Whole Exome Sequencing
Gene package Movement disorders, version 6.1, 31-1-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Cartagenia software and classified with Alamut Visual. 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, C9orf72, 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).

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	% covered >30x
AAAS	Achalasia-addisonianism-alacrimia syndrome, 231550	605378	102	100	100	100
AARS1	Charcot-Marie-Tooth disease, axonal, type 2N, 613287 Epileptic encephalopathy, early infantile, 29, 616339	601065	89	100	100	96
AARS2	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889	612035	122	100	100	100
ABCB7	Anemia, sideroblastic, with ataxia, 301310	300135	59	100	98	87
ABCD1	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100	300371	86	84	78	75
ABHD12	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674	613599	61	100	100	94
ACTB	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371	102630	192	100	100	100
ADAR	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400	146920	90	100	100	100
ADCY5	Dyskinesia, familial, with facial myokymia, 606703	600293	111	98	95	93

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ADGRG1	Polymicrogyria, bilateral frontoparietal, 606854 Polymicrogyria, bilateral perisylvian, 615752	604110	120	100	100	100
ADPRHL2	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170	610624	129	100	100	100
AFG3L2	Spastic ataxia 5, 614487 Spinocerebellar ataxia 28, 610246	604581	77	99	95	90
AGTPBP1	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276	606830	64	100	95	87
AIMP1	Leukodystrophy, hypomyelinating, 3, 260600	603605	78	100	100	96
ALDH18A1	Cutis laxa 3, 616603 Cutis laxa, type IIIA, 219150 Spastic paraplegia 9A, 601162 Spastic paraplegia 9B, 616586	138250	80	100	100	98
ALDH3A2	Sjogren-Larsson syndrome, 270200	609523	64	100	99	93
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency, 271980	610045	66	100	97	91
ALS2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225	606352	72	100	100	96
ANO10	Spinocerebellar ataxia 10, 613728	613726	57	100	99	88
ANO3	Dystonia 24, 615034	610110	61	93	92	87
AP4B1	Spastic paraplegia 47, 614066	607245	81	100	100	100
AP4E1	Spastic paraplegia 51, 613744 Stuttering, familial persistent, 1, 184450	607244	64	100	100	96
AP4M1	Spastic paraplegia 50, 612936	602296	130	100	100	99
AP4S1	Spastic paraplegia 52, 614067	607243	44	100	99	87
AP5Z1	Spastic paraplegia 48, 613647	613653	111	100	100	99
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920	606350	76	100	99	92
ARG1	Argininemia, 207800	608313	75	100	100	98
ARL6IP1	?Spastic paraplegia 61, 615685	607669	87	100	100	99
ARSA	Metachromatic leukodystrophy, 250100	607574	136	100	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	46	89	79	68
ASPA	Canavan disease, 271900	608034	55	100	99	91
ATCAY	Ataxia, cerebellar, Cayman type, 601238	608179	116	100	100	96

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ATG5	?Spinocerebellar ataxia 25, 617584	604261	47	100	97	81
ATL1	Neuropathy, hereditary sensory, type ID, 613708 Spastic paraplegia 3A, 182600	606439	72	100	100	93
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	75	100	99	95
ATN1	Congenital hypotonia, epilepsy, developmental delay, and digital anomalies, 618494 Dentatorubral-pallidoluysian atrophy, 125370	607462	155	100	100	99
ATP13A2	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, 617225	610513	117	100	100	98
ATP1A2	Alternating hemiplegia of childhood 1, 104290 Migraine, familial basilar, 602481 Migraine, familial hemiplegic, 2, 602481	182340	130	100	100	100
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	144	100	100	100
ATP2B3	?Spinocerebellar ataxia 1, 302500	300014	96	100	100	99
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	46	100	91	64
ATP7B	Wilson disease, 277900	606882	99	100	100	99
ATP8A2	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268	605870	69	100	99	94
ATXN1	Spinocerebellar ataxia 1, 164400	601556	116	100	100	100
ATXN2	{Amyotrophic lateral sclerosis, susceptibility to, 13}, 183090 {Parkinson disease, late-onset, susceptibility to}, 168600 Spinocerebellar ataxia 2, 183090	601517	75	97	93	86
ATXN3	Machado-Joseph disease, 109150	607047	87	100	99	92
ATXN7	Spinocerebellar ataxia 7, 164500	607640	90	100	97	93
AUH	3-methylglutaconic aciduria, type I, 250950	600529	94	100	100	98
B4GALNT1	Spastic paraplegia 26, 609195	601873	111	100	100	100
BCAP31	Deafness, dystonia, and cerebral hypomyelination, 300475	300398	83	100	100	97
BCKDHA	Maple syrup urine disease, type Ia, 248600	608348	160	100	100	100
BCKDHB	Maple syrup urine disease, type Ib, 248600	248611	64	100	99	94

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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	101	100	100	100
C12orf65	Combined oxidative phosphorylation deficiency 7, 613559 Spastic paraplegia 55, 615035	613541	78	100	100	100
C19orf12	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, 615043	614297	186	100	100	100
C9orf72	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550	614260	56	100	100	96
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227	114815	79	100	100	94
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	87	100	98	93
CACNA1B	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements, 618497	601012	116	99	97	96
CACNA1E	Epileptic encephalopathy, early infantile, 69, 618285	601013	106	100	99	97
CACNA1G	Spinocerebellar ataxia 42, 616795 Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087	604065	146	100	100	100
CACNB4	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 Episodic ataxia, type 5, 613855	601949	57	100	100	94
CAMTA1	Cerebellar ataxia, nonprogressive, with mental retardation, 614756	611501	161	100	99	97
CAPN1	Spastic paraplegia 76, 616907	114220	135	100	100	100
CCDC88C	Hydrocephalus, congenital, 1, 236600 ?Spinocerebellar ataxia 40, 616053	611204	104	100	100	97
CCT5	Neuropathy, hereditary sensory, with spastic paraplegia, 256840	610150	85	100	100	91
CDKL5	Epileptic encephalopathy, early infantile, 2, 300672	300203	58	100	97	87
CHCHD2	Parkinson disease 22, 616710	616244	67	100	88	68
CHMP1A	Pontocerebellar hypoplasia, type 8, 614961	164010	102	100	100	100
CHP1	?Spastic ataxia 9, 618438	606988	40	99	84	66
CIZ1	No OMIM phenotype	611420	100	99	95	91

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CLCN2	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 Hyperaldosteronism, familial, type II, 605635 Leukoencephalopathy with ataxia, 615651	600570	100	100	100	100
CLCN4	Raynaud-Claes syndrome, 300114	302910	80	100	100	98
CLP1	Pontocerebellar hypoplasia, type 10, 615803	608757	99	100	100	100
CLPB	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271	616254	114	100	100	100
COASY	Neurodegeneration with brain iron accumulation 6, 615643 Pontocerebellar hypoplasia, type 12, 618266	609855	139	100	100	100
COL6A3	Bethlem myopathy 1, 158810 Dystonia 27, 616411 Ullrich congenital muscular dystrophy 1, 254090	120250	104	100	100	99
COQ2	Coenzyme Q10 deficiency, primary, 1, 607426 {Multiple system atrophy, susceptibility to}, 146500	609825	66	100	100	94
COQ8A	Coenzyme Q10 deficiency, primary, 4, 612016	606980	129	100	100	100
COQ9	Coenzyme Q10 deficiency, primary, 5, 614654	612837	105	100	100	100
CP	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290	117700	63	100	100	96
CPT1C	?Spastic paraplegia 73, 616282	608846	107	100	100	98
CSF1R	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	99	100	100	100
CSTB	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800	601145	84	100	100	100
CWF19L1	Spinocerebellar ataxia 17, 616127	616120	59	100	100	92
CYB5R3	Methemoglobinemia, type I, 250800 Methemoglobinemia, type II, 250800	613213	127	100	100	100
CYP27A1	Cerebrotendinous xanthomatosis, 213700	606530	131	100	100	100
CYP2U1	Spastic paraplegia 56, 615030	610670	66	100	100	96
CYP7B1	Bile acid synthesis defect, congenital, 3, 613812 Spastic paraplegia 5A, 270800	603711	67	100	100	98
DAB1	Spinocerebellar ataxia 37, 615945	603448	70	100	100	97
DBT	Maple syrup urine disease, type II, 248600	248610	115	100	100	99
DCAF17	Woodhouse-Sakati syndrome, 241080	612515	75	100	100	94

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DCC	Colorectal cancer, somatic, 114500 Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600	120470	70	100	100	98
DCTN1	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	601143	95	100	100	99
DDC	Aromatic L-amino acid decarboxylase deficiency, 608643	107930	84	100	99	92
DDHD1	Spastic paraplegia 28, 609340	614603	107	100	98	92
DDHD2	Spastic paraplegia 54, 615033	615003	76	100	100	97
DHDDS	?Congenital disorder of glycosylation, type 1bb, 613861 Developmental delay and seizures with or without movement abnormalities, 617836 Retinitis pigmentosa 59, 613861	608172	69	100	100	99
DLAT	Pyruvate dehydrogenase E2 deficiency, 245348	608770	74	100	98	90
DLD	Dihydrolipoamide dehydrogenase deficiency, 246900	238331	77	100	100	98
DNAJC13	No OMIM phenotype	614334	71	100	99	94
DNAJC3	?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192	601184	66	100	99	91
DNAJC6	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	608375	84	100	100	98
DNAL4	?Mirror movements 3, 616059	610565	121	100	100	100
DNMT1	Cerebellar ataxia, deafness, and narcolepsy, 604121 Neuropathy, hereditary sensory, type IE, 614116	126375	93	100	100	97
DSTYK	Congenital anomalies of kidney and urinary tract 1, 610805 Spastic paraplegia 23, 270750	612666	77	100	98	93
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277	602292	79	100	100	100
EEF2	?Spinocerebellar ataxia 26, 609306	130610	117	100	100	100
EIF2B1	Leukoencephalopathy with vanishing white matter, 603896	606686	79	100	100	100
EIF2B2	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606454	77	100	98	91
EIF2B3	Leukoencephalopathy with vanishing white matter, 603896	606273	58	100	100	94
EIF2B4	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	606687	89	100	100	100
EIF2B5	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896	603945	72	100	100	98
EIF4G1	{Parkinson disease 18}, 614251	600495	100	100	100	100

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ELOVL4	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110	605512	71	100	100	98
ELOVL5	Spinocerebellar ataxia 38, 615957	611805	93	100	100	98
ENTPD1	Spastic paraplegia 64, 615683	601752	68	100	100	96
ERLIN1	Spastic paraplegia 62, 615681	611604	66	100	100	95
ERLIN2	Spastic paraplegia 18, 611225	611605	65	100	100	95
EXOSC3	Pontocerebellar hypoplasia, type 1B, 614678	606489	114	100	100	96
FA2H	Spastic paraplegia 35, 612319	611026	83	100	99	91
FAM126A	Leukodystrophy, hypomyelinating, 5, 610532	610531	63	100	100	98
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154	616107	74	100	100	96
FARS2	Combined oxidative phosphorylation deficiency 14, 614946 Spastic paraplegia 77, 617046	611592	111	100	100	100
FAT2	Spinocerebellar ataxia 45, 617769	604269	95	100	100	99
FBXO7	Parkinson disease 15, 260300	605648	86	100	100	97
FGF14	Spinocerebellar ataxia 27, 609307	601515	68	100	100	98
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa, 609033	609144	98	100	100	96
FMR1	Fragile X syndrome, 300624 Fragile X tremor/ataxia syndrome, 300623 Premature ovarian failure 1, 311360	309550	41	100	96	76
FOLR1	Neurodegeneration due to cerebral folate transport deficiency, 613068	136430	111	100	100	100
FOXG1	Rett syndrome, congenital variant, 613454	164874	127	97	90	84
FRMD7	Nystagmus 1, congenital, 310700 Nystagmus, infantile periodic alternating, 310700	300628	50	100	98	89
FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	144	100	100	100
FUS	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782	137070	77	100	100	97
FXN	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300	606829	41	100	91	64
GALC	Krabbe disease, 245200	606890	54	100	99	89
GAN	Giant axonal neuropathy-1, 256850	605379	86	100	100	99

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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	172	100	100	100
GBA2	Spastic paraplegia 46, 614409	609471	137	100	100	100
GCDH	Glutaricaciduria, type I, 231670	608801	112	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	52	100	100	93
GFAP	Alexander disease, 203450	137780	81	100	100	99
GIGYF2	{Parkinson disease 11}, 607688	612003	66	100	99	94
GJC2	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, 613206	608803	85	96	86	76
GLB1	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010	611458	105	100	100	98
GNAL	Dystonia 25, 615073	139312	71	100	100	94
GOSR2	Epilepsy, progressive myoclonic 6, 614018	604027	66	100	95	90
GPR143	Nystagmus 6, congenital, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500	300808	43	100	89	76
GRID2	Spinocerebellar ataxia 18, 616204	602368	78	100	100	99
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, 614254 Neurodevelopmental disorder with or without hyperkinetic movements and seizures, 617820	138249	132	100	100	99
GRIN2B	Epileptic encephalopathy, early infantile, 27, 616139 Mental retardation 6, 613970	138252	122	100	100	99
GRM1	Spinocerebellar ataxia 44, 617691 Spinocerebellar ataxia 13, 614831	604473	136	100	100	99
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures, 616756	610876	80	100	100	94
HEXB	Sandhoff disease, infantile, juvenile, and adult forms, 268800	606873	119	100	100	97
HPCA	Dystonia 2, torsion, 224500	142622	248	100	100	100

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HPRT1	HPRT-related gout, 300323 Lesch-Nyhan syndrome, 300322	308000	43	100	99	81
HSD17B4	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400	601860	62	100	98	93
HSPD1	Leukodystrophy, hypomyelinating, 4, 612233 Spastic paraplegia 13, 605280	118190	65	100	99	85
HTRA2	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297	606441	143	100	100	98
IBA57	Multiple mitochondrial dysfunctions syndrome 3, 615330 ?Spastic paraplegia 74, 616451	615316	124	100	100	97
IFRD1	No OMIM phenotype	603502	68	100	100	95
ISCA2	Multiple mitochondrial dysfunctions syndrome 4, 616370	615317	89	100	100	100
ITPR1	Gillespie syndrome, 206700 Spinocerebellar ataxia 15, 606658 Spinocerebellar ataxia 29, congenital nonprogressive, 117360	147265	90	100	100	98
KATNB1	Lissencephaly 6, with microcephaly, 616212	602703	141	100	100	100
KCNA1	Episodic ataxia/myokymia syndrome, 160120	176260	126	100	100	100
KCNA2	Epileptic encephalopathy, early infantile, 32, 616366	176262	108	100	100	100
KCNC1	Epilepsy, progressive myoclonic 7, 616187	176258	138	100	100	99
KCNC3	Spinocerebellar ataxia 13, 605259	176264	91	94	78	64
KCND3	Brugada syndrome 9, 616399 Spinocerebellar ataxia 19, 607346	605411	167	100	100	99
KCNJ10	Enlarged vestibular aqueduct, digenic, 600791 SESAME syndrome, 612780	602208	171	100	100	100
KCNMA1	?Cerebellar atrophy, developmental delay, and seizures, 617643 Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446	600150	88	100	100	96
KCTD17	Dystonia 26, myoclonic, 616398	616386	89	100	100	97
KCTD7	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726	611725	159	100	100	100
KIDINS220	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296	615759	59	100	98	90
KIF1A	Mental retardation 9, 614255 Neuropathy, hereditary sensory, type IIC, 614213 Spastic paraplegia 30, 610357	601255	101	100	100	98
KIF1C	Spastic ataxia 2, 611302	603060	141	100	100	100

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KIF5A	{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, 604187	602821	90	100	100	96
KMT2B	Dystonia 28, childhood-onset, 617284	606834	141	97	95	94
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	101	100	100	100
LAMA1	Poretti-Boltshauser syndrome, 615960	150320	87	100	100	97
LAMB1	Lissencephaly 5, 615191	150240	94	100	100	97
LMNB1	Leukodystrophy, adult-onset, 169500	150340	66	100	100	97
LRPPRC	Leigh syndrome, French-Canadian type, 220111	607544	60	100	99	91
LRRK2	{Parkinson disease 8}, 607060	609007	78	100	100	96
MAG	Spastic paraplegia 75, 616680	159460	156	100	100	100
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	120	100	100	100
MARS2	?Combined oxidative phosphorylation deficiency 25, 616430 Spastic ataxia 3, 611390	609728	157	100	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	113	100	100	96
MECR	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282	608205	97	100	100	99
MED20	No OMIM phenotype	612915	148	100	100	100
MICU1	Myopathy with extrapyramidal signs, 615673	605084	52	100	98	82
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts, 604004	605908	79	100	100	94

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
MMADHC	Homocystinuria, cblD type, variant 1, 277410 Methylmalonic aciduria and homocystinuria, cblD type, 277410 Methylmalonic aciduria, cblD type, variant 2, 277410	611935	63	100	100	90
MME	Charcot-Marie-Tooth disease, axonal, type 2T, 617017 ?Spinocerebellar ataxia 43, 617018	120520	58	100	99	91
MRE11	Ataxia-telangiectasia-like disorder 1, 604391	600814	55	100	97	83
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	99	100	100	99
MTPAP	?Spastic ataxia 4, 613672	613669	97	100	100	97
MTTP	Abetalipoproteinemia, 200100 {Metabolic syndrome, protection against}, 605552	157147	67	100	100	97
MVK	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900	251170	100	100	100	100
MYBPC1	Arthrogryposis, distal, type 1B, 614335 Lethal congenital contracture syndrome 4, 614915 Myopathy, congenital, with tremor, 618524	160794	62	100	100	94
MYORG	Basal ganglia calcification, idiopathic, 7, 618317	618255	205	100	100	99
NEFL	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 Charcot-Marie-Tooth disease, type 1F, 607734 Charcot-Marie-Tooth disease, type 2E, 607684	162280	157	100	100	100
NEU1	Sialidosis, type I, 256550 Sialidosis, type II, 256550	608272	155	100	100	100
NEXMIF	Mental retardation 98, 300912	300524	48	100	100	95
NFASC	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356	609145	101	100	100	99
NIPA1	Spastic paraplegia 6, 600363	608145	107	100	100	97
NKX2-1	Chorea, hereditary benign, 118700 Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 {Thyroid cancer, nonmedullary, 1}, 188550	600635	95	100	100	100
NKX6-2	Spastic ataxia 8, with hypomyelinating leukodystrophy, 617560	605955	107	99	88	83
NOL3	?Myoclonus, familial, 1, 614937	605235	132	100	100	100

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NPC1	Niemann-Pick disease, type C1, 257220 Niemann-Pick disease, type D, 257220	607623	87	100	100	99
NPC2	Niemann-pick disease, type C2, 607625	601015	95	100	100	100
NT5C2	Spastic paraplegia 45, 613162	600417	54	100	98	89
NUP62	Striatonigral degeneration, infantile, 271930	605815	107	100	100	100
OCLN	Pseudo-TORCH syndrome 1, 251290	602876	61	96	84	79
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	59	100	98	89
OPHN1	Mental retardation, with cerebellar hypoplasia and distinctive facial appearance, 300486	300127	52	100	96	84
PANK2	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200	606157	81	100	100	99
PARK7	Parkinson disease 7 early-onset, 606324	602533	68	100	99	87
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	73	100	100	96
PCNA	?Ataxia-telangiectasia-like disorder 2, 615919	176740	93	100	100	92
PDE10A	Dyskinesia, limb and orofacial, infantile-onset, 616921 Striatal degeneration, 616922	610652	62	100	99	94
PDE8B	Pigmented nodular adrenocortical disease, primary, 3, 614190 Striatal degeneration, 609161	603390	72	100	99	96
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174	190040	101	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
PDGFRB	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	173410	116	100	100	100
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency, 312170	300502	49	100	94	84
PDHX	Lacticacidemia due to PDX1 deficiency, 245349	608769	79	100	100	97
PDSS1	Coenzyme Q10 deficiency, primary, 2, 614651	607429	60	100	93	85
PDSS2	Coenzyme Q10 deficiency, primary, 3, 614652	610564	75	100	97	87
PDYN	Spinocerebellar ataxia 23, 610245	131340	134	100	100	100
PEX10	Peroxisome biogenesis disorder 6A (Zellweger), 614870 Peroxisome biogenesis disorder 6B, 614871	602859	100	100	100	97
PEX2	Peroxisome biogenesis disorder 5A (Zellweger), 614866 Peroxisome biogenesis disorder 5B, 614867	170993	63	100	100	100
PEX7	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100	601757	57	100	100	95
PHYH	Refsum disease, 266500	602026	121	100	100	94
PIK3R5	Ataxia-oculomotor apraxia 3, 615217	611317	107	100	100	100
PINK1	Parkinson disease 6, early onset, 605909	608309	100	98	93	88
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	118	100	100	99
PLD3	?Spinocerebellar ataxia 46, 617770	615698	148	100	100	100
PLP1	Pelizaeus-Merzbacher disease, 312080 Spastic paraplegia 2, 312920	300401	93	100	99	97
PMM2	Congenital disorder of glycosylation, type Ia, 212065	601785	72	100	100	97
PMPCA	Spinocerebellar ataxia 2, 213200	613036	133	100	100	100
PNKD	Paroxysmal nonkinesigenic dyskinesia 1, 118800	609023	112	100	100	100
PNKP	Ataxia-oculomotor apraxia 4, 616267 Microcephaly, seizures, and developmental delay, 613402	605610	104	100	100	98
PNPLA6	Boucher-Neuhauser syndrome, 215470 ?Laurence-Moon syndrome, 245800 Oliver-McFarlane syndrome, 275400 Spastic paraplegia 39, 612020	603197	123	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	114	100	100	99
POLR1C	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390	610060	92	100	100	98
POLR3A	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090	614258	84	100	100	98
POLR3B	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381	614366	76	100	99	93
PPP2R2B	Spinocerebellar ataxia 12, 604326	604325	77	100	99	95
PRF1	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027	170280	120	100	100	100
PRICKLE1	Epilepsy, progressive myoclonic 1B, 612437	608500	76	100	100	98
PRKCG	Spinocerebellar ataxia 14, 605361	176980	122	100	100	99
PRKN	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116	602544	96	100	100	99
PRKRA	Dystonia 16, 612067	603424	81	100	100	96
PRRT2	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 Episodic kinesigenic dyskinesia 1, 128200 Seizures, benign familial infantile, 2, 605751	614386	123	100	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	59	100	100	93
PTRHD1	No OMIM phenotype	617342	119	100	100	100
PUM1	Spinocerebellar ataxia 47, 617931	607204	67	100	100	98
PYCR2	Leukodystrophy, hypomyelinating, 10, 616420	616406	114	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
RAB18	Warburg micro syndrome 3, 614222	602207	85	100	100	97
RAB29	No OMIM phenotype	603949	68	100	100	99
RAB3GAP1	Warburg micro syndrome 1, 600118	602536	67	100	100	97
RAB3GAP2	Martsolf syndrome, 212720 Warburg micro syndrome 2, 614225	609275	66	100	99	92
RAD51	{Breast cancer, susceptibility to}, 114480 ?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508	179617	52	88	88	85
RARS1	Leukodystrophy, hypomyelinating, 9, 616140	107820	72	100	98	90
RARS2	Pontocerebellar hypoplasia, type 6, 611523	611524	66	100	99	93
REEP1	?Neuronopathy, distal hereditary motor, type VB, 614751 Spastic paraplegia 31, 610250	609139	72	100	100	99
RETREG1	Neuropathy, hereditary sensory and autonomic, type IIB, 613115	613114	73	100	98	86
RNASEH2A	Aicardi-Goutieres syndrome 4, 610333	606034	109	100	100	100
RNASEH2B	Aicardi-Goutieres syndrome 2, 610181	610326	62	100	98	88
RNASEH2C	Aicardi-Goutieres syndrome 3, 610329	610330	323	100	100	100
RNF170	Ataxia, sensory, 1, 608984	614649	69	100	97	85
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840	609948	66	100	97	86
ROBO3	Gaze palsy, familial horizontal, with progressive scoliosis, 1, 607313	608630	106	100	100	100
RTN2	Spastic paraplegia 12, 604805	603183	135	100	100	100
RUBCN	?Spinocerebellar ataxia 15, 615705	613516	85	100	98	96
SACS	Spastic ataxia, Charlevoix-Saguenay type, 270550	604490	70	100	100	99
SAMD9L	Ataxia-pancytopenia syndrome, 159550	611170	63	100	100	99
SAMHD1	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415	606754	64	100	99	88
SCN11A	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548	604385	88	100	100	97
SCN8A	Cognitive impairment with or without cerebellar ataxia, 614306 Epileptic encephalopathy, early infantile, 13, 614558 ?Myoclonus, familial, 2, 618364 Seizures, benign familial infantile, 5, 617080	600702	110	100	100	98
SCYL1	Spinocerebellar ataxia 21, 616719	607982	110	100	100	98
SEPSECS	Pontocerebellar hypoplasia type 2D, 613811	613009	78	100	100	96
SERAC1	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739	614725	65	100	99	86
SETX	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, with axonal neuropathy 2, 606002	608465	70	100	100	97

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SGCE	Dystonia-11, myoclonic, 159900	604149	84	100	100	94
SIL1	Marinesco-Sjogren syndrome, 248800	608005	100	100	100	100
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy, 218000	604878	67	100	99	94
SLC16A2	Allan-Herndon-Dudley syndrome, 300523	300095	75	100	100	95
SLC17A5	Salla disease, 604369 Sialic acid storage disorder, infantile, 269920	604322	86	100	100	97
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483	606152	73	100	100	98
SLC1A3	Episodic ataxia, type 6, 612656	600111	99	100	100	100
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	99	100	100	95
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970	603861	132	100	100	98
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	127	100	100	100
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	148	100	100	100
SLC33A1	Congenital cataracts, hearing loss, and neurodegeneration, 614482 Spastic paraplegia 42, 612539	603690	68	100	98	86
SLC39A14	Hypermanganesemia with dystonia 2, 617013 ?Hyperostosis cranalis interna, 144755	608736	92	100	100	97
SLC52A2	Brown-Vialetto-Van Laere syndrome 2, 614707	607882	173	100	100	100
SLC52A3	Brown-Vialetto-Van Laere syndrome 1, 211530 ?Fazio-Londe disease, 211500	613350	109	100	100	100
SLC6A19	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600	608893	115	100	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135	126455	102	100	100	100
SLC9A1	?Lichtenstein-Knorr syndrome, 616291	107310	119	100	100	100
SLC9A6	Mental retardation syndromic, Christianson type, 300243	300231	64	100	96	84
SMPD1	Niemann-Pick disease, type A, 257200 Niemann-Pick disease, type B, 607616	607608	131	100	100	99

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
SNCA	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	163890	52	100	100	98
SNORD118	Leukoencephalopathy, brain calcifications, and cysts, 614561	616663	Geen coverage data			
SNX14	Spinocerebellar ataxia 20, 616354	616105	64	100	99	91
SOX10	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266	602229	72	100	97	89
SPART	Troyer syndrome, 275900	607111	71	100	100	98
SPAST	Spastic paraplegia 4, 182601	604277	64	100	100	94
SPG11	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, 604360	610844	84	100	100	97
SPG21	Mast syndrome, 248900	608181	66	100	95	88
SPG7	Spastic paraplegia 7, 607259	602783	125	100	100	97
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	102	100	100	100
SPTBN2	Spinocerebellar ataxia 5, 600224 Spinocerebellar ataxia 14, 615386	604985	121	100	100	100
STUB1	?Spinocerebellar ataxia 48, 618093 Spinocerebellar ataxia 16, 615768	607207	137	100	100	100
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073	603921	56	100	97	86
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400	611224	88	100	100	100
SUMF1	Multiple sulfatase deficiency, 272200	607939	88	100	100	99
SUOX	Sulfite oxidase deficiency, 272300	606887	136	100	100	100
SURF1	Charcot-Marie-Tooth disease, type 4K, 616684 Leigh syndrome, due to COX IV deficiency, 256000	185620	88	94	89	87
SYNE1	Arthrogryposis multiplex congenita, myogenic type, 618484 Emery-Dreifuss muscular dystrophy 4, 612998 Spinocerebellar ataxia 8, 610743	608441	77	100	100	97
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	62	100	99	92
SYT14	?Spinocerebellar ataxia 11, 614229	610949	61	100	95	86
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	54	100	97	87

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
TANGO2	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878	616830	96	100	100	99
TBC1D20	Warburg micro syndrome 4, 615663	611663	74	100	93	93
TBC1D23	Pontocerebellar hypoplasia, type 11, 617695	617687	57	100	99	90
TBCD	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193	604649	104	100	99	96
TBP	{Parkinson disease, susceptibility to}, 168600 Spinocerebellar ataxia 17, 607136	600075	80	100	100	96
TDP1	?Spinocerebellar ataxia, with axonal neuropathy 1, 607250	607198	53	100	100	92
TDP2	Spinocerebellar ataxia 23, 616949	605764	120	100	100	99
TECPR2	Spastic paraplegia 49, 615031	615000	102	100	100	99
TENM4	Essential tremor, hereditary, 5, 616736	610084	97	100	99	97
TFG	Hereditary motor and sensory neuropathy, Okinawa type, 604484 ?Spastic paraplegia 57, 615658	602498	87	100	100	94
TGM6	Spinocerebellar ataxia 35, 613908	613900	117	100	100	99
TH	Segawa syndrome, recessive, 605407	191290	93	100	99	94
THAP1	Dystonia 6, torsion, 602629	609520	87	100	100	98
TIMM8A	Mohr-Tranebjaerg syndrome, 304700	300356	138	100	100	100
TMEM106B	Leukodystrophy, hypomyelinating, 16, 617964	613413	72	100	99	89
TMEM230	No OMIM phenotype	617019	30	74	52	36
TMEM240	Spinocerebellar ataxia 21, 607454	616101	108	100	100	100
TMEM67	{Bardet-Biedl syndrome 14, modifier of}, 615991 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 ?RHYNS syndrome, 602152	609884	77	100	100	94
TOE1	Pontocerebellar hypoplasia, type 7, 614969	613931	110	100	100	98
TOR1A	{Dystonia-1, modifier of} Dystonia-1, torsion, 128100	605204	73	100	100	98
TPP1	Ceroid lipofuscinosis, neuronal, 2, 204500 Spinocerebellar ataxia 7, 609270	607998	97	100	100	100
TREM2	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2, 618193	605086	107	100	100	100

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
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	234	100	100	100
TRPC3	?Spinocerebellar ataxia 41, 616410	602345	112	100	99	95
TSEN2	Pontocerebellar hypoplasia type 2B, 612389	608753	63	87	86	78
TSEN54	Pontocerebellar hypoplasia type 2A, 277470 Pontocerebellar hypoplasia type 4, 225753 ?Pontocerebellar hypoplasia type 5, 610204	608755	105	100	96	96
TTBK2	Spinocerebellar ataxia 11, 604432	611695	74	100	100	99
TTC19	Mitochondrial complex III deficiency, nuclear type 2, 615157	613814	53	100	92	77
TTPA	Ataxia with isolated vitamin E deficiency, 277460	600415	61	100	98	91
TUBA1A	Lissencephaly 3, 611603	602529	110	100	100	100
TUBB4A	Dystonia 4, torsion, 128101 Leukodystrophy, hypomyelinating, 6, 612438	602662	242	100	100	99
TUBG1	Cortical dysplasia, complex, with other brain malformations 4, 615412	191135	190	100	100	100
TWNK	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Perrault syndrome 5, 616138 Progressive external ophthalmoplegia with mitochondrial DNA deletions 3, 609286	606075	145	100	100	100
TYROBP	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1, 221770	604142	108	100	100	100
UBA5	Epileptic encephalopathy, early infantile, 44, 617132 ?Spinocerebellar ataxia 24, 617133	610552	46	100	91	67
UBAP1	Spastic paraplegia 80, 618418	609787	73	98	92	86
UBQLN2	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857	300264	79	100	100	100
UCHL1	{?Parkinson disease 5, susceptibility to}, 613643 Spastic paraplegia 79, 615491	191342	79	100	100	99
VAMP1	Myasthenic syndrome, congenital, 25, 618323 Spastic ataxia 1, 108600	185880	104	100	100	100
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	90	100	100	98
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050	192977	70	100	100	97
VPS13A	Choreoacanthocytosis, 200150	605978	72	100	99	91
VPS13C	Parkinson disease 23, early onset, 616840	608879	50	100	97	85
VPS13D	Spinocerebellar ataxia 4, 607317	608877	64	100	98	92
VPS35	{Parkinson disease 17}, 614203	601501	68	100	99	94

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VPS37A	Spastic paraplegia 53, 614898	609927	46	100	99	86
VPS53	Pontocerebellar hypoplasia, type 2E, 615851	615850	82	100	100	96
VRK1	Pontocerebellar hypoplasia type 1A, 607596	602168	53	100	99	92
VWA3B	?Spinocerebellar ataxia 22, 616948	614884	66	100	98	92
WASHC5	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, 603563	610657	62	100	100	94
WDR26	Skraban-Deardorff syndrome, 617616	617424	50	100	94	81
WDR45	Neurodegeneration with brain iron accumulation 5, 300894	300526	92	100	100	100
WDR73	Galloway-Mowat syndrome 1, 251300	616144	152	100	100	97
WDR81	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 Hydrocephalus, congenital, 3, with brain anomalies, 617967	614218	155	100	100	100
WFS1	?Cataract 41, 116400 Deafness 6/14/38, 600965 {Diabetes mellitus, noninsulin-dependent, association with}, 125853 Wolfram syndrome 1, 222300 Wolfram-like syndrome, 614296	606201	196	100	100	100
WWOX	Epileptic encephalopathy, early infantile, 28, 616211 Esophageal squamous cell carcinoma, somatic, 133239 Spinocerebellar ataxia 12, 614322	605131	92	100	100	100
XK	McLeod syndrome with or without chronic granulomatous disease, 300842	314850	62	100	100	96
XPR1	Basal ganglia calcification, idiopathic, 6, 616413	605237	64	100	100	99
XRCC1	?Spinocerebellar ataxia 26, 617633	194360	97	100	100	100
ZC4H2	Wieacker-Wolff syndrome, 314580	300897	78	100	99	93
ZFYVE26	Spastic paraplegia 15, 270700	612012	90	100	100	98
ZFYVE27	Spastic paraplegia 33, 610244	610243	99	100	100	100

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "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 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x