EPILEPSY GENE PANEL DG 2.17 (336 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS	109.2	100.0%	99.7%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABAT	86.1	99.9%	98.4%	GABA-transaminase deficiency, 613163
ABCC8	134.7	100.0%	99.9%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTL6B	144.9	100.0%	100.0%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	128.5	99.9%	99.1%	Aminoacylase 1 deficiency, 609924
ADSL	147.2	99.2%	98.9%	Adenylosuccinase deficiency, 103050
ALDH7A1	68.0	94.2%	86.7%	Epilepsy, pyridoxine-dependent, 266100
ALG1	51.3	53.6%	52.1%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	132.1	96.8%	96.5%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	77.7	98.6%	92.4%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG3	117.9	100.0%	100.0%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98.1	98.9%	94.9%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	168.4	100.0%	100.0%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMPD2	146.3	100.0%	100.0%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	151.3	100.0%	100.0%	Glycine encephalopathy, 605899
ANKRD11	131.8	99.6%	97.6%	KBG syndrome, 148050
AP3B2	135.0	99.8%	97.9%	Epileptic encephalopathy, early infantile, 48, 617276
APOPT1	80.4	82.1%	82.1%	Mitochondrial complex IV deficiency, 220110
ARHGEF9	52.4	76.3%	72.8%	Epileptic encephalopathy, early infantile, 8, 300607

ARID1B	150.6	99.5%	99.3%	Coffin-Siris syndrome 1, 135900
ARX	58.2	90.9%	83.3%	Proud syndrome, 300004 Lissencephaly, X-linked 2, 300215 Partington syndrome, 309510 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	124.7	99.6%	96.8%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	135.7	99.9%	99.2%	Argininosuccinic aciduria, 207900
ASNS	81.9	97.9%	91.0%	Asparagine synthetase deficiency, 615574
ASXL3	141.2	99.8%	99.1%	Bainbridge-Ropers syndrome, 615485
ATP1A2	173.7	100.0%	99.8%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	173.9	100.0%	100.0%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP6AP2	45.5	89.4%	67.3%	Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	86.2	99.1%	95.1%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	143.1	99.8%	98.7%	Mental retardation, autosomal dominant 26, 615834
BOLA3	50.9	99.8%	94.6%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	155.4	100.0%	99.6%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BTD	135.6	100.0%	99.8%	Biotinidase deficiency, 253260
CACNA1A	101.2	98.2%	96.2%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500
CACNA1E	129.3	100.0%	99.5%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA2D2	139.2	96.6%	94.3%	Cerebellar atrophy with seizures and variable developmental delay, 618501

CACNB4	98.5	97.3%	96.2%	Episodic ataxia, type 5, 613855
CAD	147.4	100.0%	99.6%	Epileptic encephalopathy, early infantile, 50, 616457
CASK	84.5	99.4%	94.0%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CDKL5	102.7	95.0%	92.9%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	126.1	99.4%	99.1%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	192.3	100.0%	100.0%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	125.3	100.0%	99.3%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB2	176.5	100.0%	98.9%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	83.4	64.8%	63.4%	Mental retardation, autosomal dominant 45, 617600
CLCN4	111.7	99.9%	98.8%	Raynaud-Claes syndrome, 300114
CLDN16	132.1	100.0%	100.0%	Hypomagnesemia 3, renal, 248250
CLDN19	141.2	99.8%	97.2%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	123.4	92.5%	92.2%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	139.4	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	141.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	156.2	83.5%	83.5%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	222.5	100.0%	100.0%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	132.4	92.7%	92.7%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	129.8	100.0%	99.8%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COL4A1	100.3	99.8%	98.0%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A3BP	132.6	99.6%	97.3%	Mental retardation, autosomal dominant 34, 616351
COLGALT1	157.8	98.6%	93.8%	Brain small vessel disease 3, 618360
COQ2	107.7	97.7%	97.0%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	116.2	91.7%	90.8%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	177.7	100.0%	100.0%	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	110.2	99.7%	97.2%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPS1	133.4	100.0%	99.9%	Carbamoylphosphate synthetase I deficiency, 237300

CPT2	152.7	98.3%	98.3%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CSTB	74.8	99.1%	93.0%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	187.3	100.0%	99.0%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	116.2	94.6%	83.7%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	75.8	97.7%	88.2%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX2	134.5	100.0%	99.4%	Epileptic encephalopathy, early infantile, 67, 618141
D2HGDH	157.7	100.0%	99.8%	D-2-hydroxyglutaric aciduria, 600721
DCX	93.7	99.8%	98.9%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDX3X	74.5	86.2%	82.8%	Mental retardation, X-linked 102, 300958
DENND5A	101.5	99.8%	98.9%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	129.9	100.0%	99.8%	Epilepsy, familial focal, with variable foci 1, 604364
DHDDS	84.5	97.3%	94.0%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DLAT	104.9	100.0%	99.4%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	203.8	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	151.2	94.9%	93.3%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	120.8	99.9%	97.7%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DOCK7	118.3	99.6%	97.8%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	93.2	100.0%	100.0%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	134.2	95.5%	87.7%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	95.4	100.0%	99.1%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	140.7	99.4%	96.2%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	121.4	100.0%	99.9%	Dihydropyrimidinuria, 222748
DYNC1H1	149.0	100.0%	99.8%	Mental retardation, autosomal dominant 13, 614563 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 Charcot-Marie-Tooth disease, axonal, type 20, 614228
DYRK1A	134.0	100.0%	100.0%	Mental retardation, autosomal dominant 7, 614104
EEF1A2	209.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFHC1	117.7	93.1%	91.1%	No OMIM disease ID

EGF	111.5	100.0%	99.8%	Hypomagnesemia 4, renal, 611718
EHMT1	138.4	94.7%	94.5%	Kleefstra syndrome 1, 610253
EPM2A	125.2	94.8%	90.2%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ETHE1	105.9	99.9%	97.9%	Ethylmalonic encephalopathy, 602473
EXOSC3	135.7	96.5%	87.0%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	101.5	99.3%	95.1%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	169.5	100.0%	100.0%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FGD1	93.2	98.7%	94.4%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	105.7	100.0%	100.0%	Epileptic encephalopathy, early infantile, 47, 617166
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FOLR1	115.7	100.0%	100.0%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	162.5	99.7%	96.6%	Rett syndrome, congenital variant, 613454
FOXRED1	129.1	99.9%	99.0%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	115.6	99.7%	97.7%	Mental retardation, X-linked 104, 300983
FRRS1L	100.0	89.3%	81.8%	Epileptic encephalopathy, early infantile, 37, 616981
FXYD2	118.4	100.0%	100.0%	Hypomagnesemia 2, renal, 154020
GABRA1	164.8	100.0%	100.0%	Epileptic encephalopathy, early infantile, 19, 615744
GABRB3	139.5	99.7%	98.5%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	126.9	91.0%	90.0%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAMT	125.7	99.7%	94.3%	Cerebral creatine deficiency syndrome 2, 612736
GCK	152.9	100.0%	100.0%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176 MODY, type II, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485

GCSH	32.0	94.3%	74.1%	?Glycine encephalopathy, 605899
GLDC	60.8	91.8%	80.4%	Glycine encephalopathy, 605899
GLRA1	103.2	100.0%	99.8%	Hyperekplexia 1, 149400
GLRB	103.4	99.5%	94.9%	Hyperekplexia 2, 614619
GLUD1	66.3	98.0%	88.9%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	160.9	93.8%	93.8%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	108.1	95.9%	94.1%	Epilepsy, progressive myoclonic 6, 614018
GPC3	76.8	98.9%	93.5%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	147.8	99.8%	98.8%	Molybdenum cofactor deficiency C, 615501
GRIA3	83.0	98.9%	93.2%	Mental retardation, X-linked 94, 300699
GRIN1	186.6	100.0%	100.0%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	139.8	100.0%	100.0%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	168.9	99.9%	99.2%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	92.8	96.7%	85.3%	Epileptic encephalopathy, early infantile, 46, 617162
GRN	193.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HACE1	135.7	100.0%	99.4%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	118.2	99.3%	99.2%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	114.5	99.5%	96.5%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	144.1	100.0%	99.9%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	154.7	100.0%	99.3%	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	98.0	100.0%	99.5%	HSD10 mitochondrial disease, 300438
HSD17B4	106.4	95.5%	93.1%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ICK	111.0	100.0%	98.8%	Endocrine-cerebroosteodysplasia, 612651
IDH2	107.4	100.0%	99.6%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	108.7	88.3%	80.0%	Microcephaly, epilepsy, and diabetes syndrome, 614231

IFIH1	110.9	99.8%	98.2%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	81.5	96.6%	90.5%	Mental retardation, X-linked 1/78, 309530
IRF2BPL	197.8	99.6%	97.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	142.5	100.0%	100.0%	Epileptic encephalopathy, early infantile, 35, 616647
JAM3	132.0	100.0%	100.0%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	156.7	99.8%	99.1%	Koolen-De Vries syndrome, 610443
KATNB1	170.5	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA1	164.5	100.0%	100.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	132.6	100.0%	99.4%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	141.6	100.0%	99.8%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	189.4	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	159.4	98.7%	98.4%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	157.5	89.3%	88.6%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	222.1	100.0%	100.0%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	107.9	94.9%	93.6%	Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ2	133.0	91.5%	90.4%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	116.5	100.0%	98.7%	Seizures, benign neonatal, 2, 121201
KCNT1	145.3	96.3%	95.2%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	171.1	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	111.1	99.6%	97.5%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KPTN	163.9	100.0%	100.0%	Mental retardation, autosomal recessive 41, 615637
LAMB1	147.7	100.0%	99.7%	Lissencephaly 5, 615191
LGI1	132.9	98.5%	97.4%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	124.4	100.0%	98.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
MBD5	151.8	99.9%	99.8%	Mental retardation, autosomal dominant 1, 156200

MECP2	135.2	100.0%	99.5%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MED12	89.4	99.6%	96.5%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEF2C	131.2	99.5%	95.7%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFF	86.8	93.5%	88.5%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	117.4	100.0%	99.6%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	102.4	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	101.3	99.3%	96.6%	Molybdenum cofactor deficiency A, 252150
MOCS2	134.2	99.6%	99.6%	Molybdenum cofactor deficiency B, 252160
MPDU1	110.1	100.0%	99.8%	Congenital disorder of glycosylation, type If, 609180
MPDZ	127.3	99.6%	98.1%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTHFR	124.2	98.5%	96.7%	Homocystinuria due to MTHFR deficiency, 236250
MTOR	116.6	100.0%	99.2%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTRR	135.6	100.0%	99.2%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
NACC1	188.0	100.0%	100.0%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	105.0	100.0%	99.3%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NDUFA1	195.4	99.9%	98.7%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	129.7	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100.2	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	56.3	94.0%	79.5%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	159.3	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	98.6	99.4%	93.8%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	128.0	99.8%	99.3%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	22.8	89.3%	61.0%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	114.2	98.3%	94.1%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	140.7	100.0%	99.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	102.9	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	132.3	90.7%	90.6%	Mitochondrial complex I deficiency, nuclear type 8, 618230

NDUFS4	148.6	100.0%	99.6%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	120.0	100.0%	100.0%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	154.4	100.0%	99.5%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	71.5	91.8%	77.9%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	108.4	100.0%	100.0%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	96.3	72.4%	71.5%	Periventricular nodular heterotopia 7, 617201
NEU1	150.1	99.5%	96.5%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	135.1	99.9%	99.4%	Mental retardation, X-linked 98, 300912
NGLY1	134.1	100.0%	99.9%	Congenital disorder of deglycosylation, 615273
NHLRC1	184.6	100.0%	100.0%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	150.6	100.0%	100.0%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	129.2	100.0%	99.9%	Epilepsy, familial focal, with variable foci 3, 617118
NRXN1	147.9	97.6%	97.3%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	102.4	98.9%	94.2%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OCLN	179.8	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPHN1	80.7	99.0%	95.5%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PAFAH1B1	78.7	91.7%	82.7%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAK3	85.7	99.0%	94.4%	Mental retardation, X-linked 30/47, 300558
PC	170.2	99.9%	98.8%	Pyruvate carboxylase deficiency, 266150
PCDH19	192.4	99.9%	98.9%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	88.0	98.6%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	113.7	99.6%	97.7%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDP1	134.9	100.0%	100.0%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	83.7	99.8%	96.8%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392
PET100	95.2	99.7%	90.6%	Mitochondrial complex IV deficiency, 220110
PEX1	126.3	100.0%	99.1%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	123.8	100.0%	98.4%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870

PEX12	125.4	100.0%	100.0%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	189.6	100.0%	100.0%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	144.7	99.8%	98.8%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	157.0	98.9%	95.7%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	85.8	100.0%	98.9%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	105.1	100.0%	100.0%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	108.6	100.0%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	115.8	100.0%	99.4%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	117.6	99.1%	93.9%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PGAP3	74.5	63.7%	60.6%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	57.7	96.9%	84.1%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	103.7	93.6%	90.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	157.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	88.3	95.6%	86.3%	?Epileptic encephalopathy, early infantile, 55, 617599
PIGT	169.4	98.1%	98.1%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLCB1	134.5	100.0%	99.8%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	113.6	99.9%	98.2%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	97.2	99.9%	97.2%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	130.3	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065

PNKP	123.1	100.0%	100.0%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	78.3	100.0%	99.2%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	124.4	100.0%	99.8%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	138.5	91.6%	91.6%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	145.7	100.0%	100.0%	Mental retardation, autosomal dominant 35, 616355
PPT1	140.2	90.3%	89.2%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	173.3	100.0%	100.0%	Renpenning syndrome, 309500
PRF1	154.3	91.2%	90.7%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRICKLE1	104.3	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	124.1	100.0%	99.7%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PSAP	103.3	100.0%	99.5%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PUM1	132.0	100.0%	99.7%	Spinocerebellar ataxia 47, 617931
PURA	233.4	99.9%	98.8%	Mental retardation, autosomal dominant 31, 616158
PYCR2	129.0	99.7%	97.6%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	137.7	100.0%	100.0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
RAB39B	108.8	100.0%	100.0%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RARS2	102.7	100.0%	99.3%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	143.0	100.0%	100.0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	98.0	99.8%	96.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	314.2	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	141.6	100.0%	99.9%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	84.8	98.2%	91.5%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600

RRM2B	142.6	100.0%	99.4%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCARB2	106.4	99.9%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	120.1	99.9%	99.0%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
SCN1B	186.5	100.0%	99.3%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	135.0	99.5%	97.4%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	139.1	99.9%	98.8%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	162.9	100.0%	99.9%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SEPSECS	160.6	100.0%	100.0%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	99.0	99.8%	97.6%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	143.0	98.1%	93.8%	Phelan-McDermid syndrome, 606232
SIK1	131.1	99.8%	98.1%	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	121.0	86.3%	84.2%	Epileptic encephalopathy, early infantile, 34, 616645
SLC13A5	155.1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	140.0	100.0%	99.3%	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC19A3	139.2	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	100.8	99.5%	97.2%	Epileptic encephalopathy, early infantile, 41, 617105
SLC25A1	114.2	99.8%	97.0%	?Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182

SLC25A15	152.1	98.4%	94.4%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	138.5	100.0%	99.7%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	160.0	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC35A2	114.2	100.0%	99.1%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	136.0	100.0%	100.0%	Myoclonic-atonic epilepsy, 616421
SLC6A8	58.5	97.6%	87.8%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	104.0	98.5%	93.3%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	109.3	97.3%	96.3%	Nicolaides-Baraitser syndrome, 601358
SMC1A	93.2	99.9%	98.3%	Cornelia de Lange syndrome 2, 300590
SMPD4	108.3	99.9%	96.9%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	63.5	88.9%	74.1%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	121.8	100.0%	99.7%	?Myasthenic syndrome, congenital, 18, 616330
SPATA5	142.5	100.0%	99.8%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPTAN1	118.5	99.1%	98.6%	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	143.4	100.0%	99.8%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	104.4	89.3%	85.5%	Salt and pepper developmental regression syndrome, 609056
STX1B	173.7	100.0%	100.0%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	108.2	96.8%	96.5%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	180.8	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SYN1	73.1	93.6%	84.0%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	152.9	98.5%	98.0%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	126.3	99.9%	98.5%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530
SYP	87.1	99.9%	98.6%	Mental retardation, X-linked 96, 300802
SZT2	146.3	99.6%	99.5%	Epileptic encephalopathy, early infantile, 18, 615476
TANGO2	139.6	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	89.8	98.8%	94.7%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	199.9	100.0%	100.0%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338

				Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBCD	145.8	98.8%	95.5%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	117.0	99.3%	95.6%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TCF4	111.6	100.0%	99.8%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	175.8	100.0%	99.9%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	153.2	100.0%	100.0%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	130.2	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRPM6	128.5	99.9%	99.0%	Hypomagnesemia 1, intestinal, 602014
TSC1	117.4	99.6%	98.4%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	155.5	100.0%	100.0%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	93.1	99.9%	96.9%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100.3	100.0%	99.1%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	129.0	99.7%	97.9%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	82.5	99.9%	97.8%	Lissencephaly 3, 611603
TUBB2A	83.6	99.8%	98.0%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	88.3	100.0%	99.9%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB4A	114.3	97.8%	95.9%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBG1	162.6	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	79.3	96.9%	83.9%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE3A	80.9	98.8%	92.5%	Angelman syndrome, 105830
UBTF	127.3	100.0%	99.8%	Neurodegeneration, childhood-onset, with brain atrophy, 617672

UGP2	125.9	99.1%	98.5%	No OMIM Disease ID
VPS53	117.2	91.3%	89.9%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	97.7	99.9%	98.4%	Skraban-Deardorff syndrome, 617616
WDR45	74.7	97.1%	90.6%	Neurodegeneration with brain iron accumulation 5, 300894
WWOX	122.0	100.0%	100.0%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XK	88.8	99.9%	99.5%	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	181.4	100.0%	100.0%	Epileptic encephalopathy, early infantile, 56, 617665
ZEB2	145.0	99.7%	98.6%	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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors