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Clinical Exome Sequencing Analysis – Carrier Screening

Patient name	**: Mr. XXX**	**Mrs. YYY**
Gender/ Age	**: Male/ 36 years**	**Female/ 30 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**	
Hospital/Clinic	**: XX**	

Clinical history

Mr. XXX and Mrs. YYY are third degree consanguineous couple. Their first pregnancy was IUD of a male fetus at 26 weeks GA. Their second pregnancy was a female baby and succumbed to Epidermolysis Bullosa. 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 likely pathogenic variant in the *ITGB4* gene (p.Ser1111ArgfsTer50) related to phenotype.

Mr. XXX is found to be carrier of likely pathogenic variant in the *APC2* gene (p. Ser311Ter).

Mr. XXX and Mrs. YYY are found to be carriers of uncertain significant variant in the *ATP7B* gene (p. Pro350Leu) and *PKD1L1* gene (p. Cys2188Trp).

List of common significant carrier variant identified related to the phenotype:

Disease	Mr. XXX	Mrs. YYY
Epidermolysis bullosa, junctional 5A, intermediate **(OMIM#619816)** Epidermolysis bullosa, junctional 5B, with pyloric atresia **(OMIM#226730)** **Mode of inheritance: AR**	**CARRIER** Gene: *ITGB4* Exon 28, c.3331_3332dupAG p. Ser1111ArgfsTer50 Heterozygous **Classification: Likely Pathogenic**	**CARRIER** Gene: *ITGB4* Exon 28, c.3331_3332dupAG p. Ser1111ArgfsTer50 Heterozygous **Classification: Likely Pathogenic**

List of significant carrier variant identified:

Disease	Mr. XXX	Mrs. YYY
Cortical dysplasia, complex, with other brain malformations 10 **(OMIM#618677)** Intellectual developmental disorder, autosomal recessive 74 **(OMIM#617169)** **Mode of inheritance: AR**	**CARRIER** Gene: *APC2* Exon 9, c.932C>A p. Ser311Ter, Heterozygous **Classification: Likely Pathogenic**	**NON - CARRIER**

List of common uncertain significant carrier variants identified:

Disease	Mr. XXX	Mrs. YYY
Wilson disease **(OMIM#277900)** **Mode of inheritance: AR**	**CARRIER** Gene: *ATP7B* Exon 2, c.1049C>T p. Pro350Leu Heterozygous **Classification: Uncertain Significance**	**CARRIER** Gene: *ATP7B* Exon 2, c.1049C>T p. Pro350Leu Heterozygous **Classification: Uncertain Significance**

Heterotaxy, visceral, 8, autosomal **(OMIM#617205)** **Mode of inheritance: AR**	**CARRIER** Gene: *PKD1L1* Exon 44, c.6564C>G p. Cys2188Trp Heterozygous **Classification: Uncertain Significance**	**CARRIER** Gene: *PKD1L1* Exon 44, c.6564C>G p. Cys2188Trp 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.

Interpretation

Interpretation for the common significant carrier variant identified related to the phenotype in Mr. XXX and Mrs. YYY

***ITGB4:* c.3331_3332dupAG**

Variant summary: A heterozygous two base pairs insertion in exon 28 of the *ITGB4* gene (chr17:g.75750124insGA, NM_000213.5, Depth: >115x) that results in a frameshift and premature truncation of the protein 50 amino acids downstream to codon 1111 (p.Ser1111ArgfsTer50) was detected. This variant is a frameshift variant which occurs in an exon of *ITGB4* 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.

***In-silico* prediction:** The *in-silico* predictions of the variant is deleterious by MutationTaster2.

OMIM phenotype: Epidermolysis bullosa, junctional 5A, intermediate (OMIM#619816) and Epidermolysis bullosa, junctional 5B, with pyloric atresia (OMIM#226730) are caused by homozygous or compound heterozygous mutation in the *ITGB4* gene (OMIM*147557). Intermediate junctional epidermolysis bullosa 5A (JEB5A) is an autosomal recessive blistering disease of skin and mucous membranes. Blistering is less severe than in severe JEB. The plane of skin cleavage is through the lamina lucida of the cutaneous basement membrane zone. Nails may be dystrophic and dental enamel defects are present. Blistering does not affect the life span of affected individuals. Junctional epidermolysis bullosa 5B with pyloric atresia (JEB5B) is an autosomal recessive blistering disease of skin and mucous membranes. Severity of skin involvement ranges from extensive full thickness skin loss (aplasia cutis congenita) to mild epidermolysis bullosa that improves with age. The plane of skin cleavage is through the lamina lucida of the cutaneous basement membrane zone. Pyloric atresia is usually evident within a few days to weeks of life. Atresia may occur at other gastrointestinal sites including the esophagus and duodenum. JEB5B is usually lethal within

the first few weeks of life despite surgical correction of pyloric atresia. Milder, non-lethal forms with less skin blistering have been reported. These diseases follow 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 variant identified in Mr. XXX

***APC2:* c.932C>A**

Variant summary: A heterozygous stop gained variation in exon 9 of the *APC2* gene (chr19:g.1456968C>A, NM_005883.3, Depth: 149x) that results in the premature truncation of the protein at codon 311 (p.Ser311Ter) was detected. This variant is a stop gained variant which occurs in an exon of *APC2* 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.

***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: Cortical dysplasia, complex, with other brain malformations 10 (OMIM#618677) is caused by homozygous or compound heterozygous mutation in the *APC2* gene (OMIM*612034). Intellectual developmental disorder, autosomal recessive 74 (OMIM#617169) is caused by homozygous mutation in the *APC2* gene (OMIM*612034). 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 carrier variant identified in Mr. XXX and Mrs. YYY

***ATP7B:* c.1049C>T**

Variant summary: A heterozygous missense variation in exon 2 of the *ATP7B* gene (chr13:g.51974171G>A, NM_000053.4, Depth: >194x) that results in the amino acid substitution of Leucine for Proline at codon 350 (p.Pro350Leu) was detected.

Population frequency: This variant has minor allele frequency of 0.002% in gnomAD database and has minor allele frequency of 0.02% in 1000 genomes databases.

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

OMIM phenotype: Wilson disease (OMIM#277900) is caused by homozygous or compound heterozygous mutation in the *ATP7B* gene (OMIM*606882). 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.

***PKD1L1:* c.6564C>G**

Variant summary: A heterozygous missense variation in exon 44 of the *PKD1L1* gene (chr7:g.47829596G>C, NM_138295.5, Depth: >108x) that results in the amino acid substitution of Tryptophan for Cysteine at codon 2188 (p.Cys2188Trp) 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 damaging by SIFT and PolyPhen-2(HumDiv).

OMIM phenotype: Heterotaxy, visceral, 8, autosomal (OMIM#617205) is caused by homozygous mutation in the *PKD1L1* gene (OMIM*609721). 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.

Additional Variant(s)

The additional variants identified may or may not be related to patient's phenotype. Phenotype – genotype correlation is recommended.

List of additional significant variants identified:

Disease	Mr. XXX	Mrs. YYY
Brain small vessel disease 2 **(OMIM#614483)** **Mode of inheritance: AD**	**NON - CARRIER**	Gene: *COL4A2* Exon 24, c.1679delG p. Gly560AspfsTer11 Heterozygous **Classification: Likely Pathogenic**

Recommendations

- The *ATP7B* gene has pseudogene in the human genome. Validation of the variant(s) by Sanger sequencing is strongly recommended to rule out false positives. Sequencing the variant(s) in the parents is recommended to confirm the significance.

- 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 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	6.59 Gb	7.49 Gb
Data ≥ Q30	96.81%	96.79%

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 [4].

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/VCV000861197.7

4. 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

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,AEB P1,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,A MPD1,AMPD2,AMPD3,AMT,ANGPTL3,ANO10,ANO5,ANTXR1,ANTXR2,AP4B1,AP4E1,AP5Z1,APOC2,APOE,APRT,AQP2,AQP7,ARFGEF2,ARHGDIA,AR HGEF6,ARL2BP,ARL6,ARL6IP1,ASL,ASNS,ASPA,ASPM,ASS1,ATCAY,ATG5,ATIC,ATM,ATP2A1,ATP2B3,ATP6AP2,ATP6V0A2,ATP6V1A,ATP6V1B1,ATP7 A,ATP8A2,ATPAF2,ATR,AURKC,AVPR2,B2M,B3GAT3,B4GALNT1,B4GALT1,B4GAT1,B9D1,B9D2,BANF1,BBIP1,BBS1,BBS10,BBS12,BBS2,BBS4,BBS5,B BS7,BCAP31,BCL10,BCS1L,BGN,BIN1,BLNK,BLOC1S3,BLVRA,BMP1,BMP2,BMPR1B,BPGM,BRAT1,BRF1,BSCL2,BSND,BTK,BVES,C12orf57,C12orf65,C 19orf12,C1QA,C1QB,C1QC,C3,C8A,C8B,C8orf37,CA12,CA2,CA5A,CA8,CABP2,CABP4,CACNA1D,CACNA1F,CACNA2D4,CAD,CANT1,CAPN1,CARD11,C ARD9,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,CDK5R AP2,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,COG 4,COG6,COL17A1,COL4A3,COL4A4,COL6A1,COL6A2,COL6A3,COL7A1,COL9A2,COLEC11,COLQ,COQ4,COQ6,COQ7,COQ9,COX10,COX14,COX20,COX4 I2,COX6A1,COX6B1,COX8A,CPA6,CPLX1,CPN1,CPT1A,CPT2,CRADD,CRAT,CRB1,CRIPT,CRTAP,CRYAB,CRYBB1,CRYBB3,CSF2RB,CSF3R,CSTA,CSTB,CTC 1,CTH,CTNNB1,CTNS,CTPS1,CTSA,CTSC,CTSD,CTSF,CTSK,CUBN,CUL7,CWF19L1,CYB5A,CYBA,CYBB,CYP11B1,CYP11B2,CYP17A1,CYP1B1,CYP24A1,CY P26C1,CYP27A1,CYP27B1,CYP2C19,CYP2D6,CYP2R1,CYP2U1,CYP4F22,CYP4V2,CYP7B1,D2HGDH,DAG1,DARS2,DBH,DBT,DCC,DCHS1,DCPS,DCTN1,D CXR,DDB2,DDC,DDHD1,DDHD2,DDOST,DDR2,DDRGK1,DDX11,DDX59,DES,DGKE,DGUOK,DHCR7,DHH,DHODH,DHTKD1,DIS3L2,DKC1,DLAT,DLEC1,D LG3,DLX5,DMD,DNA2,DNAAF1,DNAAF3,DNAH1,DNAH11,DNAI1,DNAJB13,DNAJB2,DNAJC19,DNAJC3,DNAJC6,DNAL4,DNASE1L3,DNM2,DNMT3B,D OCK2,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,ELOVL 4,ELP2,EMC1,EMD,EMG1,EMP2,ENAM,ENO3,ENTPD1,EPCAM,EPG5,EPO,EPS8,EPS8L2,EPX,ERBB3,ERCC1,ERCC2,ERCC3,ERCC4,ERCC5,ERCC6,ERLIN 1,ERLIN2,ESCO2,ESR1,ESRRB,ETFB,ETFDH,ETHE1,EVC2,EXOSC2,EXOSC3,EXOSC8,EXPH5,EXT1,EXT2,EXTL3,EYS,F12,F13A1,F13B,F2,F5,F7,F9,FA2H,FA DD,FAH,FAM126A,FAM20A,FAM20C,FAN1,FANCI,FANCL,FAR1,FARS2,FASLG,FASTKD2,FAT4,FBP1,FBXL4,FBXO7,FCGR2A,FCGR3A,FCN3,FECH,FERM T1,FERMT3,FEZF1,FGA,FGB,FGD4,FGF16,FGF20,FGFR3,FH,FHL1,FIBP,FIG4,FKBP10,FKBP14,FKRP,FKTN,FLAD1,FLI1,FLNB,FLVCR1,FLVCR2,FMO3,FOL R1,FOXI1,FOXN1,FOXP3,FOXRED1,FRAS1,FREM1,FREM2,FRMPD4,FSHB,FSHR,FTL,FTSJ1,FUCA1,FUT1,FUT8,FXN,FYCO1,FZD6,G6PC,G6PC3,GAA,GA B1,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,G NMT,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,H 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1,LPL,LRAT,LRBA,LRIG2,LRIT3,LRP2,LRPAP1,LRPPRC,LRRC6,LRSAM1,LRTOMT,LSS,LTBP2,LZTFL1,MAB21L2,MAD2L2,MAG,MAMLD1,MAN1B1,MAN2 B1,MAOA,MAP3K8,MAPKBP1,MAPT,MARS2,MASP1,MAT1A,MBOAT7,MBTPS2,MC1R,MC2R,MC4R,MCCC1,MCEE,MCM5,MCM9,MECR,MED12,ME D25,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,MR PL44,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,NDR G1,NDST1,NDUFA1,NDUFA10,NDUFA11,NDUFA12,NDUFA13,NDUFA2,NDUFA9,NDUFAF1,NDUFAF2,NDUFAF3,NDUFAF4,NDUFAF5,NDUFAF6,NDU FB11,NDUFB3,NDUFB9,NDUFS1,NDUFS2,NDUFS6,NDUFS8,NDUFV1,NDUFV2,NECAP1,NEFL,NEK8,NEK9,NEU1,NEUROG3,NGF,NGLY1,NHLRC1,NHP 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