

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:

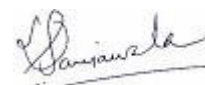
GLB1: c.1577dupG:

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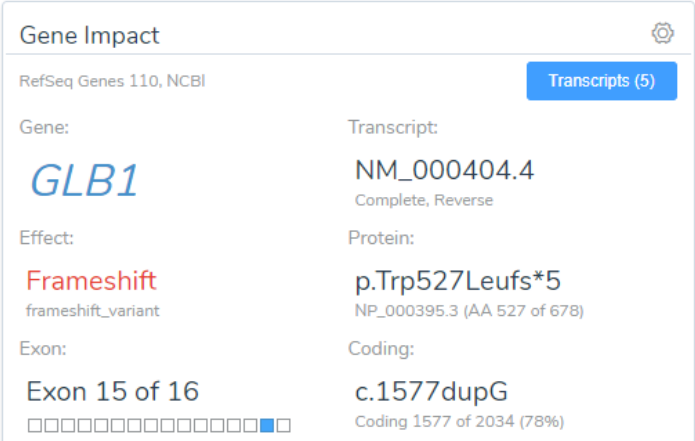
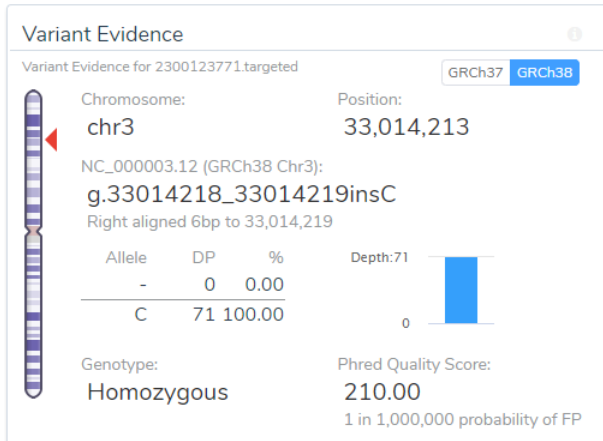
Dr. Pratap N. Mukhopadhyaya
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Based on above evidence, this variant (GLB1: c.1577dupG) is classified as Pathogenic variant.

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:

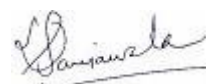
1. Segregation analysis of the variant by Sanger sequencing is recommended in affected and unaffected members of the family.
2. **Targeted mutation analysis of reported variant in parents.**

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:

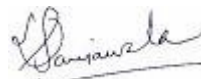
- Next Generation Sequencing: DNA extracted from Blood, Saliva, Amniotic fluid, CVS or any other standard source is used for targeted capture-based Library preparation. Targeted capture provides an efficient and sensitive means for sequencing specific genomic regions in a high-throughput manner. The 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.

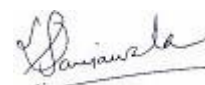
References:

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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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

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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%
DLD	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%

Case ID

Name

Sex/Age

Bill. Loc.

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Indication

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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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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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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%

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**

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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Sample Type : EDTA PERIPHERAL BLOOD

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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%
THR3	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%

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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%	FOXE1	100.00%	FOXE3	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

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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%

Case ID

Name

Sex/Age

Bill. Loc.

Ref. By

Indication

Sample Type : EDTA PERIPHERAL BLOOD

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

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

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

Report Version : 1

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

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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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