Supplementary Table 1. List of IMDs presenting with epilepsies, treatablilty, laboratory investigations, OMIM references and IEMbase IDs.

Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
INTERMEDIARY METABOLISM: NUTRIENTS (n=39)											
Urea cycle disorders and inherited hyperammonemias											
Glutamate dehydrogenase superactivity	*GLUD1*	AD				EEG, abnormal	Drug-resistant epilepsy		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	606762	IEM0165
Carbamoyl phosphate synthetase I deficiency	*CPS1*	AR						Treatable	Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	237300	IEM0057
Ornithine transcarbamylase deficiency	*OTC*	XL						Treatable	Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	311250	IEM0058
Argininosuccinate synthetase deficiency	*ASS1*	AR	Neonatal seizures			Burst-suppression patten	Drug-resistant epilepsy		Ammonia (B), ASAT/ALAT (P), Urea (P), Amino acids (P), Orotic acid (U)	215700	IEM0059
Arginase 1 deficiency	*ARG1*	AR						Treatable	Ammonia (P); Amino acids (P); Purines and pyrimidines (U)	207800	IEM0061
Organic acidurias											
Propionic acidemia due to propionyl-CoA carboxylase subunit alpha deficiency	*PCCA*	AR	Neonatal seizures			Comb-like rhythm	Drug-resistant epilepsy		Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	232000	IEM0124
Propionic acidemia due to propionyl-CoA carboxylase subunit beta deficiency	*PCCB*	AR	Neonatal seizures			Comb-like rhythm	Drug-resistant epilepsy		Acyglycines (U), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	232000	IEM0125
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	*MMUT*	AR	Neonatal seizures				Drug-resistant epilepsy		Amino acids (P); Organic acids (U); Acylcarnitines (U, P, DBS); Anion gap	251000	IEM0127
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR	Neonatal seizures				Drug-resistant epilepsy		Organic acids (U), Acylcarnitines (DBS, P), Lactate (P), Glucose (S), Lipid panel (S)	248360	IEM0129
Disorders of branched-chain amino acid metabolism											
Branched-chain ketoacid dehydrogenase E1 alpha deficiency MSUD1a	*BCKDHA*	AR	Neonatal seizures			Comb-like rhythm	Drug-resistant epilepsy		Amino acids (P), Organic acids (U)	248600	IEM0108
Branched-chain ketoacid dehydrogenase E1 beta deficiency MSUD1b	*BCKDHB*	AR	Neonatal seizures			Comb-like rhythm	Drug-resistant epilepsy		Amino acids (P), Organic acids (U)	248600	IEM0109
Dihydrolipoyl transacylase deficiency MSUD2	*DBT*	AR				Comb-like rhythm	Drug-resistant epilepsy		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Acylglycines (U), ASAT/ALAT (P), CK (P), Lactate (P), Ammonia (P), Glucose (S), Urinalysis	248600	IEM0110
Disorders of phenylalanine metabolism											
Phenylalanine hydroxylase deficiency Classic PKU (late diagnosed, untreated)	*PAH*	AR			Myoclonic seizures			Treatable	Amino acids (P)	261600	IEM0082
Disorders of the metabolism of sulfur-containing amino acids and hydrogen sulfide											
Cystathionine beta-synthase deficiency Classical homocystinuria	*CBS*	AR						Treatable	SAM & SAH (P), Amino acids (P)	236200	IEM0102
Isolated sulfite oxidase deficiency	*SUOX*	AR	Neonatal seizures				Drug-resistant epilepsy		a-aminosemialdehyde (CSF), PLP (CSF), Sulfite (U), Amino acids (P)	272300	IEM0105
Disorders of glycine and serine metabolism											
Nonketotic hyperglycinemia due to glycine decarboxylase deficiency	*GLDC*	AR	Hiccups	Infantile spasms	Myoclonic seizures	Hypsarrhythmia (EEG) Burst-suppression (EEG)		Treatable	Amino acids (P, CSF)	238300	IEM0185
3-phosphoglycerate dehydrogenase deficiency	*PHGDH*	AR	Neonatal seizures	Infantile spasms	Myoclonic seizures	Hypsarrhythmia (EEG)	Drug-resistant epilepsy		5-Methyl-THF (CSF), Amino acids (P)	606879;601851	IEM0181
Phosphoserine aminotransferase deficiency	*PSAT1*	AR	Neonatal seizures					Treatable	Amino acids (P, CSF)	610992;610936	IEM0182
Nonketotic hyperglycinemia due to aminomethyltransferase deficiency	*AMT*	AR	Hiccups	Infantile spasms	Myoclonic seizures	Hypsarrhythmia (EEG) Burst-suppression (EEG)	Drug-resistant epilepsy		Amino acids (P, CSF)	605899	IEM0186
Phosphoserine phosphatase deficiency	*PSPH*	AR					Drug-resistant epilepsy	Treatable	Amino acids (P, CSF)	614023	IEM0183
ASCT1 transporter deficiency	*SLC1A4*	AR		Infantile spasms		Hypsarrhythmia (EEG) Epileptic spikes (EEG)	Drug-resistant epilepsy		DNA	616657	IEM0184
Disorders of ornithine, proline and hydroxyproline metabolism											
Pyrroline-5-carboxylate dehydrogenase deficiency Hyperprolinemia type 2	*ALDH4A1*	AR					Drug-resistant epilepsy		Amino acids (P, U); B6 vitaminers (U)	239510	IEM0142
Disorders of lysine, hydroxylysine, and tryptophan metabolism											

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Hydroxylysinuria	*HYKK*	AD			Myoclonic epilepsy		Drug-resistant epilepsy		Hydroxylysine (U)	614681	IEM1111
Disorders of glutamate/glutamine and aspartate/asparagine metabolism											
Glutamine synthetase deficiency	*GLUL*	AR	Neonatal seizures			EEG, abnormal	Epilepsy, intractable		Amino acids (P, CSF); Ammonia (B)	610015	IEM0179
Asparagine synthetase deficiency	*ASNS*	AR	Neonatal eizures	Infantile spasms		Hypsarrhythmia (EEG) Burst-suppression (EEG)	Drug-resistant epilepsy		Amino acids (P)	615574	IEM0180
Glutaminase deficiency	*GLS*	AR	Neonatal seizures			Burst-suppression (EEG)	Drug-resistant epilepsy		Amino acids (U, CSF)	618328	IEM1083
Disorders of amino acid transport											
Nephropathic cystinosis	*CTNS*	AR					Drug-resistant epilepsy		Amino acids (P), Cystine (WBC, PMN)	219800;219900;219750	IEM0873
Astroglial glutamate aspartate transporter deficiency	*SLC1A2*	AD		Infantile spasms		Hypsarrhythmia (EEG) EEG, abnormal	Drug-resistant epilepsy		DNA	617105	IEM0168
GABA transporter deficiency	*SLC6A1*	AD			Seizures, myoclonic-atonic				DNA	616421	IEM0154
Sodium-coupled neutral amino acid transporter 3 deficiency	*SLC38A3*	AR					Drug-resistant epilepsy		DNA	619881	IEM1924
Other disorders of amino acid metabolism											
Dipeptidase deficiency	*DPEP1*	?			Myoclonic seizures	EEG, abnormal	Drug-resistant epilepsy		Acyglycines (P, U); Cystine (U); Leucotriens (U)	179780	IEM1508
Disorders of gluconeogenesis											
Pyruvate carboxylase deficiency	*PC*	AR					Drug-resistant epilepsy		Amino acids (P), Lactate (P), Glucose (P), 3-OH-Butyrate/Acetoacetate (P)	266150	IEM0372
Disorders of glycogen metabolism											
Laforin deficiency	*EPM2A*	AR			Myoclonic epilepsy Myoclonus	Giant somatosensory evoked potentials	Drug-resistant epilepsy		DNA	254780	IEM0368
Malin deficiency	*NHLRC1*	AR			Myoclonic epilepsy	EEG, abnormal	Drug-resistant epilepsy		DNA	254780	IEM0369
Disorders of carbohydrate transmembrane transport and absorption											
Glucose transporter 1 deficiency	*SLC2A1*	AD, AR	Neonatal seizures					Treatable	Glucose (P, CSF)	606777;612126;601042;614847	IEM0314
Disorders of carnitine metabolism											
Carnitine palmitoyl-transferase 1C deficiency	*CPT1C*	AD	Neonatal seizures		Seizures	Evokedpotentials+/-, abnormal (EEG)	Drug-resistant epilepsy		DNA	616282	IEM1165
Disorders of ketone body metabolism											
3-Hydroxy-3-methylglutaryl-CoA synthase deficiency	*HMGCS2*	AR	Neonatal seizures					Treatable	Acyglycines (U), Organic acids (U), Acylcarnitines (P), Fatty acids and ketones (P)	605911	IEM0641
Mitochondrial acetoacetyl-CoA thiolase deficiency	*ACAT1*	AR					Drug-resistant epilepsy		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), Lactate (P), Acetoacetate (P, U), 3-Hydroxy-n-butyric acid (P, U)	203750	IEM0643
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	AR	Neonatal seizures					Treatable	Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), ASAT/ALAT (P), Lactate (P), Ammonia (P)	246450	IEM0122
INTERMEDIARY METABOLISM: ENERGY (n=42)											
Disorders of pyruvate metabolism											
Pyruvate dehydrogenase E1 alpha deficiency	*PDHA1*	XL					Drug-resistant epilepsy		Lactate (P); Pyruvate (P); Alanine (P)	312170	IEM0389
Pyruvate dehydrogenase E3-binding protein deficiency	*PDHX*	AR					Drug-resistant epilepsy		Lactate (P); Pyruvate (P); Alanine (P); Fatty acids and ketones (S, U)	245349	IEM0392
Disorders of the Krebs cycle											
Fumarate hydratase deficiency	*FH*	AR				EEG, abnormal	Drug-resistant epilepsy		Lactate (P), Organic acids (U), Catecholamines (P, U)	606812	IEM0401

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Mitochondrial malate dehydrogenase deficiency	*MDH2*	AR			Myoclonus				Lactate (P), Organic acids (U), Catecholamines (P, U)	617339	IEM0403
OGDHL deficiency	*OGDHL*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	617513	IEM1907
Disorders of creatine metabolism											
Arginine:glycine amidinotransferase deficiency	*GATM*	AR						Treatable	Creatinine (P, U), Guanidino compounds (P, U)	612718	IEM0042
Guanidinoacetate methyltransferase deficiency	*GAMT*	AR						Treatable	Creatinine (P, U), Guanidino compounds (P, U)	612736	IEM0044
Creatine transporter deficiency	*SLC6A8*	XL					Drug-resistant epilepsy		Creatinine (P, U), Guanidino compounds (P, U)	300352	IEM0045
Disorders of mtDNA-encoded oxidative phosphorylation proteins											
Mitochondrial ATP synthase F0 subunit 6 deficiency	*MT-ATP6*	MIT	Neonatal seizures			Burst-suppression pattern	Drug-resistant epilepsy		Lactate (P)	516060	IEM0484
Disorders of mtDNA-encoded tRNA and rRNA											
Mitochondrial tRNA(Cys) deficiency	*MT-TC*	MIT			Myoclonic jerks		Drug-resistant epilepsy		Lactate (P)	590020	IEM0534
Mitochondrial tRNA(Lys) deficiency	*MT-TK*	MIT			Myoclonic epilepsy		Drug-resistant epilepsy		Lactate (P)	590060	IEM0542
Disorders of complex I subunits and assembly factors											
Complex I assembly factor 8 deficiency	*NDUFAF8*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		Lactate (P)	618776	IEM1530
Disorders of complex II subunits and assembly factors											
Succinate dehydrogenase complex assembly factor 1 deficiency	*SDHAF1*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		Lactate (P), Organic acids (U)	252011	IEM0454
Succinate dehydrogenase subunit D deficiency	*SDHD*	AR			Myoclonus		Drug-resistant epilepsy		Catecholamines (P, U)	252011	IEM0452
Disorders of complex IV subunits and assembly factors											
Cytochrome c oxidase assembly factor 15 deficiency	*COX15*	AR	Neonatal seizures				Drug-resistant epilepsy		Lactate (P)	615119;256000	IEM0471
Cytochrome c oxidase subunit 6B1 deficiency	*COX6B1*	AR	Neonatal seizures				Drug-resistant epilepsy		Lactate (P)	220110	IEM0467
Cytochrome c oxidase subunit 8A deficiency	*COX8A*	MIT	Neonatal seizures				Drug-resistant epilepsy		DNA	220110	IEM1145
LRPPRC deficiency	*LRPPRC*	AR	Neonatal seizures			Burst-suppression pattern	Drug-resistant epilepsy		Lactate (P)	220111	IEM0476
COX11 deficiency	*COX11*	AR	Neonatal seizures				Drug-resistant epilepsy		Lactate (P)	603648	IEM1938
Disorders of coenzyme Q10 biosynthesis											
Coenzyme Q2 polyprenyltranferase deficiency	*COQ2*	AR					Drug-resistant epilepsy		Lactate (P), CoQ10 (M, P, WBC)	609825;607426	IEM0620
Coenzyme Q4 deficiency	*COQ4*	AR					Drug-resistant epilepsy		Lactate (P), CoQ10 (M, P, WBC)	616276	IEM0621
Coenzyme Q6 monooxygenase deficiency	*COQ6*	AR					Drug-resistant epilepsy		DNA	614650	IEM0622
Disorders of lipoic acid and iron-sulfur metabolism											
Lipoyltransferase 2 deficiency	*LIPT2*	AR				EEG, abnormal	Drug-resistant epilepsy		Amino acids (CSF, P), Lactate (P), Protein bound lipoic acid (FB)	617668	IEM0192
BOLA3 deficiency	*BOLA3*	AR			Seizures (myoclonic)				Amino acids (CSF, P), Lactate (P)	614299	IEM0196
Ferredoxin reductase deficiency	*FDXR*	AR					Drug-resistant epilepsy		Complexes I - IV activity (muscle), DNA	617717	IEM0203
Disorders of mitochondrial nucleotide pool maintenance											

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
SAMHD1 deficiency Aicardi-Goutières syndrome type 5	*SAMHD1*	AR			Exaggerated startle reaction		Drug-resistant epilepsy		ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	612952	IEM0031
Mitochondrial DNA polymerase gamma catalytic subunit deficiency	*POLG*	AD, AR				RHADS	Epilepsy, intractable		Lactate (P), Organic acids (U), ASAT/ALAT (P)	203700	IEM0491
TWINKLE mitochondrial DNA helicase deficiency	*TWNK*	AR					Drug-resistant epilepsy		DNA	271245;616138	IEM0495
Disorders of mitochondrial aminoacyl-tRNA synthetases											
Mitochondrial arginyl-tRNA synthetase deficiency	*RARS2*	AR			Myoclonic epilepsy		Drug-resistant epilepsy		Lactate (P)	611523	IEM0553
Mitochondrial asparaginyl-tRNA synthetase deficiency	*NARS2*	AR					Drug-resistant epilepsy		Lactate (P)	616239	IEM0554
Mitochondrial cysteinyl-tRNA synthetase deficiency	*CARS2*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		Lactate (P)	616672	IEM0556
Mitochondrial phenylalanyl-tRNA synthetase deficiency	*FARS2*	AR					Drug-resistant epilepsy		Lactate (P)	614946	IEM0562
Mitochondrial valyl-tRNA synthetase deficiency	*VARS2*	AR			Seizures (myoclonic)				Lactate (P)	615917	IEM0565
Mitochondrial prolyl-tRNA synthetase deficiency	*PARS2*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Epilepsy, refractory		Lactate (P)	612036	IEM1078
Disorders of the mitoribosome											
GUF1 deficiency	*GUF1*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Epilepsy, refractory		DNA	617065	IEM1080
Mitochondrial rRNA methyltransferase 2 deficiency	*MRM2*	?					Epilepsy, refractory		Citrulline (P)		IEM1220
Mitochondrial ribosomal small subunit 39 deficiency	*PTCD3*	AR			Myoclonus		Drug-resistant epilepsy		Lactate (P)	619057	IEM1540
Disorders of mitochondrial shuttles and carriers											
Mitochondrial glutamate transporter deficiency	*SLC25A22*	AR			Myoclonic epilepsy	EEG, abnormal	Drug-resistant epilepsy		DNA	609304	IEM0166
Mitochondrial dicarboxylate transporter deficiency	*SLC25A10*	AR		Infantile spasms		EEG, abnormal	Drug-resistant epilepsy		DNA	606794	IEM1139
Cytosolic malate dehydrogenase deficiency	*MDH1*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		Lactate (P)	618959	IEM1541
Disorders of mitochondrial protein import											
TIMM50 deficiency	*TIMM50*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		Lactate (P), Organic acids (U)	607381	IEM0588
Disorders of mitochondrial protein quality control											
HSPE1 deficiency	*HSPE1*	AD		Infantile spasms			Drug-resistant epilepsy		Pyridoxal 5'-phosphate, PLP (CSF)	600141	IEM1221
LIPID METABOLISM AND TRANSPORT (n=13)											
Disorders of peroxisomal fatty acid oxidation											
X-linked adrenoleukodystrophy and adrenomyeloneuropathy	*ABCD1*	XL				EEG, abnormal		Treatable	VLCFA (P)	300100	IEM0883
Disorders of eicosanoid metabolism											
Leukotriene C4 synthase deficiency	*LTC4S*	AR				EEG, abnormal	Drug-resistant epilepsy		Leucotriens (P, U,, CSF)	246530	IEM1179
Disorders of glycerolipid metabolism											
Seipin deficiency	*BSCL2*	AR			Myoclonus		Drug-resistant epilepsy		ASAT/ALAT (P), Triglyceride (S), Insulin (S)	615924;269700	IEM1054
Disorders of glycerophospholipid metabolism											
Phosphatidylinositol 3,5-bisphosphate-5-phosphatase deficiency	*FIG4*	AD				EEG, abnormal	Drug-resistant epilepsy		DNA	612577;611228	IEM0690

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Synaptojanin 1 deficiency	*SYNJ1*	AR				EEG, abnormal	Seizures, refractory		Lactate (P)	617389	IEM0693
Fatty Acyl-CoA reductase 1deficiency	*FAR1*	AR					Drug-resistant epilepsy		Plasmalogens (RBC)	616154	IEM0881
Peroxin 5 deficiency Zellweger	*PEX5*	AR		Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		ASAT/ALAT (P), VLCFA (P), Pipecolic acid (P, U)	214110	IEM0892
Phospholipase A2 group 6 deficiency	*PLA2G6*	AR				High-amplitude 16- to 24-Hz activity	Drug-resistant epilepsy		DNA	256600	IEM0669
Phosphatidylinositol 4,5-bisphosphate phospholipase C β1 deficiency	*PLCB1*	AR		Infantile spasms		Hypsarrhythmia (EEG) EEG, abnormal	Seizures, refractory		DNA	613722	IEM0707
Choline kinase, alpha deficiency	*CHKA*	AR					Drug-resistant epilepsy		DNA	620023	IEM1936
Disorders of sphingolipid synthesis and recycling											
Ceramide synthase 2 deficiency	*CERS2*	AD			Myoclonic epilepsy		Drug-resistant epilepsy		DNA	606920	IEM1176
Ceramide synthase 1 deficiency	*CERS1*	AR			Myoclonic epilepsy		Drug-resistant epilepsy		DNA	616230	IEM1175
Disorders of bile acid metabolism											
Sterol 27-hydroxylase deficiency Cerebrotendinous xanthomatosis	*CYP27A1*	AR				Evokedpotentials+/-, abnormal (EEG)		Treatable	Lipid panel (S); Sterols (P); Cholestane pentol glucuronide (U); 25-Hydroxy-Vitamin D (P)	213700	IEM0782
METABOLISM OF HETEROCYCLIC COMPOUNDS (n=17)											
Disorders of pyrimidine metabolism											
Pyrimidine nucleoside transporter deficiency	*SLC28A1*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		Purines and pyrimidines (U, P)	606207	IEM1089
CAD trifunctional protein deficiency	*CAD*	AR						Treatable	Purines and pyrimidines (U, P)	616457	IEM0001
Disorders of purine metabolism											
Adenylosuccinate lyase deficiency	*ADSL*	AR	Neonatal seizures				Drug-resistant epilepsy		Purines (CSF, U)	103050;608222	IEM0009
Disorders of ectonucleotide and nucleic acid metabolism											
3' Repair exonuclease 1 deficiency Aicardi-Goutières syndrome type 1	*TREX1*	AR			Exaggerated startle reaction				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 lysophosphatidylcholine	225750	IEM0026
Ribonuclease H2 subunit B deficiency Aicardi-Goutières syndrome type 2	*RNASEH2B*	AR		Infantile spasms	Myoclonic seizures				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	610181	IEM0027
Ribonuclease H2 subunit C deficiency Aicardi-Goutières syndrome type 3	*RNASEH2C*	AR		Infantile spasms	Myoclonic seizures				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	610329	IEM0028
Ribonuclease H2 subunit A deficiency Aicardi-Goutières syndrome type 4	*RNASEH2A*	AR		Infantile spasms	Myoclonic seizures				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	610333	IEM0029
RNA-specific adenosine deaminase deficiency Aicardi-Goutières syndrome type 6	*ADAR*	AR			Exaggerated startle reaction				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF)	615010	IEM0032
MDA5 superactivity Aicardi-Goutières syndrome type 7	*IFIH1*	AD			Exaggerated startle reaction				ASAT/ALAT (P), Blood count (B), Interferon-alpha (B), Pterins (CSF), C26:0 fatty acids (P)	615846	IEM0033
PRUNE1 deficiency	*PRUNE1*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	617481	IEM1481
Disorders of non-mitochondrial tRNA processing and aminoacyl-tRNA synthetases											
CLP1 deficiency	*CLP 1*	AR					Seizures, refractory		DNA	615803	IEM1284
DALRD3 deficiency	*DALRD3*	AR			Seizures (myoclonic)				DNA	618910	IEM1287
Alanyl-tRNA synthetase 1 deficiency	*AARS1*	AR			Seizures (myoclonic)		Seizures, refractory		DNA	613287;616339	IEM1301
Glutaminyl-tRNA synthetase 1 deficiency	*QARS1*	AR					Drug-resistant epilepsy		DNA	615760	IEM1306
AIMP2/p38 deficiency	*AIMP2*	AR				EEG, abnormal	Seizures, intractable		DNA	618006	IEM1321

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Disorders of heme degradation and bilirubin metabolism											
UDP-glucuronosyltransferase A1 deficiency	*UGT1A1*	AR					Drug-resistant epilepsy		Bilirubin (P)	218800;606785	IEM0802
NADH-cytochrome b5 reductase deficiency	*CYB5R3*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		DNA	250800	IEM0798
COMPLEX MOLECULE AND ORGANELLE METABOLISM (n=87)											
Disorders of N-linked protein glycosylation											
PMM2-CDG Phosphomannomutase 2 deficiency	*PMM2*	AR	Neonatal seizures					Treatable	ASAT/ALAT (P), Lipid panel (S), Sialotransferrins (S), Albumin (S), Factor XI (B),	601785	IEM0908
DPAGT1-CDG UDP-GlcNAc:Dol-P-GlcNac-P transferase deficiency	*DPAGT1*	AR	Neonatal seizures				Drug-resistant epilepsy		Sialotrasferrins (S)	608093	IEM0910
ALG13-CDG UDP-N-acetylglucosamine transferase catalytic subunit deficiency	*ALG13*	XL	Neonatal seizures				Epilepsy, refractory		Sialotrasferrins (S); Coagulation factors (P)	300884	IEM0912
ALG14-CDG UDP-N-acetylglucosamine transferase subunit deficeincy	*ALG14*	AR	Neonatal seizures				Drug-resistant epilepsy		Sialotrasferrins (S)	616227;612866	IEM0913
ALG1-CDG Mannosyltransferase 1 deficiency	*ALG1*	AR	Neonatal seizures				Drug-resistant epilepsy		Sialotransferrins (S), IGG (P), B cells, circulating (blood), Protein C (S)	608540	IEM0914
ALG2-CDG Mannosyltransferase 2 deficiency	*ALG2*	AR	Abnormal jitter				Drug-resistant epilepsy		Factor XI (B), Sialotrasferrins (S)	607906	IEM0915
ALG11-CDG Mannosyltransferase 4-5 deficiency	*ALG11*	AR	Neonatal seizures				Drug-resistant epilepsy		Factor XI (B), Antithrombin III (P), Sialotrasferrins (S)	613661	IEM0916
ALG3-CDG Mannosyltransferase 6 deficiency	*ALG3*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		Antithrombin III (P), Protein S (S), Sialotrasferrins (S)	601110	IEM0918
ALG6-CDG Glucosyltransferase 1 deficiency	*ALG6*	AR	Neonatal seizures				Drug-resistant epilepsy		Factor XI (B), Sialotrasferrins (S), ASAT/ALAT (P)	603147	IEM0921
ALG8-CDG Glucosyltransferase 2 deficiency	*ALG8*	AD, AR	Neonatal seizures				Drug-resistant epilepsy		CK (P), Sialotransferrins (S), Albumin (S), Factor IX and XII (B), Antithrombin III (P)	608104	IEM0922
GCS1-CDG Glucosidase 1 deficiency	*MOGS*	AR	Neonatal seizures			Burst-suppression (EEG)	Drug-resistant epilepsy		ASAT/ALAT (P), Oligosaccharide (U), Sialotransferins (S)	606056	IEM0927
MGAT2-CDG	*MGAT2*	AR					Epilepsy, intractable		ASAT/ALAT (P), CK (P), Sialotransferrins (S), Factor IX and XII (B), Antithrombin III (P)	212066	IEM0931
FCSK-CDG Fucokinase deficiency	*FCSK*	AR	Neonatal seizures				Drug-resistant epilepsy		Sialotrasferrins (S)	618324	IEM1068
OSTC-CDG Oligosaccharyltransferase complex deficiency	*OSTC*	AR	Neonatal seizures	Infantile spasms			Drug-resistant epilepsy		Sialotrasferrins (S)	619023	IEM1255
Disorders of O-linked protein glycosylation											
POMT1-CDG O-Mannosyltransferase 1 deficiency	*POMT1*	AR	Neonatal seizures				Drug-resistant epilepsy		CK (P), Sialotrasferrins (S)	236670;613555;609308	IEM0933
POMT2-CDG O-Mannosyltransferase 2 deficiency	*POMT2*	AR	Neonatal seizures				Drug-resistant epilepsy		CK (P), DNA	613150;613156;613158	IEM0934
EOGT-CDG	*EOGT*	AR	Neonatal seizures				Drug-resistant epilepsy		DNA	615297	IEM0966
Disorders of lipid glycosylation											
ST3GAL5-CDG Lactosylceramide alpha-2,3-sialyltransferase deficiency	*ST3GAL5*	AR					Drug-resistant seizures		Lactosylceramide (P), GM3 ganglioside (P)	609056	IEM0987
PIGA-CDG	*PIGA*	XL	Neonatal seizures				Drug-resistant epilepsy		ALP (P), GPI-anchored proteins (WBC, F)	300868;300818	IEM0971
PIGC-CDG	*PIGC*	AR					Seizures, intractable		Flow cytometry of GPI markers (Gr)	615716	IEM0972
PIGQ-CDG	*PIGQ*	AR					Seizures, refractory		ALP (P), DNA	605754	IEM0973
ST3GAL3-CDG	*ST3GAL3*	AR					Drug-resistant epilepsy		DNA	611090	IEM0989
PIGW-CDG	*PIGW*	AR	Neonatal seizures				Drug-resistant epilepsy		Alkaline phosphatase (P); DNA	616025	IEM0976

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
PIGM-CDG	*PIGM*	AR	Neonatal seizures				Drug-resistant epilepsy		Flow cytometry of GPI markers (Gr)	610293	IEM0977
PIGO-CDG	*PIGO*	AR					Drug-resistant epilepsy		ALP (P), GPI-anchored proteins (WBC, F)	614749	IEM0980
PIGU-CDG	*PIGU*	AR					Drug-resistant epilepsy		DNA	618590	IEM1553
PIGT-CDG	*PIGT*	AR					Drug-resistant epilepsy		ALP (P), GPI-anchored proteins (WBC, F)	615398	IEM0982
GPAA1-CDG	*GPAA1*	AR					Drug-resistant epilepsy		DNA	617810	IEM0983
PGAP1-CDG	*PGAP1*	AR					Drug-resistant epilepsy		DNA	615802	IEM0984
PGAP2-CDG	*PGAP2*	AR					Drug-resistant epilepsy		ALP (P), GPI-anchored proteins (WBC, F)	614207	IEM0986
PIGS-CDG	*PIGS*	AR					Drug-resistant epilepsy		DNA	618143	IEM1059
Disorders of multiple glycosylation pathways											
UGDH-CDG	*UGDH*	AR		Infantile spasms		Hypsarrhythmia (EEG) EEG, abnormal	Seizures, refractory		DNA	618792	IEM1556
UGP2-CDG	*UGP2*	AR		Infantile spasms		Hypsarrhythmia (EEG) EEG, abnormal	Seizures, refractory		DNA	618744	IEM1557
Disorders of mitochondrial membrane biogenesis and remodeling											
SERAC1 deficiency MEGDEL Syndrome	*SERAC1*	AR	Neonatal seizures			Burst-suppression pattern	Drug-resistant epilepsy		Lactate (P); Organic acids (U); Filipin staining	614739	IEM0582
Cardiolipin synthase 1 deficiency	*CRLS1*	AR			Myoclonic jerks		Drug-resistant epilepsy		DNA	608188	IEM1912
Disorders of mitochondrial and peroxisomal dynamics											
UGO-1 like protein deficiency	*SLC25A46*	AR			Myoclonus		Drug-resistant epilepsy		Lactate (P), Organic acids (U)	616505	IEM0576
Trafficking kinesin-binding protein 1 deficiency	*TRAK1*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		DNA	608112	IEM0614
SPATA5 deficiency	*SPATA5*	AR					Epilepsy, intractable		DNA	616577	IEM1380
Peroxisomal biogenesis disorders											
Peroxin 1 deficiency Zellweger	*PEX1*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	234580;214100;601539	IEM0889
Peroxin 2 deficiency Zellweger	*PEX2*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614866;614867	IEM0890
Peroxin 3 deficiency Zellweger	*PEX3*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	617370;614882	IEM0891
Peroxin 6 deficiency Zellweger	*PEX6*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614862;614863;616617	IEM0893
Peroxin 10 deficiency Zellweger	*PEX10*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614870;614871	IEM0894
Peroxin 14B deficiency Zellweger	*PEX11B*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614920	IEM0895
Peroxin 12 deficiency Zellweger	*PEX12*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614859;266510	IEM0896
Peroxin 13 deficiency Zellweger	*PEX13*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614883;614885	IEM0897
Peroxin 14 deficiency Zellweger	*PEX14*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614887	IEM0898
Peroxin 16 deficiency Zellweger	*PEX16*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614876;614877	IEM0899
Peroxin 19 deficiency Zellweger	*PEX19*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pipecolic acid (P)	614886	IEM0900

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Peroxin 26 deficiency Zellweger	*PEX26*	AR	Neonatal seizures	Infantile spasms		Hypsarrhythmia	Drug-resistant seizures		VLCFA (P), Pristanic acid (S), Phytanic acid (S), AST/ALT (P), Plasmalogens (RBC), Pipecolic acid (S, U)	614872;614873	IEM0901
Disorders of vesicular trafficking											
GOSR2-CDG	*GOSR2*	AR			Myoclonic epilepsy		Drug-resistant epilepsy		DNA	614018	IEM1062
TRAPPC12 deficiency	*TRAPPC12*	AR		Infantile spasms	Myoclonus	Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	617669	IEM1412
ARFGEF2 deficiency	*ARFGEF2*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	608097	IEM1422
AP3B2 deficiency	*AP3B2*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	617276	IEM1425
YIF1B deficiency	*YIF1B*	AR					Drug-resistant epilepsy		DNA	619125	IEM1517
VPS16 deficiency	*VPS16*	AD, AR			Myoclonic jerks		Drug-resistant epilepsy		Iron (BR)	619291	IEM1656
GET3-CDG Arsenical pump-driving ATPase 1	*GET3*	AR				EEG, abnormal	Drug-resistant epilepsy		Sialotransferins (S)	601913	IEM1918
Disorders of sphingolipid degradation											
Beta-hexosaminidase subunit alpha deficiency Tay-Sachs disease	*HEXA*	AR			Myoclonic seizures	Fast central spikes	Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	272800	IEM0836
Beta-hexosaminidase subunit beta deficiency Sandhoff disease	*HEXB*	AR			Myoclonic seizures		Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	268800	IEM0837
Formyl-glycine generating enzyme deficiency Multiple sulfatase deficiency	*SUMF1*	AR	Neonatal seizures				Drug-resistant epilepsy		Sulfatide (U), Glycosaminoglycans (U)	272200	IEM0843
Beta-galactosidase-1 deficiency, GM1 gangliosis	*GLB1*	AR			Myoclonic seizures		Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	253010	IEM0865
Acid ceramidase deficiency, primary neurologic phenotype Farber disease	*ASAH1*	AR			Myoclonic epilepsy		Drug-resistant epilepsy		Lysosomal enzymes (DBS)	228000	IEM0846
Combinded saposin deficiency (all forms)	*PSAP*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		Sulfatides (U), Protein (CSF), Lysosomal enzymes (DBS)	611721	IEM0847
Disorders of glycoprotein degradation											
Alpha-neuraminidase deficiency Sialidosis	*NEU1*	AR			Myoclonic epilepsy	Rhythmic vertex-positive spikes	Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	256550	IEM0848
Cathepsin A deficiency Galactosialidosis	*CTSA*	AR			Myoclonus		Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	256540	IEM0849
Alpha-N-acetylgalactosaminidase deficiency Schindler disease type I	*NAGA*	AR			Myoclonic epilepsy		Drug-resistant epilepsy		Oligosaccharides (U), Lysosomal enzymes (DBS)	609241	IEM0852
Neuronal ceroid lipofuscinosis											
Tripeptidyl-peptidase 1 deficiency	*TPP1*	AR			Myoclonic epilepsy Seizures (myoclonic)	Vanishing electroencephalogram		Treatable	Lysosomal tripeptidyl-peptidase-1 (DBS)	204500	IEM0820
CLN3 disease	*CLN3*	AR				EEG, abnormal	Drug-resistant epilepsy		Peripheral smear, DNA	204200	IEM0821
CLN4 disease	*DNAJC5*	AD			Myoclonus	EEG, abnormal	Drug-resistant epilepsy		DNA	162350	IEM0822
Palmitoyl-protein thioesterase 1 deficiency	*PPT1*	AR			Myoclonic epilepsy Seizures (myoclonic)	Marked photosensitivity	Drug-resistant epilepsy		Lysosomal enzymes (DBS)	256730	IEM0689
CLN5 disease	*CLN5*	AR			Myoclonic epilepsy	EEG, abnormal	Drug-resistant epilepsy		DNA	256731	IEM0823
CLN6 disease	*CLN6*	AR			Myoclonic epilepsy	EEG, abnormal	Drug-resistant epilepsy		DNA	601780	IEM0824
CLN7 disease	*MFSD8*	AR			Myoclonic epilepsy Myoclonus	EEG, abnormal	Drug-resistant epilepsy		DNA	610951	IEM0825
CLN8 disease	*CLN8*	AR			Myoclonic epilepsy	EEG, abnormal	Drug-resistant epilepsy		DNA	600143	IEM0826
Cathepsin D deficiency	*CTSD*	AR			Myoclonus	EEG, abnormal	Drug-resistant epilepsy		Lysosomal enzymes (DBS)	610127	IEM0827

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Progranulin deficiency	*GRN*	AD, AR				EEG, abnormal	Drug-resistant epilepsy		DNA	614706	IEM0828
ATP13A2 deficiency Kufor-Rakeb syndrome	*ATP13A2*	AR			Myoclonus		Drug-resistant epilepsy		DNA	606693	IEM0829
Cathepsin F deficiency	*CTSF*	AR					Drug-resistant epilepsy		DNA	603539	IEM0830
CLN14 disease	*KCTD7*	AR			Myoclonic epilepsy Seizures (myoclonic)	EEG, abnormal	Drug-resistant epilepsy		DNA	611726	IEM0831
Disorders of autophagy											
Spastizin deficiency	*ZFYVE26*	AR			Myoclonus		Drug-resistant epilepsy		DNA	270700	IEM0815
CHMP2B deficiency	*CHMP2B*	AD			Myoclonus		Drug-resistant epilepsy		DNA	600795	IEM1244
Hereditary spastic paraplegia CHMP3	*CHMP3*	AR			Myoclonus		Drug-resistant epilepsy		DNA	610052	IEM1943
Other disorders of complex molecule degradation											
Niemann-Pick disease type C1	*NPC1*	AR					Drug-resistant epilepsy		Oxysterols (P); Filipin staining (F)	257220	IEM0870
Niemann-Pick disease type C2	*NPC2*	AR					Drug-resistant epilepsy		Oxysterols (P); Filipin staining (F)	607625	IEM0871
Glucocerebrosidase receptor deficiency	*SCARB2*	AR			Seizures (myoclonic)		Drug-resistant epilepsy		Lysosomal enzymes (DBS)	254900	IEM0875
Alpha-glucosidase deficiency Pompe disease	*GAA*	AR				EEG, abnormal	Drug-resistant epilepsy		ASAT/ALAT (P), CK (P), Glycogen (M)	232300	IEM0356
Lysosome-associated membrane protein 2 deficiency Danon disease	*LAMP2*	XL				EEG, abnormal	Drug-resistant epilepsy		ASAT/ALAT (P), CK (P), Glycogen (M)	300257	IEM0367
COFACTOR AND MINERAL METABOLISM (n=33)											
Disorders of tetrahydrobiopterin metabolism											
Autosomal recessive GTP cyclohydrolase 1 deficiency	*GCH1*	AR	Neonatal seizures		Seizures (myoclonic)		Drug-resistant epilepsy		Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	233910	IEM0083
6-Pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	AR			Seizures (myoclonic)	EEG, abnormal		Treatable	Amino acids (P), Pterins (DBS, U, CSF), Biogenic amines (CSF)	261640	IEM0085
Dihydropteridine reductase deficiency	*QDPR*	AR			Seizures (myoclonic)	Generalized slowing (EEG) Spike wave discharges (EEG)	Drug-resistant epilepsy		Amino acids (P); Pterins (DBS,U); DHPR activity (DBS); Biogenic amines (CSF), 5-methyl-THF (CSF)	261630	IEM0087
Disorders of thiamine metabolism											
Thiamine transporter 2 deficiency	*SLC19A3*	AR					Epilepsy, intractable		Lactate (P), Organic acids (U)	606152	IEM0230
Disorders of pantothenate and CoA metabolism											
Pantothenate kinase 2 deficiency	*PANK2*	AR					Drug-resistant epilepsy		Iron (brain), DNA	234200	IEM0246
Mitochondrial coenzyme A transporter deficiency	*SLC25A42*	AR					Drug-resistant epilepsy		Lactate P)	610823	IEM0249
Disorders of pyridoxine metabolism											
Pyridox(am)ine 5´- phosphate oxidase deficiency	*PNPO*	AR	Neonatal seizures				Seizures, pharmacoresistant	Treatable	B6 vitaminers (CSF, P)	610090	IEM0250
Pyridoxal 5'-phosphate binding protein deficiency	*PLPBP*	AR	Neonatal seizures			Burst-suppression (EEG)	Drug-resistant epilepsy	Treatable	Pyridoxal 5'-phosphate, PLP (CSF), B6 vitaminers (CSF, P, U), Homovanillic acid, HVA (CSF)	617290	IEM0251
Alpha-amino adipic semialdehyde (AASA) dehydrogenase deficiency Pyridoxine-dependent seizures	*ALDH7A1*	AR	Neonatal seizures				Drug-resistant epilepsy	Treatable	Pyridoxal 5'-phosphate, PLP (CSF), B6 vitaminers (CSF, P, U), Pipecolic acid (CSF)	266100	IEM0131
Tissue-nonspecific alkaline phosphatase deficiency	*ALPL*	AR, AD	Neonatal seizures						Pyridoxal 5'-phosphate, PLP (CSF), ALP, Calcium (P), Phosphate (P)	241500	IEM0252
Disorders of biotin metabolism											
Biotinidase deficiency	*BTD*	AR						Treatable	Acyglycines (U), Lactate (P), Organic acids (U), Acylcarnitines (P,DBS)	253260	IEM0227

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Holocarboxylase synthetase deficiency	*HLCS*	AR	Neonatal seizures			Burst-suppression pattern	Drug-resistant epilepsy		Organic acids (U), Acylcarnitines (DBS, P), Lactate (P)	253270	IEM0228
Disorders of folate metabolism											
Proton-coupled folate transporter deficiency	*SLC46A1*	AR						Treatable	Folate (S), 5-Methyl-THF (CSF), Blood count (B), Immunoglobulins (S)	229050	IEM0221
Folate receptor alpha deficiency	*FOLR1*	AR	Neonatal seizures		Seizures, myoclonic-astatic	EEG, abnormal		Treatable	5-Methyltetrahydrofolate (CSF), Folate (S)	613068	IEM0222
5,10-methylenetetrahydrofolate reductase deficiency	*MTHFR*	AR		Infantile spasms	Seizures (myoclonic)		Drug-resistant epilepsy	Treatable	Amino acids (P); Homocysteine (P); 5-Methyltetrahydrofolate (CSF), Folate (S)	236250	IEM0223
Dihydrofolate reductase deficiency	*DHFR*	AR						Treatable	LDH (P), Hb (B), Folates (B, CSF)	126060	IEM0225
5,10-Methenyltetrahydrofolate synthetase deficiency	*MTHFS*	AR						Treatable	5-Methyl-THF (CSF)	604197	IEM1035
Disorders of cobalamin metabolism											
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	AR					Drug-resistant epilepsy		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P), Blood count	277380	IEM0211
Methylmalonic aciduria and homocystinuria, cblJ type	*ABCD4*	AR					Drug-resistant epilepsy		Amino acids (P), Organic acids (U), Acylcarnitines (DBS, P), tHcy (P), SAM/SAH (P)	614857	IEM0212
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR					Drug-resistant epilepsy		Homocysteine, total (P), B12 (S), Blood count, Organic acids (U), Acylcarnitines (DBS, P), Amino acids (P)	277400	IEM0213
Methylmalonic aciduria, cblDv2 type	*MMADHC*	AR	Neonatal seizures				Drug-resistant epilepsy		SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	277410	IEM0215
Homocystinuria, cblDv1 type	*MMADHC*	AR	Neonatal seizures				Drug-resistant epilepsy		SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	277410	IEM0215
Methylmalonic aciduria and homocystinuria, cblD type	*MMADHC*	AR	Neonatal seizures				Drug-resistant epilepsy		SAM (P,CSF), Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	277410	IEM0215
Methylmalonic aciduria, cblA type	*MMAA*	AR	Neonatal seizures				Drug-resistant epilepsy		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	251100	IEM0218
Methylmalonic aciduria, cblB type	*MMAB*	AR	Neonatal seizures				Drug-resistant epilepsy		Amino acids (P); Organic acids (U); Acylcarnitines (U, P, DBS); Anion gap	251110	IEM0219
Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*	XL	Neonatal seizures	Infantile spasms			Seizures, intractable		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	309541	IEM0220
Methylmalonic aciduria and homocystinuria due to Ronin deficiency	*THAP11*	AR		Infantile spasms			Seizures, intractable		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	609119	IEM1125
Methylmalonic aciduria and homocystinuria due to ZNF143 deficiency	*ZNF143*	AR		Infantile spasms			Seizures, intractable		Amino acids (P), Organic acids (U), Acylcarnitines (P,DBS)	603433	IEM1256
Disorders of molybdenum cofactor metabolism											
Cyclic pyranopterin monophosphate synthase deficiency MoCo A deficiency	*MOCS1*	AR	Neonatal seizures		Myoclonus	Burst-suppression pattern	Drug-resistant epilepsy		Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)	603707	IEM0275
Molybdopterin synthase deficiency MoCo B deficiency	*MOCS2*	AR	Neonatal seizures		Myoclonus	Burst-suppression pattern	Drug-resistant epilepsy		Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)	603708	IEM0276
Gephyrin deficiency MoCo C deficiency	*GPHN*	AR	Neonatal seizures		Myoclonic seizures	Burst-suppression pattern	Drug-resistant epilepsy		Uric acid (P), Sulfite (U), a-aminosemialdehyde (CSF), PLP (CSF), Purines and pyrimidines (U), Amino acids (P)	603930	IEM0277
Other disorders of vitamin metabolism											
Vitamin D 1-α-hydroxylase deficiency	*CYP27B1*	AR					Drug-resistant epilepsy	Treatable	Amino acids (U), ALP (P), Calcium (P), Phosphate (P), PTH (S), 1,25-Dihydroxy vitamin D (S)	264700	IEM0266
Disorders of copper metabolism											
Copper-transporting ATPase subunit alpha deficiency Menkes disease	*ATP7A*	AR	Neonatal seizures			Burst-suppression pattern	Drug-resistant epilepsy		Copper (S, U), Ceruloplasmin (S)	309400	IEM0280
METABOLIC CELL SIGNALLING (n=25)											
Monoamine neurotransmission											
Aromatic L-amino acid decarboxylase (AADC) deficiency	*DDC*	AR						Treatable	Biogenic amines (CSF, P); Vanillinlactic acid (U)	608643	IEM0077
Tyrosine hydroxylase deficiency	*TH*	AR						Treatable	Prolactin (P); Biogenic amines (CSF)	191290	IEM0076

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Disorder (n=256)	Gene	Inheritance	Neonatal seizures	Infantile spasms	Myoclonic seizures	Specific EEG pattern	Drug-resistant epilepsy	Treatable epilepsies	Laboratory investigations	OMIM	IEMcode
Dopamine transporter deficiency	*SLC6A3*	AR	Neonatal irritability				Drug-resistant epilepsy		Organic acids (U), Biogenic amines (CSF)	613135;126455	IEM0080
Gamma-aminobutyric acid neurotransmitter disorders											
GABA transaminase deficiency	*ABAT*	AR					Drug-resistant epilepsy		GABA free (CSF), β-Alanine (CSF), Homocarnosine (CSF)	137150;613163	IEM0152
Succinic semialdehyde dehydrogenase deficiency SSADH 4-Hydroxybutyric aciduria	*ALDH5A1*	AR				EEG, abnormal Generalized slowing (EEG)	Drug-resistant epilepsy		Organic acids (U)	271980;610045	IEM0153
GABA type A receptor γ2 subunit deficiency	*GABRG2*	AD			Seizures (myoclonic)				DNA	611277	IEM0159
GABA type A receptor δ subunit deficiency	*GABRD*	AR			Seizures (myoclonic)				DNA	137163	IEM1113
Glutamate neurotransmitter disorders											
Ionotropic glutamate receptor NMDA type subunit 1 dysregulation	*GRIN1*	AD, AR			Myoclonus	EEG, abnormal	Drug-resistant epilepsy		DNA	614254;617820	IEM0169
Ionotropic glutamate receptor NMDA type subunit 2A dysregulation	*GRIN2A*	AD				EEG, abnormal	Drug-resistant epilepsy		DNA	245570	IEM0170
Ionotropic glutamate receptor NMDA type subunit 2B dysregulation	*GRIN2B*	AD		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	616139;613970	IEM0171
Ionotropic glutamate receptor NMDA type subunit 2D superactivity	*GRIN2D*	AD		Infantile spasms		Hypsarrhythmia (EEG) EEG, abnormal	Epilepsy, refractory		DNA	617162	IEM0172
Ionotropic glutamate receptor AMPA type subunit 3 deficiency	*GRIA3*	XL			Myoclonic jerks		Drug-resistant epilepsy		DNA	300699	IEM0173
Ionotropic glutamate receptor AMPA type subunit 4 dysregulation	*GRIA4*	AD					Drug-resistant epilepsy		DNA	617864	IEM0174
Thorase deficiency	*ATAD1*	AR		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		GABA free (CSF)	618011	IEM0175
Intellectual developmental disorder (GRIK2)	*GRIK2*	AR			Myoclonus		Drug-resistant epilepsy		DNA	611092	IEM1901
Glycine neurotransmitter disorders											
Glycine receptor subunit alpha 1 deficiency	*GLRA1*	AD, AR			Myoclonus	EEG, abnormal	Drug-resistant epilepsy		DNA	149400	IEM0189
Glycine receptor subunit beta deficiency	*GLRB*	AR			Myoclonus		Drug-resistant epilepsy		DNA	614619	IEM0190
Intellectual developmental disorder, X-linked, syndromic, Pilorge type	*GLRA2*	XL				EEG, abnormal	Drug-resistant epilepsy		DNA	301076	IEM1922
Disorders of the synaptic vesicle cycle											
TBC1D24 deficiency	*TBC1D24*	AD, AR					Drug-resistant epilepsy		DNA	614617;616044;615338;220500;608105;605021	IEM1453
KIF5A deficiency	*KIF5A*	AD			Seizures (myoclonic)		Drug-resistant epilepsy		DNA	602821	IEM1455
Dynamin 1 deficiency	*DNM1*	AD		Infantile spasms		Hypsarrhythmia (EEG)	Drug-resistant epilepsy		DNA	616346	IEM1458
NAPB deficiency	*NAPB*	AR					Drug-resistant epilepsy		DNA	611270	IEM1460
Syntaxin-binding protein 1 deficiency	*STXBP1*	AD		Infantile spasms		Hypsarrhythmia (EEG) Burst-suppression (EEG)	Seizures, refractory		DNA	612164	IEM1465
Synaptic vesicle glycoprotein 2A deficiency	*SV2A*	AD					Epilepsy, intractable		N-acetylaspartate (MRS), Lactate (MRS)	185860	IEM1466
SORCS receptor 3 deficiency	*SORCS3*	AR					Drug-resistant epilepsy		DNA	606285	IEM1480