MOVEMENT DISORDERS GENE PANEL DG 2.9/DG 2.10 (225 genes)

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AARS2	141.5	99%	99%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCB7	151.3	99%	98%	Anemia, sideroblastic, with ataxia, 301310
ABCD1	94.7	76%	69%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ABHD12	113.2	98%	90%	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ADAR	136.8	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
ADCK3	161	99%	99%	Coenzyme Q10 deficiency, primary, 4, 612016
ADCY5	148.6	93%	91%	Dyskinesia, familial, with facial myokymia, 606703
AFG3L2	124	94%	87%	Spastic ataxia 5, autosomal recessive, 614487 Spinocerebellar ataxia 28, 610246
ALDH18A1	143.9	100%	100%	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	153.1	100%	99%	Sjogren-Larsson syndrome, 270200
ALS2	192.8	99%	99%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ANO10	136.7	99%	98%	Spinocerebellar ataxia, autosomal recessive 10, 613728
ANO3	167.4	99%	98%	Dystonia 24, 615034
AP4B1	163.4	100%	99%	Spastic paraplegia 47, autosomal recessive, 614066
AP4E1	125.7	99%	98%	Spastic paraplegia 51, autosomal recessive, 613744 Stuttering, familial persistent, 1, 184450
AP4M1	133.2	99%	97%	Spastic paraplegia 50, autosomal recessive, 612936
AP4S1	83.2	76%	70%	Spastic paraplegia 52, autosomal recessive, 614067

AP5Z1	104.5	100%	99%	Spastic paraplegia 48, autosomal recessive, 613647
APTX	127.2	94%	92%	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920
ARG1	181	100%	100%	Argininemia, 207800
ARSA	114.5	100%	99%	Metachromatic leukodystrophy, 250100
ARX	38.3	81%	67%	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	144.7	99%	94%	Canavan disease, 271900
ATCAY	165.5	100%	99%	Ataxia, cerebellar, Cayman type, 601238
ATL1	183.7	99%	98%	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, autosomal dominant, 182600
ATM	132.3	99%	96%	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic {Breast cancer, susceptibility to}, 114480
ATP13A2	129.3	100%	99%	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A2	219	100%	99%	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481
ATP1A3	204.3	100%	100%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
ATP2B3	155.7	99%	98%	?Spinocerebellar ataxia, X-linked 1, 302500
ATP7B	170.2	100%	99%	Wilson disease, 277900
B4GALNT1	167.7	99%	96%	Spastic paraplegia 26, autosomal recessive, 609195
BCAP31	75.6	94%	81%	Deafness, dystonia, and cerebral hypomyelination, 300475
BCKDHA	195.9	100%	99%	Maple syrup urine disease, type Ia, 248600
BCKDHB	147.9	93%	84%	Maple syrup urine disease, type Ib, 248600

BSCL2	129.8	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
C10orf2	192.9	100%	100%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286
C12orf65	94.5	98%	93%	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, autosomal recessive, 615035
C19orf12	110.3	100%	99%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CA8	131.4	97%	93%	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227
CACNA1A	104.4	95%	92%	Episodic ataxia, type 2, 108500 Migraine, familial hemiplegic, 1, 141500 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 Spinocerebellar ataxia 6, 183086
CACNA1G	152.3	99%	98%	Spinocerebellar ataxia 42, 616795
CACNB4	134.7	99%	97%	Episodic ataxia, type 5, 613855 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682
CAMTA1	203.2	100%	99%	Cerebellar ataxia, nonprogressive, with mental retardation, 614756
CAPN1	171.6	100%	100%	Spastic paraplegia 76, autosomal recessive, 616907
CCT5	171.1	99%	99%	Neuropathy, hereditary sensory, with spastic paraplegia, 256840
CIZ1	167.3	99%	97%	No OMIM phenotype Cervical dystonia, primary (Xiao (2012) Ann Neurol 71, 458)
COASY	173	100%	100%	Neurodegeneration with brain iron accumulation 6, 615643
COL4A1	103.5	98%	94%	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773 Brain small vessel disease with or without ocular anomalies, 607595 Porencephaly 1, 175780 ?Retinal arteries, tortuosity of, 180000 {Hemorrhage, intracerebral, susceptibility to}, 614519
COQ2	92.8	96%	93%	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500
COQ9	94.2	99%	96%	Coenzyme Q10 deficiency, primary, 5, 614654

COX20	60.1	95%	81%	Mitochondrial complex IV deficiency, 220110
CP	151.7	94%	91%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CSF1R	147.8	99%	98%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
CSTB	108.7	99%	99%	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800
CYP27A1	206.9	98%	96%	Cerebrotendinous xanthomatosis, 213700
CYP2U1	162.9	95%	93%	Spastic paraplegia 56, autosomal recessive, 615030
CYP7B1	129.5	97%	92%	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800
DBT	138.6	99%	96%	Maple syrup urine disease, type II, 248600
DCAF17	125.5	99%	97%	Woodhouse-Sakati syndrome, 241080
DCC	160.1	100%	99%	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic 133239 Mirror movements 1, 157600
DCTN1	142.6	99%	98%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DDC	121.6	99%	97%	Aromatic L-amino acid decarboxylase deficiency, 608643
DDHD1	168.6	97%	95%	Spastic paraplegia 28, autosomal recessive, 609340
DDHD2	175.7	99%	98%	Spastic paraplegia 54, autosomal recessive, 615033
DLAT	104.8	99%	98%	Pyruvate dehydrogenase E2 deficiency, 245348
DLD	157	99%	98%	Dihydrolipoamide dehydrogenase deficiency, 246900
DNAL4	62.7	99%	95%	?Mirror movements 3, 616059
DNMT1	128.6	99%	98%	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 Neuropathy, hereditary sensory, type IE, 614116
ECHS1	123.3	100%	99%	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277
EIF2B1	151	100%	99%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B2	155.4	100%	99%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF2B3	186.7	100%	100%	Leukoencephalopathy with vanishing white matter, 603896
EIF2B4	159.3	100%	99%	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896

EIF2B5	133.6	99%	98%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
EIF4G1	150.3	100%	99%	{Parkinson disease 18}, 614251
ELOVL5	129.4	100%	99%	Spinocerebellar ataxia 38, 615957
ERLIN2	172.1	100%	99%	Spastic paraplegia 18, autosomal recessive, 611225
FA2H	101.9	95%	89%	Spastic paraplegia 35, autosomal recessive, 612319
FAR1	102.4	96%	93%	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154
FBXO7	224.2	99%	98%	Parkinson disease 15, autosomal recessive, 260300
FGF14	225.2	100%	99%	Spinocerebellar ataxia 27, 609307
FLVCR1	166.7	99%	98%	Ataxia, posterior column, with retinitis pigmentosa, 609033
FOLR1	155.8	100%	100%	Neurodegeneration due to cerebral folate transport deficiency, 613068
FRMD7	140.9	99%	99%	Nystagmus 1, congenital, X-linked, 310700 Nystagmus, infantile periodic alternating, X-linked, 310700
FTL	149.2	99%	93%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GALC	115.9	99%	96%	Krabbe disease, 245200
GAN	199	100%	99%	Giant axonal neuropathy-1, 256850
GBA	227.1	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	185.6	100%	99%	Spastic paraplegia 46, autosomal recessive, 614409
GCDH	139.2	93%	91%	Glutaricaciduria, type I, 231670
GCH1	92.5	97%	89%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GFAP	119.9	99%	98%	Alexander disease, 203450
GJC2	50	88%	71%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206

GLB1	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GNAL	151.4	97%	93%	Dystonia 25, 615073
GOSR2	143.2	97%	96%	Epilepsy, progressive myoclonic 6, 614018
GPR143	73.2	90%	83%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GPR56	177.1	100%	100%	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752
GRID2	216.8	100%	99%	Spinocerebellar ataxia, autosomal recessive 18, 616204
GRM1	194.2	100%	99%	Spinocerebellar ataxia, autosomal recessive 13, 614831
HEXB	166.2	99%	95%	Sandhoff disease, infantile, juvenile, and adult forms, 268800
HPRT1	82.8	96%	89%	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322
HSPD1	105.4	98%	93%	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, autosomal dominant, 605280
ITPR1	177.7	100%	99%	Gillespie syndrome,206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360
KCNA1	179.6	100%	99%	Episodic ataxia/myokymia syndrome, 160120
KCNC1	218.2	100%	100%	Epilepsy, progressive myoclonic 7, 616187
KCNC3	148.5	72%	58%	Spinocerebellar ataxia 13, 605259
KCND3	210.5	100%	98%	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346
KCNJ10	219.9	100%	99%	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780
KCNJ6	192.3	100%	100%	Keppen-Lubinsky syndrome, 614098
KCNMA1	150.1	100%	99%	Generalized epilepsy and paroxysmal dyskinesia, 609446
KCTD7	157.8	94%	93%	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726
KIAA0196	157.5	99%	98%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
KIAA0226	117.9	98%	97%	?Spinocerebellar ataxia, autosomal recessive 15, 615705
KIAA2022	176.1	100%	99%	Mental retardation, X-linked 98, 300912

KIF1A	130.8	99%	97%	Mental retardation, autosomal dominant 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, autosomal recessive, 610357
KIF1C	131.8	99%	99%	Spastic ataxia 2, autosomal recessive, 611302
KIF5A	143.4	100%	99%	Spastic paraplegia 10, autosomal dominant, 604187
KMT2B	139.5	94%	92%	Dystonia 28, childhood-onset, 617284
L1CAM	152.5	99%	97%	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	154	100%	99%	Poretti-Boltshauser syndrome, 615960
MARS2	179.7	100%	100%	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430
MECP2	99.4	99%	95%	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	120.5	99%	96%	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282
MICU1	134.3	97%	93%	Myopathy with extrapyramidal signs, 615673
MLC1	114.2	100%	99%	Megalencephalic leukoencephalopathy with subcortical cysts, 604004
MMADHC	94.4	90%	80%	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410
MRE11A	64.3	97%	89%	Ataxia-telangiectasia-like disorder, 604391
MTHFR	161.7	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MTPAP	133.3	98%	94%	Ataxia, spastic, 4, 613672

MTTP	162.6	100%	99%	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552
NIPA1	179.3	99%	98%	Spastic paraplegia 6, autosomal dominant, 600363
NKX2-1	58.2	99%	92%	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550
NOL3	99.9	95%	88%	Myoclonus, familial cortical, 614937
NPC1	153.4	99%	99%	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220 {Nasopharyngeal carcinoma 1}
NPC2	148.6	100%	99%	Niemann-pick disease, type C2, 607625
NUP62	113.7	100%	99%	Striatonigral degeneration, infantile, 271930
OPA1	146.6	99%	97%	Behr syndrome,210000 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type),616896 {Glaucoma, normal tension, susceptibility to}, 606657
PANK2	178.4	99%	98%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PAX6	135.8	100%	100%	Aniridia, 106210 Cataract with late-onset corneal dystrophy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550 Peters anomaly, 604229 ?Morning glory disc anomaly, 120430
PDE10A	172.5	99%	99%	Dyskinesia,limb and orofacial,infantile-onset,616921 Striatal degeneration,autosomal dominant,616922
PDE8B	125	99%	99%	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161
PDGFB	110.3	100%	100%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174

PDGFRB	160.1	98%	95%	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	112.7	98%	92%	Pyruvate dehydrogenase E1-alpha deficiency, 312170
PDHX	133.8	99%	97%	Lacticacidemia due to PDX1 deficiency,245349
PDSS1	140.2	93%	87%	Coenzyme Q10 deficiency, primary, 2, 614651
PDSS2	146.3	98%	95%	Coenzyme Q10 deficiency, primary, 3, 614652
PDYN	136.3	100%	100%	Spinocerebellar ataxia 23, 610245
PEX10	124.7	97%	92%	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871
PEX2	187.6	100%	100%	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHYH	90	98%	93%	Refsum disease, 266500
PIK3R5	117.8	100%	99%	Ataxia-oculomotor apraxia 3, 615217
PLA2G6	135.5	99%	97%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
PLP1	155	99%	98%	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920
PMM2	171.3	100%	99%	Congenital disorder of glycosylation, type Ia, 212065
PMPCA	135.9	98%	95%	Spinocerebellar ataxia, autosomal recessive 2, 213200
PNKD	126.7	100%	99%	Paroxysmal nonkinesigenic dyskinesia,118800
PNKP	100.7	99%	97%	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402
PNPLA6	150.4	99%	98%	Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, autosomal recessive, 612020 ?Laurence-Moon syndrome, 245800

POLG	128.1	100%	99%	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
POLR3A	153	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	163.8	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
PRKCG	137.2	99%	95%	Spinocerebellar ataxia 14, 605361
PRKRA	179.8	99%	99%	Dystonia 16, 612067
PRRT2	91.4	100%	99%	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751
PYCR2	142.2	99%	96%	Leukodystrophy, hypomyelinating, 10, 616420
RAB18	108.6	99%	91%	Warburg micro syndrome 3, 614222
RAB3GAP1	155.9	99%	99%	Warburg micro syndrome 1, 600118
RAB3GAP2	116.8	99%	96%	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225
RAD51	141.7	89%	89%	Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244
REEP1	113.6	97%	95%	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	92%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	98%	Aicardi-Goutieres syndrome 3, 610329
RNF170	143.3	99%	95%	Ataxia, sensory, 1, autosomal dominant, 608984
RNF216	158.3	99%	98%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
RTN2	121.4	98%	96%	Spastic paraplegia 12, autosomal dominant, 604805
SACS	187.5	100%	99%	Spastic ataxia, Charlevoix-Saguenay type, 270550
SAMHD1	154.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SCN11A	165	99%	98%	Episodic pain syndrome, familial, 3, 615552

				Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN8A	209.5	99%	99%	Epileptic encephalopathy, early infantile, 13, 614558 ?Cognitive impairment with or without cerebellar ataxia, 614306
SERAC1	139.3	99%	96%	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739
SETX	197.6	100%	99%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SGCE	110.4	95%	92%	Dystonia-11, myoclonic, 159900
SIL1	175.7	99%	97%	Marinesco-Sjogren syndrome, 248800
SLC12A6	162.9	100%	100%	Agenesis of the corpus callosum with peripheral neuropathy, 218000
SLC16A2	70.9	96%	87%	Allan-Herndon-Dudley syndrome, 300523
SLC19A3	185.8	100%	100%	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483
SLC1A3	153.2	100%	100%	Episodic ataxia, type 6, 612656
SLC20A2	126.4	99%	97%	Basal ganglia calcification, idiopathic, 1, 213600
SLC25A15	238.4	98%	96%	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970
SLC2A1	191.5	100%	99%	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	194.2	100%	99%	Hypermanganesemia with dystonia 1, 613280
SLC33A1	152.6	98%	94%	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, autosomal dominant, 612539
SLC39A14	120.5	99%	97%	Hypermanganesemia with dystonia 2, 617013
SLC52A2	206.8	100%	100%	Brown-Vialetto-Van Laere syndrome 2, 614707
SLC52A3	108.8	99%	99%	Brown-Vialetto-Van Laere syndrome 1, 211530 Fazio-Londe disease, 211500
SLC6A3	150.7	100%	99%	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SLC9A1	161.7	100%	100%	?Lichtenstein-Knorr syndrome, 616291
SMPD1	151.8	99%	98%	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616

SNCA	158.8	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
SNX14	89.1	96%	88%	Spinocerebellar ataxia, autosomal recessive 20, 616354
SOX10	74.6	98%	95%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SPAST	82.7	97%	88%	Spastic paraplegia 4, autosomal dominant, 182601
SPG11	145	99%	98%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SPG20	163.8	99%	98%	Troyer syndrome, 275900
SPG21	141.5	99%	98%	Mast syndrome, 248900
SPG7	138.5	96%	92%	Spastic paraplegia 7, autosomal recessive, 607259
SPR	194	99%	90%	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716
SPTBN2	127.6	99%	99%	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia, autosomal recessive 14, 615386
STUB1	186.3	100%	99%	Spinocerebellar ataxia, autosomal recessive 16, 615768
SUOX	221.3	100%	100%	Sulfite oxidase deficiency, 272300
SYNE1	172	99%	99%	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 Spinocerebellar ataxia, autosomal recessive 8, 610743
TAF1	133.9	99%	97%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TANGO2	158.8	100%	100%	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
TBC1D20	165.4	94%	94%	Warburg micro syndrome 4, 615663
TDP1	128.3	99%	98%	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250
TECPR2	169.7	100%	99%	Spastic paraplegia 49, autosomal recessive, 615031
TENM4	165	100%	99%	Tremor, hereditary essential, 5, 616736
TGM6	163.5	99%	98%	Spinocerebellar ataxia 35, 613908
TH	87.6	97%	93%	Segawa syndrome, recessive, 605407
THAP1	162.3	100%	99%	Dystonia 6, torsion, 602629
TIMM8A	43.7	88%	71%	Jensen syndrome, 311150 Mohr-Tranebjaerg syndrome, 304700

TMEM240	136.1	100%	99%	Spinocerebellar ataxia 21, 607454
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TOR1A	210.4	100%	99%	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}
TPP1	155.4	100%	100%	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia, autosomal recessive 7, 609270
TREM2	149.5	99%	98%	Nasu-Hakola disease, 221770
TREX1	302.9	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
TSEN54	98.8	96%	93%	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204
TTBK2	165.1	100%	99%	Spinocerebellar ataxia 11, 604432
TTC19	105.8	90%	82%	Mitochondrial complex III deficiency, nuclear type 2, 615157
TTPA	128.8	94%	86%	Ataxia with isolated vitamin E deficiency, 277460
TUBA1A	115.3	99%	97%	Lissencephaly 3, 611603
TUBB4A	135.6	96%	95%	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438
TUBG1	190.6	100%	100%	Cortical dysplasia, complex, with other brain malformations 4, 615412
TYROBP	90.7	100%	100%	Nasu-Hakola disease, 221770
VAMP1	156.6	100%	100%	Spastic ataxia 1, autosomal dominant, 108600
VCP	155.7	100%	99%	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	217.6	100%	99%	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050
VPS13A	89.6	97%	91%	Choreoacanthocytosis, 200150
VPS37A	86.4	93%	76%	Spastic paraplegia 53, autosomal recessive, 614898
VRK1	157.2	99%	97%	Pontocerebellar hypoplasia type 1A, 607596

WDR45	85.2	95%	89%	Neurodegeneration with brain iron accululation 5, 300894
WDR81	187.7	100%	99%	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185
WWOX	143.3	100%	99%	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322
XK	107.1	99%	99%	McLeod syndrome with or without chronic granulomatous disease, 300842
XPR1	171.5	99%	99%	Basal ganglia calcification, idiopathic, 6, 616413
ZFYVE26	130.7	100%	99%	Spastic paraplegia 15, autosomal recessive, 270700
ZFYVE27	133.5	100%	99%	Spastic paraplegia 33, autosomal dominant, 610244
ZNF592	139.4	100%	99%	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 : April 14th 2017.
This list is accurate for panel version DG 2.9 and DG 2.10

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