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Patient Details

Name	: Mr. K ANAND	**Sex / Age**	: Male / 21 Years	**Case ID**	: 51022802630
Ref By	:	**PT. ID**	:	**Test Name**	: ORION (WES-Whole Exome Sequencing), Mitochondrial Part of ORION
Bill. Loc.	: SAGEPATH LABS PVT LTD				

Sample Details

Registration Date & Time	: 2025-10-18 15:48:26	**Sample Type**	: Whole Blood EDTA	**Sample Date & Time**	: 2025-10-18 15:48:00
Ref ID 1.	: -	**Report Date & Time**	: 2025-11-12 16:29:37		

Clinical History

Salient features: Lt UL & LL involuntary movements, Lt UL pill-rolling movement, Lt UL muscle wasting, wasting of thenar & hypothenar muscles, left UL and LL paresthesia, UTI, hypocalcemia, c/o hair fall, general weakness, h/o TIA right sided, Demyelinating disease with focal seizures, Recurrent ADEM, Peripheral neuropathy, Asymmetrical motor sensory axonal demyelinating polyneuropathy
Investigations: MRI whole spine with contrast s/o subtle T2/STIR hyperintense foci with heterogeneous enhancement noted in both halves of spinal cord (left more than right) at C5-C6 level and involving the anterior grey matter (anterolateral aspect). Mild thickening of cord noted extending from cervical levels. Mild posterior annular disc bulge indentation on anterior thecal sac at C3-C4, C4-C5 and C5-C6 level. Schmorl's node is noted at L2-L3 level; Motor nerve conduction studies s/o asymmetrical motor sensory axonal demyelinating polyneuropathy.

Test Results and Interpretation

HEMIZYGOUS PATHOGENIC VARIANT IN GJB1 GENE CONSISTENT WITH PHENOTYPE DETECTED.

ADDITIONAL HETEROZYGOUS PATHOGENIC VARIANT IN MYBPC3 GENE DETECTED. CLINICAL CORRELATION RECOMMENDED.

MITOCHONDRIAL GENOME SEQUENCING IS NEGATIVE.

Summary Of Variants

Gene and Transcript	Involved Exon-Intron/ Total Exon-Intron	Variant Nomenclature	Zygosity	Classification	OMIM Phenotype	Inheritance
GJB1 (NM_000166.6)	Exon 2/Exon 2	c.548G>A p.Arg183His [41x /41x]	Hemizygous	Pathogenic	Charcot-Marie-Tooth neuropathy, X-linked dominant,]	X-linked dominant

Summary Of Variants

Gene and Transcript	Involved Exon-Intron/ Total Exon-Intron	Variant Nomenclature	Zygosity	Classification	OMIM Phenotype	Inheritance
MYBPC3 (NM_000256.3)	Intron 23/Intron 34	c.2308+1G>A - [32x /61x]	Heterozygous	Pathogenic	Cardiomyopathy, hypertrophic, 4	Autosomal dominant, Autosomal recessive

Variant Details

GJB1

Variant Nomenclature	c.548G>A (p.Arg183His)
Genomic Nomenclature	chrX:g.70444105G>A
Zygosity	Hemizygous

Type of variant	gnomAD frequency	Computational evidences	ClinVar	LOF disease mechanism of action	Downstream LOF	Previously reported [reported zygosity]	Variant references
Missense Variant	Absent	**REVEL:** 0.959 **CADD:** 28.1	Pathogenic **(Multiple submission)**	NA	NA	Yes [Hemizygous/ Heterozygous]	Chen CX, et. al.,2020 Niu J, et. al.,2020 Chen B, et. al.,2019

Another missense variant [p.Arg183Cys] on the same residue / position of this gene has previously been reported to be disease causing (Hahn AF, et. al., 2016), suggesting that this residue might be of clinical significance. Experimental studies have shown that this missense change affects GJB1 function (Tsai PC, et. al.,2016; Chen CX, et. al., 2020). For these reasons, this variant has been classified as Pathogenic.

References:

1. Chen CX, et. al., Identification and functional characterization of novel GDAP1 variants in Chinese patients with Charcot-Marie-Tooth disease. Ann Clin Transl Neurol. 2020 Dec;7(12):2381-2392.
2. Niu J, et. al., *GJB1* Mutation-A Disease Spectrum: Report of Case Series. Front Neurol. 2020 Jan 15;10:1406.
3. Chen B, et. al., Three novel mutations in a group of Chinese patients with X-linked Charcot-Marie-Tooth disease. Clin Neurol Neurosurg. 2019 Sep;184:105430. doi: 10.1016/j.clineuro.2019.105430. Epub 2019 Jul 10. PMID: 31323543.
4. Hahn AF, et. al., Pathological findings in the x-linked form of Charcot-Marie-Tooth disease: a morphometric and ultrastructural analysis. Acta Neuropathol. 2001 Feb;101(2):129-39.
5. Tsai PC, et. al., Clinical and biophysical characterization of 19 *GJB1* mutations. Ann Clin Transl Neurol. 2016 Sep 1;3(11):854-865.

Variant Details

6. Chen CX, et. al., Identification and functional characterization of novel GDAP1 variants in Chinese patients with Charcot-Marie-Tooth disease. Ann Clin Transl Neurol. 2020 Dec;7(12):2381-2392.

Disease

CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT, 1 (OMIM gene ID: 302800)

GJB1 disorders are typically characterized by peripheral motor and sensory neuropathy with or without fixed CNS abnormalities and/or acute, self-limited episodes of transient neurologic dysfunction (especially weakness and dysarthria). Peripheral neuropathy typically manifests in affected males between ages five and 25 years. Although both men and women are affected, manifestations tend to be less severe in women, some of whom may remain asymptomatic. Less commonly, initial manifestations in some affected individuals are stroke-like episodes (acute fulminant episodes of reversible CNS dysfunction).

MYBPC3

Variant Nomenclature	c.2308+1G>A -
Genomic Nomenclature	chr11:g.47360070C>T
Zygosity	Heterozygous

Type of variant	gnomAD frequency	Computational evidences	ClinVar	LOF disease mechanism of action	Downstream LOF	Previously reported [reported zygosity]	Variant references
Splice Donor Variant	Absent	**SPLICE AI:** 0.93 [Damaging]	Pathogenic **(Multiple submission)**	Yes	Yes	Yes [Heterozygous]	Carrier L, et. al.,1997 van Lint FHM, et. al., 2019
This variant is also known as IVS24 +1 G-A. It has also been observed to segregate with disease in related individuals. For these reasons, this variant has been classified as Pathogenic.							

References:

1. Carrier L, et. al., Organization and sequence of human cardiac myosin binding protein C gene (MYBPC3) and identification of mutations predicted to produce truncated proteins in familial hypertrophic cardiomyopathy. Circ Res. 1997 Mar;80(3):427-34. PMID: 9048664.
2. van Lint FHM, et. al., Large next-generation sequencing gene panels in genetic heart disease: yield of pathogenic variants and variants of unknown significance. Neth Heart J. 2019 Jun;27(6):304-309.

Disease

CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 4 (OMIM gene ID: 600958)

Nonfamilial hypertrophic cardiomyopathy tends to be milder. This form typically begins later in life than familial hypertrophic cardiomyopathy, and affected individuals have a lower risk of serious cardiac events and sudden death than people with the familial form. While most people with familial hypertrophic cardiomyopathy are

Variant Details

symptom-free or have only mild symptoms, this condition can have serious consequences. It can cause abnormal heart rhythms (arrhythmias) that may be life threatening. People with familial hypertrophic cardiomyopathy have an increased risk of sudden death, even if they have no other symptoms of the condition. A small number of affected individuals develop potentially fatal heart failure, which may require heart transplantation. The symptoms of familial hypertrophic cardiomyopathy are variable, even within the same family. Many affected individuals have no symptoms. Other people with familial hypertrophic cardiomyopathy may experience chest pain; shortness of breath, especially with physical exertion; a sensation of fluttering or pounding in the chest (palpitations); lightheadedness; dizziness; and fainting. familial hypertrophic cardiomyopathy, cardiac thickening usually occurs in the interventricular septum, which is the muscular wall that separates the lower left chamber of the heart (the left ventricle) from the lower right chamber (the right ventricle). In some people, thickening of the interventricular septum impedes the flow of oxygen-rich blood from the heart, which may lead to an abnormal heart sound during a heartbeat (heart murmur) and other signs and symptoms of the condition. Other affected individuals do not have physical obstruction of blood flow, but the pumping of blood is less efficient, which can also lead to symptoms of the condition. Familial hypertrophic cardiomyopathy often begins in adolescence or young adulthood, although it can develop at any time throughout life. Hypertrophic cardiomyopathy is a heart condition characterized by thickening (hypertrophy) of the heart (cardiac) muscle. When multiple members of a family have the condition, it is known as familial hypertrophic cardiomyopathy. Hypertrophic cardiomyopathy also occurs in people with no family history; these cases are considered nonfamilial hypertrophic cardiomyopathy.

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.**
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.**

Test Information

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.**
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

Technical Notes

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: 15.10.6

Gene Coverage

Indication Based Analysis:

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
AAAS	100%	AARS1	100%	AARS2	100%	AASS	100%
ABCA1	100%	ABCA7	100%	ABCB11	100%	ABCB4	100%
ABCB6	100%	ABCB7	100%	ABCC8	100%	ABCC9	100%
ABCD1	100%	ABHD12	100%	ABHD16A	100%	ACADVL	100%
ACAT2	100%	ACBD5	100%	ACD	100%	ACO2	100%
ACP5	100%	ACTA1	100%	ACTA2	100%	ACTL6B	100%
ADA2	100%	ADAM22	100%	ADAMTS13	100%	ADAMTS15	100%
ADAMTS3	100%	ADAR	100%	ADCY5	100%	ADGRG1	100%
ADGRV1	100%	ADH1C	100%	ADPRS	100%	ADRA2B	100%
ADSL	100%	AFF3	100%	AFG3L2	100%	AGRN	100%
AGTPBP1	100%	AHCY	100%	AHI1	100%	AHNAK2	100%
AIFM1	100%	AIP	100%	AIRE	100%	AKR1B1	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
AKT1	100%	AKT3	100%	ALAD	100%	ALDH18A1	100%
ALDH7A1	100%	ALG12	100%	ALG2	100%	ALK	100%
ALS2	100%	AMACR	100%	AMPD2	100%	ANAPC2	99.9%
ANG	100%	ANO10	100%	ANO3	100%	ANOS1	100%
ANXA11	100%	AOC1	100%	AP1S2	100%	AP1S3	100%
AP2M1	100%	AP3B2	100%	AP5Z1	100%	APC2	100%
APOA1	100%	APOE	100%	APP	100%	APTX	100%
AR	100%	AREG	100%	ARHGEF10	100%	ARL13B	100%
ARL3	100%	ARL6IP1	100%	ARMC9	100%	ARSA	100%
ARVCF	100%	ARX	100%	ASAH1	100%	ASL	100%
ASNS	100%	ATAD1	100%	ATAD3A	99.5%	ATAT1	100%
ATCAY	100%	ATG7	100%	ATL1	100%	ATL3	100%
ATM	100%	ATN1	100%	ATP11A	100%	ATP13A2	100%
ATP1A1	100%	ATP1A2	100%	ATP1A3	100%	ATP1A4	100%
ATP2A1	100%	ATP2B3	100%	ATP6AP2	100%	ATP6V0A1	100%
ATP6V0C	100%	ATP6V1A	100%	ATP6V1B2	100%	ATP7A	100%
ATP7B	100%	ATP8B1	100%	ATXN1	100%	ATXN10	100%
ATXN2	100%	ATXN3	100%	ATXN7	100%	AUH	100%
AURKAIP1	100%	B2M	100%	B4GALNT1	100%	B9D1	100%
B9D2	100%	BAG3	100%	BANK1	100%	BAP1	100%
BAZ1B	100%	BCKDK	100%	BCL11B	100%	BEAN1	100%
BICD2	100%	BLK	100%	BMPR1A	100%	BOLA3	100%
BRAT1	100%	BRCA2	100%	BRD1	100%	BRPF1	100%
BSCL2	100%	BTK	100%	BUD23	100%	C19orf12	100%
C4A	99.7%	C4B	100%	C9orf72	100%	CA2	100%
CA8	100%	CABP2	100%	CABP4	100%	CACNA1A	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
CACNA1B	100%	CACNA1C	100%	CACNA1D	100%	CACNA1E	100%
CACNA1G	100%	CACNA1H	100%	CACNA1S	100%	CACNA2D1	98.5%
CACNA2D2	100%	CACNB4	100%	CADM3	100%	CALR	100%
CAMK2A	100%	CAMKMT	100%	CAMLG	100%	CAMTA1	100%
CAPN1	100%	CAPRIN1	100%	CARS1	100%	CARS2	100%
CASK	100%	CASR	100%	CAV1	100%	CBLIF	100%
CBY1	100%	CC2D2A	100%	CCBE1	100%	CCDC141	100%
CCDC88A	100%	CCDC88C	100%	CCL13	100%	CCL14	100%
CCM2	100%	CCND1	100%	CCNF	100%	CCR1	100%
CCT5	100%	CD28	100%	CD40LG	100%	CD59	100%
CDC42BPB	100%	CDH23	100%	CDK19	100%	CDKL5	100%
CELF2	100%	CEP104	100%	CEP120	100%	CEP126	100%
CEP41	100%	CEP85L	100%	CERS1	100%	CFAP410	100%
CFH	100%	CHAT	100%	CHCHD10	100%	CHCHD2	100%
CHD2	100%	CHD7	100%	CHEK2	100%	CHKA	100%
CHMP1A	100%	CHMP1B	100%	CHMP2B	100%	CHRNA2	100%
CHRNA4	100%	CHRNB2	100%	CIC	100%	CILK1	100%
CIZ1	100%	CLCF1	100%	CLCN2	100%	CLCN4	100%
CLCN7	100%	CLCNKB	100%	CLDN16	100%	CLIP2	100%
CLN3	100%	CLN5	100%	CLN6	100%	CLN8	100%
CLP1	100%	CLPB	100%	CLSTN1	100%	CLTC	100%
CLTCL1	100%	CLTRN	100%	CNBP	100%	CNKSR2	98.6%
CNKSR3	100%	CNP	100%	CNPY3	100%	CNTN2	100%
CNTNAP1	100%	CNTNAP2	100%	COA3	100%	COA7	100%
COA8	100%	COG8	100%	COL13A1	100%	COL25A1	100%
COL4A1	100%	COL6A3	100%	COLEC12	100%	COMP	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
COMT	100%	COQ2	100%	COQ4	100%	COQ5	100%
COQ6	100%	COQ7	100%	COQ8A	100%	COX10	100%
COX20	100%	COX4I1	100%	COX6A1	100%	CP	100%
CPA6	100%	CPLANE1	99.7%	CPLX1	100%	CPOX	100%
CPSF3	100%	CR2	100%	CRAT	100%	CREBBP	100%
CRELD1	100%	CRH	100%	CRLF1	98.9%	CRLS1	100%
CRYAB	100%	CSF1R	100%	CSPP1	100%	CSTB	100%
CTC1	100%	CTDP1	100%	CTDSP2	100%	CTH	100%
CTLA4	100%	CTNND2	100%	CTNS	100%	CTSF	100%
CTSH	100%	CUBN	100%	CUL4B	100%	CUX2	100%
CWF19L1	100%	CXXC1	100%	CYB5A	100%	CYB5R3	98.6%
CYFIP2	100%	CYP27A1	100%	CYP27B1	100%	CYP2A6	100%
CYP2R1	100%	CYP2U1	100%	CYP3A4	100%	CYP7B1	100%
DAB1	100%	DALRD3	100%	DAO	100%	DARS2	100%
DCAF17	100%	DCAF8	100%	DCC	100%	DCTN1	100%
DCTN2	100%	DDC	100%	DDHD1	100%	DDHD2	100%
DDOST	100%	DEAF1	100%	DENND5A	100%	DEPDC5	100%
DGAT2	100%	DGCR2	100%	DGCR6	100%	DGCR6L	100%
DGCR8	100%	DGUOK	100%	DHDDS	100%	DHFR	100%
DHH	100%	DHTKD1	100%	DHX30	100%	DIAPH3	100%
DLAT	100%	DLST	100%	DMD	100%	DMP1	100%
DMTF1	100%	DMXL2	100%	DNAAF3	100%	DNAJB2	100%
DNAJC13	100%	DNAJC19	100%	DNAJC3	100%	DNAJC30	100%
DNAJC5	100%	DNAJC6	100%	DNASE1	100%	DNM1	100%
DNM1L	100%	DNM2	100%	DNMT1	100%	DNMT3A	100%
DOCK7	100%	DPAGT1	100%	DPM1	100%	DPYD	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
DPYSL2	100%	DPYSL5	100%	DRD2	100%	DRD3	100%
DRP2	100%	DSCAM	99.3%	DST	100%	DTYMK	100%
DUSP6	100%	DXO	100%	DYNC1H1	100%	DYSF	100%
EBF3	100%	ECEL1	100%	EDNRB	100%	EEF1A2	100%
EEF2	100%	EFHC1	100%	EGFL7	100%	EGR2	100%
EHMT1	99.6%	EIF2AK2	100%	EIF4A3	100%	EIF4G1	100%
EIF4H	100%	ELN	100%	ELOVL1	100%	ELOVL4	100%
ELOVL5	100%	ELP1	100%	ELP2	100%	EMILIN1	100%
ENPP1	100%	EP300	100%	EPAS1	100%	EPCAM	100%
EPM2A	100%	EPRS1	100%	ERAP1	100%	ERBB3	100%
ERBB4	100%	ERCC2	100%	ERCC3	100%	ERCC4	100%
ERCC5	100%	ERCC6	100%	ERCC8	100%	ERGIC1	100%
ERLIN1	100%	ERLIN2	100%	ESS2	100%	ETHE1	100%
ETS1	100%	EXOSC3	100%	EXOSC8	100%	EXOSC9	100%
EXTL3	100%	FA2H	100%	FAM111A	100%	FAM149B1	100%
FAR1	100%	FARS2	100%	FARSB	100%	FAS	100%
FASN	100%	FAT4	100%	FBLN5	100%	FBXL4	100%
FBXO28	100%	FBXO38	100%	FBXO7	100%	FCGR2B	100%
FCGR3B	100%	FDXR	100%	FEZF1	100%	FGD4	100%
FGF12	100%	FGF13	100%	FGF14	100%	FGF17	100%
FGF23	100%	FGF8	100%	FGFR1	100%	FH	100%
FIG4	100%	FKBP6	100%	FKRP	100%	FLNC	100%
FLRT3	100%	FLVCR1	100%	FMR1	100%	FOCAD	100%
FOXG1	100%	FOXP3	100%	FOXRED1	100%	FRMD5	100%
FRMPD4	100%	FRRS1L	100%	FTH1	100%	FTL	100%
FUCA1	100%	FUS	100%	FXN	100%	FXR1	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
FZR1	100%	GAA	100%	GABBR2	100%	GABRA1	100%
GABRA2	100%	GABRA3	100%	GABRA5	100%	GABRB2	100%
GABRB3	100%	GABRD	100%	GABRG2	100%	GAL	100%
GALC	100%	GALP	100%	GALT	100%	GAMT	100%
GAN	100%	GARS1	100%	GART	100%	GATA1	100%
GATA3	100%	GBA2	100%	GBE1	100%	GBF1	100%
GCDH	100%	GCH1	95.7%	GCK	100%	GCLC	100%
GCM2	100%	GCSH	100%	GDAP1	100%	GDF1	100%
GEMIN4	100%	GET4	100%	GFAP	100%	GFM2	100%
GGT1	100%	GIGYF2	100%	GJA5	100%	GJA8	100%
GJB1	100%	GJB3	100%	GJC2	99.8%	GLA	100%
GLB1	100%	GLDC	100%	GLE1	100%	GLRA1	100%
GLRB	100%	GLRX5	100%	GLT8D1	100%	GLYCTK	100%
GM2A	100%	GMPPB	100%	GNA11	100%	GNA14	100%
GNAO1	100%	GNAS	100%	GNB1	100%	GNB2	100%
GNB4	100%	GNPTAB	100%	GOLGA2	100%	GON7	100%
GOSR2	100%	GP1BB	100%	GPAA1	100%	GPHN	100%
GPR101	100%	GPR88	100%	GPRIN1	100%	GPT2	100%
GRIA2	100%	GRIA3	100%	GRIA4	100%	GRIK2	100%
GRIK5	100%	GRIN1	100%	GRIN2A	100%	GRIN2B	100%
GRIN2D	100%	GRM1	100%	GRM7	100%	GRN	100%
GSN	100%	GSS	100%	GTF2E2	100%	GTF2H5	100%
GTF2I	99.7%	GTF2IRD1	100%	GTF2IRD2	100%	GTF2IRD2B	100%
GTPBP2	100%	GUF1	100%	HACE1	100%	HADHA	100%
HADHB	100%	HARS1	100%	HCFC1	100%	HCN1	100%
HCRT	100%	HCRTR1	100%	HESX1	100%	HEXA	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
HEXB	100%	HEXIM1	100%	HFE	100%	HIBCH	100%
HIKESHI	100%	HINT1	100%	HIRA	100%	HK1	100%
HLA-B	100%	HLA-DQA1	100%	HLA-DQB2	100%	HMBS	100%
HMGA2	100%	HMGCL	100%	HNF1A	100%	HNF4A	100%
HNRNPA1	100%	HNRNPA2B1	100%	HNRNPU	100%	HOOK1	100%
HOXD10	100%	HPCA	100%	HPDL	100%	HPRT1	100%
HS6ST1	100%	HSD17B10	100%	HSD17B4	100%	HSPB1	100%
HSPB3	100%	HSPB8	100%	HSPD1	100%	HTRA1	100%
HTRA2	100%	HTT	100%	HYLS1	100%	IARS2	100%
IBA57	100%	IDUA	100%	IER3IP1	100%	IFIH1	100%
IFNGR1	100%	IFRD1	100%	IFT122	100%	IFT74	100%
IGHMBP2	100%	IL10	100%	IL12A	100%	IL17RD	100%
IL23R	100%	IL36RN	100%	IMPDH2	100%	INF2	94.3%
INPP5E	100%	INTS11	100%	IRAK1	100%	IREB2	100%
IRF2BPL	100%	IRF4	100%	IRF5	100%	ITGAM	100%
ITM2B	100%	ITPA	100%	ITPR1	100%	ITPR3	100%
IVD	100%	IVNS1ABP	100%	JAG1	100%	JAK2	100%
JAZF1	100%	JMJD1C	100%	JPH1	100%	JPH3	100%
JRK	99.9%	KARS1	100%	KATNIP	100%	KBTBD13	100%
KCNA1	100%	KCNA2	100%	KCNA4	100%	KCNB1	100%
KCNC1	100%	KCNC2	100%	KCNC3	95.2%	KCND3	100%
KCNH5	100%	KCNJ1	100%	KCNJ10	100%	KCNJ11	100%
KCNK4	100%	KCNK9	100%	KCNMA1	100%	KCNN2	100%
KCNQ2	100%	KCNQ3	100%	KCNT1	100%	KCTD17	100%
KCTD7	100%	KDELR2	100%	KDM1A	100%	KDM6A	100%
KIAA0319L	100%	KIAA0586	100%	KIAA0753	100%	KIF1A	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
KIF1B	100%	KIF1C	100%	KIF5A	100%	KIF7	100%
KLC2	100%	KLC3	100%	KLHL41	100%	KLRC4	100%
KMT2B	99.1%	KMT2D	100%	KNSTRN	100%	KPNA3	100%
KRAS	100%	KRIT1	100%	KRT14	100%	KRT17	100%
KRT5	100%	LAMA1	100%	LAMA2	100%	LARS2	100%
LAS1L	100%	LDB3	100%	LDLR	100%	LEMD2	100%
LEMD3	100%	LGI1	100%	LGI3	100%	LIAS	100%
LIFR	100%	LIG3	100%	LIMK1	100%	LIN28B	100%
LITAF	100%	LMAN2L	100%	LMNA	100%	LMNB1	100%
LMNB2	100%	LMO1	100%	LMX1B	100%	LNPK	96.8%
LONP1	100%	LPIN1	100%	LRIG3	100%	LRP12	100%
LRPAP1	100%	LRPPRC	100%	LRRK2	100%	LRSAM1	100%
LSM11	100%	LYST	100%	LZTR1	100%	MACF1	100%
MAG	100%	MAGI1	100%	MAN1B1	100%	MAOA	100%
MAP4K2	100%	MAPK10	100%	MAPRE3	100%	MAPT	100%
MARCHF6	100%	MARS1	100%	MAST3	100%	MATR3	100%
MAX	100%	MCM2	100%	MCM3AP	100%	MCM7	100%
MDH2	100%	MECP2	99.5%	MECR	100%	MED11	100%
MED12	100%	MED17	100%	MED23	100%	MED25	100%
MED9	100%	MEFV	100%	MEGF10	100%	MEN1	100%
METTL27	100%	MFN2	100%	MICAL1	100%	MICOS13	100%
MICU1	100%	MINPP1	100%	MITF	100%	MKS1	100%
MLH1	100%	MMAA	100%	MMACHC	100%	MME	100%
MMP12	100%	MMUT	100%	MOCS1	100%	MOCS2	100%
MOG	100%	MORC2	100%	MPC1	100%	MPL	100%
MPLKIP	100%	MPV17	100%	MPZ	100%	MRAP	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
MRE11	100%	MRM2	100%	MRPL12	100%	MRPS34	100%
MSH2	100%	MSH6	100%	MSTO1	100%	MTCH1	100%
MTFMT	100%	MTHFR	100%	MTMR2	100%	MTOR	100%
MTPAP	99.4%	MTTP	100%	MUSK	100%	MUTYH	100%
MYBPC1	100%	MYBPC3	100%	MYCN	100%	MYD88	100%
MYH14	100%	MYH7	100%	MYH7B	100%	MYL2	100%
MYO5A	100%	MYO9A	100%	MYORG	100%	MYOT	100%
MYPN	100%	NADK2	100%	NAGA	100%	NAGLU	100%
NAGS	100%	NARS1	100%	NARS2	99.5%	NAT8	100%
NAXD	100%	NAXE	100%	NCF1	99.2%	NDE1	100%
NDNF	100%	NDP	100%	NDRG1	100%	NDUFA1	100%
NDUFA13	100%	NDUFA4	100%	NDUFA9	100%	NDUFAF3	100%
NDUFAF5	100%	NDUFS1	100%	NDUFS2	100%	NDUFS4	100%
NDUFS8	100%	NDUFV1	100%	NEB	100%	NECAP1	100%
NEFH	100%	NEFL	99.9%	NEK1	100%	NEMF	100%
NEU1	100%	NEUROD2	100%	NEUROG1	100%	NEXMIF	100%
NF1	100%	NF2	100%	NFASC	100%	NFE2L2	100%
NFU1	100%	NGF	100%	NGLY1	100%	NHLRC1	100%
NHLRC2	100%	NIPA1	100%	NKX2-1	100%	NKX6-2	100%
NMNAT2	100%	NOL3	100%	NONO	100%	NOP56	100%
NOTCH3	100%	NPHP1	100%	NPPC	100%	NPRL2	100%
NPRL3	100%	NPTX1	100%	NR1H4	100%	NR4A2	100%
NRAS	100%	NSD1	100%	NSUN3	100%	NT5C2	100%
NTNG1	100%	NTNG2	100%	NTRK1	100%	NTRK2	100%
NUP214	100%	NUP54	100%	NUP62	100%	NUS1	100%
OBSCN	100%	OCA2	100%	OCRL	100%	ODC1	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
OFD1	100%	OMA1	100%	OPA1	100%	OPA3	100%
OPHN1	100%	OPTN	100%	ORAI1	99.3%	OSTM1	100%
OTOF	100%	OXR1	100%	P2RY11	100%	P4HA2	100%
PACS2	100%	PAH	100%	PAK1	100%	PANK2	100%
PARK7	100%	PARS2	100%	PC	100%	PCBD1	100%
PCDH10	100%	PCDH19	100%	PDCD1	100%	PDCD10	100%
PDCD6	100%	PDE10A	100%	PDE2A	100%	PDE6D	100%
PDE8B	100%	PDGFB	100%	PDGFRB	100%	PDHA1	100%
PDK3	100%	PDP1	100%	PDSS1	100%	PDSS2	100%
PDYN	100%	PET100	100%	PEX10	100%	PEX11B	100%
PEX12	100%	PEX16	100%	PEX2	100%	PEX6	100%
PEX7	100%	PFN1	100%	PGAP1	100%	PGAP2	100%
PGAP3	100%	PGK1	100%	PGM3	100%	PHACTR1	100%
PHGDH	100%	PHIP	100%	PHOX2B	100%	PHYH	100%
PI4K2A	100%	PI4KA	100%	PIBF1	100%	PIGA	100%
PIGB	100%	PIGL	100%	PIGN	100%	PIGO	100%
PIGP	100%	PIGQ	100%	PIGT	100%	PIGV	100%
PIGW	100%	PIGY	100%	PIK3C2A	100%	PIK3CA	100%
PIK3CD	100%	PIK3R5	100%	PINK1	100%	PITRM1	100%
PITX3	100%	PJVK	100%	PKN1	100%	PLA2G6	100%
PLCB1	100%	PLEKHG5	100%	PLEKHM1	100%	PLOD3	100%
PLP1	100%	PLPBP	100%	PLPP6	100%	PLVAP	100%
PMM2	100%	PMP2	100%	PMP22	100%	PMPCA	100%
PMS1	100%	PMS2	100%	PNKD	100%	PNKP	100%
PNP	100%	PNPLA2	100%	PNPLA6	100%	PNPO	100%
PNPT1	100%	PODXL	99.6%	POLD1	100%	POLE	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
POLG	100%	POLG2	100%	POLR1A	100%	POLR1C	100%
POLR1D	100%	POLR3A	100%	POLR3B	100%	POLR3K	100%
POMGNT1	100%	POMK	100%	POMT1	100%	POMT2	100%
PON1	100%	PON2	100%	PON3	100%	POU3F3	90.1%
POU3F4	100%	POU4F1	94.6%	PPARGC1A	100%	PPFIBP1	100%
PPM1B	100%	PPOX	100%	PPP1R15B	100%	PPP2CA	100%
PPP2R2B	100%	PPP3CA	100%	PPT1	100%	PPY	100%
PRDM12	93.8%	PRDM8	100%	PRDX1	100%	PRDX3	100%
PRDX5	100%	PRDX6	100%	PREPL	100%	PRICKLE1	100%
PRKAR1B	100%	PRKCG	100%	PRKN	100%	PRKRA	100%
PRNP	100%	PROC	100%	PROK2	100%	PROKR2	100%
PRORP	100%	PRPH	100%	PRPH2	100%	PRPS1	100%
PRPS1L1	100%	PRRT2	100%	PRUNE1	100%	PRX	100%
PSAP	100%	PSAT1	100%	PSEN1	100%	PSEN2	100%
PSMC1	100%	PSMC3	100%	PTCD3	100%	PTEN	100%
PTGES2	100%	PTGS1	100%	PTGS2	100%	PTH	100%
PTPA	100%	PTPN22	100%	PTPRA	100%	PTRH2	100%
PTRHD1	100%	PTS	100%	PUM1	100%	PURA	100%
PUS1	100%	PUS3	100%	PUS7	100%	PXK	100%
PXMP2	100%	PYCR1	100%	PYCR2	100%	QDPR	100%
QRICH1	100%	RAB18	100%	RAB39B	100%	RAB3GAP2	98.4%
RAB7A	100%	RAI1	100%	RAPGEF2	100%	RARS1	100%
RARS2	100%	RBM10	100%	RBM15	100%	RBM7	100%
REEP1	100%	REEP2	100%	RELN	100%	REPS1	100%
RET	100%	RETREG1	100%	RFC1	100%	RFC2	100%
RFT1	100%	RFX7	100%	RHOBTB2	100%	RILPL1	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
RLIM	100%	RMND1	100%	RNASE12	100%	RNASEH1	100%
RNASEH2A	100%	RNASEH2B	100%	RNASEH2C	100%	RNASET2	100%
RNF113A	100%	RNF2	100%	RNF216	100%	ROR1	100%
RORA	100%	RORB	100%	RPGRIP1L	100%	RPIA	100%
RPL10	100%	RPL11	100%	RPL15	100%	RPS20	100%
RRAGD	100%	RREB1	100%	RRM1	100%	RRM2B	100%
RTN2	100%	RUSC2	100%	RYR1	100%	SACS	100%
SAMD12	100%	SAMD9L	100%	SAMHD1	100%	SATB1	100%
SBF1	100%	SBF2	100%	SC5D	100%	SCARB2	100%
SCN10A	100%	SCN11A	100%	SCN1A	100%	SCN1B	100%
SCN2A	100%	SCN3A	100%	SCN4A	100%	SCN8A	100%
SCN9A	100%	SCO2	100%	SCP2	100%	SCYL1	100%
SCYL2	100%	SDHA	100%	SDHAF2	100%	SDHB	100%
SDHC	100%	SDHD	100%	SEC24C	100%	SELENOI	100%
SEMA3A	100%	SEMA4A	100%	SEMA6B	100%	SEPSECS	100%
SEPTIN9	99.7%	SERPING1	100%	SERPINI1	100%	SETX	100%
SFTPA1	100%	SFTPA2	100%	SFXN4	100%	SGCD	100%
SGCE	100%	SGPL1	100%	SH2B1	100%	SH2B3	100%
SH2D3C	100%	SH3TC2	100%	SHQ1	100%	SIGMAR1	100%
SIK1	100%	SLA2	100%	SLC12A1	100%	SLC12A3	100%
SLC12A5	100%	SLC12A6	100%	SLC13A5	100%	SLC16A2	100%
SLC17A5	100%	SLC17A8	100%	SLC18A2	100%	SLC18A3	100%
SLC19A1	100%	SLC19A2	100%	SLC19A3	100%	SLC1A2	100%
SLC1A3	100%	SLC1A4	100%	SLC20A2	100%	SLC22A5	100%
SLC25A1	100%	SLC25A10	100%	SLC25A11	100%	SLC25A12	100%
SLC25A13	100%	SLC25A15	100%	SLC25A19	100%	SLC25A21	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
SLC25A22	100%	SLC25A23	100%	SLC25A4	100%	SLC25A42	100%
SLC25A46	100%	SLC26A2	100%	SLC2A1	100%	SLC2A3	100%
SLC30A10	100%	SLC30A9	100%	SLC32A1	100%	SLC33A1	100%
SLC38A3	100%	SLC39A13	100%	SLC39A14	100%	SLC39A4	100%
SLC3A1	100%	SLC44A1	100%	SLC46A1	99.9%	SLC4A1	100%
SLC4A10	100%	SLC52A1	100%	SLC52A2	100%	SLC52A3	100%
SLC5A6	100%	SLC5A7	100%	SLC6A1	100%	SLC6A17	100%
SLC6A19	100%	SLC6A2	100%	SLC6A3	100%	SLC6A4	100%
SLC6A5	100%	SLC6A8	100%	SLC6A9	100%	SLC7A6OS	100%
SLC9A1	100%	SLC9A3R1	100%	SMAD2	100%	SMARCB1	100%
SMARCE1	100%	SMC1A	100%	SMG9	100%	SMIM6	100%
SMN1	100%	SMN2	100%	SMO	100%	SMOX	100%
SMPD1	100%	SMS	100%	SNAP25	100%	SNAP29	100%
SNCA	100%	SNCAIP	100%	SNRPN	100%	SNX10	100%
SOAT2	100%	SOD1	100%	SORD	88.6%	SORL1	100%
SOX10	100%	SPART	100%	SPAST	100%	SPEN	100%
SPG11	100%	SPG21	100%	SPG7	100%	SPOP	100%
SPP1	100%	SPR	100%	SPRY4	100%	SPTAN1	100%
SPTBN1	100%	SPTBN2	100%	SPTBN4	100%	SPTLC1	100%
SPTLC2	100%	SQSTM1	100%	SRPX2	99.6%	ST3GAL3	100%
ST3GAL5	100%	STAMBP	100%	STARD7	100%	STAT4	100%
STRADA	100%	STUB1	100%	STX16	100%	STX1A	100%
STX1B	100%	STXBP1	100%	SUCLA2	100%	SUCLG1	100%
SUFU	100%	SUOX	100%	SUPT16H	100%	SURF1	100%
SV2A	100%	SYCP2	98.5%	SYNE1	100%	SYNGAP1	98.3%
SYNJ1	100%	SYT1	100%	SYT2	100%	SZT2	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
TAC4	100%	TACO1	100%	TACR1	100%	TACR3	100%
TADA2A	100%	TAF1	100%	TAF15	100%	TANGO2	100%
TARDBP	100%	TARS1	100%	TAT	100%	TBC1D24	100%
TBCD	100%	TBCE	100%	TBCK	100%	TBK1	100%
TBL2	100%	TBP	100%	TBX1	94.3%	TBX5	100%
TCEAL1	100%	TCIRG1	100%	TCTN1	98.8%	TCTN2	100%
TCTN3	100%	TDP1	100%	TDRKH	100%	TECPR2	100%
TECR	100%	TEFM	100%	TENM4	100%	TERT	100%
TET2	100%	TFG	100%	TGFBR2	100%	TGM5	100%
TGM6	100%	TH	100%	THAP1	100%	THG1L	100%
THOC2	100%	THPO	100%	TICAM1	100%	TIMM50	100%
TIMM8A	100%	TIMMDC1	100%	TK1	100%	TK2	100%
TLR3	100%	TLR4	100%	TLR7	100%	TM4SF20	100%
TMCO1	100%	TMEM106B	100%	TMEM127	100%	TMEM151A	100%
TMEM216	100%	TMEM218	100%	TMEM222	100%	TMEM231	100%
TMEM237	100%	TMEM240	100%	TMEM270	100%	TMEM63A	100%
TMEM67	100%	TMEM70	100%	TMPRSS3	100%	TNFAIP3	100%
TNFRSF11A	100%	TNFRSF1A	100%	TNFRSF1B	100%	TNFRSF25	100%
TNFSF11	100%	TNFSF4	100%	TNIP1	100%	TNNT1	100%
TNPO2	100%	TNR	100%	TNRC6A	100%	TOE1	100%
TOGARAM1	100%	TOMM40	100%	TOPORS	100%	TOR1A	100%
TP53	100%	TPI1	100%	TPK1	100%	TPM2	100%
TPM3	100%	TPP1	100%	TPR	100%	TRAF3	100%
TRAF7	100%	TRAK1	100%	TRAPPC11	100%	TRAPPC12	100%
TRAPPC2	89%	TRAPPC6B	100%	TREM2	100%	TREX1	100%
TRIM2	100%	TRIM32	100%	TRIM8	100%	TRIO	99.8%

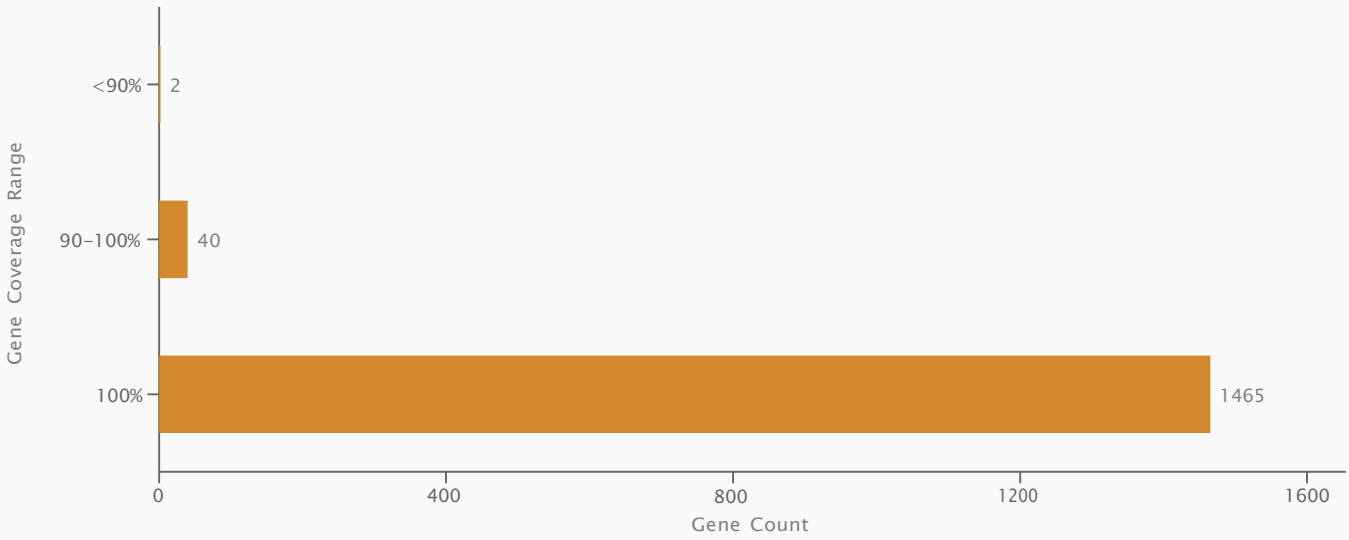
Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
TRMT5	100%	TRNP1	100%	TRPM3	100%	TRPM6	100%
TRPV4	100%	TSEN15	100%	TSEN2	100%	TSEN34	100%
TSEN54	100%	TSFM	100%	TSHR	100%	TSPOAP1	100%
TSPYL1	100%	TTC19	100%	TTI1	100%	TTPA	100%
TTR	100%	TUBA4A	100%	TUBB3	100%	TUBB4A	100%
TWNK	100%	TXN2	100%	TYMP	100%	TYROBP	100%
U2AF2	100%	UBA1	100%	UBA2	100%	UBA5	100%
UBAC2	100%	UBAP1	100%	UBAP2L	100%	UBE2L3	100%
UBE3A	100%	UBE3C	100%	UBQLN1	100%	UBQLN2	100%
UBR1	100%	UBTF	100%	UCHL1	100%	UFC1	100%
UFD1	100%	UGDH	100%	UGT1A1	100%	UGT1A10	100%
UGT1A4	100%	UGT1A6	100%	UGT1A7	100%	UGT1A8	100%
UNC13A	100%	UNC93B1	99.6%	UQCRC1	100%	UQCRQ	100%
UROC1	100%	UROD	100%	UROS	100%	USP53	100%
USP8	100%	VAC14	100%	VAMP1	100%	VAMP2	100%
VAPB	100%	VCP	100%	VDR	100%	VEGFA	100%
VHL	100%	VLDLR	100%	VPS13A	100%	VPS13C	100%
VPS13D	100%	VPS35	100%	VPS37A	100%	VPS37D	99.6%
VPS41	100%	VPS4A	100%	VPS53	100%	VRK1	100%
VSIG2	100%	VWA1	100%	VWA3B	100%	WARS1	100%
WARS2	100%	WASHC5	100%	WDR11	100%	WDR45	100%
WDR81	100%	WFS1	100%	WNK1	100%	WWOX	100%
XK	100%	XPA	100%	XPNPEP3	100%	XPR1	100%
XRCC1	100%	YARS1	100%	YARS2	100%	YEATS2	100%
YME1L1	100%	YRDC	100%	YWHAG	100%	YY1	100%
ZFHX2	100%	ZFR	100%	ZFYVE26	100%	ZFYVE27	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
ZNF142	100%	ZNF365	100%	ZNHIT3	100%		

Gene Coverage Distribution



QC Metrics

Total aligned reads	99.95 %
Total reads	61.08 (M)
Total data generated	9.08 (Gb)
Total reads which passed mapping quality cutt-off	8.68 (Gb)

Reviewed By

Dr. Neha Agrawal
Reviewed by
Clinical Geneticist

Mitochondrial Genes Mutation Next Generation Sequencing Panel

Clinical History Available:

Test requested for mitochondrial genome sequencing.

TEST RESULTS AND INTERPRETATION:

NO PATHOGENIC/LIKELY PATHOGENIC VARIANTS DETECTED IN ACCORDANCE TO THE MITOMAP DATABASE

RECOMMENDATIONS

- Please correlate clinically
- Please note that the genetic information obtained from the patient's genomic DNA was analyzed for regions of the mtDNA, and mutations in regions other than these regions have not been assessed.

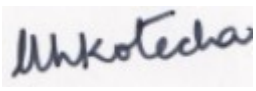
Technical Note:
Methodology: **Next Generation Sequencing**
Mutation Panel: Mitochondrial genes mutation panel
Analytic sensitivity: ~99%

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

IMPORTANT:

- These test results should be interpreted by the referring clinician only in conjunction with the patient's clinical history, other test results and any previous analysis of appropriate family members.
- 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
- 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.

For specimens received from non NCGM locations, it is presumed that it belongsto the patient as identified on the labels of the container/Test Requisition Formand it has been verified as per GCLP (Good Clinical Lab Practices) by the referrer atthe time of collection of the specimen. NCGM's responsibility is limited to theanalytical part of the assay performed.



Dr.Udhaya Kotecha
(M.D. Paeds, Fellow Med. Gen)

Limitations:

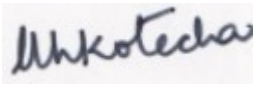
- This report is for research purposes only, not for use in clinical diagnostic or therapeutic applications.
- This test has not been validated by the FDA, NABL or CAP, and it has been determined by the accrediting bodies that such validation is not required at this time.
- For test performed on specimens received or collected from non-STMPL locations, it is presumed that the specimen belongs to the patient named or identified as labeled on the container/test request and such verification has been carried out at the point of generation of the said specimen by the sender.

Variant Analysis and Curation Performed by:

MehulMistri, PhD
Technical Analyst, Inherited Genomics

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

For specimens received from non NCGM locations, it is presumed that it belongsto the patient as identified on the labels of the container/Test Requisition Formand it has been verified as per GCLP (Good Clinical Lab Practices) by the referrer atthe time of collection of the specimen. NCGM's responsibility is limited to theanalytical part of the assay performed.



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