EPILEPSY GENE PANEL DG 2.11 (289 genes)

Gene	Median	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS	124.4	100	99	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	92.8	100	99	GABA-transaminase deficiency, 613163
ABCC8	146.9	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	129.3	99	94	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACY1	130.4	100	98	Aminoacylase 1 deficiency, 609924
ADCK3	134.7	100	99	Coenzyme Q10 deficiency, primary, 4, 612016
ADSL	185.2	100	99	Adenylosuccinase deficiency, 103050
ALDH7A1	78.1	96	88	Epilepsy, pyridoxine-dependent, 266100
ALG1	51	53	48	Congenital disorder of glycosylation, type Ik, 608540
ALG11	144.2	99	99	Congenital disorder of glycosylation, type Ip, 613661
ALG13	87.4	98	94	Epileptic encephalopathy, early infantile, 36, 300884
ALG3	133	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.3	96	93	Congenital disorder of glycosylation, type Ic, 603147
AMACR	149.1	100	99	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	173.3	100	100	Glycine encephalopathy, 605899
ANKRD11	96.4	97	94	KBG syndrome, 148050
AP3B2	135.3	97	94	Epileptic encephalopathy, early infantile, 48
APOPT1	67.9	86	83	Mitochondrial complex IV deficiency, 220110
ARHGEF9	79.2	99	97	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	157.6	94	89	Coffin-Siris syndrome 1, 135900

ARX	29.3	76	59	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	116.8	99	95	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	114.6	99	98	Argininosuccinic aciduria, 207900
ATP1A2	191.1	100	99	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	177.6	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	46.4	81	55	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	134.1	99	97	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	83	98	92	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	110.4	96	95	Mental retardation, autosomal dominant 26,615834
BOLA3	50.1	92	81	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	108.7	99	97	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	166.9	100	99	Biotinidase deficiency, 253260
CACNA1A	87.9	92	89	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	138.6	99	99	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)

CACNA2D2	135.3	93	92	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CAD	159.1	100	99	?Congenital disorder of glycosylation, type Iz, 616457
CASK	93	98	93	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	123.2	99	96	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	138.7	99	99	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	230.2	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	143.7	96	95	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	248.7	98	94	Epilepsy, nocturnal frontal lobe, 3, 605375
CLDN16	136.3	100	99	Hypomagnesemia 3, renal, 248250
CLDN19	123.8	98	93	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	121.3	98	95	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.3	98	92	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.8	98	95	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	196.4	100	100	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	188.5	100	99	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	135.5	100	99	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	148.1	100	99	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COL4A1	92.8	97	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	121.7	98	92	Mental retardation, autosomal dominant 34, 616351
COQ2	89.2	96	93	Coenzyme Q10 deficiency, primary, 1, 607426

				{Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88	84	Coenzyme Q10 deficiency, primary, 7, 616276
CPA6	118.3	99	98	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPS1	143.8	100	99	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}
CPT2	166.7	98	97	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	100	98	87	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	164	97	95	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	113	84	80	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	73.3	98	88	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
D2HGDH	132.7	97	95	D-2-hydroxyglutaric aciduria, 600721
DCX	116	100	99	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DENND5A	123	99	97	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	145.8	99	99	Epilepsy, familial focal, with variable foci, 604364
DLAT	91.6	99	96	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	200.2	100	99	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	161.4	91	90	Epileptic encephalopathy, early infantile, 31, 616346
DOCK7	114.3	97	95	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	113	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	131.2	91	86	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.2	100	99	Congenital disorder of glycosylation, type Iu, 615042
DPYD	156.6	95	93	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	180	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	158.4	100	100	Mental retardation, autosomal dominant 7, 614104

EEF1A2	178	98	93	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EGF	135.2	100	99	Hypomagnesemia 4, renal, 611718
EHMT1	147.1	98	97	Kleefstra syndrome, 610253
EPM2A	108.4	85	83	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	94.2	87	79	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	207.8	100	100	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FGD1	86.4	92	86	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	95.6	99	96	?Epileptic encephalopathy, early infantile, 47, 617166
FLNA	139.2	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	150.5	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	158	84	81	Rett syndrome, congenital variant, 613454
FOXRED1	136.9	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRMPD4	115	99	98	Mental retardation,X-linked 104, 300983
FRRS1L	103.2	68	63	Epileptic encephalopathy, early infantile, 37, 616981
FXYD2	96.9	99	99	Hypomagnesemia 2, renal, 154020
GABRA1	183.2	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	140.7	98	93	{Epilepsy, childhood absence, susceptibility to, 5}, 612269

GABRG2	147.7	92	92	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAMT	93.7	90	80	Cerebral creatine deficiency syndrome 2, 612736
GCK	141.7	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	34.2	83	67	Glycine encephalopathy, 605899
GLDC	80.8	90	82	Glycine encephalopathy, 605899
GLRA1	123.4	100	100	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	98.5	96	88	Hyperekplexia 2, autosomal recessive, 614619
GLUD1	76.7	94	84	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	180.4	100	100	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	123.2	97	96	Epilepsy, progressive myoclonic 6, 614018
GPC3	85.8	98	92	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	167.3	98	96	Molybdenum cofactor deficiency C, 615501
GRIA3	95.2	99	94	Mental retardation, X-linked 94, 300699
GRIN1	148.7	100	99	Mental retardation, autosomal dominant 8, 614254
GRIN2A	159.3	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	189.7	99	99	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRN	184.8	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	111.7	98	95	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	106.7	99	96	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	123.5	99	97	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	113.3	99	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	172.9	100	100	Holocarboxylase synthetase deficiency, 253270
HNRNPU	125.7	99	97	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	117.9	100	99	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B4	94.9	93	90	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	103.7	99	96	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	71.9	92	80	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	113.4	99	97	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	61.6	92	82	Mental retardation, X-linked 1/78, 309530
ITPA	120.4	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAM3	144.9	99	98	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	171.3	99	99	Koolen-De Vries syndrome, 610443
KCNA1	167	100	99	Episodic ataxia/myokymia syndrome, 160120
KCNA2	162.2	100	100	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	145	100	99	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	199.4	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNH1	188.2	100	99	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	238.8	100	99	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	300.2	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	131.4	100	99	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	93.2	98	94	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	111.1	98	95	Seizures, benign neonatal, type 2, 121201

KCNT1	112.2	95	92	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	148.9	94	92	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	113.5	97	95	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KIAA2022	140.1	99	99	Mental retardation, X-linked 98, 300912
KPTN	112.2	100	99	Mental retardation, autosomal recessive 41, 615637
LGI1	167.3	99	96	Epilepsy, familial temporal lobe, 1, 600512
LIAS	133.6	99	97	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	196.7	100	99	Mental retardation, autosomal dominant 1, 156200
MECP2	87.9	99	93	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	106.5	98	94	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEF2C	137	97	93	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	124.8	99	98	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MLC1	103.6	100	99	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	87.4	98	93	Molybdenum cofactor deficiency A, 252150
MOCS2	139.5	99	99	Molybdenum cofactor deficiency B, 252160
MPDU1	112	100	99	Congenital disorder of glycosylation, type If, 609180
MPDZ	148.9	98	96	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MTHFR	131.3	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	140.2	100	99	Smith-Kingsmore syndrome, 616638

MTRR	139.2	100	99	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
NACC1	168	100	99	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties and delayed brain myelination, 617393
NDUFA1	167.6	100	99	Mitochondrial complex I deficiency, 252010
NDUFA11	87.1	99	95	Mitochondrial complex I deficiency, 252010
NDUFAF1	115.7	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF2	58.6	85	70	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAF3	121.1	100	100	Mitochondrial complex I deficiency, 252010
NDUFAF4	79.5	98	91	Mitochondrial complex I deficiency, 252010
NDUFAF5	95.8	98	94	Mitochondrial complex 1 deficiency, 252010
NDUFB3	22.6	91	59	Mitochondrial complex I deficiency, 252010
NDUFB9	120.3	99	97	?Mitochondrial complex I deficiency, 252010
NDUFS1	132.1	99	98	Mitochondrial complex I deficiency, 252010
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFS3	142.6	90	90	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	147.2	100	99	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	119.1	99	99	Mitochondrial complex I deficiency, 252010
NDUFV1	137	99	97	Mitochondrial complex I deficiency, 252010
NDUFV2	69.3	78	53	Mitochondrial complex I deficiency, 252010
NECAP1	119.9	100	100	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	145.9	99	98	Periventricular nodular heterotopia 7, 617201
NGLY1	128	100	99	Congenital disorder of deglycosylation, 615273
NHLRC1	174.1	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	168.7	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	127.6	100	99	Epilepsy, familial focal, with variable foci, 3, 617118
NRXN1	166.2	99	97	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	89.6	92	85	Mitochondrial complex I deficiency, 252010

OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	89.7	99	96	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAK3	83.3	97	92	Mental retardation, X-linked 30/47, 300558
PC	149.6	97	94	Pyruvate carboxylase deficiency, 266150
PCDH19	225.8	100	99	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	110.5	98	92	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	133	99	96	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	209.6	100	100	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	35.3	89	72	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PET100	94.5	88	74	Mitochondrial complex IV deficiency, 220110
PEX1	115.7	97	95	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	112.1	96	90	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	168.4	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.2	99	98	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149.2	99	97	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137.3	97	93	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	93	99	99	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	76.4	100	99	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	98.1	99	94	Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	111.8	99	98	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716

PEX6	94.6	90	86	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	126.3	98	95	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	63.3	92	83	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	118.9	100	99	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	90.8	90	81	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	119.7	98	92	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147.2	100	99	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	101.3	91	83	No OMIM phenotype
PIGT	174.9	100	99	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	117.6	99	98	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	144.6	100	99	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	130.4	100	99	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	141.3	99	99	Congenital disorder of glycosylation, type Ia, 212065
PNKP	93.1	99	97	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	66.5	100	98	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	114.4	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	137	93	93	Mental retardation, autosomal dominant 36, 616362
PPT1	172.9	100	100	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	181.9	97	96	Renpenning syndrome, 309500
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PROSC	112.6	99	92	Epilepsy, early-onset, vitamin B6-dependent ,617290

PRRT2	78.9	99	98	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PSAP	114.3	99	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PURA	121.9	94	87	Mental retardation, autosomal dominant 31, 616158
PYCR2	127.8	100	97	Leukodystrophy, hypomyelinating, 10, 616420
QARS	165.5	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	113.5	100	99	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RARS2	107.1	100	99	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	142.2	100	99	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93	87	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.6	100	99	Aicardi-Goutieres syndrome 3, 610329
ROGDI	112.1	97	95	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	79	94	83	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	128.8	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	127.8	99	96	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCARB2	120.6	100	99	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	137	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	168.8	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	156.7	99	96	Epileptic encephalopathy, early infantile, 11, 613721

				Seizures, benign familial infantile, 3, 607745
SCN8A	198.5	100	99	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SIK1	85.7	97	92	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	149.9	99	97	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC13A5	164.5	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	156	99	98	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	178	100	99	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	128.1	99	99	Epileptic encephalopathy, early infantile, 41, 617105
SLC25A1	71.1	92	86	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	192.8	98	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	108.8	99	96	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	179.6	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	111.4	99	97	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	143.8	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A8	57	89	79	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	105	97	91	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	115.8	97	95	Nicolaides-Baraitser syndrome, 601358
SMC1A	101.3	99	98	Cornelia de Lange syndrome 2,300590
SMS	65.5	85	70	Mental retardation, X-linked, Snyder-Robinson type, 309583
SPTAN1	126.6	99	99	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	144.9	100	99	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	133.1	95	94	Amish infantile epilepsy syndrome, 609056
STX1B	152.2	100	98	Generalized epilepsy with febrile seizures plus, type 9, 616172

STXBP1	125.8	100	100	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	213.1	100	100	Sulfite oxidase deficiency, 272300
SYN1	64.8	74	63	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	70.9	49	49	Mental retardation, autosomal dominant 5, 612621
SYNJ1	127.2	99	96	Parkinson disease 20, early-onset, 615530
SYP	72.5	99	94	Mental retardation, X-linked 96, 300802
SZT2	147.4	99	99	Epileptic encephalopathy, early infantile, 18, 615476
TANGO2	145.6	100	100	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TBC1D24	179.4	100	100	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	127.9	99	98	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	128.7	99	99	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	156.8	99	98	Spinocerebellar ataxia, autosomal recessive, 616949
TPP1	146.4	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	242.7	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	150.6	99	98	Hypomagnesemia 1, intestinal, 602014
TSC1	128.9	99	98	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	131.3	100	99	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN54	83.1	95	92	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	113.3	99	97	Lissencephaly 3, 611603

TUBB2A	109.8	96	95	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB4A	121.5	96	95	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	164.6	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	75.3	94	76	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia, autosomal recessive 24, 617133
UBE3A	90.1	97	91	Angelman syndrome, 105830
WWOX	131.1	100	99	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XK	97.4	99	99	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	227	100	100	Epileptic encephalopathy, early infantile, 56,617665
ZEB2	163.4	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 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors