No 26, Rajarathinam Street, Kilpauk, Chennai - 600 010.
Helpline No: 044-4353 9444 / 99620 00048 | Email: info @andersondiagnostics.com
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Whole Exome Sequencing Analysis – Carrier Screening

Patient name	**: XX**	**XXX**
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

XX and XXX are a non-consanguineous couple. XX was diagnosed with progressive vision loss. Whole exome sequencing of XX (done elsewhere) revealed homozygous likely pathogenic variant in *ABCA4* gene causative of Retinitis pigmentosa 19, Autosomal recessive. Additionally, he was found to be a carrier for a likely pathogenic variant in *ACE* gene associated with Renal tubular dysgenesis and a likely pathogenic variant in *ODAD3* gene associated with Ciliary dyskinesia, primary, 30. XX and XXX have been evaluated for carrier status of pathogenic variations.

Results

XX is found to be affected with likely pathogenic variant in the *ABCA4* gene (p.Tyr665Ter), and found to be a carrier of likely pathogenic variants in the *ACE* gene (p. Leu273Phe) and *ODAD3* gene (p. Arg198LysTer57).

XXX is found to be a carrier of likely pathogenic variant in the *ABCG5* gene (p.Phe260ThrfsTer6).

List of significant variants identified:

Disease	XX	XXX
Retinitis pigmentosa 19 **(OMIM#601718)** **Mode of inheritance: AR**	**AFFECTED** Gene: *ABCA4* Exon 14, c.1995C>A, p. Tyr665Ter, Homozygous **Classification: Likely pathogenic**	**NON - CARRIER**
Renal tubular dysgenesis **(OMIM#267430)** **Mode of inheritance: AR**	**CARRIER** Gene: *ACE* Exon 5, c.817C>T, p. Leu273Phe, Heterozygous **Classification: Likely pathogenic**	**NON - CARRIER**
Ciliary dyskinesia, primary, 30 **(OMIM#616037)** **Mode of inheritance: AR**	**CARRIER** Gene: *ODAD3* Exon 4, c.592dupA, p. Arg198LysTer57, Heterozygous **Classification: Likely pathogenic**	**NON - CARRIER**
Sitosterolemia 2 **(OMIM#618666)** **Mode of inheritance: AR**	**NON - CARRIER**	**CARRIER** Gene: *ABCG5* Exon 7, c.777_780delCTTT, p. Phe260ThrfsTer6, Heterozygous **Classification: Likely pathogenic**

*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

***ABCA4:* c.1995C>A**

Variant summary: A homozygous stop-gained variation in exon 14 of the *ABCA4* gene (chr1:g.94060702G>T, NM_000350.3, Depth: 165x) that results in the premature truncation of the protein at codon 665 (p. Tyr665Ter) was detected. This variant is predicted to cause loss of normal protein function through protein truncation.

Population frequency: This variant has a minor allele frequency of 0.0007% in gnomAD database and has not been reported in 1000 genomes database.

Clinical and Literature evidence: This variant has been classified as pathogenic in ClinVar database [3]. This variant has been previously reported in Indian patient for *ABCA4* mutations associated with Autosomal Recessive Retinitis Pigmentosa [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++ tool.

OMIM phenotype: Retinitis pigmentosa 19 (OMIM#601718) is caused by homozygous or compound heterozygous mutation in the *ABCA4* gene (OMIM*601691). This disease follows autosomal recessive pattern of inheritance [2].

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

***ACE:* c.817C>T**

Variant summary: A heterozygous missense variation in exon 5 of the *ACE* gene (chr17:g.63480498C>T, NM_000789.4, Depth: 159x) that results in the amino acid substitution of Phenylalanine for Leucine at codon 273 (p. Leu273Phe) was detected.

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

Clinical and Literature 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 SIFT, PolyPhen-2, LRT and MutationTaster2. The reference codon is conserved across mammals in PhyloP and GERP++ tool.

OMIM phenotype: Renal tubular dysgenesis (OMIM#267430) is caused by homozygous or compound heterozygous mutation in *ACE* gene (OMIM*106180). This disease follows autosomal recessive pattern of inheritance [2].

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

***ODAD3:* c.592dupA**

Variant summary: A heterozygous single base pair insertion in exon 4 of the *ODAD3* gene (chr19:g.11426893insT, NM_145045.5, Depth: 158x) that results in a frameshift and premature truncation of the protein 57 amino acids downstream to codon 198 (p. Arg198LysTer57) was detected. This variant is predicted to cause loss of normal protein function through protein truncation caused a frameshift mutation.

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

OMIM phenotype: Ciliary dyskinesia, primary, 30 (OMIM#616037) is caused by homozygous mutation in the *ODAD3* gene (OMIM*615956). This disease follows autosomal recessive pattern of inheritance [2].

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

Interpretation for the significant variant identified in XXX

***ABCG5:* c.777_780delCTTT**

Variant summary: A heterozygous four base pair deletion in exon 7 of the *ABCG5* gene (chr2:g.43825013delAAAG, NM_022436.3, Depth: 71x) that results in the frameshift and premature truncation of the protein 6 amino acids downstream to codon 260 (p. Phe260ThrfsTer6) was detected. This variant is a frameshift variant which occurs in an exon of *ABCG5* 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: The variant has not been reported in gnomAD and 1000 genomes database.

***In-silico* prediction:** The reference codon is conserved across mammals in PhyloP and GERP++ tool.

OMIM phenotype: Sitosterolemia 2 (OMIM#618666) is caused by homozygous or compound heterozygous mutation in the *ABCG5* gene (OMIM*605459). This disease follows autosomal recessive pattern of inheritance [2].

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

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 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

	XX	XXX
Total reads generated	11.18 Gb	7.79 Gb
Data ≥ Q30	94.86%	93.36%

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 [6].

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.
2. Amberger J, Bocchini CA, Scott AF, Hamosh A. McKusick's Online Mendelian Inheritance in Man (OMIM). Nucleic Acids Res. 2009 Jan;37(Database issue):D793-6. doi: 10.1093/nar/gkn665. Epub 2008 Oct 8. PMID: 18842627; PMCID: PMC2686440.
3. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000866511.5
4. Singh HP, Jalali S, Narayanan R, Kannabiran C. Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening. Invest Ophthalmol Vis Sci. 2009 Sep;50(9):4065-71. doi: 10.1167/iovs.09-3479. Epub 2009 Apr 1. PMID: 19339744; PMCID: PMC2777646.
5. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000432142.2

6. 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.
Helpline No: 044-4353 9444 / 99620 00048 | Email: info @andersondiagnostics.com
www.andersondiagnostics.com | Home Collection : 91766 77601

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