EPILEPSY GENE PANEL DG 2.18 (346 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AARS	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 29, 616339 Charcot-Marie-Tooth disease, axonal, type 2N, 613287
ABAT	100%	99,40%	100%	100%	GABA-transaminase deficiency, 613163
ABCC8	100%	99,80%	100%	100%	Diabetes mellitus, permanent neonatal, 606176 Diabetes mellitus, noninsulin-dependent, 125853 Diabetes mellitus, transient neonatal 2, 610374 Hyperinsulinemic hypoglycemia, familial, 1, 256450 Hypoglycemia of infancy, leucine-sensitive, 240800
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTL6B	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 76, 618468 Intellectual developmental disorder with severe speech and ambulation defects, 618470
ACY1	100%	98,80%	100%	100%	Aminoacylase 1 deficiency, 609924
ADSL	99,20%	98,70%	100%	100%	Adenylosuccinase deficiency, 103050
ALDH7A1	94,40%	88,80%	100%	100%	Epilepsy, pyridoxine-dependent, 266100
ALG1	53,00%	45,80%	100%	100%	Congenital disorder of glycosylation, type Ik, 608540
ALG11	96,80%	96,80%	96,80%	96,80%	Congenital disorder of glycosylation, type Ip, 613661
ALG13	98,40%	92,60%	100%	99,60%	Epileptic encephalopathy, early infantile, 36, 300884 ?Congenital disorder of glycosylation, type Is, 300884
ALG3	100%	99,70%	100%	100%	Congenital disorder of glycosylation, type Id, 601110
ALG6	98,60%	94,80%	100%	100%	Congenital disorder of glycosylation, type Ic, 603147
AMACR	100%	100%	100%	100%	Bile acid synthesis defect, congenital, 4, 214950 Alpha-methylacyl-CoA racemase deficiency, 614307
AMPD2	99,80%	98,90%	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
AMT	100%	100%	100%	100%	Glycine encephalopathy, 605899
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
AP3B2	99,40%	95,10%	100%	100%	Epileptic encephalopathy, early infantile, 48, 617276
APOPT1	81,90%	80,70%	93,50%	93,40%	Mitochondrial complex IV deficiency, 220110

ARHGEF9	76,50%	74,10%	97,20%	97,10%	Epileptic encephalopathy, early infantile, 8, 300607
ARID1B	99,50%	98,60%	99,90%	99,20%	Coffin-Siris syndrome 1, 135900
ARX	81,00%	64,00%	91,50%	85,70%	Proud syndrome, 300004 Partington syndrome, 309510 Lissencephaly, X-linked 2, 300215 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ASAH1	99,70%	98,60%	100%	100%	Farber lipogranulomatosis, 228000 Spinal muscular atrophy with progressive myoclonic epilepsy, 159950
ASL	100%	99,60%	100%	100%	Argininosuccinic aciduria, 207900
ASNS	99,40%	95,20%	100%	100%	Asparagine synthetase deficiency, 615574
ASXL3	99,90%	99,70%	100%	100%	Bainbridge-Ropers syndrome, 615485
ATP1A2	100%	100%	100%	100%	Migraine, familial hemiplegic, 2, 602481 Migraine, familial basilar, 602481 Alternating hemiplegia of childhood 1, 104290
ATP1A3	100%	99,90%	100%	100%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
ATP6AP2	94,10%	76,60%	100%	100%	Congenital disorder of glycosylation, type IIr, 301045 Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911
ATP7A	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATRX	99,40%	96,30%	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
AUTS2	98,20%	95,80%	100%	100%	Mental retardation, autosomal dominant 26, 615834
BOLA3	99,40%	90,20%	100%	100%	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299
BRAT1	99,70%	98,20%	100%	100%	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498 Neurodevelopmental disorder with cerebellar atrophy and with or without seizures, 618056
BTD	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
CACNA1A	95,10%	91,90%	100%	100%	Spinocerebellar ataxia 6, 183086 Epileptic encephalopathy, early infantile, 42, 617106 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500

CACNA1E	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA2D2	94,00%	93,20%	99,20%	97,60%	Cerebellar atrophy with seizures and variable developmental delay, 618501
CACNB4	95,50%	94,30%	100%	100%	Episodic ataxia, type 5, 613855
CAD	100%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 50, 616457
CASK	99,60%	96,80%	100%	100%	Mental retardation, with or without nystagmus, 300422 Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749 FG syndrome 4, 300422
CDKL5	95,00%	93,50%	95,60%	95,00%	Epileptic encephalopathy, early infantile, 2, 300672
CHD2	99,40%	99,20%	100%	100%	Epileptic encephalopathy, childhood-onset, 615369
CHRNA2	100%	100%	100%	100%	Epilepsy, nocturnal frontal lobe, type 4, 610353
CHRNA4	98,30%	96,20%	100%	100%	Epilepsy, nocturnal frontal lobe, 1, 600513
CHRNB2	99,30%	96,00%	100%	100%	Epilepsy, nocturnal frontal lobe, 3, 605375
CIC	63,30%	63,30%	100%	99,90%	Mental retardation, autosomal dominant 45, 617600
CLCN4	99,90%	98,90%	100%	100%	Raynaud-Claes syndrome, 300114
CLDN16	100%	100%	100%	100%	Hypomagnesemia 3, renal, 248250
CLDN19	98,50%	93,10%	100%	100%	Hypomagnesemia 5, renal, with ocular involvement, 248190
CLN3	92,50%	91,80%	92,50%	92,50%	Ceroid lipofuscinosis, neuronal, 3, 204200
CLN5	99,40%	95,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 5, 256731
CLN6	99,90%	97,10%	100%	100%	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300 Ceroid lipofuscinosis, neuronal, 6, 601780
CLN8	83,50%	83,50%	100%	100%	Ceroid lipofuscinosis, neuronal, 8, 600143 Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003
CNNM2	100%	100%	100%	100%	Hypomagnesemia 6, renal, 613882 Hypomagnesemia, seizures, and mental retardation, 616418
CNTN2	92,70%	92,70%	100%	100%	?Epilepsy, myoclonic, familial adult, 5, 615400
CNTNAP2	100%	99,80%	100%	100%	Pitt-Hopkins like syndrome 1, 610042 Cortical dysplasia-focal epilepsy syndrome, 610042
COL4A1	98,70%	97,40%	100%	100%	?Retinal arteries, tortuosity of, 180000 Brain small vessel disease with or without ocular anomalies, 175780 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Microangiopathy and leukoencephalopathy, pontine, autosomal dominant, 618564
COL4A3BP	99,60%	96,50%	100%	100%	Mental retardation, autosomal dominant 34, 616351
COLGALT1	93,30%	89,00%	98,60%	97,00%	Brain small vessel disease 3, 618360
COQ2	98,00%	95,30%	97,20%	97,20%	Coenzyme Q10 deficiency, primary, 1, 607426
COQ4	90,90%	89,30%	100%	100%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	100%	99,50%	100%	100%	Coenzyme Q10 deficiency, primary, 4, 612016

CPA6	99,60%	97,50%	100%	100%	Febrile seizures, familial, 11, 614418 Epilepsy, familial temporal lobe, 5, 614417
CPS1	100%	99,90%	100%	100%	Carbamoylphosphate synthetase I deficiency, 237300
CPT2	98,20%	97,80%	100%	100%	CPT II deficiency, myopathic, stress-induced, 255110 CPT II deficiency, infantile, 600649 CPT II deficiency, lethal neonatal, 608836
CSTB	99,60%	89,80%	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CTSD	98,40%	95,00%	100%	100%	Ceroid lipofuscinosis, neuronal, 10, 610127
CTSF	84,00%	79,30%	100%	99,90%	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362
CUL4B	98,00%	90,90%	99,90%	99,20%	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354
CUX2	99,90%	99,10%	100%	100%	Epileptic encephalopathy, early infantile, 67, 618141
D2HGDH	99,20%	97,20%	100%	100%	D-2-hydroxyglutaric aciduria, 600721
DCX	100%	99,90%	100%	100%	Subcortical laminal heterotopia, X-linked, 300067 Lissencephaly, X-linked, 300067
DDX3X	86,80%	84,90%	100%	100%	Mental retardation, X-linked 102, 300958
DENND5A	100%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 49, 617281
DEPDC5	100%	99,80%	100%	100%	Epilepsy, familial focal, with variable foci 1, 604364
DHDDS	99,00%	95,00%	95,20%	95,20%	Retinitis pigmentosa 59, 613861 ?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836
DLAT	100%	99,70%	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DNAJC5	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350
DNM1	92,60%	89,10%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 31, 616346
DNM1L	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DOCK7	99,80%	98,20%	100%	99,90%	Epileptic encephalopathy, early infantile, 23, 615859
DPAGT1	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ij, 608093 Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750
DPM1	98,20%	91,30%	99,70%	97,10%	Congenital disorder of glycosylation, type Ie, 608799
DPM2	100%	98,70%	100%	100%	Congenital disorder of glycosylation, type Iu, 615042
DPYD	99,70%	97,70%	100%	100%	Dihydropyrimidine dehydrogenase deficiency, 274270 5-fluorouracil toxicity, 274270
DPYS	100%	99,90%	100%	100%	Dihydropyrimidinuria, 222748
DYNC1H1	99,90%	99,40%	100%	100%	Mental retardation, autosomal dominant 13, 614563 Charcot-Marie-Tooth disease, axonal, type 20, 614228 Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600
DYRK1A	100%	100%	100%	100%	Mental retardation, autosomal dominant 7, 614104

EEF1A2	100%	100%	99,90%	99,10%	Epileptic encephalopathy, early infantile, 33, 616409 Mental retardation, autosomal dominant 38, 616393
EFHC1	93,10%	91,60%	98,00%	98,00%	No OMIM disease ID
EGF	99,90%	99,70%	100%	100%	Hypomagnesemia 4, renal, 611718
EHMT1	94,50%	93,70%	99,60%	99,50%	Kleefstra syndrome 1, 610253
EPM2A	94,20%	91,50%	100%	97,70%	Epilepsy, progressive myoclonic 2A (Lafora), 254780
ETHE1	99,90%	97,40%	100%	100%	Ethylmalonic encephalopathy, 602473
EXOSC3	99,50%	94,90%	100%	100%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	92,00%	83,10%	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
FARS2	100%	100%	100%	100%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FGD1	97,30%	92,80%	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF12	99,90%	98,10%	100%	100%	Epileptic encephalopathy, early infantile, 47, 617166
FLNA	100%	99,90%	100%	100%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FOLR1	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FOXG1	88,60%	82,10%	99,20%	96,40%	Rett syndrome, congenital variant, 613454
FOXRED1	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 19, 618241
FRMPD4	99,80%	98,50%	100%	100%	Mental retardation, X-linked 104, 300983
FRRS1L	79,70%	69,10%	99,20%	95,80%	Epileptic encephalopathy, early infantile, 37, 616981
FXYD2	100%	100%	100%	100%	Hypomagnesemia 2, renal, 154020
GABRA1	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 19, 615744
GABRB3	99,60%	98,20%	100%	100%	Epileptic encephalopathy, early infantile, 43, 617113
GABRG2	90,80%	90,20%	93,00%	93,00%	Epileptic encephalopathy, early infantile, 74, 618396 Febrile seizures, familial, 8, 607681 Epilepsy, generalized, with febrile seizures plus, type 3, 607681
GAMT	93,10%	82,70%	100%	100%	Cerebral creatine deficiency syndrome 2, 612736
GCK	100%	100%	100%	100%	Diabetes mellitus, noninsulin-dependent, late onset, 125853 Diabetes mellitus, permanent neonatal, 606176

					MODY, type II, 125851 Hyperinsulinemic hypoglycemia, familial, 3, 602485
GCSH	75,70%	68,90%	100%	100%	?Glycine encephalopathy, 605899
GLDC	89,90%	82,00%	100%	99,90%	Glycine encephalopathy, 605899
GLRA1	100%	99,80%	100%	100%	Hyperekplexia 1, 149400
GLRB	99,20%	95,10%	100%	100%	Hyperekplexia 2, 614619
GLUD1	94,20%	82,90%	100%	100%	Hyperinsulinism-hyperammonemia syndrome, 606762
GNAO1	93,80%	93,80%	100%	100%	Epileptic encephalopathy, early infantile, 17, 615473 Neurodevelopmental disorder with involuntary movements, 617493
GOSR2	95,90%	94,60%	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GPC3	99,10%	94,70%	100%	100%	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GPHN	100%	99,50%	100%	100%	Molybdenum cofactor deficiency C, 615501
GRIA3	99,70%	96,10%	100%	99,60%	Mental retardation, X-linked 94, 300699
GRIN1	100%	100%	100%	100%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254
GRIN2A	100%	100%	100%	100%	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570
GRIN2B	99,80%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRIN2D	79,80%	65,40%	93,90%	88,70%	Epileptic encephalopathy, early infantile, 46, 617162
GRN	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
HACE1	100%	99,30%	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HADH	99,00%	97,50%	100%	100%	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530 Hyperinsulinemic hypoglycemia, familial, 4, 609975
HCFC1	98,30%	93,60%	100%	100%	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type), 309541
HCN1	100%	99,70%	100%	100%	Generalized epilepsy with febrile seizures plus, type 10, 618482 Epileptic encephalopathy, early infantile, 24, 615871
HLCS	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
HNRNPU	99,90%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 54, 617391
HSD17B10	100%	99,10%	100%	100%	HSD10 mitochondrial disease, 300438
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
ICK	99,90%	98,70%	100%	100%	Endocrine-cerebroosteodysplasia, 612651
IDH2	99,70%	97,40%	100%	99,80%	D-2-hydroxyglutaric aciduria 2, 613657

IER3IP1	91,90%	82,60%	100%	100%	Microcephaly, epilepsy, and diabetes syndrome, 614231
IFIH1	99,70%	98,40%	100%	100%	Aicardi-Goutieres syndrome 7, 615846 Singleton-Merten syndrome 1, 182250
IQSEC2	96,80%	88,60%	99,40%	98,40%	Mental retardation, X-linked 1/78, 309530
IRF2BPL	99,50%	95,00%	99,90%	99,20%	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088
ITPA	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 35, 616647
JAM3	100%	99,90%	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
KANSL1	99,90%	99,20%	100%	100%	Koolen-De Vries syndrome, 610443
KATNB1	100%	99,90%	100%	100%	Lissencephaly 6, with microcephaly, 616212
KCNA1	100%	99,90%	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 32, 616366
KCNB1	100%	99,60%	100%	100%	Epileptic encephalopathy, early infantile, 26, 616056
KCNC1	100%	100%	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNH1	98,70%	98,70%	98,70%	98,70%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNJ10	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ11	100%	100%	100%	100%	Maturity-onset diabetes of the young, type 13, 616329 Diabetes, permanent neonatal, with or without neurologic features, 606176 Diabetes mellitus, transient neonatal, 3, 610582 Hyperinsulinemic hypoglycemia, familial, 2, 601820
KCNMA1	94,40%	93,60%	100%	100%	Liang-Wang syndrome, 618729 Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCNQ2	91,30%	89,80%	100%	100%	Epileptic encephalopathy, early infantile, 7, 613720 Seizures, benign neonatal, 1, 121200 Myokymia, 121200
KCNQ3	100%	99,40%	99,80%	99,10%	Seizures, benign neonatal, 2, 121201
KCNT1	96,00%	95,20%	98,60%	97,30%	Epilepsy, nocturnal frontal lobe, 5, 615005 Epileptic encephalopathy, early infantile, 14, 614959
KCTD7	95,00%	95,00%	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KDM5C	99,80%	97,90%	100%	100%	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534
KDM6B	98,80%	97,90%	100%	100%	Neurodevelopmental disorder with coarse facies and mild distal skeletal abnormalities, 618505
KPTN	100%	100%	100%	100%	Mental retardation, autosomal recessive 41, 615637
LAMB1	100%	99,90%	100%	100%	Lissencephaly 5, 615191
LGI1	98,50%	97,50%	100%	100%	Epilepsy, familial temporal lobe, 1, 600512
LIAS	100%	99,10%	100%	100%	Hyperglycinemia, lactic acidosis, and seizures, 614462

MAPK8IP3	100%	99,60%	100%	100%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MBD5	99,90%	99,90%	100%	100%	Mental retardation, autosomal dominant 1, 156200
MECP2	100%	98,70%	100%	99,90%	Mental retardation, X-linked syndromic, Lubs type, 300260 Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, atypical, 312750 Rett syndrome, 312750 Rett syndrome, preserved speech variant, 312750
MED12	99,80%	96,70%	100%	100%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEF2C	99,90%	96,00%	100%	100%	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443
MFF	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MFSD8	100%	99,70%	100%	100%	Macular dystrophy with central cone involvement, 616170 Ceroid lipofuscinosis, neuronal, 7, 610951
MLC1	100%	99,00%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MOCS1	99,20%	95,40%	100%	100%	Molybdenum cofactor deficiency A, 252150
MOCS2	99,60%	99,50%	100%	100%	Molybdenum cofactor deficiency B, 252160
MPDU1	100%	100%	100%	100%	Congenital disorder of glycosylation, type If, 609180
MPDZ	99,80%	98,80%	100%	100%	Hydrocephalus, congenital, 2, with or without brain or eye anomalies, 615219
MTHFR	97,30%	96,00%	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
MTOR	100%	99,50%	100%	100%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MTRR	100%	99,60%	100%	100%	Homocystinuria-megaloblastic anemia, cbl E type, 236270
NACC1	100%	99,80%	100%	100%	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination, 617393
NANS	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NBEA	92,00%	90,60%	100%	100%	No OMIM disease ID
NDUFA1	99,90%	99,30%	100%	100%	Mitochondrial complex I deficiency, nuclear type 12, 301020
NDUFA11	100%	100%	100%	99,80%	Mitochondrial complex I deficiency, nuclear type 14, 618236
NDUFAF1	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 11, 618234
NDUFAF2	95,00%	83,40%	100%	99,90%	Mitochondrial complex I deficiency, nuclear type 10, 618233
NDUFAF3	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 18, 618240
NDUFAF4	99,80%	98,20%	100%	100%	Mitochondrial complex I deficiency, nuclear type 15, 618237
NDUFAF5	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 16, 618238
NDUFB3	95,80%	80,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 25, 618246
NDUFB9	98,40%	95,50%	100%	100%	?Mitochondrial complex I deficiency, nuclear type 24, 618245

NDUFS1	100%	99,50%	100%	100%	Mitochondrial complex I deficiency, nuclear type 5, 618226
NDUFS2	100%	100%	100%	100%	Mitochondrial complex I deficiency, nuclear type 6, 618228
NDUFS3	90,70%	90,60%	91,90%	90,70%	Mitochondrial complex I deficiency, nuclear type 8, 618230
NDUFS4	100%	99,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 1, 252010
NDUFS6	100%	99,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 9, 618232
NDUFV1	98,00%	96,10%	100%	100%	Mitochondrial complex I deficiency, nuclear type 4, 618225
NDUFV2	86,90%	76,90%	100%	100%	Mitochondrial complex I deficiency, nuclear type 7, 618229
NECAP1	100%	100%	100%	100%	?Epileptic encephalopathy, early infantile, 21, 615833
NEDD4L	72,00%	71,50%	100%	100%	Periventricular nodular heterotopia 7, 617201
NEU1	99,70%	97,70%	100%	100%	Sialidosis, type II, 256550 Sialidosis, type I, 256550
NEXMIF	100%	99,50%	100%	100%	Mental retardation, X-linked 98, 300912
NGLY1	100%	99,80%	100%	100%	Congenital disorder of deglycosylation, 615273
NHLRC1	100%	98,70%	100%	100%	Epilepsy, progressive myoclonic 2B (Lafora), 254780
NPRL2	100%	100%	100%	100%	Epilepsy, familial focal, with variable foci 2, 617116
NPRL3	100%	99,60%	100%	100%	Epilepsy, familial focal, with variable foci 3, 617118
NRXN1	97,40%	96,90%	100%	99,80%	Pitt-Hopkins-like syndrome 2, 614325
NUBPL	99,70%	98,40%	100%	100%	Mitochondrial complex I deficiency, nuclear type 21, 618242
OCLN	100%	100%	100%	100%	Pseudo-TORCH syndrome 1, 251290
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OPHN1	99,50%	97,60%	99,90%	98,80%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PACS1	98,80%	96,90%	100%	100%	Schuurs-Hoeijmakers syndrome, 615009
PACS2	99,30%	96,20%	100%	99,80%	Epileptic encephalopathy, early infantile, 66, 618067
PAFAH1B1	94,10%	87,10%	100%	100%	Subcortical laminar heterotopia, 607432 Lissencephaly 1, 607432
PAK3	99,30%	95,90%	100%	99,80%	Mental retardation, X-linked 30/47, 300558
PC	99,80%	97,30%	100%	100%	Pyruvate carboxylase deficiency, 266150
PCDH19	100%	98,90%	100%	100%	Epileptic encephalopathy, early infantile, 9, 300088
PDHA1	99,40%	97,10%	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHB	99,10%	97,50%	100%	100%	Pyruvate dehydrogenase E1-beta deficiency, 614111
PDHX	99,90%	99,40%	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDP1	100%	100%	100%	100%	Pyruvate dehydrogenase phosphatase deficiency, 608782
PDX1	93,00%	82,40%	100%	100%	Pancreatic agenesis 1, 260370 MODY, type IV, 606392

PET100	100%	99,60%	100%	100%	Mitochondrial complex IV deficiency, 220110
PEX1	99,90%	99,40%	100%	100%	Heimler syndrome 1, 234580 Peroxisome biogenesis disorder 1B (NALD/IRD), 601539 Peroxisome biogenesis disorder 1A (Zellweger), 214100
PEX10	96,80%	89,70%	100%	99,90%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX12	100%	100%	100%	100%	Peroxisome biogenesis disorder 3A (Zellweger), 614859 Peroxisome biogenesis disorder 3B, 266510
PEX13	100%	100%	100%	100%	Peroxisome biogenesis disorder 11A (Zellweger), 614883 Peroxisome biogenesis disorder 11B, 614885
PEX14	96,70%	90,80%	100%	100%	Peroxisome biogenesis disorder 13A (Zellweger), 614887
PEX16	97,90%	94,20%	100%	100%	Peroxisome biogenesis disorder 8A (Zellweger), 614876 Peroxisome biogenesis disorder 8B, 614877
PEX19	99,90%	98,50%	100%	100%	Peroxisome biogenesis disorder 12A (Zellweger), 614886
PEX26	100%	100%	100%	100%	Peroxisome biogenesis disorder 7A (Zellweger), 614872 Peroxisome biogenesis disorder 7B, 614873
PEX3	100%	99,30%	100%	100%	Peroxisome biogenesis disorder 10A (Zellweger), 614882 ?Peroxisome biogenesis disorder 10B, 617370
PEX5	99,90%	99,00%	100%	100%	Peroxisome biogenesis disorder 2B, 202370 Rhizomelic chondrodysplasia punctata, type 5, 616716 Peroxisome biogenesis disorder 2A (Zellweger), 214110
PEX6	94,50%	86,70%	100%	100%	Peroxisome biogenesis disorder 4B, 614863 Heimler syndrome 2, 616617 Peroxisome biogenesis disorder 4A (Zellweger), 614862
PGAP3	63,50%	59,60%	100%	100%	Hyperphosphatasia with mental retardation syndrome 4, 615716
PHF6	97,80%	88,30%	99,90%	98,90%	Borjeson-Forssman-Lehmann syndrome, 301900
PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D23	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	93,80%	91,50%	98,80%	98,80%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	99,90%	100%	100%	Hyperphosphatasia with mental retardation syndrome 2, 614749
PIGP	95,80%	87,30%	100%	100%	Epileptic encephalopathy, early infantile, 55, 617599
PIGT	98,10%	98,10%	100%	100%	?Paroxysmal nocturnal hemoglobinuria 2, 615399 Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398

PLA2G6	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
PLCB1	100%	99,80%	100%	100%	Epileptic encephalopathy, early infantile, 12, 613722
PLP1	100%	99,20%	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PLPBP	98,20%	90,10%	100%	100%	Epilepsy, early-onset, vitamin B6-dependent, 617290
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKP	100%	100%	100%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPO	99,90%	97,70%	100%	100%	Pyridoxamine 5'-phosphate oxidase deficiency, 610090
POLG	100%	99,30%	100%	100%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PPP2R1A	91,60%	91,50%	93,60%	93,60%	Mental retardation, autosomal dominant 36, 616362
PPP2R5D	100%	100%	100%	100%	Mental retardation, autosomal dominant 35, 616355
PPT1	90,30%	90,30%	82,50%	82,50%	Ceroid lipofuscinosis, neuronal, 1, 256730
PQBP1	100%	100%	100%	100%	Renpenning syndrome, 309500
PRF1	91,20%	90,80%	100%	100%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
PRICKLE1	100%	100%	100%	100%	Epilepsy, progressive myoclonic 1B, 612437
PRRT2	100%	99,60%	100%	100%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PSAP	100%	100%	100%	100%	Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Combined SAP deficiency, 611721 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PTS	99,90%	99,10%	100%	100%	Hyperphenylalaninemia, BH4-deficient, A, 261640
PUM1	100%	99,90%	100%	100%	Spinocerebellar ataxia 47, 617931
PURA	99,00%	95,20%	100%	99,80%	Mental retardation, autosomal dominant 31, 616158
PYCR2	100%	99,10%	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420
QARS	100%	100%	100%	100%	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760
QDPR	100%	99,70%	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630

RAB39B	100%	100%	100%	100%	Waisman syndrome, 311510 Mental retardation, X-linked 72, 300271
RARS2	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
RNASEH2A	100%	100%	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	96,00%	92,50%	100%	99,80%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	99,50%	100%	100%	Aicardi-Goutieres syndrome 3, 610329
ROGDI	98,40%	95,20%	99,90%	99,10%	Kohlschutter-Tonz syndrome, 226750
RPS6KA3	98,40%	94,40%	99,90%	98,80%	Mental retardation, X-linked 19, 300844 Coffin-Lowry syndrome, 303600
RRM2B	100%	99,70%	100%	100%	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077 Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075 Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCARB2	100%	99,80%	100%	100%	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900
SCN1A	99,90%	99,50%	100%	100%	Febrile seizures, familial, 3A, 604403 Migraine, familial hemiplegic, 3, 609634 Epilepsy, generalized, with febrile seizures plus, type 2, 604403 Epileptic encephalopathy, early infantile, 6 (Dravet syndrome), 607208
SCN1B	98,00%	96,40%	99,80%	99,30%	Epileptic encephalopathy, early infantile, 52, 617350 Atrial fibrillation, familial, 13, 615377 Cardiac conduction defect, nonspecific, 612838 Epilepsy, generalized, with febrile seizures plus, type 1, 604233 Brugada syndrome 5, 612838
SCN2A	99,60%	97,60%	100%	100%	Epileptic encephalopathy, early infantile, 11, 613721 Seizures, benign familial infantile, 3, 607745
SCN3A	99,90%	99,30%	100%	100%	Epilepsy, familial focal, with variable foci 4, 617935 Epileptic encephalopathy, early infantile, 62, 617938
SCN8A	100%	99,80%	100%	100%	Seizures, benign familial infantile, 5, 617080 Cognitive impairment with or without cerebellar ataxia, 614306 ?Myoclonus, familial, 2, 618364 Epileptic encephalopathy, early infantile, 13, 614558
SEMA6B	80,60%	73,60%	100%	100%	No OMIM disease ID
SEPSECS	100%	100%	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERPINI1	99,90%	99,00%	100%	100%	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218
SHANK3	92,20%	82,10%	98,00%	94,40%	Phelan-McDermid syndrome, 606232
SIK1	98,70%	94,40%	100%	100%	Epileptic encephalopathy, early infantile, 30, 616341
SLC12A5	83,90%	83,80%	97,40%	97,40%	Epileptic encephalopathy, early infantile, 34, 616645

SLC13A5	100%	99,90%	100%	100%	Epileptic encephalopathy, early infantile, 25, 615905
SLC16A1	100%	99,30%	100%	100%	Monocarboxylate transporter 1 deficiency, 616095 Hyperinsulinemic hypoglycemia, familial, 7, 610021 Erythrocyte lactate transporter defect, 245340
SLC19A3	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A2	100%	99,30%	100%	100%	Epileptic encephalopathy, early infantile, 41, 617105
SLC25A1	95,80%	88,60%	99,50%	97,80%	Myasthenic syndrome, congenital, 23, presynaptic, 618197 Combined D-2- and L-2-hydroxyglutaric aciduria, 615182
SLC25A15	99,80%	98,10%	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC25A22	98,60%	95,80%	100%	100%	Epileptic encephalopathy, early infantile, 3, 609304
SLC2A1	92,80%	92,80%	100%	100%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 GLUT1 deficiency syndrome 2, childhood onset, 612126
SLC35A2	99,90%	98,40%	100%	100%	Congenital disorder of glycosylation, type IIm, 300896
SLC6A1	100%	100%	100%	100%	Myoclonic-atonic epilepsy, 616421
SLC6A8	93,50%	81,60%	100%	99,80%	Cerebral creatine deficiency syndrome 1, 300352
SLC9A6	99,30%	95,50%	100%	98,30%	Mental retardation, X-linked syndromic, Christianson type, 300243
SMARCA2	96,70%	96,20%	97,40%	96,80%	Nicolaides-Baraitser syndrome, 601358
SMC1A	100%	98,70%	100%	99,80%	Cornelia de Lange syndrome 2, 300590 Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044
SMPD4	99,40%	94,20%	100%	100%	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622
SMS	91,50%	78,50%	100%	99,90%	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNAP25	100%	99,90%	100%	100%	?Myasthenic syndrome, congenital, 18, 616330
SPATA5	100%	99,70%	100%	100%	Epilepsy, hearing loss, and mental retardation syndrome, 616577
SPTAN1	99,10%	98,60%	100%	100%	Epileptic encephalopathy, early infantile, 5, 613477
ST3GAL3	100%	99,80%	100%	100%	Mental retardation, autosomal recessive 12, 611090 ?Epileptic encephalopathy, early infantile, 15, 615006
ST3GAL5	85,00%	84,20%	98,70%	98,40%	Salt and pepper developmental regression syndrome, 609056
STX1B	100%	100%	100%	100%	Generalized epilepsy with febrile seizures plus, type 9, 616172
STXBP1	96,80%	96,50%	100%	100%	Epileptic encephalopathy, early infantile, 4, 612164
SUOX	100%	100%	100%	100%	Sulfite oxidase deficiency, 272300
SYN1	81,90%	73,20%	100%	99,60%	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491
SYNGAP1	99,40%	98,10%	100%	100%	Mental retardation, autosomal dominant 5, 612621
SYNJ1	99,90%	99,40%	100%	100%	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530

SYP	99,90%	96,70%	100%	100%	Mental retardation, X-linked 96, 300802
SZT2	99,60%	99,50%	100%	99,90%	Epileptic encephalopathy, early infantile, 18, 615476
TBC1D24	100%	100%	100%	100%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBCD	96,20%	94,40%	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TBCE	99,80%	97,30%	100%	100%	Kenny-Caffey syndrome, type 1, 244460 Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207
TCF4	100%	99,80%	100%	100%	Corneal dystrophy, Fuchs endothelial, 3, 613267 Pitt-Hopkins syndrome, 610954
TDP2	100%	99,40%	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TOE1	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TPP1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TREX1	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRPM3	100%	99,50%	100%	100%	No OMIM disease ID
TRPM6	99,90%	99,50%	100%	100%	Hypomagnesemia 1, intestinal, 602014
TSC1	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSEN15	99,80%	97,50%	100%	100%	Pontocerebellar hypoplasia, type 2F, 617026
TSEN2	100%	99,60%	100%	100%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	96,30%	94,30%	99,90%	98,90%	Pontocerebellar hypoplasia type 4, 225753 Pontocerebellar hypoplasia type 2A, 277470 ?Pontocerebellar hypoplasia type 5, 610204
TUBA1A	99,90%	97,00%	100%	100%	Lissencephaly 3, 611603
TUBB2A	97,00%	95,70%	100%	100%	Cortical dysplasia, complex, with other brain malformations 5, 615763
TUBB2B	100%	99,50%	100%	100%	Cortical dysplasia, complex, with other brain malformations 7, 610031

TUBB4A	95,90%	94,00%	97,10%	96,00%	Leukodystrophy, hypomyelinating, 6, 612438 Dystonia 4, torsion, autosomal dominant, 128101
TUBG1	100%	100%	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
UBA5	97,80%	86,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 24, 617133 Epileptic encephalopathy, early infantile, 44, 617132
UBE3A	99,10%	94,80%	100%	100%	Angelman syndrome, 105830
UBTF	100%	99,40%	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
UGP2	99,00%	98,60%	96,30%	96,30%	Epileptic encephalopathy, early infantile, 83, 618744
VPS53	91,50%	90,70%	100%	99,30%	Pontocerebellar hypoplasia, type 2E, 615851
WDR26	99,00%	96,60%	100%	100%	Skraban-Deardorff syndrome, 617616
WDR45	96,40%	89,70%	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
WWOX	100%	100%	100%	100%	Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211
XK	99,80%	98,10%	100%	100%	McLeod syndrome with or without chronic granulomatous disease, 300842
YWHAG	100%	100%	100%	100%	Epileptic encephalopathy, early infantile, 56, 617665
ZEB2	99,90%	99,10%	97,40%	97,40%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors