EPILEPSY GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABAT	66,2	100%	92%	GABA-transaminase deficiency, 613163
ABCC8	83,1	100%	97%	Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800 Diabetes mellitus, transient neonatal 2, 610374 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176
ACY1	80,1	100%	97%	Aminoacylase 1 deficiency, 609924
ADSL	127,3	100%	98%	Adenylosuccinase deficiency, 103050
ALDH7A1	72,9	94%	92%	Epilepsy, pyridoxine-dependent, 266100
ALG13	58,2	92%	84%	Congenital disorder of glycosylation, type Is, 300884
AMACR	83,9	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	123,9	100%	99%	Glycine encephalopathy, 605899
ARHGEF9	50,9	98%	88%	Epileptic encephalopathy, early infantile, 8, 300607
ARX	31,5	76%	62%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASAH1	102,4	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950

ATP1A2	100,1	100%	98%	Migraine, familial hemiplegic, 2, 602481 Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481
ATP6AP2	31,7	93%	74%	Mental retardation, X-linked, with epilepsy, 300423
ATP7A	60,3	100%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	73,2	100%	98%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
BOLA3	63,4	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BTD	141,7	100%	100%	Biotinidase deficiency, 253260
CACNA1A	81	98%	90%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CASK	51,4	98%	94%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422 Mental retardation, with or without nystagmus, 300422
CDKL5	67,4	99%	96%	Epileptic encephalopathy, early infantile, 2, 300672 Angelman syndrome-like, 105830
CHD2	128	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	115,4	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	93,2	99%	98%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	142,5	95%	93%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	126,8	98%	95%	Hypomagnesemia 3, renal, 248250
CLDN19	77,7	100%	97%	Hypomagnesemia 5, renal, with ocular involvement, 248190

CLN3	85,1	100%	99%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139,7	100%	90%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	63,7	98%	81%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	133,8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	139,2	100%	99%	Hypomagnesemia 6, renal, 613882
CNTN2	86,3	100%	99%	Epilepsy, familial adult myoclonic, 5, 615400
CNTNAP2	100,2	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 {Autism susceptibility 15}, 612100 Pitt-Hopkins like syndrome 1, 610042
COQ2	75,5	99%	96%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
CPA6	122,8	100%	100%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPS1	107,7	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	109,4	92%	91%	Myopathy due to CPT II deficiency, 255110 CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CSTB	166,9	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	96,2	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	109,8	96%	82%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	61,7	100%	93%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	57,7	96%	86%	D-2-hydroxyglutaric aciduria, 600721

DCX	60,6	100%	94%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DCX	60,6	100%	94%	Pentosuria, 260800 (1)
DEPDC5	111,4	100%	99%	Epilepsy, familial focal, with variable foci, 604364
DLAT	107	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	69,7	91%	79%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DPYD	120,4	98%	96%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DYNC1H1	117,4	99%	96%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	138,5	99%	99%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	104,4	100%	100%	No OMIM phenotype Epileptic Encephalopathy (Veeramah (2013) Epilepsia 54: 1270-1281)
EGF	111,7	99%	98%	Hypomagnesemia 4, renal, 611718
EHMT1	100,4	98%	95%	Kleefstra syndrome, 610253
EPM2A	63,3	87%	81%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FARS2	97,6	98%	94%	Combined oxidative phosphorylation deficiency 14, 614946
FGD1	47	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400

FLNA	62,1	99%	91%	Heterotopia, periventricular, 300049 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, ED variant, 300537 FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048
FOLR1	82,4	100%	98%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	91,1	93%	80%	Rett syndrome, congenital variant, 613454
FOXRED1	95,2	100%	97%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXYD2	64,3	99%	80%	Hypomagnesemia-2, renal, 154020
GABRA1	120,6	100%	95%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136 {Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRG2	124,6	97%	93%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681 Febrile seizures, familial, 8, 611277
GAMT	103,3	98%	94%	Cerebral creatine deficiency syndrome 2, 612736
GCK	81	100%	97%	MODY, type II, 125851 Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, gestational, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485 Diabetes mellitus, permanent neonatal, 606176
GCSH	14,8	52%	39%	Glycine encephalopathy, 605899
GLDC	60,1	98%	85%	Glycine encephalopathy, 605899

GLRA1	113,9	100%	98%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	123,8	100%	99%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	111,8	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	109,4	100%	99%	Epileptic encephalopathy, early infantile, 17, 615473
GPC3	52,7	100%	97%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	120	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GRIA3	56,7	91%	84%	Mental retardation, X-linked 94, 300699
GRIN1	83	99%	93%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	132,6	99%	98%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	140,6	99%	98%	Mental retardation, autosomal dominant 6, 613970
GRN	115,7	100%	99%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
GRN	115,7	100%	99%	Myasthenia, limb-girdle, familial, 254300
HADH	93,4	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HDAC4	70,2	95%	88%	Brachydactyly-mental retardation syndrome, 600430
HLCS	141,8	100%	100%	Holocarboxylase synthetase deficiency, 253270

HNRNPU	110,9	97%	97%	No OMIM phenotype Developmental delay and intellectual disability (King (2014) Genome Res 24, 673) Infantile spasms (Du (2014) BMC Med Genet 15, 62) Speech delay, seizures & CNS anomalies (Caliebe (2010) Eur J Med Genet 53, 179) Seizures (Ballif (2012) Hum Genet 131, 145) Epileptic encephalopathy (Mefford (2011) Ann Neurol 70, 974) Intellectual disability & seizures (Thierry (2012) Am J Med Genet A 158A, 1633) Thin corpus callosum, psychomotor delay & seizures (Selmer (2012) Eur J Med Genet 55,715)
HSD17B10	54,1	95%	91%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 Mental retardation, X-linked syndromic 10, 300220 Mental retardation, X-linked 17/31, microduplication, 300705
HSD17B4	96,9	100%	99%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	106,8	100%	94%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	67,1	100%	99%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IQSEC2	42,2	86%	69%	Mental retardation, X-linked 1, 309530
KANSL1	49,7	78%	65%	Koolen-De Vries syndrome, 610443
KCNA1	112,2	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNJ10	151,4	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNJ11	144	100%	100%	Hyperinsulinemic hypoglycemia, familial, 2, 601820 Diabetes, permanent neonatal, 606176 Diabetes mellitus, permanent neonatal, with neurologic features, 606176 {Diabetes mellitus, type 2, susceptibility to}, 125853 Diabetes mellitus, transient neonatal, 3, 610582
KCNMA1	82,6	99%	95%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	75,6	100%	98%	Seizures, benign neonatal, 1, 121200 Myokymia, 121200 Epileptic encephalopathy, early infantile, 7, 613720

KCNQ3	100,7	100%	98%	Seizures, benign neonatal, type 2, 121201
KCNT1	75	99%	93%	Epileptic encephalopathy, early infantile, 14, 614959 Epilepsy, nocturnal frontal lobe, 5, 615005
KCTD7	106,3	92%	87%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	54,7	100%	93%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534 -3
LGI1	136,6	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	101,7	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
MBD5	140,8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MECP2	89,1	97%	90%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260
MED12	62,1	94%	85%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MEF2C	113,4	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443 Chromosome 5q14.3 deletion syndrome, 613443 (4)
MFSD8	120	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MOCS1	73,9	100%	96%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	99,4	100%	100%	Molybdenum cofactor deficiency, type B, 252150

MTHFR	93,8	100%	98%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTOR	99	99%	98%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798 -3
NDUFA1	116,6	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	103,4	99%	82%	Mitochondrial complex I deficiency, 252010
NDUFAF1	116,9	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	53,6	100%	95%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	131,2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	81,2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	129,6	100%	100%	Mitochondrial complex 1 deficiency, 252010
NDUFB3	2,3	0%	0%	Mitochondrial complex I deficiency, 252010
NDUFB9	106,8	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS1	80,4	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	123,1	100%	97%	Mitochondrial complex I deficiency, 252010
NDUFS3	153,9	100%	100%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	127,6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	118,7	90%	77%	Complex I, mitochondrial respiratory chain, deficiency of, 252010
NDUFV1	63,4	100%	92%	Mitochondrial complex I deficiency, 252010

NDUFV2	124,6	100%	100%	Mitochondrial complex I deficiency, 252010
NECAP1	103,4	100%	100%	?Epileptic encephalopathy, early infantile,21, 615833
NEDD4L	111,7	100%	100%	No OMIM phenotype Epilepsy, photosensitive generalized (Dibbens (2007) Genes Brain Behav 6, 750) Impaired ENaC regulation (Fouladkou (2004) Am J Physiol Renal Physiol 287, F550)
NHLRC1	109,3	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	123,9	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	87,4	100%	100%	Mitochondrial complex I deficiency, 252010
OFD1	37	88%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
OPHN1	48,4	99%	88%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	51,1	100%	97%	Mental retardation, X-linked 30/47, 300558
PC	99,2	95%	92%	Pyruvate carboxylase deficiency, 266150
PCDH19	67,3	100%	98%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	61,6	98%	89%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDHB	98,7	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	157,7	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PEX1	124,5	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	73	95%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	133	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510

PEX13	134,6	100%	95%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	92,3	100%	100%	Peroxisome biogenesis disorder 14B, 614920
PEX14	92,3	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	96,8	99%	89%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	104,7	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	107,1	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	141,1	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	85,2	97%	95%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370
PEX6	91,3	94%	85%	Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PHF6	71,5	100%	98%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	84,6	100%	97%	Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	67,1	100%	98%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PIGN	102,9	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	107,5	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PLA2G6	76,5	99%	93%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953
PLCB1	113	100%	98%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	45,9	100%	94%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PNKP	70,4	100%	99%	Epileptic encephalopathy, early infantile, 10, 613402

PNPO	71,8	100%	97%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	90,5	98%	92%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459
PPT1	72	100%	96%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	68,1	100%	99%	Renpenning syndrome, 309500
PRICKLE1	104,2	99%	98%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	74,9	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
RAB39B	108,1	100%	100%	Mental retardation, X-linked 72, 300271
RARS2	83	100%	98%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	90,2	100%	88%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	113	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	161,5	100%	100%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	94,4	96%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	50,9	98%	91%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	113,1	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, AD, 5, 613077 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SAMHD1	122,5	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SCARB2	88,5	100%	95%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900

SCN1A	118,1	99%	98%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Dravet syndrome, 607208 Migraine, familial hemiplegic, 3, 609634 Febrile seizures, familial, 3A, 604403
SCN1B	96,1	94%	93%	Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Atrial fibrillation, familial, 13, 615377
SCN2A	128,1	100%	98%	Seizures, benign familial infantile, 3, 607745 Epileptic encephalopathy, early infantile, 11, 613721
SCN8A	136,3	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SCN9A	113,8	100%	99%	Erythermalgia, primary, 133020 Insensitivity to pain, channelopathy-associated, 243000 Paroxysmal extreme pain disorder, 167400 Febrile seizures, familial, 3B, 613863 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SLC16A1	139,9	100%	99%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021
SLC19A3	110,7	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	77,6	88%	82%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	100,6	88%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC25A22	75,4	100%	92%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	94,3	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC35A2	53,6	100%	98%	Congenital disorder of glycosylation, type IIm, 300896

SLC6A8	4,4	13%	5%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	57,3	98%	90%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMS	13,1	56%	30%	Smith-Magenis syndrome, 182290
SPTAN1	95,4	99%	97%	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	46,7	97%	82%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 -3
ST3GAL3	113,8	100%	100%	Mental retardation, autosomal recessive 12, 611090 Epileptic encephalopathy, early infantile, 15, 615006
STXBP1	93,2	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164 (2)
SUOX	170,4	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	33	69%	52%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	53,6	92%	77%	Mental retardation, autosomal dominant 5, 612621
SYP	50,2	99%	92%	Mental retardation, X-linked 96, 300802
SZT2	100,9	99%	95%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	106	100%	100%	Myoclonic epilepsy, infantile, familial, 605021 Epileptic encephalopathy, early infantile, 16, 615338
TBCE	118,9	100%	100%	Kenny-Caffey syndrome-1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410
TCF4	101,3	100%	100%	Pitt-Hopkins syndrome, 610954
TDP2	127,3	100%	100%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420 -3
TPP1	135,3	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500

TREX1	120,7	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRPM6	121,4	99%	98%	Hypomagnesemia 1, intestinal, 602014
UBE3A	103,5	100%	100%	Angelman syndrome, 105830
ZEB2	155,1	100%	99%	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors