MOVEMENT DISORDERS GENE PANEL DG 2.18 (338 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
AARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCB7	99,50%	98,20%	99,80%	99,30%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	75,80%	71,60%	100%	100%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ABHD12	98,70%	92,30%	100%	99,30%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADAR	100%	99,80%	100%	100%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCY5	95,10%	91,20%	99,20%	98,00%	Dyskinesia, familial, with facial myokymia, 606703
ADGRG1	100%	100%	100%	100%	Polymicrogyria, bilateral perisylvian, 615752 Polymicrogyria, bilateral frontoparietal, 606854
ADPRHL2	100%	99,80%	100%	100%	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170
AFG3L2	95,00%	91,10%	100%	99,90%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGTPBP1	96,00%	94,10%	100%	100%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIMP1	99,20%	94,50%	100%	99,90%	Leukodystrophy, hypomyelinating, 3, 260600
ALDH18A1	100%	99,90%	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH3A2	95,30%	94,60%	100%	100%	Sjogren-Larsson syndrome, 270200
ALS2	100%	99,90%	100%	100%	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
AMPD2	99,80%	98,90%	100%	100%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
ANO10	99,80%	97,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	99,50%	97,80%	100%	100%	Dystonia 24, 615034

AP4B1	99,90%	98,70%	100%	100%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	99,80%	98,70%	100%	100%	Stuttering, familial persistent, 1, 184450 Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	99,90%	98,90%	100%	100%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	78,90%	71,30%	87,90%	87,90%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	94,90%	92,50%	100%	100%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	100%	100%	100%	100%	Argininemia, 207800
ARSA	100%	99,80%	100%	100%	Metachromatic leukodystrophy, 250100
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
ASPA	99,90%	98,30%	100%	100%	Canavan disease, 271900
ATCAY	100%	99,80%	100%	100%	Ataxia, cerebellar, Cayman type, 601238
ATL1	100%	99,70%	100%	100%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATM	99,80%	98,10%	100%	100%	Ataxia-telangiectasia, 208900 Lymphoma, mantle cell, somatic, 0 Lymphoma, B-cell non-Hodgkin, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0
ATP13A2	100%	99,50%	100%	100%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
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
ATP2B3	99,50%	97,50%	100%	100%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	99,90%	99,20%	100%	100%	Wilson disease, 277900
B4GALNT1	99,30%	95,00%	100%	100%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	92,60%	83,20%	100%	99,90%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	99,90%	99,20%	100%	100%	Maple syrup urine disease, type Ia, 248600
BCKDHB	99,50%	94,40%	100%	100%	Maple syrup urine disease, type Ib, 248600

BCL11B	99,10%	95,60%	98,80%	97,30%	Immunodeficiency 49, 617237 Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092
BSCL2	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
C12orf65	99,80%	98,50%	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035 Combined oxidative phosphorylation deficiency 7, 613559
C19orf12	100%	99,80%	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CA8	99,60%	97,30%	100%	100%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
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
CACNA1G	100%	99,60%	100%	100%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNB4	95,50%	94,30%	100%	100%	Episodic ataxia, type 5, 613855
CAMTA1	100%	99,50%	100%	100%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN1	100%	100%	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	100%	99,70%	100%	100%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHMP1A	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 8, 614961
CLCN2	100%	99,50%	100%	100%	Leukoencephalopathy with ataxia, 615651 Hyperaldosteronism, familial, type II, 605635
CLCN4	99,90%	98,90%	100%	100%	Raynaud-Claes syndrome, 300114
CLPB	100%	99,90%	100%	100%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COASY	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 12, 618266 Neurodegeneration with brain iron accumulation 6, 615643
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
COL6A1	100%	99,40%	100%	100%	Ullrich congenital muscular dystrophy 1, 254090 Bethlem myopathy 1, 158810

COL6A2	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 ?Myosclerosis, congenital, 255600 Ullrich congenital muscular dystrophy 1, 254090
COL6A3	100%	99,80%	100%	100%	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090
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
COQ9	100%	97,90%	100%	100%	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	97,80%	88,30%	100%	100%	Mitochondrial complex IV deficiency, 220110
CP	94,80%	88,90%	100%	100%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CSF1R	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSTB	99,60%	89,80%	100%	100%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	98,90%	96,70%	100%	100%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	94,80%	91,50%	100%	99,90%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	98,00%	92,80%	100%	100%	Spastic paraplegia 5A, autosomal recessive, 270800 Bile acid synthesis defect, congenital, 3, 613812
DARS2	100%	99,30%	100%	100%	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105
DBT	99,80%	98,00%	100%	100%	Maple syrup urine disease, type II, 248600
DCAF17	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
DCC	100%	100%	100%	100%	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500
DCTN1	100%	98,80%	100%	100%	Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DDC	99,70%	96,40%	100%	100%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	97,90%	95,80%	100%	100%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	100%	99,60%	100%	100%	Spastic paraplegia 54, autosomal recessive, 615033
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
DLD	100%	99,70%	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAJC12	87,40%	87,40%	100%	100%	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384

DNAJC3	100%	99,70%	100%	100%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAL4	100%	98,90%	100%	100%	?Mirror movements 3, 616059
DNM1L	99,90%	98,50%	100%	100%	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1, 614388 Optic atrophy 5, 610708
DNMT1	99,20%	99,00%	99,70%	99,20%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
DPYS	100%	99,90%	100%	100%	Dihydropyrimidinuria, 222748
ECHS1	99,90%	99,00%	100%	100%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2B1	100%	100%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	99,90%	99,50%	100%	100%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
EIF2B3	100%	100%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	100%	99,90%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	100%	99,00%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
ELOVL4	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ELOVL5	100%	99,80%	100%	100%	Spinocerebellar ataxia 38, 615957
ERLIN2	100%	99,90%	100%	100%	Spastic paraplegia 18, autosomal recessive, 611225
ETHE1	99,90%	97,40%	100%	100%	Ethylmalonic encephalopathy, 602473
EXOSC3	99,50%	94,90%	100%	100%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	100%	No OMIM disease ID
FA2H	92,00%	83,10%	100%	100%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	100%	99,40%	100%	100%	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	97,60%	92,80%	100%	100%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	100%	100%	100%	100%	Spastic paraplegia 77, autosomal recessive, 617046 Combined oxidative phosphorylation deficiency 14, 614946
FBXO7	99,80%	97,90%	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FGF14	100%	100%	100%	100%	Spinocerebellar ataxia 27, 609307
FLVCR1	100%	98,90%	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	100%	100%	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	99,90%	99,10%	100%	99,60%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700

FTL	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
GALC	99,80%	98,30%	100%	100%	Krabbe disease, 245200
GAN	100%	99,60%	100%	100%	Giant axonal neuropathy-1, 256850
GBA	100%	100%	100%	100%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GBA2	100%	99,70%	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	100%	99,20%	100%	100%	Glutaricaciduria, type I, 231670
GCH1	99,90%	95,50%	100%	100%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GDAP2	100%	99,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GFAP	91,80%	89,70%	100%	100%	Alexander disease, 203450
GJC2	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GLB1	99,90%	97,40%	100%	100%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GNAL	96,80%	93,40%	100%	100%	Dystonia 25, 615073
GOSR2	95,90%	94,60%	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GPR143	85,80%	76,40%	99,80%	97,90%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GRID2	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive 18, 616204
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
GRIN2B	99,80%	99,20%	100%	100%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRM1	100%	99,70%	100%	100%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
HACE1	100%	99,30%	100%	100%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HEXB	99,60%	96,90%	100%	99,90%	Sandhoff disease, infantile, juvenile, and adult forms, 268800

HK1	100%	100%	100%	100%	Hemolytic anemia due to hexokinase deficiency, 235700 Neuropathy, hereditary motor and sensory, Russe type, 605285 Neurodevelopmental disorder with visual defects and brain anomalies, 618547 Retinitis pigmentosa 79, 617460
HPRT1	99,30%	91,80%	100%	99,30%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPD1	98,80%	93,70%	100%	100%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
IBA57	93,70%	90,10%	100%	100%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
MR E11	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
ISCA2	100%	98,80%	100%	100%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITPR1	100%	99,90%	100%	100%	Spinocerebellar ataxia 15, 606658 Gillespie syndrome, 206700 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
JAM3	100%	99,90%	100%	100%	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730
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
KCNC1	100%	100%	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	81,10%	69,40%	94,70%	89,00%	Spinocerebellar ataxia 13, 605259
KCND3	100%	99,40%	100%	100%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNJ10	89,30%	89,00%	100%	100%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	100%	100%	100%	100%	Keppen-Lubinsky syndrome, 614098
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
KCTD7	95,00%	95,00%	100%	100%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA1161	100%	100%	100%	100%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
KIDINS220	100%	100%	100%	100%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF1A	99,40%	97,10%	100%	100%	NESCAV syndrome, 614255 Spastic paraplegia 30, autosomal dominant, 610357

					Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	100%	100%	100%	100%	Spastic ataxia 2, autosomal recessive, 611302
KIF5A	100%	99,90%	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
KMT2B	95,80%	94,00%	98,70%	97,90%	Dystonia 28, childhood-onset, 617284
L1CAM	99,90%	99,10%	100%	100%	MASA syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000
LAMA1	100%	99,70%	100%	100%	Poretti-Boltshauser syndrome, 615960
LAMB1	100%	99,90%	100%	100%	Lissencephaly 5, 615191
LMNB1	99,90%	98,90%	100%	100%	Leukodystrophy, adult-onset, autosomal dominant, 169500
MAPK8IP3	100%	99,60%	100%	100%	Neurodevelopmental disorder with or without variable brain abnormalities, 618443
MARS2	100%	100%	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
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
MECR	100%	98,90%	100%	100%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MFF	94,30%	89,90%	100%	100%	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086
MICU1	98,90%	95,20%	100%	100%	Myopathy with extrapyramidal signs, 615673
MLC1	100%	99,00%	100%	100%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMADHC	94,40%	83,50%	89,70%	89,70%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MTHFR	97,30%	96,00%	100%	100%	Homocystinuria due to MTHFR deficiency, 236250
MTPAP	99,50%	96,10%	100%	100%	?Spastic ataxia 4, autosomal recessive, 613672
MTTP	100%	99,60%	100%	100%	Abetalipoproteinemia, 200100
NANS	100%	99,90%	100%	100%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NEFL	99,90%	98,20%	100%	100%	Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 2E, 607684

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
NF2	100%	99,90%	100%	100%	Meningioma, NF2-related, somatic, 607174 Schwannomatosis, somatic, 162091 Neurofibromatosis, type 2, 101000
NIPA1	100%	100%	99,80%	98,50%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	98,60%	85,60%	100%	100%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NKX6-2	89,00%	81,80%	100%	100%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NOL3	93,70%	84,20%	100%	100%	?Myoclonus, familial, 1, 614937
NPC1	99,60%	98,70%	100%	100%	Niemann-Pick disease, type D, 257220 Niemann-Pick disease, type C1, 257220
NPC2	100%	99,60%	100%	100%	Niemann-pick disease, type C2, 607625
NT5C2	98,00%	96,50%	100%	100%	Spastic paraplegia 45, autosomal recessive, 613162
NUP62	100%	100%	100%	100%	Striatonigral degeneration, infantile, 271930
OCLN	100%	100%	100%	100%	Pseudo-TORCH syndrome 1, 251290
OPA1	99,70%	97,60%	100%	100%	Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896
OPHN1	99,50%	97,60%	99,90%	98,80%	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486
PACS2	99,30%	96,20%	100%	99,80%	Epileptic encephalopathy, early infantile, 66, 618067
PANK2	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX6	100%	100%	100%	100%	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Cataract with late-onset corneal dystrophy, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229
PCYT2	99,80%	97,10%	100%	98,80%	Spastic paraplegia 82, autosomal recessive, 618770
PDE10A	81,20%	80,50%	100%	100%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922

PDE8B	99,90%	99,70%	100%	100%	Striatal degeneration, autosomal dominant, 609161 Pigmented nodular adrenocortical disease, primary, 3, 614190
PDGFB	100%	99,30%	100%	100%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRB	99,20%	97,50%	100%	100%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDHA1	99,40%	97,10%	100%	100%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	99,90%	99,40%	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	94,70%	87,60%	97,30%	96,60%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	99,80%	97,10%	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	100%	100%	100%	100%	Spinocerebellar ataxia 23, 610245
PEX10	96,80%	89,70%	100%	99,90%	Peroxisome biogenesis disorder 6B, 614871 Peroxisome biogenesis disorder 6A (Zellweger), 614870
PEX2	100%	100%	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	100%	99,60%	100%	100%	Refsum disease, 266500
TAF1	99,80%	97,70%	100%	100%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TANGO2	100%	99,30%	100%	100%	metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D20	94,20%	94,20%	100%	99,90%	Warburg micro syndrome 4, 615663
TBC1D23	99,70%	97,20%	100%	100%	Pontocerebellar hypoplasia, type 11, 617695
PIK3R5	100%	99,90%	100%	100%	Ataxia-oculomotor apraxia 3, 615217
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
PLP1	100%	99,20%	100%	100%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	100%	100%	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	97,70%	94,20%	100%	100%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	100%	99,90%	100%	100%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	100%	100%	100%	100%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402

PNPLA6	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
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
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR3A	100%	99,70%	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,90%	98,60%	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
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
PRKCG	99,90%	98,40%	100%	100%	Spinocerebellar ataxia 14, 605361
PRKRA	100%	99,40%	100%	100%	Dystonia 16, 612067
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
PYCR2	100%	99,10%	100%	100%	Leukodystrophy, hypomyelinating, 10, 616420
QDPR	100%	99,70%	100%	100%	Hyperphenylalaninemia, BH4-deficient, C, 261630
RAB18	99,50%	97,40%	100%	100%	Warburg micro syndrome 3, 614222
RAB3GAP1	99,40%	98,90%	99,40%	99,40%	Warburg micro syndrome 1, 600118
RAB3GAP2	99,50%	97,00%	100%	100%	Warburg micro syndrome 2, 614225 Martsolf syndrome, 212720

RAD51	89,40%	89,40%	89,40%	89,40%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508
RARS	94,20%	91,60%	94,40%	94,30%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	100%	99,80%	100%	100%	Pontocerebellar hypoplasia, type 6, 611523
REEP1	78,70%	76,10%	100%	100%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
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
RNF170	99,60%	97,60%	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF216	99,80%	98,70%	100%	100%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	100%	99,20%	100%	100%	Spastic paraplegia 12, autosomal dominant, 604805
RUBCN	99,40%	97,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	100%	100%	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	100%	100%	100%	100%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCN11A	99,80%	98,30%	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
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
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
SEPSECS	100%	100%	100%	100%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	99,90%	99,50%	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	100%	99,80%	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	98,70%	94,00%	95,20%	95,20%	Dystonia-11, myoclonic, 159900
SIL1	99,20%	96,70%	100%	100%	Marinesco-Sjogren syndrome, 248800
SLC12A6	100%	100%	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	99,20%	93,70%	100%	100%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	100%	99,80%	98,70%	98,70%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	100%	99,90%	100%	100%	Episodic ataxia, type 6, 612656

SLC20A2	100%	99,20%	100%	100%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	99,80%	98,10%	100%	100%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
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
SLC30A10	100%	100%	100%	100%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	99,90%	98,90%	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC39A14	100%	99,40%	93,50%	93,50%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC52A2	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	100%	100%	100%	100%	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500
SLC6A3	100%	100%	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
SLC9A1	100%	100%	100%	100%	?Lichtenstein-Knorr syndrome, 616291
SMPD1	100%	100%	100%	100%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	100%	100%	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNORD118	NC	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX14	99,60%	95,90%	100%	100%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SPART	99,70%	96,80%	100%	100%	Troyer syndrome, 275900
SPAST	99,80%	98,70%	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	100%	99,30%	100%	100%	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SPG21	99,40%	96,80%	100%	100%	Mast syndrome, 248900
SPG7	94,90%	92,60%	100%	100%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	99,80%	96,30%	100%	100%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	100%	99,30%	99,90%	99,90%	Spinocerebellar ataxia, autosomal recessive 14, 615386 Spinocerebellar ataxia 5, 600224
STUB1	100%	98,70%	100%	100%	Spinocerebellar ataxia, autosomal recessive 16, 615768 ?Spinocerebellar ataxia 48, 618093

SUMF1	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
SUOX	100%	100%	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	98,30%	98,00%	98,80%	98,80%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TBCD	96,20%	94,40%	100%	100%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TDP1	99,90%	99,50%	100%	100%	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	100%	99,40%	100%	100%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	100%	100%	100%	100%	Spastic paraplegia 49, autosomal recessive, 615031
TENM4	100%	99,60%	100%	100%	Essential tremor, hereditary, 5, 616736
TGM6	99,70%	97,30%	100%	100%	Spinocerebellar ataxia 35, 613908
TH	99,30%	96,10%	100%	100%	Segawa syndrome, recessive, 605407
THAP1	100%	100%	100%	100%	Dystonia 6, torsion, 602629
TIMM8A	98,00%	90,10%	100%	100%	Mohr-Tranebjaerg syndrome, 304700
TMEM106B	99,90%	98,80%	100%	100%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM240	100%	100%	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TOE1	100%	100%	100%	100%	Pontocerebellar hypoplasia, type 7, 614969
TOR1A	100%	99,90%	100%	100%	Dystonia-1, torsion, 128100
TPP1	100%	100%	100%	100%	Spinocerebellar ataxia, autosomal recessive 7, 609270 Ceroid lipofuscinosis, neuronal, 2, 204500
TREM2	100%	99,80%	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
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
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
TTBK2	99,80%	97,60%	100%	100%	Spinocerebellar ataxia 11, 604432
TTC19	81,50%	73,80%	100%	99,20%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	94,70%	87,10%	100%	100%	Ataxia with isolated vitamin E deficiency, 277460

TUBA1A	99,90%	97,00%	100%	100%	Lissencephaly 3, 611603
TUBB	97,30%	93,90%	99,80%	99,80%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
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
TWNK	100%	100%	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TYROBP	100%	100%	100%	100%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
UBAP1	98,80%	93,40%	100%	100%	Spastic paraplegia 80, autosomal dominant, 618418
UBTF	100%	99,40%	100%	100%	Neurodegeneration, childhood-onset, with brain atrophy, 617672
VAMP1	100%	100%	100%	100%	Spastic ataxia 1, autosomal dominant, 108600 Myasthenic syndrome, congenital, 25, 618323
VARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 20, 615917
VCP	100%	99,20%	100%	100%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VLDLR	100%	99,80%	100%	100%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	99,40%	95,60%	100%	100%	Choreoacanthocytosis, 200150
VPS13D	100%	99,70%	100%	100%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS16	100%	100%	100%	100%	No OMIM disease ID
VPS37A	91,30%	78,20%	100%	100%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	91,50%	90,70%	100%	99,30%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	99,70%	98,50%	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
WASHC5	100%	99,80%	100%	100%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
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
WDR73	100%	100%	100%	100%	Galloway-Mowat syndrome 1, 251300
WDR81	100%	100%	100%	100%	Hydrocephalus, congenital, 3, with brain anomalies, 617967 Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
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
XPR1	100%	99,90%	100%	100%	Basal ganglia calcification, idiopathic, 6, 616413

XRCC1	100%	98,80%	100%	100%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
ZC4H2	100%	99,00%	100%	100%	Wieacker-Wolff syndrome, 314580 Wieacker-Wolff syndrome, female-restricted, 301041
ZFYVE26	100%	99,10%	100%	100%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	100%	100%	100%	100%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	100%	99,60%	100%	100%	No OMIM disease ID

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