MOVEMENT DISORDERS GENE PANEL DG 2.14 (304 genes)

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS2	126	100	99.3	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCB7	132	99.9	98.4	Anemia, sideroblastic, with ataxia, 301310
ABCD1	76	74.7	68	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	107	97.3	88	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	129	99.1	94.2	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADAR	125	100	99.8	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCY5	129	92.4	89.2	Dyskinesia, familial, with facial myokymia, 606703
ADGRG1	150	100	100	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
AFG3L2	121	91.9	84.9	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
AIMP1	84.8	97.3	89.7	Leukodystrophy, hypomyelinating, 3, 260600
ALDH18A1	131	100	99.9	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	126	95.3	94.6	Sjogren-Larsson syndrome, 270200
ALS2	170	99.9	99.2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
AMPD2	136	99.9	99.2	?Spastic paraplegia 63, 615686 Pontocerebellar hypoplasia, type 9, 615809
ANO10	117	98.8	96.5	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	139	99.7	98.2	Dystonia 24, 615034
AP4B1	147	100	99.8	Spastic paraplegia 47, autosomal recessive, 614066

AP4E1	98.7	99.7	97.9	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	127	99.1	96.4	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	65.8	71.8	69.3	Spastic paraplegia 52, autosomal recessive, 614067
APTX	119	94.2	91.1	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	168	100	100	Argininemia, 207800
ARSA	97.8	100	99.7	Metachromatic leukodystrophy, 250100
ARX	29.1	75.8	59.5	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	128	99.1	95.8	Canavan disease, 271900
ATCAY	146	100	99.7	Ataxia, cerebellar, Cayman type, 601238
ATL1	161	99.7	97.9	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATM	110	99	94	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	117	100	98.8	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A2	191	100	99.6	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	177	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2B3	135	99.5	97.7	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	169	100	99.8	Wilson disease, 277900
B4GALNT1	151	95.6	90.1	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	70.6	93.1	82.5	Deafness, dystonia, and cerebral hypomyelination, 300475

BCKDHA	172	100	99.5	Maple syrup urine disease, type Ia, 248600
BCKDHB	113	88.9	81.3	Maple syrup urine disease, type Ib, 248600
BSCL2	114	100	100	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	88.2	97.3	91.9	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	93.9	100	99.7	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CA8	115	96.8	93	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	87.8	92.7	89.1	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	139	99.8	99.3	No OMIM phenotype ?Epileptic encephalopathy with infantile spasms (Helbig (2016) Genet Med Epub,Epub) ?Autism (O'Roak (2012) Nature 485,246)
CACNA1G	133	99	97.5	Spinocerebellar ataxia 42, 616795
CACNB4	106	96.3	94.6	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CAMTA1	186	99.6	98.8	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN1	146	100	100	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	165	99.9	99.1	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CHMP1A	134	100	100	Pontocerebellar hypoplasia, type 8, 614961
CIZ1	153	98.9	95.4	No OMIM phenotype Cervical dystonia, primary (Xiao (2012) Ann Neurol 71, 458)
CLCN2	109	100	99.4	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

CLPB	140	100	99.5	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
COASY	169	100	100	Neurodegeneration with brain iron accumulation 6, 615643
COL4A1	92.8	97.9	94	?Retinal arteries, tortuosity of, 180000 Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 Schizencephaly, 269160 {Hemorrhage, intracerebral, susceptibility to}, 614519
COQ2	89.3	96.1	93.2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ4	89.8	88.4	84.9	Coenzyme Q10 deficiency, primary, 7, 616276
COQ8A	134	100	99.1	Coenzyme Q10 deficiency, primary, 4, 612016
COQ9	91.4	99.9	96.6	Coenzyme Q10 deficiency, primary, 5, 614654
COX20	58.1	83	65.4	Mitochondrial complex IV deficiency, 220110
CP	120	93.9	89.6	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CSF1R	140	99.5	98.4	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSTB	82.5	97.1	82.7	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	175	98.3	96.1	Cerebrotendinous xanthomatosis, 213700
CYP2U1	119	93.7	91.2	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	93.2	94.7	87.7	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	102	97.3	93.8	Maple syrup urine disease, type II, 248600
DCAF17	91.9	95.6	89.3	Woodhouse-Sakati syndrome, 241080
DCC	139	100	99.9	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	132	99.7	98.3	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DDC	101	99.1	95	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	142	97.1	94.8	Spastic paraplegia 28, autosomal recessive, 609340

DDHD2	150	99.9	98	Spastic paraplegia 54, autosomal recessive, 615033
DHDDS	93.5	97.8	94.8	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861
DLAT	91.6	99.1	96	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	124	99.9	98.6	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAJC3	116	99.9	98.1	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192
DNAL4	72.6	99.3	93.5	?Mirror movements 3, 616059
DNMT1	113	99.2	98.3	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
DPYS	134	100	99.5	Dihydropyrimidinuria, 222748
ECHS1	113	99.8	97.8	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2B1	150	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	132	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	164	100	100	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	146	100	99.5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	128	99.6	97.9	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
ELOVL4	91.9	99.9	98	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ELOVL5	121	100	99.8	Spinocerebellar ataxia 38, 615957
ERLIN2	156	100	99.3	Spastic paraplegia 18, autosomal recessive, 611225
EXOSC3	88.5	97.3	89.4	Pontocerebellar hypoplasia, type 1B, 614678
FA2H	94.1	87.9	79.9	Spastic paraplegia 35, autosomal recessive, 612319
FAM126A	125	97.3	95.2	Leukodystrophy, hypomyelinating, 5, 610532
FAR1	80.4	96.3	92.4	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FARS2	208	100	100	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, autosomal recessive, 617046
FBXO7	189	98.5	93.3	Parkinson disease 15, autosomal recessive, 260300
FGF14	190	100	99.7	Spinocerebellar ataxia 27, 609307
FLVCR1	140	99.2	95.8	Ataxia, posterior column, with retinitis pigmentosa, 609033

FOLR1	150	100	100	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	114	99.9	98.8	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	148	99	93.2	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GALC	101	98.9	94.6	Krabbe disease, 245200
GAN	190	100	99.9	Giant axonal neuropathy-1, 256850
GBA	240	100	100	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	176	99.9	99.3	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	148	99.9	99.1	Glutaricaciduria, type I, 231670
GCH1	74.4	97	86.5	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GDAP2	131	99.7	97.2	No OMIM phenotype
GFAP	103	91.7	90.3	Alexander disease, 203450
GJC2	41.9	68.9	58.6	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLB1	94.3	99.6	97	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	135	94.5	91.6	Dystonia 25, 615073
GOSR2	127	95.9	95	Epilepsy, progressive myoclonic 6, 614018
GPR143	61.5	85.3	75.5	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GRID2	175	100	99.9	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRIN1	151	100	99.5	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	189	99.9	99.3	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation, autosomal dominant 6, 613970
GRM1	186	100	99.9	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	130	99.4	94	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	58.2	96	84.8	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSD17B4	95.1	93.9	90.8	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSPD1	96.5	98.3	93.2	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
IBA57	113	93.3	89.5	?Spastic paraplegia 74, autosomal recessive, 616451 Multiple mitochondrial dysfunctions syndrome 3, 615330
ITPR1	161	100	99.9	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KATNB1	142	100	100	Lissencephaly 6, with microcephaly, 616212
KCNA1	167	100	99.7	Episodic ataxia/myokymia syndrome, 160120
KCNA2	158	100	99.9	Epileptic encephalopathy, early infantile, 32, 616366
KCNC1	199	100	100	Epilepsy, progressive myoclonic 7, 616187
KCNC3	144	68.5	59	Spinocerebellar ataxia 13, 605259
KCND3	183	99.9	99.1	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNJ10	213	89.3	89.1	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	157	100	99.9	Keppen-Lubinsky syndrome, 614098
KCNMA1	120	94.4	93.2	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446
KCTD7	167	95	95	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIDINS220	155	99.9	99.5	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296

KIF1A	114	99.2	96.1	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	121	99.9	99.1	Spastic ataxia 2, autosomal recessive, 611302
KIF5A	136	100	99.9	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
KMT2B	120	94	91.2	Dystonia 28, childhood-onset, 617284
L1CAM	133	99.8	98.4	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	138	100	99.6	Poretti-Boltshauser syndrome, 615960
LAMB1	170	100	99.6	Lissencephaly 5, 615191
LMNB1	123	99.9	99.1	Leukodystrophy, adult-onset, autosomal dominant, 169500
MARS2	173	100	100	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, autosomal recessive, 611390
MECP2	87.3	99.1	93.1	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	98.8	96.1	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MICU1	134	96	88.8	Myopathy with extrapyramidal signs, 615673
MLC1	103	100	99.8	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMADHC	77	89.3	75	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MRE11	51.2	95.3	82.3	Ataxia-telangiectasia-like disorder 1, 604391

MTHFR	126	98.4	97.2	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	110	98.9	93.5	?Spastic ataxia 4, autosomal recessive, 613672
MTTP	132	99.9	98.8	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
NANS	106	100	99.9	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442
NEFL	165	99.7	98.1	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684
NEXMIF	139	99.9	99	Mental retardation, X-linked 98, 300912
NF2	100	100	99.9	Meningioma, NF2-related, somatic, 607174 Neurofibromatosis, type 2, 101000 Schwannomatosis, somatic, 162091
NIPA1	174	99.9	99.1	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	52	96.6	83.3	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550
NKX6-2	52	79	74.5	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560
NOL3	76.3	93.5	83.9	Myoclonus, familial cortical, 614937
NPC1	148	99.2	97.8	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	141	100	99.9	Niemann-pick disease, type C2, 607625
NT5C2	125	97.1	92.7	Spastic paraplegia 45, autosomal recessive, 613162
NUP62	112	100	99.9	Striatonigral degeneration, infantile, 271930
OCLN	221	100	100	Pseudo-TORCH syndrome 1, 251290
OPA1	123	99.1	94.1	?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	147	99.3	93.1	HARP syndrome, 607236

				Neurodegeneration with brain iron accumulation 1, 234200
PAX6	120	100	99.9	?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	120	81.2	80.8	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, autosomal dominant, 616922
PDE8B	111	99.9	98.9	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	95.1	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	147	99.1	96.5	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	110	98.1	92.1	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	133	98.9	94.6	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	117	88.8	78.7	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	127	96.5	93.5	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	107	100	99.9	Spinocerebellar ataxia 23, 610245
PEX10	112	96.1	90.1	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX2	147	100	100	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	114	89.6	82	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	74.6	97.5	90.8	Refsum disease, 266500

PIK3R5	110	100	99.8	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	118	99.9	98.4	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLP1	129	100	99.4	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	141	99.9	99.4	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	121	99.4	96.8	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	99.8	100	99.2	Paroxysmal nonkinesigenic dyskinesia 1, 118800
PNKP	93	99.8	97.7	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPLA6	122	99.7	98.5	?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020
POLG	114	100	99.5	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	117	99.7	96.1	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR3A	137	100	99.9	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	146	99.9	98.5	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRF1	123	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
PRICKLE1	117	100	100	Epilepsy, progressive myoclonic 1B, 612437
PRKCG	116	99	94.5	Spinocerebellar ataxia 14, 605361
PRKRA	180	99.8	98.4	Dystonia 16, 612067
PRRT2	78.9	99.9	98.4	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200

				Seizures, benign familial infantile, 2, 605751
PSAP	114	99.9	99	Combined SAP deficiency, 611721 Gaucher disease, atypical, 610539 Krabbe disease, atypical, 611722 Metachromatic leukodystrophy due to SAP-b deficiency, 249900
PUM1	158	100	99.9	Spinocerebellar ataxia 47, 617931
PYCR2	128	100	97.6	Leukodystrophy, hypomyelinating, 10, 616420
RAB18	82.7	97.1	86.4	Warburg micro syndrome 3, 614222
RAB3GAP1	124	99.4	98.8	Warburg micro syndrome 1, 600118
RAB3GAP2	94.1	98.4	93.9	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAD51	123	89.4	89.4	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RARS	86.4	92.7	85.9	Leukodystrophy, hypomyelinating, 9, 616140
RARS2	107	100	99.1	Pontocerebellar hypoplasia, type 6, 611523
REEP1	78.3	76.3	75.7	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, autosomal dominant, 610250
RNASEH2A	142	100	99.9	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	104	93.2	87.5	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	209	100	99.9	Aicardi-Goutieres syndrome 3, 610329
RNF170	147	98.3	91.2	Ataxia, sensory, 1, autosomal dominant, 608984
RNF216	137	99.8	98.6	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	105	99.2	96.7	Spastic paraplegia 12, autosomal dominant, 604805
RUBCN	104	98	97.5	?Spinocerebellar ataxia, autosomal recessive 15, 615705
SACS	155	100	99.7	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMD9L	166	100	99.9	Ataxia-pancytopenia syndrome, 159550
SAMHD1	128	99.6	96.6	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SCN11A	138	99.2	97.6	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548

SCN8A	198	100	99.7	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080
SEPSECS	159	100	100	Pontocerebellar hypoplasia type 2D, 613811
SERAC1	113	98.8	94.6	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	163	99.9	99.1	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SGCE	88.9	93.7	90	Dystonia-11, myoclonic, 159900
SIL1	154	99.8	98	Marinesco-Sjogren syndrome, 248800
SLC12A6	142	100	99.9	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	60.3	92.8	82.1	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	186	100	99.9	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	122	100	99.8	Episodic ataxia, type 6, 612656
SLC20A2	119	99.7	97.3	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	193	98.8	95	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC2A1	190	92.9	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	164	100	100	Hypermanganesemia with dystonia 1, 613280
SLC33A1	141	96.8	90.1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC39A14	108	99.8	97.9	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC52A2	178	100	100	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	120	100	100	?Fazio-Londe disease, 211500 Brown-Vialetto-Van Laere syndrome 1, 211530
SLC6A3	146	100	99.8	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SLC9A1	161	100	100	?Lichtenstein-Knorr syndrome, 616291
SMPD1	124	99.6	97.9	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

SNCA	130	100	100	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNORD118				Leukoencephalopathy, brain calcifications, and cysts, 614561
SNX14	70.1	95.2	82.9	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPART	132	99.8	98.2	Troyer syndrome, 275900
SPAST	63.8	93.1	81.9	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	129	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG21	121	98.6	94.8	Mast syndrome, 248900
SPG7	119	93.3	92.4	Spastic paraplegia 7, autosomal recessive, 607259
SPR	167	98.9	90	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	118	99.9	99.3	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	176	100	98.9	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUMF1	103	98.6	91.1	Multiple sulfatase deficiency, 272200
SUOX	213	100	100	Sulfite oxidase deficiency, 272300
SYNE1	137	98.2	97.6	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TAF1	112	99.4	96.6	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TANGO2	145	100	100	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878
TBC1D20	146	94.2	94.1	Warburg micro syndrome 4, 615663
TBC1D23	86	95.7	91.5	Pontocerebellar hypoplasia, type 11, 617695
TDP1	123	98.7	95.3	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TDP2	165	99.9	98.8	Spinocerebellar ataxia, autosomal recessive 23, 616949
TECPR2	161	100	99.9	Spastic paraplegia 49, autosomal recessive, 615031
TENM4	161	99.9	99.2	Essential tremor, hereditary, 5, 616736
TGM6	150	99.7	98	Spinocerebellar ataxia 35, 613908

TH	68.2	97.6	88.7	Segawa syndrome, recessive, 605407
THAP1	122	100	100	Dystonia 6, torsion, 602629
TIMM8A	46	94.5	78.8	Mohr-Tranebjaerg syndrome, 304700
TMEM106B	120	99.8	96.4	Leukodystrophy, hypomyelinating, 16, 617964
TMEM240	112	99.8	97.4	Spinocerebellar ataxia 21, 607454
TMEM67	72.9	93.3	83.4	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOE1	165	100	100	Pontocerebellar hypoplasia, type 7, 614969
TOR1A	185	100	99.8	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}, 0
TPP1	146	100	100	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREM2	149	99.9	99.6	Nasu-Hakola disease, 221770
TREX1	242	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TSEN2	124	100	99.8	Pontocerebellar hypoplasia type 2B, 612389
TSEN54	82.9	95.9	92.9	?Pontocerebellar hypoplasia type 5, 610204 Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753
TTBK2	123	100	98.9	Spinocerebellar ataxia 11, 604432
TTC19	92.1	80.6	72.5	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	102	83.6	76.6	Ataxia with isolated vitamin E deficiency, 277460
TUBA1A	113	99.9	97.8	Lissencephaly 3, 611603
TUBB4A	121	96	95.3	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	164	100	100	Cortical dysplasia, complex, with other brain malformations 4, 615412
TWNK	179	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, dominant 3, 609286

TYROBP	95.2	100	99.9	Nasu-Hakola disease, 221770
VAMP1	132	100	100	Spastic ataxia 1, autosomal dominant, 108600
VCP	145	99.9	99.5	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	201	99.9	99.4	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	69.5	95.3	85.3	Choreoacanthocytosis, 200150
VPS13D	159	99.9	99.4	No OMIM phenotype Schizophrenia (McCarthy (2014) Mol Psychiatry 19, 652)
VPS37A	73.6	86.6	66.4	Spastic paraplegia 53, autosomal recessive, 614898
VPS53	129	91.4	90.4	Pontocerebellar hypoplasia, type 2E, 615851
VRK1	125	97.5	94.2	Pontocerebellar hypoplasia type 1A, 607596
WASHC5	147	99.6	98.1	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WDR45	75	97.4	90.1	Neurodegeneration with brain iron accumulation 5, 300894
WDR73	139	100	100	Galloway-Mowat syndrome 1, 251300
WDR81	163	99.9	99.4	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967
WWOX	131	100	99.7	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia, autosomal recessive 12, 614322
XK	96.8	99.9	99.1	McLeod syndrome with or without chronic granulomatous disease, 300842
XPR1	132	100	99.8	Basal ganglia calcification, idiopathic, 6, 616413
XRCC1	107	99.7	97.5	?Spinocerebellar ataxia, autosomal recessive 26, 617633
ZC4H2	78.6	99.8	98.1	Wieacker-Wolff syndrome, 314580
ZFYVE26	120	99.9	99.4	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	118	100	100	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	150	100	99.9	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 : September 11th, 2018.

This list is accurate for panel version DG 2.14

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