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Patient Details

Name	: Baby NEEHA	**Sex / Age**	: Female / 8 months	**Case ID**	: 50722804816
	:		:	**Test Name**	: ORION (WES-Whole Exome Sequencing)

Sample Details

Registration Date &	: 2025-07-26	**Sample Type**	: Whole Blood EDTA	**Sample Date & Time**	: 2025-07-26

Clinical History

Salient features: Failure to thrive, No weight gain, Skeletal dysplasia, Syndromic facies, Bronchopneumonia, Difficulty in breathing, Osteogenesis imperfecta

Test Results and Interpretation

HOMOZYGOUS PATHOGENIC VARIANT DETECTED: CLINICAL CORRELATION RECOMMENDED.

Summary Of Variants

Gene and Transcript	Exon/Intron Number	Variant Nomenclature	Zygosity	Classification	OMIM Phenotype	Inheritance
ALPL (NM_000478.6)	Exon 12	c.1348C>T p.Arg450Cys [133x /133x]	Homozygous	Pathogenic	Hypophosphatasia, infantile	Autosomal recessive

Variant Details

ALPL

Variant Nomenclature	c.1348C>T (p.Arg450Cys)
Genomic Nomenclature	chr1:g.21903914C>T
Zygosity	Homozygous

Variant Details

Type of variant	gnomAD frequency	Computational evidences	ClinVar	LOF disease mechanism of action	Downstream LOF	Previously reported [reported zygosity]	Variant references
Missense Variant	0.0002%	**REVEL:** 0.756 **CADD:** 24.2	Likely pathogenic/ Pathogenic (multiple submissions)	NA	NA	Yes [Homozygous/ Compound heterozygous]	1. Del Angel G, et al., 2020 2. Costain G, et al., 2017
This variant is also known as Arg433Cys. Experimental studies have shown that this missense change affects ALPL function (Nasu M, et al., 2006).							

References:

1. Del Angel G, et al. Large-scale in vitro functional testing and novel variant scoring via protein modeling provide insights into alkaline phosphatase activity in hypophosphatasia. *Hum Mutat*. 2020;41(7):1250-1262.
2. Costain G, et al. Enzyme replacement therapy in perinatal hypophosphatasia: Case report of a negative outcome and lessons for clinical practice. Mol Genet Metab Rep. 2017;14:22-26. Published 2017 Nov 7.
3. Nasu M, et al. Aberrant interchain disulfide bridge of tissue-nonspecific alkaline phosphatase with an Arg433-->Cys substitution associated with severe hypophosphatasia. *FEBS J*. 2006;273(24):5612-5624.

Disease

HYPOPHOSPHATASIA, INFANTILE [OMIM GENE ID: 171760]

Hypophosphatasia is characterized by defective mineralization of growing or remodeling bone, with or without root-intact tooth loss, in the presence of low activity of serum and bone alkaline phosphatase. Clinical features range from stillbirth without mineralized bone at the severe end to pathologic fractures of the lower extremities in later adulthood at the mild end. While the disease spectrum is a continuum, seven clinical forms of hypophosphatasia are usually recognized based on age at diagnosis and severity of features: Perinatal (severe): characterized by pulmonary insufficiency and hypercalcemia. Perinatal (benign): prenatal skeletal manifestations that slowly resolve into one of the milder forms. Infantile: onset between birth and age six months of clinical features of rickets without elevated serum alkaline phosphatase activity. Severe childhood (juvenile): variable presenting features progressing to rickets. Mild childhood: low bone mineral density for age, increased risk of fracture, and premature loss of primary teeth with intact roots. Adult: characterized by stress fractures and pseudofractures of the lower extremities in middle age, sometimes associated with early loss of adult dentition. Odontohypophosphatasia: characterized by premature exfoliation of primary teeth and/or severe dental caries without skeletal manifestations. **Variants in ALPL gene are also associated with autosomal recessive Hypophosphatasia, childhood; autosomal recessive and dominant Hypophosphatasia, adult and Odontohypophosphatasia.**

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: 15.9.5

Gene Coverage

Indication Based Analysis:

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
ABCC6	100%	ABCC9	100%	ABL1	100%	ACAN	100%
ACP5	100%	ACTA2	100%	ACVR1	100%	ADAMTS10	100%
ADAMTS17	100%	ADAMTS2	100%	ADAMTSL2	100%	ADAMTSL4	100%
ADCY10	100%	AEBP1	100%	AFF3	100%	AFF4	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
AGA	100%	AGPS	100%	AIFM1	100%	AIP	100%
AKT1	100%	ALB	100%	ALDH18A1	100%	ALG3	100%
ALPI	100%	ALPL	100%	ALPP	100%	ALX1	100%
ALX3	100%	ALX4	100%	AMER1	100%	ANAPC1	100%
ANAPC2	100%	ANKH	100%	ANKRD11	100%	ANO5	100%
ANTXR1	98.3%	ANTXR2	100%	AP2S1	100%	APC2	100%
ARCN1	100%	ARHGAP31	100%	ARID1B	97.9%	ARIH1	100%
ARMC5	100%	ARSB	100%	ARSL	100%	ASAH1	100%
ASCC1	100%	ASIC2	100%	ASPM	100%	ASXL1	100%
ASXL2	100%	ATP6V0A1	100%	ATP6V0A2	100%	ATP6V1A	100%
ATP6V1E1	100%	ATP7A	100%	ATP7B	100%	ATP8B1	100%
ATR	100%	ATRIP	100%	AURKAIP1	100%	AVP	100%
B3GALT6	91.3%	B3GAT3	100%	B4GALT7	100%	BANF1	100%
BAZ1B	100%	BCL7B	100%	BGN	100%	BHLHA9	100%
BMP1	100%	BMP15	100%	BMP2	100%	BMPER	100%
BMPR1B	100%	BNC1	100%	BPNT2	100%	BRAF	100%
BUD23	100%	BVES	100%	C1S	100%	C2CD3	100%
CA2	100%	CALCR	100%	CANT1	100%	CASR	100%
CAVIN1	100%	CBL	100%	CBS	100%	CC2D2A	100%
CCBE1	100%	CCDC8	100%	CCN6	100%	CCND1	100%
CCNQ	100%	CDC45	100%	CDC6	100%	CDC73	100%
CDH1	100%	CDH15	100%	CDH23	100%	CDH3	100%
CDK5RAP2	100%	CDKN1C	100%	CDT1	100%	CENPE	100%
CENPJ	100%	CEP120	100%	CEP135	100%	CEP152	100%
CEP290	100%	CEP63	100%	CEP97	100%	CFAP410	100%
CHD7	100%	CHRNG	100%	CHST14	100%	CHST3	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
CHSY1	99.1%	CHUK	100%	CILK1	100%	CKAP2L	100%
CLCN5	100%	CLCN7	100%	CLIP2	100%	CLPB	100%
CLSTN1	100%	COG1	100%	COG4	100%	COG7	100%
COL10A1	100%	COL11A1	100%	COL11A2	100%	COL12A1	100%
COL1A1	100%	COL1A2	99.6%	COL27A1	100%	COL2A1	100%
COL3A1	100%	COL4A1	100%	COL5A1	100%	COL5A2	100%
COL7A1	100%	COL9A1	100%	COL9A2	100%	COL9A3	100%
COLEC10	100%	COLEC11	100%	COMP	100%	COPB2	100%
COX4I2	100%	CPLX1	100%	CREB3L1	100%	CREBBP	100%
CRIPT	100%	CRTAP	100%	CSF1R	100%	CSGALNACT1	100%
CSPP1	100%	CTBP1	100%	CTC1	100%	CTCF	100%
CTDP1	100%	CTNS	100%	CTSA	100%	CTSK	100%
CTU2	100%	CUL7	100%	CWC27	100%	CYB5A	100%
CYP11A1	100%	CYP17A1	100%	CYP19A1	100%	CYP24A1	100%
CYP26B1	100%	CYP27A1	100%	CYP27B1	100%	CYP2R1	100%
CYP3A4	100%	DCAF17	100%	DCHS1	100%	DDOST	100%
DDR2	100%	DDRGK1	100%	DDX58	100%	DHCR24	100%
DHCR7	100%	DHODH	100%	DHX37	100%	DIP2C	100%
DKC1	100%	DKK1	100%	DLL1	100%	DLL3	100%
DLL4	100%	DLX3	100%	DLX5	100%	DLX6	100%
DMP1	100%	DMRT2	100%	DMRT3	100%	DMTF1	100%
DNA2	100%	DNAJC21	100%	DNAJC30	100%	DNMT3A	100%
DOCK6	100%	DONSON	100%	DPAGT1	100%	DPF2	100%
DPM2	100%	DSE	100%	DSPP	100%	DUSP6	100%
DVL1	100%	DVL3	100%	DXO	100%	DYM	100%
DYNC2H1	100%	DYNC2I1	100%	DYNC2I2	100%	DYNC2LI1	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
DYNLT2B	100%	EBP	100%	EDN1	100%	EDNRA	100%
EED	100%	EFEMP2	100%	EFL1	100%	EFNA1	100%
EFNB1	100%	EFTUD2	100%	EGFL7	100%	EHHADH	100%
EIF2AK3	100%	EIF4A3	100%	EIF4H	100%	ELANE	100%
ELMO2	100%	ELN	100%	ENPP1	99.5%	EOGT	100%
EP300	100%	ERCC2	100%	ERCC3	100%	ERCC4	100%
ERCC6	100%	ERF	100%	ESCO2	100%	ESR1	100%
ESR2	100%	ETF1	100%	EVC	100%	EVC2	100%
EXOC6B	100%	EXOSC2	100%	EXT1	100%	EXT2	100%
EXTL3	100%	EZH2	100%	FAH	100%	FAM111A	100%
FAM20B	100%	FAM20C	100%	FANCA	100%	FANCB	100%
FANCC	100%	FANCD2	100%	FANCE	100%	FANCG	100%
FANCI	100%	FANCL	100%	FAR1	100%	FARSB	100%
FAT4	100%	FBLN1	100%	FBLN5	100%	FBN1	100%
FBN2	100%	FERMT3	100%	FGD1	100%	FGF10	100%
FGF16	100%	FGF17	100%	FGF23	100%	FGF8	100%
FGF9	100%	FGFR1	100%	FGFR2	100%	FGFR3	100%
FIG4	100%	FKBP10	100%	FKBP14	100%	FKBP6	100%
FLCN	100%	FLNA	100%	FLNB	100%	FLRT3	100%
FMN1	100%	FN1	100%	FOXA2	100%	FOXE3	100%
FREM1	100%	FSHR	100%	FTO	100%	FUCA1	100%
FUT8	100%	FUZ	100%	FZD2	100%	FZR1	100%
GALNS	100%	GALNT3	100%	GALT	100%	GATA1	100%
GATA4	100%	GATM	100%	GBA	100%	GCK	100%
GCM2	100%	GDF3	100%	GDF5	100%	GDF6	100%
GEMIN4	100%	GFI1	100%	GGCX	100%	GHR	100%

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Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
GHRHR	100%	GHSR	100%	GJA1	100%	GK	100%
GLB1	100%	GLI2	100%	GLI3	100%	GLIS3	100%
GMNN	100%	GNA11	100%	GNAI3	100%	GNAS	100%
GNE	100%	GNPAT	100%	GNPTAB	100%	GNPTG	100%
GNRH1	100%	GNRHR	100%	GNS	100%	GORAB	100%
GPAA1	100%	GPC3	100%	GPC4	100%	GPC6	100%
GPR35	100%	GPX4	100%	GREM1	100%	GSC	100%
GTF2E2	100%	GTF2H5	100%	GTF2I	100%	GTF2IRD1	100%
GTF2IRD2	100%	GTF2IRD2B	100%	GUSB	100%	GZF1	100%
HAAO	100%	HAMP	100%	HBB	100%	HCN4	100%
HDAC4	100%	HDAC8	100%	HECW2	100%	HERC2	100%
HES7	100%	HESX1	100%	HFE	100%	HGSNAT	100%
HJV	100%	HNRNPK	100%	HOXA11	100%	HOXA13	89.8%
HOXD13	100%	HPGD	100%	HRAS	100%	HS6ST1	100%
HSD17B4	100%	HSD3B7	100%	HSPA9	100%	HSPG2	99.5%
HTRA1	100%	HYAL1	100%	IARS2	100%	ID4	100%
IDH1	100%	IDH2	100%	IDS	100%	IDUA	100%
IER3IP1	100%	IFIH1	100%	IFITM5	100%	IFNAR1	100%
IFT122	100%	IFT140	100%	IFT172	100%	IFT43	100%
IFT52	100%	IFT57	100%	IFT74	100%	IFT80	100%
IFT81	100%	IGF1	100%	IGF1R	100%	IGF2	100%
IHH	100%	IKBKG	100%	IL11RA	100%	IL12A	100%
IL12RB1	100%	IL17RD	100%	IL1RN	100%	IL6ST	100%
INPPL1	100%	INTU	100%	IRF5	100%	IRF6	100%
IRX5	100%	JAG1	100%	KAT6B	100%	KCNJ1	100%
KCNJ2	100%	KCNJ8	100%	KDELR2	100%	KDM1A	100%

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Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
KIAA0586	100%	KIAA0753	100%	KIF22	100%	KIF5B	100%
KIF7	100%	KISS1	100%	KISS1R	100%	KIT	100%
KL	98.7%	KMT2A	100%	KNSTRN	100%	KRAS	100%
KYNU	100%	LAMA3	100%	LAMA4	100%	LAMA5	100%
LAMB3	100%	LAMC2	100%	LARP7	100%	LARS2	100%
LBR	100%	LEMD3	100%	LETM1	100%	LFNG	95.6%
LHX4	100%	LIFR	100%	LIG4	100%	LIMK1	100%
LMBR1	100%	LMNA	100%	LMX1B	100%	LONP1	100%
LOX	100%	LOXL3	100%	LPIN2	100%	LRP4	100%
LRP5	100%	LRP6	100%	LRRK1	100%	LTBP2	100%
LTBP3	100%	LTBP4	100%	LZTS1	100%	MAB21L2	100%
MAFB	100%	MAGEL2	100%	MALT1	100%	MAN2B1	100%
MANBA	100%	MAP2K1	100%	MAP3K1	100%	MAP3K20	100%
MAP3K7	100%	MAP4K2	100%	MASP1	100%	MAT2A	100%
MATN3	100%	MBTPS1	100%	MBTPS2	100%	MCM3	100%
MCM5	100%	MCM7	100%	MCPH1	100%	MDM4	100%
MECOM	100%	MED12	100%	MEGF8	100%	MEN1	100%
MEOX1	100%	MEPE	100%	MESD	100%	MESP2	100%
MET	100%	METTL27	100%	MFAP5	100%	MGAT2	100%
MGP	100%	MIA3	100%	MITF	100%	MKRN3	100%
MKS1	100%	MLXIPL	100%	MMAB	100%	MMEL1	100%
MMP1	100%	MMP13	100%	MMP14	100%	MMP2	100%
MMP9	100%	MNX1	83.5%	MOGAT2	100%	MOGS	100%
MPLKIP	100%	MRPS22	100%	MST1	100%	MSX2	100%
MTAP	100%	MTCH1	100%	MTRR	100%	MTTP	100%
MTX2	100%	MYCN	100%	MYH11	100%	MYH3	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
MYLK	100%	MYO18B	100%	NAA20	100%	NAGA	100%
NAGLU	100%	NANS	100%	NBAS	100%	NCF1	100%
NDN	100%	NDUFAF1	100%	NDUFAF6	100%	NEK1	100%
NELFA	100%	NEU1	100%	NF1	100%	NFIX	100%
NGLY1	100%	NHP2	100%	NIN	100%	NIPBL	100%
NKX3-1	100%	NKX3-2	100%	NLRP3	100%	NOG	100%
NOP10	100%	NOTCH1	100%	NOTCH2	100%	NPAP1	100%
NPM1	100%	NPPC	100%	NPR2	100%	NPR3	100%
NPRL2	100%	NPRL3	100%	NR0B1	100%	NR5A1	100%
NRAS	100%	NSD1	100%	NSD2	99.9%	NSDHL	100%
NSMCE2	100%	NSMF	100%	NTRK1	100%	NUP107	100%
NXN	100%	OBSL1	100%	OCA2	100%	OCRL	100%
OFD1	100%	ORC1	100%	ORC4	100%	ORC6	100%
OSTM1	100%	OTX2	100%	P3H1	100%	P4HB	100%
PACS1	100%	PALB2	100%	PAM16	100%	PAPPA2	100%
PAPSS2	100%	PARN	100%	PAX3	100%	PCCA	100%
PCCB	100%	PCGF2	100%	PCNT	100%	PCYT1A	100%
PDE11A	100%	PDE3A	100%	PDE4D	100%	PDE8B	100%
PDGFRB	100%	PDLIM4	100%	PEX12	100%	PEX5	100%
PEX7	100%	PGM3	100%	PHEX	100%	PHF6	100%
PHIP	100%	PHKA2	100%	PHKB	100%	PHKG2	100%
PIEZO2	100%	PIGG	100%	PIGT	100%	PIGU	100%
PIGV	100%	PIGY	100%	PIK3C2A	100%	PIK3CA	100%
PIK3CD	100%	PISD	100%	PITX1	100%	PKD2	100%
PKDCC	100%	PLCB3	100%	PLCB4	100%	PLEKHM1	100%
PLK4	100%	PLOD1	100%	PLOD2	100%	PLOD3	100%

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Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
PLS3	100%	PMM2	100%	POC1A	100%	POF1B	100%
POLD1	100%	POLE	100%	POLG	100%	POLG2	100%
POLR1A	100%	POLR1C	100%	POLR1D	100%	POLR3A	100%
POLR3B	100%	POLR3GL	100%	POLR3H	100%	POP1	100%
POU1F1	100%	POU2AF1	100%	PPIB	100%	PPP3CA	100%
PRDM5	100%	PRKACA	100%	PRKAR1A	100%	PRKCA	100%
PRKD2	100%	PRKG1	100%	PRLR	100%	PRMT7	100%
PROK2	100%	PROKR2	100%	PROP1	100%	PSAP	100%
PSMC3IP	100%	PTDSS1	100%	PTH1R	100%	PTHLH	100%
PTPN11	100%	PYCR1	100%	PYDC1	100%	PYGL	100%
RAB23	100%	RAB33B	100%	RAB3GAP1	100%	RAD21	100%
RAI1	100%	RBBP8	100%	RBM8A	100%	RBPJ	100%
RECQL4	100%	RFC2	100%	RIN2	100%	RIPK4	100%
RIPPLY2	100%	RNASE12	100%	RNF113A	100%	RNF125	100%
RNMT	100%	ROR2	100%	RORA	100%	RPGRIP1L	100%
RPL10	100%	RPL11	100%	RPL15	100%	RRM2B	100%
RSPO2	100%	RSPRY1	100%	RTEL1	100%	RTTN	100%
RUNX1	100%	RUNX2	100%	SALL1	100%	SALL4	100%
SATB2	100%	SBDS	100%	SC5D	100%	SCARB2	100%
SCARF2	100%	SEC23A	100%	SEC24D	100%	SEMA4D	100%
SERPINF1	100%	SERPINH1	100%	SETBP1	100%	SETD2	100%
SF3B4	100%	SFI1	100%	SFRP4	100%	SFTPA1	100%
SFTPA2	100%	SGMS2	100%	SGSH	100%	SH3BP2	100%
SH3D19	100%	SH3PXD2B	100%	SHH	100%	SHOX	100%
SIK3	100%	SIM1	100%	SKI	100%	SLC10A1	100%
SLC10A7	100%	SLC12A1	100%	SLC17A5	100%	SLC25A15	100%

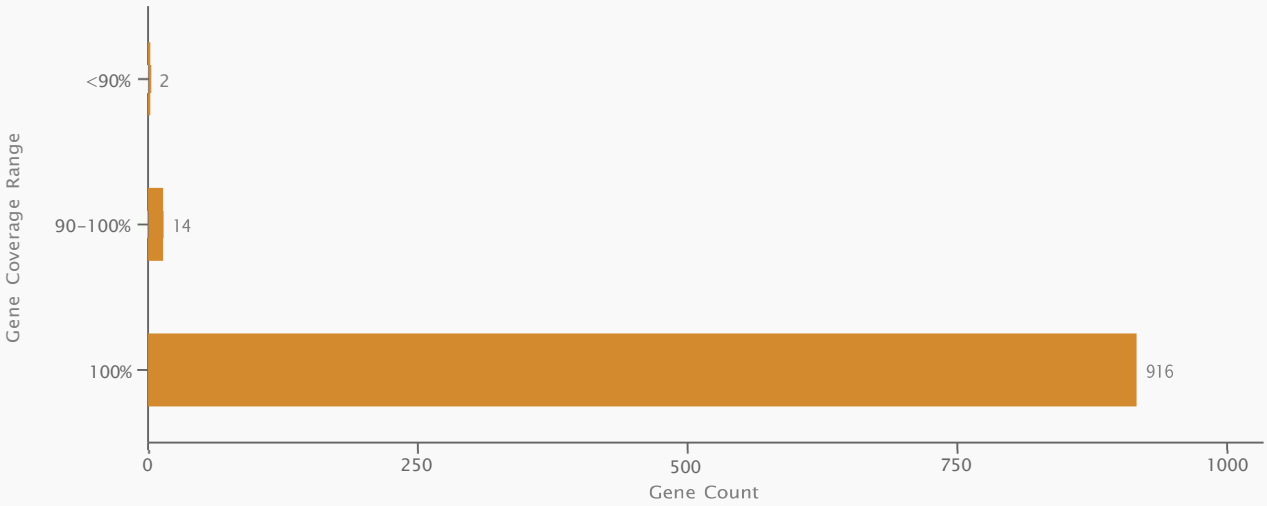
Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
SLC25A19	100%	SLC25A23	100%	SLC25A4	100%	SLC26A2	100%
SLC29A3	100%	SLC2A10	100%	SLC2A2	100%	SLC34A1	100%
SLC34A3	100%	SLC35A2	100%	SLC35D1	100%	SLC37A4	99.9%
SLC39A13	100%	SLC39A8	100%	SLC4A1	100%	SLC5A6	100%
SLC7A7	100%	SLC9A3R1	100%	SLCO2A1	100%	SLCO5A1	100%
SLX4	100%	SMAD2	100%	SMAD3	100%	SMAD4	100%
SMAD6	100%	SMARCA4	100%	SMARCAL1	100%	SMARCB1	100%
SMARCD2	100%	SMARCE1	100%	SMC1A	100%	SMC3	100%
SMIM6	100%	SMOC1	100%	SMPD1	100%	SMS	100%
SNRPB	100%	SNRPN	100%	SNX10	100%	SOST	100%
SOX3	100%	SOX9	100%	SP7	100%	SPAM1	100%
SPARC	100%	SPECC1L	100%	SPIB	100%	SPIDR	100%
SPRY4	100%	SQSTM1	100%	SRC	100%	SRCAP	100%
SRP54	100%	SRSF2	100%	STAG2	100%	STAT1	100%
STAT3	100%	STK4	100%	STN1	100%	STON1	100%
STX1A	100%	STX3	100%	SUCO	100%	SULF1	100%
SUMF1	100%	SUZ12	100%	SYK	100%	TAB2	100%
TAC3	100%	TACR3	100%	TAF1	100%	TALDO1	100%
TAPT1	100%	TARS1	100%	TBCD	100%	TBCE	100%
TBCK	100%	TBL2	100%	TBX15	100%	TBX3	100%
TBX4	100%	TBX5	100%	TBX6	100%	TBXAS1	100%
TCF12	100%	TCF4	100%	TCF7L2	100%	TCIRG1	100%
TCOF1	100%	TCTN3	100%	TENT5A	100%	TENT5B	100%
TERT	100%	TET2	100%	TFAP2A	100%	TGDS	100%
TGFB1	100%	TGFB2	100%	TGFB3	100%	TGFBR1	100%
TGFBR2	100%	THPO	100%	TINF2	100%	TMEM165	100%

Gene Coverage

Gene	Coverage	Gene	Coverage	Gene	Coverage	Gene	Coverage
TMEM216	100%	TMEM270	100%	TMEM38B	100%	TMEM53	100%
TMEM67	100%	TNFRSF11A	99.9%	TNFRSF11B	100%	TNFSF11	100%
TNFSF15	100%	TNPO3	100%	TNXB	100%	TOM1	100%
TONSL	100%	TP53INP2	100%	TP63	100%	TRAF3IP1	100%
TRAF6	100%	TRAIP	100%	TRAPPC2	100%	TREM2	100%
TRIM37	100%	TRIP11	100%	TRIP4	100%	TRMT10A	100%
TRNP1	100%	TRPS1	100%	TRPV4	100%	TRPV6	100%
TTC21B	100%	TTC26	100%	TUBGCP4	100%	TUBGCP6	100%
TWIST1	100%	TWNK	100%	TXNL4A	100%	TYROBP	100%
UBA1	100%	UBE3B	100%	UFSP2	100%	UNC80	100%
UPF3B	100%	UROD	100%	UROS	100%	USB1	100%
USP8	100%	USP9X	100%	VAC14	100%	VAMP7	100%
VCP	100%	VDAC2	100%	VDR	100%	VPS33A	100%
VPS37D	100%	VPS53	100%	WDR11	100%	WDR19	100%
WDR35	100%	WDR4	100%	WDR72	100%	WNT1	100%
WNT10B	100%	WNT3	100%	WNT3A	100%	WNT5A	100%
WNT6	100%	WNT7A	100%	WRAP53	100%	WRN	100%
WT1	100%	WWOX	100%	XRCC4	100%	XYLT1	100%
XYLT2	100%	YY1AP1	100%	ZBTB20	100%	ZFPM2	100%
ZIC1	100%	ZMPSTE24	100%	ZNF462	100%	ZNF469	100%
ZNF687	100%	ZNF699	100%	ZSWIM6	96%	ZSWIM7	100%

Gene Coverage Distribution



QC Metrics

Total aligned reads	99.95 %
Total reads	78.88 (M)
Total data generated	11.63 (Gb)
Total reads which passed mapping quality cutt-off	11.21 (Gb)

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