

MOVEMENT DISORDERS GENE PANEL DG 2.4.x

<i>Gene</i>	<i>Median coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM ID</i>
ABCB7	60.8	100%	97%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	30.2	72%	59%	Adrenoleukodystrophy, 300100; Adrenomyeloneuropathy, adult, 300100
ABHD12	55.3	94%	79%	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
ADCY5	85.5	99%	94%	Dyskinesia, familial, with facial myokymia, 606703
AFG3L2	69.5	95%	91%	Spinocerebellar ataxia 28, 610246; Ataxia, spastic, 5, autosomal recessive, 614487
ALDH3A2	101.9	100%	100%	Sjogren-Larsson syndrome, 270200
ALS2	120.9	98%	95%	Amyotrophic lateral sclerosis 2,juvenile,205100 Primary lateral sclerosis, juvenile, 606353 Spastic paraplegia, infantile onset ascending, 607225
ANO10	98.1	100%	97%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	108.3	100%	99%	Dystonia 24, 615034
AP4B1	95.5	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	132.4	100%	100%	Spastic paraplegia 51, autosomal recessive, 613744
AP4M1	96.7	100%	99%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	69.9	91%	86%	Spastic paraplegia 52, autosomal recessive, 614067
AP5Z1	71.7	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	99.3	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 {Breast cancer, susceptibility to}, 114480
ATP13A2	75	96%	91%	Parkinson disease 9, 606693
ATP1A2	96.2	100%	97%	Alternating hemiplegia of childhood, 104290 Migraine,familial basilar and hemiplegic,602481
ATP1A3	101	99%	98%	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820
ATP2B3	59.3	97%	90%	Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	115.7	100%	97%	Wilson disease, 277900
B4GALNT1	83.6	94%	88%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	40.2	73%	66%	Deafness, dystonia and cerebellar hypomyelination, 300475
BCKDHA	101.6	99%	97%	Maple syrup urine disease, type Ia, 248600
BCKDHB	89.7	100%	98%	Maple syrup urine disease, type Ib, 248600
BSCL2	101.5	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type V, 600794
C10orf2	134.1	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia, autosomal dominant, 3, 609286
C12orf65	163.2	100%	100%	Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	86.4	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	77.9	96%	90%	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500
CACNA1G	96.5	99%	96%	No OMIM disease phenotype
CACNB4	91.5	99%	95%	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855
CCT5	78.7	92%	83%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	92.9	99%	96%	Dystonia 23, 614860

COASY	116.7	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COQ2	69.8	94%	81%	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	108.4	100%	98%	Maple syrup urine disease, type II, 248600
DCAF17	92.3	100%	96%	Woodhouse-Sakati syndrome, 241080
DCTN1	107.7	100%	95%	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605
DDC	86.6	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	96.2	98%	94%	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121
EIF2B1	106.5	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	90.8	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	85.2	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	111	100%	100%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B5	92	100%	97%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	102.5	100%	99%	Parkinson disease 18, 614251
ELOVL5	87.6	99%	96%	Spinocerebellar ataxia 38, 615957

ERLIN2	105.5	99%	96%	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	61.6	89%	73%	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	104.1	99%	95%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBXO7	149.9	100%	99%	Parkinson disease 15, autosomal recessive, 260300
FGF14	108.2	100%	100%	Spinocerebellar ataxia 27, 609307
FLVCR1	92.4	100%	100%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	78	99%	96%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FTL	84.2	100%	95%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
GALC	92.9	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
GBA2	118.5	100%	100%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	70.9	91%	86%	Glutaricaciduria, type I, 231670
GCH1	98.2	100%	100%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	73.7	100%	97%	Alexander disease, 203450
GJC2	54.1	95%	81%	Leukodystrophy, hypomyelinating, 2, 608804 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480
GLB1	75.6	100%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	91.5	100%	98%	Dystonia 25, 615073
GOSR2	93.4	97%	91%	Epilepsy, progressive myoclonic 6, 614018
GPR56	84.3	100%	98%	Polymicrogyria, bilateral frontoparietal, 606854
GRID2	130.6	100%	98%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRM1	138.1	99%	98%	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	14.8	60%	35%	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233
ITPR1	105.2	100%	98%	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	113.2	100%	100%	Episodic ataxia/myokymia syndrome, 160120
KCNC1	138	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	76.6	80%	69%	Spinocerebellar ataxia 13, 605259
KCNJ10	158.3	100%	100%	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791
KCNMA1	86.3	99%	93%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCTD7	80.3	71%	68%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	97.5	100%	98%	Spastic paraplegia 8, autosomal dominant, 603563
KIAA0226	90.4	100%	99%	?Spinocerebellar ataxia, autosomal recessive 15
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	61.8	99%	91%	Hydrocephalus due to aqueductal stenosis, 307000 MASA syndrome, 303350 CRASH syndrome, 303350 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000
MARS2	151.3	100%	100%	Spastic ataxia 3, autosomal recessive, 611390
MECP2	85.3	98%	87%	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
MICU1	93.1	100%	99%	Myopathy with extrapyramidal signs

MMADHC	65.3	89%	89%	Homocystinuria, cbID type, variant 1, 277410 Methylmalonic aciduria, cbID type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cbID type, 277410
MRE11A	90.2	99%	99%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	92	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	98.9	93%	93%	Ataxia, spastic, 4, 613672
MTPP	104.3	99%	97%	Abetalipoproteinemia, 200100; {Metabolic syndrome, protection against}, 605552
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	130.2	100%	100%	Myoclonus, familial cortical, 614937
NPC1	87.5	99%	97%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220
NPC2	61.9	100%	100%	Niemann-pick disease, type C2, 607625
NUP62	89.1	99%	98%	Striatonigral degeneration, infantile, 271930
OPA1	133.1	99%	99%	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
PDE10A	99.9	98%	95%	No OMIM disease phenotype

PDE8B	94.7	100%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	61.3	100%	95%	Basal ganglia calcification, idiopathic, 5, 615483
PDGFRB	83.9	100%	97%	Basal ganglia calcification, idiopathic, 4, 615007 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550
PDHA1	61.1	100%	88%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	100.4	100%	100%	Lacticacidemia due to PDX1 deficiency, 245349
PDSS1	86.7	89%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	92.8	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	96.3	89%	83%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHYH	78.3	100%	100%	Refsum disease, 266500
PIK3R5	75.6	100%	98%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	75.5	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	63.1	98%	86%	Paroxysmal nonkinesigenic dyskinesia, 118800
PNKP	68.3	100%	98%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures and developmental delay, 613402
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), 20
POLR3A	85	99%	94%	Leukodystrophy, hypomyelinating,7 w/wo oligodontia and/or hypogonadotropic hypogonadism,607694
POLR3B	106.4	100%	98%	Leukodystrophy, hypomyelinating,8, w/wo oligodontia and/or hypogonadotropic hypogonadism,614381

PRKCG	96	97%	94%	Spinocerebellar ataxia 14, 605361
PRKRA	117.5	100%	100%	Dystonia 16, 612067
PRRT2	80.4	100%	99%	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066
REEP1	84.4	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	134.2	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNF170	112.3	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	100%	99%	Aicardi-Goutieres syndrome 5, 612952 Chilblain lupus 2, 614415 -3
SCN8A	136.5	100%	99%	Cognitive impairment w/wo cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558
SERAC1	86.2	100%	100%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	149.8	100%	100%	Ataxia-ocular apraxia-2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SGCE	83.1	95%	91%	maternally imprinted Dystonia-11, myoclonic, 159900
SIL1	95.6	100%	99%	Marinesco-Sjogren syndrome, 248800
SLC12A6	90.6	100%	99%	Agenesis of corpus callosum with peripheral neuropathy, 218000
SLC16A2	44.7	94%	77%	Allan-Herndon-Dudley syndrome, 300523
SLC18A2	106.7	100%	100%	No OMIM disease phenotype
SLC19A3	110.9	100%	100%	Thiamine metabolism dysfunction syndrome 2, 607483
SLC1A3	113.2	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	83.2	99%	97%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	99.4	86%	85%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 -3
SLC2A1	90	100%	100%	GLUT1 deficiency syndrome 1, 606777 GLUT1 deficiency syndrome 2, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 Dystonia 9, 601042

SLC30A10	133	100%	99%	Hyper manganeseemia 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
SLC52A3	77	100%	100%	Brown-Vialetto-Van Laere syndrome 1,211530 Fazio-Londe disease,211500
SLC6A3	73.9	100%	99%	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135
SLC9A1	103.6	100%	97%	?Lichtenstein-Knorr syndrome,616291
SMPD1	100.5	96%	90%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616
SNCA	147.6	100%	100%	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601
SNX14	98.6	99%	95%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SPAST	102	100%	100%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	105.9	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	78	97%	87%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	68.9	100%	99%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	92.4	99%	96%	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	105.7	99%	97%	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998
TAF1	74.2	100%	99%	Dystonia-Parkinsonism, X-linked, 314250
TDP1	106.5	100%	100%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	105.8	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	31	90%	73%	Deafness, X-linked 1, progressive Mohr-Tranebjaerg syndrome, 304700 Jensen syndrome, 311150
TMEM240	111.6	100%	100%	Spinocerebellar ataxia 21, 607454
TMEM67	115.7	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	116.7	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	67.1	92%	81%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	89.3	99%	86%	Ataxia with isolated vitamin E deficiency, 277460
TUBB4A	58.2	85%	77%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
VAMP1	102.9	95%	95%	spastic ataxia 1, autosomal dominant
VCP	106.9	100%	97%	Spastic Ataxia 1, autosomal dominant, 108600
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	74.5	99%	93%	Spastic paraplegia 53, autosomal recessive, 614898
VRK1	136.9	100%	100%	Pontocerebellar hypoplasia type 1A, 607596
WDR45	38.3	91%	85%	?Neurodegeneration with brain iron accululation 5, 300894
WDR81	105.4	99%	98%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	98.3	97%	97%	Esophageal squamous cell carcinoma, 133239; Spinocerebellar ataxia 12, 614322
XPR1	104.1	100%	100%	Basal ganglia calcification, idiopathic, 6, 616413
ZFYVE26	86.9	97%	93%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	80.4	100%	96%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	103.2	93%	90%	Spinocerebellar ataxia, autosomal recessive 5, 606937

Gene symbols used follow HGNC 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

OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015

This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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