EPILEPSY GENE PANEL DG 2.16 (328 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
AARS	103,7	100.0%	99.5%	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339
ABAT	83,2	99.9%	98.3%	GABA-transaminase deficiency, 613163
ABCC8	125,8	100.0%	99.9%	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	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACY1	118,5	100.0%	98.6%	Aminoacylase 1 deficiency, 609924
ADSL	138,6	99.2%	98.6%	Adenylosuccinase deficiency, 103050
ALDH7A1	66,7	93.5%	86.1%	Epilepsy, pyridoxine-dependent, 266100
ALG1	46,5	53.2%	50.2%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	129,3	96.8%	96.3%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	77,3	98.5%	92.1%	?Congenital disorder of glycosylation, type Is, 300884 Epileptic encephalopathy, early infantile, 36, 300884
ALG3	106,5	100.0%	99.9%	Congenital disorder of glycosylation, type Id, 601110
ALG6	101,6	99.1%	95.6%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	157,7	100.0%	100.0%	Alpha-methylacyl-CoA racemase deficiency, 614307 Bile acid synthesis defect, congenital, 4, 214950
AMPD2	132,3	100.0%	99.9%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	142,7	100.0%	100.0%	Glycine encephalopathy, 605899
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
AP3B2	125,6	99.4%	97.6%	Epileptic encephalopathy, early infantile, 48, 617276
APOPT1	NC	NC	NC	Mitochondrial complex IV deficiency, 220110
ARHGEF9	51,5	76.1%	71.3%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	139,6	99.5%	99.2%	Coffin-Siris syndrome 1, 135900

ARX	49,3	87.3%	79.2%	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	125,7	99.3%	97.2%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASNS	82,8	98.6%	92.2%	Asparagine synthetase deficiency, 615574
ASXL3	138,1	99.7%	99.1%	Bainbridge-Ropers syndrome, 615485
ATP1A2	161,7	100.0%	99.5%	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	159,8	100.0%	100.0%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP6AP2	44,9	88.4%	64.2%	?Parkinsonism with spasticity, X-linked, 300911 Mental retardation, X-linked, syndromic, Hedera type, 300423
ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	89,2	99.1%	95.5%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	130,6	99.5%	97.7%	Mental retardation, autosomal dominant 26, 615834
BOLA3	48,1	99.9%	92.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	142	100.0%	99.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056 Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
CACNA1A	92,4	97.8%	94.7%	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	120,9	99.8%	99.2%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA2D2	126	95.8%	94.0%	No OMIM phenotype

				Epileptic encephalopathy (Pippucci (2013) PLoS One 8,e82154) ?Schizophrenia (Purcell (2014) Nature 506, 185)
CACNB4	97,8	97.2%	95.5%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CAD	136,7	99.9%	99.2%	Epileptic encephalopathy, early infantile, 50, 616457
CASK	85,1	99.5%	94.5%	FG syndrome 4, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 Mental retardation, with or without nystagmus, 300422
CDKL5	100	95.1%	93.1%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	123,9	99.3%	99.0%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	174,4	100.0%	100.0%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	109,8	99.9%	99.2%	Epilepsy, nocturnal frontal lobe, 1, 600513 {Nicotine addiction, susceptibility to}, 188890
CHRNB2	160,6	99.7%	98.0%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	72,7	64.7%	63.3%	Mental retardation, autosomal dominant 45, 617600
CLCN4	105,7	99.9%	98.9%	Raynaud-Claes syndrome, 300114
CLDN16	126,6	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	125,4	99.1%	95.1%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	114,7	92.6%	91.9%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	138,7	99.9%	98.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	132,3	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 6, 601780 Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300
CLN8	144,5	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	199,8	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	123,7	92.7%	92.7%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	127,1	100.0%	99.8%	Cortical dysplasia-focal epilepsy syndrome, 610042 Pitt-Hopkins like syndrome 1, 610042 {Autism susceptibility 15}, 612100
COL4A1	95,7	99.6%	97.3%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COL4A3BP	133,1	99.7%	97.8%	Mental retardation, autosomal dominant 34, 616351

COLGALT1	149,5	97.8%	92.1%	Brain small vessel disease 3, 618360
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	109,1	99.4%	96.9%	Epilepsy, familial temporal lobe, 5, 614417 Febrile seizures, familial, 11, 614418
CPS1	133,8	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300 {Pulmonary hypertension, neonatal, susceptibility to}, 615371 {Venoocclusive disease after bone marrow transplantation}, 0
CPT2	139,2	98.3%	98.2%	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	70	99.3%	90.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	171	99.8%	97.8%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	107	91.3%	81.8%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	78	97.6%	89.3%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX2	120,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 67, 618141
D2HGDH	142	100.0%	99.4%	D-2-hydroxyglutaric aciduria, 600721
DCX	90,5	99.9%	98.4%	Lissencephaly, X-linked, 300067 Subcortical laminal heterotopia, X-linked, 300067
DDX3X	73,5	86.2%	82.9%	Mental retardation, X-linked 102, 300958
DENND5A	99,2	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	124,5	99.9%	99.7%	Epilepsy, familial focal, with variable foci 1, 604364
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DLAT	100,2	99.8%	99.2%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	188,2	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	139,8	94.7%	92.3%	Epileptic encephalopathy, early infantile, 31, 616346
DOCK7	120,7	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	87,5	100.0%	99.9%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	134,7	95.2%	88.2%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	88,5	99.8%	97.6%	Congenital disorder of glycosylation, type Iu, 615042

DPYD	141,6	99.5%	96.4%	5-fluorouracil toxicity, 274270 Dihydropyrimidine dehydrogenase deficiency, 274270
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748
DYNC1H1	140,6	100.0%	99.6%	Charcot-Marie-Tooth disease, axonal, type 20, 614228 Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	130,4	100.0%	99.9%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	188,3	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFHC1	117,2	93.0%	90.7%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631 {Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770
EGF	110,5	100.0%	99.7%	Hypomagnesemia 4, renal, 611718
EHMT1	127,7	94.6%	94.2%	Kleefstra syndrome 1, 610253
EPM2A	116,5	90.9%	88.8%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
EXOSC3	125,1	96.4%	87.8%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	161,9	100.0%	100.0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FGD1	86,7	98.4%	93.0%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF12	100,4	100.0%	99.9%	Epileptic encephalopathy, early infantile, 47, 617166
FLNA	142,7	100.0%	99.9%	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	142	97.8%	88.7%	Rett syndrome, congenital variant, 613454
FOXRED1	121	99.8%	98.2%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	108,4	99.6%	97.2%	Mental retardation, X-linked 104, 300983
FRRS1L	99,1	85.5%	79.1%	Epileptic encephalopathy, early infantile, 37, 616981
FXYD2	108,1	100.0%	100.0%	Hypomagnesemia 2, renal, 154020

GABRA1	164,2	100.0%	99.8%	Epileptic encephalopathy, early infantile, 19, 615744 {Epilepsy, childhood absence, susceptibility to, 4}, 611136 {Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136
GABRB3	131,7	99.5%	97.8%	Epileptic encephalopathy, early infantile, 43, 617113 {Epilepsy, childhood absence, susceptibility to, 5}, 612269
GABRG2	126,4	91.1%	89.7%	Epilepsy, generalized, with febrile seizures plus, type 3, 611277 Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 611277 {Epilepsy, childhood absence, susceptibility to, 2}, 607681
GAMT	112,5	98.3%	91.5%	Cerebral creatine deficiency syndrome 2, 612736
GCK	138,6	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 Hyperinsulinemic hypoglycemia, familial, 3, 602485 MODY, type II, 125851
GCSH	32,1	88.4%	69.8%	?Glycine encephalopathy, 605899
GLDC	59,2	90.6%	79.2%	Glycine encephalopathy, 605899
GLRA1	96,8	100.0%	99.7%	Hyperekplexia 1, 149400
GLRB	104,6	99.5%	94.5%	Hyperekplexia 2, 614619
GLUD1	65,5	98.1%	87.5%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	152,8	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	102,6	95.8%	93.7%	Epilepsy, progressive myoclonic 6, 614018
GPC3	75,7	98.7%	92.7%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	144,7	99.9%	98.8%	Molybdenum cofactor deficiency C, 615501
GRIA3	82,4	98.6%	92.1%	Mental retardation, X-linked 94, 300699
GRIN1	166,1	100.0%	99.9%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2A	131,2	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	158	99.8%	99.0%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	82,8	91.9%	79.5%	Epileptic encephalopathy, early infantile, 46, 617162
GRN	174,1	100.0%	100.0%	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485

HACE1	136,2	99.9%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	111,1	99.3%	98.8%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	104,4	99.3%	95.8%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	137,5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 24, 615871
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	149,9	99.9%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	92,4	100.0%	98.4%	HSD10 mitochondrial disease, 300438
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ICK	110	99.8%	99.3%	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
IDH2	98,5	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	106,3	94.3%	82.8%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	110,8	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	73,1	95.8%	87.9%	Mental retardation, X-linked 1/78, 309530
ITPA	130,2	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647 [Inosine triphosphatase deficiency], 613850
JAM3	126,9	100.0%	99.9%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	139,1	99.8%	98.6%	Koolen-De Vries syndrome, 610443
KATNB1	154,2	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA1	150,1	100.0%	100.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	126,3	100.0%	99.7%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	129,7	100.0%	99.7%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	170,9	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	148,4	98.7%	98.3%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	199,7	100.0%	100.0%	Diabetes mellitus, transient neonatal, 3, 610582 Diabetes, permanent neonatal, with or without neurologic features, 606176 Hyperinsulinemic hypoglycemia, familial, 2, 601820 Maturity-onset diabetes of the young, type 13, 616329 {Diabetes mellitus, type 2, susceptibility to}, 125853
KCNMA1	102,3	94.8%	93.4%	?Cerebellar atrophy, developmental delay, and seizures, 617643

				Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ2	118,3	91.5%	90.2%	Epileptic encephalopathy, early infantile, 7, 613720 Myokymia, 121200 Seizures, benign neonatal, 1, 121200
KCNQ3	110,4	99.9%	98.7%	Seizures, benign neonatal, 2, 121201
KCNT1	131,2	96.0%	95.1%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	154,9	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	102,8	99.5%	97.0%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	145,7	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
LAMB1	142,9	100.0%	99.7%	Lissencephaly 5, 615191
LGI1	133,3	98.3%	97.2%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	125,3	99.9%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	147,7	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200
MECP2	124,8	100.0%	98.5%	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	85,1	99.5%	95.5%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEF2C	127,9	99.4%	95.5%	Chromosome 5q14.3 deletion syndrome, 613443 Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFSD8	121,3	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 7, 610951 Macular dystrophy with central cone involvement, 616170
MLC1	96,7	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	91,2	98.8%	95.7%	Molybdenum cofactor deficiency A, 252150
MOCS2	137,7	99.6%	99.5%	Molybdenum cofactor deficiency B, 252160
MPDU1	102,4	100.0%	99.6%	Congenital disorder of glycosylation, type If, 609180
MPDZ	128,3	99.6%	98.2%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050

				{Vascular disease, susceptibility to}, 0
MTOR	112	99.9%	99.1%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MTRR	131,1	100.0%	99.0%	Homocystinuria-megaloblastic anemia, cbl E type, 236270 {Neural tube defects, folate-sensitive, susceptibility to}, 601634
NACC1	169,5	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NDUFA1	184,9	99.9%	99.2%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	116	99.8%	97.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	98,5	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	54,1	94.3%	82.0%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	141	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98,3	99.2%	94.5%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	124,9	99.9%	99.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	23,3	89.7%	62.5%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	105,2	97.8%	93.3%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	143,5	99.9%	99.8%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100,1	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	124,8	90.7%	90.5%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	144,5	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	111,9	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	141,7	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	74,2	92.4%	77.3%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	102,3	100.0%	100.0%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	93,7	72.3%	71.5%	Periventricular nodular heterotopia 7, 617201
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEXMIF	132	100.0%	99.5%	Mental retardation, X-linked 98, 300912
NGLY1	135,4	100.0%	99.7%	Congenital disorder of deglycosylation, 615273
NHLRC1	169,7	100.0%	100.0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	138,7	100.0%	100.0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	120,3	100.0%	99.8%	Epilepsy, familial focal, with variable foci 3, 617118
NRXN1	141,6	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325 {Schizophrenia, susceptibility to, 17}, 614332
NUBPL	102	98.9%	95.5%	Mitochondrial complex I deficiency, nuclear type 21, 618242

OCLN	173,9	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPHN1	78,3	98.9%	95.0%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAFAH1B1	77	92.0%	82.8%	Lissencephaly 1, 607432 Subcortical laminar heterotopia, 607432
PAK3	85,3	98.6%	93.7%	Mental retardation, X-linked 30/47, 300558
PC	155,4	99.9%	98.7%	Pyruvate carboxylase deficiency, 266150
PCDH19	176,6	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	85,3	98.9%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	111,4	99.2%	97.2%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	129,1	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	72,9	99.1%	95.2%	MODY, type IV, 606392 Pancreatic agenesis 1, 260370 {Diabetes mellitus, type II, susceptibility to}, 125853
PET100	87,9	98.0%	87.6%	Mitochondrial complex IV deficiency, 220110
PEX1	127,9	99.9%	99.3%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1A (Zellweger), 214100 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539
PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX12	120,6	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	179,6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	130,5	99.8%	97.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	140,8	98.6%	94.8%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	84,9	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	94,3	100.0%	99.6%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	113,9	99.9%	99.2%	?Peroxisome biogenesis disorder 10B, 617370 Peroxisome biogenesis disorder 10A (Zellweger), 614882
PEX5	107,9	100.0%	99.2%	Peroxisome biogenesis disorder 2A (Zellweger), 214110 Peroxisome biogenesis disorder 2B, 202370

				Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	106,5	98.5%	92.0%	Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862 Peroxisome biogenesis disorder 4B, 614863
PGAP3	70,3	63.5%	59.9%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	60,3	98.2%	87.9%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	144,5	100.0%	99.9%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	89,3	94.8%	86.0%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGT	159,3	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLCB1	134,9	100.0%	99.7%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	112,8	99.7%	97.7%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	95,3	99.6%	95.3%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	109	100.0%	99.9%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	74,4	100.0%	99.3%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	113,9	100.0%	99.6%	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	129,1	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	136,6	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPT1	136,6	90.2%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	163,5	100.0%	100.0%	Renpenning syndrome, 309500
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553

				Lymphoma, non-Hodgkin, 605027
PRICKLE1	100	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	111,8	100.0%	99.0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PUM1	126,9	100.0%	99.5%	Spinocerebellar ataxia 47, 617931
PURA	207,1	99.5%	96.9%	Mental retardation, autosomal dominant 31, 616158
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	129,2	100.0%	99.8%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	102	100.0%	99.9%	?Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	127,6	100.0%	99.4%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	87,8	98.3%	93.0%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RRM2B	143,9	99.9%	99.4%	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	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCARB2	105,8	99.8%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	121,4	100.0%	99.1%	Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208 Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634
SCN1B	169,7	99.9%	98.1%	Atrial fibrillation, familial, 13, 615377 Brugada syndrome 5, 612838 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Epileptic encephalopathy, early infantile, 52, 617350

SCN2A	132,4	99.6%	97.7%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	138,7	99.9%	99.1%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	154,3	100.0%	99.7%	?Myoclonus, familial, 2, 618364 Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	101,2	99.8%	97.4%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	123,9	97.5%	91.6%	Phelan-McDermid syndrome, 606232 {Schizophrenia 15}, 613950
SIK1	118,5	99.6%	96.7%	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	111,9	86.1%	84.1%	Epileptic encephalopathy, early infantile, 34, 616645 {Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685
SLC13A5	141,9	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	138,1	100.0%	99.2%	Erythrocyte lactate transporter defect, 245340 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	97,1	99.3%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC25A1	103,2	99.3%	95.1%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	123,8	100.0%	99.1%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	148,9	92.8%	92.8%	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	104,8	99.8%	98.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	126	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A8	53,5	96.1%	83.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	101	98.6%	94.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	105,9	96.8%	95.9%	Nicolaides-Baraitser syndrome, 601358
SMC1A	87,8	99.9%	97.8%	Cornelia de Lange syndrome 2, 300590
SMS	63	88.9%	73.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583

SNAP25	119	99.9%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SPATA5	139,5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPTAN1	112	99.1%	98.3%	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	134,7	100.0%	99.5%	?Epileptic encephalopathy, early infantile, 15, 615006 Mental retardation, autosomal recessive 12, 611090
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STX1B	157,7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	103,7	96.8%	96.4%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SYN1	66,6	90.6%	79.1%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	140,7	98.4%	97.7%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126,6	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	79,9	99.9%	98.1%	Mental retardation, X-linked 96, 300802
SZT2	135,6	99.6%	99.4%	Epileptic encephalopathy, early infantile, 18, 615476
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	92,7	99.2%	95.4%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	177,7	100.0%	100.0%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	116,4	98.7%	94.7%	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Kenny-Caffey syndrome, type 1, 244460
TCF4	109,2	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700

TRPM6	126,6	99.9%	99.1%	Hypomagnesemia 1, intestinal, 602014
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSEN15	89,8	99.7%	96.4%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	95,6	99.9%	98.9%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	114,4	99.4%	96.8%	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TUBA1A	77,6	99.8%	97.1%	Lissencephaly 3, 611603
TUBB2A	77,1	99.7%	97.2%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	78,2	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB4A	101,2	97.1%	95.6%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	154,3	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	79,9	97.7%	86.6%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE3A	81,5	98.6%	93.3%	Angelman syndrome, 105830
VPS53	111,3	91.1%	89.6%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	98,2	99.5%	97.4%	Skraban-Deardorff syndrome, 617616
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
WWOX	116,1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	85,4	100.0%	99.4%	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	167,1	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
ZEB2	140,1	99.7%	98.4%	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors