | 100.00% | OBSCN | 100.00% | ODF2L | 100.00% |
| OGDH | 100.00% | OR5M1 | 100.00% | OTOGL | 100.00% |
| OTUD7A | 100.00% | OTULIN | 100.00% | OXA1L | 100.00% |
| P2RY4 | 100.00% | P2RY8 | 200.00% | PABPC5 | 100.00% |
| PABPN1 | 100.00% | PALB2 | 100.00% | PARK7 | 100.00% |
| PARP1 | 100.00% | PASD1 | 100.00% | PAX2 | 100.00% |
| PAX9 | 100.00% | PBRM1 | 100.00% | PCDH10 | 100.00% |
| PCLO | 100.00% | PDCD10 | 100.00% | PDE6G | 100.00% |
| PDGFB | 100.00% | PDK1 | 100.00% | PDK2 | 100.00% |
| PDK4 | 100.00% | PDP2 | 100.00% | PDPR | 96.40% |
| PDXK | 100.00% | PDYN | 100.00% | PECR | 100.00% |
| PEX11A | 100.00% | PGRMC1 | 100.00% | PHC1 | 99.30% |
| PHF10 | 31.46% | PHKG1 | 100.00% | PHOX2B | 100.00% |
| PHYKPL | 100.00% | PIGF | 82.73% | PIGY | 100.00% |
| PIK3C3 | 100.00% | PIN4 | 10.86% | PITX2 | 100.00% |
| PITX3 | 100.00% | PKD1L1 | 100.00% | PLCE1 | 100.00% |
| PLCXD1 | 100.00% | PLEKHG5 | 100.00% | PLOD3 | 100.00% |
| PLXNB3 | 100.00% | PMP22 | 100.00% | PMS2 | 100.00% |
| PNKD | 100.00% | PNLIP | 100.00% | PNPLA2 | 100.00% |
| PNPLA4 | 100.00% | POC1B | 100.00% | POLD1 | 100.00% |
| PPM1B | 100.00% | PPM1K | 100.00% | PPP1R1B | 100.00% |
| PPP2R2B | 100.00% | PRDM12 | 100.00% | PRDX4 | 100.00% |
| PREPL | 100.00% | PRICKLE1 | 100.00% | PRICKLE2 | 100.00% |
| PRICKLE3 | 100.00% | PRKCG | 100.00% | PRKN | 100.00% |
| PRKRA | 100.00% | PRMT9 | 100.00% | PROP1 | 100.00% |
| PROX2 | 100.00% | PRRG1 | 100.00% | PRRG3 | 100.00% |
| PRSS56 | 100.00% | PRX | 100.00% | PSEN1 | 100.00% |
| PSMA7 | 100.00% | PSMD10 | 100.00% | PTCD1 | 100.00% |
| PTPN21 | 100.00% | PTPRZ1 | 100.00% | PUDP | 100.00% |
| QKI | 100.00% | RAB27A | 100.00% | RAB40AL | 100.00% |
| RABL6 | 88.13% | RAD50 | 100.00% | RAD51 | 100.00% |
| RAD51C | 100.00% | RALGDS | 100.00% | RANBP17 | 100.00% |
| RAP1GDS1 | 93.86% | RAPGEF1 | 100.00% | RASA1 | 100.00% |
| RBFOX1 | 100.00% | RBL2 | 100.00% | RBM28 | 100.00% |
| REEP2 | 100.00% | RENBP | 100.00% | RET | 100.00% |
| RETREG1 | 100.00% | RFX6 | 100.00% | RGN | 100.00% |
| RGS7 | 100.00% | RIMS1 | 100.00% | RING1 | 100.00% |
| RIPK4 | 100.00% | RNF135 | 100.00% | RNF168 | 100.00% |

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|----------|---------|----------|---------|----------|---------|
| RNF216 | 100.00% | RNPC3 | 100.00% | ROBO3 | 100.00% |
| RORB | 100.00% | RPS19 | 100.00% | RRAS | 100.00% |
| RSPH1 | 100.00% | RSPH3 | 100.00% | RSP04 | 100.00% |
| RTL9 | 100.00% | RTN2 | 100.00% | RUBCN | 100.00% |
| SAMD9L | 100.00% | SARDH | 100.00% | SBF2 | 100.00% |
| SCARB1 | 100.00% | SCARB2 | 100.00% | SCN11A | 100.00% |
| SCN9A | 100.00% | SCRIB | 100.00% | SELENOI | 100.00% |
| SETDB2 | 100.00% | SGCE | 100.00% | SH3TC2 | 100.00% |
| SHANK1 | 100.00% | SHPK | 100.00% | SHROOM2 | 100.00% |
| SIGMAR1 | 100.00% | SIX1 | 100.00% | SIX5 | 100.00% |
| SLC20A2 | 100.00% | SLC22A12 | 100.00% | SLC22A4 | 100.00% |
| SLC25A2 | 100.00% | SLC25A24 | 100.00% | SLC25A40 | 100.00% |
| SLC25A53 | 100.00% | SLC25A6 | 100.00% | SLC26A9 | 100.00% |
| SLC27A5 | 100.00% | SLC2A10 | 100.00% | SLC2A9 | 100.00% |
| SLC30A9 | 100.00% | SLC31A1 | 82.37% | SLC36A2 | 100.00% |
| SLC52A1 | 100.00% | SLC5A2 | 100.00% | SLC6A4 | 100.00% |
| SLC6A5 | 100.00% | SLC9A9 | 100.00% | SLCO1B1 | 100.00% |
| SLCO1B3 | 100.00% | SMARCA1 | 100.00% | SMARCC1 | 100.00% |
| SMARCD3 | 100.00% | SMO | 100.00% | SNCA | 100.00% |
| SNIP1 | 100.00% | SNRPN | 100.00% | SNTG1 | 100.00% |
| SNX3 | 100.00% | SOBP | 100.00% | SOX17 | 100.00% |
| SPAG1 | 100.00% | SPAST | 100.00% | SPG21 | 100.00% |
| SPRTN | 100.00% | SPRY3 | 100.00% | SREBF2 | 100.00% |
| SRPK3 | 100.00% | SRPX2 | 100.00% | SRRT | 100.00% |
| SRY | 100.00% | STAB2 | 100.00% | STAR | 100.00% |
| STARD8 | 100.00% | STAT1 | 100.00% | STAT5B | 100.00% |
| STIM2 | 100.00% | STT3B | 100.00% | STUB1 | 100.00% |
| STX11 | 100.00% | STX3 | 100.00% | SUCLG2 | 100.00% |
| SUGCT | 100.00% | SYNCRIP | 99.31% | SYT14 | 93.49% |
| SYT15 | 85.15% | SYTL4 | 100.00% | SYTL5 | 100.00% |
| TAB2 | 100.00% | TAF2 | 100.00% | TAF7L | 100.00% |
| TARDBP | 100.00% | TBC1D8B | 100.00% | TBP | 93.63% |
| TBX20 | 100.00% | TBX22 | 100.00% | TCEAL3 | 100.00% |
| TCN1 | 100.00% | TDO2 | 100.00% | TECR | 100.00% |
| TEK | 100.00% | TENM1 | 100.00% | TEPSIN | 100.00% |
| TFAP2A | 100.00% | TFAP2B | 100.00% | TFB2M | 100.00% |
| TFG | 100.00% | TGFB3 | 100.00% | TGM6 | 100.00% |
| THAP1 | 100.00% | THUMPD1 | 100.00% | TIMM44 | 100.00% |

Case ID

Sample Type : EDTA PERIPHERAL BLOOD

Name

Date & Time Collected : 10-May-2023 11:00 AM

Sex/Age

Date & Time Received : 12-May-2023 03:29 PM

Bill. Loc.

Date & Time Reported : 01-Jun-2023 07:01 PM

Ref. By

Report Version : 1

Indication

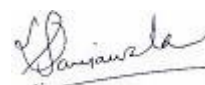
| | | | | | |
|----------|---------|---------|---------|----------|---------|
| TINF2 | 100.00% | TKTL1 | 100.00% | TLR8 | 100.00% |
| TM4SF20 | 100.00% | TM6SF2 | 100.00% | TMEM126A | 100.00% |
| TMEM132E | 100.00% | TMEM135 | 100.00% | TMEM260 | 100.00% |
| TMEM43 | 100.00% | TMLHE | 100.00% | TMPRSS6 | 100.00% |
| TMPRSS9 | 100.00% | TNKS2 | 100.00% | TNPO2 | 100.00% |
| TP73 | 100.00% | TPH2 | 100.00% | TPMT | 100.00% |
| TRAPPC6A | 100.00% | TREH | 100.00% | TREX2 | 100.00% |
| TRHR | 100.00% | TRIP13 | 100.00% | TRPM1 | 100.00% |
| TSC22D3 | 100.00% | TSHR | 100.00% | TSPAN8 | 100.00% |
| TTBK2 | 100.00% | TTC7A | 100.00% | TTR | 100.00% |
| TUBA8 | 100.00% | TUBAL3 | 100.00% | TXN2 | 100.00% |
| TXNL4A | 100.00% | TYR | 100.00% | TYRP1 | 100.00% |
| UBQLN1 | 100.00% | UBR4 | 100.00% | UNC13A | 100.00% |
| UQCRC1 | 100.00% | UQCRFS1 | 89.94% | UQCRH | 90.22% |
| USB1 | 100.00% | USF1 | 100.00% | USP18 | 100.00% |
| UTP14A | 100.00% | UVSSA | 100.00% | VAMP7 | 100.00% |
| VIP | 100.00% | VPS13C | 100.00% | VPS35 | 100.00% |
| VSX2 | 100.00% | WDR11 | 100.00% | WDR13 | 100.00% |
| WDR83OS | 100.00% | WIPI2 | 100.00% | WNK3 | 100.00% |
| WNT4 | 100.00% | WRAP53 | 100.00% | WT1 | 100.00% |
| WWC3 | 89.81% | XIAP | 100.00% | XIST | 100.00% |
| XK | 100.00% | XKRX | 100.00% | XPC | 100.00% |
| YAP1 | 100.00% | ZBTB40 | 100.00% | ZCCHC12 | 100.00% |
| ZCCHC8 | 100.00% | ZDHHC15 | 100.00% | ZFX | 100.00% |
| ZMYM3 | 100.00% | ZMYM6 | 100.00% | ZMYND12 | 100.00% |
| ZNF41 | 100.00% | ZNF425 | 100.00% | ZNF592 | 100.00% |
| ZNF599 | 100.00% | ZNF674 | 100.00% | ZNF713 | 100.00% |
| ZNF81 | 100.00% | ACAT2 | 100.00% | ANK2 | 100.00% |
| AP1G1 | 100.00% | ATP9A | 100.00% | CAMK4 | 100.00% |
| CLCN3 | 100.00% | EHHADH | 100.00% | GGPS1 | 100.00% |
| GIPC1 | 100.00% | HID1 | 100.00% | IMPDH2 | 100.00% |
| LINGO4 | 100.00% | PARP6 | 100.00% | PIDD1 | 100.00% |
| POPDC3 | 100.00% | RFX3 | 100.00% | RFX4 | 100.00% |
| RFX7 | 100.00% | | | | |

*Percentage of coding region covered.

----- End Of Report -----


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MD (Path. & Bact.)



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