

Whole Exome Sequencing Analysis

Patient name	: Baby. XY	PIN	: XXX
Gender/ Age	: Male/ 8 Months	Sample number	: XXX
Hospital/ Clinic	: XXXX	Sample collection date	: 01-02-2025
Hospital/ Clinic	: XXXX	Sample received date	: 01-02-2025
Specimen	: Peripheral Blood	Report date	: 06-05-2025

Clinical history

Proband, Baby. XY is presented with chief complaints of jerky movements since 8 months of age and infantile spasms. His EEG indicative of frequent bilateral generalized spikes and waves. His MRI indicative of gliosis with blooming artifact involving the right frontotemporal region, with ex vacuo dilatation of the right lateral ventricle and gliosis in the left periventricular white matter region. Proband, Baby. XY is suspected to be affected with inborn errors of metabolism and has been evaluated for pathogenic variations.

Results

No pathogenic or likely pathogenic variant causative of the reported phenotype was detected

List of uncertain significant variant identified:

Gene	Region	Variant*	Allele Status	Disease	Classification*	Inheritance pattern
SERPINI1 (+)	Exon 4	c.529T>C (p.Tyr177His)	Heterozygous	Encephalopathy, familial, with neuroserpin inclusion bodies (OMIM#604218)	Uncertain Significance (PM2)	Autosomal Dominant

*Genetic test results are 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.

Whole Mitochondrial Genome Sequencing

No pathogenic or likely pathogenic variant causative of the reported phenotype was detected

CNV Findings

No significant Copy Number Variations (CNV) related to phenotype was detected.

Interpretation

SERPINI1: c.529T>C

Variant summary: A heterozygous missense variation in exon 4 of the *SERPINI1* gene (chr3:g.167792637T>C, NM_001122752.2, Depth: 172x) that results in the amino acid substitution of Histidine for Tyrosine at codon 177 (p.Tyr177His) was detected.

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

Clinical evidence: A missense variant in the same amino acid position (c.530A>C; p.Tyr177Ser) has been classified as uncertain significance in ClinVar database [3].

OMIM phenotype: Encephalopathy, familial, with neuroserpin inclusion bodies (OMIM#604218) is caused by heterozygous mutation in the *SERPINI1* gene (OMIM*602445). Familial encephalopathy with neuroserpin inclusion bodies (FENIB) is an autosomal dominant disorder characterized by progressive epilepsy and dementia. Onset of symptoms ranges from the second to fifth decades of life. Severity is variable. This disease follows autosomal dominant pattern of inheritance [2].

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

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 counselling is advised.

Methodology

DNA extracted from the blood was used to perform whole exome using 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 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 (v3.1,2.1.1), 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

Total reads generated	10.34 Gb
Data ≥ Q30	95.62%

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 are not assessed using this assay.
- This assay has a sensitivity of 70-75% in detecting large deletions/duplications of more than 10 base pairs or copy number variations (CNV). However, it is important to note that any CNVs detected must be confirmed using an alternate method.
- 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.

- 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.
3. <https://www.ncbi.nlm.nih.gov/clinvar/variation/VCV002009830.2>
4. 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 American College of Medical Genetics and Genomics (ACMG)." Genetics in Medicine 24.7 (2022): 1407-1414.

This report has been reviewed and approved by:



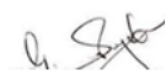
Sivasankar.S, Ph.D
Molecular Biologist



Muthukumaran. S, Ph.D
Clinical Bioinformatician



Sachin. D.Honguntikar, Ph.D,
Molecular Geneticist



Dr. G. Suriyakumar
Director

Appendix I: Gene Coverage

Indication Based Analysis:

Gene_Name	Percentage of coding region covered	Gene_Name	Percentage of coding region covered	Gene_Name	Percentage of coding region covered	Gene_Name	Percentage of coding region covered
AAAS	100.0	AARS1	100.0	AARS2	100.0	AASS	100.0
ABCA1	100.0	ABCA2	100.0	ABCA7	100.0	ABCB7	100.0
ABCC8	100.0	ABCD1	100.0	ABHD12	100.0	ABHD5	100.0
ACAD8	100.0	ACADM	100.0	ACADS	100.0	ACADSB	100.0
ACADVL	100.0	ACAT1	100.0	ACAT2	100.0	ACBD5	100.0
ACO2	100.0	ACOX1	100.0	ACOX2	100.0	ACP5	100.0
ACTB	100.0	ACTL6B	100.0	ADA2	100.0	ADAM22	100.0
ADAMTS13	100.0	ADAR	100.0	ADCY5	99.93	ADCY6	100.0
ADD3	100.0	ADGRG1	100.0	ADGRV1	100.0	ADH1C	100.0
ADNP-AS1	100.0	ADPRS	100.0	ADSL	100.0	AFG3L2	100.0
AGA	100.0	AGTPBP1	100.0	AHCY	100.0	AHI1	100.0
AIFM1	100.0	AIMP1	100.0	ALAD	100.0	ALDH18A1	100.0
ALDH3A2	100.0	ALDH5A1	100.0	ALDH6A1	100.0	ALDH7A1	100.0
ALG13	100.0	ALG6	100.0	ALS2	100.0	AMACR	100.0
AMPD2	100.0	ANG	100.0	ANK3	100.0	ANKLE2	100.0
ANO10	100.0	ANO3	100.0	ANOS1	100.0	AP1S2	99.52
AP3B2	100.0	AP3D1	100.0	AP4B1	100.0	AP4E1	100.0
AP4M1	100.0	AP4S1	100.0	AP5Z1	100.0	APOB	100.0
APP	100.0	APTX	100.0	ARCN1	100.0	ARFGEF1	100.0
ARFGEF2	100.0	ARG1	100.0	ARHGEF15	100.0	ARHGEF9	100.0
ARL13B	100.0	ARL6	100.0	ARL6IP1	100.0	ARMC9	100.0
ARNT2	100.0	ARSA	100.0	ARV1	100.0	ARX	99.16
ASAH1	100.0	ASL	100.0	ASNS	100.0	ASS1	100.0
ATCAY	100.0	ATG5	100.0	ATL1	100.0	ATM	100.0
ATP13A2	100.0	ATP1A2	100.0	ATP1A3	100.0	ATP2B3	100.0
ATP2B4	100.0	ATP5F1E	100.0	ATP6AP2	100.0	ATP7A	100.0
ATP7B	100.0	ATP8A2	100.0	ATPAF2	100.0	ATRX	100.0
ATXN7	100.0	AUH	100.0	B3GALT6	92.53	B4GALNT1	100.0
B4GAT1	100.0	B9D1	100.0	BBS1	100.0	BBS10	100.0
BBS12	100.0	BBS2	100.0	BBS4	100.0	BBS5	100.0
BBS7	100.0	BBS9	100.0	BCAP31	100.0	BCKDHA	100.0
BCKDHB	100.0	BCL11B	100.0	BCOR	100.0	BCS1L	100.0
BICD2	100.0	BOLA3	100.0	BRAT1	100.0	BSCL2	100.0
BTD	100.0	C12orf4	100.0	C19orf12	100.0	C6orf223	100.0
CA8	100.0	CACNA1A	100.0	CACNA1B	99.96	CACNA1D	100.0
CACNA1G	100.0	CACNA1H	100.0	CACNA2D2	100.0	CACNB4	100.0
CAD	100.0	CAMTA1	100.0	CAPN1	100.0	CARS2	100.0
CASK	100.0	CASR	100.0	CATIP-AS2	100.0	CAV1	100.0
CBS	17.7	CC2D2A	100.0	CCDC28B	100.0	CCDC88C	100.0
CCT5	100.0	CDKL5	100.0	CEP104	100.0	CEP120	100.0
CEP152	100.0	CEP290	100.0	CEP41	100.0	CERS1	99.38

CFH	100.0	CHAMP1	100.0	CHCHD10	100.0	CHD2	100.0
CHMP1A	99.69	CHMP2B	100.0	CHP1	100.0	CHRNA2	100.0
CHRNA4	100.0	CHRNA7	84.72	CHRNA2	100.0	CHROMR	100.0
CIT	100.0	CIZ1	100.0	CKAP2L	100.0	CLCN2	100.0
CLCN4	100.0	CLIC2	100.0	CLN3	100.0	CLN5	100.0
CLN6	100.0	CLN8	100.0	CLP1	100.0	CLPB	100.0
CLPP	100.0	CLTC	100.0	CNPY3	100.0	CNTN2	100.0
CNTNAP1	100.0	CNTNAP2	100.0	COA3	100.0	COA7	100.0
COA8	100.0	COASY	100.0	COG2	100.0	COG4	100.0
COG5	100.0	COL18A1	100.0	COL4A1	100.0	COL4A2	100.0
COL6A3	100.0	COQ2	100.0	COQ4	100.0	COQ5	100.0
COQ6	100.0	COQ7	100.0	COQ8A	100.0	COQ9	100.0
COX10	100.0	COX14	100.0	COX15	100.0	COX20	100.0
COX6B1	100.0	COX8A	100.0	CP	100.0	CPLANE1	100.0
CPLX1	100.0	CPS1	100.0	CPT1A	100.0	CPT1C	100.0
CPT2	100.0	CREBBP	100.0	CRLF1	100.0	CRPPA	88.62
CSF1R	100.0	CSPP1	100.0	CSTB	100.0	CTBP1	100.0
CTC1	100.0	CTDP1	100.0	CTNNB1	100.0	CTSD	100.0
CTSF	100.0	CUL4B	100.0	CUX2	100.0	CWF19L1	100.0
CYB5R3	100.0	CYC1	100.0	CYFIP2	100.0	CYP27A1	100.0
CYP2U1	100.0	CYP7B1	100.0	DAB1	100.0	DARS1	100.0
DARS2	100.0	DBT	100.0	DCAF17	100.0	DCC	100.0
DCPS	100.0	DCTN1	100.0	DCX	100.0	DDC	100.0
DDHD1	100.0	DDHD2	100.0	DDX3X	100.0	DEAF1	100.0
DECR1	100.0	DENND5A	100.0	DEPDC5	100.0	DHCR24	100.0
DHCR7	100.0	DHFR	100.0	DHPS	100.0	DKC1	100.0
DLAT	100.0	DLD	100.0	DLG3	100.0	DMXL2	100.0
DNA2	100.0	DNAJB2	100.0	DNAJC12	100.0	DNAJC13	100.0
DNAJC19	100.0	DNAJC3	100.0	DNAJC5	98.91	DNAJC6	100.0
DNM1	100.0	DNM1L	100.0	DNM2	100.0	DNMT1	100.0
DOCK3	100.0	DOCK7	100.0	DOCK8	100.0	DPM1	100.0
DPYD	100.0	DPYS	100.0	DRD3	100.0	DRD5	100.0
DYNC1H1	100.0	DYRK1A	100.0	EARS2	100.0	EBF3	100.0
ECHS1	100.0	EEF1A2	100.0	EEF2	100.0	EFHC1	97.5
EGR2	100.0	EIF2B1	100.0	EIF2B2	100.0	EIF2B3	100.0
EIF2B4	100.0	EIF2B5	100.0	EIF4G1	100.0	ELOVL1	100.0
ELOVL4	100.0	ELOVL5	100.0	ELP2	100.0	EMC1	100.0
EML1	100.0	ENSG00000284762	100.0	ENTPD1	100.0	EPM2A	100.0
ERAL1	100.0	ERBB4	100.0	ERCC1	100.0	ERCC2	100.0
ERCC3	100.0	ERCC4	100.0	ERCC5	100.0	ERCC6	100.0
ERCC8	100.0	ERLIN1	100.0	ERLIN2	100.0	ETFA	100.0
ETFB	100.0	ETFDH	100.0	ETHE1	100.0	EXOSC3	100.0
EXOSC8	100.0	EXOSC9	100.0	EXT2	100.0	EZH2	100.0
FA2H	100.0	FAH	100.0	FAR1	100.0	FARS2	100.0
FASTKD2	100.0	FAT2	100.0	FBXL4	100.0	FBXO7	100.0
FGD4	100.0	FGF12	100.0	FGF14	100.0	FGFR1	100.0
FIG4	100.0	FITM2	100.0	FKRP	100.0	FKTN	100.0
FLNA	100.0	FLNC	100.0	FLVCR1	100.0	FOLR1	100.0
FOXG1	98.87	FOXRED1	100.0	FRMD4A	100.0	FRRS1L	100.0
FTL	100.0	FUCA1	100.0	FUS	100.0	FXN	100.0
GABBR2	99.6	GABRA1	100.0	GABRA2	100.0	GABRB1	100.0
GABRB2	100.0	GABRB3	100.0	GABRD	100.0	GABRG2	100.0

<i>GAD1</i>	100.0	<i>GALC</i>	100.0	<i>GALE</i>	100.0	<i>GALK1</i>	100.0
<i>GALT</i>	100.0	<i>GAMT</i>	100.0	<i>GAN</i>	100.0	<i>GATAD2B</i>	100.0
<i>GATM</i>	100.0	<i>GBA</i>	98.29	<i>GBA2</i>	100.0	<i>GBE1</i>	100.0
<i>GCDH</i>	100.0	<i>GCH1</i>	100.0	<i>GCLC</i>	100.0	<i>GDAP2</i>	100.0
<i>GEMIN4</i>	100.0	<i>GFAP</i>	100.0	<i>GFM1</i>	100.0	<i>GFM2</i>	100.0
<i>GIGYF2</i>	100.0	<i>GJA1</i>	100.0	<i>GJB1</i>	100.0	<i>GJC2</i>	100.0
<i>GLB1</i>	100.0	<i>GLRA1</i>	100.0	<i>GLRX5</i>	100.0	<i>GLS</i>	100.0
<i>GLUD2</i>	100.0	<i>GLYCK</i>	100.0	<i>GM2A</i>	100.0	<i>GMPPA</i>	100.0
<i>GMPPB</i>	100.0	<i>GNAL</i>	100.0	<i>GNAO1</i>	100.0	<i>GNMT</i>	100.0
<i>GOSR2</i>	100.0	<i>GPAA1</i>	100.0	<i>GPR88</i>	100.0	<i>GRIA3</i>	100.0
<i>GRID2</i>	100.0	<i>GRIK2</i>	100.0	<i>GRIN1</i>	100.0	<i>GRIN2A</i>	100.0
<i>GRIN2B</i>	100.0	<i>GRIN2D</i>	98.52	<i>GRM1</i>	100.0	<i>GRN</i>	100.0
<i>GSS</i>	100.0	<i>GUF1</i>	100.0	<i>HACE1</i>	100.0	<i>HADH</i>	100.0
<i>HADHA</i>	100.0	<i>HADHB</i>	100.0	<i>HARS1</i>	100.0	<i>HARS2</i>	100.0
<i>HCFC1</i>	100.0	<i>HCN1</i>	100.0	<i>HDAC4</i>	100.0	<i>HEPACAM</i>	100.0
<i>HEPN1</i>	100.0	<i>HERC1</i>	100.0	<i>HEXA</i>	100.0	<i>HEXB</i>	100.0
<i>HIBCH</i>	100.0	<i>HIKESHI</i>	100.0	<i>HK1</i>	100.0	<i>HLCS</i>	100.0
<i>HMBS</i>	100.0	<i>HMGCL</i>	100.0	<i>HNRNPA1</i>	100.0	<i>HNRNPA2B1</i>	100.0
<i>HNRNPH2</i>	100.0	<i>HNRNPU</i>	100.0	<i>HPCA</i>	100.0	<i>HPD</i>	100.0
<i>HPDL</i>	100.0	<i>HPRT1</i>	100.0	<i>HSD17B10</i>	100.0	<i>HSD17B4</i>	100.0
<i>HSPD1</i>	100.0	<i>HSPG2</i>	99.98	<i>HTRA1</i>	99.66	<i>HTRA2</i>	100.0
<i>IBA57</i>	100.0	<i>IFIH1</i>	100.0	<i>IFRD1</i>	100.0	<i>IFT140</i>	100.0
<i>IFT172</i>	100.0	<i>IFT27</i>	100.0	<i>INPP5E</i>	100.0	<i>INTS8</i>	100.0
<i>IQSEC2</i>	100.0	<i>IRF2BPL</i>	100.0	<i>ISCA2</i>	100.0	<i>ITM2B</i>	100.0
<i>ITPA</i>	100.0	<i>ITPR1</i>	100.0	<i>IVD</i>	100.0	<i>ABCA1</i>	100.0
<i>ARHGEF9</i>	100.0	<i>ARMC9</i>	100.0	<i>ASS1</i>	100.0	<i>ATP8A2</i>	100.0
<i>BCOR</i>	100.0	<i>CACNA1B</i>	99.96	<i>CIZ1</i>	100.0	<i>COQ2</i>	100.0
<i>CRLF1</i>	100.0	<i>CRPPA</i>	88.62	<i>CSF1R</i>	100.0	<i>CTDP1</i>	100.0
<i>DMXL2</i>	100.0	<i>DNAJC3</i>	100.0	<i>ENSG00000272442</i>	100.0	<i>FAT2</i>	100.0
<i>FRMD4A</i>	100.0	<i>GATM</i>	100.0	<i>GFM2</i>	100.0	<i>GLB1</i>	100.0
<i>HERC1</i>	100.0	<i>KCNMA1</i>	100.0	<i>KLC2</i>	100.0	<i>LAMA2</i>	100.0
<i>LRRK2</i>	100.0	<i>LYST</i>	100.0	<i>MME</i>	100.0	<i>NDUFAF5</i>	100.0
<i>NDUFS4</i>	100.0	<i>NOS3</i>	100.0	<i>NRXN1</i>	100.0	<i>NT5C2</i>	100.0
<i>OFD1</i>	100.0	<i>PEX2</i>	100.0	<i>PIEZO2</i>	100.0	<i>PLCB1</i>	100.0
<i>PNKD</i>	100.0	<i>PPP3CA</i>	100.0	<i>PRICKLE2</i>	100.0	<i>PRKN</i>	100.0
<i>PROK2</i>	100.0	<i>RORA</i>	100.0	<i>SLC20A2</i>	100.0	<i>SYNE1</i>	100.0
<i>SZT2</i>	100.0	<i>TPK1</i>	100.0	<i>TRAPPC11</i>	100.0	<i>TTBK2</i>	100.0
<i>TTR</i>	100.0	<i>UNC80</i>	100.0	<i>WWOX</i>	100.0	<i>JAM3</i>	100.0
<i>KANK1</i>	100.0	<i>KANSL1</i>	100.0	<i>KARS1</i>	100.0	<i>KATNIP</i>	100.0
<i>KCNA1</i>	100.0	<i>KCNA2</i>	100.0	<i>KCNA4</i>	100.0	<i>KCNB1</i>	100.0
<i>KCNC1</i>	100.0	<i>KCNC3</i>	98.74	<i>KCND3</i>	100.0	<i>KCNH5</i>	100.0
<i>KCNJ10</i>	100.0	<i>KCNJ11</i>	100.0	<i>KCNJ6</i>	100.0	<i>KCNMA1</i>	100.0
<i>KCNQ2</i>	100.0	<i>KCNQ3</i>	100.0	<i>KCNT1</i>	99.72	<i>KCNT2</i>	100.0
<i>KCTD17</i>	100.0	<i>KCTD7</i>	100.0	<i>KDM5C</i>	100.0	<i>KIAA0586</i>	100.0
<i>KIDINS220</i>	100.0	<i>KIF1A</i>	100.0	<i>KIF1B</i>	100.0	<i>KIF1C</i>	100.0
<i>KIF2A</i>	100.0	<i>KIF5A</i>	100.0	<i>KIF5C</i>	100.0	<i>KIF7</i>	99.97
<i>KLC2</i>	100.0	<i>KMT2B</i>	99.43	<i>KY</i>	100.0	<i>L1CAM</i>	100.0
<i>L2HGDH</i>	100.0	<i>LAGE3</i>	100.0	<i>LAMA2</i>	100.0	<i>LAMB2</i>	100.0
<i>LARGE1</i>	100.0	<i>LGI1</i>	100.0	<i>LIAS</i>	100.0	<i>LIPT1</i>	100.0
<i>LIPT2</i>	100.0	<i>LMBRD1</i>	100.0	<i>LMNB1</i>	100.0	<i>LMNB2</i>	98.42
<i>LOC124901323</i>	100.0	<i>LRP4</i>	100.0	<i>LRPPRC</i>	100.0	<i>LRRK2</i>	100.0
<i>LRSAM1</i>	100.0	<i>LYRM7</i>	100.0	<i>LYST</i>	100.0	<i>MAG</i>	100.0

MAGI2	99.07	MAN2B1	100.0	MAPT	100.0	MARS1	100.0
MARS2	100.0	MAT1A	100.0	MATR3	100.0	MBD5	100.0
MCCC1	100.0	MCCC2	100.0	MCOLN1	100.0	MDH2	100.0
MECP2	100.0	MECR	100.0	MED13L	100.0	MED17	100.0
MED25	100.0	MEF2C	100.0	MFF	100.0	MFN2	100.0
MFSD2A	100.0	MFSD8	100.0	MGME1	100.0	MICU1	100.0
MIPEP	100.0	MIR3936HG	100.0	MKS1	100.0	MLC1	100.0
MLYCD	100.0	MMAA	100.0	MMAB	100.0	MMACHC	100.0
MMADHC	100.0	MME	100.0	MMUT	100.0	MOC51	100.0
MOC52	100.0	MORC2	100.0	MPDU1	100.0	MPV17	100.0
MPZ	100.0	MRE11	100.0	MRPS22	100.0	MRPS34	100.0
MTHFR	100.0	MT01	100.0	MTFAP	100.0	MTR	100.0
MTRR	100.0	MTTP	100.0	MVK	100.0	MYBPC1	100.0
MYH14	100.0	MYO1C	100.0	MYO7A	100.0	NADK2	99.44
NAGA	100.0	NAGLU	100.0	NALCN	100.0	NANS	100.0
NAT8L	97.96	NAXE	100.0	NDE1	100.0	NDRG1	100.0
NDUFA1	100.0	NDUFA10	100.0	NDUFA11	100.0	NDUFA12	100.0
NDUFA13	100.0	NDUFA2	100.0	NDUFA4	100.0	NDUFA9	100.0
NDUFAF1	100.0	NDUFAF2	100.0	NDUFAF3	100.0	NDUFAF4	100.0
NDUFAF5	100.0	NDUFAF6	100.0	NDUFB3	100.0	NDUFB9	100.0
NDUFS1	100.0	NDUFS2	100.0	NDUFS3	100.0	NDUFS4	100.0
NDUFS6	100.0	NDUFS7	100.0	NDUFS8	100.0	NDUFV1	100.0
NDUFV2	100.0	NECAP1	100.0	NEFH	100.0	NEFL	100.0
NEU1	100.0	NEXMIF	100.0	NF1	100.0	NF2	100.0
NFASC	100.0	NGLY1	100.0	NHLRC1	100.0	NIPA1	100.0
NMNAT1	100.0	NOL3	100.0	NOS3	100.0	NOTCH3	95.68
NPC1	100.0	NPC2	100.0	NPHP1	100.0	NPHP3- ACAD11	100.0
NR2F1	100.0	NR4A2	100.0	NRXN1	100.0	NSUN2	100.0
NT5C2	100.0	NTRK2	100.0	NUBPL	100.0	NUP62	100.0
OCLN	90.0	ODF3B	100.0	OFD1	100.0	OPA1	100.0
OPA3	100.0	OPHN1	100.0	OPTN	100.0	OSGEP	100.0
OTC	100.0	PACS2	100.0	PAFAH1B1	100.0	PAH	100.0
PANK2	100.0	PANO1	100.0	PARK7	100.0	PARN	100.0
PARS2	100.0	PC	100.0	PCBD1	100.0	PCCA	100.0
PCCB	100.0	PCDH12	100.0	PCDH19	100.0	PCLO	100.0
PCNA	100.0	PDE10A	99.54	PDE6D	100.0	PDE8B	100.0
PDGFB	100.0	PDGFRB	100.0	PDHA1	100.0	PDHB	100.0
PDHX	100.0	PDP1	100.0	PDSS1	100.0	PDSS2	100.0
PDYN	100.0	PET100	100.0	PEX1	100.0	PEX10	100.0
PEX11B	100.0	PEX12	100.0	PEX13	100.0	PEX14	100.0
PEX16	100.0	PEX19	100.0	PEX2	100.0	PEX26	100.0
PEX3	100.0	PEX5	100.0	PEX6	100.0	PEX7	100.0
PFN1	100.0	PGAP1	100.0	PGK1	100.0	PGM3	100.0
PHGDH	100.0	PHKA1	100.0	PHYH	100.0	PIBF1	100.0
PIEZO2	100.0	PIGA	100.0	PIGG	100.0	PIGN	100.0
PIGO	100.0	PIGP	100.0	PIGQ	100.0	PIGS	100.0
PIGV	100.0	PIK3R5	100.0	PINK1	99.91	PITRM1	100.0
PLA2G6	100.0	PLAA	100.0	PLCB1	100.0	PLD3	100.0
PLEKHG2	100.0	PLEKHG4	100.0	PLP1	100.0	PLXND1	99.93
PMM2	100.0	PMP22	100.0	PMPCA	100.0	PNKD	100.0
PNKP	100.0	PNP	100.0	PNPLA6	100.0	PNPLA8	100.0

<i>PNPO</i>	100.0	<i>PNPT1</i>	100.0	<i>PODXL</i>	100.0	<i>POLG</i>	100.0
<i>POLG2</i>	100.0	<i>POLR1C</i>	100.0	<i>POLR3A</i>	100.0	<i>POLR3B</i>	100.0
<i>POMGNT1</i>	100.0	<i>POMGNT2</i>	100.0	<i>POMK</i>	100.0	<i>POMT1</i>	100.0
<i>POMT2</i>	100.0	<i>PON1</i>	100.0	<i>PPP1R15B</i>	100.0	<i>PPP3CA</i>	100.0
<i>PPT1</i>	100.0	<i>PQBP1</i>	100.0	<i>PRDM8</i>	100.0	<i>PRF1</i>	100.0
<i>PRICKLE1</i>	100.0	<i>PRICKLE2</i>	100.0	<i>PRKCG</i>	100.0	<i>PRKN</i>	100.0
<i>PRKRA</i>	100.0	<i>PROK2</i>	100.0	<i>PROKR2</i>	100.0	<i>PRPH</i>	100.0
<i>PRPS1</i>	100.0	<i>PRRT2</i>	100.0	<i>PRX</i>	100.0	<i>PSAP</i>	100.0
<i>PSAT1</i>	100.0	<i>PSEN1</i>	100.0	<i>PSEN2</i>	100.0	<i>PSMB8</i>	100.0
<i>PTEN</i>	100.0	<i>PTF1A</i>	100.0	<i>PTPN23</i>	100.0	<i>PTRH1</i>	100.0
<i>PTRH2</i>	100.0	<i>PTRHD1</i>	100.0	<i>PTS</i>	100.0	<i>PUM1</i>	100.0
<i>PURA</i>	100.0	<i>PYCR2</i>	100.0	<i>PYROXD1</i>	100.0	<i>QARS1</i>	100.0
<i>QDPR</i>	98.15	<i>RAB18</i>	100.0	<i>RAB27A</i>	100.0	<i>RAB39B</i>	100.0
<i>RAB3GAP1</i>	100.0	<i>RAD50</i>	100.0	<i>RARS1</i>	100.0	<i>RARS2</i>	100.0
<i>REEP1</i>	100.0	<i>REEP2</i>	100.0	<i>RELN</i>	100.0	<i>RETREG1</i>	100.0
<i>RFT1</i>	100.0	<i>RHOBTB2</i>	100.0	<i>RLIM</i>	100.0	<i>RNASEH1</i>	100.0
<i>RNASEH2A</i>	100.0	<i>RNASEH2B</i>	100.0	<i>RNASEH2B-AS1</i>	100.0	<i>RNASEH2C</i>	100.0
<i>RNASET2</i>	100.0	<i>RNF113A</i>	100.0	<i>RNF125</i>	100.0	<i>RNF168</i>	100.0
<i>RNF170</i>	100.0	<i>RNF216</i>	100.0	<i>ROGDI</i>	97.38	<i>RORA</i>	100.0
<i>RPGRIP1L</i>	100.0	<i>RPIA</i>	100.0	<i>RPL10</i>	95.0	<i>RRM2B</i>	100.0
<i>RTN2</i>	100.0	<i>RTN4IP1</i>	100.0	<i>RTTN</i>	100.0	<i>RUBCN</i>	100.0
<i>SACS</i>	100.0	<i>SAMD9L</i>	100.0	<i>SAMHD1</i>	100.0	<i>SARS1</i>	100.0
<i>SCARB2</i>	100.0	<i>SCN1A</i>	100.0	<i>SCN1A-AS1</i>	100.0	<i>SCN1B</i>	100.0
<i>SCN2A</i>	100.0	<i>SCN3A</i>	100.0	<i>SCN8A</i>	100.0	<i>SCN9A</i>	100.0
<i>SCO1</i>	100.0	<i>SCO2</i>	100.0	<i>SCP2</i>	100.0	<i>SCYL1</i>	100.0
<i>SDHA</i>	100.0	<i>SDHAF1</i>	100.0	<i>SDHB</i>	100.0	<i>SDHD</i>	100.0
<i>SELENOI</i>	100.0	<i>SEPSECS</i>	100.0	<i>SERAC1</i>	100.0	<i>SETX</i>	100.0
<i>SFXN4</i>	100.0	<i>SGCE</i>	100.0	<i>SGPL1</i>	100.0	<i>SH3TC2</i>	100.0
<i>SHMT2</i>	100.0	<i>SIGMAR1</i>	100.0	<i>SIK1</i>	24.29	<i>SIL1</i>	100.0
<i>SLC12A5</i>	100.0	<i>SLC12A6</i>	100.0	<i>SLC13A5</i>	100.0	<i>SLC16A2</i>	100.0
<i>SLC17A5</i>	100.0	<i>SLC18A2</i>	100.0	<i>SLC19A2</i>	94.44	<i>SLC19A3</i>	100.0
<i>SLC1A2</i>	100.0	<i>SLC1A3</i>	100.0	<i>SLC1A4</i>	100.0	<i>SLC20A2</i>	100.0
<i>SLC22A5</i>	100.0	<i>SLC25A12</i>	100.0	<i>SLC25A13</i>	100.0	<i>SLC25A15</i>	100.0
<i>SLC25A20</i>	100.0	<i>SLC25A22</i>	100.0	<i>SLC25A4</i>	100.0	<i>SLC25A46</i>	100.0
<i>SLC2A1</i>	100.0	<i>SLC30A10</i>	100.0	<i>SLC30A9</i>	100.0	<i>SLC33A1</i>	100.0
<i>SLC35A2</i>	100.0	<i>SLC39A14</i>	100.0	<i>SLC39A4</i>	100.0	<i>SLC44A1</i>	100.0
<i>SLC46A1</i>	100.0	<i>SLC52A2</i>	100.0	<i>SLC52A3</i>	100.0	<i>SLC6A1</i>	100.0
<i>SLC6A17</i>	100.0	<i>SLC6A19</i>	100.0	<i>SLC6A3</i>	100.0	<i>SLC6A8</i>	100.0
<i>SLC9A1</i>	100.0	<i>SLC9A6</i>	100.0	<i>SMAD4</i>	100.0	<i>SMC1A</i>	100.0
<i>SMG9</i>	100.0	<i>SMPD1</i>	100.0	<i>SMS</i>	98.95	<i>SNAP25</i>	100.0
<i>SNCA</i>	100.0	<i>SNHG14</i>	100.0	<i>SNX14</i>	100.0	<i>SOD1</i>	100.0
<i>SOX2</i>	100.0	<i>SOX6</i>	100.0	<i>SPART</i>	100.0	<i>SPAST</i>	100.0
<i>SPG11</i>	100.0	<i>SPG21</i>	100.0	<i>SPG7</i>	100.0	<i>SPR</i>	100.0
<i>SPTAN1</i>	100.0	<i>SPTBN2</i>	100.0	<i>SPTBN4</i>	100.0	<i>SPTLC2</i>	100.0
<i>SQSTM1</i>	100.0	<i>SRPX2</i>	100.0	<i>SRY</i>	100.0	<i>ST3GAL3</i>	100.0
<i>STAMBP</i>	100.0	<i>STN1</i>	100.0	<i>STUB1</i>	100.0	<i>STXBP1</i>	100.0
<i>SUCLA2</i>	100.0	<i>SUFU</i>	100.0	<i>SUMF1</i>	100.0	<i>SUOX</i>	100.0
<i>SURF1</i>	100.0	<i>SVBP</i>	100.0	<i>SYN1</i>	100.0	<i>SYNE1</i>	100.0
<i>SYNGAP1</i>	100.0	<i>SYNJ1</i>	100.0	<i>SYT14</i>	100.0	<i>SYT2</i>	100.0
<i>SZT2</i>	100.0	<i>TACO1</i>	100.0	<i>TAF1</i>	100.0	<i>TAF2</i>	100.0
<i>TANGO2</i>	100.0	<i>TARDBP</i>	100.0	<i>TAT</i>	100.0	<i>TBC1D20</i>	100.0
<i>TBC1D23</i>	100.0	<i>TBC1D24</i>	100.0	<i>TBCD</i>	100.0	<i>TBCE</i>	100.0

<i>TBK1</i>	100.0	<i>TCF4</i>	100.0	<i>TCN2</i>	100.0	<i>TCTN1</i>	100.0
<i>TCTN2</i>	100.0	<i>TCTN3</i>	100.0	<i>TDP1</i>	100.0	<i>TDP2</i>	100.0
<i>TECPR2</i>	100.0	<i>TELO2</i>	100.0	<i>TENM4</i>	100.0	<i>TFG</i>	100.0
<i>TGM6</i>	100.0	<i>TH</i>	100.0	<i>THAP1</i>	100.0	<i>THG1L</i>	100.0
<i>THOC2</i>	100.0	<i>TIMM8A</i>	100.0	<i>TIMMDC1</i>	100.0	<i>TINF2</i>	100.0
<i>TMEM107</i>	100.0	<i>TMEM138</i>	100.0	<i>TMEM216</i>	100.0	<i>TMEM231</i>	100.0
<i>TMEM237</i>	100.0	<i>TMEM240</i>	100.0	<i>TMEM67</i>	100.0	<i>TMEM70</i>	100.0
<i>TMX2</i>	100.0	<i>TOR1A</i>	100.0	<i>TP53RK</i>	100.0	<i>TPI1</i>	100.0
<i>TPK1</i>	100.0	<i>TPM2</i>	100.0	<i>TPP1</i>	100.0	<i>TRAPPC11</i>	100.0
<i>TRAPPC12</i>	100.0	<i>TRAPPC4</i>	100.0	<i>TREM2</i>	100.0	<i>TREX1</i>	100.0
<i>TRNT1</i>	100.0	<i>TRPC3</i>	100.0	<i>TRPM7</i>	100.0	<i>TSC1</i>	100.0
<i>TSC2</i>	100.0	<i>TSEN15</i>	100.0	<i>TSEN2</i>	100.0	<i>TSEN54</i>	99.07
<i>TSFM</i>	100.0	<i>TTBK2</i>	100.0	<i>TTC19</i>	100.0	<i>TTC21B</i>	100.0
<i>TTPA</i>	100.0	<i>TTR</i>	100.0	<i>TUBA1A</i>	100.0	<i>TUBA4A</i>	100.0
<i>TUBB</i>	100.0	<i>TUBB3</i>	100.0	<i>TUBB4A</i>	100.0	<i>TUBG1</i>	100.0
<i>TWNK</i>	100.0	<i>TXN2</i>	100.0	<i>TYMP</i>	100.0	<i>TYROBP</i>	100.0
<i>UBA5</i>	100.0	<i>UBE2A</i>	100.0	<i>UBE3A</i>	100.0	<i>UBQLN2</i>	100.0
<i>UBR4</i>	100.0	<i>UBTF</i>	100.0	<i>UCHL1</i>	100.0	<i>UFC1</i>	100.0
<i>UNC80</i>	100.0	<i>UPB1</i>	100.0	<i>UQCC2</i>	100.0	<i>UQCC3</i>	100.0
<i>UQCRB</i>	100.0	<i>UQCRC2</i>	100.0	<i>UQCRFS1</i>	100.0	<i>UQCRQ</i>	100.0
<i>UROC1</i>	100.0	<i>USP18</i>	90.0	<i>VAC14</i>	100.0	<i>VAMP1</i>	100.0
<i>VAPB</i>	100.0	<i>VARS2</i>	100.0	<i>VCP</i>	100.0	<i>VLDLR</i>	100.0
<i>VPS11</i>	100.0	<i>VPS13A</i>	100.0	<i>VPS13C</i>	100.0	<i>VPS13D</i>	100.0
<i>VPS16</i>	100.0	<i>VPS35</i>	100.0	<i>VPS37A</i>	100.0	<i>VPS51</i>	100.0
<i>VPS53</i>	100.0	<i>VRK1</i>	100.0	<i>VWA3B</i>	100.0	<i>WARS2</i>	100.0
<i>WASHC5</i>	100.0	<i>WDR26</i>	100.0	<i>WDR45</i>	100.0	<i>WDR62</i>	100.0
<i>WDR73</i>	100.0	<i>WDR81</i>	100.0	<i>WFS1</i>	100.0	<i>WNK1</i>	100.0
<i>WWOX</i>	100.0	<i>XK</i>	100.0	<i>XPA</i>	100.0	<i>XPNPEP3</i>	100.0
<i>XPR1</i>	100.0	<i>XRCC1</i>	100.0	<i>XRCC4</i>	100.0	<i>YME1L1</i>	100.0
<i>YWHAG</i>	100.0	<i>ZC4H2</i>	100.0	<i>ZEB2</i>	100.0	<i>ZFYVE26</i>	100.0
<i>ZFYVE27</i>	100.0	<i>ZNF142</i>	100.0	<i>ZNF335</i>	100.0	<i>ZNF423</i>	100.0
<i>ZNHIT3</i>	100.0	<i>SERPINI1</i>	100.0				