MOVEMENT DISORDERS GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCB7	60,8	100%	97%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	29,1	70%	56%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	56,7	94%	82%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	56,1	99%	93%	Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ADCK3	98,1	99%	97%	Coenzyme Q10 deficiency, primary, 4, 612016
AFG3L2	69,5	95%	91%	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487
ALDH3A2	101	100%	100%	Sjogren-Larsson syndrome, 270200
ANO10	101,4	100%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	108,2	100%	99%	Dystonia 24, 615034
AP4B1	95,9	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	132,4	100%	100%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	97,3	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	79,4	100%	97%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	71,4	91%	90%	Spastic paraplegia 48, autosomal recessive, 613647
APTX	121	99%	94%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920

ARSA	81,1	95%	93%	Metachromatic leukodystrophy, 250100
ARX	29,5	82%	62%	Epileptic encephalopathy, early infantile, 1, 308350 Lissencephaly, X-linked 2, 300215 Mental retardation, X-linked 29 and others, 300419 Proud syndrome, 300004 Partington syndrome, 309510 Hydranencephaly with abnormal genitalia, 300215
ASPA	110,9	100%	100%	Canavan disease, 271900
ATCAY	100,4	100%	97%	Ataxia, cerebellar, Cayman type, 601238
ATL1	107,1	99%	98%	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708
ATM	115,2	100%	99%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell T-cell prolymphocytic leukemia, somatic
ATP13A2	74,9	96%	91%	Parkinson disease 9, 606693
ATP1A3	102,1	99%	98%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2B3	59,2	97%	90%	Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	115,7	100%	97%	Wilson disease, 277900
B4GALNT1	76	93%	86%	Spastic paraplegia 26, autosomal recessive, 609195
BCKDHA	97,7	99%	96%	Maple syrup urine disease, type Ia, 248600
BCKDHB	88,9	100%	98%	Maple syrup urine disease, type Ib, 248600
BSCL2	102,2	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794

C10orf2	134	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia, autosomal dominant, 3, 609286
C19ORF12	86,3	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298
CA8	80,1	100%	94%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	79	97%	91%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNB4	92,9	100%	97%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CCT5	79,3	91%	82%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	94,1	99%	96%	Dystonia 23, 614860
COQ2	71,7	97%	84%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ9	83,4	90%	84%	Coenzyme Q10 deficiency, primary, 5, 614654
CP	86,8	99%	93%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CSTB	173,8	100%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	112,4	100%	96%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	99,9	99%	94%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	104,2	100%	96%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	108,2	100%	98%	Maple syrup urine disease, type II, 248600
DCAF17	92,3	100%	96%	Woodhouse-Sakati syndrome, 241080

DCTN1	107,9	100%	95%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DDC	87,4	100%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	133	100%	99%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	98,7	100%	99%	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	100,9	100%	100%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	136,8	100%	100%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNMT1	96,4	98%	94%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
EIF2B1	102,6	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	90,8	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	86,1	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	111	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	92,1	100%	97%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	102,5	100%	99%	Parkinson disease 18, 614251
ERLIN2	104,4	99%	95%	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	61,6	89%	73%	Spastic paraplegia 35, autosomal recessive, 612319
FBXO7	149,9	100%	99%	Parkinson disease 15, autosomal recessive, 260300
FGF14	111,5	100%	100%	Spinocerebellar ataxia 27, 609307
FLVCR1	92,4	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033

FTL	84,2	100%	95%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
GALC	90,2	99%	98%	Krabbe disease, 245200
GAN	135	100%	100%	Giant axonal neuropathy-1, 256850
GBA	60,4	59%	57%	Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, perinatal lethal, 608013 {Parkinson disease, late-onset, susceptibility to}, 168600 {Lewy body dementia, susceptibility to}, 127750
GBA2	113,5	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	77	92%	90%	Glutaricaciduria, type I, 231670
GCH1	99,7	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	71,2	100%	98%	Alexander disease, 203450
GJC2	54,1	95%	81%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GLB1	73,5	100%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	89,9	100%	98%	Dystonia 25, 615073
GOSR2	97,6	100%	100%	Epilepsy, progressive myoclonic 6, 614018
GPR56	84,3	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GRM1	138,5	100%	97%	Spinocerebellar ataxia, autosomal recessive 13, 614831

HEXB	115,4	100%	100%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	50	97%	81%	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323
HSPD1	14,4	59%	33%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
ITPR1	105	100%	98%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	113,2	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNC3	83,9	78%	72%	Spinocerebellar ataxia 13, 605259
KCNJ10	158,3	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KIAA0196	97,5	100%	98%	Spastic paraplegia 8, autosomal dominant, 603563
KIF1A	68,2	98%	90%	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255
KIF1C	106,3	97%	94%	Spastic ataxia 2,autosomal recessive, 611302
KIF5A	89,4	99%	95%	Spastic paraplegia 10, autosomal dominant, 604187
L1CAM	62,1	99%	92%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 Corpus callosum, partial agenesis of, 304100

MARS2	151,3	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MECP2	88,3	98%	89%	Rett syndrome, 312750 Mental retardation, X-linked, syndromic 13, 300055 Rett syndrome, preserved speech variant, 312750 Encephalopathy, neonatal severe, 300673 {Autism susceptibility, X-linked 3}, 300496 Angelman syndrome, 105830 Mental retardation, X-linked syndromic, Lubs type, 300260
MMADHC	72,7	100%	99%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410
MRE11A	90,7	100%	100%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	93,4	99%	97%	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050
MTPAP	111,8	91%	91%	Ataxia, spastic, 4, 613672
NIPA1	101,6	90%	82%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	86,5	100%	100%	Goiter, familial, due to TTF-1 defect (1) Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978
NOL3	131,6	100%	100%	Myoclonus, familial cortical, 614937
NPC1	87,2	99%	97%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	69	100%	100%	Niemann-pick disease, type C2, 607625
NUP62	89,1	99%	98%	Striatonigral degeneration, infantile, 271930

OPA1	132,7	98%	98%	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250
PANK2	106,5	100%	94%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PAX6	85,7	100%	100%	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hyperplasia, 136520 Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700
PDE8B	94,7	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDHA1	60,1	100%	88%	Pyruvate dehydrogenase E1-alpha deficiency, 312170 Leigh syndrome, X-linked, 308930
PDSS1	86,5	89%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	85	100%	100%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	137,5	100%	100%	Spinocerebellar ataxia 23, 610245
PEX10	65,6	93%	87%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX7	103,8	97%	91%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	79	100%	100%	Refsum disease, 266500
PIK3R5	75,6	100%	98%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	74,8	98%	90%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953

PLP1	42,1	94%	71%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	92,6	100%	100%	Congenital disorder of glycosylation, type Ia, 212065
PNKD	50,1	98%	81%	Paroxysmal nonkinesigenic dyskinesia 2 (2)
PNPLA6	81,9	100%	96%	Spastic paraplegia 39, autosomal recessive, 612020
POLG	83,7	98%	94%	Progressive external ophthalmoplegia, autosomal recessive, 258450 Progressive external ophthalmoplegia, autosomal dominant, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459
PRKCG	95,4	97%	94%	Spinocerebellar ataxia 14, 605361
PRKRA	117,5	100%	100%	Dystonia 16, 612067
PRRT2	75,7	100%	98%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
PSEN1	100	99%	97%	Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694 Acne inversa, familial, 3, 613737
REEP1	77,9	100%	99%	Spastic paraplegia 31, autosomal dominant, 610250 Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	90,9	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	108,4	100%	99%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	135,2	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF170	110,7	100%	100%	Ataxia, sensory, 1, autosomal dominant, 608984

RTN2	70,8	99%	94%	Spastic paraplegia 12, autosomal dominant, 604805
SACS	152,9	100%	100%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	116,1	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SERAC1	86,5	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	150,1	100%	100%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	86,5	95%	91%	maternally imprinted Dystonia-11, myoclonic, 159900
SIL1	95,3	100%	99%	Marinesco-Sjogren syndrome, 248800
SLC16A2	49,2	99%	87%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	115,9	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	111,3	100%	100%	Episodic ataxia, type 6, 612656
SLC25A15	99,4	86%	85%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC2A1	93,6	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, suscpetibility to, 12}, 614847 Dystonia 9, 601042
SLC30A10	132,7	100%	99%	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280
SLC33A1	90,9	100%	100%	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482
SLC52A2	115,4	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC6A3	73,9	100%	99%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SMPD1	100,1	96%	90%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

SNCA	136,7	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SPAST	102	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	105,2	100%	98%	Spastic paraplegia 11, autosomal recessive, 604360
SPG20	119,1	100%	100%	Troyer syndrome, 275900
SPG21	95,9	100%	100%	Mast syndrome, 248900
SPG7	77,2	97%	86%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	68,9	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	91,9	99%	95%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	95,5	99%	93%	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	163,4	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	105,6	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TAF1	74,2	100%	99%	SVA retrotransposon insertion Dystonia-Parkinsonism, X-linked, 314250
TDP1	105,4	100%	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	106,3	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TGM6	63,5	91%	87%	Spinocerebellar ataxia 35, 613908
TH	83	98%	89%	Segawa syndrome, recessive, 605407
THAP1	131,5	100%	98%	Dystonia 6, torsion, 602629

TIMM8A	30,1	91%	69%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMEM67	115,2	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOR1A	130,2	100%	97%	Dystonia-1, torsion, 128100 Dystonia, early-onset atypical, with myoclonic features {Dystonia-1, modifier of}
TREX1	129,5	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
TTBK2	117	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	72,4	90%	75%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	89,3	99%	86%	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	60,3	87%	81%	?Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
VAMP1	108,3	100%	99%	Spastic Ataxia 1, autosomal dominant, 108600
VCP	106,9	100%	97%	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954
VLDLR	109,1	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	119,6	100%	98%	Choreoacanthocytosis, 200150
VPS37A	73,9	99%	92%	Spastic paraplegia 53, autosomal recessive, 614898
WDR45	43	97%	90%	?Neurodegeneration with brain iron accululation 5, 300894
WDR81	105,4	99%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185

WWOX	104,3	100%	99%	Esophageal squamous cell carcinoma, 133239
ZFYVE26	86,9	97%	93%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	80,7	100%	96%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	103,2	93%	90%	Spinocerebellar ataxia, autosomal recessive 5, 606937

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors