EPILEPSY PANEL DG-4.3.0 (451 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AARS1	100%	100%	100%	100%	99%	Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691
ABAT	100%	100%	100%	100%	98.7%	GABA-transaminase deficiency, 613163

ABCC8	100%	100%	100%	99.9%	98.8%	Diabetes mellitus, permanent neonatal 3, 618857;Maturity-onset diabetes of the young, type 12, 621196;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450
ACO2	93.3%	90.8%	100%	100%	98.9%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559

ACTB	100%	100%	100%	100%	99.1%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTL6B	100%	100%	100%	99.9%	98.3%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100%	100%	100%	99.9%	98.3%	Aminoacylase 1 deficiency, 609924
ADAM22	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 61, 617933

ADARB1	94.9%	94.7%	100%	100%	99.1%	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures, 618862
ADSL	100%	100%	100%	99.9%	98.7%	Adenylosuccinase deficiency, 103050
AFG2A	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AGA	100%	100%	100%	100%	99.8%	Aspartylglucosaminuria, 208400
ALDH4A1	100%	100%	100%	99.7%	98.4%	Hyperprolinemia, type II, 239510
ALDH5A1	100%	100%	100%	99.8%	98.9%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100%	100%	100%	100%	99.3%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALG1	100%	100%	100%	99.8%	98.7%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	91%	91%	100%	100%	99.6%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	100%	100%	98.8%	90%	72.6%	Developmental and epileptic encephalopathy 36, 300884

ALG14	100%	100%	100%	99.8%	99.4%	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies, 619031;Myopathy, epilepsy, and progressive cerebral atrophy, 619036;?Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227
ALG3	100%	100%	100%	99.9%	98.5%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	100%	100%	100%	99.9%	98.9%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMPD2	100%	100%	100%	99.8%	98.4%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686

AMT	100%	100%	100%	100%	98.1%	Glycine encephalopathy 2, 620398
ANKRD11	100%	100%	100%	99.9%	98.4%	KBG syndrome, 148050
AP1G1	100%	100%	100%	100%	99.8%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP2M1	100%	100%	100%	100%	98.9%	Intellectual developmental disorder 60 with seizures, 618587
AP3B2	100%	100%	100%	99.9%	98.4%	Developmental and epileptic encephalopathy 48, 617276
ARHGEF9	96.9%	96.8%	98.6%	89.3%	72.2%	Developmental and epileptic encephalopathy 8, 300607
ARID1B	98.5%	98.2%	99.4%	96.3%	90.4%	Coffin-Siris syndrome 1, 135900
ARV1	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 38, 617020

ARX	98%	94.5%	91%	72.3%	51.8%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419
ASAH1	100%	100%	100%	99.9%	99.3%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASL	100%	100%	100%	99.8%	97.6%	Argininosuccinic aciduria, 207900
ASNS	100%	100%	100%	100%	99.5%	Asparagine synthetase deficiency, 615574
ASXL3	100%	100%	100%	99.9%	99%	Bainbridge-Ropers syndrome, 615485

ATN1	100%	100%	100%	99.4%	96.3%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP1A1	100%	100%	100%	100%	99.2%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100%	100%	100%	99.9%	98.4%	Developmental and epileptic encephalopathy 98, 619605;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602;Alternating hemiplegia of childhood 1, 104290;Migraine, familial basilar, 602481;Migraine, familial hemiplegic, 2, 602481

ATP1A3	100%	100%	100%	99.9%	97.6%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP6AP2	100%	100%	99.2%	90.8%	72.4%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP6V0A1	92.9%	92.8%	100%	100%	99.5%	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971;Developmental and epileptic encephalopathy 104, 619970
ATP6V0C	100%	100%	100%	99.9%	97.5%	Epilepsy, early-onset, 3, with or without developmental delay, 620465

ATP6V1A	100%	100%	100%	100%	99.7%	Cutis laxa, autosomal recessive, type IID, 617403;Developmental and epileptic encephalopathy 93, 618012
ATP7A	95%	94.7%	98.9%	90.5%	73.2%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATRX	100%	100%	99%	92.1%	75.8%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
AUTS2	100%	100%	100%	99.9%	97.9%	Intellectual developmental disorder, autosomal dominant 26, 615834
BOLA3	100%	100%	100%	99.7%	98.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299

BRAT1	100%	100%	100%	99.8%	98.3%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	94.2%	94.2%	100%	100%	99.7%	Biotinidase deficiency, 253260
CACNA1A	100%	100%	99.9%	99.6%	97.2%	Spinocerebellar ataxia 6, 183086;Episodic ataxia, type 2, 108500;Developmental and epileptic encephalopathy 42, 617106;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500;Migraine, familial hemiplegic, 1, 141500
CACNA1E	100%	100%	100%	99.9%	98.7%	Developmental and epileptic encephalopathy 69, 618285
CACNA2D1	100%	100%	100%	99.9%	99%	Developmental and epileptic encephalopathy 110, 620149

CACNA2D2	100%	100%	100%	99.7%	98.1%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CAD	100%	100%	100%	99.9%	98.2%	Developmental and epileptic encephalopathy 50, 616457
CASK	100%	100%	98.7%	90.4%	73.1%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CASQ2	100%	100%	100%	100%	99.7%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CCM2	100%	100%	100%	99.8%	97.7%	Cerebral cavernous malformations-2, 603284
CDK19	100%	100%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 87, 618916
CDKL5	95.7%	95.6%	97.9%	87.7%	70.1%	Developmental and epileptic encephalopathy 2, 300672

CELF2	100%	100%	100%	100%	99.1%	Developmental and epileptic encephalopathy 97, 619561
CEP85L	100%	100%	100%	99.9%	99.2%	Lissencephaly 10, 618873
CERT1	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with hypotonia, speech delay, and dysmorphic facies, 616351
CHD2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 94, 615369
CHD5	100%	100%	100%	99.6%	97.5%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHRNA2	100%	100%	100%	99.6%	96%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	100%	100%	100%	99.2%	93.3%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB2	100%	100%	100%	99.8%	97.7%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	100%	100%	100%	99.7%	97.3%	Intellectual developmental disorder, autosomal dominant 45, 617600

CLCN4	100%	99.9%	98.4%	86.9%	68.6%	Raynaud-Claes syndrome, 300114
CLDN16	100%	100%	100%	100%	99.2%	Hypomagnesemia 3, renal, 248250
CLDN19	100%	100%	100%	99.9%	99.2%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	93.2%	93.1%	100%	99.9%	98.5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83%	83%	100%	100%	99.2%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100%	100%	100%	99.8%	98.2%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780
CLN8	100%	100%	100%	100%	97.9%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLTC	99.2%	99.2%	100%	100%	99.5%	Intellectual developmental disorder, autosomal dominant 56, 617854
CNNM2	100%	100%	100%	99.8%	98%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418

CNTN2	100%	100%	100%	99.9%	98.3%	Epilepsy, early-onset, 5, with or without developmental delay, 615400
CNTNAP2	100%	100%	100%	100%	99.4%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA8	100%	100%	100%	100%	98.8%	Mitochondrial complex IV deficiency, nuclear type 17, 619061
COL4A1	100%	100%	100%	100%	98.9%	?Retinal arteries, tortuosity of, 180000;{Hemorrhage, intracerebral, susceptibility to}, 614519;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773;Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564;Brain small vessel disease with or without ocular anomalies, 175780
COLGALT1	100%	100%	100%	99.9%	97.6%	Brain small vessel disease 3, 618360

COQ2	96.3%	96.3%	100%	100%	99.3%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100%	100%	100%	99.8%	98.5%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ8A	100%	100%	100%	99.7%	98%	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	100%	100%	100%	100%	99.5%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417
CPLX1	100%	100%	100%	100%	97.6%	Developmental and epileptic encephalopathy 63, 617976
CPS1	100%	100%	100%	100%	99.3%	Carbamoylphosphate synthetase I deficiency, 237300

CPT2	100%	100%	100%	99.9%	98.9%	{Encephalopathy, acute, infection-induced, 4, susceptibility to}, 614212;CPT II deficiency, infantile, 600649;CPT II deficiency, lethal neonatal, 608836;CPT II deficiency, myopathic, stress-induced, 255110
CSNK2B	100%	100%	100%	100%	99.5%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSTB	100%	100%	100%	100%	98.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	100%	100%	100%	99.8%	98.3%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100%	100%	100%	100%	99.1%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CUL4B	96.7%	96.7%	98.8%	90.9%	74.6%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUX2	100%	100%	100%	99.8%	98.5%	Developmental and epileptic encephalopathy 67, 618141

CYFIP2	98.1%	98.1%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 65, 618008
D2HGDH	100%	100%	100%	99.8%	98.1%	D-2-hydroxyglutaric aciduria, 600721
DARS1	100%	100%	100%	100%	99.3%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100%	100%	100%	99.9%	99.2%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCX	100%	100%	98.5%	89%	72.1%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DDX3X	99.5%	98.7%	98.3%	88.3%	71.5%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DENND5A	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 49, 617281

DEPDC5	100%	100%	100%	100%	99.1%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504
DHDDS	73.7%	73.7%	100%	99.9%	98.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861
DIAPH1	100%	100%	100%	99.8%	98.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DLAT	100%	100%	100%	99.9%	99.3%	Pyruvate dehydrogenase E2 deficiency, 245348

DMXL2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 81, 618663;?Deafness, autosomal dominant 71, 617605;?Polyendocrine-polyneuropathy syndrome, 616113
DNAJC5	100%	100%	100%	99.7%	98.1%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350
DNM1	100%	99.9%	100%	99.6%	97.5%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346
DNM1L	100%	100%	100%	100%	99.5%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DOCK7	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 23, 615859

DPAGT1	100%	100%	100%	100%	99.5%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type lj, 608093
DPM1	99.3%	96.2%	97.9%	94.7%	90.8%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	100%	100%	100%	98.9%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	100%	100%	100%	100%	99.7%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100%	100%	100%	99.9%	98.9%	Dihydropyrimidinuria, 222748
DTYMK	100%	100%	100%	99.7%	98.3%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847

DYNC1H1	99.3%	99.3%	100%	99.9%	99%	Charcot-Marie-Tooth disease, axonal, type 2O, 614228;Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600;Cortical dysplasia, complex, with other brain malformations 13, 614563
DYRK1A	100%	100%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
EBP	100%	99.9%	97.5%	83.6%	66.6%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
EEF1A2	99.7%	97.4%	100%	99.6%	97.6%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EFHC1	97.8%	97.5%	100%	100%	99.5%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770

EGF	100%	100%	100%	100%	99.5%	?Hypomagnesemia 4, renal, 611718
EHMT1	99.9%	99.6%	100%	99.8%	98.3%	Kleefstra syndrome 1, 610253
EIF2B1	100%	100%	100%	99.9%	99.5%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100%	100%	99.9%	99.4%	96.7%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312
EIF2B3	100%	100%	100%	99.9%	99.5%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100%	100%	100%	99.9%	99.3%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314
EIF2B5	100%	100%	100%	99.9%	98.9%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EPM2A	100%	100%	100%	99.5%	96.9%	Myoclonic epilepsy of Lafora 1, 254780
ETHE1	100%	100%	100%	99.7%	98.1%	Ethylmalonic encephalopathy, 602473

EXOC7	100%	100%	100%	99.9%	98.8%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOSC3	100%	100%	100%	99.8%	99.2%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	100%	100%	100%	99.8%	98.1%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100%	100%	100%	99.9%	99.1%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FBXO28	100%	100%	100%	99.9%	99.3%	Developmental and epileptic encephalopathy 100, 619777
FCSK	100%	100%	100%	99.9%	98.6%	Congenital disorder of glycosylation with defective fucosylation 2, 618324
FGD1	100%	99.3%	97.9%	85.1%	67.4%	Aarskog-Scott syndrome, 305400
FGF12	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 47, 617166

FGF13	100%	99.8%	99.1%	90.1%	73.4%	Developmental and epileptic encephalopathy 90, 301058;Intellectual developmental disorder, X-linked 110, 301095
FLNA	100%	99.9%	97.7%	85.4%	66.8%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620
FOLR1	100%	100%	100%	100%	99%	Neurodegeneration due to cerebral folate transport deficiency, 613068

FOXG1	100%	99.9%	100%	99.2%	95.3%	Rett syndrome, congenital variant, 613454
FOXRED1	100%	100%	100%	99.8%	98.7%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	100%	99.9%	97.4%	86.2%	69%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100%	100%	99.8%	97.6%	92.9%	Developmental and epileptic encephalopathy 37, 616981
FXYD2	100%	100%	100%	99.8%	98%	Hypomagnesemia 2, renal, 154020
FZR1	100%	100%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 109, 620145
GABBR2	99.9%	99.5%	100%	99.8%	98%	{Nicotine dependence, protection against}, 188890;{Nicotine dependence, susceptibility to}, 188890;Developmental and epileptic encephalopathy 59, 617904;Neurodevelopmental disorder with poor language and loss of hand skills, 617903

GABRA1	100%	100%	100%	99.8%	98.8%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRA2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 78, 618557;{Alcohol dependence, susceptibility to}, 103780
GABRA3	100%	100%	99.1%	89.9%	71.6%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRB1	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 45, 617153
GABRB2	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 92, 617829

GABRB3	100%	100%	100%	99.8%	98.2%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113
GABRG2	92.9%	92.9%	100%	100%	99.6%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAD1	100%	100%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 89, 619124
GAMT	100%	100%	100%	99.9%	98%	Cerebral creatine deficiency syndrome 2, 612736
GCK	100%	100%	100%	99.8%	98%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853

GCSH	100%	100%	100%	100%	99.6%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GLDC	100%	100%	100%	99.9%	99.3%	Glycine encephalopathy1, 605899
GLRA1	100%	100%	100%	99.9%	99%	Hyperekplexia 1, 149400
GLRB	100%	100%	100%	100%	99.5%	Hyperekplexia 2, 614619
GLS	100%	100%	100%	100%	99.2%	CASGID syndrome, 618339;Global developmental delay, progressive ataxia, and elevated glutamine, 618412;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100%	100%	100%	99.8%	98.6%	Hyperinsulinism-hyperammonemia syndrome, 606762
GLUL	100%	100%	100%	99.8%	98.7%	Glutamine deficiency, congenital, 610015;Developmental and epileptic encephalopathy 116, 620806

GNAO1	100%	100%	100%	99.9%	98.3%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	100%	100%	100%	100%	99.3%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GOT2	100%	100%	100%	99.9%	99.1%	Developmental and epileptic encephalopathy 82, 618721
GPC3	100%	99.9%	98.6%	88.7%	70.6%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPHN	100%	100%	100%	100%	99.2%	Molybdenum cofactor deficiency C, 615501
GRIA3	100%	100%	98.3%	88.5%	70.7%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699

GRIN1	100%	100%	100%	99.7%	97.4%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820;Developmental and epileptic encephalopathy 101, 619814;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100%	100%	100%	100%	99%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99.6%	97.9%	99.8%	98.2%	92%	Developmental and epileptic encephalopathy 46, 617162

GRN	100%	100%	100%	100%	99.4%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
HACE1	100%	100%	100%	99.8%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100%	100%	100%	100%	99.3%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HCFC1	100%	99.9%	97.8%	85.9%	67.7%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541
HCN1	100%	100%	100%	99.5%	97%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482

HCN2	92.3%	87.8%	96%	90.8%	80.8%	Febrile seizures, familial, 2, 602477;{Epilepsy, idiopathic generalized, susceptibility to, 17}, 602477;Generalized epilepsy with febrile seizures plus, type 11, 602477
HECW2	100%	100%	100%	100%	99.3%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HLCS	100%	99.9%	100%	99.9%	98.2%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	100%	100%	100%	99.5%	96.6%	Developmental and epileptic encephalopathy 54, 617391
HSD17B10	100%	100%	98.5%	88.7%	70.3%	HSD10 mitochondrial disease, 300438
HSD17B4	100%	100%	100%	100%	99.5%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
IDH2	100%	100%	100%	99.9%	98.6%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	100%	100%	100%	100%	99.3%	Microcephaly, epilepsy, and diabetes syndrome, 614231

IFIH1	100%	100%	100%	100%	99.7%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IQSEC2	99.4%	96.9%	94.9%	80.3%	62.4%	Intellectual developmental disorder, X-linked 1, 309530
IRF2BPL	100%	100%	99%	95.6%	86.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	100%	100%	100%	100%	99.1%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
JAM3	100%	100%	100%	100%	99.4%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	100%	100%	100%	100%	99.7%	Koolen-De Vries syndrome, 610443
KATNB1	100%	100%	100%	99.9%	98.6%	Lissencephaly 6, with microcephaly, 616212

KCNA1	100%	100%	100%	100%	98.5%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100%	100%	100%	99.9%	98.5%	
KCNB1	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100%	100%	100%	99.9%	98.3%	Epilepsy, progressive myoclonic 7, 616187
KCND1	100%	99.9%	97.9%	85.4%	65.4%	
KCND3	100%	100%	100%	99.6%	96.6%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399
KCNH1	98.6%	98.6%	100%	99.9%	99.1%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNJ10	100%	100%	100%	100%	99.6%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780

KCNJ11	100%	100%	100%	99.8%	98.1%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	100%	100%	100%	99.9%	98.7%	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596;Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446;Cerebellar atrophy, developmental delay, and seizures, 617643;Liang-Wang syndrome, 618729
KCNQ2	100%	100%	100%	99.8%	97.8%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200

KCNQ3	100%	100%	100%	99.9%	98.9%	Seizures, benign neonatal, 2, 121201
KCNT1	100%	99.9%	100%	99.9%	97.6%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 57, 617771
KCTD7	100%	100%	100%	99.8%	98.4%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	97.9%	97.6%	98%	86.8%	68%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6B	100%	100%	100%	99.5%	96.3%	Stolerman neurodevelopmental syndrome, 618505
KIF5A	100%	100%	100%	100%	98.9%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187

KMT5B	100%	100%	100%	99.9%	98.9%	Intellectual developmental disorder, autosomal dominant 51, 617788
KPTN	100%	100%	100%	99.7%	97.2%	Intellectual developmental disorder, autosomal recessive 41, 615637
KRIT1	100%	100%	100%	99.9%	99.6%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860
LAMB1	100%	100%	100%	100%	99.4%	Lissencephaly 5, 615191
LGI1	100%	100%	100%	100%	99.9%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	100%	100%	100%	100%	99.7%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT2	100%	100%	100%	99.9%	97.1%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668

MAPK8IP3	100%	100%	100%	99.9%	98.3%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MAST3	100%	100%	100%	99.8%	97.4%	Developmental and epileptic encephalopathy 108, 620115
MBD5	100%	100%	100%	100%	99.6%	Intellectual developmental disorder, autosomal dominant 1, 156200
MBOAT7	100%	100%	100%	99.9%	98%	Intellectual developmental disorder, autosomal recessive 57, 617188
MDH2	100%	100%	100%	99.8%	98.1%	Developmental and epileptic encephalopathy 51, 617339

MECP2	100%	99.8%	96.6%	81.9%	64%	Rett syndrome, atypical, 312750;Encephalopathy, neonatal severe, 300673;Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260;{Autism susceptibility, X-linked 3}, 300496;Intellectual developmental disorder, X-linked syndromic 13, 300055;Rett syndrome, 312750;Rett syndrome, preserved speech variant, 312750
MED12	100%	99.9%	97.8%	86.4%	68.3%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED17	100%	100%	100%	100%	99.5%	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668

MEF2C	100%	100%	100%	99.9%	98.6%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443
MFF	95.9%	95.9%	100%	99.9%	99.3%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100%	100%	100%	100%	99.7%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	100%	100%	100%	100%	98.5%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MOCS1	100%	100%	100%	99.8%	98.2%	Molybdenum cofactor deficiency A, 252150
MOCS2	100%	100%	100%	100%	99.5%	Molybdenum cofactor deficiency B1, 252160
MPDU1	100%	100%	100%	100%	97.8%	Congenital disorder of glycosylation, type If, 609180

MPDZ	100%	100%	100%	99.9%	99.3%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTFMT	100%	100%	100%	99.4%	97.8%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248
MTHFR	100%	100%	100%	99.9%	99.2%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MTOR	100%	100%	100%	99.9%	99%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MTRR	100%	100%	100%	100%	99.4%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634

NACC1	100%	100%	100%	100%	98.7%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	100%	100%	100%	100%	99.1%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NARS2	92.3%	92.3%	100%	99.9%	99.7%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434
NAXE	97.9%	93%	100%	100%	98.8%	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186
NBEA	97.6%	97.6%	100%	99.8%	99%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NCDN	100%	100%	100%	99.9%	98.8%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NDUFA1	100%	100%	98.6%	87.7%	69.4%	Mitochondrial complex I deficiency, nuclear type 12, 301020

NDUFA11	100%	99.3%	100%	99.8%	98.4%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100%	100%	100%	99.9%	99.4%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100%	99.9%	98.9%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	100%	100%	99.9%	98.4%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	100%	100%	100%	99.6%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	100%	100%	100%	100%	99.9%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	100%	100%	100%	99.9%	99.2%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100%	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 5, 618226

NDUFS2	99.9%	98%	100%	100%	98.9%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	98.5%	93.4%	100%	99.9%	98.7%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	100%	100%	100%	99.1%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100%	100%	100%	100%	99.5%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	100%	100%	100%	99.7%	97.2%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100%	100%	100%	100%	99.7%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	100%	100%	100%	99.9%	99.3%	Developmental and epileptic encephalopathy 21, 615833
NEDD4L	100%	100%	100%	99.9%	98.8%	Periventricular nodular heterotopia 7, 617201
NEU1	100%	100%	100%	99.9%	99.3%	Sialidosis, type II, 256550;Sialidosis, type I, 256550

NEUROD2	100%	100%	100%	98.2%	90.8%	Developmental and epileptic encephalopathy 72, 618374
NEXMIF	100%	100%	98.9%	89.5%	72.8%	Intellectual developmental disorder, X-linked 98, 300912
NGLY1	100%	100%	100%	100%	99.5%	Congenital disorder of deglycosylation 1, 615273
NHLRC1	100%	100%	100%	100%	99%	Myoclonic epilepsy of Lafora 2, 620681
NPRL2	100%	100%	100%	100%	98.9%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100%	100%	100%	99.8%	98.4%	Epilepsy, familial focal, with variable foci 3, 617118
NR2F1	100%	99.8%	100%	98.4%	92.5%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	100%	100%	100%	99.9%	99%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911

NRROS	100%	100%	100%	99.9%	98.9%	Seizures, early-onset, with neurodegeneration and brain calcification, 618875
NRXN1	100%	100%	100%	99.8%	98.5%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332
NUBPL	100%	100%	100%	100%	99.3%	Mitochondrial complex I deficiency, nuclear type 21, 618242
NUS1	100%	100%	100%	100%	98.5%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
OCLN	94.5%	94.5%	100%	99.8%	98.2%	Pseudo-TORCH syndrome 1, 251290
OFD1	100%	100%	98.9%	90.6%	73.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDHL	100%	100%	100%	99.9%	99.1%	

OPHN1	94%	93.9%	99%	90.1%	72.6%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
PACS1	100%	100%	100%	99.8%	98.2%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100%	100%	100%	99.8%	97.9%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100%	100%	100%	99.9%	99.4%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432
PAK3	100%	100%	98.8%	90.4%	73%	Intellectual developmental disorder, X-linked 30, 300558
PARS2	100%	100%	100%	100%	98.9%	Developmental and epileptic encephalopathy 75, 618437
PC	100%	100%	100%	100%	98.6%	Pyruvate carboxylase deficiency, 266150
PCDH19	100%	99.9%	97.1%	84.2%	65.9%	Developmental and epileptic encephalopathy 9, 300088
PDCD10	100%	100%	100%	100%	99.3%	Cerebral cavernous malformations-3, 603285

PDHA1	99.3%	94.3%	98%	87.6%	71.1%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100%	100%	100%	100%	99%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100%	100%	100%	100%	99%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100%	100%	100%	100%	99.7%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	100%	100%	100%	100%	98.8%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PET100	100%	100%	100%	100%	98.6%	Mitochondrial complex IV deficiency, nuclear type 12, 619055
PEX1	100%	100%	100%	100%	99.6%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100

PEX10	100%	100%	100%	100%	99.2%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX12	100%	100%	100%	99.9%	98.9%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100%	100%	100%	100%	99.7%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100%	100%	100%	100%	98.5%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100%	100%	100%	100%	98.6%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100%	100%	100%	100%	99.3%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	100%	100%	100%	99.6%	97.1%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872

PEX3	100%	100%	100%	100%	99.6%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100%	100%	99.9%	99.6%	98.1%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100%	100%	100%	99.9%	98.1%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PGAP3	100%	100%	100%	99.8%	98%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PHACTR1	100%	100%	99.9%	99.5%	96.7%	Developmental and epileptic encephalopathy 70, 618298

PHF21A	100%	100%	100%	99.9%	98.6%	Intellectual developmental disorder with behavioral abnormalities and craniofacial dysmorphism with or without seizures, 618725
PHF6	100%	100%	99%	91.8%	74.8%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	100%	100%	100%	100%	99.2%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PI4K2A	100%	100%	100%	99.8%	97.2%	Neurodevelopmental disorder with hyperkinetic movements, seizures and structural brain abnormalities, 620732
PIGA	100%	100%	99%	88.5%	70.7%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072

PIGB	100%	100%	100%	100%	99.3%	Developmental and epileptic encephalopathy 80, 618580
PIGG	100%	100%	100%	99.9%	98.9%	[Blood group, EMM system], 619812;Neurodevelopmental disorder with or without hypotonia, seizures, and cerebellar atrophy, 616917
PIGK	100%	100%	100%	100%	99.6%	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures, 618879
PIGN	100%	100%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	99.9%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 55, 617599
PIGQ	100%	100%	100%	99.9%	98.9%	Multiple congenital anomalies-hypotonia-seizures syndrome 4, 618548

PIGS	98.8%	95.2%	100%	99.9%	98.3%	Developmental and epileptic encephalopathy 95, 618143
PIGT	100%	100%	100%	99.8%	98.5%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398
PIGU	100%	100%	100%	100%	99.2%	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis, 618590
PLA2G6	100%	99.9%	100%	99.9%	98.8%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLCB1	100%	100%	100%	100%	99.4%	Developmental and epileptic encephalopathy 12, 613722
PLP1	100%	99.7%	99.4%	89%	70.3%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920

PLPBP	100%	100%	100%	99.9%	98.7%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PMM2	100%	100%	100%	100%	99.3%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100%	100%	100%	99.9%	97.8%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNPO	100%	100%	100%	99.8%	98.6%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090

POLG	100%	100%	100%	99.9%	99%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPFIBP1	100%	100%	100%	100%	99.7%	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024
PPP2R1A	93.9%	93.9%	100%	100%	99%	Houge-Janssens syndrome 2, 616362
PPP2R5D	100%	100%	100%	99.9%	98.7%	Houge-Janssens syndrome 1, 616355

PPP3CA	100%	100%	100%	100%	99.1%	Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development, 618265;Developmental and epileptic encephalopathy 91, 617711
PPT1	90.3%	90.3%	100%	99.9%	99.1%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	98.3%	87.8%	65.5%	Renpenning syndrome, 309500
PRF1	100%	100%	100%	100%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRICKLE1	100%	100%	100%	99.9%	99.1%	Epilepsy, progressive myoclonic 1B, 612437
PRODH	100%	100%	100%	99.9%	98.8%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500

PRRT2	100%	100%	100%	99.8%	97%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200
PSAP	100%	100%	100%	99.9%	98.4%	Combined SAP deficiency, 611721;Krabbe disease, atypical, 611722;Metachromatic leukodystrophy due to SAP-b deficiency, 249900;Gaucher disease, atypical, 610539;{Parkinson disease 24, autosomal dominant, susceptibility to}, 619491
PTRH2	100%	100%	100%	100%	99.6%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	100%	100%	100%	99.9%	99.1%	Hyperphenylalaninemia, BH4-deficient, A, 261640

PUM1	100%	100%	100%	99.6%	98.4%	Neurodevelopmental disorder with motor abnormalities, and facial dysmorphism, 620719
PURA	100%	100%	100%	99.9%	98.3%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PYCR2	100%	100%	100%	99.9%	99%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100%	100%	100%	99.9%	98.7%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100%	100%	100%	99.9%	98.5%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB39B	100%	100%	98.5%	90.9%	75.3%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RARS2	94.1%	93.7%	100%	99.9%	99.7%	Pontocerebellar hypoplasia, type 6, 611523

RHOBTB2	98.7%	98.7%	100%	99.9%	98.6%	Developmental and epileptic encephalopathy 64, 618004
RNASEH2A	100%	100%	100%	99.9%	98.8%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100%	99.9%	99.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	100%	100%	99.4%	96.7%	Aicardi-Goutieres syndrome 3, 610329
RNF13	100%	100%	100%	100%	99.7%	Developmental and epileptic encephalopathy 73, 618379
RNU4-2						ReNU syndrome, 620851
ROGDI	100%	100%	100%	99.9%	98.5%	Kohlschutter-Tonz syndrome, 226750
RORA	100%	100%	100%	99.4%	97.5%	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060
RPS6KA3	100%	100%	98.8%	91.2%	73%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600

RRM2B	100%	100%	100%	100%	99.5%	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075;Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075;Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction, 268315;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077
SAMHD1	100%	100%	100%	99.9%	99.2%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SCAF4	100%	100%	100%	99.9%	98.9%	Fliedner-Zweier syndrome, 620511
SCARB2	100%	100%	100%	99.9%	99.5%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900

SCN1A	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317;Migraine, familial hemiplegic, 3, 609634;Dravet syndrome, 607208;Febrile seizures, familial, 3A, 604403;Generalized epilepsy with febrile seizures plus, type 2, 604403
SCN1B	100%	99.4%	100%	99.6%	98%	Generalized epilepsy with febrile seizures plus, type 1, 604233;Developmental and epileptic encephalopathy 52, 617350;Cardiac conduction defect, nonspecific, 612838;Atrial fibrillation, familial, 13, 615377;Brugada syndrome 5, 612838
SCN2A	100%	100%	100%	100%	99.8%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924

SCN3A	100%	100%	100%	100%	99.5%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938
SCN8A	100%	100%	100%	99.9%	98.8%	?Myoclonus, familial, 2, 618364;Seizures, benign familial infantile, 5, 617080;Cognitive impairment with or without cerebellar ataxia, 614306;Developmental and epileptic encephalopathy 13, 614558
SEMA6B	100%	100%	100%	99.8%	97.1%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	99.8%	97.1%	100%	100%	99.3%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	100%	100%	100%	100%	99.8%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218

SETD1A	100%	100%	100%	99.8%	97.5%	Epilepsy, early-onset, 2, with or without developmental delay, 618832;Neurodevelopmental disorder with speech impairment and dysmorphic facies, 619056
SETD1B	100%	100%	99.9%	99.1%	94.9%	Intellectual developmental disorder with seizures and language delay, 619000
SHANK3	99.3%	97%	99.8%	99.2%	95.5%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SIK1	100%	100%	100%	100%	98.3%	Developmental and epileptic encephalopathy 30, 616341
SLC12A5	100%	100%	100%	99.9%	98%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC13A5	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905

SLC16A1	100%	100%	100%	99.9%	99%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	99.9%	98.8%	100%	100%	99.4%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A2	100%	100%	100%	100%	99.5%	Developmental and epileptic encephalopathy 41, 617105
SLC25A1	100%	100%	100%	99.7%	95.7%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A15	100%	100%	100%	100%	99.3%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100%	100%	100%	99.7%	97.6%	Developmental and epileptic encephalopathy 3, 609304

SLC2A1	100%	100%	100%	100%	98.8%	Dystonia 9, 601042;GLUT1 deficiency syndrome 1, infantile onset, severe, 606777;Stomatin-deficient cryohydrocytosis with neurologic defects, 608885;{Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847;GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC32A1	100%	100%	100%	99.9%	98.9%	Generalized epilepsy with febrile seizures plus, type 12, 620755;Developmental and epileptic encephalopathy 114, 620774
SLC35A2	100%	99.9%	98.2%	87.9%	70.6%	Congenital disorder of glycosylation, type IIm, 300896
SLC38A3	100%	100%	100%	100%	98.2%	Developmental and epileptic encephalopathy 102, 619881
SLC6A1	100%	100%	100%	100%	98.6%	Myoclonic-atonic epilepsy, 616421

SLC6A8	100%	99.6%	97.5%	84.9%	65.9%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	100%	100%	99.1%	89.3%	71.5%	Neurodegenerative disorder, X-linked, female-restricted, with parkinsonism and cognitive impairement, 301142;Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SMARCA2	98%	97.2%	100%	99.9%	99%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMC1A	100%	99.9%	98.4%	87%	68.7%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMPD4	100%	100%	100%	99.9%	98.8%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622

SMS	99.8%	98.8%	98.7%	88.9%	72.7%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAP25	100%	100%	100%	100%	99.2%	Developmental and epileptic encephalopathy 117, 616330
SPTAN1	99.4%	98.8%	100%	99.9%	99%	Developmental delay with or without epilepsy, 620540;Developmental and epileptic encephalopathy 5, 613477;Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538;Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528
ST3GAL3	95.6%	95%	100%	99.9%	98.5%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.3%	98.3%	100%	99.9%	99.1%	Salt and pepper developmental regression syndrome, 609056

STRADA	100%	100%	100%	99.9%	99%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STX1B	100%	100%	100%	99.2%	94.4%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 4, 612164
SUOX	100%	100%	100%	99.8%	98.7%	Sulfite oxidase deficiency, 272300
SYN1	100%	100%	96.5%	82.9%	62.9%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNGAP1	100%	100%	100%	99.7%	97.3%	Intellectual developmental disorder, autosomal dominant 5, 612621
SYNJ1	100%	100%	100%	100%	99.5%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389

SYP	100%	100%	98.3%	84.9%	65.4%	Intellectual developmental disorder, X-linked 96, 300802
SZT2	100%	100%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 18, 615476
TANGO2	100%	100%	100%	99.9%	98%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 11, 617695

TBC1D24	100%	100%	100%	99.9%	98.9%	Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBCD	91.2%	90.6%	100%	99.8%	98.1%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100%	100%	100%	100%	99.4%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207

TCF4	100%	100%	100%	100%	99.3%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TDP2	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TIMM50	100%	100%	100%	99.9%	98.7%	3-methylglutaconic aciduria, type IX, 617698
TMTC3	100%	100%	100%	100%	99.6%	Lissencephaly 8, 617255
TOE1	100%	100%	100%	99.9%	98.7%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	100%	100%	100%	99.8%	98.6%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270
TRAK1	100%	100%	100%	99.9%	98.9%	Developmental and epileptic encephalopathy 68, 618201

TREX1	100%	100%	100%	100%	99.8%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TRIT1	100%	100%	100%	100%	99.3%	Combined oxidative phosphorylation deficiency 35, 617873
TRPM3	97.8%	97.8%	100%	100%	99.3%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TRPM6	100%	100%	100%	100%	99.4%	Hypomagnesemia 1, intestinal, 602014
TSC1	100%	100%	100%	100%	99.2%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690

TSC2	100%	100%	100%	99.8%	98.7%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSEN15	100%	100%	100%	100%	99.1%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100%	99.9%	99.4%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100%	100%	100%	99.7%	97.5%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	100%	100%	100%	99.8%	97.9%	Lissencephaly 3, 611603
TUBB2A	100%	100%	100%	99.9%	97.8%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	100%	100%	99.9%	97.6%	Cortical dysplasia, complex, with other brain malformations 7, 610031

TUBB4A	99.7%	96.9%	100%	99.9%	98.2%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100%	100%	100%	100%	98.6%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	99.9%	98.3%	100%	99.9%	99.4%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBE3A	100%	100%	100%	100%	99.6%	Angelman syndrome, 105830
UBTF	100%	100%	100%	99.9%	98.2%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UGDH	100%	100%	100%	100%	99.6%	Developmental and epileptic encephalopathy 84, 618792
UGP2	98.9%	96.9%	100%	99.9%	99.4%	Developmental and epileptic encephalopathy 83, 618744

VPS11	100%	100%	100%	99.9%	98.2%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS53	83.3%	81.9%	100%	100%	99.1%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	93.8%	93.8%	100%	99.8%	98.6%	Skraban-Deardorff syndrome, 617616
WDR45	100%	100%	97.9%	86.1%	66.9%	Neurodegeneration with brain iron accumulation 5, 300894
WFS1	91.2%	91.2%	100%	99.8%	98.2%	Deafness, autosomal dominant 6/14/38, 600965;?Cataract 41, 116400;Wolfram-like syndrome, autosomal dominant, 614296;{Diabetes mellitus, noninsulin-dependent, association with}, 125853;Wolfram syndrome 1, 222300
WWOX	100%	100%	100%	99.9%	99.1%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322

XK	100%	99.7%	98.7%	89.3%	71.5%	McLeod syndrome, 300842
YWHAG	100%	100%	100%	100%	98.3%	Developmental and epileptic encephalopathy 56, 617665
ZEB2	100%	100%	100%	99.6%	97.9%	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.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors