

Ataxia Spasticity panel		
versie	16-Oct-2018 (260 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABCB7</i>	300135	Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive
<i>ABCD1</i>	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
<i>ABHD12</i>	613599	Polymyopathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ADAR</i>	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
<i>ADGRG1</i>	604110	Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive; Polymicrogyria, bilateral perisylvian, 615752 (3)
<i>AFG3L2</i>	604581	Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>ALDH18A1</i>	138250	Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive, Isolated cases; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive
<i>ALDH5A1</i>	610045	Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive
<i>ALG6</i>	604566	Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive
<i>AMPD2</i>	102771	Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive; ?Spastic paraplegia 63, 615686 (3), Autosomal recessive
<i>ANO10</i>	613726	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive
<i>AP4B1</i>	607245	Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive
<i>AP4E1</i>	607244	Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive; Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant
<i>AP4M1</i>	602296	Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive
<i>AP4S1</i>	607243	Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive

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<i>AP5Z1</i>	613653	Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive
<i>APTX</i>	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive
<i>ARG1</i>	608313	Argininemia, 207800 (3), Autosomal recessive
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6IP1</i>	607669	?Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive
<i>ARSA</i>	607574	Metachromatic leukodystrophy, 250100 (3), Autosomal recessive
<i>ARSI</i>	610009	No OMIM phenotype
<i>ARX</i>	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
<i>ASPA</i>	608034	Canavan disease, 271900 (3), Autosomal recessive
<i>ATCAY</i>	608179	Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive
<i>ATL1</i>	606439	Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant; Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant
<i>ATM</i>	607585	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); T-cell prolymphocytic leukemia, somatic (3)
<i>ATP13A2</i>	610513	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive
<i>ATP2B3</i>	300014	?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive
<i>ATP7B</i>	606882	Wilson disease, 277900 (3), Autosomal recessive
<i>ATP8A2</i>	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive
<i>AUH</i>	600529	3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive
<i>B4GALNT1</i>	601873	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive
<i>BCKDHA</i>	608348	Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive
<i>BCKDHB</i>	248611	Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive
<i>BICD2</i>	609797	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290 (3), Autosomal dominant

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<i>BSCL2</i>	606158	Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive; Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant
<i>BTD</i>	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
<i>C12orf65</i>	613541	Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive; Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive
<i>C19orf12</i>	614297	Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive
<i>CA8</i>	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive
<i>CACNA1A</i>	601011	Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant
<i>CACNA1G</i>	604065	Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant
<i>CACNB4</i>	601949	{Epilepsy, idiopathic generalized, susceptibility to, 9}, 607682 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 6}, 607682 (3), Autosomal dominant; Episodic ataxia, type 5, 613855 (3), Autosomal dominant
<i>CAMTA1</i>	611501	Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant
<i>CAPN1</i>	114220	Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive
<i>CC2D2A</i>	612013	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive
<i>CCDC88C</i>	611204	Hydrocephalus, nonsyndromic, autosomal recessive, 236600 (3), Autosomal recessive; ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant
<i>CCT5</i>	610150	Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive

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<i>CLCN2</i>	600570	{Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive
<i>COQ2</i>	609825	Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive; {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal recessive, Autosomal dominant
<i>COQ4</i>	612898	Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive
<i>COQ8A</i>	606980	Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive
<i>COQ9</i>	612837	Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive
<i>COX20</i>	614698	Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial
<i>CSTB</i>	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive
<i>CWF19L1</i>	616120	Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>CYP2U1</i>	610670	Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive
<i>CYP7B1</i>	603711	Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive; Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive
<i>DARS2</i>	610956	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive
<i>DBT</i>	248610	Maple syrup urine disease, type II, 248600 (3), Autosomal recessive
<i>DDHD1</i>	614603	Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive
<i>DDHD2</i>	615003	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive
<i>DLAT</i>	608770	Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive
<i>DLD</i>	238331	Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive
<i>DNAJC19</i>	608977	3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive
<i>DNMT1</i>	126375	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant
<i>EEF2</i>	130610	?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant
<i>ELOVL4</i>	605512	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive; Spinocerebellar ataxia 34, 133190 (3),

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		Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant
<i>ELOVL5</i>	611805	Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant
<i>ENTPD1</i>	601752	Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive
<i>EPM2A</i>	607566	Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive
<i>ERCC4</i>	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; ?XFE progeroid syndrome, 610965 (3); Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
<i>ERLIN1</i>	611604	Spastic paraplegia 62, 615681 (3), Autosomal recessive
<i>ERLIN2</i>	611605	Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive
<i>EXOSC3</i>	606489	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive
<i>FA2H</i>	611026	Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive
<i>FGF14</i>	601515	Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant
<i>FLRT1</i>	604806	No OMIM phenotype
<i>FLVCR1</i>	609144	Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive
<i>FOLR1</i>	136430	Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive
<i>FRMD7</i>	300628	Nystagmus 1, congenital, X-linked, 310700 (3), X-linked; Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked
<i>GALC</i>	606890	Krabbe disease, 245200 (3), Autosomal recessive
<i>GAN</i>	605379	Giant axonal neuropathy-1, 256850 (3), Autosomal recessive
<i>GFAP</i>	137780	Alexander disease, 203450 (3), Autosomal dominant
<i>GJC2</i>	608803	Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive; Lymphedema, hereditary, IC, 613480 (3), Autosomal dominant; Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive
<i>GLB1</i>	611458	GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive; GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive
<i>GLRX5</i>	609588	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive
<i>GOSR2</i>	604027	Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive
<i>GPR143</i>	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked

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<i>GRID2</i>	602368	Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive
<i>GRM1</i>	604473	Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive
<i>GRN</i>	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant
<i>HEXA</i>	606869	GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive
<i>HEXB</i>	606873	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSPD1</i>	118190	Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive; Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant
<i>IBA57</i>	615316	Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive; ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive
<i>IFIH1</i>	606951	Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant
<i>IFRD1</i>	603502	No OMIM phenotype
<i>IRF2BPL</i>	611720	No OMIM phenotype
<i>ITM2B</i>	603904	Dementia, familial British, 176500 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant
<i>ITPR1</i>	147265	Gillespie syndrome, 206700 (3), Autosomal recessive, Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant
<i>KCNA1</i>	176260	Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant
<i>KCNA2</i>	176262	Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant
<i>KCNC3</i>	176264	Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant
<i>KCND3</i>	605411	Brugada syndrome 9, 616399 (3), Autosomal dominant; Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant
<i>KCNJ10</i>	602208	Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive
<i>KCNJ6</i>	600877	Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant

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<i>KCNMA1</i>	600150	?Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant
<i>KCTD7</i>	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive
<i>KIDINS220</i>	615759	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant
<i>KIF1A</i>	601255	Mental retardation, autosomal dominant 9, 614255 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal recessive
<i>KIF1C</i>	603060	Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive
<i>KIF5A</i>	602821	{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
<i>L1CAM</i>	308840	CRASH syndrome, 303350 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; MASA syndrome, 303350 (3), X-linked recessive
<i>L2HGDH</i>	609584	L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive
<i>LAMA1</i>	150320	Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive
<i>LAMB1</i>	150240	Lissencephaly 5, 615191 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>MAG</i>	159460	Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive
<i>MAN2B1</i>	609458	Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive
<i>MARS</i>	156560	Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive
<i>MARS2</i>	609728	?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive; Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive
<i>MECP2</i>	300005	{Autism susceptibility, X-linked 3}, 300496 (3), Isolated cases, X-linked, Multifactorial; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, atypical,

312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant

<i>MLC1</i>	605908	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive
<i>MMADHC</i>	611935	Homocystinuria, cblD type, variant 1, 277410 (3), Autosomal recessive; Methylmalonic aciduria and homocystinuria, cblD type, 277410 (3), Autosomal recessive; Methylmalonic aciduria, cblD type, variant 2, 277410 (3), Autosomal recessive
<i>MRE11 (MRE11A)</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
<i>MSTO1</i>	617619	Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal recessive, Autosomal dominant
<i>MTPAP</i>	613669	?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive
<i>MTTP</i>	157147	Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant
<i>NANS</i>	605202	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive
<i>NEFL</i>	162280	Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal recessive, Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant
<i>NEU1</i>	608272	Sialidosis, type I, 256550 (3), Autosomal recessive; Sialidosis, type II, 256550 (3), Autosomal recessive
<i>NEXMIF</i>	300524	Mental retardation, X-linked 98, 300912 (3), X-linked dominant
<i>NF2</i>	607379	Meningioma, NF2-related, somatic, 607174 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 162091 (3)
<i>NHLRC1</i>	608072	Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive
<i>NIPA1</i>	608145	Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant
<i>NOL3</i>	605235	Myoclonus, familial cortical, 614937 (3), Autosomal dominant
<i>NPC1</i>	607623	Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive; Niemann-Pick disease, type D, 257220 (3), Autosomal recessive
<i>NPC2</i>	601015	Niemann-pick disease, type C2, 607625 (3), Autosomal recessive
<i>NT5C2</i>	600417	Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive
<i>OCLN</i>	602876	Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive

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<i>OFD1</i>	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>OPA1</i>	605290	Behr syndrome, 210000 (3), Autosomal recessive; {Glaucoma, normal tension, susceptibility to}, 606657 (3); ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3); Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant
<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>OPHN1</i>	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive
<i>OTC</i>	300461	Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive
<i>PDHX</i>	608769	Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive
<i>PDYN</i>	131340	Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant
<i>PEX10</i>	602859	Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive; Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive
<i>PEX16</i>	603360	Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PGAP1</i>	611655	Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PIK3R5</i>	611317	Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive
<i>PLA2G6</i>	603604	Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive
<i>PLP1</i>	300401	Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive
<i>PMM2</i>	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
<i>PMPCA</i>	613036	Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive

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<i>PNPLA6</i>	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
<i>POLG</i>	174763	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POLR3B</i>	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive
<i>PRF1</i>	170280	Aplastic anemia, 609135 (3); Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Lymphoma, non-Hodgkin, 605027 (3)
<i>PRKCG</i>	176980	Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant
<i>PRNP</i>	176640	Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (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); Prion disease with protracted course, 606688 (3), Autosomal dominant
<i>PRRT2</i>	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant; Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant
<i>PSAP</i>	176801	Combined SAP deficiency, 611721 (3), Autosomal recessive; Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive
<i>PSEN1</i>	104311	?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant, Isolated cases

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<i>PTRH2</i>	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive
<i>PUM1</i>	607204	Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant
<i>RAB18</i>	602207	Warburg micro syndrome 3, 614222 (3), Autosomal recessive
<i>RAB3GAP1</i>	602536	Warburg micro syndrome 1, 600118 (3), Autosomal recessive
<i>RAB3GAP2</i>	609275	Martsolf syndrome, 212720 (3), Autosomal recessive; Warburg micro syndrome 2, 614225 (3), Autosomal recessive
<i>REEP1</i>	609139	?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant; Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant
<i>REEP2</i>	609347	?Spastic paraplegia 72, autosomal dominant, 615625 (3), Autosomal recessive, Autosomal dominant; ?Spastic paraplegia 72, autosomal recessive, 615625 (3), Autosomal recessive, Autosomal dominant
<i>RETREG1 (FAM134B)</i>	613114	Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive
<i>RNASEH2A</i>	606034	Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive
<i>RNASEH2B</i>	610326	Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive
<i>RNASEH2C</i>	610330	Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive
<i>RNF168</i>	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
<i>RNF170</i>	614649	Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant
<i>RNF216</i>	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive
<i>RTN2</i>	603183	Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant
<i>RUBCN</i>	613516	?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive
<i>SACS</i>	604490	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive
<i>SAMD9L</i>	611170	Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant
<i>SAMHD1</i>	606754	Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive; ?Chilblain lupus 2, 614415 (3), Autosomal dominant
<i>SCN8A</i>	600702	?Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant; Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant
<i>SDHA</i>	600857	Cardiomyopathy, dilated, 1GG, 613642 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive; Paragangliomas 5, 614165 (3), Autosomal dominant

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<i>SETX</i>	608465	Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 1, 606002 (3), Autosomal recessive
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SLC16A2</i>	300095	Allan-Herndon-Dudley syndrome, 300523 (3), X-linked
<i>SLC17A5</i>	604322	Salla disease, 604369 (3), Autosomal recessive; Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive
<i>SLC19A3</i>	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive
<i>SLC1A3</i>	600111	Episodic ataxia, type 6, 612656 (3), Autosomal dominant
<i>SLC25A15</i>	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive
<i>SLC2A1</i>	138140	Dystonia 9, 601042 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal recessive, Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant
<i>SLC33A1</i>	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
<i>SLC52A2</i>	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
<i>SLC52A3</i>	613350	Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive
<i>SLC9A1</i>	107310	?Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive
<i>SMPD1</i>	607608	Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive
<i>SNX14</i>	616105	Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive
<i>SOX10</i>	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
<i>SPART (SPG20)</i>	607111	Troyer syndrome, 275900 (3), Autosomal recessive
<i>SPAST</i>	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
<i>SPG11</i>	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

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<i>SPG21</i> (<i>ACP33</i>)	608181	Mast syndrome, 248900 (3), Autosomal recessive
<i>SPG7</i>	602783	Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal recessive, Autosomal dominant
<i>SPR</i>	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), ?Autosomal dominant, Autosomal recessive
<i>SPTBN2</i>	604985	Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive
<i>SRD5A3</i>	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
<i>STUB1</i>	607207	Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive
<i>SUOX</i>	606887	Sulfite oxidase deficiency, 272300 (3), Autosomal recessive
<i>SYNE1</i>	608441	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive
<i>TANGO2</i>	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive
<i>TBC1D20</i>	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
<i>TDP1</i>	607198	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 (3)
<i>TECPR2</i>	615000	Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive
<i>TFG</i>	602498	Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant; ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive
<i>TGM6</i>	613900	Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant
<i>TMEM216</i>	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
<i>TMEM231</i>	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
<i>TMEM240</i>	616101	Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant
<i>TMEM67</i>	609884	{Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive
<i>TPP1</i>	607998	Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive
<i>TSEN54</i>	608755	Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; Pontocerebellar hypoplasia type 4, 225753 (3),

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		Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive
<i>TTBK2</i>	611695	Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant
<i>TTC19</i>	613814	Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive
<i>TTPA</i>	600415	Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive
<i>TUBA1A</i>	602529	Lissencephaly 3, 611603 (3), Autosomal dominant
<i>TUBB4A</i>	602662	Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant
<i>TUBG1</i>	191135	Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant
<i>TWNK</i>	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
<i>UCHL1</i>	191342	{?Parkinson disease 5, susceptibility to}, 613643 (3); Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive
<i>USP8</i>	603158	Pituitary adenoma 4, ACTH-secreting, somatic, 219090 (3)
<i>VAMP1</i>	185880	Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant
<i>VCP</i>	601023	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3); 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
<i>VLDLR</i>	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive
<i>VPS13B</i>	607817	Cohen syndrome, 216550 (3), Autosomal recessive
<i>VPS13D</i>	608877	No OMIM phenotype
<i>VPS37A</i>	609927	Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive
<i>VRK1</i>	602168	Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive
<i>VWA3B</i>	614884	?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive
<i>WASHC5</i>	610657	Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant
<i>WDR48</i>	612167	No OMIM phenotype
<i>WDR73</i>	616144	Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive

<i>WDR81</i>	614218	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive; Hydrocephalus, nonsyndromic, autosomal recessive 3, 617967 (3)
<i>WWOX</i>	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
<i>XRCC1</i>	194360	?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive
<i>ZC4H2</i>	300897	Wieacker-Wolff syndrome, 314580 (3), X-linked recessive
<i>ZFR</i>	615635	No OMIM phenotype
<i>ZFYVE26</i>	612012	Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive
<i>ZFYVE27</i>	610243	Spastic paraplegia 33, autosomal dominant, 610244 (3), Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

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

OMIM release used for OMIM disease identifiers and descriptions: July 04, 2018

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.