EPILEPSY GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype and OMIM ID
ABAT	74.7	99%	94%	GABA-transaminase deficiency, 613163 GABA-transaminase deficiency
ABCC8	104.5	100%	99%	Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACY1	92.0	100%	98%	Aminoacylase 1 deficiency, 609924
ADSL	147.4	100%	100%	Adenylosuccinase deficiency, 103050
ALDH7A1	87.7	100%	94%	Epilepsy, pyridoxine-dependent, 266100
ALG13	134.2	96%	95%	Congenital disorder of glycosylation, type Is, 300884
AMACR	97.2	100%	100%	Alpha-methylacyl-CoA racemase deficiency, 614307
AMT	147.7	100%	100%	Glycine encephalopathy, 605899
APOPT1	111.2	98%	87%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	108.9	100%	99%	Epileptic encephalopathy, early infantile, 8, 300607
ARX	75.1	84%	82%	Epileptic encephalopathy, early infantile 1 Hydraencephaly with abnormal genitalia Lissencephaly, X-linked 2 Mental retardation, X-linked 29 Partington syndrome Proud syndrome
ASAH1	104.3	100%	100%	Farber lipogranulomatosis, 228000
ATP1A2	122.4	100%	99%	Migraine, familial hemiplegic, 2, 602481
ATP6AP2	74.3	99%	96%	Mental retardation, X-linked, with epilepsy, 300423
ATP7A	148.7	100%	100%	Menkes disease, 309400
ATRX	165.3	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040
BOLA3	77.2	100%	99%	Multiple mitochondrial dysfunctions syndrome 2, 614299
BTD	173.1	100%	100%	Biotinidase deficiency, 253260
CACNA1A	90.9	98%	92%	Migraine, familial hemiplegic, 1, 141500
CASK	108.3	100%	98%	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749
CDKL5	148.2	100%	100%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	142.8	99%	99%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	148.8	100%	99%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	117.7	100%	99%	Epilepsy, nocturnal frontal lobe, 1, 600513

CHRNB2	171.8	95%	94%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	141.1	100%	97%	Hypomagnesemia 3, renal, 248250
CLDN19	98.8	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	103.3	100%	100%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	156.3	100%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	84.2	100%	92%	Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	147.5	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143
CNNM2	150.6	100%	100%	Hypomagnesemia 6, renal, 613882
CNTN2	110.6	100%	100%	Epilepsy, familial adult myoclonic, 5, 615400
CNTNAP2	121.9	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042
COQ2	84.1	98%	93%	Coenzyme Q10 deficiency, primary, 1, 607426
CPA6	141.1	100%	100%	Epilepsy, familial temporal lobe, 5, 614417
CPS1	126.2	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300
CPT2	119.9	92%	92%	Myopathy due to CPT II deficiency, 255110
CSTB	185.7	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	113.0	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	130.3	95%	84%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	135.7	100%	100%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	74.9	98%	91%	D-2-hydroxyglutaric aciduria, 600721
DCX	132.0	100%	100%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	133.3	100%	99%	Epilepsy, familial focal, with variable foci, 604364
DLAT	114.1	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	73.0	94%	84%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	88.4	95%	80%	No OMIM phenotype Epileptic encephalopathy (Allen (2013) Nature 501, 217)
DOCK7	122.7	100%	100%	Epileptic encephalopathy, early infantile, 23, 615859
DPYD	133.1	99%	98%	Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	136.4	99%	97%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant, AD, 158600
DYRK1A	161.4	100%	99%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	117.1	100%	100%	No OMIM phenotype
EGF	123.5	100%	98%	Hypomagnesemia 4, renal, 611718
EHMT1	112.3	98%	96%	Kleefstra syndrome, 610253
EPM2A	84.5	88%	84%	Epilepsy, progressive myoclonic 2A (Lafora), 254780

FA2H	73.8	91%	82%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	115.7	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946
FASN	106.4	99%	97%	No OMIM phenotype Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95,360) Intellectual disability (Najmabadi (2011) Nature 478, 57) Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360)
FGD1	104.7	99%	96%	Aarskog-Scott syndrome, 305400
FLNA	143.5	100%	99%	Heterotopia, periventricular, 300049
FOLR1	106.4	100%	99%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	104.8	89%	78%	Rett syndrome, congenital variant, 613454
FOXRED1	120.8	100%	94%	Leigh syndrome due to mitochondrial complex I deficiency, 256000
FXYD2	77.3	94%	88%	Hypomagnesemia-2, renal, 154020
GABBR2	100.1	98%	96%	No OMIM phenotype Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360)
GABRA1	146.4	99%	96%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRG2	146.1	98%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277
GAMT	123.8	100%	94%	Cerebral creatine deficiency syndrome 2, 612736
GCK	97.2	100%	100%	MODY, type II, 125851
GCSH	17.7	52%	46%	Glycine encephalopathy, 605899
GLDC	69.3	97%	91%	Glycine encephalopathy, 605899
GLRA1	126.4	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	136.2	100%	98%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	134.1	88%	88%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	124.0	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GPC3	121.3	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870
GPHN	134.0	100%	100%	Molybdenum cofactor deficiency, type C, 252150
GRIA3	130.4	99%	98%	Mental retardation, X-linked 94, 300699
GRIN1	96.1	99%	96%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	158.9	100%	99%	Epilepsy with neurodevelopmental defects, 613971
GRIN2B	163.0	100%	99%	Mental retardation, autosomal dominant 6, 613970
GRN	148.6	100%	99%	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Myasthenia, limb-girdle, familial, 254300
HADH	104.4	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	117.7	100%	100%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	82.7	94%	90%	Brachydactyly-mental retardation syndrome, 600430

HLCS	168.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	133.5	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	131.0	99%	99%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438
HSD17B4	117.3	100%	99%	D-bifunctional protein deficiency, 261515
IDH2	131.7	100%	99%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	73.9	100%	99%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	148.8	100%	100%	Aicardi-Goutieres syndrome 7, 615846
IQSEC2	95.7	94%	87%	Mental retardation, X-linked 1, 309530
KANSL1	81.0	98%	91%	Koolen-De Vries syndrome, 610443
KCNA1	138.7	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNJ10	176.9	100%	100%	SESAME syndrome, 612780
KCNJ11	163.6	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	97.2	99%	97%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	87.5	100%	99%	Seizures, benign neonatal, 1, 121200
KCNQ3	112.8	99%	98%	Seizures, benign neonatal, type 2, 121201
KCNT1	91.3	100%	95%	Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	116.6	93%	86%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	132.1	100%	99%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	90.7	100%	100%	Mental retardation, autosomal recessive 41, 615637
LGI1	164.7	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	122.0	100%	100%	Pyruvate dehydrogenase lipoic acid synthetase deficiency, 614462
MBD5	171.4	100%	100%	Mental retardation, autosomal dominant 1, 156200
MECP2	198.9	100%	99%	Rett syndrome, 312750
MED12	143.5	99%	95%	Opitz-Kaveggia syndrome, 305450
MEF2C	125.4	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443

MFSD8	137.3	100%	100%	Ceroid lipofuscinosis, neuronal, 7, 610951
MOCS1	91.3	100%	96%	Molybdenum cofactor deficiency, type A, 252150
MOCS2	113.6	100%	100%	Molybdenum cofactor deficiency, type B, 252150
MTHFR	112.3	100%	98%	Homocystinuria due to MTHFR deficiency, 236250
MTOR	113.4	100%	99%	No OMIM phenotype Lennox-Gastaut syndrome (Allen(2013) Nature 501, 217)
NDUFA1	229.7	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	125.4	97%	85%	Mitochondrial complex I deficiency, 252010
NDUFAF1	131.5	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	62.2	100%	99%	Mitochondrial complex I deficiency, 252010 Leigh syndrome, 256000
NDUFAF3	149.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	68.2	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF5	140.5	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFB3	2.3	%	%	Mitochondrial complex I deficiency, 252010
NDUFB9	118.7	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS1	94.2	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS2	154.3	100%	98%	Mitochondrial complex I deficiency, 252010
NDUFS3	182.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS4	155.8	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFS6	142.6	99%	82%	Mitochondrial complex I deficiency, 252010
NDUFV1	74.8	99%	92%	Mitochondrial complex I deficiency, 252010
NDUFV2	134.0	100%	100%	Mitochondrial complex I deficiency, 252010
NECAP1	117.3	100%	100%	?Epileptic encephalopathy, early infantile,21, 615833
NEDD4L	133.4	100%	100%	No OMIM phenotype
NHLRC1	138.7	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	147.1	100%	98%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	112.4	100%	100%	Mitochondrial complex I deficiency, 252010
OFD1	86.3	95%	89%	Oral-facial-digital syndrome 1, 311200
OPHN1	113.7	100%	98%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	114.4	100%	100%	Mental retardation, X-linked 30/47, 300558
PC	121.6	95%	92%	Pyruvate carboxylase deficiency, 266150
PCDH19	165.1	99%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	133.0	100%	99%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	125.2	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111

PDP1	203.1	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PET100	81.3	100%	99%	Mitochondrial complex IV deficiency, 220110
PEX1	141.3	100%	100%	Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	87.3	92%	86%	Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX12	138.8	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	160.2	98%	92%	Peroxisome biogenesis disorder 11A (Zellweger), 614883
PEX14	105.9	100%	100%	Peroxisome biogenesis disorder 14B, 614920 Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	122.3	92%	91%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876
PEX19	119.3	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	132.3	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	162.6	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	103.6	98%	97%	Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	107.0	98%	91%	Peroxisome biogenesis disorder 4A (Zellweger), 614862
PGAP3	67.7	98%	88%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	148.2	100%	100%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	95.1	100%	100%	Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	158.6	100%	100%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	122.3	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	132.6	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	156.4	100%	100%	?Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	83.6	99%	94%	Infantile neuroaxonal dystrophy 1, 256600
PLCB1	129.5	100%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	92.2	100%	98%	Pelizaeus-Merzbacher disease, 312080
PNKP	80.8	100%	99%	Epileptic encephalopathy, early infantile, 10, 613402
PNPO	80.3	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	102.8	99%	95%	Progressive external ophthalmoplegia, autosomal recessive, 258450
PPT1	83.8	100%	98%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	158.0	100%	100%	Renpenning syndrome, 309500
PRICKLE1	124.1	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	86.4	100%	99%	Episodic kinesigenic dyskinesia 1, 128200
QARS	141.3	100%	100%	Microcephaly, progressive,seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	227.2	100%	100%	Mental retardation, X-linked 72, 300271
RARS2	101.5	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	114.6	100%	96%	Aicardi-Goutieres syndrome 4, 610333

RNASEH2B	122.8	100%	100%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	179.6	100%	100%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	112.4	98%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	112.3	100%	100%	Coffin-Lowry syndrome, 303600
RRM2B	133.5	100%	100%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic,renal tubulopathy), 612075
RYR3	115.7	100%	99%	No OMIM phenotype Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360) Hyperinsulinism (Proverbio (2013) PLoS One 8, e68740) Schizophrenia (Fromer (2014) Nature 506, 179) Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95, 360)
SAMHD1	137.9	100%	98%	Aicardi-Goutieres syndrome 5, 612952
SCARB2	114.7	100%	96%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	134.6	100%	99%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403
SCN1B	105.2	95%	93%	Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	142.8	100%	100%	Seizures, benign familial infantile, 3, 607745
SCN8A	161.4	100%	99%	Cognitive impairment with or without cerebellar ataxia, 614306
SCN9A	133.6	100%	99%	Erythermalgia, primary, 133020
SLC13A5	95.1	100%	99%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	158.1	100%	100%	Erythrocyte lactate transporter defect, 245340
SLC19A3	130.9	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	86.4	89%	83%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	122.4	96%	83%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	89.1	100%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	114.2	100%	100%	GLUT1 deficiency syndrome 1, 606777
SLC35A2	117.6	100%	100%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A8	9.4	22%	15%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	131.2	100%	98%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMC1A	164.3	99%	97%	Cornelia de Lange syndrome 2, 300590
SMS	35.3	88%	68%	Smith-Magenis syndrome, 182290
SPTAN1	115.5	99%	98%	Epileptic encephalopathy, early infantile, 5, 613477
SRPX2	95.2	100%	99%	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643
ST3GAL3	130.5	100%	100%	Mental retardation, autosomal recessive 12, 611090
ST3GAL5	116.7	93%	93%	Amish infantile epilepsy syndrome, 609056
STXBP1	106.6	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	204.6	100%	100%	Sulfite oxidase deficiency, 272300

SYN1	78.0	98%	93%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	64.2	93%	82%	Mental retardation, autosomal dominant 5, 612621
SYP	107.4	100%	99%	Mental retardation, X-linked 96, 300802
SZT2	122.1	99%	97%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	140.0	100%	100%	Myoclonic epilepsy, infantile, familial, 605021
TBCE	136.5	100%	100%	Kenny-Caffey syndrome-1, 244460
TCF4	111.9	100%	100%	Pitt-Hopkins syndrome, 610954
TDP2	141.2	100%	100%	Dentinogenesis imperfecta, Shields type II, 125490
TPP1	154.3	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	179.4	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750
TRPM6	139.7	99%	98%	Hypomagnesemia 1, intestinal, 602014
TSC1	105.1	99%	98%	Focal cortical dysplasia, Taylor balloon cell type, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TUBB2A	50.1	100%	94%	Cortical dysplasia, complex, with other brain malformations 5, 615763
UBE3A	119.2	100%	100%	Angelman syndrome, 105830
ZEB2	179.6	100%	100%	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors