

Supplementary table 1. List of 172 targeted genes included in the developmental and epileptic encephalopathy panel

Gene	OMIM	Full name	Cytogenetic location
<i>AARS</i>	601065	ALANYL-tRNA SYNTHETASE	16q22.1
<i>ABAT</i>	137150	4-AMINOBTYRATE AMINOTRANSFERASE	16p13.2
<i>ACADL</i>	609576	ACYL-CoA DEHYDROGENASE, LONG-CHAIN	2q34
<i>ACADM</i>	607008	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN	1p31.1
<i>ACADS</i>	606885	ACYL-CoA DEHYDROGENASE, SHORT-CHAIN	12q24.31
<i>ACY1</i>	104620	AMINOACYLASE 1	3p21.2
<i>ADGRV1</i>	602851	ADHESION G PROTEIN-COUPLED RECEPTOR V1	5q14.3
<i>ADSL</i>	608222	ADENYLOSUCCINATE LYASE	22q13.1
<i>ALAD</i>	125270	DELTA-AMINOLEVULINATE DEHYDRATASE	9q32
<i>ALAS2</i>	301300	DELTA-AMINOLEVULINATE SYNTHASE 2	Xp11.21
<i>ALDH4A1</i>	606811	ALDEHYDE DEHYDROGENASE, FAMILY 4, SUBFAMILY A, MEMBER 1	1p36.13
<i>ALDH7A1</i>	107323	ALDEHYDE DEHYDROGENASE 7 FAMILY, MEMBER A1	5q23.2
<i>ALG13</i>	300776	ASPARAGINE-LINKED GLYCOSYLATION 13	Xq23
<i>ALPL</i>	171760	ALKALINE PHOSPHATASE, LIVER	1p36.12
<i>AMT</i>	238310	AMINOMETHYLTRANSFERASE	3p21.31
<i>ARHGEF15</i>	608504	RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 15	17p13.1
<i>ARHGEF9</i>	300429	RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 9	Xq11.1
<i>ARX</i>	300382	ARISTALESS-RELATED HOMEBOX, X-LINKED	Xp21.3
<i>ASNS</i>	108370	ASPARAGINE SYNTHETASE	7q21.3
<i>ASPM</i>	605481	ABNORMAL SPINDLE-LIKE, MICROCEPHALY-ASSOCIATED	1q31.3
<i>ATP13A2</i>	610513	ATPase, TYPE 13A2	1p36.13
<i>ATP6AP2</i>	300556	ATPase, H ⁺ TRANSPORTING, LYSOSOMAL, ACCESSORY PROTEIN 2	Xp11.4
<i>BRAT1</i>	614506	BRCA1-ASSOCIATED ATM ACTIVATOR 1	7p22.3
<i>BTD</i>	609019	BIOTINIDASE	3p25.1
<i>CACNA1A</i>	601011	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT	19p13.13
<i>CACNB4</i>	601949	CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT	2q23.3
<i>CASK</i>	300172	CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE	Xp11.4
<i>CASR</i>	601199	CALCIUM-SENSING RECEPTOR	3q13.3-q21.1
<i>CBS</i>	613381	CYSTATHIONINE BETA-SYNTHASE	21q22.3
<i>CDKL5</i>	300203	CYCLIN-DEPENDENT KINASE-LIKE 5	Xp22.13
<i>CHD2</i>	602119	CHROMODOMAIN HELICASE DNA-BINDING PROTEIN 2	15q26.1
<i>CHRNA2</i>	118502	CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC,	8p21.2

		ALPHA POLYPEPTIDE 2	
<i>CHRNA4</i>	118504	CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 4	20q13.33
<i>CHRNA7</i>	118511	CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 7	15q13.3
<i>CHRNA2</i>	118507	CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, BETA POLYPEPTIDE 2	1q21.3
<i>CLCN4</i>	302910	CHLORIDE CHANNEL 4	Xp22.2
<i>CLN3</i>	607042	CLN3 GENE	16p12.1
<i>CLN5</i>	608102	CLN5 GENE	13q22.3
<i>CLN6</i>	606725	CLN6 GENE	15q23
<i>CLN8</i>	607837	CLN8 GENE	8p23.3
<i>CNTNAP2</i>	604569	CONTACTIN-ASSOCIATED PROTEIN-LIKE 2	7q35-q36
<i>COL4A1</i>	120130	COLLAGEN, TYPE IV, ALPHA-1	13q34
<i>CPOX</i>	612732	COPROPORPHYRINOGEN OXIDASE	3q11.2
<i>CPT1A</i>	600528	CARNITINE PALMITOYLTRANSFERASE I, LIVER	11q13.3
<i>CPT1B</i>	601987	CARNITINE PALMITOYLTRANSFERASE I, MUSCLE	22q13.33
<i>CPT2</i>	600650	CARNITINE PALMITOYLTRANSFERASE II	1p32.3
<i>CSTB</i>	601145	CYSTATIN B	21q22.3
<i>CTSD</i>	116840	CATHEPSIN D	11p15.5
<i>CTSF</i>	603539	CATHEPSIN F	11q13.2
<i>DNAJC5</i>	611203	DNAJ/HSP40 HOMOLOG, SUBFAMILY C, MEMBER 5	20q13.33
<i>DNM1</i>	602377	DYNAMIN 1	9q34.11
<i>DOCK7</i>	615730	DEDICATOR OF CYTOKINESIS 7	1p31.3
<i>DYRK1A</i>	600855	DUAL-SPECIFICITY TYROSINE PHOSPHORYLATION-REGULATED KINASE 1A	21q22.13
<i>EEF1A2</i>	602959	EUKARYOTIC TRANSLATION ELONGATION FACTOR 1, ALPHA-2	20q13.33
<i>EPM2A</i>	607566	EPM2A GENE	6q24.3
<i>FARS2</i>	611592	PHENYLALANYL-tRNA SYNTHETASE 2, MITOCHONDRIAL	6p25.1
<i>FECH</i>	612386	FERROCHELATASE	18q21.31
<i>FOLR1</i>	136430	FOLATE RECEPTOR 1, ADULT	11q13.4
<i>FOXG1</i>	164874	FORKHEAD BOX G1	14q12
<i>GABBR2</i>	607340	GAMMA-AMINOBUTYRIC ACID B RECEPTOR 2	9q22.33
<i>GABRA1</i>	137160	GAMMA-AMINOBUTYRIC ACID RECEPTOR, ALPHA-1	5q34
<i>GABRB3</i>	137192	GAMMA-AMINOBUTYRIC ACID RECEPTOR, BETA-3	15q12
<i>GABRG2</i>	137164	GAMMA-AMINOBUTYRIC ACID RECEPTOR, GAMMA-2	5q34
<i>GAMT</i>	601240	GUANIDINOACETATE METHYLTRANSFERASE	19p13.3
<i>GATM</i>	602360	L-ARGININE:GLYCINE AMIDINOTRANSFERASE	15q21.1
<i>GCSH</i>	238330	GLYCINE CLEAVAGE SYSTEM H PROTEIN	16q23.2
<i>GLDC</i>	238300	GLYCINE DECARBOXYLASE	9p24.1
<i>GNAO1</i>	139311	GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-	16q13

		ACTIVATING ACTIVITY POLYPEPTIDE O	
<i>GOSR2</i>	604027	GOLGI SNAP RECEPTOR COMPLEX MEMBER 2	17q21.32
<i>GRIN1</i>	138249	GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL-D-ASPARTATE, SUBUNIT 1	9q34.3
<i>GRIN2A</i>	138253	GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL-D-ASPARTATE, SUBUNIT 2A	16p13.2
<i>GRIN2B</i>	138252	GLUTAMATE RECEPTOR, IONOTROPIC, N-METHYL-D-ASPARTATE, SUBUNIT 2B	12p13.1
<i>GRN</i>	138945	GRANULIN PRECURSOR	17q21.31
<i>HADH</i>	601609	3-HYDROXYACYL-CoA DEHYDROGENASE	4q25
<i>HADHA</i>	600890	HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT	2p23.3
<i>HCN1</i>	602780	HYPERPOLARIZATION-ACTIVATED CYCLIC NUCLEOTIDE-GATED POTASSIUM CHANNEL 1	5p12
<i>HCN4</i>	605206	HYPERPOLARIZATION-ACTIVATED CYCLIC NUCLEOTIDE-GATED POTASSIUM CHANNEL 4	15q24.1
<i>HFE</i>	613609	HFE GENE	6p22.2
<i>HLCS</i>	609018	HOLOCARBOXYLASE SYNTHETASE	21q22.13
<i>HMBS</i>	609806	HYDROXYMETHYLBILANE SYNTHASE	11q23.3
<i>HNRNPU</i>	602869	HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN U	1q44
<i>IQSEC2</i>	300522	IQ MOTIF- AND SEC7 DOMAIN-CONTAINING PROTEIN 2	Xp11.22
<i>KANSL1</i>	612452	KAT8 REGULATORY NSL COMPLEX, SUBUNIT 1	17q21.31
<i>KCNA1</i>	176260	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1	12p13.32
<i>KCNA2</i>	176262	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 2	1p13.3
<i>KCNB1</i>	600397	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAB-RELATED SUBFAMILY, MEMBER 1	20q13.13
<i>KCNC1</i>	176258	POTASSIUM CHANNEL, VOLTAGE-GATED, SHAW-RELATED SUBFAMILY, MEMBER 1	11p15.1
<i>KCNH5</i>	605716	POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY H, MEMBER 5	14q23.2
<i>KCNJ10</i>	602208	POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 10	1q23.2
<i>KCNJ11</i>	600937	POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 11	11p15.1
<i>KCNMA1</i>	600150	POTASSIUM CHANNEL, CALCIUM-ACTIVATED, LARGE CONDUCTANCE, SUBFAMILY M, ALPHA MEMBER 1	10q22.3
<i>KCNQ2</i>	602235	POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2	20q13.33
<i>KCNQ3</i>	602232	POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE	8q24.22

		SUBFAMILY, MEMBER 3	
<i>KCNT1</i>	608167	POTASSIUM CHANNEL, SUBFAMILY T, MEMBER 1	9q34.3
<i>KCTD7</i>	611725	POTASSIUM CHANNEL TETRAMERIZATION DOMAIN-CONTAINING PROTEIN 7	7q11.21
<i>KPNA7</i>	614107	KARYOPHERIN ALPHA-7	7q22.1
<i>LGI1</i>	604619	LEUCINE-RICH GENE, GLIOMA-INACTIVATED, 1	10q23.33
<i>LIAS</i>	607031	LIPOIC ACID SYNTHASE	4p14
<i>MAGI2</i>	606382	MEMBRANE-ASSOCIATED GUANYLATE KINASE, WW AND PDZ DOMAINS-CONTAINING, 2	7q21.11
<i>MBD5</i>	611472	METHYL-CpG-BINDING DOMAIN PROTEIN 5	2q23.1
<i>MECP2</i>	300005	METHYL-CpG-BINDING PROTEIN 2	Xq28
<i>MEF2C</i>	600662	MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE C	5q14.3
<i>MFSD8</i>	611124	MAJOR FACILITATOR SUPERFAMILY DOMAIN-CONTAINING PROTEIN 8	4q28.2
<i>MMADHC</i>	611935	MMADHC GENE	2q23.2
<i>MTHFR</i>	607093	5,10-METHYLENETETRAHYDROFOLATE REDUCTASE	1p36.22
<i>MTR</i>	156570	5-METHYLTETRAHYDROFOLATE-HOMOCYSTEINE S-METHYLTRANSFERASE	1q43
<i>MTRR</i>	602568	METHIONINE SYNTHASE REDUCTASE	5p15.31
<i>NECAP1</i>	611623	NECAP ENDOCYTOSIS-ASSOCIATED PROTEIN 1	12p13.31
<i>NHLRC1</i>	608072	NHL REPEAT-CONTAINING 1 GENE	6p22.3
<i>NRXN1</i>	600565	NEUREXIN I	2p16.3
<i>OPHN1</i>	300127	OLIGOPHRENIN 1	Xq12
<i>PAH</i>	612349	PHENYLALANINE HYDROXYLASE	12q23.2
<i>PC</i>	608786	PYRUVATE CARBOXYLASE	11q13.2
<i>PCDH19</i>	300460	PROTOCADHERIN 19	Xq22.1
<i>PHGDH</i>	606879	PHOSPHOGLYCERATE DEHYDROGENASE	1p12
<i>PIGA</i>	311770	PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS A PROTEIN	Xp22.2
<i>PIGQ</i>	605754	PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS Q PROTEIN	16p13.3
<i>PLCB1</i>	607120	PHOSPHOLIPASE C, BETA-1	20p12.3
<i>PNKP</i>	605610	POLYNUCLEOTIDE KINASE 3-PRIME PHOSPHATASE	19q13.33
<i>PNPO</i>	603287	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE	17q21.32
<i>POLG</i>	174763	POLYMERASE, DNA, GAMMA	15q26.1
<i>PPOX</i>	600923	PROTOPORPHYRINOGEN OXIDASE	1q23.3
<i>PPT1</i>	600722	PALMITOYL-PROTEIN THIOESTERASE 1	1p34.2
<i>PRICKLE1</i>	608500	PRICKLE, DROSOPHILA, HOMOLOG OF, 1	12q12
<i>PRICKLE2</i>	608501	PRICKLE, DROSOPHILA, HOMOLOG OF, 2	3p14.1
<i>PRODH</i>	606810	PROLINE DEHYDROGENASE (OXIDASE) 1	22q11.21
<i>PRRT2</i>	614386	PROLINE-RICH TRANSMEMBRANE PROTEIN 2	16p11.2
<i>PURA</i>	600473	PURINE-RICH ELEMENT-BINDING PROTEIN A	5q31.3

<i>QARS</i>	603727	GLUTAMINYL-tRNA SYNTHETASE	3p21.31
<i>SCARB2</i>	602257	SCAVENGER RECEPTOR CLASS B, MEMBER 2	4q21.1
<i>SCN1A</i>	182389	SODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT	2q24.3
<i>SCN1B</i>	600235	SODIUM CHANNEL, VOLTAGE-GATED, TYPE I, BETA SUBUNIT	19q13.11
<i>SCN2A</i>	182390	SODIUM CHANNEL, VOLTAGE-GATED, TYPE II, ALPHA SUBUNIT	2q24.3
<i>SCN3A</i>	182391	SODIUM CHANNEL, VOLTAGE-GATED, TYPE III, ALPHA SUBUNIT	2q24.3
<i>SCN8A</i>	600702	SODIUM CHANNEL, VOLTAGE-GATED, TYPE VIII, ALPHA SUBUNIT	12q13.13
<i>SCN9A</i>	603415	SODIUM CHANNEL, VOLTAGE-GATED, TYPE IX, ALPHA SUBUNIT	2q24.3
<i>SETBP1</i>	611060	SET-BINDING PROTEIN 1	18q12.3
<i>SIK1</i>	605705	SALT-INDUCIBLE KINASE 1	21q22.3
<i>SLC13A5</i>	608305	SOLUTE CARRIER FAMILY 13 (SODIUM-DEPENDENT CITRATE TRANSPORTER), MEMBER 5	17p13.1
<i>SLC19A3</i>	606152	SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 3	2q36.3
<i>SLC22A5</i>	603377	SOLUTE CARRIER FAMILY 22 (ORGANIC CATION TRANSPORTER), MEMBER 5	5q31.1
<i>SLC25A20</i>	613698	SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, GLUTAMATE), MEMBER 20	3p21.31
<i>SLC25A22</i>	609302	SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, GLUTAMATE), MEMBER 22	11p15.5
<i>SLC25A29</i>	615064	SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, GLUTAMATE), MEMBER 29	14q32.2
<i>SLC2A1</i>	138140	SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1	1p34.2
<i>SLC46A1</i>	611672	SOLUTE CARRIER FAMILY 46 (FOLATE TRANSPORTER), MEMBER 1	17q11.2
<i>SLC6A1</i>	137165	SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, GABA), MEMBER 1	3p25.3
<i>SLC6A8</i>	300036	SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, CREATINE), MEMBER 8	Xq28
<i>SLC9A6</i>	300231	SOLUTE CARRIER FAMILY 9, MEMBER 6	Xq26.3
<i>SMARCA2</i>	600014	SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, MEMBER 2	9p24.3
<i>SPTAN1</i>	182810	SPECTRIN, ALPHA, NONERYTHROCYTIC 1	9q34.11

<i>SRPX2</i>	300642	SUSHI REPEAT-CONTAINING PROTEIN, X-LINKED, 2	Xq22.1
<i>ST3GAL3</i>	606494	ST3 BETA-GALACTOSIDE ALPHA-2,3-SIALYLTRANSFERASE 3	1p34.1
<i>ST3GAL5</i>	604402	ST3 BETA-GALACTOSIDE ALPHA-2,3-SIALYLTRANSFERASE 5	2p11.2
<i>STX1B</i>	601485	SYNTAXIN 1B	16p11.2
<i>STXBP1</i>	602926	SYNTAXIN-BINDING PROTEIN 1	9q34.11
<i>SYN1</i>	313440	SYNAPSIN I	Xp11.3-p11.2
<i>SYNGAP1</i>	603384	SYNAPTIC RAS-GTPase-ACTIVATING PROTEIN 1	6p21.32
<i>SZT2</i>	615463	SEIZURE THRESHOLD 2, MOUSE, HOMOLOG OF	1p34.2
<i>TBC1D24</i>	613577	TBC1 DOMAIN FAMILY, MEMBER 24	16p13.3
<i>TBL1XR1</i>	608628	TRANSDUCIN-BETA-LIKE 1 RECEPTOR 1	3q26.32
<i>TCF4</i>	602272	TRANSCRIPTION FACTOR 4	18q21.2
<i>TNK2</i>	606994	TYROSINE KINASE, NONRECEPTOR, 2	3q29
<i>TPP1</i>	607998	TRIPTEPTIDYL PEPTIDASE I	11p15.4
<i>TSEN54</i>	608755	tRNA SPLICING ENDONUCLEASE 54, S. CEREVISIAE, HOMOLOG OF	17q25.1
<i>UBE2A</i>	312180	UBIQUITIN-CONJUGATING ENZYME E2A	Xq24
<i>UBE3A</i>	601623	UBIQUITIN-PROTEIN LIGASE E3A	15q11.2
<i>UROD</i>	613521	UROPORPHYRINOGEN DECARBOXYLASE	1p34.1
<i>UROS</i>	606938	UROPORPHYRINOGEN III SYNTHASE	10q26.2
<i>WDR62</i>	613583	WD REPEAT-CONTAINING PROTEIN 62	19q13.12
<i>WWOX</i>	605131	WW DOMAIN-CONTAINING OXIDOREDUCTASE	16q23.1-q23.2
<i>ZEB2</i>	605802	ZINC FINGER E BOX-BINDING HOMEBOX 2	2q22.3