EPILEPSY GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS	145.1	100%	99%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	103.8	100%	99%	GABA-transaminase deficiency, 613163
ABCC8	161.8	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	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACY1	162.7	99%	98%	Aminoacylase 1 deficiency, 609924
ADCK3	161	99%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADSL	196.2	100%	100%	Adenylosuccinase deficiency, 103050
ALDH7A1	88.5	97%	91%	Epilepsy, pyridoxine-dependent, 266100
ALG1	57.5	53%	49%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	189.4	100%	99%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	104.2	99%	96%	Epileptic encephalopathy, early infantile, 36, 300884
ALG3	116.7	100%	99%	Congenital disorder of glycosylation, type Id, 601110
ALG6	126.3	98%	95%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	173.1	100%	99%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	187.9	100%	100%	Glycine encephalopathy, 605899
APOPT1	90.9	87%	86%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	99.5	99%	98%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	159.9	96%	92%	Coffin-Siris syndrome 1, 135900
ARX	38.3	81%	67%	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	147.1	99%	97%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	128.8	100%	98%	Argininosuccinic aciduria, 207900
ATP1A2	219	100%	99%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	204.3	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	55.5	91%	71%	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	148.2	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	105.7	98%	95%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	125.8	97%	96%	Mental retardation, autosomal dominant 26,615834
BOLA3	63.6	92%	83%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	121.6	99%	98%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	168.1	100%	100%	Biotinidase deficiency, 253260
CACNA1A	104.4	95%	92%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	163.7	99%	99%	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)
CACNA2D2	158.8	94%	93%	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CASK	112.2	99%	95%	FG syndrome 4, 300422

				Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	135.2	99%	96%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	145.9	99%	99%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	229.9	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	169.5	97%	96%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	250.6	99%	98%	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	171.5	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	144.2	99%	96%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	133	99%	96%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	155.4	99%	97%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	144.8	98%	94%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	273.8	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	228.4	100%	99%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	161	100%	99%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	168.7	100%	99%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COL4A1	103.5	98%	94%	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	153.5	99%	98%	Mental retardation, autosomal dominant 34, 616351
COQ2	92.8	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	103.5	87%	84%	Coenzyme Q10 deficiency, primary, 7, 616276
CPA6	153.2	100%	99%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPS1	183.3	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300

				{Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	182	98%	96%	CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836 CPT II deficiency, myopathic, stress-induced 255110 {Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212
CSTB	108.7	99%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	197.2	99%	98%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	119	87%	79%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	88.6	98%	92%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	153.2	98%	96%	D-2-hydroxyglutaric aciduria, 600721
DCX	123.8	100%	99%	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DEPDC5	164.8	99%	99%	Epilepsy, familial focal, with variable foci, 604364
DLAT	104.8	99%	98%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	212	100%	99%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	167.4	94%	90%	Epileptic encephalopathy, early infantile, 31, 616346
DOCK7	147.1	98%	96%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	119.1	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	149.2	92%	87%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	99	99%	96%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	190.3	97%	95%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	189.4	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	168	100%	99%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	213.2	100%	98%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EGF	165.2	100%	99%	Hypomagnesemia 4, renal, 611718
EHMT1	163	99%	98%	Kleefstra syndrome, 610253
EPM2A	121.8	85%	84%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	101.9	95%	89%	Spastic paraplegia 35, autosomal recessive, 612319

FARS2	234.6	100%	100%	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FGD1	98.4	94%	88%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FLNA	160.7	100%	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	155.8	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	139.8	87%	81%	Rett syndrome, congenital variant, 613454
FOXRED1	144.3	100%	99%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FXYD2	104.4	100%	99%	Hypomagnesemia 2, renal, 154020
GABRA1	219.1	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	158	97%	93%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	196.1	94%	92%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAMT	123	98%	91%	Cerebral creatine deficiency syndrome 2, 612736
GCK	151.4	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	41.7	85%	70%	Glycine encephalopathy, 605899
GLDC	88.5	92%	84%	Glycine encephalopathy, 605899
GLRA1	124.8	100%	99%	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400

GLRB	119.6	98%	93%	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	84.5	94%	85%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	197.4	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	143.2	97%	96%	Epilepsy, progressive myoclonic 6, 614018
GPC3	106.4	98%	94%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	186.4	99%	97%	Molybdenum cofactor deficiency C, 615501
GRIA3	102.9	99%	95%	Mental retardation, X-linked 94, 300699
GRIN1	169.2	100%	99%	Mental retardation, autosomal dominant 8, 614254
GRIN2A	170.4	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	194.9	99%	99%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRN	214.6	100%	100%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	126.2	97%	94%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCN1	155.1	100%	99%	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	121.1	100%	99%	No OMIM phenotype Anorexia nervosa/bulimia nervosa (Cui (2013) J Clin Invest 123,4706) Brachydactyly mental retardation syndrome (Williams (2010) Am J Hum Genet 87, 219) ?Autism spectrum disorder (Pinto (2014) Am J Hum Genet 94, 677)
HLCS	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	153.2	99%	98%	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	119.7	100%	98%	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B4	125.3	95%	93%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	108	99%	98%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	72.1	92%	80%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	143.7	99%	98%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	72.6	94%	86%	Mental retardation, X-linked 1/78, 309530
JAM3	158.3	99%	98%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730

KANSL1	90.1	95%	89%	Koolen-De Vries syndrome, 610443
KCNA1	179.6	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	178.9	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	150.8	100%	99%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	218.2	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	195.4	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	219.9	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	281.8	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	150.1	100%	99%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	114.3	98%	95%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	118.7	99%	96%	Seizures, benign neonatal, type 2, 121201
KCNT1	133.4	95%	94%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	157.8	94%	93%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	123.7	98%	95%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KIAA2022	176.1	100%	99%	Mental retardation, X-linked 98, 300912
KPTN	122.9	100%	99%	Mental retardation, autosomal recessive 41, 615637
LGI1	207	99%	98%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	175	100%	99%	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	204.8	100%	99%	Mental retardation, autosomal dominant 1, 156200
MECP2	99.4	99%	95%	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	111.4	98%	94%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEF2C	150.6	99%	96%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	143.4	100%	99%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MLC1	114.2	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	92.7	98%	92%	Molybdenum cofactor deficiency A, 252150
MOCS2	183	99%	99%	Molybdenum cofactor deficiency B, 252160
MPDU1	129.4	100%	99%	Congenital disorder of glycosylation, type If, 609180
MTHFR	161.7	100%	100%	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	152.6	100%	99%	Smith-Kingsmore syndrome, 616638
NDUFA1	229	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFA11	95.2	99%	95%	Mitochondrial complex I deficiency, 252010
NDUFAF1	128.7	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF2	48.3	87%	76%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	130.4	100%	100%	Mitochondrial complex I deficiency, 252010
NDUFAF4	125.1	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFAF5	108.8	99%	98%	Mitochondrial complex 1 deficiency, 252010
NDUFB3	29.3	96%	76%	Mitochondrial complex I deficiency, 252010
NDUFB9	123.2	99%	97%	?Mitochondrial complex I deficiency, 252010
NDUFS1	165.9	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS2	121.6	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFS3	149.4	90%	90%	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	200.6	100%	100%	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010

NDUFS6	146.2	100%	99%	Mitochondrial complex I deficiency, 252010
NDUFV1	158	99%	97%	Mitochondrial complex I deficiency, 252010
NDUFV2	88.7	90%	73%	Mitochondrial complex I deficiency, 252010
NECAP1	135.2	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	159.4	99%	99%	Periventricular nodular heterotopia 7, 617201
NGLY1	162.3	100%	99%	Congenital disorder of deglycosylation, 615273
NHLRC1	192.8	100%	99%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	197.3	100%	99%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	147.1	100%	99%	Epilepsy, familial focal, with variable foci, 3, 617118
NRXN1	193.5	99%	98%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	102.9	92%	88%	Mitochondrial complex I deficiency, 252010
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	103.5	99%	97%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	94.1	98%	93%	Mental retardation, X-linked 30/47, 300558
PC	166.1	99%	97%	Pyruvate carboxylase deficiency, 266150
PCDH19	219.8	99%	99%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	112.7	98%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	148.5	99%	97%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	197.1	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	39.1	93%	75%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PET100	98.1	98%	86%	Mitochondrial complex IV deficiency, 220110
PEX1	139.9	98%	97%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	124.7	97%	92%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	157.9	100%	99%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510

PEX13	240	100%	99%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	145.5	99%	98%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	146.3	97%	93%	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	118.8	100%	99%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	87.2	100%	99%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	129.6	99%	97%	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	132.1	99%	98%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	95.9	91%	85%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	126.1	98%	94%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	74.9	94%	86%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	138	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	130.2	99%	94%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	143.9	100%	99%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGT	195.8	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	135.5	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	173.5	100%	99%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	155	99%	98%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100.7	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402

PNPO	80.9	100%	99%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	128.1	100%	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	158.2	93%	93%	Mental retardation, autosomal dominant 36, 616362
PPT1	203.5	100%	100%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	180.7	97%	96%	Renpenning syndrome, 309500
PRICKLE1	138.6	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRICKLE2	149.2	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	91.4	100%	99%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PURA	135.6	98%	93%	Mental retardation, autosomal dominant 31, 616158
PYCR2	142.2	99%	96%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	163.6	100%	99%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	125.7	100%	99%	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RARS2	137.6	100%	99%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	92%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	98%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	127.1	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	97.9	96%	89%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	163	99%	98%	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	154.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415

SCARB2	141.5	100%	99%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	170.2	99%	99%	Dravet syndrome, 607208 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	189.8	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	186.5	99%	98%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN8A	209.5	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SIK1	111.9	98%	96%	Epileptic encephalopathy, early infantile, 30, 616341
SLC13A5	167.9	100%	99%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	180.7	99%	98%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	185.8	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC25A1	90.7	98%	92%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	238.4	98%	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	120.5	99%	97%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	191.5	100%	99%	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	111.5	99%	97%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	167	100%	99%	Myoclonic-atonic epilepsy, 616421
SLC6A8	59	91%	81%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	119.2	98%	94%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	127.7	97%	96%	Nicolaides-Baraitser syndrome, 601358
SMC1A	114.2	100%	98%	Cornelia de Lange syndrome 2,300590
SMS	79.4	89%	77%	Mental retardation, X-linked, Snyder-Robinson type, 309583

SPTAN1	137.5	99%	99%	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	195.5	100%	99%	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	144.8	95%	94%	Amish infantile epilepsy syndrome, 609056
STX1B	191.4	100%	99%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	146.2	100%	99%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	221.3	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	73.7	84%	71%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	77.1	95%	86%	Mental retardation, autosomal dominant 5, 612621
SYP	81.3	99%	96%	Mental retardation, X-linked 96, 300802
SZT2	158.8	99%	99%	Epileptic encephalopathy, early infantile, 18, 615476
TANGO2	158.8	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TBC1D24	203.2	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	162.8	99%	99%	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	150.3	100%	99%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	203.7	100%	99%	Spinocerebellar ataxia, autosomal recessive, 616949
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	302.9	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	166.1	100%	99%	Hypomagnesemia 1, intestinal, 602014
TSC1	140.4	99%	97%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	150.1	99%	99%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254

TSEN54	98.8	96%	93%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	115.3	99%	97%	Lissencephaly 3, 611603
TUBB2A	111.8	99%	96%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBG1	190.6	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBE3A	117.2	99%	96%	Angelman syndrome, 105830
WWOX	143.3	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XK	107.1	99%	99%	McLeod syndrome with or without chronic granulomatous disease, 300842
ZEB2	195.1	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 : April 14th 2017
This list is accurate for panel version DG 2.9

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