SANJAY GANDHI POSTGRADUATE INSTITUTE OF MEDICAL SCIENCES, LUCKNOW

Madhuri Bhaskar

32/F

2022—908396

Department	Unit/Consultant
Cardio	↓ Dr A. Saha Sir

Referring Doctor/Hospital:

Name:

Address:

History

te

As

No cardiac symptoms at present

↓

Routine ~~Lab~~ investigation

Dyslipidemia

H/O

↳ Diabetes ×1 yrs Back

DM
HTN
CKD
CVA ⊖
CAD

addiction ⊖

Mother died at 47 yr age unknown reason

History

Treatment History

↳ main ~~c~~ complaint

↳ presyncopal symptoms on exertion

↳ New onset palpitation on normal activity

rial/Social/Family History

P2+2+2

1st → FTHVD

2nd → LSCS

Physical Examination

Nutrition Status: Good/Adequate/Reduced

Anaemia	No	✓	Yes	
Dyspnea	No	✓	Yes	
Cyanosis	No	✓	Yes	
Jaundice	No	✓	Yes	
Edema	No	✓	Yes	
Exanthem	No	✓	Yes	

W		Kg
H		Cm
BP	114/81	mm
Pulse	72	/m

Extremities (Regular
(Normal
(No RR/RF def

Skin

Thyroid

Lymph Nodes | ND

Head & Neck JVP

Chest BIL VBS

CVS

Apex – (N) 5th ICS mcl tone

S_1 – (N)

S_2 < A_2 / P_2 (N)

No S3/S4/ murm (N)

Abdomen Soft BS

CNS conscious

Provisional Diagnosis Dyslipidemia Ex.d., NSR, ē Dizziness Ex.d.

Kg
Cm
mmHg
/min

32/F

Persistently elevated S. Triglyceridemia

Plan

Tg = 786
TC = 144
HDL = 36
LDL = 58
VLDL = 157
S. uric acid = 9.3
TSH = 2.5

No xanthomas

DM ⊖
ECG - (N)
? Familial Hypertriglyceridemia

Follow up Notes

ECG (29/10) → NSR. WNL

Adv
- Echo
- Genetics Cx

① T. Rozard 40mg HS
② T. Trascepa 1000mg BD
③ T. Fenolip 160mg HS
④ T. Febuxostat 40mg OD

29/10/22

Labs

	Dec 21	March 22	~~Feb~~ 22
T. Chol	147	203	156
Tg	298	873	1098
LDL	88	71	152

SGOT → 113
SGPT → 125

HbA1c → 5.6

T3 → 1.0
T4 → 0.5
~~TSH → 2.5~~

29/10/22

[20229008396]

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Rae Bareli Road, Lucknow 226 014

Name: Madhuri Bhatkar CR No. □□□□□□□□□□□□□□□□

Diagnosis: Persistent hypertriglyceridemia c HSR, LV function

Investigations Ordered

- ☐ Heamatology
- ☐ Coagulation
- ☐ Cl. Chemistry
- ☐ Urinalysis & Fluids
- ☐ Cytology
- ☐ Bacteriology
- ☐ Serology
- ☐ Plain X-ray
- ☐ Ultrasound
- ☐ CT
- ☐ MRI
- ☐ Nuclear Medicine
- ☐ Immunology
- ☐ Medical Genetics
- ☐ Endocrinology
- ☐ GI Endoscopy

Rx

1) T. Rozavel 40mg 1 HS
2) T. Traxcefa 1000mg BD
3) T. Fenolip 160mg HS
4) T. Febustat 40mg 1 OD

29/10/22

Others

- ☐ R/V after 1 month c lipid profile / uric acid / report
- ☐
- ☐
- ☐

Next Appointment on...at......in

(Signature)

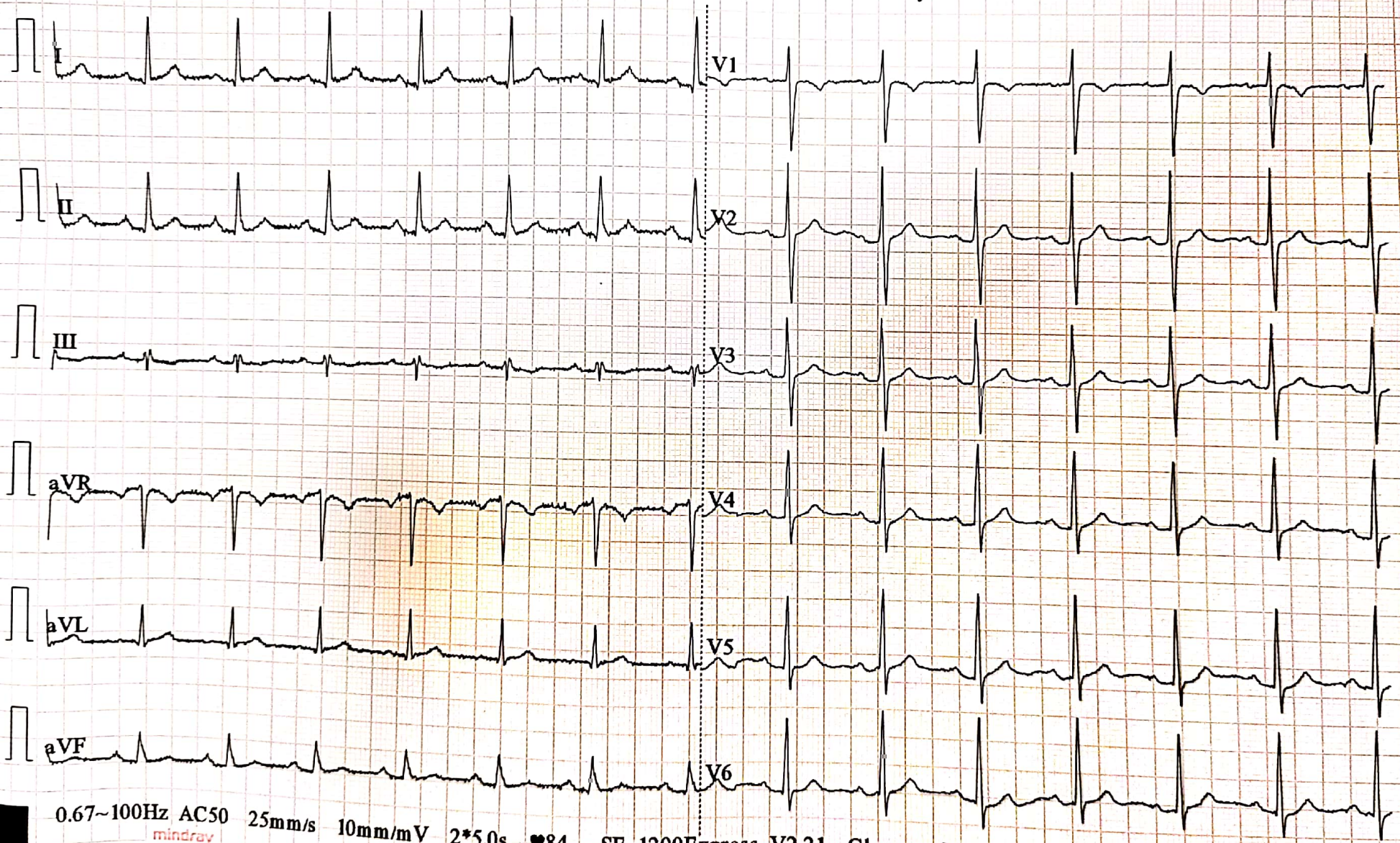
ID: 503
manoj
Male Years

29-10-2022 08:27:48

HR	:	84	bpm
P	:	108	ms
PR	:	182	ms
QRS	:	92	ms
QT/QTc	:	324/383	ms
P/QRS/T	:	56/30/21	°
RV5/SV1	:	1.411/1.215	mV

Diagnosis Information:
Sinus rhythm
Normal ECG

Report Confirmed by:



0.67~100Hz AC50 25mm/s 10mm/mV 2*5.0s ♥84 SE-1200Express V2.21 Glasgow V28.6.0

mindray

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REQUISITION FORM FOR CONSULTATION

Madhuri Bhasker

32/F

2022 9083 96

Ward No. OPD

Bed No.

Dr. Deepti Mandal
Department of Medical Genetics
Sanjay Gandhi Postgraduate Institute
Lucknow-226014 (U.P.) INDIA
9/11/22

Dr. Deepti Saxena
Department of Medical Genetics
Sanjay Gandhi Postgraduate Institute
Lucknow-226014 (U.P.) INDIA

Consultation required from : Medical Genetics

Urgent ☐

Routine ✓

Diagnosis/Specific problem : persistent hypertriglyceridemia
? Familial hyperTG

Consultation/Opinion required in respect of : ? familial HyperTG

Request

Opinion only ☐

Opinion + Follow up ☐

Transfer ☐

Date 29/10/22 Time .....

Signature ..... Designation .....

Name Dr. A Saha

Report/Opinion of the consultant *

Date..... Time ..... Signature..... Designation .....

Name .....

* Use reverse side if required.

G-2

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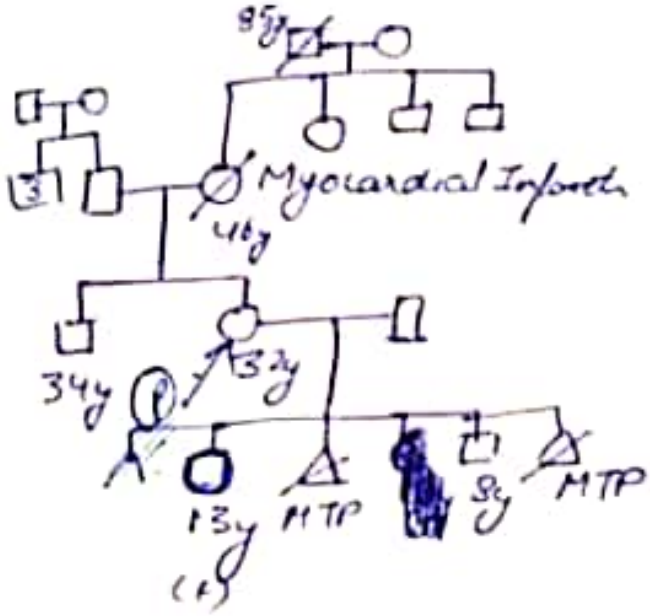
4 Dr. Deepti

OUT PATIENT RECORD

DEPARTMENT OF MEDICAL GENETICS
SANJAY GANDHI POSTGRADUATE INSTITUTE
OF MEDICAL SCIENCES, LUCKNOW– 226014

Tel. : 2494076 (OPD)
: 2494325 (Office)
: 2494348/2494334

Name Madhuri Chauhan 33/F CR No. 20229083 96 Diagnosis

Date	Clinical Condition	Advice
08/11/2022		H/O weakness, syncopal attacks 3-4 times within 3-4 months on exertion - 1 year back. H/O Night blindness No DM/HTN No xanthoma Wt: 59.7 kg Tg 786 mg/dl, S. Cholesterol 144 mg/dl HDL 36 mg/dl, LDL 58.8 mg/dl VLDL 157.2, * RBS 97 mg/dl, HbA1C 5.6
07/12/22		

Please bring this Card at each visit.

Date	Clinical Condition	Advice
	Δ Hypertriglyceridemia.	
	No family history.	
	Referred from cardiology.	
	Adv- Whole exome sequencing.	
	to look for monogenic causes.	
	(test not available in SCPGI).	
	- If any genetic cause is identified, then risk of recurrence to siblings and offsprings = 50%.	
	- Lipid profile of at-risk relatives can be done.	
	- F/c with reports after 2 months (Tue/wed).	

Dr Soma.

Patient Details

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

Sample Details

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

Clinical History

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

Test Results and Interpretation

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

Summary of Variants

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

Variant Details

APOA5

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

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

Disease

HYPERTRIGLYCERIDEMIA, FAMILIAL

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

References

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

Special Recommendations

Parental testing for the above variant is recommended

Recommendations

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

Technical Notes

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

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

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

Bioinformatics pipeline version: 5.0.0.

Gene Coverage

Indication Based Analysis:

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

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

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

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

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

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

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

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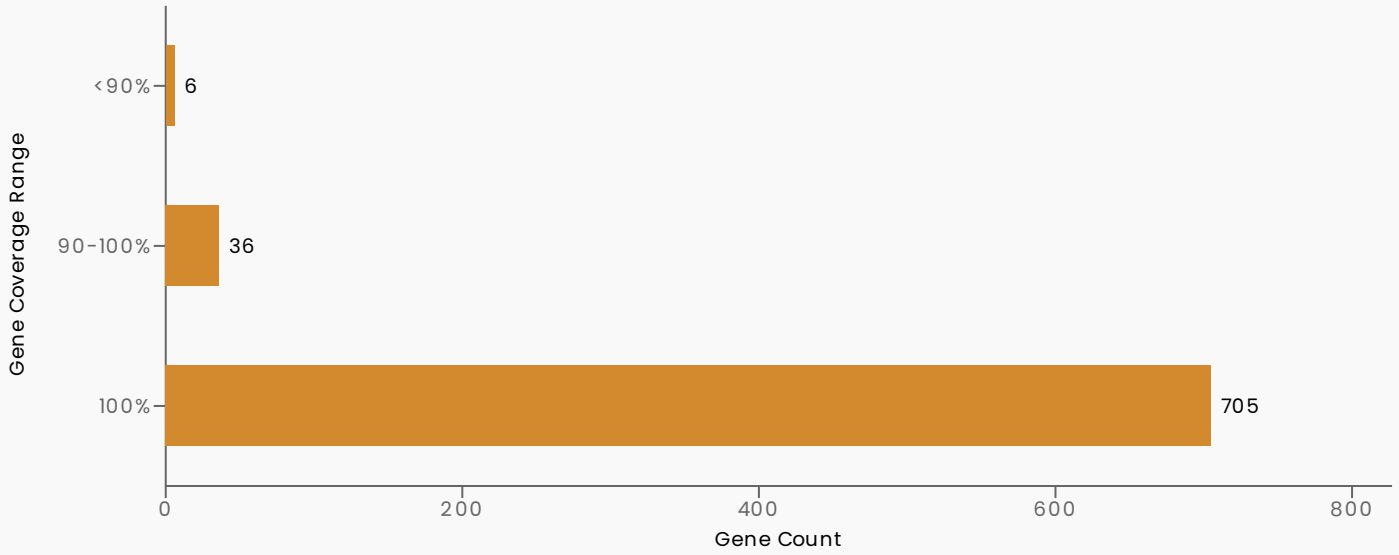
Gene Coverage Distribution



QC Metrics

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

Test Limitations

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

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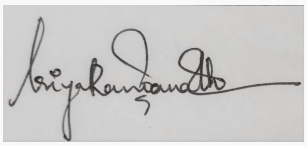
- The classification and interpretation of all the variants in this assay reflects the current state of scientific understanding at the time this report was issued. In some instances, the classification and interpretation of such variants may change as new scientific information comes to light. We recommend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

Reviewed By

Bijal Vyas PhD
Analyzed by
Senior Genome Analyst



Priya Ranganath
Reviewed by
Clinical Geneticist

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

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