MOVEMENT DISORDERS GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
KCNC3	56	48	605259	Spinocerebellar ataxia 13
AP4E1	130	95	613744	Spastic paraplegia 51 autosomal recessive
FGF14	132	85	609307	Spinocerebellar ataxia 27
SLC6A3	78	94	613135	Parkinsonism-dystonia infantile
PNKD	61	86	200	-
SNCA	144	96	127750	Dementia Lewy body
L1CAM	95	93	304100	Corpus callosum partial agenesis of
WDR81	95	94	610185	Cerebellar ataxia mental retardation and dysequilibrium syndrome 2
SETX	153	96	602433	Amyotrophic lateral sclerosis 4 juvenile
ATCAY	88	94	601238	Ataxia cerebellar Cayman type
ATP2B3	88	83	302500	Spinocerebellar ataxia X-linked 1
ASPA	118	96	271900	Canavan disease
PRRT2	71	96	602066	Convulsions familial infantile with paroxysmal choreoathetosis
SLC2A1	90	94	601042	Dystonia 9
VPS13A	110	96	200150	Choreoacanthocytosis
SPR	69	90	612716	Dystonia dopa-responsive due to sepiapterin reductase deficiency
PDHA1	105	85	308930	Leigh syndrome X-linked
EIF2B2	109	86	603896	Leukoencephalopathy with vanishing white matter
EIF2B3	105	94	603896	Leukoencephalopathy with vanishing white matter
EIF2B4	130	86	603896	Leukoencephaly with vanishing white matter
EIF2B5	112	94	603896	Leukoencephalopathy with vanishing white matter
FLVCR1	95	96	609033	Ataxia posterior column with retinitis pigmentosa
FTL	130	48	600886	Hyperferritinemia-cataract syndrome
AP5Z1	74	86	613647	Spastic paraplegia 48 autosomal recessive
OPA1	121	99	165500	Optic atrophy 1
ZNF592	128	86	606937	Spinocerebellar ataxia autosomal recessive 5
CIZ1	101	91	200	-
SLC33A1	93	96	614482	Congenital cataracts hearing loss and neurodegeneration

KIF5A	113	82	604187	Spastic paraplegia 10 autosomal dominant
POLG	91	85	203700	Mitochondrial DNA depletion syndrome 4A (Alpers type)
FA2H	61	88	612319	Spastic paraplegia 35 autosomal recessive
MECP2	107	100	105830	Angelman syndrome
MTPAP	113	94	613672	Ataxia spastic 4
AP4M1	104	91	612936	Spastic paraplegia 50 autosomal recessive
PDYN	153	92	610245	Spinocerebellar ataxia 23
RNASEH2A	103	87	610333	Aicardi-Goutieres syndrome 4
RNASEH2B	94	95	610181	Aicardi-Goutieres syndrome 2
DCTN1	133	90	607641	Neuropathy distal hereditary motor type VIIB
RNASEH2C	134	100	610329	Aicardi-Goutieres syndrome 3
PNPLA6	90	87	612020	Spastic paraplegia 39 autosomal recessive
TIMM8A	56	60	200	Deafness X-linked 1
NPC1	110	84	257220	Niemann-Pick disease type C1
NPC2	76	86	607625	Niemann-pick disease type C2
DLD	141	94	246900	Dihydrolipoamide dehydrogenase deficiency
GPR56	91	95	606854	Polymicrogyria bilateral frontoparietal
CSTB	146	96	254800	Epilepsy progressive myoclonic 1A (Unverricht and Lundborg)
LRRK2	117	94	607060	Parkinson disease 8
VAMP1	120	77	200	-
SLC30A10	125	97	613280	Hypermanganesemia with dystonia polycythemia and cirrhosis
MRE11A	98	80	604391	Ataxia-telangiectasia-like disorder
KCNA1	124	48	160120	Episodic ataxia/myokymia syndrome
CACNB4	100	92	613855	Episodic ataxia type 5
PDSS1	109	86	614651	Coenzyme Q10 deficiency primary 2
PDSS2	94	92	614652	Coenzyme Q10 deficiency primary 3
ARX	37	94	308350	Epileptic encephalopathy early infantile 1
C12orf65	159	100	613559	Combined oxidative phosphorylation deficiency 7
TTBK2	131	96	604432	Spinocerebellar ataxia 11
PRKRA	103	97	612067	Dystonia 16
GALC	98	93	245200	Krabbe disease
NKX2-1	88	93	118700	Chorea hereditary benign
FBXO7	151	92	260300	Parkinson disease 15 autosomal recessive

TAF1	128	71	314250	Dystonia-Parkinsonism X-linked
GCDH	89	86	231670	Glutaricaciduria type I
MMADHC	79	80	200	-
SACS	165	100	270550	Spastic ataxia Charlevoix-Saguenay type
TREX1	122	100	225750	Aicardi-Goutieres syndrome 1 dominant and recessive
NIPA1	112	89	600363	Spastic paraplegia 6 autosomal dominant
REEP1	93	87	614751	Neuronopathy distal hereditary motor type VB
DLAT	116	89	245348	Pyruvate dehydrogenase E2 deficiency
ABCB7	99	90	301310	Anemia sideroblastic with ataxia
AFG3L2	93	83	614487	Ataxia spastic 5 autosomal recessive
ATP13A2	81	88	606693	Parkinson disease 9
ATM	116	93	208900	Ataxia-telangiectasia
CYP7B1	98	88	613812	Bile acid synthesis defect congenital 3
C10orf2	136	97	271245	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
SYNE1	120	87	612998	Emery-Dreifuss muscular dystrophy 4 autosomal dominant
CCT5	119	60	256840	Neuropathy hereditary sensory with spastic paraplegia
VCP	131	86	613954	Amyotrophic lateral sclerosis 14 with or without frontotemporal dementia
NUP62	84	97	271930	Striatonigral degeneration infantile
SPTBN2	99	89	600224	Spinocerebellar ataxia 5
DNMT1	105	89	604121	Cerebellar ataxia deafness and narcolepsy autosomal dominant
GOSR2	115	82	614018	Epilepsy progressive myoclonic 6
HSPD1	43	20	612233	Leukodystrophy hypomyelinating 4
SLC25A15	146	88	238970	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
CACNA1A	89	88	108500	Episodic ataxia type 2
ATP7B	130	86	277900	Wilson disease
SPG7	87	90	200	-
GFAP	89	89	203450	Alexander disease
KIAA0226	97	87	200	-
ATL1	121	97	613708	Neuropathy hereditary sensory type ID
HEXB	106	97	268800	Sandhoff disease infantile juvenile and adult forms
TTPA	97	97	277460	Ataxia with isolated vitamin E deficiency
VPS37A	86	76	614898	Spastic paraplegia 53 autosomal recessive
BSCL2	119	87	269700	Lipodystrophy congenital generalized type 2

PAX6	103	82	106210	Aniridia
PINK1	89	83	605909	Parkinson disease 6 early onset
KCNJ10	158	94	600791	Enlarged vestibular aqueduct digenic
RTN2	74	89	604805	Spastic paraplegia 12 autosomal dominant
PEX7	112	91	614879	Peroxisome biogenesis disorder 9B
GLB1	88	84	230500	GM1-gangliosidosis type I
SLC1A3	119	88	612656	Episodic ataxia type 6
SERAC1	95	95	614739	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome
SPG20	127	95	275900	Troyer syndrome
ANO3	118	92	615034	Dystonia 24
SPG21	114	87	200	-
PDE8B	102	89	614190	Pigmented nodular adrenocortical disease primary 3
ABHD12	72	98	612674	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract
ADCK3	97	87	200	-
DCAF17	96	97	241080	Woodhouse-Sakati syndrome
ANO10	118	94	613728	Spinocerebellar ataxia autosomal recessive 10
DDC	102	93	608643	Aromatic L-amino acid decarboxylase deficiency
SUOX	174	99	272300	Sulfite oxidase deficiency
NOL3	132	95	614937	Myoclonus familial cortical
APTX	152	95	208920	Ataxia early-onset with oculomotor apraxia and hypoalbuminemia
ERLIN2	125	94	611225	Spastic paraplegia 18 autosomal recessive
COQ9	113	83	614654	Coenzyme Q10 deficiency primary 5
GAN	146	91	256850	Giant axonal neuropathy-1
PRKCG	117	86	605361	Spinocerebellar ataxia 14
PMM2	118	89	212065	Congenital disorder of glycosylation type Ia
RNF170	130	94	608984	Ataxia sensory 1 autosomal dominant
TMEM67	104	95	216360	COACH syndrome
EIF4G1	116	92	614251	Parkinson disease 18
GNAL	82	99	615073	Dystonia 25
COQ2	79	93	607426	Coenzyme Q10 deficiency primary 1
PANK2	152	93	607236	HARP syndrome
KIF1A	68	91	614255	Mental retardation autosomal dominant 9
CYP27A1	126	86	213700	Cerebrotendinous xanthomatosis

ZFYVE27	95	78	610244	Spastic paraplegia 33 autosomal dominant
PSEN1	104	90	613737	Acne inversa familial 3
CA8	75	99	613227	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3
ZFYVE26	101	82	270700	Spastic paraplegia 15 autosomal recessive
SIL1	111	82	248800	Marinesco-Sjogren syndrome
PLA2G6	77	86	256600	Infantile neuroaxonal dystrophy 1
SGCE	92	92	159900	Dystonia-11 myoclonic
SPAST	87	99	182601	Spastic paraplegia 4 autosomal dominant
C19orf12	69	95	614298	Neurodegeneration with brain iron accumulation 4
WDR45	75	68	300894	Neurodegeneration with brain iron accululation 5
CYP2U1	109	94	615030	Spastic paraplegia 56 autosomal recessive
ABCD1	59	84	300100	Adrenoleukodystrophy
KIAA0196	108	89	603563	Spastic paraplegia 8 autosomal dominant
TH	83	90	605407	Segawa syndrome recessive
SLC19A3	123	91	607483	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)
HPRT1	75	80	300323	HPRT-related gout
GJC2	34	99	608804	Leukodystrophy hypomyelinating 2
ALDH3A2	103	85	270200	Sjogren-Larsson syndrome
GCH1	85	98	128230	Dystonia DOPA-responsive with or without hyperphenylalaninemia
PLEKHG4	87	96	200	-
DDHD2	112	91	615033	Spastic paraplegia 54 autosomal recessive
DDHD1	117	99	609340	Spastic paraplegia 28 autosomal recessive
MTHFR	98	91	236250	Homocystinuria due to MTHFR deficiency
TOR1A	144	96	200	-
MARS2	154	100	200	-
THAP1	112	94	602629	Dystonia 6 torsion
PIK3R5	82	91	615217	Ataxia-oculomotor apraxia 3
TGM6	66	87	613908	Spinocerebellar ataxia 35
PEX10	77	90	614870	Peroxisome biogenesis disorder 6A (Zellweger)
PDHX	99	95	200	-
B4GALNT1	89	86	609195	Spastic paraplegia 26 autosomal recessive
GBA	145	76	608013	Gaucher disease perinatal lethal
ACTB	101	17	243310	Baraitser-Winter syndrome 1

BCKDHA	109	92	248600	Maple syrup urine disease type Ia
PLP1	79	79	312080	Pelizaeus-Merzbacher disease
BCKDHB	89	98	248600	Maple syrup urine disease type Ib
TECPR2	116	86	615031	Spastic paraplegia 49 autosomal recessive
ATP1A3	115	84	614820	Alternating hemiplegia of childhood 2
SAMHD1	127	89	612952	Aicardi-Goutieres syndrome 5
PARK2	89	82	200	-
AP4S1	75	84	614067	Spastic paraplegia 52 autosomal recessive
EIF2B1	139	82	603896	Leukoencephalopathy with vanishing white matter
PHYH	90	93	266500	Refsum disease
ITPR1	117	81	606658	Spinocerebellar ataxia 15
PARK7	112	86	200	-
MTTP	111	90	545000	Merff syndrome
SLC16A2	89	90	300523	Allan-Herndon-Dudley syndrome
GBA2	142	96	614409	Spastic paraplegia 46 autosomal recessive
DBT	100	93	248600	Maple syrup urine disease type II
TDP1	127	90	607250	Spinocerebellar ataxia autosomal recessive with axonal neuropathy
ARSA	83	94	250100	Metachromatic leukodystrophy
SMPD1	95	94	257200	Niemann-Pick disease type A
CP	94	79	604290	Cerebellar ataxia
AP4B1	106	96	614066	Spastic paraplegia 47 autosomal recessive
SPG11	113	89	604360	Spastic paraplegia 11 autosomal recessive
VLDLR	131	89	224050	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors