Whole Exome Sequencing
Gene package Movement disorders, version 2, 25-9-2015

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 *ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN10, FMR1, PPP2R2B, C9orf72* 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).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
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	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310	102630	77	100	100
ADCK3	Coenzyme Q10 deficiency, primary, 4, 612016	606980	95	100	100
AFG3L2	Spinocerebellar ataxia 28, 610246 Ataxia, spastic, 5, autosomal recessive, 614487	604581	78	100	100
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	68	90	84
ANO10	Spinocerebellar ataxia, autosomal recessive 10, 613728	613726	91	100	100
ANO3	Dystonia 24, 615034	610110	73	100	100
AP4B1	Spastic paraplegia 47, autosomal recessive, 614066	607245	68	100	97
AP4E1	Spastic paraplegia 51, autosomal recessive, 613744	607244	83	100	100
AP4M1	Spastic paraplegia 50, autosomal recessive, 612936	602296	61	100	98
AP4S1	Spastic paraplegia 52, autosomal recessive, 614067	607243	75	100	100
AP5Z1	Spastic paraplegia 48, autosomal recessive, 613647	613653	40	97	85
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	69	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ARSA	Metachromatic leukodystrophy, 250100	607574	86	100	98
ARX	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	300382	79	100	100
ASPA	Canavan disease, 271900	608034	24	84	61
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	85	100	97
ATL1	Spastic paraplegia 3A, autosomal dominant, 182600 Neuropathy, hereditary sensory, type ID, 613708	606439	99	100	100
ATM	Ataxia-telangiectasia, 208900 Lymphoma, B-cell non-Hodgkin, somatic {Breast cancer, susceptibility to}, 114480 Lymphoma, mantle cell, somatic T-cell prolymphocytic leukemia, somatic	607585	68	100	100
ATN1	Dentatorubro-pallidoluysian atrophy, 125370	607462	64	100	97
ATP13A2	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693	610513	67	100	99
ATP1A3	Dystonia-12, 128235 Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338	182350	96	100	99
ATP2B3	?Spinocerebellar ataxia, X-linked 1, 302500	300014	57	100	97
ATP6AP2	?Mental retardation, X-linked, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, X-linked, 300911	300556	39	100	90
ATP7B	Wilson disease, 277900	606882	90	100	100
ATXN1	Spinocerebellar ataxia 1, 164400	601556	no coverage data, repeat expansion		
ATXN10	Spinocerebellar ataxia 10, 603516	611150	no coverage data, repeat expansion		
ATXN2	Spinocerebellar ataxia 2, 183090 {Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600	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		
B4GALNT1	Spastic paraplegia 26, autosomal recessive, 609195	601873	86	100	100
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	112	100	99
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	93	100	92

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
BSCL2	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924	606158	82	100	100
C10orf2	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138	606075	103	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043	614297	71	100	100
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	614260	no coverage data, repeat expansion		
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	66	100	100
CACNA1A	Migraine, familial hemiplegic, 1, 141500 Episodic ataxia, type 2, 108500 Spinocerebellar ataxia 6, 183086 Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500	601011	74	100	95
CACNB4	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 Episodic ataxia, type 5, 613855	601949	74	100	96
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 ?Spinocerebellar ataxia 40, 616053	611204	77	100	100
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	610150	85	99	96
CIZ1	No OMIM phenotype	611420	58	96	87
COMT	{Schizophrenia, susceptibility to}, 181500 {Panic disorder, susceptibility to}, 167870	116790	104	100	100
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	60	100	94
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	75	100	95
CP	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290	117700	50	87	77
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	124	100	97
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	95	99	95
CYP2U1	Spastic paraplegia 56, autosomal recessive, 615030	610670	89	96	93
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, autosomal recessive, 270800	603711	56	100	95
DBT	Maple syrup urine disease, type II, 248600	248610	70	100	95
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	71	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
DCTN1	Neuropathy, distal hereditary motor, type VIIB, 607641 {Amyotrophic lateral sclerosis, susceptibility to}, 105400 Perry syndrome, 168605	601143	84	100	98
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	95	100	100
DDHD1	Spastic paraplegia 28, autosomal recessive, 609340	614603	84	98	95
DDHD2	Spastic paraplegia 54, autosomal recessive, 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	Parkinson disease 21, 616361	614334	44	95	80
DNAJC6	Parkinson disease 19, juvenile-onset, 615528	608375	88	100	100
DNMT1	Neuropathy, hereditary sensory, type IE, 614116 Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121	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
ERLIN2	Spastic paraplegia 18, autosomal recessive, 611225	611605	92	100	100
FA2H	Spastic paraplegia 35, autosomal recessive, 612319	611026	55	98	87
FBXO7	Parkinson disease 15, autosomal recessive, 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
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	no coverage data, repeat expansion		
FTL	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604	134790	62	94	76
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
GBA	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	606463	110	100	100
GBA2	Spastic paraplegia 46, autosomal recessive, 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 Spastic paraplegia 44, autosomal recessive, 613206 Lymphedema, hereditary, IC, 613480	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
GPR56	Polymicrogyria, bilateral frontoparietal, 604110	606854	no coverage data		
GRM1	Spinocerebellar ataxia, autosomal recessive 13, 614831	604473	89	100	100
GRN	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706	138945	95	100	100
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	88	100	95
HPRT1	Lesch-Nyhan syndrome, 300322 HPRT-related gout, 300323	308000	36	100	85
HSPD1	Spastic paraplegia 13, autosomal dominant, 605280 Leukodystrophy, hypomyelinating, 4, 612233	118190	27	84	58
HTRA2	{Parkinson disease 13}, 610297	606441	86	100	100
IFRD1	No OMIM phenotype	603502	66	100	97
ITPR1	Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	96	100	100
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	81	100	100
KCNC3	Spinocerebellar ataxia 13, 605259	176264	56	80	66

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
KCND3	Spinocerebellar ataxia 19, 607346 Brugada syndrome 9, 616399	605411	104	100	99
KCNJ10	SESAME syndrome, 612780 Enlarged vestibular aqueduct, digenic, 600791	602208	122	100	100
KCNMA1	Generalized epilepsy and paroxysmal dyskinesia, 609446	600150	73	100	97
KIAA0196	Spastic paraplegia 8, autosomal dominant, 603563 Ritscher-Schinzel syndrome 1, 220210	610657	96	100	99
KIF1A	Spastic paraplegia 30, autosomal recessive, 610357 Neuropathy, hereditary sensory, type IIC, 614213 Mental retardation, autosomal dominant 9, 614255	601255	75	100	98
KIF1C	Spastic ataxia 2, autosomal recessive, 611302	603060	55	99	92
KIF5A	Spastic paraplegia 10, autosomal dominant, 604187	602821	81	100	100
L1CAM	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	308840	52	100	100
LRRK2	{Parkinson disease 8}, 607060	609007	75	100	97
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600	157140	55	97	91
MARS2	Spastic ataxia 3, autosomal recessive, 611390 ?Combined oxidative phosphorylation deficiency 25, 616430	609728	93	100	100
MECP2	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 Mental retardation, X-linked syndromic, Lubs type, 300260 Rett syndrome, atypical, 312750	300005	52	99	81
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410	611935	60	100	97
MRE11A	Ataxia-telangiectasia-like disorder, 604391	600814	53	95	85

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
MTHFR	Homocystinuria due to MTHFR deficiency, 236250 {Schizophrenia, susceptibility to}, 181500 {Vascular disease, susceptibility to} {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050	607093	94	100	100
MTPAP	Ataxia, spastic, 4, 613672	613669	74	100	99
NIPA1	Spastic paraplegia 6, autosomal dominant, 600363	608145	116	100	97
NKX2-1	{Thyroid cancer, monmedullary, 1}, 188550 Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978	600635	70	100	97
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	Optic atrophy 1, 165500 {Glaucoma, normal tension, susceptibility to}, 606657 Optic atrophy plus syndrome, 125250	605290	69	100	93
PANK2	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236	606157	90	100	100
PARK2	Parkinson disease, juvenile, type 2, 600116 Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 {Leprosy, susceptibility to}, 607572	602544	69	100	99
PARK7	Parkinson disease 7, autosomal recessive early-onset, 606324	602533	63	100	99
PAX6	Aniridia, 106210 Peters anomaly, 604229 Cataract with late-onset corneal dystrophy, 106210 Keratitis, 148190 Foveal hypoplasia 1, 136520 ?Morning glory disc anomaly, 120430 Optic nerve hypoplasia, 165550 Coloboma, ocular, 120200 Coloboma of optic nerve, 120430 Gillespie syndrome, 206700	607108	62	100	99
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, autosomal dominant, 609161	603390	66	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PDGFB	Meningioma, SIS-related, 607174 Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483	190040	73	100	100
PDGFRB	Myeloproliferative disorder with eosinophilia, 131440 Basal ganglia calcification, idiopathic, 4, 615007 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592	173410	92	100	99
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	64	99	92
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	65	91	83
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	69	99	94
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
PEX7	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879	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, autosomal recessive, 612953	603604	88	100	99
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, X-linked, 312920	300401	71	100	99
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	93	100	100
PNKD	Paroxysmal nonkinesigenic dyskinesia, 118800	609023	80	100	100
PNPLA6	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400	603197	72	100	98
POLG	Progressive external ophthalmoplegia, autosomal recessive 1, 258450 Progressive external ophthalmoplegia, autosomal dominant 1, 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	174763	86	100	100
PPP2R2B	Spinocerebellar ataxia 12, 604326	604325	no coverage data, repeat expansion		
PRKCG	Spinocerebellar ataxia 14, 605361	176980	84	100	99
PRKRA	Dystonia 16, 612067	603424	55	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
PRRT2	Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751 Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066	614386	58	100	100
PSEN1	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	104311	71	100	98
RAB29	No OMIM phenotype	603949	71	100	100
REEP1	Spastic paraplegia 31, autosomal dominant, 610250 ?Neuronopathy, distal hereditary motor, type VB, 614751	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, autosomal dominant, 608984	614649	58	100	99
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	609948	54	98	90
RTN2	Spastic paraplegia 12, autosomal dominant, 604805	603183	77	100	100
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
SCN8A	?Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558	600702	97	100	99
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	69	100	99
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002	608465	76	100	99
SGCE	Dystonia-11, myoclonic, 159900	604149	64	94	93
SIL1	Marinesco-Sjogren syndrome, 248800	608005	85	100	100
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	42	99	89
SLC18A2	No OMIM phenotype	193001	42	99	86
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

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
SLC2A1	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 GLUT1 deficiency syndrome 2, childhood onset, 612126 {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 Dystonia 9, 601042 Stomatin-deficient cryohydrocytosis with neurologic defects, 608885	138140	104	100	100
SLC30A10	Hypermanganesemia with dystonia, polycythemia, and cirrhosis, 613280	611146	103	100	100
SLC33A1	Spastic paraplegia 42, autosomal dominant, 612539 Congenital cataracts, hearing loss, and neurodegeneration, 614482	603690	62	100	96
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	103	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 613135	126455	87	100	100
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	110	100	100
SNCA	Parkinson disease 4, 605543 Dementia, Lewy body, 127750 Parkinson disease 1, 168601	163890	82	100	100
SPAST	Spastic paraplegia 4, autosomal dominant, 182601	604277	49	100	91
SPG11	Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668	610844	73	100	99
SPG20	Troyer syndrome, 275900	607111	71	100	99
SPG21	Mast syndrome, 248900	608181	85	100	99
SPG7	Spastic paraplegia 7, autosomal recessive, 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, autosomal recessive 14, 615386	604985	82	100	99
STUB1	Spinocerebellar ataxia, autosomal recessive 16, 615768	607207	71	100	96
SUOX	Sulfite oxidase deficiency, 272300	606887	104	100	100
SYNE1	Spinocerebellar ataxia, autosomal recessive 8, 610743 Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998	608441	87	100	100
SYNJ1	Parkinson disease 20, early-onset, 615530	604297	42	96	79
SYT14	Spinocerebellar ataxia, autosomal recessive 11, 614229	610949	92	93	89
TAF1	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966	313650	78	100	100
TARDBP	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069	605078	75	100	100
TBP	Spinocerebellar ataxia 17, 607136 {Parkinson disease, susceptibility to}, 168600	600075	no coverage data, repeat expansion		
TDP1	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250	607198	77	100	96

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
TECPR2	Spastic paraplegia 49, autosomal recessive, 615031	615000	92	100	100
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 Jensen syndrome, 311150	300356	42	100	100
TMEM240	Spinocerebellar ataxia 21, 607454	616101	62	100	100
TMEM67	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Nephronophthisis 11, 613550	609884	51	99	90
TOR1A	Dystonia-1, torsion, 128100 {Dystonia-1, modifier of}	605204	71	100	100
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700	606609	129	100	100
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
TUBB4A	Dystonia 4, torsion, autosomal dominant, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	60	97	93
UCHL1	?{Parkinson disease 5, susceptibility to}, 613643 ?Neurodegeneration with optic atrophy, childhood onset, 615491	191342	70	100	99
VAMP1	Spastic ataxia 1, autosomal dominant, 108600	185880	55	100	95
VCP	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 Charcot-Marie-Tooth disease, type 2Y, 616687	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
VPS35	{Parkinson disease 17}, 614203	601501	52	89	79
VPS37A	Spastic paraplegia 53, autosomal recessive, 614898	609927	46	76	55
WDR45	Neurodegeneration with brain iron accululation 5, 300894	300526	32	89	78
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185	614218	90	100	100
WWOX	Esophageal squamous cell carcinoma, somatic, 133239 Spinocrebellar ataxia, autosomal recessive 12, 614322 Epileptic encephalopathy, early infantile, 28, 616211	605131	72	100	100
ZFYVE26	Spastic paraplegia 15, autosomal recessive, 270700	612012	79	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x
ZFYVE27	Spastic paraplegia 33, autosomal dominant, 610244	610243	71	100	100
ZNF592	No OMIM phenotype	613624	80	100	99

- Gene symbols according HGCN
- OMIM release used: 17-3-2016
- "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