EPILEPSY GENE PANEL DG 2.7/DG 2.8

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS	144.7	99%	99%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	103	100%	99%	GABA-transaminase deficiency, 613163
ABCC8	167	100%	99%	Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ACTB	134.1	98%	93%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACY1	155.9	99%	98%	Aminoacylase 1 deficiency, 609924
ADCK3	146	99%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
ADSL	205.9	100%	99%	Adenylosuccinase deficiency, 103050
ALDH7A1	83.1	97%	89%	Epilepsy, pyridoxine-dependent, 266100
ALG1	60.3	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	182.7	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	107.4	98%	94%	Epileptic encephalopathy, early infantile, 36, 300884
ALG3	125.8	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	104.1	95%	92%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	161.6	99%	98%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	174	100%	99%	Glycine encephalopathy, 605899
APOPT1	80.9	87%	84%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	109.7	99%	98%	Epileptic encephalopathy, early infantile, 8, 300607
ARX	39.1	82%	70%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215

				Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASAH1	141	98%	92%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ATP1A2	209.6	100%	100%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	205.7	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	55.4	83%	63%	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	157.3	99%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	94.5	97%	93%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	115.2	96%	95%	Mental retardation, autosomal dominant 26,615834
BOLA3	59	91%	82%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	112.9	99%	96%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	163.8	100%	99%	Biotinidase deficiency, 253260
CACNA1A	105.6	95%	91%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	158.5	99%	99%	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)
CACNA2D2	160.6	94%	92%	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)

CASK	116.5	98%	94%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	140.7	98%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	148.5	99%	98%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	251.6	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	168.2	97%	95%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	267.9	99%	98%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	161.4	100%	99%	Hypomagnesemia 3, renal, 248250
CLDN19	143.1	98%	95%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	125.9	98%	94%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	163.1	98%	93%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	142.3	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	252.4	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	213.2	99%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	148.2	100%	99%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	156.8	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COL4A1	101.8	98%	93%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	140.6	99%	96%	Mental retardation, autosomal dominant 34, 616351
COQ2	84.5	95%	92%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	94	86%	82%	Coenzyme Q10 deficiency, primary, 7, 616276
CPA6	139.5	99%	98%	Epilepsy, familial temporal lobe, 5, 614417

				Febrile seizures, familial, 11, 614418
CPS1	169.3	100%	99%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	168.6	98%	96%	CPT deficiency, hepatic, type II, 600649 CPT II deficiency, lethal neonatal, 608836 Myopathy due to CPT II deficiency, 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CSTB	120.6	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	183.6	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	117.4	89%	82%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	83.8	96%	90%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	147.9	97%	95%	D-2-hydroxyglutaric aciduria, 600721
DCX	137.9	100%	99%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	162.3	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DLAT	102.4	99%	95%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	205.6	100%	99%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	171.5	94%	91%	Epileptic encephalopathy, early infantile, 31, 616346
DOCK7	128.8	97%	94%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	134.5	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	136.9	89%	84%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	115.2	100%	99%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	177.9	95%	94%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	196.8	100%	99%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	176.5	100%	99%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	208.6	99%	98%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393

EGF	157.6	99%	99%	Hypomagnesemia 4, renal, 611718
EHMT1	161.2	98%	96%	Kleefstra syndrome, 610253
EPM2A	123.7	85%	83%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	108.9	94%	88%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	224.8	100%	99%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FASN	136.8	99%	99%	No OMIM phenotype Intellectual disability (Najmabadi (2011) Nature 478, 57) ?Epileptic encephalopathy (Appenzeller (2014) Am J Hum Genet 95, 360) ?Lennox-Gastaut syndrome (Appenzeller (2014) Am J Hum Genet 95,360)
FGD1	98.1	95%	89%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FLNA	161.1	99%	99%	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FOLR1	166.3	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	130.8	89%	82%	Rett syndrome, congenital variant, 613454
FOXRED1	145.1	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXYD2	105.5	100%	99%	Hypomagnesemia 2, renal, 154020
GABRA1	199.9	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	165.3	97%	93%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	178	93%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277

				{Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAMT	119.1	97%	91%	Cerebral creatine deficiency syndrome 2, 612736
GCK	155.4	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	38.8	83%	62%	Glycine encephalopathy, 605899
GLDC	90.1	91%	84%	Glycine encephalopathy, 605899
GLRA1	136.5	100%	100%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	100.7	96%	91%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	82.4	94%	86%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	204.8	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	143.1	97%	95%	Epilepsy, progressive myoclonic 6, 614018
GPC3	121	98%	94%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	192.1	98%	97%	Molybdenum cofactor deficiency C, 615501
GRIA3	112.8	99%	95%	Mental retardation, X-linked 94, 300699
GRIN1	166.9	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	176.5	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	213.2	99%	99%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRN	206.3	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	120.8	97%	95%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	142.6	99%	98%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	119.7	99%	99%	No OMIM phenotype
HLCS	193.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	136.6	99%	97%	No OMIM phenotype Lennox-Gastaut syndrome (Allen (2013) Nature 501,217) Fever-associated epilepsy (Hartmann (2015) Epilepsia 56,e26)

				Infantile spasms (Du (2014) BMC Med Genet 15,62) Speech delay,seizures and 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) Preaxial polydactyly (Gupta (2014) Am J Med Genet A 164A,186) 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) ?Developmental delay and intellectual disability (King (2014) Genome Res 24,673) ?Intellectual disability (Hamdan (2014) PLoS Genet 10,e1004772) ?Intellectual disability,epilepsy,panhypopituitarism,hypertension & other anomalies (Zhu (2015) Genet Med)
HSD17B10	120.3	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B4	110.3	94%	91%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	108.8	99%	98%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	60.9	86%	78%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	130	98%	96%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	72.2	94%	86%	Mental retardation, X-linked 1/78, 309530
JAM3	166.6	99%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	94.7	94%	89%	Koolen-De Vries syndrome, 610443
KCNA1	178.7	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	180.7	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	149.4	100%	99%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	213.9	100%	99%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	198.9	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	229	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	302.7	100%	100%	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, 606176

				Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNMA1	158.7	100%	99%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	103.9	98%	96%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	117.6	99%	96%	Seizures, benign neonatal, type 2, 121201
KCNT1	129.2	95%	93%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	144.9	93%	92%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	128.3	98%	95%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KIAA2022	183	100%	99%	Mental retardation, X-linked 98, 300912
KPTN	118.2	100%	99%	Mental retardation, autosomal recessive 41, 615637
LGI1	203.4	98%	95%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	159.6	99%	95%	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	202.7	100%	99%	Mental retardation, autosomal dominant 1, 156200
MECP2	100	99%	94%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MED12	116.5	98%	95%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEF2C	142.4	98%	93%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	137.1	99%	98%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MOCS1	92.2	98%	93%	Molybdenum cofactor deficiency A, 252150
MOCS2	156.7	99%	98%	Molybdenum cofactor deficiency B, 252160

MPDU1	131.9	100%	99%	Congenital disorder of glycosylation, type If, 609180
MTHFR	153.2	100%	99%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTOR	152.8	100%	99%	Smith-Kingsmore syndrome, 616638
NDUFA1	236.8	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFA11	95.1	99%	94%	Mitochondrial complex I deficiency, 252010
NDUFAF1	120.7	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF2	59.8	81%	67%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	122.5	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFAF4	103.9	98%	91%	Mitochondrial complex I deficiency, 252010
NDUFAF5	104.7	97%	94%	Mitochondrial complex 1 deficiency, 252010
NDUFB3	23.3	91%	56%	Mitochondrial complex I deficiency, 252010
NDUFB9	128.2	99%	97%	?Mitochondrial complex I deficiency, 252010
NDUFS1	154.7	99%	98%	Mitochondrial complex I deficiency, 252010
NDUFS2	120.4	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	151.1	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	175.1	100%	98%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	138.1	99%	99%	Mitochondrial complex I deficiency, 252010
NDUFV1	168.6	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	74.1	84%	62%	Mitochondrial complex I deficiency, 252010
NECAP1	133.2	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	163.6	99%	97%	No OMIM phenotype {Essential hypertension, association with} (Russo (2005) Hypertension 46,488) Epilepsy,photosensitive generalised (Dibbens (2007),Genes Brain Behav 6,750) Infantile spasms (Allen (2013) Nature 501,217) Impaired ENaC regulation (Fouladkou (2004) Am J Physiol Renal Physiol 287,F550)

NGLY1	140.8	99%	98%	Congenital disorder of deglycosylation, 615273
NHLRC1	174.4	100%	99%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NRXN1	182	99%	97%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	101.7	90%	85%	Mitochondrial complex I deficiency, 252010
OFD1	56.1	84%	71%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	113.4	99%	96%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	95.9	97%	91%	Mental retardation, X-linked 30/47, 300558
PC	162.8	99%	97%	Pyruvate carboxylase deficiency, 266150
PCDH19	226.4	100%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	127.8	97%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	144	98%	95%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	209.6	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	35.9	91%	71%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PET100	127.6	95%	82%	Mitochondrial complex IV deficiency, 220110
PEX1	123.4	97%	95%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	118.3	97%	93%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	165.4	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	212.2	99%	98%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	143.6	99%	98%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	138.5	96%	92%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	121	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX26	86.5	99%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.8	98%	94%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	126.7	99%	97%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	92	90%	84%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	135.7	98%	95%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	77.9	92%	83%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	138.6	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	128.6	95%	89%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	140.6	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	174.6	99%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	132.4	99%	98%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	177.3	99%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	162.1	100%	99%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	178.4	99%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	98.4	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	84.2	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	126.2	99%	99%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640

				Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	151.6	93%	92%	Mental retardation, autosomal dominant 36, 616362
PPT1	190.4	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	174.3	97%	96%	Renpenning syndrome, 309500
PRICKLE1	137.2	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	143.3	100%	99%	No OMIM phenotype ?Autism spectrum disorder (Sowers (2013) Mol Psychiatry 18, 1077) ?Myoclonus epilepsy (Tao (2011) Am J Hum Genet 88,138)
PRRT2	80.7	99%	98%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PURA	125.2	98%	95%	Mental retardation, autosomal dominant 31, 616158
PYCR2	137.6	99%	98%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	167.3	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	139.4	100%	99%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RARS2	126.3	99%	98%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	149.3	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	125.1	94%	84%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	207.6	99%	97%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	134	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	99.5	94%	87%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	148.4	99%	97%	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SAMHD1	149.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCARB2	138.9	99%	98%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	157.3	99%	98%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403

				Migraine, familial hemiplegic, 3, 609634
SCN1B	180.2	97%	96%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233
SCN2A	170.4	99%	97%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	224.3	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SIK1	99.2	98%	94%	Epileptic encephalopathy, early infantile, 30, 616341
SLC13A5	176.9	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	167.2	99%	97%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	191.3	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	84.7	97%	90%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	228.7	98%	95%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	117.1	99%	96%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	183.5	100%	100%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC35A2	118.7	99%	96%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	166.1	100%	99%	Myoclonic-atonic epilepsy, 616421
SLC6A8	61.1	92%	82%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	126.9	97%	90%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	131.1	96%	94%	Nicolaides-Baraitser syndrome, 601358
SMC1A	120	100%	99%	Cornelia de Lange syndrome 2,300590
SMS	70.1	88%	76%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SPTAN1	139.4	99%	98%	Epileptic encephalopathy, early infantile, 5, 613477

ST3GAL3	193.7	100%	100%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	138.4	95%	94%	Amish infantile epilepsy syndrome, 609056
STXBP1	147.9	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	219.5	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	79.6	83%	70%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	75	95%	86%	Mental retardation, autosomal dominant 5, 612621
SYP	77.8	99%	95%	Mental retardation, X-linked 96, 300802
SZT2	157.5	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	178.8	100%	99%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBCE	151.6	99%	98%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	160.1	99%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	177.7	99%	97%	Spinocerebellar ataxia, autosomal recessive, 616949
TPP1	158.7	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	272.2	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	172.4	99%	98%	Hypomagnesemia 1, intestinal, 602014
TSC1	149.6	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	144.5	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TUBB2A	119.2	99%	97%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBG1	185.9	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBE3A	103.9	98%	93%	Angelman syndrome, 105830

WWOX	148	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
ZEB2	181.5	100%	99%	Mowat-Wilson syndrome, 235730

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
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.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors