EPILEPSY PANEL DG-4.0.0 (395 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS1	100.0%	100.0%	100.0%	99.2%	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	100.0%	100.0%	100.0%	98.7%	GABA-transaminase deficiency, 613163
ABCC8	100.0%	100.0%	100.0%	99.4%	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857;Diabetes mellitus, transient neonatal 2, 610374;Diabetes mellitus, noninsulin-dependent, 125853;Hypoglycemia of infancy, leucine-sensitive, 240800;Hyperinsulinemic hypoglycemia, familial, 1, 256450

ACO2	92.4%	89.8%	100.0%	99.3%	Optic atrophy 9, 616289;Infantile cerebellar-retinal degeneration, 614559
ACTB	100.0%	100.0%	100.0%	99.0%	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, 620479
ACTL6B	100.0%	100.0%	100.0%	98.9%	Developmental and epileptic encephalopathy 76, 618468;Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100.0%	100.0%	100.0%	99.4%	Aminoacylase 1 deficiency, 609924
ADSL	100.0%	100.0%	100.0%	99.0%	Adenylosuccinase deficiency, 103050
AFG2A	100.0%	100.0%	100.0%	98.9%	Neurodevelopmental disorder with hearing loss, seizures, and brain abnormalities, 616577
AGA	100.0%	100.0%	100.0%	98.3%	Aspartylglucosaminuria, 208400

ALDH4A1	100.0%	100.0%	100.0%	98.6%	Hyperprolinemia, type II, 239510
ALDH5A1	100.0%	100.0%	100.0%	97.8%	Succinic semialdehyde dehydrogenase deficiency, 271980
ALDH7A1	100.0%	100.0%	100.0%	99.0%	Epilepsy, early-onset, 4, vitamin B6-dependent, 266100
ALG1	100.0%	100.0%	100.0%	99.4%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	91.0%	91.0%	100.0%	98.4%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	99.7%	99.0%	97.0%	70.4%	Developmental and epileptic encephalopathy 36, 300884
ALG3	100.0%	100.0%	100.0%	97.8%	Congenital disorder of glycosylation, type Id, 601110
ALG6	100.0%	100.0%	99.9%	96.4%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	100.0%	100.0%	100.0%	97.1%	Alpha-methylacyl-CoA racemase deficiency, 614307;Bile acid synthesis defect, congenital, 4, 214950
AMPD2	100.0%	100.0%	99.9%	98.7%	Pontocerebellar hypoplasia, type 9, 615809;?Spastic paraplegia 63, autosomal recessive, 615686

AMT	100.0%	100.0%	100.0%	99.5%	Glycine encephalopathy 2, 620398
ANKRD11	100.0%	100.0%	100.0%	98.0%	KBG syndrome, 148050
AP1G1	100.0%	100.0%	100.0%	97.9%	Usmani-Riazuddin syndrome, autosomal recessive, 619548;Usmani-Riazuddin syndrome, autosomal dominant, 619467
AP3B2	100.0%	100.0%	100.0%	98.8%	Developmental and epileptic encephalopathy 48, 617276
ARHGEF9	96.7%	95.8%	98.4%	72.4%	Developmental and epileptic encephalopathy 8, 300607
ARID1B	98.6%	98.4%	98.0%	86.3%	Coffin-Siris syndrome 1, 135900
ARX	99.0%	96.7%	89.5%	50.5%	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	100.0%	100.0%	100.0%	97.8%	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950;Farber lipogranulomatosis, 228000
ASL	100.0%	100.0%	100.0%	99.3%	Argininosuccinic aciduria, 207900

ASNS	100.0%	100.0%	100.0%	98.2%	Asparagine synthetase deficiency, 615574
ASXL3	100.0%	100.0%	100.0%	97.4%	Bainbridge-Ropers syndrome, 615485
ATN1	100.0%	100.0%	99.9%	96.6%	Dentatorubral-pallidoluysian atrophy, 125370;Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494
ATP1A1	100.0%	100.0%	100.0%	99.1%	Hypomagnesemia, seizures, and impaired intellectual development 2, 618314;Charcot-Marie-Tooth disease, axonal, type 2DD, 618036
ATP1A2	100.0%	100.0%	100.0%	99.3%	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	100.0%	100.0%	100.0%	98.9%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
ATP6AP2	100.0%	100.0%	97.4%	70.6%	Intellectual developmental disorder, X-linked syndromic, Hedera type, 300423;?Parkinsonism with spasticity, X-linked, 300911;Congenital disorder of glycosylation, type IIr, 301045
ATP7A	94.9%	94.5%	98.1%	71.7%	Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATRX	99.9%	99.7%	96.4%	65.4%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
AUTS2	100.0%	100.0%	100.0%	98.4%	Intellectual developmental disorder, autosomal dominant 26, 615834

BOLA3	100.0%	100.0%	100.0%	97.5%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	100.0%	100.0%	100.0%	99.7%	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056;Rigidity and multifocal seizure syndrome, lethal neonatal, 614498
BTD	94.2%	94.2%	100.0%	99.5%	Biotinidase deficiency, 253260
CACNA1A	100.0%	100.0%	100.0%	97.5%	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	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 69, 618285
CACNA2D2	100.0%	100.0%	100.0%	97.8%	Cerebellar atrophy with seizures and variable developmental delay, 618501

CACNB4	100.0%	100.0%	100.0%	98.9%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682;?Episodic ataxia, type 5, 613855;{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682
CAD	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 50, 616457
CASK	100.0%	100.0%	97.4%	71.5%	Intellectual developmental disorder, with or without nystagmus, 300422;Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749;FG syndrome 4, 300422
CASQ2	100.0%	100.0%	100.0%	98.6%	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938
CCM2	100.0%	100.0%	99.9%	98.4%	Cerebral cavernous malformations-2, 603284
CDKL5	95.7%	95.3%	97.4%	68.8%	Developmental and epileptic encephalopathy 2, 300672
CERT1	100.0%	100.0%	100.0%	98.2%	Intellectual developmental disorder, autosomal dominant 34, 616351
CHD2	100.0%	100.0%	100.0%	98.6%	Developmental and epileptic encephalopathy 94, 615369

CHD5	100.0%	100.0%	100.0%	98.1%	Parenti-Mignot neurodevelopmental syndrome, 619873
CHRNA2	100.0%	100.0%	100.0%	99.1%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	100.0%	100.0%	100.0%	96.9%	{Nicotine addiction, susceptibility to}, 188890;Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB2	100.0%	100.0%	100.0%	99.2%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	100.0%	100.0%	100.0%	99.1%	Intellectual developmental disorder, autosomal dominant 45, 617600
CLCN4	100.0%	100.0%	98.1%	70.4%	Raynaud-Claes syndrome, 300114
CLDN16	100.0%	100.0%	100.0%	98.8%	Hypomagnesemia 3, renal, 248250
CLDN19	100.0%	100.0%	100.0%	99.9%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	93.2%	93.1%	100.0%	98.5%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	83.1%	83.0%	100.0%	96.8%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	100.0%	100.0%	100.0%	97.6%	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300;Ceroid lipofuscinosis, neuronal, 6A, 601780

CLN8	100.0%	100.0%	100.0%	99.6%	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003;Ceroid lipofuscinosis, neuronal, 8, 600143
CLTC	99.2%	99.2%	100.0%	98.8%	Intellectual developmental disorder, autosomal dominant 56, 617854
CNNM2	100.0%	100.0%	100.0%	97.4%	Hypomagnesemia 6, renal, 613882;Hypomagnesemia, seizures, and impaired intellectual development 1, 616418
CNTN2	100.0%	100.0%	99.9%	99.4%	Epilepsy, early-onset, 5, with or without developmental delay, 615400
CNTNAP2	100.0%	100.0%	100.0%	99.0%	Pitt-Hopkins like syndrome 1, 610042;{Autism susceptibility 15}, 612100
COA8	100.0%	99.9%	100.0%	97.0%	Mitochondrial complex IV deficiency, nuclear type 17, 619061

COL4A1	100.0%	100.0%	100.0%	98.4%	?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	100.0%	100.0%	99.9%	95.2%	Brain small vessel disease 3, 618360
COQ2	96.3%	96.3%	100.0%	98.5%	{Multiple system atrophy, susceptibility to}, 146500;Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	100.0%	100.0%	100.0%	99.6%	Coenzyme Q10 deficiency, primary, 7, 616276;Spastic ataxia 10, autosomal recessive, 620666
COQ8A	100.0%	100.0%	100.0%	99.7%	Coenzyme Q10 deficiency, primary, 4, 612016
CPA6	100.0%	100.0%	100.0%	99.4%	Febrile seizures, familial, 11, 614418;Epilepsy, familial temporal lobe, 5, 614417

CPS1	100.0%	100.0%	100.0%	98.5%	Carbamoylphosphate synthetase I deficiency, 237300;{Pulmonary hypertension, neonatal, susceptibility to}, 615371
CPT2	100.0%	100.0%	100.0%	98.7%	{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	100.0%	100.0%	100.0%	99.2%	Poirier-Bienvenu neurodevelopmental syndrome, 618732
CSTB	100.0%	100.0%	100.0%	95.3%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	100.0%	100.0%	100.0%	99.5%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	100.0%	100.0%	100.0%	98.6%	Ceroid lipofuscinosis, neuronal, 13 (Kufs type), 615362
CUL4B	96.7%	96.6%	97.1%	66.8%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUX2	100.0%	100.0%	99.9%	98.2%	Developmental and epileptic encephalopathy 67, 618141

D2HGDH	100.0%	100.0%	100.0%	99.2%	D-2-hydroxyglutaric aciduria, 600721
DARS1	100.0%	100.0%	100.0%	97.8%	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281
DARS2	100.0%	100.0%	100.0%	96.8%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DCX	98.8%	98.7%	98.2%	71.0%	Subcortical laminal heterotopia, X-linked, 300067;Lissencephaly, X-linked, 300067
DDX3X	99.2%	98.4%	98.0%	70.1%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DENND5A	100.0%	100.0%	100.0%	97.8%	Developmental and epileptic encephalopathy 49, 617281
DEPDC5	100.0%	100.0%	100.0%	99.0%	Epilepsy, familial focal, with variable foci 1, 604364;Developmental and epileptic encephalopathy 111, 620504
DHDDS	73.8%	73.7%	100.0%	98.8%	Developmental delay and seizures with or without movement abnormalities, 617836;?Congenital disorder of glycosylation, type 1bb, 613861;Retinitis pigmentosa 59, 613861

DIAPH1	100.0%	100.0%	99.9%	95.3%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DLAT	100.0%	100.0%	100.0%	98.9%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	100.0%	100.0%	100.0%	99.8%	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350
DNM1	100.0%	100.0%	100.0%	98.1%	Developmental and epileptic encephalopathy 31B, autosomal recessive, 620352;Developmental and epileptic encephalopathy 31A, autosomal dominant, 616346
DNM1L	100.0%	100.0%	100.0%	98.6%	Optic atrophy 5, 610708;Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388
DOCK7	100.0%	100.0%	100.0%	98.1%	Developmental and epileptic encephalopathy 23, 615859
DPAGT1	100.0%	100.0%	100.0%	99.3%	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750;Congenital disorder of glycosylation, type Ij, 608093

DPM1	99.2%	96.7%	100.0%	98.2%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100.0%	100.0%	100.0%	99.3%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	99.8%	99.6%	100.0%	98.6%	Dihydropyrimidine dehydrogenase deficiency, 274270;5-fluorouracil toxicity, 274270
DPYS	100.0%	100.0%	100.0%	98.3%	Dihydropyrimidinuria, 222748
DTYMK	100.0%	100.0%	100.0%	99.1%	Neurodegeneration, childhood-onset, with progressive microcephaly, 619847
DYNC1H1	99.3%	99.3%	100.0%	98.9%	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	100.0%	100.0%	100.0%	98.7%	Intellectual developmental disorder, autosomal dominant 7, 614104
EBP	100.0%	100.0%	98.7%	72.8%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960

EEF1A2	99.6%	98.5%	100.0%	97.4%	Developmental and epileptic encephalopathy 33, 616409;Intellectual developmental disorder, autosomal dominant 38, 616393
EFHC1	97.8%	97.5%	100.0%	98.7%	{Epilepsy, juvenile absence, susceptibility to, 1}, 607631;{Myoclonic epilepsy, juvenile, susceptibility to, 1}, 254770
EGF	100.0%	100.0%	100.0%	98.8%	?Hypomagnesemia 4, renal, 611718
EHMT1	100.0%	99.9%	99.9%	98.9%	Kleefstra syndrome 1, 610253
EIF2B1	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896
EIF2B2	100.0%	100.0%	100.0%	98.1%	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312
EIF2B3	100.0%	100.0%	100.0%	97.5%	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313
EIF2B4	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314

EIF2B5	100.0%	100.0%	100.0%	98.9%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EPM2A	100.0%	100.0%	99.5%	90.8%	Myoclonic epilepsy of Lafora 1, 254780
ETHE1	100.0%	100.0%	100.0%	97.9%	Ethylmalonic encephalopathy, 602473
EXOC7	100.0%	100.0%	100.0%	98.6%	Neurodevelopmental disorder with seizures and brain atrophy, 619072
EXOSC3	100.0%	100.0%	100.0%	98.8%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	100.0%	100.0%	100.0%	98.8%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100.0%	100.0%	100.0%	99.1%	Combined oxidative phosphorylation deficiency 14, 614946;Spastic paraplegia 77, autosomal recessive, 617046
FGD1	99.9%	99.5%	97.6%	69.5%	Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400
FGF12	100.0%	100.0%	100.0%	99.7%	Developmental and epileptic encephalopathy 47, 617166

FLNA	100.0%	99.9%	99.0%	78.6%	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	100.0%	100.0%	100.0%	99.8%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	100.0%	99.9%	100.0%	94.3%	Rett syndrome, congenital variant, 613454
FOXRED1	100.0%	100.0%	100.0%	98.6%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	100.0%	99.7%	97.5%	68.2%	Intellectual developmental disorder, X-linked 104, 300983
FRRS1L	100.0%	100.0%	99.9%	90.5%	Developmental and epileptic encephalopathy 37, 616981
FXYD2	100.0%	100.0%	100.0%	99.5%	Hypomagnesemia 2, renal, 154020

GABRA1	100.0%	100.0%	100.0%	99.1%	{Epilepsy, juvenile myoclonic, susceptibility to, 5}, 611136;Developmental and epileptic encephalopathy 19, 615744;{Epilepsy, childhood absence, susceptibility to, 4}, 611136
GABRA3	100.0%	99.9%	98.1%	72.1%	Epilepsy, X-linked 2, with or without impaired intellectual development and dysmorphic features, 301091
GABRB3	100.0%	100.0%	99.9%	97.2%	{Epilepsy, childhood absence, susceptibility to, 5}, 612269;Developmental and epileptic encephalopathy 43, 617113
GABRG2	92.9%	92.6%	100.0%	99.3%	Developmental and epileptic encephalopathy 74, 618396;Febrile seizures, familial, 8, 607681;Generalized epilepsy with febrile seizures plus, type 3, 607681
GAMT	100.0%	100.0%	100.0%	97.5%	Cerebral creatine deficiency syndrome 2, 612736

GCK	100.0%	100.0%	100.0%	99.6%	MODY, type II, 125851;Diabetes mellitus, permanent neonatal 1, 606176;Hyperinsulinemic hypoglycemia, familial, 3, 602485;Diabetes mellitus, noninsulin-dependent, late onset, 125853
GCSH	100.0%	100.0%	100.0%	98.1%	Multiple mitochondrial dysfunctions syndrome 7, 620423
GLDC	100.0%	100.0%	100.0%	98.6%	Glycine encephalopathy1, 605899
GLRA1	100.0%	100.0%	100.0%	99.3%	Hyperekplexia 1, 149400
GLRB	100.0%	100.0%	100.0%	98.3%	Hyperekplexia 2, 614619
GLS	100.0%	100.0%	100.0%	97.9%	Global developmental delay, progressive ataxia, and elevated glutamine, 618412;?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339;Developmental and epileptic encephalopathy 71, 618328
GLUD1	100.0%	100.0%	100.0%	94.8%	Hyperinsulinism-hyperammonemia syndrome, 606762

GNAO1	100.0%	100.0%	100.0%	98.8%	Developmental and epileptic encephalopathy 17, 615473;Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	100.0%	100.0%	100.0%	99.7%	Epilepsy, progressive myoclonic 6, 614018;Muscular dystrophy, congenital, with or without seizures, 620166
GPC3	99.6%	98.9%	97.7%	68.3%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GPHN	100.0%	99.9%	100.0%	98.1%	Molybdenum cofactor deficiency C, 615501
GRIA3	99.7%	99.2%	97.4%	68.8%	Intellectual developmental disorder, X-linked syndromic, Wu type, 300699
GRIN1	100.0%	100.0%	100.0%	97.9%	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	99.8%	99.3%	100.0%	98.9%	Epilepsy, focal, with speech disorder and with or without impaired intellectual development, 245570
GRIN2B	99.9%	99.8%	100.0%	99.4%	Developmental and epileptic encephalopathy 27, 616139;Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970
GRIN2D	99.7%	98.7%	99.7%	88.2%	Developmental and epileptic encephalopathy 46, 617162
GRN	100.0%	100.0%	100.0%	99.7%	Aphasia, primary progressive, 607485;Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
HACE1	100.0%	100.0%	100.0%	97.4%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	100.0%	100.0%	100.0%	98.1%	Hyperinsulinemic hypoglycemia, familial, 4, 609975;3-hydroxyacyl-CoA dehydrogenase deficiency, 231530
HCFC1	100.0%	99.9%	98.4%	75.7%	Methylmalonic aciduria and homocysteinemia, cblX type, 309541

HCN1	99.9%	99.7%	99.8%	96.0%	Developmental and epileptic encephalopathy 24, 615871;Generalized epilepsy with febrile seizures plus, type 10, 618482
HECW2	100.0%	100.0%	100.0%	98.8%	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268
HLCS	100.0%	100.0%	99.9%	97.6%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	100.0%	100.0%	100.0%	97.5%	Developmental and epileptic encephalopathy 54, 617391
HSD17B10	100.0%	99.8%	98.0%	70.3%	HSD10 mitochondrial disease, 300438
HSD17B4	100.0%	100.0%	100.0%	98.2%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
IDH2	100.0%	100.0%	100.0%	98.1%	D-2-hydroxyglutaric aciduria 2, 613657
IER3IP1	100.0%	100.0%	100.0%	98.9%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	100.0%	100.0%	100.0%	98.2%	Immunodeficiency 95, 619773;Aicardi-Goutieres syndrome 7, 615846;Singleton-Merten syndrome 1, 182250
IQSEC2	98.6%	96.4%	93.5%	59.6%	Intellectual developmental disorder, X-linked 1, 309530

IRF2BPL	100.0%	100.0%	99.2%	91.8%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	100.0%	100.0%	100.0%	97.8%	[Inosine triphosphatase deficiency], 613850;Developmental and epileptic encephalopathy 35, 616647
JAM3	100.0%	100.0%	100.0%	98.5%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	100.0%	100.0%	100.0%	99.4%	Koolen-De Vries syndrome, 610443
KATNB1	100.0%	100.0%	100.0%	99.7%	Lissencephaly 6, with microcephaly, 616212
KCNA1	100.0%	100.0%	100.0%	99.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 32, 616366
KCNA3	100.0%	100.0%	100.0%	95.4%	
KCNB1	100.0%	100.0%	100.0%	99.2%	Developmental and epileptic encephalopathy 26, 616056
KCNC1	100.0%	100.0%	100.0%	99.4%	Epilepsy, progressive myoclonic 7, 616187
KCND3	100.0%	100.0%	100.0%	99.3%	Spinocerebellar ataxia 19, 607346;Brugada syndrome 9, 616399

KCNH1	98.6%	98.6%	100.0%	98.8%	Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNJ10	100.0%	100.0%	100.0%	99.5%	Enlarged vestibular aqueduct, digenic, 600791;SESAME syndrome, 612780
KCNJ11	100.0%	100.0%	100.0%	99.7%	Diabetes, permanent neonatal 2, with or without neurologic features, 618856;{Diabetes mellitus, type 2, susceptibility to}, 125853;Maturity-onset diabetes of the young, type 13, 616329;Diabetes mellitus, transient neonatal 3, 610582;Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	100.0%	99.9%	100.0%	97.8%	{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	100.0%	100.0%	100.0%	99.0%	Developmental and epileptic encephalopathy 7, 613720;Seizures, benign neonatal, 1, 121200;Myokymia, 121200
KCNQ3	100.0%	100.0%	100.0%	97.7%	Seizures, benign neonatal, 2, 121201
KCNT1	100.0%	100.0%	100.0%	98.0%	Developmental and epileptic encephalopathy 14, 614959;Epilepsy nocturnal frontal lobe, 5, 615005
KCNT2	99.7%	99.4%	100.0%	98.7%	Developmental and epileptic encephalopathy 57, 617771
KCTD7	100.0%	100.0%	100.0%	98.9%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	97.8%	97.6%	97.5%	69.9%	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534
KDM6B	100.0%	100.0%	100.0%	97.2%	Stolerman neurodevelopmental syndrome, 618505
KIF5A	100.0%	100.0%	100.0%	97.9%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187

KMT5B	100.0%	100.0%	100.0%	97.7%	Intellectual developmental disorder, autosomal dominant 51, 617788
KPTN	100.0%	100.0%	100.0%	97.9%	Intellectual developmental disorder, autosomal recessive 41, 615637
KRIT1	100.0%	100.0%	100.0%	98.1%	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860;Cerebral cavernous malformations-1, 116860;Cavernous malformations of CNS and retina, 116860
LAMB1	100.0%	99.7%	100.0%	98.8%	Lissencephaly 5, 615191
LGI1	100.0%	100.0%	100.0%	97.9%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	100.0%	100.0%	100.0%	99.2%	Hyperglycinemia, lactic acidosis, and seizures, 614462
LIPT2	100.0%	100.0%	100.0%	98.2%	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668
MAPK8IP3	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443

MAST3	100.0%	100.0%	100.0%	98.9%	Developmental and epileptic encephalopathy 108, 620115
MBD5	100.0%	100.0%	100.0%	98.0%	Intellectual developmental disorder, autosomal dominant 1, 156200
MECP2	100.0%	99.7%	97.9%	72.3%	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	100.0%	99.8%	97.5%	69.0%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MEF2C	100.0%	100.0%	100.0%	99.3%	Chromosome 5q14.3 deletion syndrome, 613443;Neurodevelopmental disorder with hypotonia, stereotypic hand movements, and impaired language, 613443

MFF	95.9%	95.9%	100.0%	98.8%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100.0%	100.0%	100.0%	99.2%	Macular dystrophy with central cone involvement, 616170;Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	100.0%	100.0%	100.0%	99.3%	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004
MOCS1	100.0%	100.0%	100.0%	98.8%	Molybdenum cofactor deficiency A, 252150
MOCS2	100.0%	100.0%	100.0%	98.5%	Molybdenum cofactor deficiency B, 252160
MPDU1	100.0%	100.0%	100.0%	97.2%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99.5%	99.1%	100.0%	98.8%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTFMT	100.0%	100.0%	100.0%	97.8%	Combined oxidative phosphorylation deficiency 15, 614947;Mitochondrial complex I deficiency, nuclear type 27, 618248

MTHFR	100.0%	100.0%	100.0%	98.3%	Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634;{Vascular disease, susceptibility to},
MTOR	100.0%	100.0%	100.0%	99.3%	Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MTRR	100.0%	100.0%	100.0%	98.4%	Homocystinuria-megaloblastic anemia, cbl E type, 236270;{Neural tube defects, folate-sensitive, susceptibility to}, 601634
NACC1	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	100.0%	100.0%	100.0%	98.1%	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442
NARS2	92.3%	92.3%	100.0%	98.5%	Combined oxidative phosphorylation deficiency 24, 616239;?Deafness, autosomal recessive 94, 618434

NBEA	97.4%	96.8%	100.0%	98.2%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NCDN	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with infantile epileptic spasms, 619373
NDUFA1	100.0%	100.0%	95.8%	64.4%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100.0%	98.8%	100.0%	96.7%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100.0%	100.0%	100.0%	98.3%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	67.4%	67.4%	100.0%	97.3%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100.0%	100.0%	100.0%	95.9%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	100.0%	100.0%	100.0%	95.3%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100.0%	100.0%	99.9%	96.1%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	100.0%	100.0%	100.0%	99.9%	Mitochondrial complex I deficiency, nuclear type 25, 618246

NDUFB9	100.0%	100.0%	100.0%	98.9%	?Mitochondrial complex I deficiency, nuclear type 24, 618245
NDUFS1	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	99.5%	96.5%	100.0%	98.3%	?Leber-like hereditary optic neuropathy, autosomal recessive 2, 620569;Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	96.5%	91.2%	100.0%	99.2%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100.0%	99.9%	100.0%	98.0%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	100.0%	100.0%	99.9%	98.8%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	100.0%	100.0%	100.0%	98.5%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	100.0%	100.0%	100.0%	98.5%	Developmental and epileptic encephalopathy 21, 615833
NEDD4L	100.0%	100.0%	100.0%	97.9%	Periventricular nodular heterotopia 7, 617201

NEU1	100.0%	100.0%	100.0%	99.4%	Sialidosis, type II, 256550;Sialidosis, type I, 256550
NEXMIF	100.0%	99.9%	97.4%	68.7%	Intellectual developmental disorder, X-linked 98, 300912
NGLY1	100.0%	100.0%	100.0%	98.7%	Congenital disorder of deglycosylation 1, 615273
NHLRC1	100.0%	100.0%	100.0%	99.3%	Myoclonic epilepsy of Lafora 2, 620681
NPRL2	100.0%	100.0%	100.0%	99.2%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100.0%	100.0%	100.0%	98.8%	Epilepsy, familial focal, with variable foci 3, 617118
NR2F1	100.0%	99.9%	99.9%	91.8%	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722
NR4A2	100.0%	100.0%	100.0%	98.2%	Intellectual developmental disorder with language impairment and early-onset DOPA-responsive dystonia-parkinsonism, 619911
NRXN1	99.8%	99.7%	100.0%	99.0%	Pitt-Hopkins-like syndrome 2, 614325;{Schizophrenia, susceptibility to, 17}, 614332
NUBPL	100.0%	100.0%	100.0%	98.7%	Mitochondrial complex I deficiency, nuclear type 21, 618242

NUS1	100.0%	100.0%	100.0%	98.7%	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831;?Congenital disorder of glycosylation, type 1aa, 617082
OCLN	94.5%	94.5%	100.0%	97.1%	Pseudo-TORCH syndrome 1, 251290
OFD1	100.0%	100.0%	96.1%	66.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OGDHL	100.0%	100.0%	100.0%	99.2%	Yoon-Bellen neurodevelopmental syndrome, 619701
OPHN1	93.9%	93.9%	98.0%	70.9%	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486
PACS1	100.0%	100.0%	99.9%	95.6%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	100.0%	100.0%	99.5%	96.7%	Developmental and epileptic encephalopathy 66, 618067
PAFAH1B1	100.0%	100.0%	100.0%	98.4%	Subcortical laminar heterotopia, 607432;Lissencephaly 1, 607432

PAK3	99.8%	99.3%	96.9%	70.1%	Intellectual developmental disorder, X-linked 30, 300558
PC	100.0%	100.0%	100.0%	99.7%	Pyruvate carboxylase deficiency, 266150
PCDH19	100.0%	99.9%	98.4%	72.7%	Developmental and epileptic encephalopathy 9, 300088
PDCD10	100.0%	100.0%	100.0%	96.9%	Cerebral cavernous malformations-3, 603285
PDHA1	99.6%	96.5%	97.6%	72.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	100.0%	100.0%	100.0%	98.8%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	100.0%	99.8%	99.9%	98.1%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100.0%	100.0%	100.0%	99.4%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	100.0%	100.0%	100.0%	97.3%	{Diabetes mellitus, type II, susceptibility to}, 125853;Pancreatic agenesis 1, 260370;MODY, type IV, 606392
PET100	100.0%	100.0%	100.0%	99.3%	Mitochondrial complex IV deficiency, nuclear type 12, 619055

PEX1	100.0%	100.0%	100.0%	98.5%	Heimler syndrome 1, 234580;Peroxisome biogenesis disorder 1B (NALD/IRD), 601539;Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	100.0%	100.0%	100.0%	99.8%	Peroxisome biogenesis disorder 6A (Zellweger), 614870;Peroxisome biogenesis disorder 6B, 614871
PEX12	100.0%	100.0%	100.0%	98.8%	Peroxisome biogenesis disorder 3B, 266510;Peroxisome biogenesis disorder 3A (Zellweger), 614859
PEX13	100.0%	100.0%	100.0%	97.8%	Peroxisome biogenesis disorder 11A (Zellweger), 614883;Peroxisome biogenesis disorder 11B, 614885
PEX14	100.0%	100.0%	100.0%	99.1%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	100.0%	100.0%	100.0%	99.2%	Peroxisome biogenesis disorder 8B, 614877;Peroxisome biogenesis disorder 8A (Zellweger), 614876
PEX19	100.0%	100.0%	100.0%	99.0%	Peroxisome biogenesis disorder 12A (Zellweger), 614886

PEX26	100.0%	100.0%	100.0%	98.0%	Peroxisome biogenesis disorder 7B, 614873;Peroxisome biogenesis disorder 7A (Zellweger), 614872
PEX3	100.0%	100.0%	100.0%	97.9%	Peroxisome biogenesis disorder 10A (Zellweger), 614882;?Peroxisome biogenesis disorder 10B, 617370
PEX5	100.0%	100.0%	100.0%	98.8%	Peroxisome biogenesis disorder 2B, 202370;Peroxisome biogenesis disorder 2A (Zellweger), 214110;Rhizomelic chondrodysplasia punctata, type 5, 616716
PEX6	100.0%	100.0%	100.0%	97.9%	Peroxisome biogenesis disorder 4B, 614863;Peroxisome biogenesis disorder 4A (Zellweger), 614862;Heimler syndrome 2, 616617
PGAP3	100.0%	100.0%	100.0%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716
PHF6	100.0%	100.0%	98.0%	73.9%	Borjeson-Forssman-Lehmann syndrome, 301900

PHGDH	100.0%	100.0%	100.0%	99.2%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PIGA	100.0%	100.0%	97.7%	73.6%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGN	100.0%	99.9%	100.0%	98.6%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100.0%	100.0%	100.0%	99.2%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGP	100.0%	100.0%	100.0%	96.8%	Developmental and epileptic encephalopathy 55, 617599
PIGT	100.0%	100.0%	100.0%	99.3%	?Paroxysmal nocturnal hemoglobinuria 2, 615399;Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398

PLA2G6	100.0%	99.9%	100.0%	99.2%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
PLCB1	100.0%	100.0%	100.0%	97.7%	Developmental and epileptic encephalopathy 12, 613722
PLP1	99.9%	98.9%	98.2%	69.4%	Pelizaeus-Merzbacher disease, 312080;Spastic paraplegia 2, X-linked, 312920
PLPBP	100.0%	100.0%	100.0%	99.2%	Epilepsy, early-onset, 1, vitamin B6-dependent, 617290
PMM2	100.0%	100.0%	100.0%	98.2%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100.0%	100.0%	100.0%	98.8%	?Charcot-Marie-Tooth disease, type 2B2, 605589;Ataxia-oculomotor apraxia 4, 616267;Microcephaly, seizures, and developmental delay, 613402
PNPO	100.0%	100.0%	100.0%	99.1%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090

POLG	100.0%	100.0%	100.0%	99.4%	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
PPP2R1A	94.0%	93.9%	100.0%	99.4%	Houge-Janssens syndrome 2, 616362
PPP2R5D	100.0%	100.0%	100.0%	98.7%	Houge-Janssens syndrome 1, 616355
PPT1	90.3%	90.3%	100.0%	97.8%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100.0%	100.0%	97.9%	68.4%	Renpenning syndrome, 309500
PRF1	100.0%	100.0%	100.0%	99.4%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PRICKLE1	100.0%	100.0%	100.0%	98.5%	Epilepsy, progressive myoclonic 1B, 612437

PRODH	100.0%	100.0%	100.0%	99.3%	{Schizophrenia, susceptibility to, 4}, 600850;Hyperprolinemia, type I, 239500
PRRT2	100.0%	100.0%	100.0%	97.7%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066;Seizures, benign familial infantile, 2, 605751;Episodic kinesigenic dyskinesia 1, 128200
PSAP	100.0%	100.0%	100.0%	99.1%	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	100.0%	100.0%	100.0%	98.8%	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263
PTS	100.0%	100.0%	100.0%	95.8%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	100.0%	100.0%	100.0%	98.6%	Spinocerebellar ataxia 47, 617931;Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719

PURA	100.0%	100.0%	100.0%	94.3%	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties, 616158
PYCR2	100.0%	100.0%	100.0%	98.6%	Leukodystrophy, hypomyelinating, 10, 616420
QARS1	100.0%	100.0%	100.0%	99.0%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100.0%	100.0%	100.0%	97.6%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB39B	100.0%	100.0%	97.8%	68.9%	Intellectual developmental disorder, X-linked 72, 300271;Waisman syndrome, 311510
RARS2	94.2%	93.1%	100.0%	98.6%	Pontocerebellar hypoplasia, type 6, 611523
RHOBTB2	98.7%	98.7%	100.0%	98.8%	Developmental and epileptic encephalopathy 64, 618004
RNASEH2A	100.0%	100.0%	100.0%	99.3%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91.4%	91.4%	100.0%	97.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100.0%	100.0%	100.0%	97.1%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	100.0%	100.0%	100.0%	99.1%	Kohlschutter-Tonz syndrome, 226750

RPS6KA3	99.9%	99.5%	97.7%	70.8%	Intellectual developmental disorder, X-linked 19, 300844;Coffin-Lowry syndrome, 303600
RRM2B	100.0%	100.0%	100.0%	97.7%	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	100.0%	100.0%	100.0%	98.1%	?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SCAF4	100.0%	100.0%	100.0%	98.6%	Fliedner-Zweier syndrome, 620511
SCARB2	100.0%	100.0%	100.0%	99.1%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900

SCN1A	100.0%	100.0%	100.0%	98.7%	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	100.0%	99.9%	100.0%	98.1%	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	100.0%	100.0%	100.0%	98.2%	Seizures, benign familial infantile, 3, 607745;Developmental and epileptic encephalopathy 11, 613721;Episodic ataxia, type 9, 618924
SCN3A	100.0%	100.0%	100.0%	98.2%	Epilepsy, familial focal, with variable foci 4, 617935;Developmental and epileptic encephalopathy 62, 617938

SCN8A	100.0%	100.0%	100.0%	98.6%	?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	100.0%	100.0%	100.0%	98.5%	Epilepsy, progressive myoclonic, 11, 618876
SEPSECS	98.6%	94.4%	100.0%	98.2%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	100.0%	100.0%	100.0%	98.9%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	99.8%	99.3%	99.8%	97.1%	Phelan-McDermid syndrome, 606232;{Schizophrenia 15}, 613950
SIK1	100.0%	100.0%	100.0%	99.5%	Developmental and epileptic encephalopathy 30, 616341
SLC12A5	100.0%	100.0%	100.0%	98.0%	{Epilepsy, idiopathic generalized, susceptibility to, 14}, 616685;Developmental and epileptic encephalopathy 34, 616645
SLC13A5	100.0%	100.0%	100.0%	98.3%	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905

SLC16A1	100.0%	100.0%	100.0%	99.7%	Hyperinsulinemic hypoglycemia, familial, 7, 610021;Erythrocyte lactate transporter defect, 245340;Monocarboxylate transporter 1 deficiency, 616095
SLC19A3	99.6%	98.4%	100.0%	98.1%	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483
SLC1A2	100.0%	99.8%	100.0%	99.2%	Developmental and epileptic encephalopathy 41, 617105
SLC25A1	100.0%	100.0%	100.0%	93.2%	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182;Myasthenic syndrome, congenital, 23, presynaptic, 618197
SLC25A15	100.0%	100.0%	100.0%	99.1%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	100.0%	100.0%	100.0%	99.6%	Developmental and epileptic encephalopathy 3, 609304

SLC2A1	100.0%	100.0%	100.0%	99.4%	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
SLC35A2	100.0%	100.0%	98.8%	74.8%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	100.0%	100.0%	100.0%	99.6%	Myoclonic-atonic epilepsy, 616421
SLC6A8	100.0%	99.6%	95.4%	67.7%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	100.0%	99.9%	97.6%	69.8%	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243
SMARCA2	98.0%	97.9%	100.0%	98.8%	Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293

SMC1A	100.0%	99.8%	96.9%	67.2%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMPD4	100.0%	100.0%	100.0%	99.4%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	100.0%	99.4%	97.6%	72.4%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNAP25	100.0%	100.0%	100.0%	98.5%	?Myasthenic syndrome, congenital, 18, 616330
SPTAN1	99.1%	98.8%	100.0%	98.7%	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	97.4%	95.3%	100.0%	99.3%	Developmental and epileptic encephalopathy 15, 615006;Intellectual developmental disorder, autosomal recessive 12, 611090
ST3GAL5	98.3%	98.3%	100.0%	97.8%	Salt and pepper developmental regression syndrome, 609056
STRADA	100.0%	100.0%	100.0%	98.8%	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087
STX1B	100.0%	100.0%	100.0%	97.4%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	100.0%	100.0%	100.0%	98.6%	Developmental and epileptic encephalopathy 4, 612164
SUOX	100.0%	100.0%	100.0%	99.0%	Sulfite oxidase deficiency, 272300
SYN1	100.0%	100.0%	96.2%	65.8%	Epilepsy, X-linked 1, with variable learning disabilities and behavior disorders, 300491;Intellectual developmental disorder, X-linked 50, 300115
SYNGAP1	100.0%	100.0%	100.0%	96.3%	Intellectual developmental disorder, autosomal dominant 5, 612621

SYNJ1	100.0%	100.0%	100.0%	98.2%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
SYP	100.0%	99.8%	98.1%	71.2%	Intellectual developmental disorder, X-linked 96, 300802
SZT2	100.0%	100.0%	100.0%	99.3%	Developmental and epileptic encephalopathy 18, 615476
TANGO2	100.0%	100.0%	100.0%	99.4%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	100.0%	100.0%	100.0%	98.5%	Pontocerebellar hypoplasia, type 11, 617695
TBC1D24	100.0%	100.0%	100.0%	99.6%	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	91.1%	90.1%	100.0%	98.7%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	100.0%	100.0%	100.0%	98.9%	Kenny-Caffey syndrome, type 1, 244460;Hypoparathyroidism-retardation-dysmorphism syndrome, 241410;Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TCF4	100.0%	100.0%	100.0%	98.4%	Pitt-Hopkins syndrome, 610954;Corneal dystrophy, Fuchs endothelial, 3, 613267
TDP2	100.0%	100.0%	100.0%	97.8%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	100.0%	100.0%	100.0%	98.9%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	100.0%	100.0%	100.0%	99.2%	Ceroid lipofuscinosis, neuronal, 2, 204500;Spinocerebellar ataxia, autosomal recessive 7, 609270

TREX1	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	98.4%	Combined oxidative phosphorylation deficiency 35, 617873
TRPM3	97.8%	97.8%	100.0%	98.5%	?Cataract 50 with or without glaucoma, 620253;Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224
TRPM6	100.0%	100.0%	100.0%	98.4%	Hypomagnesemia 1, intestinal, 602014
TSC1	100.0%	100.0%	100.0%	98.8%	Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690

TSC2	100.0%	100.0%	100.0%	99.5%	Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSEN15	100.0%	100.0%	99.9%	97.9%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	88.4%	88.4%	100.0%	98.3%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	100.0%	100.0%	100.0%	98.3%	Pontocerebellar hypoplasia type 2A, 277470;Pontocerebellar hypoplasia type 4, 225753;?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	100.0%	100.0%	100.0%	99.3%	Lissencephaly 3, 611603
TUBB2A	100.0%	100.0%	100.0%	99.5%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100.0%	100.0%	100.0%	99.7%	Cortical dysplasia, complex, with other brain malformations 7, 610031
TUBB4A	98.9%	95.9%	100.0%	98.9%	Dystonia 4, torsion, autosomal dominant, 128101;Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	100.0%	100.0%	100.0%	98.8%	Cortical dysplasia, complex, with other brain malformations 4, 615412

UBA5	99.6%	96.8%	100.0%	97.1%	?Spinocerebellar ataxia, autosomal recessive 24, 617133;Developmental and epileptic encephalopathy 44, 617132
UBE3A	100.0%	100.0%	100.0%	98.3%	Angelman syndrome, 105830
UBTF	100.0%	100.0%	99.9%	97.7%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UGP2	97.9%	96.6%	100.0%	98.1%	Developmental and epileptic encephalopathy 83, 618744
VPS11	100.0%	100.0%	100.0%	99.2%	?Dystonia 32, 619637;Leukodystrophy, hypomyelinating, 12, 616683
VPS53	82.7%	80.4%	100.0%	98.6%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	93.8%	93.8%	99.9%	93.3%	Skraban-Deardorff syndrome, 617616
WDR45	100.0%	100.0%	98.9%	76.4%	Neurodegeneration with brain iron accumulation 5, 300894

WFS1	91.2%	91.2%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.2%	Esophageal squamous cell carcinoma, somatic, 133239;Developmental and epileptic encephalopathy 28, 616211;Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	100.0%	99.9%	97.9%	71.5%	McLeod syndrome, 300842
YWHAG	100.0%	100.0%	100.0%	98.8%	Developmental and epileptic encephalopathy 56, 617665
ZEB2	100.0%	99.9%	100.0%	96.7%	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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

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