anderson Clinical Genetics
No 26, Rajarathinam Street, Kilpauk, Chennai - 600 010.
Helpline No: 044-4353 9444 / 99620 00048 | Email: info @andersondiagnostics.com
www.andersondiagnostics.com | Home Collection : 91766 77601

Whole Exome Sequencing Analysis – Carrier Screening

Patient name	**: XY**	**XX**
Gender/ Age	**: XXX**	**XXX**
PIN	**: XXX**	**XXX**
Sample no	**: XXX**	**XXX**
Specimen	**: Peripheral blood**	**Peripheral blood**
Sample collection date	**: XXX**	**XXX**
Sample receipt date	**: XXX**	**XXX**
Report date	**: XXX**	**XXX**
Referring clinician	**: XXX**	
Hospital/Clinic	**: XXX**	

Clinical history

XY and XX are a nonconsanguineous couple. At 14 years of age, XX had weakness in her lower limbs and had an abnormal gait. The muscle biopsy was indicative of muscular dystrophy (report unavailable). They have an ongoing pregnancy of 15 weeks 2 days GA. XY and XX have been evaluated for pathogenic variations.

Results

XX is found to be affected with likely pathogenic variant in the *DYSF* gene (p.Leu1341Pro), and found to be a carrier of likely pathogenic variant in the *EPRS1* gene (p.Tyr377Ter), *FCSK* gene (p.Val225CysfsTer42) and found to be a carrier of uncertain significance variant in the *L1CAM* gene (p.Arg491Ser).

XX is found to be a carrier of likely pathogenic variant in the *CEP152* gene (c.3345+1G>A), *CTSF* gene (p.Glu387Lys) and *DNAAF2* gene (p.Glu437Ter).

List of significant variant identified related to the phenotype:

Disease	XY	XX
Miyoshi muscular dystrophy 1 **(OMIM#254130)** Muscular dystrophy, limb-girdle, autosomal recessive 2 **(OMIM#253601)** Myopathy, distal, with anterior tibial onset **(OMIM#606768)** **Mode of inheritance: AR**	**NON-CARRIER**	**AFFECTED** Gene: *DYSF* Exon 38, c.4076T>C, p.Leu1359Pro , Homozygygous **Classification: Likely pathogenic**

List of carrier variants identified:

Disease	XY	XX
Ceroid lipofuscinosis, neuronal, 13 (Kufs type) (**OMIM#615362)** **Mode of inheritance: AR**	**CARRIER** Gene: *CTSF* Exon 9, c.1159G>A, p.Glu387Lys, Heterozygous **Classification: Likely pathogenic**	**NON-CARRIER**
Nephrotic syndrome, type 12 **(OMIM#616892)** **Mode of inheritance: AR**	**CARRIER** Gene: *DNAAF2* Exon 1, c.1309G>T, p.Glu437Ter, Heterozygous **Classification: Likely pathogenic**	**NON-CARRIER**
Microcephaly 9, primary,autosomal recessive **(OMIM#614852)** Seckel syndrome 5 **(OMIM#613823)** **Mode of inheritance: AR**	**CARRIER** Gene: *CEP152* Intron 20, c.3345+1G>A, 5' splice site, Heterozygous **Classification: Likely pathogenic**	**NON-CARRIER**

Leukodystrophy, hypomyelinating, 15 **(OMIM# 617951)** **Mode of inheritance: AR**	**NON-CARRIER**	**CARRIER** Gene: *EPRS1* Exon 10, c.1130_1131delAT, p.Tyr377Ter, Heterozygous **Classification: Likely pathogenic**
Congenital disorder of glycosylation with defective fucosylation 2 **(OMIM#618324)** **Mode of inheritance: AR**	**NON-CARRIER**	**CARRIER** Gene: *FCSK* Exon 9, c.669_670delTG, p.Val225CysfsTer42, Heterozygous **Classification: Likely pathogenic**
Hydrocephalus, congenital, X-linked **(OMIM#307000)** MASA syndrome **(OMIM#303350)** **Mode of inheritance: XLR**	**NON-CARRIER**	**CARRIER** Gene: *L1CAM* Intron 9, c.860G>A, p.Arg287His , Heterozygous **Classification: Uncertain Significance**

*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 variants identified in XX

***DYSF*:c.4076T>C**

Variant summary: A homozygous missense variation in exon 38 of the *DYSF* gene (chr2:g.71611481T>C, NM_001130987.2, Depth: 154x) that results in the amino acid substitution of Proline for Leucine acid at codon 1359 (p.Leu1359Pro) was detected.

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

Clinvar and Literature evidence: This variant has been previously classified as pathogenic in ClinVar database [3]. This variant has been previously reported in a patients affected with limb girdle muscular dystrophy 2B in homozygous state [4].

***In-silico* prediction:** The in-silico predictions of the variant are damaging by PolyPhen-2 (HumDiv), SIFT, LRT and MutationTaster2. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Miyoshi muscular dystrophy 1 (OMIM#254130) and Myopathy, distal, with anterior tibial onset (OMIM#606768) are caused by homozygous mutation in the *DYSF* gene (OMIM*603009). Muscular dystrophy, limb-girdle, autosomal recessive 2 (OMIM#253601), is caused by homozygous or compound heterozygous mutation in the *DYSF gene* (OMIM*603009). This disease follows autosomal recessive pattern of inheritance [2].

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

Interpretation for the significant variants identified in XY

***CTSF*:c.1159G>A**

Variant summary: A heterozygous missense variation in exon 9 of the *CTSF* gene (chr11:g.66564893C>T, NM_003793.4, Depth: 180x) that results in the amino acid substitution of Lysine for Glutamic acid at codon 387 (p.Glu387Lys) was detected.

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

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

***In-silico* prediction:** The *in-silico* predictions of the variant are damaging by PolyPhen-2 (HumDiv), SIFT, LRT and MutationTaster2. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Ceroid lipofuscinosis, neuronal, 13 (Kufs type) (OMIM#615362), caused by homozygous or compound heterozygous mutation in the CTSF gene (OMIM*603539). This disease follows autosomal recessive pattern of inheritance [2].

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

***DNAAF2*:c.1309G>T**

Variant summary: A heterozygous stop gained variation in exon 1 of the *DNAAF2* gene (chr14:g.49633841C>A, NM_018139.3, Depth: 215x) that results in the premature truncation of the protein at codon 437 (p.Glu437Ter) was detected. This variant is a stop gained variant which occurs in an exon of *DNAAF2* upstream of 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.

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

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

OMIM phenotype: Ciliary dyskinesia, primary, 10 (OMIM# 612518) is caused by homozygous or compound heterozygous mutation in the *DNAAF2* gene (OMIM*612517). This disease follows autosomal recessive pattern of inheritance [2].

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

***CEP152*:c.3345+1G>A**

Variant summary: A heterozygous 5' splice site variation in intron 20 of the *CEP152* gene (chr15:g.48755902C>T, NM_001194998.2, Depth: 125x) that affects the invariant GT donor splice site of exon 5(c.3345+1G>A) 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. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Microcephaly 9, primary, autosomal recessive(OMIM#614852) and Seckel syndrome 5 (OMIM#613823) are caused by homozygous or compound heterozygous mutation in the *CEP152* gene (OMIM*613529). 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 significant variants identified in XX

***EPRS1*:c.1130_1131delAT**

Variant summary: A heterozygous stop gained variation in exon 10 of the *EPRS1* gene (chr1:g.220020206delAT, NM_004446.3, Depth: 131x) that results in the premature truncation of the protein at codon 377 (p.Tyr377Ter) was detected. This variant is a stop gained variant which occurs in an exon of *EPRS1* upstream of 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.

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

OMIM phenotype: Leukodystrophy, hypomyelinating, 15 (OMIM#617951) is caused by homozygous or compound heterozygous mutation in the *EPRS1* gene (OMIM*138295). This disease follows autosomal recessive pattern of inheritance [2].

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

***FCSK*:c.669_670delTG**

Variant summary: A heterozygous two base pair deletion in exon 9 of the *FCSK* gene (chr16:g.70468854delTG, NM_145059.3, Depth: 163x) that results in a frameshift and premature truncation of the protein 42 amino acids downstream to codon 225 (p.Val225CysfsTer42) was detected. This variant is a frameshift variant which occurs in an exon of *FCSK* 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 a minor allele frequency of 0.0013% in gnomAD database and has not been reported in 1000 genomes database.

Clinvar evidence: This variant has been classified as a likely pathogenic variant in ClinVar database [7].

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

OMIM phenotype: Congenital disorder of glycosylation with defective fucosylation 2 (OMIM#618324) is caused by homozygous or compound heterozygous mutation in the *FCSK* gene (OMIM*608675). This disease follows autosomal recessive pattern of inheritance [2].

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

Interpretation for the uncertain significant variant identified in XX

***L1CAM*:c.860G>A**

Variant summary: A heterozygous missense variation in exon 9 of the *L1CAM* gene (chrX:g.153870187C>T, NM_001278116.2, Depth: 282x) that results in the amino acid substitution of Histidine for Arginine at codon 287 (p.Arg287His) was detected.

Population frequency: This variant has a minor allele frequency of 0.0027% in gnomAD database and has a minor allele frequency of 0.0265% in 1000 genomes database.

Clinvar evidence: This variant has been classified as variant of uncertain significance in ClinVar database [8].

***In-silico* prediction:** The *in-silico* predictions of the variant are damaging by PolyPhen-2 (HumDiv), SIFT and LRT. The reference codon is conserved across mammals in PhyloP and GERP++ tools.

OMIM phenotype: Hydrocephalus, congenital, X-linked (OMIM#307000) and MASA syndrome (OMIM#303350) are caused by mutation in the *L1CAM* gene (OMIM*308840) This disease follows X-linked recessive pattern of inheritance [2].

Variant classification: Based on the evidence, this variant is classified as 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 Illumina 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 haplotype caller 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

	XY	XX
Total reads generated	14.70 Gb	13.91 Gb
Data ≥ Q30	95.04%	95.04%

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

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 [9].

References

1. Richards, S, et al. Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in medicine: official journal of the American College of Medical Genetics. 17.5 (2015): 405-424.
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7. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV001703172.3
8. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000092935.5
9. Kalia S.S. et al., Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2016 update (ACMG SF v2.0): a policy statement of the American College of Medical Genetics and Genomics. Genet Med., 19(2):249-255, 2017.

This report has been reviewed and approved by:

Sivasankar.S, Ph.D Molecular Biologist	Muthukumaran. S, Ph.D Clinical Bioinformatician	Sachin. D.Honguntikar, Ph.D, Molecular Geneticist	Dr. G. Suriyakumar Director

No 26, Rajarathinam Street, Kilpauk, Chennai - 600 010.
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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