

## EPILEPSY GENE PANEL DG 2.13 (299 genes)

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
AARS	124.3	100	99	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	92.7	100	99	GABA-transaminase deficiency, 613163
ABCC8	146.6	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	99	94	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACY1	132.8	99	98	Aminoacylase 1 deficiency, 609924
ADSL	183.6	99	99	Adenylosuccinase deficiency, 103050
ALDH7A1	77.1	93	85	Epilepsy, pyridoxine-dependent, 266100
ALG1	50.9	53	48	Congenital disorder of glycosylation, type Ik, 608540
ALG11	139.6	96	96	Congenital disorder of glycosylation, type Ip, 613661
ALG13	86.7	98	94	Epileptic encephalopathy, early infantile, 36, 300884
ALG3	132.9	100	100	Congenital disorder of glycosylation, type Id, 601110
ALG6	96.4	96	93	Congenital disorder of glycosylation, type Ic, 603147
AMACR	157.9	100	100	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMT	173.1	100	100	Glycine encephalopathy, 605899
ANKRD11	96.3	97	94	KBG syndrome, 148050
AP3B2	135.1	97	94	Epileptic encephalopathy, early infantile, 48
APOPT1	63.8	81	78	Mitochondrial complex IV deficiency, 220110
ARHGEF9	60.2	76	74	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	156.7	94	89	Coffin-Siris syndrome 1, 135900
ARX	29.1	75	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	105.9	97	92	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	114.4	99	98	Argininosuccinic aciduria, 207900
ATP1A2	190.8	100	99	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	177.3	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	46.1	81	55	?Parkinsonism with spasticity, X-linked, 300911 ?Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	133.2	99	97	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	82.6	98	92	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	110.3	96	95	Mental retardation, autosomal dominant 26,615834
BOLA3	50.1	92	81	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	108.5	99	97	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	166.6	100	99	Biotinidase deficiency, 253260
CACNA1A	87.8	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	139.3	99	99	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)
CACNA2D2	135.2	93	92	No OMIM phenotype Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CAD	158.9	100	99	?Congenital disorder of glycosylation, type Iz, 616457

CASK	92.3	98	93	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	114.4	94	91	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	137.7	99	98	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	229.3	100	100	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	142.1	96	95	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRN2	247.5	98	94	Epilepsy, nocturnal frontal lobe, 3, 605375
CLCN4	123	100	99	Mental retardation, X-linked 49/15, 300114
CLDN16	136.3	100	99	Hypomagnesemia 3, renal, 248250
CLDN19	123.7	98	93	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	114.9	92	90	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	146.1	98	92	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	131.6	98	95	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	163.9	83	83	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	188.4	100	99	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	125.4	92	92	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	148	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	121.6	98	92	Mental retardation, autosomal dominant 34, 616351
COQ2	89.3	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
COQ8A	134.3	100	99	Coenzyme Q10 deficiency, primary, 4, 612016

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	162.8	97	95	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	82.5	97	82	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	163.7	98	95	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	112.8	84	80	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	72.8	98	88	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX2	85.6	99	97	No OMIM phenotype
D2HGDH	134.5	97	95	D-2-hydroxyglutaric aciduria, 600721
DCX	113.2	100	99	Lissencephaly, X-linked, 300067 Subcortical laminal heteropia, X-linked, 300067
DENND5A	123	99	97	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	148.3	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	156.7	89	87	Epileptic encephalopathy, early infantile, 31, 616346
DOCK7	114.4	97	95	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	110.7	100	100	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	131.1	91	86	Congenital disorder of glycosylation, type Ie, 608799
DPM2	102.1	100	99	Congenital disorder of glycosylation, type Iu, 615042
DPYD	158.3	95	93	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DYNC1H1	179.8	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	159.6	100	100	Mental retardation, autosomal dominant 7, 614104
EEF1A2	177.7	98	93	Epileptic encephalopathy, early infantile, 33, 616409

				Mental retardation, autosomal dominant 38, 616393
EGF	135.2	100	99	Hypomagnesemia 4, renal, 611718
EHMT1	141.4	94	92	Kleefstra syndrome, 610253
EPM2A	110.1	86	84	Epilepsy, progressive myoclonic 2A (Lafora), 254780
FA2H	94.1	87	79	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	207.7	100	100	Combined oxidative phosphorylation deficiency 14, 614946 ?Spastic paraplegia 77, autosomal recessive, 617046
FGD1	85.7	92	86	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	95.5	99	96	?Epileptic encephalopathy, early infantile, 47, 617166
FLNA	138.1	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	150.4	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	157.5	84	81	Rett syndrome, congenital variant, 613454
FOXRED1	136.6	100	99	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
FRMPD4	114.1	99	98	Mental retardation,X-linked 104, 300983
FRRS1L	103.1	68	63	Epileptic encephalopathy, early infantile, 37, 616981
FXVD2	96.7	99	99	Hypomagnesemia 2, renal, 154020
GABRA1	179.5	100	100	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	140.5	98	93	{Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	137.9	91	90	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Febrile seizures, familial, 8, 611277

				{Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAMT	93.5	90	80	Cerebral creatine deficiency syndrome 2, 612736
GCK	141.4	100	100	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	34.2	83	67	Glycine encephalopathy, 605899
GLDC	78.9	90	82	Glycine encephalopathy, 605899
GLRA1	123.3	100	100	Hyperreflexia, hereditary 1, autosomal dominant or recessive, 149400
GLRB	98.6	96	88	Hyperreflexia 2, autosomal recessive, 614619
GLUD1	74.5	94	84	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	167.8	93	93	Epileptic encephalopathy, early infantile, 17, 615473
GOSR2	127.2	95	95	Epilepsy, progressive myoclonic 6, 614018
GPC3	85.1	98	92	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	167.2	98	96	Molybdenum cofactor deficiency C, 615501
GRIA3	98.2	99	94	Mental retardation, X-linked 94, 300699
GRIN1	150.7	100	99	Mental retardation, autosomal dominant 8, 614254
GRIN2A	159.1	100	100	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	189.4	99	99	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	73.1	69	62	Epileptic encephalopathy, early infantile, 46, 617162
GRN	184.5	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HADH	110.8	98	95	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	105.8	99	96	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cbIX type ), 309541
HCN1	122.4	99	97	Epileptic encephalopathy, early infantile, 24, 615871
HDAC4	111.9	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	172.8	100	100	Holocarboxylase synthetase deficiency, 253270

HNRNPU	126.6	99	97	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	117.1	100	99	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438 ?Mental retardation, X-linked syndromic 10, 300220
HSD17B4	95.1	93	90	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
IDH2	103.5	99	96	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	73	93	82	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	113.5	99	97	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	61.2	92	82	Mental retardation, X-linked 1/78, 309530
ITPA	120.2	100	100	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAM3	158.6	100	100	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	172.3	99	99	Koolen-De Vries syndrome, 610443
KATNB1	141.7	100	100	Lissencephaly 6, with microcephaly, 616212
KCNA1	167	100	99	Episodic ataxia/myokymia syndrome, 160120
KCNA2	157.7	100	99	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	145	100	99	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	199.2	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNH1	185.8	98	98	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	213.4	89	89	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	299.5	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	120.4	94	93	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCNQ2	85.4	90	86	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110.7	98	95	Seizures, benign neonatal, type 2, 121201

KCNT1	112	95	92	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	166.7	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	112.6	97	95	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	112.1	100	99	Mental retardation, autosomal recessive 41, 615637
LAMB1	169.8	100	99	Lissencephaly 5, 615191
LGI1	163.8	97	94	Epilepsy, familial temporal lobe, 1, 600512
LIAS	133.7	99	97	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	196.2	99	99	Mental retardation, autosomal dominant 1, 156200
MECP2	87.3	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	105.7	98	94	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEF2C	137.7	97	93	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	125.1	99	98	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MLC1	103.4	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	111.8	100	99	Congenital disorder of glycosylation, type If, 609180
MPDZ	149	98	96	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219
MTHFR	126.1	98	97	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	100	99	Smith-Kingsmore syndrome, 616638



MTRR	139.1	100	99	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
NACC1	167.7	100	99	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties and delayed brain myelination, 617393
NANS	106.1	100	99	Sponyloepimetaphyseal dysplasia, Genevieve type,610442
NDUFA1	166.8	100	99	Mitochondrial complex I deficiency, 252010
NDUFA11	86.9	99	95	Mitochondrial complex I deficiency, 252010
NDUFAB1	115.6	100	100	Mitochondrial complex I deficiency, 252010
NDUFAB2	58.6	85	70	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFAB3	120.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFAB4	79.4	98	91	Mitochondrial complex I deficiency, 252010
NDUFAB5	95.7	98	94	Mitochondrial complex 1 deficiency, 252010
NDUFB3	22.6	91	59	Mitochondrial complex I deficiency, 252010
NDUFB9	120.1	99	97	?Mitochondrial complex I deficiency, 252010
NDUFS1	132.2	99	98	Mitochondrial complex I deficiency, 252010
NDUFS2	117.8	100	100	Mitochondrial complex I deficiency, 252010
NDUFS3	142.4	90	90	Leigh syndrome due to mitochondrial complex I deficiency, 256000 Mitochondrial complex I deficiency, 252010
NDUFS4	147.3	100	99	Leigh syndrome, 256000 Mitochondrial complex I deficiency, 252010
NDUFS6	119.1	99	99	Mitochondrial complex I deficiency, 252010
NDUFV1	136.7	99	97	Mitochondrial complex I deficiency, 252010
NDUFV2	69.5	78	53	Mitochondrial complex I deficiency, 252010
NECAP1	116.9	100	99	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	105.2	71	70	Periventricular nodular heterotopia 7, 617201
NEXMIF	139.2	99	99	Mental retardation, X-linked 98, 300912
NGLY1	128	100	99	Congenital disorder of deglycosylation, 615273
NHLRC1	174.2	100	100	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	168.5	100	100	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	127.5	100	99	Epilepsy, familial focal, with variable foci, 3, 617118
NRXN1	160.9	96	95	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	89.8	92	85	Mitochondrial complex I deficiency, 252010

OCLN	220.7	100	100	Band-like calcification with simplified gyration and polymicrogyria, 251290
OFD1	51.5	84	67	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OPHN1	89	99	96	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAFAH1B1	105.2	89	81	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAK3	82.8	97	91	Mental retardation, X-linked 30/47, 300558
PC	149.3	97	94	Pyruvate carboxylase deficiency, 266150
PCDH19	224.1	100	99	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	109.8	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	35.4	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	115.8	97	95	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	111.8	96	90	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	168.3	100	100	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	197.6	99	98	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	149	99	97	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	137	97	93	Peroxisome biogenesis disorder 8A, (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	92.9	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	111.7	99	98	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	94.5	90	86	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	72.8	62	58	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	62.8	92	83	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	115.6	100	99	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	90.5	90	81	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	111.3	92	87	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	147	100	99	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	101.4	91	83	?Epileptic encephalopathy, early infantile, 55, 617599
PIGT	171.3	98	98	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 ?Paroxysmal nocturnal hemoglobinuria 2, 615399
PLA2G6	117.5	99	98	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	142.8	100	99	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	129.2	100	99	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	112.6	99	92	Epilepsy, early-onset, vitamin B6-dependent ,617290
PMM2	141.1	99	99	Congenital disorder of glycosylation, type Ia, 212065
PNKP	93	99	97	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	66.4	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	134	91	91	Mental retardation, autosomal dominant 36, 616362

PPT1	144.5	90	87	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	186.1	100	100	Renpenning syndrome, 309500
PRF1	122.5	91	90	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRICKLE1	117.3	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	78.9	99	98	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PSAP	114.4	99	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PUM1	158	100	99	No OMIM phenotype
PURA	121.8	94	87	Mental retardation, autosomal dominant 31, 616158
PYCR2	127.6	100	97	Leukodystrophy, hypomyelinating, 10, 616420
QARS	166.6	100	100	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	113	100	99	Mental retardation, X-linked 72, 300271 ?Waisman syndrome, 311510
RARS2	107.2	100	99	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	142.1	100	99	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	103.8	93	87	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209.2	100	99	Aicardi-Goutieres syndrome 3, 610329
ROGDI	112.2	97	95	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	79.3	94	83	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	128.6	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	127.9	99	96	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCARB2	121.2	100	99	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	135.2	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	168.3	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	198.3	100	99	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SIK1	85.6	97	92	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	125.9	85	81	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC13A5	164.1	100	100	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	157.1	99	98	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	186.4	100	99	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	128	99	99	Epileptic encephalopathy, early infantile, 41, 617105
SLC25A1	71	92	87	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	192.5	98	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	108.7	99	96	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	190.1	92	92	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	108.8	99	96	Congenital disorder of glycosylation, type II <sub>m</sub> , 300896
SLC6A1	143.6	100	100	Myoclonic-atonic epilepsy, 616421
SLC6A8	56.5	89	79	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	104.2	97	91	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	113.8	95	93	Nicolaidis-Baraitser syndrome, 601358

SMC1A	99.4	99	98	Cornelia de Lange syndrome 2,300590
SMS	67.8	88	73	Mental retardation, X-linked, Snyder-Robinson type, 309583
SPTAN1	125.5	99	98	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	144.5	100	99	Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	121.9	84	84	Amish infantile epilepsy syndrome, 609056
STX1B	152.2	100	98	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	124.5	96	96	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	212.6	100	100	Sulfite oxidase deficiency, 272300
SYN1	64.2	74	63	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	141.3	98	98	Mental retardation, autosomal dominant 5, 612621
SYNJ1	127.2	99	96	Parkinson disease 20, early-onset, 615530
SYP	72	99	94	Mental retardation, X-linked 96, 300802
SZT2	149.5	99	99	Epileptic encephalopathy, early infantile, 18, 615476
TANGO2	145.3	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias and neurodegeneration, 616878
TBC1D24	179.2	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	128	99	98	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	128	99	99	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	165.1	99	98	Spinocerebellar ataxia, autosomal recessive, 616949
TPP1	146.3	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	242.4	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	151.1	99	98	Hypomagnesemia 1, intestinal, 602014
TSC1	128.8	99	98	Lymphangi leiomyomatosis, 606690

				Tuberous sclerosis-1, 191100
TSC2	131.2	100	99	Lymphangioliomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN54	82.9	95	92	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	113.2	99	97	Lissencephaly 3, 611603
TUBB2A	109.9	96	95	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB4A	121.2	96	95	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	164.2	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	75.4	94	77	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia, autosomal recessive 24, 617133
UBE3A	89.8	97	91	Angelman syndrome, 105830
WWOX	130.9	100	99	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	96.8	99	99	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	226.6	100	100	Epileptic encephalopathy, early infantile, 56,617665
ZEB2	157	99	98	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 18th, 2018.

This list is accurate for panel version DG 2.13

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