

Whole Exome Sequencing Analysis – Carrier Screening

Patient name	: Mr. XY	Mrs. XX
Gender/ Age	: Male/37 years	Female/28 years
PIN	: XXXXX	XXXXX
Sample no	: XXX	XXX
Specimen	: Peripheral blood	Peripheral blood
Sample collection date	: XX	XX
Sample receipt date	: XX	XX
Report date	: 08-03-2025	08-03-2025
Referring clinician	: XXX	
Hospital/Clinic	: XXXX	

Clinical history

Mr. XY and Mrs. XX are third degree consanguineous couple with bad obstetric history. Their first pregnancy was a missed abortion. Their second pregnancy conceived by ART conception with self-gametes. Fetal Echocardiography at 28 weeks 1 day GA indicative of mild cardiomegaly, biventricular chamber dilatation, biventricular dysfunction, functional tricuspid and pulmonary regurgitation and dilated cardiomyopathy. FTLSCS - male baby with delayed cry and succumbed to illness at 8 hours of birth. His Whole Mitochondrial Genome Sequencing indicative of no pathogenic or likely pathogenic variants. Clinical Exome Sequencing revealed a homozygous pathogenic variant c.2947C>T in *ALPK3* gene causative of familial hypertrophic cardiomyopathy – 27. Mr. XY and Mrs. XX have been evaluated for pathogenic variations.

Results

Mr. XY and Mrs. XX are found to be carriers of common likely pathogenic variants in *ALPK3* gene (p. Gln983Ter) and *ADAMTS13* gene (p. Val88Met).

Mr. XY is found to be a carrier of likely pathogenic variants in *GP6* gene (p. Trp160Ter), *NPR3* gene (c.769+1G>C) and found to have uncertain significance variant in *SZT2* gene (p. Ser1812Gly).

Mrs. XX is found to be a carrier of likely pathogenic variants in *ANO5* gene (p. Phe578Ser) and found to have uncertain significance variant in *SZT2* gene (p. Glu1755Lys).

Mr. XY and Mrs. XX are found to be carriers of common uncertain significance variant in *DNAH17* gene (p. Glu2382Gln).

List of common significant carrier variant identified to related phenotype:

Disease	Mr. XY	Mrs. XX
Cardiomyopathy, familial hypertrophic 27 (OMIM#618052)	CARRIER Gene: <i>ALPK3</i> Exon 6, c.2947C>T p. Gln983Ter Heterozygous	CARRIER Gene: <i>ALPK3</i> Exon 6, c.2947C>T p. Gln983Ter Heterozygous
Mode of inheritance: AR	Classification: Likely Pathogenic	Classification: Likely Pathogenic

List of significant carrier variants identified:

Disease	Mr. XY	Mrs. XX
Thrombotic thrombocytopenic purpura, hereditary (OMIM#274150)	CARRIER Gene: <i>ADAMTS13</i> Exon 3, c.262G>A p. Val88Met Heterozygous	CARRIER Gene: <i>ADAMTS13</i> Exon 3, c.262G>A p. Val88Met Heterozygous
Mode of inheritance: AR	Classification: Likely Pathogenic	Classification: Likely Pathogenic
Bleeding disorder, platelet-type, 11 (OMIM#614201)	CARRIER Gene: <i>GP6</i> Exon 4, c.480G>A p. Trp160Ter Heterozygous	NON - CARRIER
Mode of inheritance: AR	Classification: Likely Pathogenic	
Boudin-Mortier syndrome (OMIM#619543)	CARRIER Gene: <i>NPR3</i> Intron 1, c.769+1G>C 5' Splice site, Heterozygous	NON - CARRIER
Mode of inheritance: AR	Classification: Likely Pathogenic	

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Gnathodiaphyseal dysplasia (OMIM#166260) Mode of inheritance: AD Miyoshi muscular dystrophy 3 (OMIM#613319) Muscular dystrophy, limb-girdle, autosomal recessive 12 (OMIM#611307) Mode of inheritance: AR	NON - CARRIER	CARRIER Gene: <i>ANO5</i> Exon 16, c.1733T>C p. Phe578Ser, Heterozygous Classification: Likely Pathogenic
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List of uncertain significant carrier variants identified:

Disease	Mr. XY	Mrs. XX
Spermatogenic failure 39 (OMIM#618643)	CARRIER Gene: <i>DNAH17</i> Exon 46, c.7144G>C p. Glu2382Gln Heterozygous Classification: Uncertain Significance	CARRIER Gene: <i>DNAH17</i> Exon 46, c.7144G>C p. Glu2382Gln Heterozygous Classification: Uncertain Significance
Developmental and epileptic encephalopathy 18 (OMIM#615476) Mode of inheritance: AR	CARRIER Gene: <i>SZT2</i> Exon 48, c.6769A>G p. Ser2257Gly Heterozygous Classification: Uncertain Significance	CARRIER Gene: <i>SZT2</i> Exon 37, c.5434G>A p. Glu1812Lys Heterozygous Classification: 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 uncertain significant variants identified:

Disease	Mr. XY	Mrs. XX
Spherocytosis, type 2 (OMIM#616649)	NON - CARRIER	CARRIER Gene: <i>SPTB</i> Exon 14, c.2407G>T p. Glu803Ter, Heterozygous
Mode of inheritance: AD		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 common significant variant identified to related phenotype in Mr. XY and Mrs. XX

***ALPK3*: c.2947C>T**

Variant summary: A heterozygous stop gained variation in exon 6 of the *ALPK3* gene (chr15:g.84857685C>T, NM_020778.5, Depth: >129) that results in the premature truncation of the protein at codon 983 (p.Gln983Ter) was detected. This variant is a stop gained variant which occurs in an exon of *ALPK3* upstream of where nonsense mediated decay is predicted to occur. This variant is predicted to cause loss of normal protein function through protein truncation.

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

In-silico prediction: The *in-silico* predictions of the variant are deleterious by CADD. The reference codon is conserved across GERP++ tool.

OMIM phenotype: Cardiomyopathy, familial hypertrophic 27 (OMIM#618052) is caused by homozygous mutation in the *ALPK3* gene (OMIM*617608). 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 common significant variant identified in Mr. XY and Mrs. XX

ADAMTS13: c.262G>A

Variant summary: A heterozygous missense variation in exon 3 of the **ADAMTS13** gene (chr9:g.133424410G>A, NM_139027.6, Depth: 121x) that results in the amino acid substitution of Methionine for Valine at codon 88 (p.Val88Met) was detected.

Population frequency: This variant has minor allele frequency of 0.0013% in gnomAD database and has not been reported in 1000 genomes database.

Clinical and Literature evidence: This variant has been previously classified as variant of uncertain significance in ClinVar database [3]. This missense variant with the same amino acid position has been previously reported in patients affected with Familial Thrombotic Thrombocytopenic Purpura in compound heterozygous state [4].

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

OMIM phenotype: Thrombotic thrombocytopenic purpura, hereditary (OMIM#274150) is caused by homozygous or compound heterozygous mutation in the **ADAMTS13** gene (OMIM*604134). This disease follows autosomal recessive pattern of inheritance [2].

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

Interpretation for the significant carrier variant identified in Mr. XY

GP6: c.480G>A

Variant summary: A heterozygous stop gained variation in exon 4 of the **GP6** gene (chr19:g.55027708C>T, NM_001083899.2, Depth: 142x) that results in the premature truncation of the protein at codon 160(p.Trp160Ter) was detected. This variant is a stop gained variant which occurs in an exon of **GP6** upstream of where nonsense mediated decay is predicted to occur. This variant is predicted to cause loss of normal protein function through protein truncation.

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

In-silico prediction: The *in-silico* predictions of the variant are deleterious by CADD.

OMIM phenotype: Bleeding disorder, platelet-type, 11 (OMIM#614201) can be caused by compound heterozygous mutation in the **GP6** gene (OMIM*605546). This disease follows autosomal recessive pattern of inheritance [2].

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

NPR3: c.769+1G>C

Variant summary: A heterozygous 5' splice site variation in intron 1 of the *NPR3* gene (chr5:g.32712546G>C, NM_001204375.2, Depth: 44x) that affects the invariant GT donor splice site of exon 1 (c.769+1G>C) was detected.

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

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

OMIM phenotype: Boudin-Mortier syndrome (OMIM#619543) is caused by homozygous or compound heterozygous mutation in the *NPR3* gene (OMIM*108962). This disease follows autosomal recessive pattern of inheritance [2].

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

Interpretation for the significant carrier variant identified in Mrs. XX

ANOS: c.1733T>C

Variant summary: A heterozygous missense variation in exon 16 of the *ANOS* gene (chr11:g.22262231T>C, NM_213599.3, Depth: 67x) that results in the amino acid substitution of Serine for Phenylalanine at codon 578 (p.Phe578Ser) was detected.

Population frequency: This variant has minor allele frequency of 0.0105% in gnomAD database and has minor allele frequency of 0.020% in 1000 genomes database.

Clinical and Literature evidence: This variant has been previously classified as pathogenic in ClinVar database [5]. This variant has been previously reported in patients affected with limb girdle muscular dystrophy in compound heterozygous state [6].

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

OMIM phenotype: Gnathodiaphyseal dysplasia (OMIM#166260) is caused by heterozygous mutation in the *ANOS* gene (OMIM*608662). This disease follows autosomal dominant pattern of inheritance [2]. Miyoshi muscular dystrophy 3 (OMIM#613319), Muscular dystrophy, limb-girdle, autosomal recessive 12 (OMIM#611307) are caused by homozygous or compound heterozygous mutation in the *ANOS* gene (OMIM*608662). 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. XY and Mrs. XX***DNAH17: c.7144G>C***

Variant summary: A heterozygous missense variation in exon 46 of the *DNAH17* gene (chr17:g.78486091C>G, NM_173628.4, Depth: >96x) that results in the amino acid substitution of Glutamine for Glutamic acid at codon 2382 (p. Glu2382Gln) was detected.

Population frequency: This variant has minor allele frequency of 0.008% in gnomAD database and has a minor allele frequency of 0.02% in 1000 genomes database.

Clinical evidence: This variant has been previously classified as variant of uncertain significance in ClinVar database [7].

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: Spermatogenic failure 39 (OMIM#618643) is caused by homozygous or compound heterozygous mutation in the *DNAH17* gene (OMIM*610063). 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 carrier variant identified in Mr. XY***SZT2: c.6769A>G***

Variant summary: A heterozygous missense variation in exon 48 of the *SZT2* gene (chr1:g.43439070A>G, NM_001365999.1, Depth: 80x) that results in the amino acid substitution of Serine for Glycine at codon 2257 (p.Ser2257Gly) was detected.

Population frequency: This variant has minor allele frequency of 0.0026% in gnomAD database and has not been reported in 1000 genomes database.

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

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

OMIM phenotype: Developmental and epileptic encephalopathy 18 (OMIM#615476) is caused by homozygous or compound heterozygous mutation in the *SZT2* gene (OMIM*615463). 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 carrier variant identified in Mrs. XX

SZT2: c.5434G>A

Variant summary: A heterozygous missense variation in exon 37 of the *SZT2* gene (chr1:g.43432431G>A, NM_001365999.1, Depth: 164x) that results in the amino acid substitution of Lysine for Glutamic acid at codon 1812(p.Glu1812Lys) was detected.

Population frequency: This variant has minor allele frequency of 0.0013% in gnomAD database and has not been reported in 1000 genomes database.

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

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

OMIM phenotype: Developmental and epileptic encephalopathy 18 (OMIM#615476) is caused by homozygous or compound heterozygous mutation in the *SZT2* gene (OMIM*615463). 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 uncertain significant variants identified:

Disease	Mr. XY	Mrs. XX
Spherocytosis, type 2 (OMIM#616649)	NON - CARRIER	CARRIER Gene: <i>SPTB</i> Exon 14, c.2407G>T p. Glu803Ter, Heterozygous
Mode of inheritance: AD		Classification: 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 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. SentieonDNAScope 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. XY	Mrs. XX
Total reads generated	8.33Gb	10.68Gb
Data ≥ Q30	96.14%	96.24%

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

1. Richards, S, et al. Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in medicine: official journal of the American College of Medical Genetics. 17.5 (2015): 405-424.
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3. <https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000068812.6>
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6. Hicks D, et al. A founder mutation in Anoctamin 5 is a major cause of limb-girdle muscular dystrophy. Brain. 2011 Jan;134(Pt 1):171-182. doi: 10.1093/brain/awq294. PMID: 21186264; PMCID: PMC4038512.
7. <https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV002401284.2>
8. <https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV000940206.6>
9. <https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV001517124.5>
10. Miller, David T., et al. "ACMG SF v3. 1 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the Americans College of Medical Genetics and Genomics (ACMG)." Genetics in Medicine 24.7 (2022): 1407-1414.

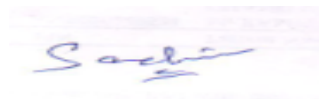
This report has been reviewed and approved by:



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:

AARS2, ABAT, ABCA1, ABCA12, ABCA3, ABCA4, ABCC4, 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, AH11, AHSG, AICDA, AIFM1, AIP1, 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, APSZ1, APOC2, APOE, APR1, AQP2, AQP7, ARFGF2, ARHGDI, ARHGFE6, 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, CDF6, CDH11, CDH3, CDHR1, CDK10, CDK5, CDK5R, ADK, CDK6, CDSN, CEBPE, CENPF, CENPJ, CEP104, CEP152, CEP164, CEP19, CEP41, CEP57, CERS3, CFH, CFL2, CFC, CFTR, CHAT, CHIT1, CHKB, CHMP1A, CHRD1, CHRM3, CHRNA1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHST8, CIB2, CIB5, CIDEF, 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, COA5Y, COG4, COG6, COL17A1, COL4A3, COL4A4, 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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, FAR52, FASLG, FASTKD2, FAT4, FBP1, FBXL4, FBXO7, FCGR2A, FCGR3A, FCN3, FECH, FERMT1, FERMT3, FEZF1, FGA, FGB, FGD4, FGF16, FGF20, FGFTR3, FH, FHL1, FIBP, FIG4, FKBP10, FKBP14, FKRP, FUT1, FLAD1, FYI1, FLNB, FLVCR1, FLVCR2, FMO3, FOLR1, FOXI3, FOXF1, FOGX3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FSHB, FSHR, FTL, FTSJ1, FUCA1, FUTN, FLAT8, FYL18, FXN, FYL1C1, FZD6, G6PC, G6PC3, GAA, GAB1, GAD1, GALE, GALK1, GALNT3, GALT, GAMT, GAN, GATA1, GATA5, GBA2, GBE1, GCLC, GCNT2, GDAP1, GDF5, GDF6, GDF9, GEMIN4, GF1B, GFM1, GPT1, GGCX, GH1, GHRL, GHSR, 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