

# Ataxia Spasticity

Gene panel

## Gene panel information

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

## Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>AAAS</b>	99.88 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
<b>AARS1</b>	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
<b>ABCA2</b>	100 %	600047	Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive
<b>ABCB7</b>	99.58 %	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked
<b>ABCD1</b>	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<b>ABHD12</b>	99.98 %	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<b>ABHD16A</b>	100 %	142620	Spastic paraplegia 86, autosomal recessive, 619735 (3), Autosomal recessive
<b>ACBD6</b>	98.86 %	616352	Neurodevelopmental disorder with progressive movement abnormalities, 620785 (3), Autosomal recessive
<b>ACER3</b>	99.76 %	617036	?Leukodystrophy, progressive, early childhood-onset, 617762 (3), Autosomal recessive
<b>ACO2</b>	99.99 %	100850	Optic atrophy 9, 616289 (3), Autosomal dominant, Autosomal recessive; Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive
<b>ADAR</b>	99.84 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
<b>ADCY5</b>	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
<b>ADGRG1</b>	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
<b>ADPRS</b>	99.94 %	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive
<b>AFG3L2</b>	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
<b>AGTPBP1</b>	99.68 %	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive
<b>AHI1</b>	99.86 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive

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<b>AIFM1</b>	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
<b>AIMP1</b>	99.97 %	603605	Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive
<b>ALDH18A1</b>	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
<b>ALDH3A2</b>	99.95 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
<b>ALDH5A1</b>	96.19 %	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<b>ALG6</b>	93.37 %	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
<b>ALS2</b>	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
<b>AMFR</b>	99.74 %	603243	Spastic paraplegia 89, autosomal recessive, 620379 (3), Autosomal recessive
<b>AMPD2</b>	99.91 %	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, autosomal recessive, 615686 (3), Autosomal recessive
<b>ANO10</b>	99.93 %	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<b>AP1S2</b>	99.56 %	300629	Pettigrew syndrome, 304340 (3), X-linked recessive
<b>AP4B1</b>	96.92 %	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<b>AP4E1</b>	99.94 %	607244	Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive
<b>AP4M1</b>	99.98 %	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<b>AP4S1</b>	87.89 %	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive
<b>AP5Z1</b>	100 %	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
<b>APOB</b>	99.99 %	107730	Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant; Hypobetalipoproteinemia, 615558 (3), Autosomal recessive
<b>APTX</b>	99.92 %	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<b>ARG1</b>	99.95 %	608313	Argininemia, 207800 (3), Autosomal recessive
<b>ARL13B</b>	99.53 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<b>ARL6IP1</b>	99.51 %	607669	Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
<b>ARSA</b>	99.99 %	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<b>ARX</b>	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
<b>ASPA</b>	99.98 %	608034	Canavan disease, 271900 (3), Autosomal recessive
<b>ATAD3A</b>	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
<b>ATCAY</b>	100 %	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<b>ATG5</b>	99.81 %	604261	?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive
<b>ATG7</b>	99.9 %	608760	Spinocerebellar ataxia, autosomal recessive 31, 619422 (3), Autosomal recessive

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Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>ATL1</b>	99.95 %	606439	Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant
<b>ATM</b>	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)
<b>ATP13A2</b>	99.96 %	610513	Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive; Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive
<b>ATP1A2</b>	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
<b>ATP1A3</b>	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
<b>ATP2B3</b>	99.98 %	300014	?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive
<b>ATP5MC3</b>	99.97 %	602736	Dystonia, early-onset, and/or spastic paraplegia, 619681 (3), Autosomal dominant
<b>ATP6V0A1</b>	99.85 %	192130	Neurodevelopmental disorder with epilepsy and brain atrophy, 619971 (3), Autosomal recessive; Developmental and epileptic encephalopathy 104, 619970 (3), Autosomal dominant
<b>ATP7B</b>	100 %	606882	Wilson disease, 277900 (3), Autosomal recessive
<b>ATP8A2</b>	100 %	605870	Cerebellar ataxia, impaired intellectual development, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<b>AUH</b>	99.95 %	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<b>B3GALNT2</b>	92.79 %	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11, 615181 (3), Autosomal recessive
<b>B4GALNT1</b>	99.97 %	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<b>B4GAT1</b>	100 %	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287 (3), Autosomal recessive
<b>BBS1</b>	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
<b>BCAS3</b>	99.31 %	607470	Hengel-Marooftan-Schols syndrome, 619641 (3), Autosomal recessive
<b>BCKDHA</b>	99.97 %	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<b>BCKDHB</b>	99.73 %	248611	Maple syrup urine disease, type Ib, 620698 (3), Autosomal recessive
<b>BCL11B</b>	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
<b>BCS1L</b>	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
<b>BICD2</b>	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
<b>BORCS8</b>	99.93 %	616601	<i>No OMIM phenotypes</i>
<b>BSCL2</b>	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

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<b>BTD</b>	100 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<b>C12orf57</b>	100 %	615140	Temtamy syndrome, 218340 (3), Autosomal recessive
<b>C19orf12</b>	99.99 %	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
<b>CA8</b>	99.71 %	114815	Spinocerebellar ataxia, autosomal recessive 34, 613227 (3), Autosomal recessive
<b>CACNA1A</b>	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
<b>CACNA1E</b>	99.82 %	601013	Developmental and epileptic encephalopathy 69, 618285 (3), Autosomal dominant
<b>CACNA1G</b>	99.95 %	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant
<b>CACNA2D2</b>	99.99 %	607082	Cerebellar atrophy with seizures and variable developmental delay, 618501 (3), Autosomal recessive
<b>CACNB4</b>	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
<b>CAMTA1</b>	99.97 %	611501	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756 (3), Autosomal dominant
<b>CAPN1</b>	99.99 %	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
<b>CAPRN1</b>	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
<b>CASK</b>	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
<b>CC2D2A</b>	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
<b>CCDC88C</b>	100 %	611204	?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive
<b>CCT5</b>	99.99 %	610150	?Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
<b>CEP290</b>	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
<b>CHMP1A</b>	100 %	164010	Pontocerebellar hypoplasia, type 8, 614961 (3), Autosomal recessive
<b>CHP1</b>	99.9 %	606988	?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive
<b>CLCN2</b>	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
<b>CLDN11</b>	100 %	601326	Leukodystrophy, hypomyelinating, 22, 619328 (3), Autosomal dominant
<b>CLN5</b>	100 %	608102	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive

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<b>CLN6</b>	100 %	606725	Ceroid lipofuscinosis, neuronal, 6B (Kufs type), 204300 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 6A, 601780 (3), Autosomal recessive
<b>CLP1</b>	99.98 %	608757	Pontocerebellar hypoplasia, type 10, 615803 (3), Autosomal recessive
<b>CNTNAP1</b>	99.98 %	602346	Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive
<b>COA7</b>	99.91 %	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive
<b>COA8</b>	99.94 %	616003	Mitochondrial complex IV deficiency, nuclear type 17, 619061 (3), Autosomal recessive
<b>COASY</b>	99.98 %	609855	Pontocerebellar hypoplasia, type 12, 618266 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 6, 615643 (3), Autosomal recessive
<b>COG5</b>	99.92 %	606821	Congenital disorder of glycosylation, type Iii, 613612 (3), Autosomal recessive
<b>COQ2</b>	99.9 %	609825	{Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive
<b>COQ4</b>	100 %	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive; Spastic ataxia 10, autosomal recessive, 620666 (3), Autosomal recessive
<b>COQ8A</b>	100 %	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<b>COQ9</b>	99.62 %	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<b>COX10</b>	99.99 %	602125	Mitochondrial complex IV deficiency, nuclear type 3, 619046 (3), Autosomal recessive
<b>COX15</b>	100 %	603646	Mitochondrial complex IV deficiency, nuclear type 6, 615119 (3), Autosomal recessive
<b>COX20</b>	99.67 %	614698	Mitochondrial complex IV deficiency, nuclear type 11, 619054 (3), Autosomal recessive
<b>CP</b>	99.95 %	117700	Aceruloplasminemia, 604290 (3), Autosomal recessive
<b>CPT1C</b>	99.99 %	608846	?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant
<b>CRPPA</b>	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
<b>CSTB</b>	100 %	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<b>CTBP1</b>	99.98 %	602618	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant
<b>CTNNB1</b>	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)
<b>CWF19L1</b>	99.91 %	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
<b>CYP27A1</b>	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<b>CYP2U1</b>	99.99 %	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<b>CYP7B1</b>	99.82 %	603711	Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive
<b>DAB1</b>	97.87 %	603448	Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant
<b>DAG1</b>	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
<b>DAGLA</b>	99.93 %	614015	Neuroocular syndrome 2, paroxysmal type, 168885 (3), Autosomal dominant
<b>DARS1</b>	98.85 %	603084	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive

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<b>DARS2</b>	98.31 %	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<b>DBT</b>	94.51 %	248610	Maple syrup urine disease, type II, 620699 (3), Autosomal recessive
<b>DDHD1</b>	99.93 %	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
<b>DDHD2</b>	99.97 %	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<b>DDX3X</b>	99.01 %	300160	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958 (3), X-linked recessive, X-linked dominant
<b>DEGS1</b>	99.99 %	615843	Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive
<b>DHDDS</b>	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
<b>DLAT</b>	99.65 %	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<b>DLD</b>	99.89 %	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<b>DNAJC19</b>	99.76 %	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<b>DNAJC3</b>	99.92 %	601184	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive
<b>DNAJC5</b>	99.99 %	611203	Ceroid lipofuscinosis, neuronal, 4 (Kufs type), autosomal dominant, 162350 (3), Autosomal dominant
<b>DNMT1</b>	99.13 %	126375	Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant; Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant
<b>DOCK3</b>	99.96 %	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive
<b>DPYSL5</b>	99.93 %	608383	Ritscher-Schinzel syndrome 4, 619435 (3), Autosomal dominant
<b>DSTYK</b>	99.83 %	612666	Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant
<b>DYNC1H1</b>	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 20, 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
<b>EARS2</b>	99.96 %	612799	Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive
<b>EBF3</b>	99.99 %	607407	Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant
<b>ECHS1</b>	100 %	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive
<b>EEF2</b>	99.95 %	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
<b>EEFSEC</b>	99.96 %	607695	<i>No OMIM phenotypes</i>
<b>EIF2AK1</b>	99.9 %	613635	?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3), Autosomal dominant
<b>EIF2B1</b>	99.98 %	606686	Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure, 603896 (3), Autosomal recessive
<b>EIF2B2</b>	99.9 %	606454	Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure, 620312 (3), Autosomal recessive
<b>EIF2B3</b>	97.26 %	606273	Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure, 620313 (3), Autosomal recessive
<b>EIF2B4</b>	99.96 %	606687	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure, 620314 (3), Autosomal recessive

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>EIF2B5</b>	99.98 %	603945	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315 (3), Autosomal recessive
<b>ELOVL1</b>	100 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant, Autosomal recessive
<b>ELOVL4</b>	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
<b>ELOVL5</b>	99.87 %	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant
<b>EMC1</b>	99.85 %	616846	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive
<b>ENTPD1</b>	99.98 %	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<b>EPM2A</b>	99.99 %	607566	Myoclonic epilepsy of Lafora 1, 254780 (3), Autosomal recessive
<b>ERCC1</b>	99.96 %	126380	Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive
<b>ERCC4</b>	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
<b>ERCC8</b>	99.79 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
<b>ERLIN1</b>	99.98 %	611604	Spastic paraplegia 62, autosomal recessive, 615681 (3), Autosomal recessive
<b>ERLIN2</b>	99.94 %	611605	Spastic paraplegia 18A, autosomal dominant, 620512 (3), Autosomal dominant; Spastic paraplegia 18B, autosomal recessive, 611225 (3), Autosomal recessive
<b>EXOSC3</b>	100 %	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<b>EXOSC5</b>	99.98 %	606492	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576 (3), Autosomal recessive
<b>EXOSC8</b>	99.91 %	606019	Pontocerebellar hypoplasia, type 1C, 616081 (3), Autosomal recessive
<b>EXOSC9</b>	94.91 %	606180	Pontocerebellar hypoplasia, type 1D, 618065 (3), Autosomal recessive
<b>FA2H</b>	99.98 %	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<b>FAR1</b>	99.82 %	616107	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154 (3), Autosomal recessive; Cataracts, spastic paraparesis, and speech delay, 619338 (3), Autosomal dominant
<b>FARS2</b>	100 %	611592	Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive; Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive
<b>FASTKD2</b>	99.93 %	612322	Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive
<b>FAT2</b>	99.99 %	604269	Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant
<b>FBXL4</b>	100 %	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive
<b>FBXO7</b>	99.98 %	605648	Parkinson disease 15, autosomal recessive, 260300 (3), Autosomal recessive
<b>FDXR</b>	99.99 %	103270	Multiple mitochondrial dysfunctions syndrome 9B, 620887 (3); Auditory neuropathy and optic atrophy, 617717 (3), Autosomal recessive
<b>FGF14</b>	99.99 %	601515	Spinocerebellar ataxia 27A, 193003 (3), Autosomal dominant; Spinocerebellar ataxia 27B, late-onset, 620174 (3), Autosomal dominant
<b>FICD</b>	100 %	620875	Spastic paraplegia 92, autosomal recessive, 620911 (3), Autosomal recessive
<b>FITM2</b>	99.99 %	612029	Siddiqi syndrome, 618635 (3), Autosomal recessive
<b>FKRP</b>	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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>FKTN</b>	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
<b>FLVCR1</b>	99.91 %	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<b>FOLR1</b>	100 %	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<b>FRMD4A</b>	100 %	616305	?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive
<b>FRMD5</b>	100 %	616309	Neurodevelopmental disorder with eye movement abnormalities and ataxia, 620094 (3), Autosomal dominant
<b>FRMD7</b>	99.97 %	300628	Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked; Nystagmus 1, congenital, X-linked, 310700 (3), X-linked
<b>GAD1</b>	99.92 %	605363	Developmental and epileptic encephalopathy 89, 619124 (3), Autosomal recessive
<b>GALC</b>	99.92 %	606890	Krabbe disease, 245200 (3), Autosomal recessive
<b>GAN</b>	99.98 %	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<b>GBA2</b>	99.99 %	609471	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive
<b>GBE1</b>	99.73 %	607839	Glycogen storage disease IV, 232500 (3), Autosomal recessive; Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive
<b>GCH1</b>	99.94 %	600225	Dystonia, DOPA-responsive, 128230 (3), Autosomal dominant, Autosomal recessive; Hyperphenylalaninemia, BH4-deficient, B, 233910 (3), Autosomal recessive
<b>GDAP2</b>	95.02 %	618128	Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive
<b>GEMIN5</b>	99.96 %	607005	Neurodevelopmental disorder with cerebellar atrophy and motor dysfunction, 619333 (3), Autosomal recessive
<b>GFAP</b>	99.99 %	137780	Alexander disease, 203450 (3), Autosomal dominant
<b>GFM2</b>	99.87 %	606544	Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive
<b>GJA1</b>	100 %	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Syndactyly, keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
<b>GJC2</b>	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
<b>GLB1</b>	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
<b>GLRA1</b>	100 %	138491	Hyperekplexia 1, 149400 (3), Autosomal dominant, Autosomal recessive
<b>GLRB</b>	99.79 %	138492	Hyperekplexia 2, 614619 (3), Autosomal recessive
<b>GLRX5</b>	100 %	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
<b>GLS</b>	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
<b>GM2A</b>	100 %	613109	GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive



# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>GMPPB</b>	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
<b>GOSR2</b>	98.92 %	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive; Muscular dystrophy, congenital, with or without seizures, 620166 (3), Autosomal recessive
<b>GPAA1</b>	100 %	603048	Glycosylphosphatidylinositol biosynthesis defect 15, 617810 (3), Autosomal recessive
<b>GPT2</b>	99.97 %	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia, 616281 (3), Autosomal recessive
<b>GRID2</b>	99.97 %	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
<b>GRIN1</b>	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
<b>GRM1</b>	100 %	604473	Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive; Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant
<b>GRN</b>	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
<b>GSX2</b>	100 %	616253	Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive
<b>HACE1</b>	99.76 %	610876	Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive
<b>HECTD4</b>	99.96 %	620209	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum, 620250 (3), Autosomal recessive
<b>HEXA</b>	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
<b>HEXB</b>	99.91 %	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<b>HIKESHI</b>	99.78 %	614908	Leukodystrophy, hypomyelinating, 13, 616881 (3), Autosomal recessive
<b>HMBS</b>	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
<b>HPDL</b>	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
<b>HSD17B10</b>	99.98 %	300256	HSD10 mitochondrial disease, 300438 (3), X-linked dominant
<b>HSD17B4</b>	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<b>HSPD1</b>	83.42 %	118190	Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive
<b>IBA57</b>	100 %	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
<b>IFIH1</b>	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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>IFRD1</b>	99.67 %	603502	<i>No OMIM phenotypes</i>
<b>IFT140</b>	100 %	614620	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive
<b>INPP5E</b>	99.85 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
<b>INTS11</b>	100 %	611354	Neurodevelopmental disorder with motor and language delay, ocular defects, and brain abnormalities, 620428 (3), Autosomal recessive
<b>INTS8</b>	99.89 %	611351	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive
<b>IRF2BPL</b>	99.21 %	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant
<b>ISCA1</b>	99.79 %	611006	Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive
<b>ITM2B</b>	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
<b>ITPR1</b>	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
<b>JAM2</b>	91.82 %	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive
<b>KCNA1</b>	100 %	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<b>KCNA2</b>	99.99 %	176262	Developmental and epileptic encephalopathy 32, 616366 (3), Autosomal dominant
<b>KCNA4</b>	100 %	176266	Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive
<b>KCNC3</b>	99.98 %	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
<b>KCND3</b>	99.98 %	605411	Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant; Brugada syndrome 9, 616399 (3), Autosomal dominant
<b>KCNJ10</b>	99.98 %	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<b>KCNJ6</b>	100 %	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant
<b>KCNMA1</b>	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
<b>KCNN2</b>	91.25 %	605879	?Dystonia 34, myoclonic, 619724 (3), Autosomal dominant; Neurodevelopmental disorder with or without variable movement or behavioral abnormalities, 619725 (3), Autosomal dominant
<b>KCNQ2</b>	100 %	602235	Developmental and epileptic encephalopathy 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant
<b>KCNQ3</b>	99.98 %	602232	Seizures, benign neonatal, 2, 121201 (3), Autosomal dominant
<b>KCTD7</b>	99.98 %	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<b>KDM5C</b>	99.98 %	314690	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type, 300534 (3), X-linked recessive

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>KIDINS220</b>	99.94 %	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant; Ventriculomegaly and arthrogyrosis, 619501 (3), Autosomal recessive
<b>KIF1A</b>	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
<b>KIF1C</b>	99.99 %	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
<b>KIF5A</b>	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
<b>KLC2</b>	100 %	611729	Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive
<b>KPNA3</b>	99.89 %	601892	Spastic paraplegia 88, autosomal dominant, 620106 (3), Autosomal dominant
<b>L1CAM</b>	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
<b>L2HGDH</b>	99.92 %	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<b>LAMA1</b>	99.98 %	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<b>LAMB1</b>	99.87 %	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<b>LARGE1</b>	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
<b>LARS2</b>	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
<b>LETM1</b>	99.97 %	604407	Neurodegeneration, childhood-onset, with multisystem involvement due to mitochondrial dysfunction, 620089 (3), Autosomal recessive
<b>LIG3</b>	99.99 %	600940	Mitochondrial DNA depletion syndrome 20 (MNGIE type), 619780 (3), Autosomal recessive
<b>LIPT2</b>	99.99 %	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive
<b>LNPK</b>	92.81 %	610236	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum, 618090 (3), Autosomal recessive
<b>LYRM7</b>	99.98 %	615831	Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive
<b>LYST</b>	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<b>MAG</b>	99.99 %	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
<b>MAN2B1</b>	99.99 %	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<b>MAPK8IP3</b>	100 %	605431	Neurodevelopmental disorder with or without variable brain abnormalities, 618443 (3), Autosomal dominant
<b>MARS1</b>	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
<b>MARS2</b>	100 %	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
<b>MCOLN1</b>	100 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucoylipidosis IV, 252650 (3), Autosomal recessive

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>MECP2</b>	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
<b>MFSD8</b>	99.7 %	611124	Macular dystrophy with central cone involvement, 616170 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 7, 610951 (3), Autosomal recessive
<b>MINPP1</b>	99.73 %	605391	{Thyroid carcinoma, follicular}, 188470 (3), Somatic mutation, Autosomal dominant; Pontocerebellar hypoplasia, type 16, 619527 (3), Autosomal recessive
<b>MLC1</b>	99.99 %	605908	Megalencephalic leukoencephalopathy with subcortical cysts 1, 604004 (3), Autosomal recessive
<b>MMADHC</b>	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
<b>MME</b>	97.17 %	120520	?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal dominant, Autosomal recessive
<b>MORC2</b>	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
<b>MPV17</b>	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
<b>MRE11</b>	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
<b>MRPS34</b>	100 %	611994	Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive
<b>MSTO1</b>	76.34 %	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive
<b>MTCL1</b>	100 %	615766	<i>No OMIM phenotypes</i>
<b>MTFMT</b>	99.98 %	611766	Combined oxidative phosphorylation deficiency 15, 614947 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 27, 618248 (3), Autosomal recessive
<b>MTPAP</b>	99.97 %	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
<b>MTRFR</b>	99.87 %	613541	Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive
<b>MTTP</b>	99.92 %	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive
<b>MVK</b>	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
<b>MYORG</b>	99.99 %	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive
<b>NAA60</b>	99.99 %	614246	Basal ganglia calcification, idiopathic, 9, autosomal recessive, 620786 (3), Autosomal recessive
<b>NANS</b>	100 %	605202	Spondyloepimetaphyseal dysplasia, Genevieve type, 610442 (3), Autosomal recessive
<b>NAXE</b>	99.99 %	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186 (3), Autosomal recessive
<b>NDUFA12</b>	99.21 %	614530	Mitochondrial complex I deficiency, nuclear type 23, 618244 (3), Autosomal recessive
<b>NDUFA9</b>	100 %	603834	Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive
<b>NDUFAF4</b>	99.95 %	611776	Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive
<b>NDUFAF6</b>	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
<b>NDUFS1</b>	99.79 %	157655	Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive
<b>NDUFS3</b>	100 %	603846	Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive
<b>NDUFS7</b>	99.99 %	601825	Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive
<b>NEFL</b>	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
<b>NEU1</b>	99.98 %	608272	Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive
<b>NEXMIF</b>	99.99 %	300524	Intellectual developmental disorder, X-linked 98, 300912 (3), X-linked dominant
<b>NF2</b>	100 %	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3)
<b>NFASC</b>	99.94 %	609145	Neurodevelopmental disorder with central and peripheral motor dysfunction, 618356 (3), Autosomal recessive
<b>NHLRC1</b>	100 %	608072	Myoclonic epilepsy of Lafora 2, 620681 (3), Autosomal recessive
<b>NIPA1</b>	99.91 %	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant
<b>NKX2-1</b>	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
<b>NKX6-2</b>	100 %	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive
<b>NOL3</b>	100 %	605235	?Myoclonus, familial, 1, 614937 (3), Autosomal dominant
<b>NPC1</b>	99.99 %	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<b>NPC2</b>	100 %	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<b>NPTX1</b>	100 %	602367	Spinocerebellar ataxia 50, 620158 (3), Autosomal dominant
<b>NRCAM</b>	99.82 %	601581	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities, 619833 (3), Autosomal recessive
<b>NSRP1</b>	99.96 %	616173	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities, 620001 (3), Autosomal recessive
<b>NT5C2</b>	99.96 %	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
<b>NTNG2</b>	99.98 %	618689	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive
<b>NUP93</b>	99.87 %	614351	Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive
<b>NUS1</b>	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
<b>OCLN</b>	82.91 %	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive
<b>OFD1</b>	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
<b>OGDHL</b>	99.95 %	617513	Yoon-Bellen neurodevelopmental syndrome, 619701 (3), Autosomal recessive
<b>OPA1</b>	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

# Ataxia Spasticity

Gene panel

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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>PNKD</b>	100 %	609023	Paroxysmal nonkinesigenic dyskinesia 1, 118800 (3), Autosomal dominant
<b>PNKP</b>	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
<b>PNPLA6</b>	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
<b>PNPT1</b>	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
<b>POLG</b>	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
<b>POLG2</b>	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 (neurophthalmic type), 619425 (3), Autosomal recessive
<b>POLR3A</b>	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
<b>POLR3B</b>	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
<b>POLR3K</b>	100 %	606007	Leukodystrophy, hypomyelinating, 21, 619310 (3), Autosomal recessive
<b>POMGNT1</b>	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
<b>POMGNT2</b>	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
<b>POMT1</b>	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
<b>POMT2</b>	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
<b>POU4F1</b>	99.71 %	601632	Ataxia, intention tremor, and hypotonia syndrome, childhood-onset, 619352 (3), Autosomal dominant
<b>PPFIBP1</b>	99.11 %	603141	Neurodevelopmental disorder with seizures, microcephaly, and brain abnormalities, 620024 (3), Autosomal recessive

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>PRDM13</b>	99.99 %	616741	Pontocerebellar hypoplasia, type 17, 619909 (3), Autosomal recessive; Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761 (3), Autosomal recessive
<b>PRDX3</b>	99.94 %	604769	Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant
<b>PRF1</b>	100 %	170280	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3)
<b>PRICKLE1</b>	99.87 %	608500	Epilepsy, progressive myoclonic 1B, 612437 (3), Autosomal recessive
<b>PRKCG</b>	99.99 %	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
<b>PRNP</b>	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
<b>PRPS1</b>	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
<b>PRRT2</b>	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
<b>PSAP</b>	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
<b>PSEN1</b>	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
<b>PTF1A</b>	100 %	607194	Pancreatic and cerebellar agenesis, 609069 (3), Autosomal recessive; Pancreatic agenesis 2, 615935 (3), Autosomal recessive
<b>PTPN23</b>	100 %	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive
<b>PTRH2</b>	99.99 %	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<b>PUM1</b>	98.14 %	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant; Neurodevelopmental disorder with motor abnormalities, seizures, and facial dysmorphism, 620719 (3), Autosomal dominant
<b>PYCR2</b>	99.95 %	616406	Leukodystrophy, hypomyelinating, 10, 616420 (3), Autosomal recessive
<b>RAB11B</b>	100 %	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant
<b>RAB18</b>	99.76 %	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<b>RAB1A</b>	99.93 %	179508	<i>No OMIM phenotypes</i>
<b>RAB3GAP1</b>	99.73 %	602536	Martsolf syndrome 2, 619420 (3), Autosomal recessive; Warburg micro syndrome 1, 600118 (3), Autosomal recessive



# Ataxia Spasticity

Gene panel

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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>SCN1A</b>	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
<b>SCN2A</b>	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
<b>SCN8A</b>	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
<b>SCYL1</b>	100 %	607982	Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive
<b>SDHA</b>	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
<b>SDHAF1</b>	99.99 %	612848	Mitochondrial complex II deficiency, nuclear type 2, 619166 (3), Autosomal recessive
<b>SDHD</b>	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
<b>SELENOI</b>	99.86 %	607915	Spastic paraplegia 81, autosomal recessive, 618768 (3), Autosomal recessive
<b>SEPSECS</b>	99.78 %	613009	Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive
<b>SERAC1</b>	99.9 %	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739 (3), Autosomal recessive
<b>SETX</b>	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
<b>SIL1</b>	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<b>SLC13A5</b>	99.99 %	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta, 615905 (3), Autosomal recessive
<b>SLC16A2</b>	99.97 %	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<b>SLC17A5</b>	99.71 %	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<b>SLC19A3</b>	99.95 %	606152	Thiamine metabolism dysfunction syndrome 2 (biotin/thiamine-responsive basal ganglia disease type), 607483 (3), Autosomal recessive
<b>SLC1A3</b>	99.98 %	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
<b>SLC1A4</b>	99.97 %	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive
<b>SLC25A15</b>	100 %	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
<b>SLC25A46</b>	99.88 %	610826	Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 1E, 619303 (3), Autosomal recessive
<b>SLC2A1</b>	99.93 %	138140	Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatid-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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>SLC33A1</b>	99.67 %	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive
<b>SLC44A1</b>	99.9 %	606105	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive
<b>SLC52A2</b>	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
<b>SLC52A3</b>	99.94 %	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
<b>SLC6A8</b>	99.99 %	300036	Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive
<b>SLC9A1</b>	99.96 %	107310	Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
<b>SLC9A6</b>	99.42 %	300231	Intellectual developmental disorder, X-linked syndromic, Christianson type, 300243 (3), X-linked
<b>SMPD1</b>	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
<b>SMPD4</b>	99.9 %	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies, 618622 (3), Autosomal recessive
<b>SNAP25</b>	99.89 %	600322	?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant
<b>SNAPC4</b>	100 %	602777	Neurodevelopmental disorder with motor regression, progressive spastic paraplegia, and oromotor dysfunction, 620515 (3), Autosomal recessive
<b>SNX14</b>	99.73 %	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<b>SOX10</b>	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
<b>SPART</b>	99.98 %	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<b>SPAST</b>	99.77 %	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<b>SPATA5L1</b>	99.91 %	619578	Deafness, autosomal recessive 119, 619615 (3), Autosomal recessive; Neurodevelopmental disorder with hearing loss and spasticity, 619616 (3), Autosomal recessive
<b>SPG11</b>	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
<b>SPG21</b>	99.97 %	608181	Mast syndrome, 248900 (3), Autosomal recessive
<b>SPG7</b>	99.99 %	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive
<b>SPR</b>	99.99 %	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
<b>SPTAN1</b>	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
<b>SPTBN2</b>	99.98 %	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
<b>SPTSSA</b>	99.88 %	613540	Spastic paraplegia 90A, autosomal dominant, 620416 (3), Autosomal dominant; ?Spastic paraplegia 90B, autosomal recessive, 620417 (3), Autosomal dominant
<b>SQSTM1</b>	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

# Ataxia Spasticity

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
<b>SRD5A3</b>	99.94 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive
<b>STUB1</b>	99.99 %	607207	Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
<b>SUCLA2</b>	99.96 %	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive
<b>SUFU</b>	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
<b>SUOX</b>	100 %	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<b>SURF1</b>	100 %	185620	Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive; Mitochondrial complex IV deficiency, nuclear type 1, 220110 (3), Autosomal recessive
<b>SVBP</b>	99.04 %	617853	Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive
<b>SYNE1</b>	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
<b>SYT14</b>	99.94 %	610949	?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive
<b>TAF8</b>	99.91 %	609514	Neurodevelopmental disorder with severe motor impairment, absent language, cerebral hypomyelination, and brain atrophy, 619972 (3), Autosomal recessive
<b>TANGO2</b>	99.85 %	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<b>TBC1D20</b>	100 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<b>TBC1D23</b>	98.7 %	617687	Pontocerebellar hypoplasia, type 11, 617695 (3), Autosomal recessive
<b>TDP1</b>	99.97 %	607198	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive
<b>TDP2</b>	99.97 %	605764	Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive
<b>TECPR2</b>	99.96 %	615000	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay, 615031 (3), Autosomal recessive
<b>TENM3</b>	99.99 %	610083	Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive
<b>TFG</b>	98.68 %	602498	?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant
<b>TGM6</b>	99.99 %	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
<b>THG1L</b>	99.99 %	618802	Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive
<b>TINF2</b>	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<b>TMEM106B</b>	99.92 %	613413	Leukodystrophy, hypomyelinating, 16, 617964 (3), Autosomal dominant
<b>TMEM216</b>	99.98 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<b>TMEM231</b>	88.88 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<b>TMEM240</b>	99.99 %	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
<b>TMEM63B</b>	99.95 %	619952	<i>No OMIM phenotypes</i>
<b>TMEM63C</b>	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
<b>TMEM67</b>	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
<b>TMX2</b>	99.99 %	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive
<b>TOE1</b>	99.96 %	613931	Pontocerebellar hypoplasia, type 7, 614969 (3), Autosomal recessive
<b>TPP1</b>	99.99 %	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<b>TRAK1</b>	99.98 %	608112	Developmental and epileptic encephalopathy 68, 618201 (3), Autosomal recessive
<b>TRAPPC12</b>	99.96 %	614139	Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive
<b>TRAPPC4</b>	100 %	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive
<b>TRIT1</b>	99.25 %	617840	Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive
<b>TRPC3</b>	99.97 %	602345	?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant
<b>TSEN15</b>	99.57 %	608756	Pontocerebellar hypoplasia, type 2F, 617026 (3), Autosomal recessive
<b>TSEN2</b>	99.98 %	608753	Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive
<b>TSEN54</b>	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
<b>TTBK2</b>	99.93 %	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
<b>TTC19</b>	99.99 %	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
<b>TTPA</b>	99.88 %	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<b>TTR</b>	100 %	176300	Amyloidosis, hereditary, transthyretin-related, 105210 (3), Autosomal dominant; Carpal tunnel syndrome, familial, 115430 (3), Autosomal dominant; [Dystransthyretinemic hyperthyroxinemia], 145680 (3), Autosomal dominant
<b>TUBA1A</b>	99.97 %	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
<b>TUBB3</b>	100 %	602661	Fibrosis of extraocular muscles, congenital, 3A, 600638 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 1, 614039 (3), Autosomal dominant
<b>TUBB4A</b>	100 %	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<b>TUBG1</b>	99.95 %	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
<b>TWNK</b>	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
<b>UBA5</b>	99.95 %	610552	?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Developmental and epileptic encephalopathy 44, 617132 (3), Autosomal recessive
<b>UBAP1</b>	99.51 %	609787	Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant
<b>UBTF</b>	99.99 %	600673	Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant
<b>UCHL1</b>	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
<b>UFC1</b>	99.35 %	610554	Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive
<b>UFM1</b>	99.25 %	610553	Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive
<b>UGP2</b>	99.85 %	191760	Developmental and epileptic encephalopathy 83, 618744 (3), Autosomal recessive
<b>VAMP1</b>	100 %	185880	Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive; Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
<b>VCP</b>	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
<b>VLDLR</b>	99.99 %	192977	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome 1, 224050 (3), Autosomal recessive
<b>VPS13B</b>	99.9 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<b>VPS13D</b>	99.96 %	608877	Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive
<b>VPS37A</b>	99.96 %	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
<b>VPS41</b>	99.92 %	605485	Spinocerebellar ataxia, autosomal recessive 29, 619389 (3), Autosomal recessive
<b>VPS53</b>	100 %	615850	Pontocerebellar hypoplasia, type 2E, 615851 (3), Autosomal recessive
<b>VRK1</b>	99.98 %	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal recessive 10, 620542 (3), Autosomal recessive
<b>VWA3B</b>	98.82 %	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
<b>WARS2</b>	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
<b>WASHC5</b>	99.98 %	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
<b>WDR45B</b>	100 %	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive
<b>WDR73</b>	99.92 %	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive
<b>WDR81</b>	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
<b>WFS1</b>	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
<b>WWOX</b>	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
<b>XRCC1</b>	99.96 %	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
<b>ZC4H2</b>	99.98 %	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant
<b>ZFYVE26</b>	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.