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Whole Exome Sequencing Analysis – Carrier Screening

Patient name	: Mr. XXX	Mrs. YYY
Gender/ Age	: Male/ 32 years	Female/ 26 years
PIN	: AND24380038709	AND24380038710
Sample no	: 382431351	382431352
Specimen	: Peripheral blood	Peripheral blood
Sample collection date	: 27-05-2024	27-05-2024
Sample receipt date	: 27-05-2024	27-05-2024
Report date	: 22-06-2024	22-06-2024
Hospital/Clinic	: ARC International Fertility And Research Centre - Chennai	

Clinical history

Mr. XXX and Mrs. YYY are non-consanguineous couple and they presented with hearing loss and mutism. Mr. XXX has a history of fever at 6 months of age and then was diagnosed with bilateral hearing loss. Mrs. YYY was born to third degree consanguineous parents and was diagnosed with congenital bilateral hearing loss. She has a history of Hepatitis B at 2 years of age, hypothyroidism and PCOD. Mr. XXX and Mrs. YYY have been evaluated for carrier status of pathogenic variations.

Results

Mr. XXX is found to be affected with likely pathogenic variant in the *GJB2* gene (p.Trp24Ter).

Mrs. YYY is found to be affected with likely pathogenic variant in the *MYO3A* gene (p.Arg1495Ter).

Mr. XXX is found to be carrier of likely pathogenic variants in the *NDUFA13* gene (p.Tyr16LeufsTer41), *OCA2* gene (c.2079+1G>A) and *PLAA* gene (c.1039+2T>G)

Mrs. YYY is found to be carrier of uncertain significant variant in *PLAA* gene (p.Asn333Ser).

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List of significant variants identified related to the phenotype:

Disease	Mr. XXX	Mrs. YYY
Deafness, autosomal recessive 1A (OMIM#220290) Mode of inheritance: AR Reproductive risk: 1 in 59,277	**AFFECTED** Gene: GJB2 Exon 2, c.71G>A p.Trp24Ter Homozygous Classification: **Likely Pathogenic**	**NON - CARRIER**
Deafness, autosomal dominant 90 (OMIM#620722) Mode of inheritance: AD Deafness, autosomal recessive 30 (OMIM#607101) Mode of inheritance: AR	**NON - CARRIER**	**AFFECTED** Gene: MYO3A Exon 32, c.4483C>T p.Arg1495Ter Heterozygous Classification: **Likely Pathogenic**

List of carrier variants identified:

Disease	Mr. XXX	Mrs. YYY
?Mitochondrial complex I deficiency, nuclear type 28 (OMIM#618249) Mode of inheritance: AR	**CARRIER** Gene: NDUFA13 Exon 1, c.44dupG p.Tyr16LeufsTer41 Heterozygous Classification: **Likely Pathogenic**	**NON - CARRIER**
Albinism, brown oculocutaneous / Albinism, oculocutaneous, type II (OMIM#203200) Mode of inheritance: AR Reproductive risk : 1 in 12,008	**CARRIER** Gene: OCA2 Intron 19, c.2079+1G>A 5' Splice site Heterozygous Classification: **Likely Pathogenic**	**NON - CARRIER**
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies (OMIM#617527) Mode of inheritance: AR	**CARRIER** Gene: PLAA Intron 7, c.1039+2T>G 5' Splice site Heterozygous Classification: **Likely Pathogenic**	**CARRIER** Gene: PLAA Exon 7, c.998A>G p.Asn333Ser Heterozygous Classification: **Uncertain significance**

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*Genetic test results are reported based on the recommendation of American College of Medical Genetics [1]. No other variant that warrants to be reported for the given clinical indication was identified.

Variant Interpretation

Interpretation for the significant variant related to the phenotype identified in Mr. XXX

GJB2: c.71G>A

Variant summary: A homozygous stop gained variation in exon 2 of the *GJB2* gene (chr13:g.20189511C>T, NM_004004.6, Depth: 116x) that results in the premature truncation of the protein at codon 24 (p.Trp24Ter) was detected. This variant is a stop gained variant which occurs in an exon of *GJB2* upstream where nonsense mediated decay is predicted to occur. This variant is predicted to cause loss of normal protein function through protein truncation.

Population frequency: This variant has minor allele frequency of 0.0138% in gnomAD database and has minor allele frequency of 0.0399% in 1000 genomes databases.

Clinical evidence: This variant has been previously classified as pathogenic in ClinVar database [3]. This variant has been previously reported in Indian patients affected with nonsyndromic hearing loss in both homozygous and compound heterozygous state [4].

In-silico prediction: The *in-silico* predictions of the variant are deleterious by CADD. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Deafness, autosomal recessive 1A (OMIM#220290) is caused by homozygous or compound heterozygous mutation in the *GJB2* gene (OMIM*121011). This disease follows autosomal recessive and Digenic dominant pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a likely pathogenic variant. **In this view, clinical correlation and familial segregation analysis are strongly recommended to establish the significance of the finding. If the results do not correlate, additional testing may be considered based on the phenotype observed.**

Interpretation for the significant variant related to the phenotype identified in Mrs. YYY

MYO3A: c.4483C>T

Variant summary: A heterozygous stop gained variation in exon 32 of the *MYO3A* gene (chr10:g.26193249C>T, NM_017433.5, Depth:118x) that results in the premature truncation of the protein at codon 1495 (p.Arg1495Ter) was detected. This variant is a stop gained variant which occurs in an exon of *MYO3A* upstream where nonsense mediated decay is predicted to occur. This variant is predicted to cause loss of normal protein function through protein truncation.

Population frequency: This variant has minor allele frequency of 0.0053% in gnomAD database and has minor allele frequency of 0.0399% in 1000 genomes databases.

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Clinical evidence: This variant has been previously classified as pathogenic in ClinVar database [5].

In-silico prediction: The in-silico predictions of the variant are deleterious by CADD.

OMIM phenotype: Deafness, autosomal dominant 90 (OMIM#620722) is caused by heterozygous mutation in the MYO3A gene (OMIM*606808). This disease follows autosomal dominant pattern of inheritance [2]. Deafness, autosomal recessive 30 (OMIM#607101) is caused by homozygous or compound heterozygous mutation in the MYO3A gene (OMIM#606808). This disease follows autosomal recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a likely pathogenic variant. **In this view, clinical correlation and familial segregation analysis are strongly recommended to establish the significance of the finding. If the results do not correlate, additional testing may be considered based on the phenotype observed.**

Interpretation for the significant carrier variants identified in Mr. XXX

NDUFA13: c.44dupG

Variant summary: A heterozygous single base pair insertion in exon 1 of the NDUFA13 gene (chr19:g.19516277insG, NM_015965.7, Depth:133x) that results in a frameshift and premature truncation of the protein 41 amino acids downstream to codon 16 (p.Tyr16LeufsTer41) was detected. This variant is a frameshift variant which occurs in an exon of NDUFA13 upstream where nonsense mediated decay is predicted to occur. This variant is predicted to cause loss of normal protein function through protein truncation.

Population frequency: This variant has not been reported in gnomAD database and 1000 genomes database.

Clinical evidence: This variant has been previously classified as likely pathogenic in ClinVar database [6].

In-silico prediction: The in-silico predictions of the variant are deleterious by MutationTaster2. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: ?Mitochondrial complex I deficiency, nuclear type 28 (OMIM#618249) is caused by homozygous mutation in the NDUFA13 gene (OMIM*609435). This disease follows autosomal recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a likely pathogenic variant.

OCA2: c.2079+1G>A

Variant summary: A heterozygous 5' splice site variation in intron 19 of the OCA2 gene (chr15:g.27926126C>T, NM_000275.3, Depth: 53x) that affects the invariant GT donor splice site of exon 19 (c.2079+1G>A) was detected.
Population frequency: This variant has not been reported in gnomAD database and 1000 genomes database.

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Clinical evidence: This variant has been previously classified as likely pathogenic in ClinVar database [7]. This variant has been previously reported in patient affected with Oculocutaneous albinism in compound heterozygous state [8].

In-silico prediction: The *in-silico* predictions of the variant are deleterious by CADD and disrupting by GeneSplicer, MaxEntScan and NNSplice. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Albinism, brown oculocutaneous / Albinism, oculocutaneous, type II (OMIM#203200) are caused by homozygous or compound heterozygous mutation in the *OCA2* gene (OMIM*611409). These diseases follow autosomal recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a likely pathogenic variant.

PLAA: c.1039+2T>G

Variant summary: A heterozygous 5' splice site variation in intron 7 of the *PLAA* gene (chr9:g.26923176A>C, NM_001031689.3, Depth: 100x) that affects the invariant GT donor splice site of exon 7 (c.1039+2T>G) was detected.

Population frequency: This variant has not been reported in gnomAD database and 1000 genomes database.

In-silico prediction: The *in-silico* predictions of the variant are deleterious by CADD and disrupting by GeneSplicer, MaxEntScan and NNSplice. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies (OMIM#617527) is caused by homozygous or compound heterozygous mutation in the *PLAA* gene (OMIM*603873). This disease follows autosomal recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a likely pathogenic variant.

Interpretation for the uncertain significant carrier variant identified in Mrs. YYY

PLAA: c.998A>G

Variant summary: A heterozygous missense variation in exon 4 of the *PLAA* gene (chr9:g.26923219T>C, NM_001031689.3; depth: 107x) that results in the amino acid substitution of Serine for Asparagine at codon 333 (p.Asn333Ser) was detected.

Population frequency: This variant has minor allele frequency of 0.0118% in gnomAD database and has not been reported in 1000 genomes databases.

Clinical evidence: This variant has been previously classified as uncertain significance in ClinVar database [9].

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In-silico prediction: The *in-silico* predictions of the variant are damaging by SIFT and LRT. The reference codon is conserved across mammals in PhyloP tool.

OMIM phenotype: Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies (OMIM#617527) is caused by homozygous or compound heterozygous mutation in the *PLAA* gene (OMIM*603873). This disease follows autosomal recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as a variant of uncertain significance.

Recommendations

- Sequencing the variant(s) in the parents and the other affected and unaffected members of the family is recommended to confirm the significance.
- Alternative test is strongly recommended to rule out the deletion/duplication.
- Genetic counseling is recommended.

Methodology

DNA extracted from the blood was used to perform whole exome using a whole exome capture kit. The targeted libraries were sequenced to a targeted depth of 80 to 100X using GenoLab M sequencing platform. This kit has deep exonic coverage of all the coding regions including the difficult to cover regions. The sequences obtained are aligned to human reference genome (GRCh38.p13) using Sentieon aligner and analyzed using Sentieon for removing duplicates, recalibration and re-alignment of indels. Sentieon DNAscope has been used to call the variants. Detected variants were annotated and filtered using the VarSeq software with the workflow implementing the ACMG guidelines for variant classification. The variants were annotated using 1000 genomes(V2), gnomAD (v3.1.2,2.1.1), ClinVar, OMIM, dbSNP, NCBI RefSeq Genes. *In-silico* predictions of the variant was carried out using VS-SIFT, VS-PolyPhen2, PhyloP, GERP++, GeneSplicer, MaxEntScan, NNSplice, PWM Splice Predictor. Only non-synonymous and splice site variants found in the coding regions were used for clinical interpretation. Silent variations that do not result in any change in amino acid in the coding region are not reported.

Sequence data attributes

	Mr. XXX	**Mrs. YYY**
Total reads generated	9.56 Gb	8.62 Gb
Data ≥ Q30	95.74%	97.19%

Genetic test results are reported based on the recommendations of American College of Medical Genetics [1], as described below:

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Classification	Interpretation
Pathogenic	A disease-causing variation in a gene which can explain the patients' symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed
Likely Pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.
Variant of Uncertain Significance	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non- disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

Disclaimer

- The classification of variants of unknown significance can change over time. Anderson Diagnostics and Labs cannot be held responsible for it.
- Intronic variants, UTR, Promoter region variants and CNV are not assessed using this assay.
- Certain genes may not be covered completely, and few mutations could be missed. Variants not detected by this assay may impact the phenotype.
- The variations have not been validated by Sanger sequencing.
- The above findings and result interpretation was done based on the clinical indication provided at the time of reporting.
- It is also possible that a pathogenic variant is present in a gene that was not selected for analysis and/or interpretation in cases where insufficient phenotypic information is available.
- Genes with pseudogenes, paralog genes and genes with low complexity may have decreased sensitivity and specificity of variant detection and interpretation due to inability of the data and analysis tools to unambiguously determine the origin of the sequence data in such regions.
- The variants of uncertain significance and variations with high minor allele frequencies which are likely to be benign will be given upon request.
- Incidental or secondary findings that meet the ACMG guidelines can be given upon request [10].

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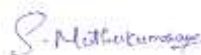www.andersondiagnostics.com | Home Collection : 91766 77601

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APPENDIX: Current gene list used for carrier screening of pathogenic and likely pathogenic variants:

AAAS,AARS2,ABAT,ABCA1,ABCA12,ABCA3,ABCA4,ABCB4,ABCC8,ABCD1,ABCD4,ABCG5,ABCG8,ACAD9,ACADS,ACADSB,ACADVL,ACAT1,ACD,ACKR1, ACO2,ACOX1,ACOX2,ACP2,ACP5,ACTA1,ADAMTS10,ADAMTSL2,ADAMTSL4,ADAR,ADAT3,ADCY6,ADD3,ADGRG6,ADGRV1,ADRB2,ADRB3,ADSL,AEBP1,AGA,AGBL5,AGPAT2,AGRP,AGT,AGTR1,AGXT,AGXT2,AHCY,AHI1,AHSG,AICDA,AIFM1,AIPL1,AIRE,AK1,AKR1C4,AKR1D1,ALAD,ALAS2,ALDH18A1, ALDH6A1,ALDOA,ALDOB,ALG1,ALG12,ALG2,ALG3,ALG9,ALMS1,ALOX12B,ALOXE3,ALPL,ALS2,ALX3,ALX4,AMACR,AMBN,AMHR2,AMMECR1,AMN,AMPD1,AMPD2,AMPD3,AMT,ANGPTL3,ANO10,ANO5,ANTXR1,ANTXR2,AP4B1,AP4E1,AP5Z1,APOC2,APOE,APRT,AQP2,AQP7,ARFGEF2,ARHGDIA,ARHGEF6,ARL2BP,ARL6,ARL6IP1,ASL,ASNS,ASPA,ASPM,ASS1,ATCAY,ATG5,ATIC,ATM,ATP2A1,ATP2B3,ATP6AP2,ATP6V0A2,ATP6V1A,ATP6V1B1,ATP7A,ATP8A2,ATPAF2,ATR,AURKC,AVPR2,B2M,B3GAT3,B4GALNT1,B4GALT1,B4GAT1,B9D1,B9D2,BANF1,BBIP1,BBS1,BBS10,BBS12,BBS2,BBS4,BBS5,BBS7,BCAP31,BCL10,BCS1L,BGN,BIN1,BLNK,BLOC1S3,BLVRA,BMP1,BMP2,BMPR1B,BPGM,BRAT1,BRF1,BSCL2,BSND,BTK,BVES,C12orf57,C12orf65,C19orf12,C1QA,C1QB,C1QC,C3,C8A,C8B,C8orf37,CA12,CA2,CA5A,CA8,CABP2,CABP4,CACNA1D,CACNA1F,CACNA2D4,CAD,CANT1,CAPN1,CARD11,CARD9,CARTPT,CASP14,CASQ2,CASR,CATSPER1,CAV3,CBX2,CCBE1,CCDC103,CCDC114,CCDC115,CCDC22,CCDC28B,CCDC65,CCDC8,CCDC88C,CCNO,CCT5,CD19,CD247,CD27,CD36,CD3D,CD3E,CD3G,CD40,CD40LG,CD46,CD79A,CD79B,CD8A,CDAN1,CDC6,CDH11,CDH3,CDHR1,CDK10,CDK5,CDK5RAP2,CDK6,CDSN,CEBPE,CENPF,CENPJ,CEP104,CEP152,CEP164,CEP19,CEP41,CEP57,CERS3,CFH,CFL2,CFP,CFTR,CHAT,CHIT1,CHKB,CHMP1A,CHRDL1,CHRM3,CHRNA1,CHRND,CHRNE,CHRNG,CHST14,CHST3,CHST6,CHST8,CIB2,CIDEC,CIITA,CIT,CKAP2L,CLCN1,CLCN4,CLCNKA,CLCNKB,CLDN1,CLDN14,CLDN16,CLDN19,CLIC2,CLMP,CLN8,CLP1,CLPB,CLPP,CLRN1,CNGA3,CNGB1,CNGB3,CNNM2,CNPY3,CNTN1,CNTN2,CNTNAP1,CNTNAP2,COASY,COG4,COG6,COL17A1,COL4A3,COL4A4,COL6A1,COL6A2,COL6A3,COL7A1,COL9A2,COLEC11,COLQ,COQ4,COQ6,COQ7,COQ9,COX10,COX14,COX20,COX4I2,COX6A1,COX6B1,COX8A,CPA6,CPLX1,CPN1,CPT1A,CPT2,CRADD,CRAT,CRB1,CRIPT,CRTAP,CRYAB,CRYBB1,CRYBB3,CSF2RB,CSF3R,CSTA,CSTB,CTC1,CTH,CTNNB1,CTNS,CTPS1,CTSA,CTSC,CTSD,CTSF,CTSK,CUBN,CUL7,CWF19L1,CYB5A,CYBA,CYBB,CYP11B1,CYP11B2,CYP17A1,CYP1B1,CYP24A1,CYP26C1,CYP27A1,CYP27B1,CYP2C19,CYP2D6,CYP2R1,CYP2U1,CYP4F22,CYP4V2,CYP7B1,D2HGDH,DAG1,DARS2,DBH,DBT,DCC,DCHS1,DCPS,DCTN1,DCXR,DDB2,DDC,DDHD1,DDHD2,DDOST,DDR2,DDRGK1,DDX11,DDX59,DES,DGKE,DGUOK,DHCR7,DHH,DHODH,DHTKD1,DIS3L2,DKC1,DLAT,DLEC1,DLG3,DLX5,DMD,DNA2,DNAAF1,DNAAF3,DNAH1,DNAH11,DNAI1,DNAJB13,DNAJB2,DNAJC19,DNAJC3,DNAJC6,DNAL4,DNASE1L3,DNM2,DNMT3B,DOCK2,DOK7,DOLK,DPAGT1,DPH1,DPM2,DPYD,DPYS,DRAM2,DRC1,DSC3,DSE,DSG1,DSG4,DSP,DST,DSTYK,DTNBP1,DUOXA2,DYSF,EBP,ECEL1,ECHS1,ECM1,EDAR,EDARADD,EDC3,EDN1,EDN3,EDNRB,EFEMP2,EGFR,EGR2,EIF2B1,EIF2B2,EIF2B3,EIF2B4,EIF2S3,EIF4A3,ELAC2,ELMO2,ELMOD3,ELOVL4,ELP2,EMC1,EMD,EMG1,EMP2,ENAM,ENO3,ENTPD1,EPCAM,EPG5,EPO,EPS8,EPS8L2,EPX,ERBB3,ERCC1,ERCC2,ERCC3,ERCC4,ERCC5,ERCC6,ERLIN1,ERLIN2,ESCO2,ESR1,ESRRB,ETFB,ETFDH,ETHE1,EVC2,EXOSC2,EXOSC3,EXOSC8,EXPH5,EXT1,EXT2,EXTL3,EYS,F12,F13A1,F13B,F2,F5,F7,F9,FA2H,FADD,FAH,FAM126A,FAM20A,FAM20C,FAN1,FANCI,FANCL,FAR1,FARS2,FASLG,FASTKD2,FAT4,FBP1,FBXL4,FBXO7,FCGR2A,FCGR3A,FCN3,FECH,FERMT1,FERMT3,FEZF1,FGA,FGB,FGD4,FGF16,FGF20,FGFR3,FH,FHL1,FIBP,FIG4,FKBP10,FKBP14,FKRP,FKTN,FLAD1,FLI1,FLNB,FLVCR1,FLVCR2,FMO3,FOLR1,FOXI1,FOXN1,FOXP3,FOXRED1,FRAS1,FREM1,FREM2,FRMPD4,FSHB,FSHR,FTL,FTSJ1,FUCA1,FUT1,FUT8,FXN,FYCO1,FZD6,G6PC,G6PC3,GAA,GAB1,GAD1,GALE,GALK1,GALNT3,GALT,GAMT,GAN,GATA1,GATA5,GBA2,GBE1,GCLC,GCNT2,GDAP1,GDF5,GDF6,GDF9,GEMIN4,GFI1B,GFM1,GFPT1,GGCX,GH1,GHRL,GHSR,GJA1,GJB2,GJB3,GJB6,GLB1,GLE1,GLI3,GLIS3,GLRA1,GLRB,GLUL,GLYCTK,GM2A,GMPPA,GMPPB,GNAT1,GNB3,GNB5,GNE,GNMT,GNPAT,GNPTAB,GNRH1,GNRHR,GNS,GORAB,GP9,GPC3,GPC6,GPD1,GPI,GPIHBP1,GPR179,GPR68,GPSM2,GPT2,GPX1,GRHL2,GRHPR,GRIA3,GRID2,GRIN1,GRIP1,GRM1,GRN,GRXCR1,GRXCR2,GSS,GSTZ1,GTPBP2,GTPBP3,GUCY2C,GUCY2D,GUF1,GULOP,GUSB,GYG1,GYS1,GYS2,H6PD,HADH,HADHA,HADHB,HAL,HAMP,HARS2,HAX1,HBB,HCFC1,HEPACAM,HERC1,HESX1,HEXB,HFE,HGD,HGF,HINT1,HK1,HMGCL,HMGCS2,HNF1A,HNMT,HOXA2,HOXB1,HPCA,HPD,HPGD,HPS1,HPS3,HPS4,HPS5,HPS6,HPSE2,HR,HSD17B3,HSD3B2,HSD3B7,HSPA9,HTRA2,HYAL1,HYLS1,IARS2,ICOS,IDS,IER3IP1,IFNAR2,IFNGR1,IFT140,IFT27,IFT43,IFT52,IGBP1,IGF1,IGF1R,IGHM,IGHMBP2,IGKC,IGLL1,IGSF1,IGSF3,IHH,IKBKB,IL10RA,IL10RB,IL11RA,IL12B,IL17RC,IL1RN,IL21R,IL2RA,IL2RG,IL36RN,IL7R,ILDR1,IMPG2,ING1,INPP5K,INS,INTU,INVS,IQCB1,IRF1,IRF7,IRF8,IRX5,ISCA2,ISCU,ISG15,ITGA2B,ITGA3,ITGA6,ITGA7,ITGB2,ITGB3,ITGB6,ITK,ITPA,ITPR3,IVD,IYD,JAGN1,JAK3,JAM3,JPH1,JUP,KANK2,KATNB1,KCNJ1,KCNJ10,KCNJ11,KCNJ13,KCNMA1,KCNV2,KCTD7,KERA,KHDC3L,KHK,KIAA0556,KIAA0753,KIF1A,KIF1C,KISS1,KISS1R,KIZ,KLC2,KLHDC8B,KLHL15,KLHL3,KLHL41,KLHL7,KLK4,KNG1,KPTN,KRAS,KRT1,KRT10,KRT14,KRT25,KRT5,KRT74,KRT83,KRT85,KY,KYNU,L1CAM,L2HGDH,LAMA1,LAMA2,LAMB1,LAMB2,LAMB3,LAMC2,LAMTOR2,LBR,LCK,LCT,LEMD2,LEP,LGI4,LHB,LHCGR,LHFPL5,LIG4,LIM2,LINS1,LIPC,LIPE,LIPH,LIPN,LIPT1,LMAN1,LMAN2L,LMF1,LMNA,LMOD3,LONP1,LPAR6,LPIN1,LPL,LRAT,LRBA,LRIG2,LRIT3,LRP2,LRPAP1,LRPPRC,LRRC6,LRSAM1,LRTOMT,LSS,LTBP2,LZTFL1,MAB21L2,MAD2L2,MAG,MAMLD1,MAN1B1,MAN2B1,MAOA,MAP3K8,MAPKBP1,MAPT,MARS2,MASP1,MAT1A,MBOAT7,MBTPS2,MC1R,MC2R,MC4R,MCCC1,MCEE,MCM5,MCM9,MECR,MED12,MED25,MEGF10,MEGF8,MEOX1,MET,METTL23,MFF,MFN2,MFRP,MFSD2A,MGAT2,MGME1,MGP,MID1,MID2,MITF,MKKS,MKS1,MLC1,MLPH,MMAA,MMACHC,MMADHC,MMP1,MMP2,MMP20,MOCOS,MOCS1,MOCS2,MOGS,MPDU1,MPDZ,MPI,MPL,MPLKIP,MPO,MPV17,MPZ,MRAP,MRPL3,MRPL44,MRPS16,MRPS2,MRPS22,MRPS34,MRPS7,MS4A1,MSH2,MSH3,MSH5,MSH6,MSMO1,MSN,MTFMT,MTHFD1,MTHFR,MTM1,MTMR2,MTPAP,MTR,MTRR,MTTP,MYH2,MYH7,MYL3,MYO18B,MYO1E,MYO7A,NAGA,NAGS,NANS,NARS2,NAT2,NBN,NCAPD2,NCAPH,NCF2,NCF4,NDE1,NDP,NDRG1,NDST1,NDUFA1,NDUFA10,NDUFA11,NDUFA12,NDUFA13,NDUFA2,NDUFA9,NDUFAF1,NDUFAF2,NDUFAF3,NDUFAF4,NDUFAF5,NDUFAF6,NDUFB11,NDUFB3,NDUFB9,NDUFS1,NDUFS2,NDUFS6,NDUFS8,NDUFV1,NDUFV2,NECAP1,NEFL,NEK8,NEK9,NEU1,NEUROG3,NGF,NGLY1,NHLRC1,NHP2,NIPAL4,NLRP1,NLRP7,NMNAT1,NOP10,NPC1,NPC2,NPHP1,NPHP4,NPHS1,NPHS2,NPPA,NPR2,NR0B2,NR1H4,NR2E3,NSDHL,NSUN2,NT5C3A,NT5E,NTHL1,NTRK1,NUP62,NUP93,NYX,OAT,OCA2,OGDH,OGT,OPA3,OPHN1,OPLAH,ORC1,ORC6,OSTM1,OTC,OTOF,OTOG,P2RY12,PADI3,PADI6,PAH,PAM16,PANK2,PAPSS2,PARK7,PAX3,PAX4,PAX7,PC,PCDH12,PCDH15,PCK1,PCK2,PCLO,PCNA,PCNT,PCSK1,PCYT1A,PDE6B,PDE6C,PDE6D,PDE6G,PDE6H,PDP1,PDSS2,PDX1,PEPD,PET100,PEX11B,PEX12,PEX13,PEX14,PEX16,PEX19,PEX2,PEX26,PEX3,PEX5,PEX6,PGAM2,PGAP1,PGAP2,PGAP3,PGK1,PGR,PHF6,PHGDH,PHKA1,PHKA2,PHKB,PHKG2,PHOX2A,PHYH,PIGC,PIGG,PIGL,PIGM,PIGO,PIGT,PIGV,PIGW,PIGY,PIH1D3,PIK3R1,PIK3R5,PKHD1,PKLR,PLA2G5,PLA2G6,PLA2G7,PLAA,PLCB1,PLCD1,PLCE1,PLCZ1,PLEKHG2,PLG,PLK4,PLOD1,PLOD3,PLP1,PMM2,PMP22,PMPCA,PNKP,PNLIP,PNP,PNPLA1,PNPLA2,PNPLA6,PNPLA8,PNPO,POC1B,POGLUT1,POLG,POLH,POLR1D,POLR3A,POLR3B,POMC,POMGNT1,POMGNT2,POMK,POMT1,POMT2,POP1,POR,POU3F4,PPARG,PPARGC1B,PPIB,PPP1R15B,PPP2R1B,PQBP1,PRDM5,PRDX1,PREPL,PRF1,PRG4,PRICKLE1,PRKACG,PRKRA,PRMT7,PROC,PROP1,PRPH,PRPH2,PRPS1,PRRX1,PRSS1,PRSS12,PRSS56,PRX,PSAP,PSAT1,PSMB4,PSMB8,PSMB9,PSMC3IP,PSPH,PTCH2,PTCHD1,PTGER2,PTH1R,PTPN14,PTPRF,PTRH2,PTS,PUS3,PYCR1,PYCR2,PYGL,PYGM,QDPR,RAB18,RAB23,RAB27A,RAB28,RAB33B,RAB39B,RAG1,RAG2,RAPSN,RARB,RASGRP2,RBBP8,RBCK1,RBM10,RBM8A,RBMX,RBP3,RBP4,RCBTB1,RD3,RDH11,RDH12,RDH5,RDX,REEP6,RELN,REN,RFT1,RFX5,RFX6,RFXAP,RHO,RIN2,RIPK4,

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RIPPLY2,RLBP1,RLIM,RMRP,RNASEH1,RNASEH2A,RNASEH2C,RNASET2,RNF168,RNU4ATAC,ROBO3,ROM1,ROR2,RORC,RPE65,RPIA,RPL10,RS1,RSPH1,RSPH3,RSPO1,RSPO4,RSPRY1,RTEL1,RTN4IP1,RTTN,RUSC2,S1PR2,SACS,SAG,SALL2,SAMD9,SAMHD1,SAR1B,SARS2,SBDS,SBF2,SC5D,SCARB2,SCN4A,SCN5A,SCN9A,SCNN1A,SCNN1B,SCNN1G,SCO1,SCO2,SCYL1,SDHAF1,SDR9C7,SEC23B,SEC24D,SEMA4A,SEPSECS,SERAC1,SERPINA1,SERPINA6,SERPINB7,SERPINB8,SERPINC1,SERPINE1,SERPINF2,SERPING1,SERPINH1,SETX,SFRP4,SFTPB,SFXN4,SGCA,SGCD,SGCG,SGSH,SHPK,SIGMAR1,SIL1,SIM1,SIX6,SKIV2L,SLC10A2,SLC12A3,SLC12A5,SLC13A5,SLC16A1,SLC17A5,SLC18A3,SLC19A2,SLC1A1,SLC1A4,SLC22A12,SLC22A18,SLC22A5,SLC24A4,SLC24A5,SLC25A12,SLC25A13,SLC25A15,SLC25A19,SLC25A20,SLC25A22,SLC25A26,SLC25A32,SLC25A4,SLC25A46,SLC26A1,SLC26A2,SLC26A3,SLC26A4,SLC26A5,SLC29A3,SLC2A1,SLC2A10,SLC2A2,SLC2A9,SLC30A10,SLC33A1,SLC34A1,SLC34A2,SLC34A3,SLC35A1,SLC35C1,SLC36A2,SLC37A4,SLC38A8,SLC39A13,SLC39A4,SLC45A2,SLC46A1,SLC4A11,SLC4A4,SLC52A2,SLC52A3,SLC5A1,SLC5A2,SLC5A5,SLC5A7,SLC6A17,SLC6A19,SLC6A20,SLC6A3,SLC6A5,SLC6A9,SLC7A14,SLC7A7,SLC7A9,SLC9A1,SLITRK6,SLURP1,SLX4,SMARCAL1,SMG9,SMIM1,SMOC1,SMOC2,SMPD1,SNAI2,SNAP29,SNIP1,SNORD118,SNX10,SNX14,SOD1,SOHLH1,SOST,SP110,SPARC,SPATA16,SPATA5,SPG11,SPG21,SPINK1,SPINK5,SPINT2,SPR,SPRTN,SPTA1,SQSTM1,SRD5A2,SRD5A3,SSR4,STAC3,STAMBP,STAR,STAT2,STIL,STIM1,STRA6,STRADA,STT3A,STT3B,STUB1,STX11,SUFU,SULT2B1,SUMF1,SUN5,SUOX,SYCE1,SYNE4,SZT2,TAC3,TACO1,TACR3,TACSTD2,TAF13,TAF4B,TAF6,TANGO2,TAP1,TARS2,TAT,TAZ,TBC1D24,TBC1D7,TBCD,TBX15,TBX19,TBX21,TBX6,TCAP,TCHH,TCIRG1,TCN2,TCTN2,TCTN3,TDO2,TDP2,TDRD7,TEC,TECR,TECTA,TELO2,TEX14,TEX15,TF,TFAM,TFG,TFRC,TG,TGDS,TGFB1,TGM1,TGM3,TGM5,THOC6,THRB,TIA1,TICAM1,TIMM50,TIMM8A,TJP2,TK2,TLE6,TLR3,TMC1,TMC6,TMC8,TMEM107,TMEM126A,TMEM126B,TMEM138,TMEM216,TMEM237,TMPRSS3,TMPRSS6,TNFRSF10B,TNFRSF11B,TNFRSF13B,TNFSF11,TNIK,TNNI3,TOE1,TPI1,TPK1,TPMT,TPO,TPP1,TRAC,TRAF3IP1,TRAF3IP2,TRAIP,TRAPPC11,TRAPPC9,TREH,TREM2,TREX1,TRH,TRIM32,TRIM37,TRIP13,TRIP4,TRMT10A,TRMT10C,TRMT5,TSHB,TSHR,TSPAN7,TSPEAR,TSPYL1,TSR2,TTC25,TTC37,TTI2,TTLL5,TTPA,TUB,TUBA8,TUBB8,TUBGCP4,TUBGCP6,TUFM,TULP1,TWIST2,TXN2,TXNL4A,TXNRD2,TYK2,TYR,TYROBP,TYRP1,UBE3B,UCHL1,UCP1,UCP3,UGT1A1,UMPS,UNC80,UPB1,UQCC2,UQCC3,UQCRB,UQCRQ,UROC1,UROD,UROS,USB1,USH1C,USH1G,USH2A,USP27X,USP8,UVSSA,VAC14,VARS,VARS2,VDR,VHL,VIPAS39,VLDLR,VPS11,VPS33B,VPS37A,VPS53,VRK1,VWA3B,WARS2,WAS,WBP2,WDR19,WDR35,WDR45B,WDR62,WDR81,WFS1,WNK1,WNT10B,WNT3,WNT7A,WRAP53,WRN,WWOX,XDH,XPA,XPC,XPNPEP3,XRCC1,XRCC2,XRCC4,YARS2,YME1L1,YY1AP1,ZAP70,ZBTB16,ZBTB24,ZBTB42,ZC4H2,ZFYVE26,ZIC3,ZMPSTE24,ZMYND10,ZMYND15,ZNF335,ZNF408,ZNF423,ZNF469,ZNF513,ZP1,ABCB7,ACACA,AFP,ALG6,ANK3,ATP2B2,CC2D1A,CEP120,CEP83,COL4A6,DENND5A,DLD,DMXL2,DNM1L,FANCB,FANCC,GAS8,ITPR1,KDM5C,LAMA3,LOXHD1,LRP1,MED23,MYO3A,NBAS,NDUFS7,NKX32,PHF8,PIBF1,PLEC,SLC25A38,SYNJ1,TECPR2,TOR1AIP1,UBE2T,ADAMTS18,AK2,ARL13B,BRWD3,DOCK8,DYNC2H1,EIF2AK4,GLDN,GP1BA,GTF2E2,IFT172,IFT80,IL1RAPL1,JAK2,KIF4A,LRP4,LYST,MATN3,MMP21,MYPN,ORC4,PLCB4,POC1A,PTPRQ,SLC4A1,SLCO1B3,SPTBN2,TUSC3,UNG,USP9X,WDR34,ANO6,AP1S1,ARSB,ASAH1,BCKDHB,COG2,CP,CYP21A2,KIAA1109,KRT18,LAS1L,LHX3,LIFR,MAK,MARVELD2,MASP2,MUSK,MYO5A,NALCN,NEK1,NUP107,PAK3,PLOD2,PRKCD,SUCLG1,TBXAS1,TMEM231,TRIP11,VPS13A,WNT10A,AASS,ADA,CEP290,ERCC6L2,GSC,HSPD1,IL17RD,LAMC3,MLH1,SLCO2A1,SPEG,TMTC3,TRPM6,TTC21B,ABCB11,ATP6AP1,CENPE,CEP78,COL11A2,DCLRE1C,HES7,ITPR2,LIPA,MCM8,MUTYH,NADK2,NBEAL2,P3H2,PIK3CA,RAB3GAP2,SUCLA2,AFF2,ALDH1A3,AP3D1,ATP8B1,BRAF,COL11A1,COL18A1,FGG,IDUA,ITGB4,NPHP3,NRXN1,PNPT1,PRKDC,PTEN,RECQL4,SDHA,SEC23A,SI,SLC12A1,SLC25A1,TBC1D23,ADAM22,AGRN,ALDH7A1,ATRX,CASP8,FGF5,HEXA,IBA57,ITGA8,NR0B1,POF1B,SH3PXD2B,SH3TC2,SP7,ABCD3,ALDH4A1,AUH,CD81,INPP5E,MED17,MYO15A,PLEKHG5,SLC12A6,VPS13C,WNT1,ACADM,AR,DMP1,FGD1,GOSR2,IQCE,MDH2,MIPEP,PCCB,PROM1,PTPRO,SBF1,ACE,ATP13A2,COL27A1,DCDC2,DNAJC21,FANCD2,GHR,PXDN,SH2D1A,SLC24A1,SLC6A8,CNNM4,GCH1,GP6,KRT8,MMAB,NAA10,RBM28,STAG3,TNFRSF4,ATF6,COL25A1,DOCK6,HACE1,KIF14,LAT,MCM4,OTOGL,PUS1,SLC11A2,ST14,BRCA1,CC2D2A,CFI,DNAL1,GIPC3,HIBCH,KLKB1,MCCC2,OPA1,SCP2,SPAG1,TAPBP,TBCK,UBA1,UBA5,VPS13B,CLCF1,HFM1,MAGED2,OFD1,PTPN22,SIPA1L3,SLCO1B1,TALDO1,THOC2,UBE2A,AP3B1,COA6,HTT,PPT1,RP1,TRIT1,CEP135,DSC2,IL17RA,PIEZO1,RB1,SLC3A1,CFAP53,CFD,NEK2,SLC19A3,AP3B2,DHFR,DNAJC12,GPX4,ICK,LTBP4,SASS6,CRB2,GALC,PEX1,PFKM,POLA1,RMND1,TP53RK,VPS45,ARHGEF18,COL1A2,LDHA,UPF3B,ANOS1,ASPH,HOXC13,NIN,FMN2,GATM,GCDH,GLRX5,MANBA,RUBCN,SPG7,CARS2,CR2,FLNA,NSMCE2,TMEM199,WDR73,ERBB2,OCRL,RASSF1,CDH23,MERTK,ARNT2,PGM1,TAP2,UBR1,EPM2A,GK,SLC35D1,TERT,TMEM165,TMEM70,TSEN15,XIAP,F10,GATAD1,LCAT,PPA2,TMIE,TMPRSS15,FBXO31,INPPL1,TAF1,UQCRC2,ORAI1,EML1,HMX1,PIEZO2,XYLT2,DHCR24,HPRT1,RAX,CLCN2,DOCK7,KIF7,NAGLU,NDUFS3,PIGN,NUP155,PI4KA,SURF1,TTC7A,CYP2A6,RNF216,CHRNB1,NT5C2,PYROXD1,RAD51C,TRNT1,TENM3,CTDP1,ADAMTS13,NUS1,POU1F1,TBC1D20,AGL,NDUFS4,PAX1,SLC39A14,APOB,CPS1,HSPG2,REEP2,NME8,MYO5B,DDX3X,SERPINB6,MCPH1,TCTN1,CCDC174,LMBR1,DLL3,CAST,MEFV,BFSP1,LTBP3,SMARCD2,AMH,P3H1,SARDH,TRAPPC2,BRCA2,GPHN,PINK1,RAB3GAP1,RYR1,MSRB3,PIP5K1C,TMCO1,POLE,WDR60,CDCA7,FGF3,PTF1A,SYNE1,SDCCAG8,CLEC7A,FOXE1,PCBD1,TYMP,MECP2,C2,CEP63,LARP7,HELLS,TH,POLR1C,APC2,NNT,AFG3L2,ABCC2,POMP,TMEM67,CLCN5,SGCB,PGM3,ROGDI,TRMU,CCDC151,TTN,GALNS,EIF2AK3,PMS2,TTC19,PDHX,TRDN,ACER3,AIMP2,MYBPC1,ALDH5A1,VMA21,NFU1,CLIC5,TAF2,MCOLN1,FTCD,MPC1,INSR,LTC4S,DEAF1,ARHGEF9,SLC39A8,HLADQB1,TKT,BLM,PROS1,TSEN54,PKD1L1,CYC1,MME,TRIOBP,CPAMD8,CLN6,FANCE,IMPAD1,SOBP,BCAM,NEFH,PRDM8,GPR88,HLADQA1,NCAPD3,ABCC6,F8,TCTEX1D2,DHDDS,DMGDH,CUL4B,BCKDHA,ESPN,ENPP1,TECRL,FOXO1,EIF2B5,TRIM2,BUB1B,IFT122,VWF,CAPN3,LRP5,SYN1,AK7,SCARF2,CCDC88A,EDA,ARSA,PEX10,HSD17B4,MTO1,B4GALT7,LMNB2,CRBN,COA5,DGAT1,FANCA,TBCE,AGPS,CYB5R3,TSEN2,CHEK2,GJC2,SYT14,ANKLE2,BMPER,DYM,GCSH,LDLRAP1,AP1S2,CLN5,MYO6,DPY19L2,PTPRC,MALT1,TAPT1,DIAPH1,KIAA0586,WNT4,IL12RB1,AP4M1,ADAM17,CISD2,GP1BB,ADCY1,EPB41,STS,GRM6,CSPP1,TSFM,KCNQ1,COQ2,GNPTG,DCAF17,CDC45,EARS2,TFR2,TNNT1,CHSY1,FRMD4A,IGFBP7,PCCA,CD59,ERCC8,LMBRD1,GLDC,ADGRG1,SUGCT,GSR,WDR72,AGK,BOLA3,DUOX2,RPGRIP1L,CDT1,ACY1,ADAMTS2,SLC35A3,VAX1,XYLT1,TEX11,SCN1B,COL13A1,ST3GAL5,ST3GAL3,CDC14A,ADAMTS17,LARS2,B3GLCT,ALG8,PARN,GRIK2,ANKS6,MAGI2,ATAD3A,APTX,C2CD3,PEX7,RRM2B,GCK,GPNMB,ATP7B,ITCH,LYRM7,VPS33A,ZC3H14,EVC,AIMP1,MFSD8,RNASEH2B,MMP13,DPM1,NUBPL,ARMC4,IFNGR2,TPM3,TP53,TNFRSF13C,STAT1,TTC8,BBS9,KREMEN1,C15orf41,ARV1,PIGA,SLC9A3,GBA,EOGT,PDE10A,ALDH3A2,IFT81,RARS2,CD55,LIMS2,FTO,PHC1,ABHD5,B3GALNT2,CORO1A,GDF1,HLCS,PDZD7,ARX,ARG1,ATOH7,FBLN5,WDPCP,CRLF1,USP18,IMPA1,IFT74,MVK,RFXANK,SMS,ZNF141,CLN3,PRDM12,FRRS1L,SOX18,ETFA,MICU1,SDC3,COX15,PDSS1,KCNE1,DYNC2LI1,CERS1,NAT8L,HERC2,TNXB,AKR1C2,OTULIN,BLOC1S6,NEB,ATP6V1E1,RPGR,DNAAF5,FOXE3,AP4S1,ACAN,LIAS,BHLHA9,ASCC1,HGSNAT,CFHR3,HTRA1,MLYCD,ABHD12,OCLN,SDHD,TPRN,BTD,ADK,OTOA,HSD11B2,B3GALT6,TMLHE,HYDIN,CFHR1,OPN1LW