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

Clinical Exome Sequencing Analysis – Carrier Screening

Patient name	**: Mr. XXX**	**Mrs. YYY**
Gender/ Age	**: Male/ 36 Years**	**Female/ 31 years**
PIN	**: XX**	**XX**
Sample no	**: XX**	**XX**
Specimen	**: Peripheral blood**	**Peripheral blood**
Sample collection date	**: XX**	**XX**
Sample receipt date	**: XX**	**XX**
Report date	**: XX**	**XX**
Referring clinician	**: XX**	**XX**
Hospital/Clinic	**: XX**	

Clinical history

Mr. XXX is married to Mrs. YYY and presented with bad obstetric history. Their first-born female child was affected with Joubert syndrome. Her focused exome analysis (done elsewhere) was indicative of homozygous pathogenic variant c.1094dupT in *AHI1* gene causative of joubert syndrome 3 and heterozygous variant of uncertain significance c.1816C>G in *RPGRIP1L* gene causative of COACH syndrome, Joubert syndrome 7 and Meckel syndrome 5. Targeted sequence analysis (done elsewhere) in both parents was indicative of presence of the variant c.1094dupT in *AHI1* gene in heterozygous state in both partners. In their second pregnancy (monochorionic diamniotic twin pregnancy) invasive prenatal diagnosis via chorionic villus sampling was done by sanger sequencing indicative of fetuses with homozygous pathogenic variant c.1094dupT in *AHI1* gene and terminated the pregnancy. In their third pregnancy invasive prenatal diagnosis via chorionic villus sampling was done by sanger sequencing (done elsewhere) which revealed the fetus harbour the pathogenic variant c.1094dupT in *AHI1* gene in heterozygous state. Mr. XXX and Mrs. YYY have been evaluated for carrier status of pathogenic variations.

Results

Mr. XXX and Mrs. YYY are found to be carriers of pathogenic variant in the *AHI1* gene (p.Met365IlefsTer8) and found to be carriers of uncertain significance variant in the *MECR* gene (p.Asp352Glu).

Mr. XXX is found to be carrier of likely pathogenic variants in the *GLRX5* gene (p.Leu148Ser), *TYR* gene (p.Ala355Val) and found to be carrier of uncertain significance variant in *OTOG* gene (p.Cys2828Arg).

Mrs. YYY is found to be carrier of uncertain significance variant in the *OTOG* gene (p.Arg1144Trp).

List of common significant variant identified related to the phenotype:

Disease	Mr. XXX	Mrs. YYY
Joubert syndrome 3 **(OMIM#608629)** **Mode of inheritance: AR**	**CARRIER** Gene: *AHI1* Exon 9, c.1094dupT, p.Met365IlefsTer8, Heterozygous **Classification: Pathogenic**	**CARRIER** Gene: *AHI1* Exon 9, c.1094dupT, p.Met365IlefsTer8, Heterozygous **Classification: Pathogenic**

List of carrier variants identified:

Disease	Mr. XXX	Mrs. YYY
Anemia, sideroblastic, 3, pyridoxine-refractory **(OMIM#616860)** Spasticity, childhood-onset, with hyperglycinemia **(OMIM#616859)** **Mode of inheritance: AR**	**CARRIER** Gene: *GLRX5* Exon 2, c.443T>C, p.Leu148Ser, Heterozygous **Classification: Likely Pathogenic**	**NON - CARRIER**
Albinism, oculocutaneous, type IA **(OMIM#203100)** Albinism, oculocutaneous, type IB **(OMIM#606952)** **Mode of inheritance: AR**	**CARRIER** Gene: *TYR* Exon 3, c.1064C>T, p.Ala355Val, Heterozygous **Classification: Likely Pathogenic**	**NON - CARRIER**

Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities **(OMIM#617282)** **Mode of inheritance: AR**	**CARRIER** Gene: *MECR* Exon 10, c.1056C>G, p.Asp352Glu, Heterozygous **Classification: Uncertain Significance**	**CARRIER** Gene: *MECR* Exon 10, c.1056C>G, p.Asp352Glu, Heterozygous **Classification: Uncertain Significance**
Deafness, autosomal recessive 18B **(OMIM#614945)** **Mode of inheritance: AR**	**CARRIER** Gene: *OTOG* Exon 55, c.8482T>C, p.Cys2828Arg, Heterozygous **Classification: Uncertain Significance**	**CARRIER** Gene: *OTOG* Exon 29, c.3430C>T, p.Arg1144Trp, Heterozygous **Classification: Uncertain Significance**

Previously reported heterozygous variant of uncertain significance in *RPGRIP1L* gene (c.1816C>G; p.Leu606Val) in index baby sample is found in heterozygous state in the sample of Mr. XXX in this couple carrier analysis.

*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 common significant variant identified in Mr. XXX and Mrs. YYY related to the phenotype

***AHI1*: c.1094dupT**

Variant summary: A heterozygous single base pair insertion in exon 9 of the *AHI1* gene (chr6:g.135457551insA, NM_001134831.2, Depth: >143x) that results in a frameshift and premature truncation of the protein 8 amino acids downstream to codon 365 (p.Met365IlefsTer8) was detected. This variant is a frameshift variant which occurs in an exon of *AHI1* 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 databases.

Clinical evidence: This variant has been previously classified as likely pathogenic in ClinVar database [3].

***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: Joubert syndrome 3 (OMIM#608629) is caused by homozygous mutation in the *AHI1* gene (OMIM*608894). This disease follows autosomal recessive pattern of inheritance [2].

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

This variant [*AHI1*: c.1094dupT; p.Met365IlefsTer8] has been previously reported in index child in homozygous state associated with Joubert syndrome.

Interpretation for the significant carrier variants identified in Mr. XXX

***GLRX5:* c.443T>C**

Variant summary: A heterozygous missense variation in exon 2 of the *GLRX5* gene (chr14:g.95544094T>C, NM_016417.3, Depth: 107x) that results in the amino acid substitution of Serine for Leucine at codon 148 (p.Leu148Ser) was detected.

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

Clinical and Literature evidence: This variant has been previously classified as a pathogenic in ClinVar database [4]. This variant has been previously reported in a patient affected with congenital sideroblastic anemia in compound heterozygous state [5].

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

OMIM phenotype: Anemia, sideroblastic, 3, pyridoxine-refractory (OMIM#616860) and Spasticity, childhood-onset, with hyperglycinemia (OMIM#616859) are caused by homozygous or compound heterozygous mutation in the *GLRX5* gene (OMIM*609588). These diseases follow autosomal recessive pattern of inheritance [2].

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

***TYR*: c.1064C>T**

Variant summary: A heterozygous missense variation in exon 3 of the *TYR* gene (chr11:g.89227850C>T, NM_000372.5, Depth: 125x) that results in the amino acid substitution of Valine for Alanine at codon 355 (p.Ala355Val) was detected.

Population frequency: This variant has a minor allele frequency of 0.0132% in gnomAD database and has not been reported in 1000 genomes databases.

Clinical and Literature evidence: This variant has been previously classified as a likely pathogenic in ClinVar database [6]. This variant has been previously reported in a patient affected with oculocutaneous albinism in compound heterozygous state [7].

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

OMIM phenotype: Albinism, oculocutaneous, type IA (OMIM#203100) and Albinism, oculocutaneous, type IB (OMIM#606952) are caused by homozygous or compound heterozygous mutation in the *TYR* gene (OMIM*606933). These diseases follow autosomal recessive pattern of inheritance [2].

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

Interpretation for the common uncertain significant variant identified in Mr. XXX and Mrs. YYY

***MECR*: c.1056C>G**

Variant summary: A heterozygous missense variation in exon 10 of the *MECR* gene (chr1:g.29194088G>C, NM_016011.5, Depth: >138x) that results in the amino acid substitution of Glutamic acid for Aspartic acid at codon 352 (p.Asp352Glu) was detected.

Population frequency: This variant has minor allele frequency of 0.0053% in gnomAD database and has a minor allele frequency of 0.039% in 1000 genomes databases.

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

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

OMIM phenotype: Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities (OMIM#617282) is caused by homozygous or compound heterozygous mutation in the *MECR* gene (OMIM*608205). 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.

Interpretation for the uncertain significant variant identified in Mr. XXX

***OTOG*: c.8482T>C**

Variant summary: A heterozygous missense variation in exon 55 of the *OTOG* gene (chr11:g.17645584T>C, NM_001292063.2, Depth: 120x) that results in the amino acid substitution of Arginine for Cysteine at codon 2828 (p.Cys2828Arg) was detected.

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

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

OMIM phenotype: Deafness, autosomal recessive 18B (OMIM#614945) is caused by homozygous or compound heterozygous mutation in the *OTOG* gene (OMIM*604487). 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.

Interpretation for the uncertain significant variant identified in Mrs. YYY

***OTOG*: c.3430C>T**

Variant summary: A heterozygous missense variation in exon 29 of the *OTOG* gene (chr11:g.17596059C>T, NM_001292063.2, Depth: 165x) that results in the amino acid substitution of Tryptophan for Arginine at codon 1144 (p.Arg1144Trp) was detected.

Population frequency: This variant has minor allele frequency of 0.0033% in gnomAD database and has a minor allele frequency of 0.03% in 1000 genomes databases.

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

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

OMIM phenotype: Deafness, autosomal recessive 18B (OMIM#614945) is caused by homozygous or compound heterozygous mutation in the *OTOG* gene (OMIM*604487). 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

- The *TYR* gene has pseudogene in the human genome. Validation of the variant by Sanger sequencing is strongly recommended to rule out false positives.
- 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 targeted gene capture using a custom 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++, CADD, 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	5.33 Gb	6.58 Gb
Data ≥ Q30	92.80%	92.14%

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 [10].

References

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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

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,

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