MOVEMENT DISORDERS GENE PANEL DG 2.16 (310 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	122,7	100.0%	99.8%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCB7	126,2	99.9%	98.6%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	87,4	77.2%	75.0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	93,1	100.0%	98.9%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCY5	131,8	97.8%	94.7%	Dyskinesia, familial, with facial myokymia, 606703
ADGRG1	147,2	100.0%	100.0%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
AFG3L2	98,3	95.9%	86.1%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AGTPBP1	116,9	98.7%	95.1%	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276
AIMP1	79,4	99.1%	92.4%	Leukodystrophy, hypomyelinating, 3, 260600
ALDH18A1	113,7	100.0%	99.8%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALS2	145,1	100.0%	99.8%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
AMPD2	132,3	100.0%	99.9%	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
ANO10	106	98.9%	96.3%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	118,3	99.5%	97.7%	Dystonia 24, 615034
AP4B1	121	99.9%	98.4%	Spastic paraplegia 47, autosomal recessive, 614066

AP4E1	106,6	99.8%	98.8%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	129,3	99.7%	98.1%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	66,2	78.5%	71.3%	Spastic paraplegia 52, autosomal recessive, 614067
APTX	96,3	94.1%	91.3%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	159,1	100.0%	100.0%	Argininemia, 207800
ARSA	138,5	100.0%	100.0%	Metachromatic leukodystrophy, 250100
ARX	49,3	87.3%	79.2%	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004
ASPA	116,1	99.7%	96.9%	Canavan disease, 271900
ATCAY	152,7	100.0%	99.7%	Ataxia, cerebellar, Cayman type, 601238
ATL1	134,7	99.9%	99.0%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATM	110,9	99.6%	97.2%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic, 0 Lymphoma, mantle cell, somatic, 0 T-cell prolymphocytic leukemia, somatic, 0 {Breast cancer, susceptibility to}, 114480
ATP13A2	134,1	99.9%	99.7%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A2	161,7	100.0%	99.5%	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	159,8	100.0%	100.0%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2B3	123	99.6%	97.5%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	128,7	99.9%	99.1%	Wilson disease, 277900
B4GALNT1	151,2	99.8%	97.9%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	73,4	93.2%	78.3%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	176,9	100.0%	99.8%	Maple syrup urine disease, type Ia, 248600
BCKDHB	123,3	98.6%	92.8%	Maple syrup urine disease, type Ib, 248600
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700

				Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
C12orf65	110,4	100.0%	99.6%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CA8	107,5	99.7%	97.6%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	92,4	97.8%	94.7%	Epileptic encephalopathy, early infantile, 42, 617106 Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1E	120,9	99.8%	99.2%	Epileptic encephalopathy, early infantile, 69, 618285
CACNA1G	148,6	100.0%	99.8%	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087
CACNB4	97,8	97.2%	95.5%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CAMTA1	179,5	100.0%	99.7%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN1	162,5	100.0%	100.0%	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	117,9	99.9%	98.9%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHMP1A	123,2	100.0%	99.8%	Pontocerebellar hypoplasia, type 8, 614961
CLCN2	115,1	100.0%	99.7%	Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651 {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628
CLCN4	105,7	99.9%	98.9%	Raynaud-Claes syndrome, 300114
CLPB	125,6	99.8%	97.9%	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COASY	172,7	100.0%	100.0%	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266
COL4A1	95,7	99.6%	97.3%	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COQ2	103,5	97.6%	97.1%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500

COQ4	105	91.3%	90.2%	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	161,8	100.0%	99.9%	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	73,8	100.0%	98.1%	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	66,2	96.4%	85.3%	Mitochondrial complex IV deficiency, 220110
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CSF1R	113,3	99.9%	99.1%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSTB	70	99.3%	90.9%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	173	100.0%	99.7%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	134,3	98.4%	95.5%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	103,8	99.6%	96.6%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	109,9	99.6%	96.9%	Maple syrup urine disease, type II, 248600
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DCC	118,6	100.0%	99.8%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600
DCTN1	112,6	99.9%	99.2%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DDC	97,9	99.5%	95.0%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	161,6	99.9%	98.4%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	129,6	100.0%	99.5%	Spastic paraplegia 54, autosomal recessive, 615033
DHDDS	81	97.1%	93.8%	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DLAT	100,2	99.8%	99.2%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	119,2	99.9%	99.7%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAJC3	137,4	100.0%	99.7%	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAL4	69,9	99.9%	95.3%	?Mirror movements 3, 616059
DNMT1	114,3	99.2%	98.7%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DPYS	117,5	100.0%	99.8%	Dihydropyrimidinuria, 222748
ECHS1	103,8	100.0%	99.7%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2B1	121,7	100.0%	99.9%	Leukoencephalopathy with vanishing white matter, 603896

EIF2B2	109,7	99.4%	92.4%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	134,7	100.0%	100.0%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	122	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	103,1	100.0%	99.6%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELOVL5	105,7	100.0%	99.1%	Spinocerebellar ataxia 38, 615957
ERLIN2	115,3	100.0%	99.2%	Spastic paraplegia 18, autosomal recessive, 611225
EXOSC3	125,1	96.4%	87.8%	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	92,7	98.8%	92.5%	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	125,4	100.0%	99.4%	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	73,7	97.2%	91.8%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	161,9	100.0%	100.0%	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FBXO7	152,8	99.9%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FGF14	214,2	100.0%	100.0%	Spinocerebellar ataxia 27, 609307
FLVCR1	146,1	99.9%	99.2%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	107,4	100.0%	99.9%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	101	99.8%	97.8%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GALC	102,9	99.8%	98.8%	Krabbe disease, 245200
GAN	142,2	99.9%	99.4%	Giant axonal neuropathy-1, 256850
GBA	169,8	100.0%	100.0%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GBA2	141,5	100.0%	99.6%	Spastic paraplegia 46, autosomal recessive, 614409

GCDH	145,9	100.0%	99.1%	Glutaricaciduria, type I, 231670
GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GDAP2	118,2	99.9%	99.3%	Spinocerebellar ataxia, autosomal recessive 27, 618369
GFAP	103,7	91.9%	91.4%	Alexander disease, 203450
GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	130,2	99.4%	96.5%	Dystonia 25, 615073
GOSR2	102,6	95.8%	93.7%	Epilepsy, progressive myoclonic 6, 614018
GPR143	59,5	91.0%	79.1%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GRID2	146,8	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIN1	166,1	100.0%	99.9%	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820
GRIN2B	158	99.8%	99.0%	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRM1	156,6	100.0%	99.9%	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
HACE1	136,2	99.9%	99.1%	Spastic paraplegia and psychomotor retardation with or without seizures, 616756
HEXB	163	99.7%	98.5%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	59,8	98.3%	88.2%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD17B4	109,4	96.3%	93.6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPD1	74,3	98.1%	92.5%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
IBA57	137,4	99.3%	95.9%	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ISCA2	105,1	99.8%	95.8%	Multiple mitochondrial dysfunctions syndrome 4, 616370
ITPR1	131,2	100.0%	99.7%	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360

KATNB1	154,2	100.0%	100.0%	Lissencephaly 6, with microcephaly, 616212
KCNA1	150,1	100.0%	100.0%	Episodic ataxia/myokymia syndrome, 160120
KCNA2	126,3	100.0%	99.7%	Epileptic encephalopathy, early infantile, 32, 616366
KCNC1	170,9	100.0%	100.0%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	112,7	90.4%	72.9%	Spinocerebellar ataxia 13, 605259
KCND3	162	99.9%	99.2%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNJ10	148,6	89.2%	88.1%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	157,2	100.0%	99.9%	Keppen-Lubinsky syndrome, 614098
KCNMA1	102,3	94.8%	93.4%	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCTD7	154,9	95.0%	95.0%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIDINS220	137,5	100.0%	99.8%	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296
KIF1A	115	99.7%	97.6%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	147,7	100.0%	99.4%	Spastic ataxia 2, autosomal recessive, 611302
KIF5A	116	100.0%	99.9%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KMT2B	141,1	96.9%	93.5%	Dystonia 28, childhood-onset, 617284
L1CAM	126,6	99.9%	98.6%	Corpus callosum, partial agenesis of, 304100 CRASH syndrome, 303350 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Hydrocephalus with Hirschsprung disease, 307000 MASA syndrome, 303350
LAMA1	116	100.0%	99.5%	Poretti-Boltshauser syndrome, 615960
LAMB1	142,9	100.0%	99.7%	Lissencephaly 5, 615191
LMNB1	118,4	100.0%	99.7%	Leukodystrophy, adult-onset, autosomal dominant, 169500
MARS2	178,4	100.0%	100.0%	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MECP2	124,8	100.0%	98.5%	Encephalopathy, neonatal severe, 300673 Mental retardation, X-linked syndromic, Lubs type, 300260 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750

				Rett syndrome, preserved speech variant, 312750 {Autism susceptibility, X-linked 3}, 300496
MECR	108,2	100.0%	99.7%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MICU1	103,3	98.8%	96.5%	Myopathy with extrapyramidal signs, 615673
MLC1	96,7	100.0%	99.9%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMADHC	81,2	92.7%	79.5%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MTPAP	122,3	99.0%	93.2%	?Spastic ataxia 4, autosomal recessive, 613672
MTTP	114,7	99.9%	99.4%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
NANS	97,2	99.9%	98.4%	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NEFL	178,7	99.8%	97.8%	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEU1	141,3	99.3%	96.4%	Sialidosis, type I, 256550 Sialidosis, type II, 256550
NEXMIF	132	100.0%	99.5%	Mental retardation, X-linked 98, 300912
NF2	94,2	100.0%	99.6%	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NIPA1	156,7	100.0%	99.9%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	88,8	100.0%	99.7%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NKX6-2	122,9	98.1%	91.2%	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NOL3	112,8	99.6%	96.3%	?Myoclonus, familial, 1, 614937
NPC1	117,8	100.0%	99.2%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	124,7	100.0%	99.9%	Niemann-pick disease, type C2, 607625
NT5C2	121,2	97.9%	96.3%	Spastic paraplegia 45, autosomal recessive, 613162

NUP62	111,8	100.0%	100.0%	Striatonigral degeneration, infantile, 271930
OCLN	173,9	100.0%	100.0%	Pseudo-TORCH syndrome 1, 251290
OPA1	124,7	99.7%	97.4%	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Behr syndrome, 210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 {Glaucoma, normal tension, susceptibility to}, 606657
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX6	116,5	100.0%	99.8%	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PDE10A	107,6	81.4%	80.4%	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE8B	99,7	99.9%	98.9%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	115,4	100.0%	100.0%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	126,6	99.7%	98.0%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PDHA1	85,3	98.9%	95.4%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	129	99.9%	99.5%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	104,8	96.7%	87.7%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	112,9	99.6%	96.1%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	121,8	100.0%	100.0%	Spinocerebellar ataxia 23, 610245
PEX10	113,3	99.9%	97.4%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX2	134,9	100.0%	100.0%	Peroxisome biogenesis disorder 5A (Zellweger), 614866

				Peroxisome biogenesis disorder 5B, 614867
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	74	99.9%	96.9%	Refsum disease, 266500
PIK3R5	120,7	100.0%	99.9%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLP1	112,8	99.7%	97.7%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	127,7	100.0%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	108,1	99.1%	95.9%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	126,8	100.0%	99.8%	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	109	100.0%	99.9%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPLA6	137,9	99.9%	99.5%	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POLG	113,9	100.0%	99.6%	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR1C	98,3	98.9%	94.9%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRF1	138,1	91.2%	90.6%	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRICKLE1	100	100.0%	99.8%	Epilepsy, progressive myoclonic 1B, 612437
PRKCG	129,8	100.0%	99.3%	Spinocerebellar ataxia 14, 605361
PRKRA	190,7	100.0%	100.0%	Dystonia 16, 612067
PRRT2	111,8	100.0%	99.0%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751

PSAP	98,1	100.0%	99.3%	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PUM1	126,9	100.0%	99.5%	Spinocerebellar ataxia 47, 617931
PYCR2	116,5	99.7%	96.9%	Leukodystrophy, hypomyelinating, 10, 616420
RAB18	83,4	99.7%	97.2%	Warburg micro syndrome 3, 614222
RAB3GAP1	121,7	99.4%	98.9%	Warburg micro syndrome 1, 600118
RAB3GAP2	91,6	99.7%	96.9%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAD51	100,6	89.4%	89.4%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RARS	93,4	93.6%	90.0%	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	104	100.0%	99.4%	Pontocerebellar hypoplasia, type 6, 611523
REEP1	71,5	78.6%	76.2%	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNF170	126,6	99.7%	97.7%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF216	125	99.9%	98.1%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	140,8	99.9%	98.7%	Spastic paraplegia 12, autosomal dominant, 604805
RUBCN	99,3	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	150,4	100.0%	99.9%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	171,8	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCN11A	122,1	99.3%	97.1%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN8A	154,3	100.0%	99.7%	?Myoclonus, familial, 2, 618364 Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SEPSECS	159,6	100.0%	99.6%	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	111	99.7%	99.0%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	151,6	100.0%	99.6%	Amyotrophic lateral sclerosis 4, juvenile, 602433

				Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002
SGCE	97,7	96.0%	92.3%	Dystonia-11, myoclonic, 159900
SIL1	129,5	98.9%	96.2%	Marinesco-Sjogren syndrome, 248800
SLC12A6	118,9	100.0%	99.9%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	63,3	98.7%	91.0%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	134,6	100.0%	99.9%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	99,5	100.0%	99.8%	Episodic ataxia, type 6, 612656
SLC20A2	108,6	100.0%	98.5%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	146,8	97.9%	93.6%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC2A1	148,9	92.8%	92.8%	Dystonia 9, 601042 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847
SLC30A10	176,1	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	132	99.7%	97.7%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC52A2	185,4	100.0%	100.0%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	118,8	100.0%	99.8%	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC6A3	133	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC9A1	142,4	100.0%	100.0%	?Lichtenstein-Knorr syndrome, 616291
SMPD1	146,4	100.0%	99.2%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	105	100.0%	100.0%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNORD118	NC	NC	NC	Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX14	84,1	99.0%	95.4%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPART	132,6	99.8%	98.2%	Troyer syndrome, 275900
SPAST	95,4	99.8%	97.7%	Spastic paraplegia 4, autosomal dominant, 182601

SPG11	116,1	99.7%	98.4%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	120,6	99.7%	96.8%	Mast syndrome, 248900
SPG7	115,2	99.3%	96.4%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	145,7	100.0%	99.8%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	126,2	100.0%	99.7%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	173,9	100.0%	99.5%	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia, autosomal recessive 16, 615768
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
SUOX	167,2	100.0%	100.0%	Sulfite oxidase deficiency, 272300
SYNE1	121,6	98.3%	97.8%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TAF1	86,8	99.1%	95.5%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TANGO2	127,3	100.0%	100.0%	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D20	115,7	96.3%	93.8%	Warburg micro syndrome 4, 615663
TBC1D23	92,7	99.2%	95.4%	Pontocerebellar hypoplasia, type 11, 617695
TBCD	136,2	98.2%	94.3%	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193
TDP1	103,9	99.9%	99.5%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250
TDP2	173	99.9%	99.4%	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	137,2	100.0%	100.0%	Spastic paraplegia 49, autosomal recessive, 615031
TENM4	119,9	100.0%	99.6%	Essential tremor, hereditary, 5, 616736
TGM6	130,2	99.9%	98.7%	Spinocerebellar ataxia 35, 613908
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
THAP1	141,3	100.0%	100.0%	Dystonia 6, torsion, 602629
TIMM8A	46,3	94.6%	79.9%	Mohr-Tranebjaerg syndrome, 304700
TMEM106B	121,4	99.7%	98.9%	Leukodystrophy, hypomyelinating, 16, 617964
TMEM240	163,9	100.0%	100.0%	Spinocerebellar ataxia 21, 607454
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991

TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TOR1A	142,4	100.0%	100.0%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}, 0
TPP1	123,7	100.0%	99.9%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREM2	127	100.0%	99.9%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TSEN2	95,6	99.9%	98.9%	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	114,4	99.4%	96.8%	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TTBK2	108,4	99.9%	96.8%	Spinocerebellar ataxia 11, 604432
TTC19	83,4	97.0%	82.6%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	109,2	97.6%	92.5%	Ataxia with isolated vitamin E deficiency, 277460
TUBA1A	77,6	99.8%	97.1%	Lissencephaly 3, 611603
TUBB4A	101,2	97.1%	95.6%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	154,3	100.0%	100.0%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TWNK	159,6	100.0%	100.0%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
TYROBP	83,3	100.0%	100.0%	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770
VAMP1	135,4	100.0%	100.0%	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, autosomal dominant, 108600
VCP	100,3	100.0%	99.2%	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687 Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320
VLDLR	141,4	100.0%	99.9%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	78,2	99.2%	95.3%	Choreoacanthocytosis, 200150
VPS13D	138	100.0%	99.7%	Spinocerebellar ataxia, autosomal recessive 4, 607317
VPS37A	64,3	91.3%	79.3%	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	111,3	91.1%	89.6%	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	129,6	99.8%	98.7%	Pontocerebellar hypoplasia type 1A, 607596
WASHC5	134,2	99.9%	99.5%	Ritscher-Schinzel syndrome 1, 220210

				Spastic paraplegia 8, autosomal dominant, 603563
WDR26	98,2	99.5%	97.4%	Skraban-Deardorff syndrome, 617616
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
WDR73	153,2	100.0%	99.9%	Galloway-Mowat syndrome 1, 251300
WDR81	184,8	100.0%	100.0%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WWOX	116,1	100.0%	99.9%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	85,4	100.0%	99.4%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPR1	126	100.0%	99.6%	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	111,4	99.9%	99.1%	?Spinocerebellar ataxia, autosomal recessive 26, 617633
ZC4H2	72,4	99.8%	95.9%	Wieacker-Wolff syndrome, 314580
ZFYVE26	104,8	99.9%	98.7%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	110,4	100.0%	100.0%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	142,8	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 5, 251300

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors