Whole Exome Sequencing
Gene package Movement disorders, version 3, 1-7-2017

Technical information
After DNA was enriched using Agilent Sureselect Clinical Research Exome (CRE) Capture, samples were run on the Illumina Hiseq platform. The aim is to obtain 50 million total reads per exome with a mapped fraction >0.98. The average coverage of the exome is ~50x. Data are demultiplexed by Illumina software bcl2fastq. Reads are mapped to the genome using BWA (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by Genome Analysis Toolkit (reference: http://www.broadinstitute.org/gatk/). Analysis is performed in Cartagenia using The Variant Calling File (VCF) followed by filtering. Additionally, MPLA analysis was performed for *APTX, FXN* and *SETX* (SALSA P316 Recessive Ataxias probemix; MRC Holland) and for several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). For *ATXN1, ATXN2, ATXN3, ATXN7, ATXN10, FMR1, PPP2R2B* and *TBP* a repeat expansion test was performed. For *FXN* and *CACNA1A* a repeat expansion test was performed in additional to either MPLA and/or exome sequencing. It is not excluded that pathogenic mutations are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889	612035	80	100	100
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	51	100	98
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	46	96	86
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	62	100	94
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	77	100	100
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	96	100	100
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	91	98	96
ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	78	100	100
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	68	90	84

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ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	82	100	99
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	91	100	100
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	61	97	87
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225	606352	77	100	98
ANO10	Spinocerebellar ataxia 10, 613728	613726	73	100	100
ANO3	Dystonia 24, 615034	610110	68	100	97
AP4B1	Spastic paraplegia 47, 614066	607245	83	100	100
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	61	100	98
AP4M1	Spastic paraplegia 50, 612936	602296	75	100	100
AP4S1	Spastic paraplegia 52, 614067	607243	40	97	85
AP5Z1	Spastic paraplegia 48, 613647	613653	69	100	99
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	86	100	98
ARG1	Argininemia, 207800	608313	71	100	100
ARSA	Metachromatic leukodystrophy, 250100	607574	79	100	100
ARX	Epileptic encephalopathy, early infantile, 1, 308350 Hydranencephaly with abnormal genitalia, 300215 Lissencephaly 2, 300215 Mental retardation 29 and others, 300419 Partington syndrome, 309510 Proud syndrome, 300004	300382	24	84	61
ASPA	Canavan disease, 271900	608034	85	100	97
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	99	100	100
ATL1	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, 182600	606439	68	100	100
ATM	Ataxia-telangiectasia, 208900 {Breast cancer, susceptibility to}, 114480 Lymphoma, B-cell non-Hodgkin, somatic Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	64	100	97
ATP13A2	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, 617225	610513	67	100	99

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ATP1A2	Alternating hemiplegia of childhood, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	109	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	96	100	99
ATP2B3	?Spinocerebellar ataxia 1, 302500	300014	57	100	97
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	39	100	90
ATP7B	Wilson disease, 277900	606882	90	100	100
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	82	100	98
ATXN1	Spinocerebellar ataxia 1, 164400	601556	no coverage data, repeat expansion		
ATXN10	Spinocerebellar ataxia 10, 603516	611150	no coverage data, repeat expansion		
ATXN2	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600 Spinocerebellar ataxia 2, 183090	601517	no coverage data, repeat expansion		
ATXN3	Machado-Joseph disease, 109150	607047	no coverage data, repeat expansion		
ATXN7	Spinocerebellar ataxia 7, 164500	607640	no coverage data, repeat expansion		
AUH	3-methylglutaconic aciduria, type I, 250950	600529	60	100	90
B4GALNT1	Spastic paraplegia 26, 609195	601873	86	100	100
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	22	76	46
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	112	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	93	100	92
BCS1L	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000	603647	114	100	100
BSCL2	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	606158	82	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	56	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	71	100	100
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
CACNA1A	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	601011	74	100	95
CACNA1B	?Dystonia 23, 614860	601012	56	96	88
CACNA1G	Spinocerebellar ataxia 42, 616795	604065	88	100	98
CACNB4	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855	601949	74	100	96
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	100	100	99
CAPN1	Spastic paraplegia 76, 616907	114220	68	100	99
CCDC88C	Hydrocephalus, nonsyndromic, 236600 ?Spinocerebellar ataxia 40, 616053	611204	77	100	100
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	610150	85	99	96
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	47	98	94
CHCHD2	Parkinson disease 22, 616710	616244	33	89	67
CIZ1	No OMIM phenotype	611420	58	96	87
COASY	Neurodegeneration with brain iron accumulation 6, 615643	609855	74	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	94
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	95	100	100
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	75	100	95
COX10	Leigh syndrome due to mitochondrial COX4 deficiency, 256000 Mitochondrial complex IV deficiency, 220110	602125	105	100	100
COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 Leigh syndrome due to cytochrome c oxidase deficiency, 256000	603646	86	100	100
COX20	Mitochondrial complex IV deficiency, 220110	614698	14	60	25
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	50	87	77
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	81	100	99
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	124	100	97
CWF19L1	Spinocerebellar ataxia 17, 616127	616120	41	97	76
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	95	99	95
CYP2U1	Spastic paraplegia 56, 615030	610670	89	96	93
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, 270800	603711	56	100	95
DBT	Maple syrup urine disease, type II, 248600	248610	70	100	95

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DCAF17	Woodhouse-Sakati syndrome, 241080	612515	71	100	98
DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Mirror movements 1, 157600	120470	85	100	97
DCTN1	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	601143	84	100	98
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	95	100	100
DDHD1	Spastic paraplegia 28, 609340	614603	84	98	95
DDHD2	Spastic paraplegia 54, 615033	615003	86	100	100
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	62	100	95
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	84	100	97
DNAJC13	No OMIM phenotype	614334	44	95	80
DNAJC3	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192	601184	46	98	85
DNAJC6	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	608375	88	100	100
DNAL1	Ciliary dyskinesia, primary, 16, 614017	610062	57	90	89
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	70	100	98
EEF2	?Spinocerebellar ataxia 26, 609306	130610	66	100	98
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896	606686	89	100	100
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606454	85	100	100
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896	606273	100	100	100
EIF2B4	Leukoencephaly with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606687	74	100	96
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	603945	88	100	100
EIF4G1	{Parkinson disease 18}, 614251	600495	92	100	99
ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 ?Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	57	100	98
ELOVL5	Spinocerebellar ataxia 38, 615957	611805	35	99	80
ERLIN2	Spastic paraplegia 18, 611225	611605	92	100	100
FA2H	Spastic paraplegia 35, 612319	611026	55	98	87
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	27	88	58
FBXO7	Parkinson disease 15, 260300	605648	100	100	95
FGF14	Spinocerebellar ataxia 27, 609307	601515	96	100	100
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	82	100	97

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FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	44	100	89
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	94	100	100
FOXG1	Rett syndrome, congenital variant, 613454	164874	80	83	77
FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	42	100	96
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	62	94	76
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782	137070	79	100	99
FXN	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300	606829	38	96	72
GALC	Krabbe disease, 245200	606890	64	100	98
GAN	Giant axonal neuropathy-1, 256850	605379	87	100	100
GBA	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	606463	110	100	100
GBA2	Spastic paraplegia 46, 614409	609471	62	99	93
GCDH	Glutaricaciduria, type I, 231670	608801	81	100	99
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	63	100	87
GFAP	Alexander disease, 203450	137780	62	100	100
GIGYF2	{Parkinson disease 11}, 607688	612003	65	100	97
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, 613206	608803	40	83	58
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	77	100	97
GNAL	Dystonia 25, 615073	139312	49	96	83
GOSR2	Epilepsy, progressive myoclonic 6, 614018	604027	76	100	100

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GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	36	99	75
GRID1	No OMIM phenotype	610659	88	100	99
GRM1	Spinocerebellar ataxia 13, 614831	604473	89	100	100
HEXA	GM2-gangliosidosis, several forms, 272800 [Hex A pseudodeficiency], 272800 Tay-Sachs disease, 272800	606869	85	100	100
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	88	100	95
HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	36	100	85
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	27	84	58
HTRA2	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297	606441	86	100	100
IFRD1	No OMIM phenotype	603502	66	100	97
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	96	100	100
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	81	100	100
KCNC1	Epilepsy, progressive myoclonic 7, 616187	176258	75	100	99
KCNC3	Spinocerebellar ataxia 13, 605259	176264	56	80	66
KCND3	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346	605411	104	100	99
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	122	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	73	100	97
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	90	100	100
KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	75	100	98
KIF1C	Spastic ataxia 2, 611302	603060	55	99	92
KIF5A	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, 604187	602821	81	100	100
L1CAM	CRASH syndrome, 303350 Corpus callosum, partial agenesis of, 304100 Hydrocephalus due to aqueductal stenosis, 307000 Hydrocephalus with Hirschsprung disease, 307000 Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 MASA syndrome, 303350	308840	52	100	100

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LAMA1	Poretti-Boltshauser syndrome, 615960	150320	88	100	99
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	76	100	98
LRRK2	{Parkinson disease 8}, 607060	609007	75	100	97
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540	157140	55	97	91
MARS2	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, 611390	609728	93	100	100
MECP2	{Autism susceptibility 3}, 300496 Encephalopathy, neonatal severe, 300673 Mental retardation syndromic, Lubs type, 300260 Mental retardation, syndromic 13, 300055 Rett syndrome, 312750 Rett syndrome, atypical, 312750 Rett syndrome, preserved speech variant, 312750	300005	52	99	81
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	45	98	87
MICU1	Myopathy with extrapyramidal signs, 615673	605084	37	93	80
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	71	100	96
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	60	100	97
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}	607093	94	100	100
MTPAP	?Spastic ataxia 4, 613672	613669	74	100	99
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	78	100	96
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	94	100	100
NEXMIF	Mental retardation 98, 300912	300524	44	100	97
NIPA1	Spastic paraplegia 6, 600363	608145	116	100	97
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, monmedullary, 1}, 188550	600635	70	100	97

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NOL3	Myoclonus, familial cortical, 614937	605235	54	100	99
NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	83	100	96
NPC2	Niemann-pick disease, type C2, 607625	601015	115	100	100
NUP62	Striatonigral degeneration, infantile, 271930	605815	67	100	100
OPA1	Behr syndrome, 210000 {Glaucoma, normal tension, susceptibility to}, 606657 ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 Optic atrophy 1, 165500 Optic atrophy plus syndrome, 125250	605290	69	100	93
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	90	100	100
PARK7	Parkinson disease 7 early-onset, 606324	602533	63	100	99
PAX6	Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Keratitis, 148190 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550	607108	62	100	99
PCNA	?Ataxia-telangiectasia-like disorder, 615919	176740	33	97	74
PDE10A	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, 616922	610652	49	99	95
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, 609161	603390	66	100	98
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174	190040	73	100	100
PDGFRB	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 (4) Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812	173410	92	100	99
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
PDHX	Lacticacidemia due to PDX1 deficiency, 245349	608769	77	100	100
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	65	91	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	69	99	94

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PDYN	Spinocerebellar ataxia 23, 610245	131340	76	100	100
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	60	95	89
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	71	100	100
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	68	100	89
PHYH	Refsum disease, 266500	602026	57	99	85
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	76	100	100
PINK1	Parkinson disease 6, early onset, 605909	608309	65	94	90
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	88	100	99
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	71	100	99
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	93	100	100
PMPCA	Spinocerebellar ataxia 2, 213200	613036	43	93	78
PNKD	Paroxysmal nonkinesigenic dyskinesia, 118800	609023	80	100	100
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	61	100	98
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	72	100	98
POLG	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 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	86	100	100
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694	614258	88	100	100
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	82	100	99
PPP2R2B	Spinocerebellar ataxia 12, 604326	604325	no coverage data, repeat expansion		
PRKCG	Spinocerebellar ataxia 14, 605361	176980	84	100	99
PRKN	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 {Leprosy, susceptibility to}, 607572 Parkinson disease, juvenile, type 2, 600116	602544	69	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PRKRA	Dystonia 16, 612067	603424	55	100	97
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751	614386	58	100	100
PSEN1	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700	104311	71	100	98
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	60	96	87
RAB18	Warburg micro syndrome 3, 614222	602207	59	97	91
RAB29	No OMIM phenotype	603949	71	100	100
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	68	100	98
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	69	100	96
RAD51	{Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508	179617	58	88	88
REEP1	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, 610250	609139	73	100	100
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	89	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	50	100	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	112	100	100
RNF170	Ataxia, sensory, 1, 608984	614649	58	100	99
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	609948	54	98	90
RTN2	Spastic paraplegia 12, 604805	603183	77	100	100
RUBCN	?Spinocerebellar ataxia 15, 615705	613516	76	98	96
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550	604490	68	100	99
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	76	100	94
SCN11A	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548	604385	81	100	100
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 Seizures, benign familial infantile, 5, 617080	600702	97	100	99
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	69	100	99

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SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia 1, 606002	608465	76	100	99
SGCE	Dystonia-11, myoclonic, 159900	604149	64	94	93
SIL1	Marinesco-Sjogren syndrome, 248800	608005	85	100	100
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	71	100	99
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	42	99	89
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	118	100	100
SLC1A3	Episodic ataxia, type 6, 612656	600111	92	100	100
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	83	100	97
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	106	90	86
SLC2A1	Dystonia 9, 601042 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	104	100	100
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	103	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	62	100	96
SLC39A14	Hypermanganesemia with dystonia 2, 617013	608736	51	96	83
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	103	100	100
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500	613350	60	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	87	100	100
SLC9A1	?Lichtenstein-Knorr syndrome, 616291	107310	60	100	98
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	110	100	100
SNCA	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	163890	82	100	100
SNX14	Spinocerebellar ataxia 20, 616354	616105	27	80	52
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	45	99	87
SPART	Troyer syndrome, 275900	607111	71	100	99
SPAST	Spastic paraplegia 4, 182601	604277	49	100	91

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SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	73	100	99
SPG21	Mast syndrome, 248900	608181	85	100	99
SPG7	Spastic paraplegia 7, 607259	602783	93	98	92
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	98	100	94
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	82	100	99
STUB1	Spinocerebellar ataxia 16, 615768	607207	71	100	96
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	38	86	75
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	74	100	98
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	71	88	88
SYNE1	Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	87	100	100
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	42	96	79
SYT14	Spinocerebellar ataxia 11, 614229	610949	92	93	89
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	78	100	100
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	52	100	91
TBC1D20	Warburg micro syndrome 4, 615663	611663	51	93	85
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	no coverage data, repeat expansion		
TDP1	Spinocerebellar ataxia with axonal neuropathy, 607250	607198	77	100	96
TECPR2	Spastic paraplegia 49, 615031	615000	92	100	100
TENM4	Tremor, hereditary essential, 5, 616736	610084	51	98	89
TGM6	Spinocerebellar ataxia 35, 613908	613900	82	100	100
TH	Segawa syndrome, recessive, 605407	191290	55	100	95
THAP1	Dystonia 6, torsion, 602629	609520	84	100	96
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	42	100	100
TMEM240	Spinocerebellar ataxia 21, 607454	616101	62	100	100

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TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550	609884	51	99	90
TOR1A	{Dystonia-1, modifier of} Dystonia-1, torsion, 128100	605204	71	100	100
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	79	100	98
TREM2	Nasu-Hakola disease, 221770	605086	79	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 {Systemic lupus erythematosus, susceptibility to}, 152700 Vasculopathy, retinal, with cerebral leukodystrophy, 192315	606609	129	100	100
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	67	96	96
TTBK2	Spinocerebellar ataxia 11, 604432	611695	70	100	99
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	57	90	78
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	70	96	89
TUBA1A	Lissencephaly 3, 611603	602529	34	93	78
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	60	97	93
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	65	100	99
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	103	100	100
TYROBP	Nasu-Hakola disease, 221770	604142	44	100	92
UCHL1	?{Parkinson disease 5, susceptibility to}, 613643 Spastic paraplegia 79, 615491	191342	70	100	99
VAMP1	Spastic ataxia 1, 108600	185880	55	100	95
VCP	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	601023	84	100	99
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	105	100	99
VPS13A	Choreoacanthocytosis, 200150	605978	57	99	94
VPS13C	Parkinson disease 23, early onset, 616840	608879	37	90	72
VPS35	{Parkinson disease 17}, 614203	601501	52	89	79
VPS37A	Spastic paraplegia 53, 614898	609927	46	76	55

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VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	78	100	96
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	96	100	99
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	32	89	78
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	90	100	100
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia 12, 614322	605131	72	100	100
XK	McLeod syndrome with or without chronic granulomatous disease, 300842	314850	54	100	98
XPR1	Basal ganglia calcification, idiopathic, 6, 616413	605237	50	97	84
ZFYVE26	Spastic paraplegia 15, 270700	612012	79	100	97
ZFYVE27	Spastic paraplegia 33, 610244	610243	71	100	100
ZNF592	No OMIM phenotype	613624	80	100	99

- Gene symbols according HGCN
- OMIM release used: 2-6-2017
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping.
- The statistics above are based on a set of 50 samples
- Median depth is the median of the mean sequence depth over the protein coding exons of the transcript
- % 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