# Whole Exome Sequencing Analysis – Carrier Screening

| | | |
|---|---|---|
| **Patient name** | **: Mr. XXX** | **Mrs. YYY** |
| **Gender/ Age** | **: Male/30 Years** | **Female/26 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 a non-consanguineous couple. Mrs. YYY has multiple neurofibromas and café au lait spots. She is suspected to be affected with Neurofibromatosis. Mr. XXX's semen analysis was indicative of Severe Oligoasthenoteratozoospermia. The couple has Primary Infertility and history of two failed IUI cycles with self-gamete. Mrs. YYY's mother and two brothers have neurofibromas, father and paternal grandfather succumbed to kidney disease. Mr. XXX's mother has congenital hearing loss and mutism. Mr. XXX and Mrs. YYY have been evaluated for pathogenic variations.

## Results

Mrs. YYY is found to be affected with likely pathogenic variant in *NF1* gene (p. Lys615=), found to be a carrier of likely pathogenic variant *CHRND* gene (p. Glu402Lys), *MCCC2* gene (c.1149+1G>A) and found to be a carrier of uncertain significance variant in *SYP* gene (p. Leu233Gln) and *PLP1* gene (p.Ser77Phe).

Mr. XXX is found to be a carrier of likely pathogenic variant in *CRB1* gene (p. Trp906Ter) and *ALOX12B* gene (p. Pro353Thr).

**List of significant variant identified related to the phenotype:**

| Disease | Mr. XXX | Mrs. YYY |
|---|---|---|
| Neurofibromatosis, familial spinal<br>**(OMIM#162210)**<br><br>Neurofibromatosis, type 1<br>**(OMIM#162200)**<br><br>Neurofibromatosis-Noonan syndrome<br>**(OMIM#601321)**<br><br>Watson syndrome<br>**(OMIM#193520)**<br><br>**Mode of inheritance: AD** | **NON - CARRIER** | **AFFECTED**<br>Gene: **#***NF1*<br>Exon 16, c.1845G>A,<br>p. Lys615=,<br>Heterozygous<br><br>**Classification: Pathogenic** |

**# Sanger sequencing is recommended for the *NF1* variant to rule out false positives.**

**List of additional carrier variants identified:**

| Disease | Mr. XXX | Mrs. YYY |
|---|---|---|
| Multiple pterygium syndrome, lethal type<br>**(OMIM#253290)**<br><br>Myasthenic syndrome, congenital, 3B, fast-channel<br>**(OMIM#616322)**<br><br>**Mode of inheritance: AR** | **NON - CARRIER** | **CARRIER**<br>Gene: *CHRND*<br>Exon 10, c.1204G>A,<br>p. Glu402Lys,<br>Heterozygous<br><br>**Classification: Likely pathogenic** |
| 3-Methylcrotonyl-CoA carboxylase 2 deficiency<br>**(OMIM#210210)**<br><br>**Mode of inheritance: AR** | **NON - CARRIER** | **CARRIER**<br>Gene: *MCCC2*<br>Intron 12, c.1149+1G>A,<br>5' splice site,<br>Heterozygous<br><br>**Classification: Likely pathogenic** |

| | | |
|---|---|---|
| Intellectual developmental disorder, X-linked 96<br>**(OMIM#300802)**<br><br>**Mode of inheritance: XLR** | **NON - CARRIER** | **CARRIER**<br>Gene: *SYP*<br>Exon 6, c.698T>A,<br>p. Leu233Gln,<br>Heterozygous<br><br>**Classification: Uncertain significance** |
| Pelizaeus-Merzbacher disease<br>**(OMIM#312080)**<br><br>Spastic paraplegia 2, X-linked<br>**(OMIM#312920)**<br><br>**Mode of inheritance: XLR** | **NON - CARRIER** | **CARRIER**<br>Gene: *PLP1*<br>Exon 3, c.230C>T,<br>p. Ser77Phe,<br>Heterozygous<br><br>**Classification: Uncertain significance** |
| Leber Congenital Amaurosis 8<br>**(OMIM#613835)**<br><br>Retinitis pigmentosa-12<br>**(OMIM#600105)**<br><br>**Mode of inheritance: AR** | **CARRIER**<br>Gene: *CRB1*<br>Exon 8, c.2718G>A,<br>p. Trp906Ter,<br>Heterozygous<br><br>**Classification: Likely pathogenic** | **NON - CARRIER** |
| Ichthyosis, congenital, autosomal recessive 2<br>**(OMIM#242100)**<br><br>**Mode of inheritance: AR** | **CARRIER**<br>Gene: *ALOX12B*<br>Exon 8, c.1057C>A,<br>p. Pro353Thr,<br>Heterozygous<br><br>**Classification: Likely pathogenic** | **NON - CARRIER** |

*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 Mrs. YYY**

***NF1*: c.1845G>A**

**Variant summary:** A heterozygous synonymous variation in exon 16 of the *NF1* gene (chr17:g.31223567G>A, NM_001042492.3, Depth: 19x) that results in the synonymous substitution of amino acid Lysine at codon 615 (p. Lys615=) was detected.

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

**Clinical and Literature evidence** : This variant has been classified as pathogenic in ClinVar database [3]. This gene has been previously reported in a patient affected with neurofibromatosis type 1 [4].

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

**OMIM phenotype:** Neurofibromatosis, familial spinal (OMIM#162210), Neurofibromatosis, type 1 (OMIM#162200), Neurofibromatosis-Noonan syndrome (OMIM#601321) and Watson syndrome (OMIM#193520) are caused by heterozygous mutation in the *NF1* gene (OMIM* 613113). This disorder is characterized by cafe-au-lait spots, Lisch nodules in the eye, and fibromatous tumors of the skin. Individuals with the disorder have increased susceptibility to the development of benign and malignant tumors. 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.

***CHRND*: c.1204G>A**

**Variant summary:** A heterozygous missense variation in exon 10 of the *CHRND* gene (chr2:g.232534087G>A, NM_000751.3, Depth: 147x) that results in the amino acid substitution of Lysine for Glutamic acid at codon 402 (p. Glu402Lys) was detected.

**Population frequency**: This variant has a minor allele frequency of 0.0033 % in gnomAD database and has not been reported in 1000 genomes database.

**Clinical 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 (HumDiv), LRT and MutationTaster2. The reference codon is conserved across mammals in PhyloP and GERP++ tool.

**OMIM phenotype:** Multiple pterygium syndrome, lethal type (OMIM#253290) and Myasthenic syndrome, congenital, 3B, fast-channel (OMIM#616322) is caused by homozygous or compound heterozygous mutation in the *CHRND* gene (OMIM*100720). These diseases follows autosomal recessive pattern of inheritance [2].

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

***MCCC2*: c.1149+1G>A**

**Variant summary:** A heterozygous 5' splice site variation in intron 12 of the *MCCC2* gene (chr5:g.71643896G>A, NM_022132.5, Depth: 44x) that affects the invariant GT donor splice site of exon 12 (c.1149+1G>A) was detected. This variant results in the loss of an donor splice site for the clinically relevant transcript. The c.1149+1G>A variant is a loss of function variant in the gene *MCCC2*, which is intolerant of Loss of Function variants.

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

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

***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:** 3-Methylcrotonyl-CoA carboxylase 2 deficiency (OMIM#210210) is caused by homozygous or compound heterozygous mutation in the *MCCC2* gene (OMIM*609014). This disease follows autosomal recessive pattern of inheritance [2].

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

**Interpretation for the uncertain significant variants identified in Mrs. YYY**

***SYP*: c.698T>A**

**Variant summary:** A heterozygous missense variation in exon 6 of the *SYP* gene (chrX:g.49191681A>T, NM_003179.3, Depth: 165x) that results in the amino acid substitution of Glutamine for Leucine at codon 233 (p. Leu233Gln) 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). The reference codon is conserved across mammals in PhyloP and GERP++tool.

**OMIM phenotype:** Intellectual developmental disorder, X-linked 96 (OMIM#300802) is caused by mutation in the *SYP* gene (OMIM*313475). This disease follows X-linked recessive pattern of inheritance [2].

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

***PLP1*: c.230C>T**

**Variant summary:** A heterozygous missense variation in exon 3 of the *PLP1* gene (chrX:g.103786503C>T, NM_000533.5, Depth: 108x) that results in the amino acid substitution of Phenylalanine for Serine at codon 77 (p. Ser77Phe) 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, Polyphen-2 (HumDiv) and LRT. The reference codon is conserved across mammals in PhyloP and GERP++ tool.

**OMIM phenotype:** Pelizaeus-Merzbacher disease (OMIM#312080) and Spastic paraplegia 2, X-linked (OMIM#312920) are caused by mutation in the *PLP1* gene (OMIM*300401). These diseases follow X-linked recessive pattern of inheritance [2].

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

**Interpretation for the significant variants identified in Mr. XXX**

***CRB1*: c.2718G>A**

**Variant summary:** A heterozygous stop-gained variation in exon 8 of the *CRB1* gene (chr1:g.197429490G>A, NM_201253.3, Depth: 90x) that results in the premature truncation of the protein at codon 906 (p. Trp906Ter) was detected. This variant is predicted to cause loss of normal protein function through protein truncation. This variant is a stop gained variant which occurs in an exon of *CRB1* upstream of where nonsense mediated decay is predicted to occur.

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

**Clinical evidence** : This variant has been classified as pathogenic in ClinVar database [7].

***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:** Leber congenital amaurosis 8 (OMIM#613835) and Retinitis pigmentosa-12 (OMIM#600105) are caused by homozygous or compound heterozygous mutations in the *CRB1* gene (OMIM*604210). This disease follows autosomal recessive pattern of inheritance [2].

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

***ALOX12B*: c.1057C>A**

**Variant summary:** A heterozygous missense variation in exon 8 of the *ALOX12B* gene (chr17:g.8079410G>T, NM_001139.3, Depth: 84x) that results in the amino acid substitution of Threonine for Proline at codon 353 (p. Pro353Thr) was detected.

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

**Clinical evidence** : This variant has been classified as pathogenic in ClinVar database [8].

***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++ tool.

**OMIM phenotype:** Ichthyosis, congenital, autosomal recessive 2 (OMIM#242100) is caused by homozygous or compound heterozygous mutation in the *ALOX12B* gene (OMIM* 603741). This disease follows autosomal recessive pattern of inheritance [2].

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

## Recommendations

- Sanger sequencing is recommended for the *NF1* variant to rule out false positives.
- The *NF1* 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 and the other affected and unaffected members of the family is recommended to confirm the significance.
- Alternative test is strongly recommended to rule out the deletion/duplication.
- Genetic counseling is recommended.

## Methodology

DNA extracted from the blood was used to perform whole exome using a whole exome capture kit. The targeted libraries were sequenced to a targeted depth of 80 to 100X using GenoLab M sequencing platform. This kit has deep exonic coverage of all the coding regions including the difficult to cover regions. The sequences obtained are aligned to human reference genome (GRCh38.p13) using Sentieon aligner and analyzed using Sentieon for removing duplicates, recalibration and re-alignment of indels.

Sentieon 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

| | Mr. XXX | Mrs. YYY |
|---|---|---|
| Total reads generated | 10.41 Gb | 10.05 Gb |
| Data ≥ Q30 | 93.61% | 93.38% |

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.
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/VCV000578879.8
4. Bianchessi, Donatella, et al. "126 novel mutations in Italian patients with neurofibromatosis type 1." Molecular genetics & genomic medicine 3.6 (2015): 513-525. PMID: 26740943.
5. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000189817.7
6. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV001067700.10
7. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV001070228.3
8. https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000995734.2
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.


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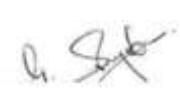

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Director

## 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,*

*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*