SagePath
Excellence In Health Care

Patient Details

Name	: Mr. ANMOL DHARGAVE	**Sex / Age**	: Male / 27 Years	**Test Name**	: ORION (WES-Whole Exome Sequencing)
Ref By	:		:		

Sample Details

Registration Date & Time	: 2025-08-31 16:47:01	**Sample Type**	: Whole Blood EDTA	**Sample Date & Time**	: 2025-08-31 16:47:00
Ref ID 1.	: -	**Acc. Remarks**	: -	**Report Date & Time**	: 2025-09-30

Clinical History

Salient features: Cough on exposure to cold air, sore throat, variant asthma

Notes

Genes associated with above mentioned disorders were evaluated.

HPO terms used for analysis: Cough, Respiratory tract infection, Recurrent lower respiratory tract infections, Immunodeficiency, Ciliary dyskinesia

Ultra rare variants with a gnomAD frequency of <0.01 were evaluated.

Test Results and Interpretation

NO SIGNIFICANT VARIANT RELATED TO PHENOTYPE DETECTED.

Test Information

1. **Clinical correlation as well as reverse phenotyping is recommended for all reports.**
2. **Genetic counseling for accurate interpretation of test results is recommended.**
3. **The reported findings are based on NGS analysis.**
4. **Analysis includes both single nucleotide (SNV) as well as copy number variant analysis (CNV).**
5. **Copy number variants when detected are included in the report.**
6. **Since CNV analysis is performed on a comparative basis, a negative result does not exclude the presence of a CNV.**
7. **The CNV pipeline is not validated for <3 exon copy number variants wherein detection is influenced by the underlying gene region and structure.**
8. **Variant calling (SNV and CNV) may be limited in low covered regions as well as in regions of low complexity and in pseudogenes.**
9. **Synonymous variants (not affecting splice site) as well as intronic variants are usually not reported.**

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Test Information

10. Analysis and reporting is focussed on the provided phenotype and based on relevant HPO (Human Phenotype Ontology) terms as well as on genes associated with provided phenotype.
11. A genotype based analysis is also performed when the above yields negative results but reporting is limited to genes wherein current available evidence suggests a possible association with the provided phenotype.
12. It may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity.
13. Disease descriptions are included from OMIM, Genereviews and PUBMED indexed articles as and where applicable.
14. 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.
15. Phenotype variability may be due to modifying genetic/non-genetic factors and is not a part of the current analysis.
16. Candidate genes and genes with limited evidence are designated as genes of uncertain significance and all variants detected therein are classified as variants of uncertain significance.
17. Typically only variants at a depth of >/= 10X are reported. Lower depth variants may be false positives.
18. Detected variants in low complexity regions as well as variants at low depth and relatively low VAF should be reconfirmed by an alternate methodology.
19. CNV confirmation via MLPA or Exon array is recommended for copy number variants involving a single gene as well as for CNV <400kb. Only large constitutional CNV can be tested via other array platforms.
20. Variant depth/Total depth has been mentioned in summary of the variants.
21. Parental/Maternal testing as applicable is recommended for variants when detected for phasing (where applicable)
22. Segregation analysis (testing of multiple affected as well as unaffected members) of detected variants (if any) is recommended. Variant classification is subject to change after segregation.
23. ACMG secondary findings as well as carrier status of variants are only provided when requested.
24. Within carrier screening risk factors/alleles as well as hypomorphic variants may not be included
25. Interobserver as well as inter-laboratory variation is known with respect to variant classification due to the subjective nature of the provided criteria. Though the laboratory follows the updated recommendations provided by the ClinGen SVI as well as ACMG, independent assessment of variant classification by the referring clinician is recommended before decision making.
26. For prenatal samples analysis is limited to provided clinical phenotypes utilizing relevant HPO terms. Typically variants of uncertain significance are only introduced if the respective gene has been associated with the observed fetal phenotype. In solo fetal exomes, variants of uncertain deemed to be disease causing based on genotype characteristics may be included for further evaluation by the referring clinician. In prenatal scenarios trio fetal testing is strongly recommended to allow better interpretation of detected variants.
27. If the above results do not correlate completely with patient phenotype, additional testing is advised based on clinician's discretion.
28. Typically, heterozygous variants of uncertain significance in genes associated with autosomal recessive disorder are not reported in the proband. Such variants are included if relevant to phenotype in carrier screening.
29. A negative report does not exclude a genetic disorder due to inherent limitations of the assay design.
30. On the background of whole exome sequencing, analysis is limited to provided indications/ requested testing. Hence analysis is limited to a single gene if the same has been requested.
31. As a part of knowledge sharing initiative, all reported variants are submitted in de-identified form in the ClinVar database.
32. Extracted DNA if available after requisitioned testing will be stored as per recommendations. Please note that DNA may degrade over time and this may affect the quality of the stored sample.
33. Collected blood samples are not stored
34. Prenatal samples (AF/CVS/cord blood) are not stored. Extracted DNA if available is stored as acknowledged above.
35. As per PCPNDT fetal gender is not revealed.
36. Maternal cell contamination is recommended for prenatal and POC (product of conception) to ensure accuracy of test results. The same is performed only when requested and on the availability of the maternal sample.
37. Discrepant maternal cell contamination results may rise with use of donor gamete and hence information regarding the same should be provided to the laboratory.
38. Reproductive decision making is not recommended based on variants of uncertain significance.
39. Raw data can be transferred on request and after due consent/assent from the involved patient/ family. Additional charges will be applicable for the same.
40. The test performed by the laboratory with the assumption that the sample belongs to the person herewith mentioned in the requisition form and appropriate consent as well as prenatal counseling including test information has been provided by the referring clinician.

Test Information

41. **Repeat sampling may be required in case of gender discrepancy (unless the same can be attributed to an underlying scientific reason) as well as in rare cases where DNA/data quality prevents further analysis.**
42. **Reanalysis of data is recommended as deemed necessary by the referring clinician. Additional charges will be applicable for the same.**

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. 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 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. 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).

Laboratory reporting protocol: The analysis is based on the provided phenotype: relevant HPO terms, curated gene panels and relevant literature is assessed for phenotype based analysis. Variant reporting is limited to exon regions and upto 10 basepairs within exon intron boundaries. Previously reported deep intronic and non coding variants will be included when detected at a depth more than 10X . Variant reporting is performed at a minimum depth of 10X. The gnomAD variant frequency reflects the liftover of hg38 to hg19.

For Mitochondrial Genome Sequencing (if requested): Only phenotype-related Pathogenic and Likely Pathogenic variations reported in the MitoMap database as well as literature are reported. Haplogroups are not analyzed. A list of variants other than the above are available on request. Analyzed genes include:MT-ND1, MT-ND2, MT-ND3, MT-ND4L, MT-ND4, MT-ND5, MT-ND6, MT-CYB, MT-CO1, MT-CO2, MT-CO3, MT-ATP6, MT-ATP8, MT-RNR2, MT-RNR1, MT-RNR2, MT-TA, MT-TR, MT-TN, MT-TD, MT-TC, MT-TE, MT-TQ, MT-TG, MT-TH, MT-TI, MT-TL1, MT-TL2, MT-TK, MT-TM, MT-TF, MT-TP, MT-TS1, MT-TS2, MT-TT, MT-TW, MT-TY, MT-TV.

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.

REVEL: The REVEL score for an individual missense variant can range from 0 to 1, with higher scores reflecting greater likelihood that the variant is disease-causing.

CADD: The variants with scores above 20 are predicted to be among the 1.0% most deleterious possible substitutions in the human genome.

Bioinformatics pipeline version: 15.10.3

Gene Coverage

Indication Based Analysis:

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
ABCA3	100%	ACAT1	100%	ACBD5	100%	ACD	100%
ACP5	100%	ACTB	100%	ADA	100%	ADA2	100%
ADAM17	100%	ADAM7	100%	ADAMTS13	100%	ADAR	100%
AGA	100%	AGR2	100%	AICDA	100%	AIRE	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
AK2	100%	AK7	100%	AKR1C1	100%	ALAS2	100%
ALG13	100%	ALPI	100%	ALPP	100%	ANKRD26	99.3%
ANTXR1	100%	AP1S3	100%	AP3B1	100%	AP3D1	100%
AP4E1	100%	APOL1	100%	ARHGEF1	100%	ARID3A	100%
ARL2BP	100%	ARPC1B	100%	ARTN	100%	ATAD3A	100%
ATG4A	100%	ATL1	100%	ATL3	100%	ATM	100%
ATP11A	100%	ATP6AP1	100%	ATR	100%	B2M	100%
BACH2	100%	BANK1	100%	BAP1	100%	BCL10	100%
BCL11B	100%	BCL2L11	100%	BCOR	100%	BLK	100%
BLM	100%	BLNK	100%	BLOC1S3	100%	BLOC1S6	100%
BMPR2	100%	BRCA1	100%	BRCA2	100%	BRIP1	100%
BRWD1	100%	BTK	100%	BTNL2	100%	C1QA	100%
C1QB	100%	C1QC	100%	C1R	100%	C1S	100%
C2	100%	C2orf69	100%	C3	100%	C4A	100%
C4B	100%	C4BPA	100%	C5	100%	C6	100%
C7	100%	C8A	100%	C8B	100%	C8G	100%
C9	100%	CAMLG	100%	CARD11	100%	CARD14	100%
CARD9	100%	CARMIL2	100%	CASP10	100%	CASP8	100%
CAVIN1	100%	CBL	100%	CCBE1	100%	CCDC103	100%
CCDC28B	100%	CCDC39	100%	CCDC40	100%	CCDC65	100%
CCNO	100%	CD19	100%	CD247	100%	CD27	100%
CD28	100%	CD3D	100%	CD3E	100%	CD3G	100%
CD4	100%	CD40	100%	CD40LG	100%	CD46	100%
CD55	100%	CD59	100%	CD70	100%	CD79A	100%
CD79B	100%	CD81	100%	CD8A	100%	CDAN1	100%
CDC42	100%	CDCA7	100%	CDIN1	100%	CDK9	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
CEBPA	100%	CEBPE	100%	CENPF	100%	CFAP221	100%
CFAP298	100%	CFAP300	100%	CFAP52	100%	CFAP74	100%
CFB	100%	CFD	100%	CFH	100%	CFHR2	100%
CFHR4	100%	CFHR5	100%	CFI	100%	CFP	100%
CFTR	100%	CHD7	100%	CHUK	100%	CIB1	100%
CIITA	100%	CLCN7	100%	CLEC16A	100%	CLEC7A	100%
CLPB	100%	CLSTN1	100%	CNBP	100%	CNKSR3	100%
COG6	100%	COL4A5	100%	COL4A6	100%	COLEC11	100%
COPA	100%	COPG1	100%	CORO1A	100%	CR2	100%
CRACR2A	100%	CREBBP	100%	CRLF1	99.8%	CRNN	100%
CSF2	100%	CSF2RA	100%	CSF2RB	100%	CSF3R	100%
CTC1	100%	CTLA4	100%	CTNNBL1	100%	CTPS1	100%
CTSC	100%	CXCL10	100%	CXCR2	100%	CXCR4	100%
CYBA	100%	CYBB	100%	CYBC1	100%	DAW1	100%
DBR1	100%	DCLRE1B	100%	DCLRE1C	100%	DDX41	100%
DDX58	100%	DEF6	100%	DEFA6	100%	DGKE	100%
DHFR	100%	DIAPH1	100%	DKC1	100%	DLEC1	100%
DNAAF1	100%	DNAAF2	100%	DNAAF3	100%	DNAAF4	100%
DNAAF5	100%	DNAAF6	100%	DNAH1	100%	DNAH11	100%
DNAH5	100%	DNAH8	100%	DNAH9	100%	DNAI1	100%
DNAI2	100%	DNAJB13	100%	DNAJC21	100%	DNAL1	100%
DNASE1L3	100%	DNASE2	100%	DNMT3B	100%	DOCK2	100%
DOCK8	100%	DPP9	100%	DRC1	100%	DSG1	100%
DSP	100%	DSPP	100%	DTNBP1	100%	DUT	100%
EBF1	100%	EFL1	100%	EIF2AK4	100%	ELANE	100%
ELF1	100%	ELF4	100%	EPCAM	100%	EPG5	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
EPO	100%	EPOR	100%	EPX	100%	ERBIN	100%
ERCC2	100%	ERCC3	100%	ERCC4	100%	ERCC6L2	100%
ETV6	100%	EXTL3	100%	F11	100%	F12	100%
F13A1	100%	F13B	100%	F5	100%	F7	100%
F8	100%	F9	100%	FAAP24	100%	FADD	100%
FAM13A	100%	FANCA	100%	FANCB	100%	FANCC	100%
FANCD2	100%	FANCE	100%	FANCF	100%	FANCG	100%
FANCI	100%	FANCL	100%	FANCM	100%	FARSA	100%
FARSB	100%	FAS	100%	FASLG	100%	FASN	100%
FAT4	100%	FBF1	100%	FBN1	100%	FCGR1A	100%
FCGR2A	100%	FCGR3A	100%	FCGR3B	100%	FCGRT	100%
FCHO1	100%	FCN3	100%	FERMT1	100%	FERMT3	100%
FGA	100%	FGB	100%	FIP1L1	100%	FNIP1	100%
FOCAD	100%	FOXJ1	100%	FOXN1	100%	FOXP3	100%
FPR1	100%	FPR2	100%	FPR3	100%	G6PC2	100%
G6PC3	100%	G6PD	100%	GAB2	100%	GAD1	100%
GAS2L2	100%	GAS8	100%	GATA1	100%	GATA2	100%
GFI1	100%	GINS1	100%	GJC2	100%	GLRX5	100%
GMPS	100%	GNAO1	100%	GNE	100%	GP1BA	100%
GP9	100%	GTF2H5	100%	GUCY2C	100%	HAVCR2	100%
HAX1	100%	HCK	100%	HELLS	100%	HLA-B	100%
HLA-DPA1	100%	HLA-DPB1	100%	HLA-DQA1	100%	HLA-DQB2	100%
HMOX1	100%	HNRNPC	100%	HOXA11	100%	HPS1	100%
HPS3	100%	HPS4	100%	HPS5	100%	HPS6	100%
HTRA2	100%	HYDIN	100%	HYOU1	100%	ICOS	100%
ICOSLG	100%	IFIH1	100%	IFNA17	100%	IFNA2	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
IFNAR1	100%	IFNAR2	100%	IFNG	100%	IFNGR1	100%
IFNGR2	100%	IGLL1	100%	IKBKB	100%	IKBKG	100%
IKZF1	100%	IKZF2	100%	IKZF3	100%	IL10	100%
IL10RA	100%	IL10RB	100%	IL12B	100%	IL12RB1	100%
IL12RB2	100%	IL17A	100%	IL17F	100%	IL17RA	100%
IL17RC	100%	IL18	100%	IL18BP	100%	IL1A	100%
IL1RN	100%	IL2	100%	IL21	100%	IL21R	100%
IL22	100%	IL23A	100%	IL23R	100%	IL2RA	100%
IL2RB	100%	IL2RG	100%	IL31RA	100%	IL36RN	100%
IL6	100%	IL6R	100%	IL6ST	100%	IL7R	100%
INO80	100%	INPP5D	100%	INSR	100%	INVS	100%
IRAK1	100%	IRAK4	100%	IRF2BP2	100%	IRF3	100%
IRF4	100%	IRF7	100%	IRF8	100%	IRF9	100%
ISG15	100%	ITCH	100%	ITGAM	100%	ITGB2	100%
ITK	100%	ITPKB	100%	IVNS1ABP	100%	JAGN1	100%
JAK1	100%	JAK2	100%	JAK3	100%	KDM6A	100%
KIF23	100%	KIT	100%	KLF1	100%	KMT2A	100%
KMT2D	100%	KRAS	100%	KRT20	100%	LAMTOR2	100%
LAT	100%	LAX1	100%	LCK	100%	LCP2	100%
LIG1	100%	LIG4	100%	LPIN2	100%	LRBA	100%
LRIG1	100%	LRRC56	100%	LRRC8A	100%	LSM11	100%
LYST	100%	LYZ	100%	MAD2L2	100%	MAGI1	100%
MAGT1	100%	MALT1	100%	MAN2B1	100%	MAN2B2	100%
MANBA	100%	MAP1LC3B2	100%	MAP3K14	100%	MAPK8	100%
MARS1	100%	MASP1	100%	MASP2	100%	MASTL	100%
MBL2	100%	MC2R	100%	MCIDAS	100%	MCM10	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
MCM4	100%	MDC1	100%	MECOM	100%	MEFV	100%
MLPH	100%	MMAB	100%	MOGS	100%	MPEG1	100%
MPI	100%	MPL	100%	MPO	100%	MPZ	100%
MRE11	100%	MRPL36	100%	MRTFA	100%	MS4A1	100%
MS4A2	100%	MSH5	100%	MSH6	100%	MSN	100%
MST1	100%	MTHFD1	100%	MUC5B	100%	MVK	100%
MX1	100%	MYD88	100%	MYH9	100%	MYO5A	100%
MYO5B	100%	MYSM1	100%	NABP1	100%	NANOS1	100%
NANOS2	100%	NBAS	100%	NBN	100%	NBPF15	100%
NCF1	100%	NCF2	100%	NCF4	100%	NCKAP1L	100%
NCSTN	100%	NEK10	100%	NFAT5	100%	NFE2L2	100%
NFKB1	100%	NFKB2	100%	NFKBIA	100%	NFKBID	100%
NHEJ1	100%	NHP2	100%	NKX2-1	100%	NKX2-5	100%
NLRC3	100%	NLRC4	100%	NLRP1	100%	NLRP12	100%
NLRP2	100%	NLRP3	100%	NME1	100%	NME2	100%
NME5	100%	NME8	100%	NOD2	100%	NOP10	100%
NOS1	100%	NOS2	100%	NPM1	100%	NRAS	100%
NSMCE3	100%	NT5C3A	100%	NUMA1	100%	OAS1	100%
ODAD1	100%	ODAD2	100%	ODAD3	100%	ODAD4	100%
OFD1	100%	ORAI1	100%	ORC3	100%	OSTM1	100%
OTULIN	100%	P2RX7	100%	P4HA2	100%	PALB2	100%
PANK2	100%	PARN	100%	PAX1	100%	PAX5	100%
PCCA	100%	PCCB	100%	PDCD1	100%	PEPD	100%
PGM3	100%	PI4KA	100%	PIGA	100%	PIK3C2A	100%
PIK3CD	100%	PIK3CG	100%	PIK3R1	100%	PIKFYVE	100%
PLCG2	100%	PLEKHM1	100%	PLG	100%	PLPPR2	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
PML	100%	PMM2	100%	PMP22	100%	PMS2	100%
PNP	100%	POLA1	100%	POLD1	100%	POLD2	100%
POLE	100%	POLE2	100%	POLR3A	100%	POLR3C	100%
POLR3F	100%	POMP	100%	POMT1	100%	POT1	100%
POTEF	99.1%	POU2AF1	100%	PPY	100%	PRF1	100%
PRG4	100%	PRIM1	100%	PRKAR1A	100%	PRKCD	100%
PRKD1	100%	PRKDC	100%	PROC	100%	PROS1	100%
PRPS1	100%	PRPS1L1	100%	PRRC2A	100%	PRTN3	100%
PSEN1	100%	PSENEN	100%	PSMA3	100%	PSMA7	100%
PSMB4	100%	PSMB8	100%	PSMB9	100%	PSMG2	100%
PSTPIP1	100%	PTEN	100%	PTPN11	100%	PTPN22	100%
PTPRC	100%	PUS1	100%	PXMP2	100%	RAB27A	100%
RAC2	100%	RAD50	100%	RAD51	100%	RAD51C	100%
RAG1	100%	RAG2	100%	RANBP2	100%	RARA	100%
RASGRP1	100%	RASGRP2	100%	RBBP6	100%	RBCK1	100%
RBM8A	100%	RECQL4	100%	REL	100%	RELA	100%
RELB	100%	RET	100%	RFC1	100%	RFWD3	100%
RFX5	100%	RFXANK	100%	RFXAP	100%	RHOG	100%
RHOH	100%	RIPK1	100%	RNASEH2A	100%	RNASEH2B	100%
RNASEH2C	100%	RNF168	100%	RNF2	100%	RNF31	100%
RNF6	100%	RORC	100%	RPGR	100%	RPL11	100%
RPL15	100%	RPL26	100%	RPL27	100%	RPL31	100%
RPL35	100%	RPL35A	100%	RPL36	100%	RPL5	100%
RPL9	100%	RPS15	100%	RPS15A	100%	RPS19	100%
RPS24	100%	RPS26	100%	RPS27	100%	RPS27A	100%
RPS28	100%	RPS29	100%	RPS7	100%	RPSA	100%

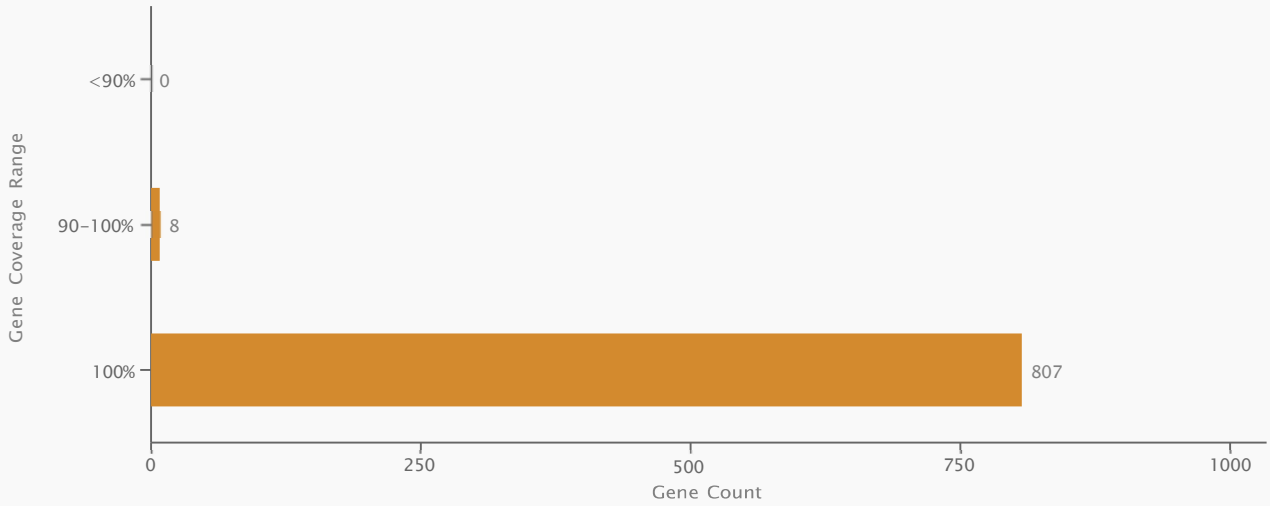
Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
RSPH1	100%	RSPH3	100%	RSPH4A	100%	RSPH9	100%
RTEL1	100%	RUNX1	100%	SAMD3	100%	SAMD9	100%
SAMD9L	100%	SAMHD1	100%	SART3	100%	SASH3	100%
SBDS	100%	SBF2	100%	SCNN1A	100%	SCNN1B	100%
SCNN1G	100%	SDK1	98.1%	SEC14L2	100%	SEC14L3	100%
SEC23B	100%	SEC61A1	100%	SEMA3E	100%	SERAC1	100%
SERPINA1	100%	SERPING1	100%	SF3B1	100%	SFTPA1	100%
SFTPA2	100%	SFTPC	100%	SGPL1	100%	SH2B3	100%
SH2D1A	100%	SH3BP2	100%	SH3KBP1	100%	SKIV2L	100%
SLC19A1	100%	SLC19A2	100%	SLC25A38	100%	SLC29A3	100%
SLC34A2	100%	SLC35A1	100%	SLC35C1	100%	SLC37A4	99.9%
SLC39A4	100%	SLC39A7	100%	SLC41A1	100%	SLC46A1	99.9%
SLC7A7	100%	SLPI	100%	SLX4	100%	SMARCAL1	100%
SMARCD2	100%	SNX10	100%	SOAT1	100%	SOCS1	100%
SP110	100%	SPAG1	100%	SPEF2	100%	SPI1	100%
SPINK5	100%	SPNS1	100%	SPPL2A	100%	SPTLC1	100%
SPTLC2	100%	SRP54	100%	SRP72	100%	STAT1	100%
STAT2	100%	STAT3	100%	STAT4	100%	STAT5A	100%
STAT5B	100%	STIM1	100%	STING1	100%	STK36	100%
STK4	100%	STN1	100%	STON1	100%	STX11	100%
STXBP2	100%	SYK	100%	TADA2A	100%	TAP1	100%
TAP2	100%	TAPBP	100%	TBK1	100%	TBL1XR1	100%
TBX1	95.4%	TBX21	100%	TCF3	100%	TCF7L1	100%
TCIRG1	100%	TCN2	100%	TERF2IP	100%	TERT	100%
TET2	100%	TFG	100%	TFRC	100%	TGFB1	100%
TGFBR1	100%	TGFBR2	100%	THBD	100%	THPO	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
TICAM1	100%	TIMP1	100%	TINF2	100%	TIRAP	100%
TLR1	100%	TLR3	100%	TLR7	100%	TLR8	100%
TMC6	100%	TMC8	100%	TNFAIP3	100%	TNFRSF11A	100%
TNFRSF13B	100%	TNFRSF13C	100%	TNFRSF1A	100%	TNFRSF4	100%
TNFRSF6B	100%	TNFRSF9	100%	TNFSF11	100%	TNFSF12	100%
TNFSF13	100%	TNIP1	100%	TOP2B	100%	TP53	100%
TPP1	100%	TPP2	100%	TRADD	100%	TRAF3	100%
TRAF3IP2	100%	TREX1	100%	TRIM22	100%	TRNT1	100%
TSC1	100%	TSC2	100%	TSR2	100%	TTC12	100%
TTC37	100%	TTC7A	100%	TUBB	100%	TUBB1	100%
TYK2	100%	UBA1	100%	UBE2T	100%	UBL4A	100%
UBXN10	100%	UNC119	100%	UNC13D	100%	UNC93B1	100%
UNG	100%	USB1	100%	USP18	100%	VPS13B	100%
VPS45	100%	WAS	100%	WDR1	100%	WIPF1	100%
WRAP53	100%	WWOX	100%	WWTR1	100%	XIAP	100%
XK	100%	XRCC2	100%	YARS2	100%	ZAP70	100%
ZBTB16	100%	ZBTB24	100%	ZMYND10	100%	ZNF160	100%
ZNF341	100%	ZNF395	100%	ZNFX1	100%		

Gene Coverage Distribution



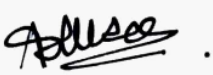
QC Metrics

Total aligned reads	99.94 %
Total reads	95.34 (M)
Total data generated	14.12 (Gb)
Total reads which passed mapping quality cutt-off	13.08 (Gb)

Reviewed By

Dr. Sweta Das
Reviewed by
Clinical Geneticist

Dr. Neha Agrawal
Reviewed by
Clinical Geneticist



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