Ataxia Spasticity

Gene panel

Gene panel information

Gene panel	**Ataxia Spasticity**
Version	4
Total genes	524
Activation date	Friday 21 march 2025
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AAAS	99.88 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AARS1	99.99 %	601065	Developmental and epileptic encephalopathy 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant; ?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661 (3), Autosomal dominant; Trichothiodystrophy 8, nonphotosensitive, 619691 (3), Autosomal recessive
ABCA2	100 %	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
ABCB7	99.58 %	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked
ABCD1	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
ABHD12	99.98 %	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ABHD16A	100 %	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
ACBD6	98.86 %	616352	Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive
ACER3	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
ACO2	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
ADAR	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADCY5	99.98 %	600293	Dyskinesia with orofacial involvement, autosomal dominant, 606703 (3), Autosomal dominant; Neurodevelopmental disorder with hyperkinetic movements and dyskinesia, 619651 (3), Autosomal recessive; Dyskinesia with orofacial involvement, autosomal recessive, 619647 (3), Autosomal recessive
ADGRG1	99.9 %	604110	Cortical dysplasia, complex, with other brain malformations 14B, (bilateral perisylvian), 615752 (3); Cortical dysplasia, complex, with other brain malformations 14A, (bilateral frontoparietal), 606854 (3), Autosomal recessive
ADPRS	99.94 %	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
AFG3L2	99.97 %	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3), Autosomal dominant; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
AGTPBP1	99.68 %	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
AHI1	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive

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AIFM1	99.92 %	300169	Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive
AIMP1	99.97 %	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
ALDH18A1	99.96 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
ALDH3A2	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALDH5A1	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
ALG6	93.37 %	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
ALS2	99.87 %	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive
AMFR	99.74 %	603243	Spastic paraplegia 89, autosomal recessive, 620379 (3), Autosomal recessive
AMPD2	99.91 %	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive
ANO10	99.93 %	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
AP1S2	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
AP4B1	96.92 %	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
AP4E1	99.94 %	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
AP4M1	99.98 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
AP4S1	87.89 %	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
AP5Z1	100 %	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
APOB	99.99 %	107730	Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
APTX	99.92 %	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
ARG1	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
ARL13B	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL6IP1	99.51 %	607669	Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
ARSA	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
ARX	95.36 %	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ASPA	99.98 %	608034	Canavan disease, 271900 (3), Autosomal recessive
ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
ATCAY	100 %	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
ATG5	99.81 %	604261	?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive
ATG7	99.9 %	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive
ATL1	99.95 %	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant

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Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ATM	99.83 %	607585	Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Somatic mutation, Autosomal dominant; T-cell prolymphocytic leukemia, somatic (3); Lymphoma, mantle cell, somatic (3)
ATP13A2	99.96 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
ATP1A2	99.85 %	182340	Developmental and epileptic encephalopathy 98, 619605 (3), Autosomal dominant; Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies, 619602 (3), Autosomal recessive; Alternating hemiplegia of childhood 1, 104290 (3), Autosomal dominant; Migraine, familial basilar, 602481 (3), Autosomal dominant; Migraine, familial hemiplegic, 2, 602481 (3), Autosomal dominant
ATP1A3	99.98 %	182350	Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant; CAPOS syndrome, 601338 (3), Autosomal dominant; Developmental and epileptic encephalopathy 99, 619606 (3), Autosomal dominant
ATP2B3	99.98 %	300014	?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive
ATP5MC3	99.97 %	602736	Dystonia, early-onset, and/or spastic paraplegia, 619681 (3), Autosomal dominant
ATP6V0A1	99.85 %	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant
ATP7B	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
ATP8A2	100 %	605870	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
AUH	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
B3GALNT2	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
B4GALNT1	99.97 %	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
B4GAT1	100 %	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BCAS3	99.31 %	607470	Hengel-Maroofian-Schols syndrome, 619641 (3), Autosomal recessive
BCKDHA	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
BCKDHB	99.73 %	248611	Maple syrup urine disease, type Ib, 620698 (3), Autosomal recessive
BCL11B	100 %	606558	Immunodeficiency 49, severe combined, 617237 (3), Autosomal dominant; Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant
BCS1L	99.99 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
BICD2	99.99 %	609797	Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant, 618291 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant, 615290 (3), Autosomal dominant
BORCS8	99.93 %	616601	*No OMIM phenotypes*
BSCL2	99.99 %	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BTD	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
C12orf57	100 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive

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C19orf12	99.99 %	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
CA8	99.71 %	114815	Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive
CACNA1A	98.16 %	601011	Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Developmental and epileptic encephalopathy 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant
CACNA1E	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
CACNA1G	99.95 %	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
CACNA2D2	99.99 %	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
CACNB4	99.2 %	601949	{Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; ?Episodic ataxia, type 5, 613855 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant
CAMTA1	99.97 %	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
CAPN1	99.99 %	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
CAPRIN1	99.56 %	601178	Neurodevelopmental disorder with language impairment, autism, and attention deficit-hyperactivity disorder, 620782 (3), Autosomal dominant; Neurodegeneration, childhood-onset, with cerebellar ataxia and cognitive decline, 620636 (3), Autosomal dominant
CASK	98.95 %	300172	Intellectual developmental disorder, with or without nystagmus, 300422 (3), X-linked recessive; Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia, 300749 (3), X-linked; FG syndrome 4, 300422 (3), X-linked recessive
CC2D2A	99.95 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCDC88C	100 %	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
CCT5	99.99 %	610150	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
CEP290	98.1 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CHMP1A	100 %	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
CHP1	99.9 %	606988	?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive
CLCN2	100 %	600570	Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant
CLDN11	100 %	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
CLN5	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive
CLN6	100 %	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive

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CLP1	99.98 %	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
CNTNAP1	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
COA7	99.91 %	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
COA8	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
COASY	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
COG5	99.92 %	606821	Congenital disorder of glycosylation, type IIi, 613612 (3), Autosomal recessive
COQ2	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
COQ4	100 %	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive
COQ8A	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
COQ9	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
COX10	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
COX15	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
COX20	99.67 %	614698	Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive
CP	99.95 %	117700	Aceruloplasminemia, 604290 (3), Autosomal recessive
CPT1C	99.99 %	608846	?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant
CRPPA	99.98 %	614631	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643 (3), Autosomal recessive
CSTB	100 %	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
CTBP1	99.98 %	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
CTNNB1	99.95 %	116806	Exudative vitreoretinopathy 7, 617572 (3), Autosomal dominant; Pilomatricoma, somatic, 132600 (3); Colorectal cancer, somatic, 114500 (3); Neurodevelopmental disorder with spastic diplegia and visual defects, 615075 (3), Autosomal dominant; Medulloblastoma, somatic, 155255 (3); Ovarian cancer, somatic, 167000 (3); Hepatocellular carcinoma, somatic, 114550 (3)
CWF19L1	99.91 %	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP2U1	99.99 %	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
CYP7B1	99.82 %	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
DAB1	97.87 %	603448	Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant
DAG1	100 %	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818 (3), Autosomal recessive
DAGLA	99.93 %	614015	Neuroocular syndrome 2, paroxysmal type, 168885 (3), Autosomal dominant
DARS1	98.85 %	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive
DARS2	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
DBT	94.51 %	248610	Maple syrup urine disease, type II, 620699 (3), Autosomal recessive

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DDHD1	99.93 %	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
DDHD2	99.97 %	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
DDX3X	99.01 %	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
DEGS1	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
DHDDS	98.65 %	608172	Developmental delay and seizures with or without movement abnormalities, 617836 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1bb, 613861 (3), Autosomal recessive; Retinitis pigmentosa 59, 613861 (3), Autosomal recessive
DLAT	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
DLD	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
DNAJC19	99.76 %	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
DNAJC3	99.92 %	601184	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
DNAJC5	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
DNMT1	99.13 %	126375	Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant; Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant
DOCK3	99.96 %	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
DPYSL5	99.93 %	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
DSTYK	99.83 %	612666	Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant
DYNC1H1	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant
EARS2	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
EBF3	99.99 %	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
ECHS1	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
EEF2	99.95 %	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
EEFSEC	99.96 %	607695	*No OMIM phenotypes*
EIF2AK1	99.9 %	613635	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3), Autosomal dominant
EIF2B1	99.98 %	606686	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive
EIF2B2	99.9 %	606454	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3), Autosomal recessive
EIF2B3	97.26 %	606273	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3), Autosomal recessive
EIF2B4	99.96 %	606687	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3), Autosomal recessive
EIF2B5	99.98 %	603945	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3), Autosomal recessive
ELOVL1	100 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant, Autosomal recessive

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ELOVL4	99.91 %	605512	Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 (3), Autosomal recessive
ELOVL5	99.87 %	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant
EMC1	99.85 %	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
ENTPD1	99.98 %	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
EPM2A	99.99 %	607566	Myoclonic epilepsy of Lafora 1, 254780 (3), Autosomal recessive
ERCC1	99.96 %	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
ERCC4	99.92 %	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
ERCC8	99.79 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
ERLIN1	99.98 %	611604	Spastic paraplegia 62, autosomal recessive, 615681 (3), Autosomal recessive
ERLIN2	99.94 %	611605	Spastic paraplegia 18A, autosomal dominant, 620512 (3), Autosomal dominant; Spastic paraplegia 18B, autosomal recessive, 611225 (3), Autosomal recessive
EXOSC3	100 %	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
EXOSC5	99.98 %	606492	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive
EXOSC8	99.91 %	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
EXOSC9	94.91 %	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
FA2H	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
FAR1	99.82 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
FARS2	100 %	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
FASTKD2	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
FAT2	99.99 %	604269	Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant
FBXL4	100 %	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
FBXO7	99.98 %	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
FDXR	99.99 %	103270	Multiple mitochondrial dysfunctions syndrome 9B, 620887 (3); Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive
FGF14	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant
FICD	100 %	620875	Spastic paraplegia 92, autosomal recessive, 620911 (3), Autosomal recessive
FITM2	99.99 %	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
FKRP	100 %	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without impaired intellectual development), type B, 5, 606612 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153 (3), Autosomal recessive
FKTN	99.94 %	607440	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800 (3), Autosomal recessive; Cardiomyopathy, dilated, 1X, 611615 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital without impaired intellectual development), type B, 4, 613152 (3), Autosomal recessive

Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FLVCR1	99.91 %	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
FOLR1	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
FRMD4A	100 %	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
FRMD5	100 %	616309	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 (3), Autosomal dominant
FRMD7	99.97 %	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
GAD1	99.92 %	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
GALC	99.92 %	606890	Krabbe disease, 245200 (3), Autosomal recessive
GAN	99.98 %	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
GBA2	99.99 %	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive
GBE1	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
GCH1	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
GDAP2	95.02 %	618128	Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive
GEMIN5	99.96 %	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
GFAP	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
GFM2	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GJC2	100 %	608803	Lymphatic malformation 3, 613480 (3), Autosomal dominant; ?Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive
GLB1	100 %	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive
GLRA1	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
GLRB	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
GLRX5	100 %	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
GLS	99.78 %	138280	Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Developmental and epileptic encephalopathy 71, 618328 (3), Autosomal recessive
GM2A	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive
GMPPB	100 %	615320	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 14, 615351 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GOSR2	98.92 %	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive
GPAA1	100 %	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
GPT2	99.97 %	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
GRID2	99.97 %	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
GRIN1	100 %	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Developmental and epileptic encephalopathy 101, 619814 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant
GRM1	100 %	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
GRN	100 %	138945	Frontotemporal dementia 2, 607485 (3), Autosomal dominant, Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant, Autosomal recessive; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
GSX2	100 %	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
HACE1	99.76 %	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
HECTD4	99.96 %	620209	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive
HEXA	99.99 %	606869	[Hex A pseudodeficiency], 272800 (3), Autosomal recessive; GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
HEXB	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
HIKESHI	99.78 %	614908	Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive
HMBS	99.97 %	609806	Leukoencephalopathy, porphyria-related, 620711 (3), Autosomal recessive; Encephalopathy, porphyria-related, 620704 (3), Autosomal recessive; Porphyria, acute intermittent, nonerythroid variant, 176000 (3), Autosomal dominant; Porphyria, acute intermittent, 176000 (3), Autosomal dominant
HPDL	99.99 %	618994	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities, 619026 (3), Autosomal recessive; Spastic paraplegia 83, autosomal recessive, 619027 (3), Autosomal recessive
HSD17B10	99.98 %	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
HSD17B4	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSPD1	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
IBA57	100 %	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
IFIH1	99.84 %	606951	Immunodeficiency 95, 619773 (3), Autosomal recessive; Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
IFRD1	99.67 %	603502	*No OMIM phenotypes*
IFT140	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive

Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
INPP5E	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INTS11	100 %	611354	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive
INTS8	99.89 %	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive
IRF2BPL	99.21 %	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
ISCA1	99.79 %	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
ITM2B	99.85 %	603904	?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant
ITPR1	99.98 %	147265	Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant
JAM2	91.82 %	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
KCNA1	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
KCNA2	99.99 %	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
KCNA4	100 %	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
KCNC3	99.98 %	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
KCND3	99.98 %	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
KCNJ10	99.98 %	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
KCNJ6	100 %	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
KCNMA1	99.89 %	600150	{Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Liang-Wang syndrome, 618729 (3), Autosomal dominant
KCNN2	91.25 %	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
KCNQ2	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
KCNQ3	99.98 %	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
KCTD7	99.98 %	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
KDM5C	99.98 %	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive
KIDINS220	99.94 %	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogryposis, 619501 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KIF1A	99.96 %	601255	NESCAV syndrome, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant; Spastic paraplegia 30, autosomal recessive, 620607 (3), Autosomal recessive
KIF1C	99.99 %	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
KIF5A	99.91 %	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
KLC2	100 %	611729	Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive
KPNA3	99.89 %	601892	Spastic paraplegia 88, autosomal dominant, 620106 (3), Autosomal dominant
L1CAM	99.98 %	308840	MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus, congenital, X-linked, 307000 (3), X-linked recessive; ?Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive
L2HGDH	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
LAMA1	99.98 %	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
LAMB1	99.87 %	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
LARGE1	100 %	603590	Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6, 608840 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154 (3), Autosomal recessive
LARS2	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
LETM1	99.97 %	604407	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive
LIG3	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
LIPT2	99.99 %	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
LNPK	92.81 %	610236	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive
LYRM7	99.98 %	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MAG	99.99 %	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
MAN2B1	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
MAPK8IP3	100 %	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
MARS1	99.97 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MARS2	100 %	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
MCOLN1	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MECP2	99.95 %	300005	Rett syndrome, atypical, 312750 (3), X-linked dominant; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Intellectual developmental disorder, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Intellectual developmental disorder, X-linked syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant
MFSD8	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
MINPP1	99.73 %	605391	{Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive
MLC1	99.99 %	605908	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive
MMADHC	99.76 %	611935	Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive
MME	97.17 %	120520	?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal dominant, Autosomal recessive
MORC2	100 %	616661	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688 (3), Autosomal dominant; Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy, 619090 (3), Autosomal dominant
MPV17	99.98 %	137960	Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive
MRE11	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MRPS34	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
MSTO1	76.34 %	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
MTCL1	100 %	615766	*No OMIM phenotypes*
MTFMT	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
MTPAP	99.97 %	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
MTRFR	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
MTTP	99.92 %	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYORG	99.99 %	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
NAA60	99.99 %	614246	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 (3), Autosomal recessive
NANS	100 %	605202	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive
NAXE	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
NDUFA12	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
NDUFA9	100 %	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
NDUFAF4	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
NDUFAF6	99.86 %	612392	Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive; Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive

Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NDUFS1	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
NDUFS3	100 %	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
NDUFS7	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
NEFL	100 %	162280	Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
NEU1	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
NEXMIF	99.99 %	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
NF2	100 %	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3)
NFASC	99.94 %	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
NHLRC1	100 %	608072	Myoclonic epilepsy of Lafora 2, 620681 (3), Autosomal recessive
NIPA1	99.91 %	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant
NKX2-1	100 %	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NKX6-2	100 %	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
NOL3	100 %	605235	?Myoclonus, familial, 1, 614937 (3), Autosomal dominant
NPC1	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
NPC2	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
NPTX1	100 %	602367	Spinocerebellar ataxia 50, 620158 (3), Autosomal dominant
NRCAM	99.82 %	601581	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive
NSRP1	99.96 %	616173	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive
NT5C2	99.96 %	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
NTNG2	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
NUP93	99.87 %	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
NUS1	99.9 %	610463	Intellectual developmental disorder, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive
OCLN	82.91 %	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
OFD1	99.68 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
OGDHL	99.95 %	617513	Yoon-Bellen neurodevelopmental syndrome, 619701 (3), Autosomal recessive
OPA1	99.95 %	605290	Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Behr syndrome, 210000 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
OPA3	100 %	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
OPHN1	99.92 %	300127	Intellectual developmental disorder, X-linked syndromic, Billuart type, 300486 (3), X-linked recessive
OTC	99.42 %	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked
PACS2	99.99 %	610423	Developmental and epileptic encephalopathy 66, 618067 (3), Autosomal dominant
PARS2	99.99 %	612036	Developmental and epileptic encephalopathy 75, 618437 (3), Autosomal recessive
PC	99.99 %	608786	Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive
PCDH12	100 %	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive
PCYT2	100 %	602679	Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive
PDHA1	99.04 %	300502	Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant
PDHX	99.64 %	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
PDYN	100 %	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
PEX10	100 %	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
PEX16	99.94 %	603360	Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive; Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive
PEX2	100 %	170993	Peroxisome biogenesis disorder 5A (Zellweger), 614866 (3), Autosomal recessive; Peroxisome biogenesis disorder 5B, 614867 (3), Autosomal recessive
PEX6	99.99 %	601498	Peroxisome biogenesis disorder 4B, 614863 (3), Autosomal dominant, Autosomal recessive; Peroxisome biogenesis disorder 4A (Zellweger), 614862 (3), Autosomal recessive; Heimler syndrome 2, 616617 (3), Autosomal recessive
PEX7	99.72 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PGAP1	99.56 %	611655	Neurodevelopmental disorder with dysmorphic features, spasticity, and brain abnormalities, 615802 (3), Autosomal recessive
PHGDH	99.79 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHYH	100 %	602026	Refsum disease, 266500 (3), Autosomal recessive
PI4KA	99.76 %	600286	Spastic paraplegia 84, autosomal recessive, 619621 (3), Autosomal recessive; Gastrointestinal defects and immunodeficiency syndrome 2, 619708 (3), Autosomal recessive; Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531 (3), Autosomal recessive
PIK3R5	99.99 %	611317	Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive
PITRM1	99.89 %	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
PLA2G6	99.98 %	603604	Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive
PLAA	99.79 %	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive
PLD3	99.99 %	615698	?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant
PLP1	99.98 %	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
PMM2	99.93 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
PMPCA	99.99 %	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive
PMPCB	99.94 %	603131	Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PNKD	100 %	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
PNKP	100 %	605610	?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive
PNPLA6	99.99 %	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
PNPT1	99.56 %	610316	Spinocerebellar ataxia 25, 608703 (3), Autosomal dominant; Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 13, 614932 (3), Autosomal recessive
POLG	100 %	174763	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
POLG2	99.51 %	604983	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 16 (hepatic type), 618528 (3), Autosomal recessive; ?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type), 619425 (3), Autosomal recessive
POLR3A	99.97 %	614258	Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive; Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POLR3B	99.94 %	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 (3), Autosomal dominant
POLR3K	100 %	606007	Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive
POMGNT1	99.69 %	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 3, 613151 (3), Autosomal recessive; Retinitis pigmentosa 76, 617123 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280 (3), Autosomal recessive
POMGNT2	100 %	614828	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 8, 614830 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8, 618135 (3), Autosomal recessive
POMT1	99.96 %	607423	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155 (3), Autosomal recessive
POMT2	99.98 %	607439	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150 (3), Autosomal recessive; Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 2, 613156 (3), Autosomal recessive
POU4F1	99.71 %	601632	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 (3), Autosomal dominant
PPFIBP1	99.11 %	603141	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PRDM13	99.99 %	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
PRDX3	99.94 %	604769	Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant
PRF1	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
PRICKLE1	99.87 %	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
PRKCG	99.99 %	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
PRNP	100 %	176640	Spongiform encephalopathy with neuropsychiatric features, 606688 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant; Huntington disease-like 1, 603218 (3), Autosomal dominant; Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant
PRPS1	99.95 %	311850	Arts syndrome, 301835 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Gout, PRPS-related, 300661 (3), X-linked recessive
PRRT2	99.97 %	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant
PSAP	99.94 %	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); {Parkinson disease 24, autosomal dominant, susceptibility to}, 619491 (3), Autosomal dominant
PSEN1	100 %	104311	Pick disease, 172700 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant
PTF1A	100 %	607194	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive
PTPN23	100 %	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
PTRH2	99.99 %	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
PUM1	98.14 %	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant; Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719 (3), Autosomal dominant
PYCR2	99.95 %	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
RAB11B	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
RAB18	99.76 %	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
RAB1A	99.93 %	179508	*No OMIM phenotypes*
RAB3GAP1	99.73 %	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAB3GAP2	99.69 %	609275	Martsolf syndrome 1, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
RARS2	99.88 %	611524	Pontocerebellar hypoplasia, type 6, 611523 (3), Autosomal recessive
REEP1	99.97 %	609139	Neuronopathy, distal hereditary motor, autosomal recessive 6, 620011 (3), Autosomal recessive; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, autosomal dominant 12, 614751 (3), Autosomal dominant
REEP2	99.99 %	609347	Spastic paraplegia 72A, autosomal dominant, 615625 (3), Autosomal dominant; ?Spastic paraplegia 72B, autosomal recessive, 620606 (3), Autosomal recessive
RETREG1	99.99 %	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
RFC1	99.87 %	102579	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575 (3), Autosomal recessive
RINT1	99.99 %	610089	Infantile liver failure syndrome 3, 618641 (3), Autosomal recessive
RNASEH2A	99.95 %	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
RNASEH2B	99.94 %	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
RNASEH2C	99.99 %	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
RNF168	99.97 %	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
RNF170	99.9 %	614649	Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant; Spastic paraplegia 85, autosomal recessive, 619686 (3), Autosomal recessive
RNF216	99.99 %	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RNF220	99.68 %	616136	Leukodystrophy, hypomyelinating, 23, with ataxia, deafness, liver dysfunction, and dilated cardiomyopathy, 619688 (3), Autosomal recessive
RNU7-1	33.9 %	617876	Aicardi-Goutieres syndrome 9, 619487 (3), Autosomal recessive
RORA	99.97 %	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant
RPGRIP1L	96.35 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RTN2	99.98 %	603183	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity, 620854 (3), Autosomal recessive; Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant
RTN4IP1	99.98 %	610502	Optic atrophy 10 with or without ataxia, impaired intellectual development and seizures, 616732 (3), Autosomal recessive
RUBCN	100 %	613516	Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
RXYLT1	99.48 %	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041 (3), Autosomal recessive
SACS	99.97 %	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
SAMD9L	99.95 %	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant; ?Spinocerebellar ataxia 49, 619806 (3), Autosomal dominant; Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 (3), Autosomal dominant
SAMHD1	99.98 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SARS1	98.53 %	607529	Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SCN1A	99.94 %	182389	Developmental and epileptic encephalopathy 6B, non-Dravet, 619317 (3), Autosomal dominant; Migraine, familial hemiplegic, 3, 609634 (3), Autosomal dominant; Dravet syndrome, 607208 (3), Autosomal dominant; Febrile seizures, familial, 3A, 604403 (3), Autosomal dominant; Generalized epilepsy with febrile seizures plus, type 2, 604403 (3), Autosomal dominant
SCN2A	99.86 %	182390	Seizures, benign familial infantile, 3, 607745 (3), Autosomal dominant; Developmental and epileptic encephalopathy 11, 613721 (3), Autosomal dominant; Episodic ataxia, type 9, 618924 (3), Autosomal dominant
SCN8A	99.77 %	600702	?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Developmental and epileptic encephalopathy 13, 614558 (3), Autosomal dominant
SCYL1	100 %	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
SDHA	99.98 %	600857	Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Mitochondrial complex II deficiency, nuclear type 1, 252011 (3), Autosomal recessive; Neurodegeneration with ataxia and late-onset optic atrophy, 619259 (3), Autosomal dominant; Pheochromocytoma/paraganglioma syndrome 5, 614165 (3), Autosomal dominant
SDHAF1	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
SDHD	82.93 %	602690	Pheochromocytoma/paraganglioma syndrome 1, 168000 (3), Autosomal dominant; Paraganglioma and gastric stromal sarcoma, 606864 (3); Mitochondrial complex II deficiency, nuclear type 3, 619167 (3), Autosomal recessive
SELENOI	99.86 %	607915	Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive
SEPSECS	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
SERAC1	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
SETX	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SIL1	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SLC13A5	99.99 %	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive
SLC16A2	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
SLC17A5	99.71 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
SLC19A3	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive
SLC1A3	99.98 %	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
SLC1A4	99.97 %	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
SLC25A15	100 %	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
SLC25A46	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
SLC2A1	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SLC33A1	99.67 %	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive
SLC44A1	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
SLC52A2	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	99.94 %	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
SLC6A8	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
SLC9A1	99.96 %	107310	Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
SLC9A6	99.42 %	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SMPD4	99.9 %	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive
SNAP25	99.89 %	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
SNAPC4	100 %	602777	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 (3), Autosomal recessive
SNX14	99.73 %	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
SOX10	100 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
SPART	99.98 %	607111	Troyer syndrome, 275900 (3), Autosomal recessive
SPAST	99.77 %	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPATA5L1	99.91 %	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
SPG11	99.89 %	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPG21	99.97 %	608181	Mast syndrome, 248900 (3), Autosomal recessive
SPG7	99.99 %	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive
SPR	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
SPTAN1	99.96 %	182810	Developmental delay with or without epilepsy, 620540 (3), Autosomal dominant; Developmental and epileptic encephalopathy 5, 613477 (3), Autosomal dominant; Spastic paraplegia 91, autosomal dominant, with or without cerebellar ataxia, 620538 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, autosomal dominant 11, 620528 (3), Autosomal dominant
SPTBN2	99.98 %	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
SPTSSA	99.88 %	613540	Spastic paraplegia 90A, autosomal dominant, 620416 (3), Autosomal dominant; ?Spastic paraplegia 90B, autosomal recessive, 620417 (3), Autosomal dominant
SQSTM1	100 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SRD5A3	99.94 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
STUB1	99.99 %	607207	Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
SUCLA2	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
SUFU	100 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal dominant, Autosomal recessive
SUOX	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
SURF1	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
SVBP	99.04 %	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
SYNE1	99.95 %	608441	Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
SYT14	99.94 %	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
TAF8	99.91 %	609514	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive
TANGO2	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
TBC1D20	100 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TBC1D23	98.7 %	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
TDP1	99.97 %	607198	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive
TDP2	99.97 %	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
TECPR2	99.96 %	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
TENM3	99.99 %	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
TFG	98.68 %	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
TGM6	99.99 %	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
THG1L	99.99 %	618802	Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TMEM106B	99.92 %	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
TMEM216	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM231	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM240	99.99 %	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
TMEM63B	99.95 %	619952	*No OMIM phenotypes*
TMEM63C	99.99 %	619953	Spastic paraplegia 87, autosomal recessive, 619966 (3), Autosomal recessive

Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TMEM67	99.69 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TMX2	99.99 %	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
TOE1	99.96 %	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
TPP1	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
TRAK1	99.98 %	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
TRAPPC12	99.96 %	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
TRAPPC4	100 %	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
TRIT1	99.25 %	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
TRPC3	99.97 %	602345	?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant
TSEN15	99.57 %	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
TSEN2	99.98 %	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
TSEN54	100 %	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
TTBK2	99.93 %	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
TTC19	99.99 %	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
TTPA	99.88 %	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
TTR	100 %	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
TUBA1A	99.97 %	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
TUBB3	100 %	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
TUBB4A	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
TUBG1	99.95 %	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
TWNK	100 %	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
UBA5	99.95 %	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
UBAP1	99.51 %	609787	Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant
UBTF	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
UCHL1	99.99 %	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant; Spastic paraplegia 79A, autosomal dominant, 620221 (3), Autosomal dominant; Spastic paraplegia 79B, autosomal recessive, 615491 (3), Autosomal recessive

Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
UFC1	99.35 %	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
UFM1	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
UGP2	99.85 %	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
VAMP1	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant
VLDLR	99.99 %	192977	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive
VPS13B	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
VPS13D	99.96 %	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
VPS37A	99.96 %	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
VPS41	99.92 %	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
VPS53	100 %	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
VRK1	99.98 %	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive
VWA3B	98.82 %	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
WARS2	97.72 %	604733	Parkinsonism-dystonia 3, childhood-onset, 619738 (3), Autosomal recessive; Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive
WASHC5	99.98 %	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
WDR45B	100 %	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
WDR73	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
WDR81	100 %	614218	Cerebellar ataxia, impaired intellectual development, and dysquilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive
WWOX	100 %	605131	Esophageal squamous cell carcinoma, somatic, 133239 (3); Developmental and epileptic encephalopathy 28, 616211 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
XRCC1	99.96 %	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
ZC4H2	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
ZFYVE26	100 %	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.