# Ataxia Spasticity panel

| **versie** | V2 (390 genen) | Centrum voor Medische Genetica Gent |
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| Gene | OMIM gene ID | Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern |
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| *AAAS* | 605378 | Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive |
| *AARS1 (AARS)* | 601065 | Epileptic encephalopathy, early infantile, 29, 616339 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2N, 613287 (3), Autosomal dominant |
| *ABCA2* | 600047 | Intellectual developmental disorder with poor growth and with or without seizures or ataxia, 618808 (3), Autosomal recessive |
| *ABCB7* | 300135 | Anemia, sideroblastic, with ataxia, 301310 (3), X-linked recessive |
| *ABCD1* | 300371 | Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive; Adrenoleukodystrophy, 300100 (3), X-linked recessive |
| *ABHD12* | 613599 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive |
| *ACO2* | 100850 | Infantile cerebellar-retinal degeneration, 614559 (3), Autosomal recessive; ?Optic atrophy 9, 616289 (3), Autosomal recessive |
| *ADAR* | 146920 | Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive |
| *ADGRG1* | 604110 | Polymicrogyria, bilateral perisylvian, 615752 (3); Polymicrogyria, bilateral frontoparietal, 606854 (3), Autosomal recessive |
| *ADPRS (ADPRHL2)* | 610624 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures, 618170 (3), Autosomal recessive |
| *AFG3L2* | 604581 | Spastic ataxia 5, autosomal recessive, 614487 (3), Autosomal recessive; Optic atrophy 12, 618977 (3); Spinocerebellar ataxia 28, 610246 (3), Autosomal dominant |
| *AGTPBP1* | 606830 | Neurodegeneration, childhood-onset, with cerebellar atrophy, 618276 (3), Autosomal recessive |
| *AHI1* | 608894 | Joubert syndrome 3, 608629 (3), Autosomal recessive |
| *AIFM1* | 300169 | Cowchock syndrome, 310490 (3), X-linked recessive; Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy, 300232 (3), X-linked recessive; Combined oxidative phosphorylation deficiency 6, 300816 (3), X-linked recessive; Deafness, X-linked 5, 300614 (3), X-linked recessive |
| *AIMP1* | 603605 | Leukodystrophy, hypomyelinating, 3, 260600 (3), Autosomal recessive |
| *ALDH18A1* | 138250 | Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant; Spastic paraplegia 9B, autosomal recessive, 616586 (3), |

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| | | Autosomal recessive; Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant |
| *ALDH5A1* | 610045 | Succinic semialdehyde dehydrogenase deficiency, 271980 (3), Autosomal recessive |
| *ALG6* | 604566 | Congenital disorder of glycosylation, type Ic, 603147 (3), Autosomal recessive |
| *ALS2* | 606352 | Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive |
| *AMPD2* | 102771 | ?Spastic paraplegia 63, 615686 (3), Autosomal recessive; Pontocerebellar hypoplasia, type 9, 615809 (3), Autosomal recessive |
| *ANO10* | 613726 | Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive |
| *AP4B1* | 607245 | Spastic paraplegia 47, autosomal recessive, 614066 (3), Autosomal recessive |
| *AP4E1* | 607244 | Stuttering, familial persistent, 1, 184450 (3), Autosomal dominant; Spastic paraplegia 51, autosomal recessive, 613744 (3), Autosomal recessive |
| *AP4M1* | 602296 | Spastic paraplegia 50, autosomal recessive, 612936 (3), Autosomal recessive |
| *AP4S1* | 607243 | Spastic paraplegia 52, autosomal recessive, 614067 (3), Autosomal recessive |
| *AP5Z1* | 613653 | Spastic paraplegia 48, autosomal recessive, 613647 (3), Autosomal recessive |
| *APOB* | 107730 | Hypobetalipoproteinemia, 615558 (3), Autosomal recessive; Hypercholesterolemia, familial, 2, 144010 (3), Autosomal dominant |
| *APTX* | 606350 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3), Autosomal recessive |
| *ARG1* | 608313 | Argininemia, 207800 (3), Autosomal recessive |
| *ARL13B* | 608922 | Joubert syndrome 8, 612291 (3), Autosomal recessive |
| *ARL6IP1* | 607669 | ?Spastic paraplegia 61, autosomal recessive, 615685 (3), Autosomal recessive |
| *ARSA* | 607574 | Metachromatic leukodystrophy, 250100 (3), Autosomal recessive |
| *ARSI* | 610009 | No OMIM phenotype |
| *ARX* | 300382 | Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Proud syndrome, 300004 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked |
| *ASPA* | 608034 | Canavan disease, 271900 (3), Autosomal recessive |
| *ATCAY* | 608179 | Ataxia, cerebellar, Cayman type, 601238 (3), Autosomal recessive |

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| *ATG5* | 604261 | ?Spinocerebellar ataxia, autosomal recessive 25, 617584 (3), Autosomal recessive |
| *ATL1* | 606439 | Spastic paraplegia 3A, autosomal dominant, 182600 (3), Autosomal dominant; Neuropathy, hereditary sensory, type ID, 613708 (3), Autosomal dominant |
| *ATM* | 607585 | Lymphoma, mantle cell, somatic (3); Lymphoma, B-cell non-Hodgkin, somatic (3); Ataxia-telangiectasia, 208900 (3), Autosomal recessive; {Breast cancer, susceptibility to}, 114480 (3), Autosomal dominant, Somatic mutation; T-cell prolymphocytic leukemia, somatic (3) |
| *ATP13A2* | 610513 | Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive |
| *ATP1A3* | 182350 | CAPOS syndrome, 601338 (3), Autosomal dominant; Alternating hemiplegia of childhood 2, 614820 (3), Autosomal dominant; Dystonia-12, 128235 (3), Autosomal dominant |
| *ATP2B3* | 300014 | ?Spinocerebellar ataxia, X-linked 1, 302500 (3), X-linked recessive |
| *ATP7B* | 606882 | Wilson disease, 277900 (3), Autosomal recessive |
| *ATP8A2* | 605870 | ?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268 (3), Autosomal recessive |
| *AUH* | 600529 | 3-methylglutaconic aciduria, type I, 250950 (3), Autosomal recessive |
| *B4GALNT1* | 601873 | Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive |
| *BCKDHA* | 608348 | Maple syrup urine disease, type Ia, 248600 (3), Autosomal recessive |
| *BCKDHB* | 248611 | Maple syrup urine disease, type Ib, 248600 (3), Autosomal recessive |
| *BCL11B* | 606558 | Intellectual developmental disorder with dysmorphic facies, speech delay, and T-cell abnormalities, 618092 (3), Autosomal dominant; Immunodeficiency 49, 617237 (3), Autosomal dominant |
| *BCS1L* | 603647 | Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; GRACILE syndrome, 603358 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive |
| *BICD2* | 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 |
| *BSCL2* | 606158 | Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Neuropathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive |

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| *BTD* | 609019 | Biotinidase deficiency, 253260 (3), Autosomal recessive |
| *C12orf65* | 613541 | Spastic paraplegia 55, autosomal recessive, 615035 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 7, 613559 (3), Autosomal recessive |
| *C19orf12* | 614297 | Neurodegeneration with brain iron accumulation 4, 614298 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 43, autosomal recessive, 615043 (3), Autosomal recessive |
| *CA8* | 114815 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227 (3), Autosomal recessive |
| *CACNA1A* | 601011 | Spinocerebellar ataxia 6, 183086 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 42, 617106 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500 (3), Autosomal dominant; Episodic ataxia, type 2, 108500 (3), Autosomal dominant; Migraine, familial hemiplegic, 1, 141500 (3), Autosomal dominant |
| *CACNA1E* | 601013 | Epileptic encephalopathy, early infantile, 69, 618285 (3), Autosomal dominant |
| *CACNA1G* | 604065 | Spinocerebellar ataxia 42, 616795 (3), Autosomal dominant; Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits, 618087 (3), Autosomal dominant |
| *CACNB4* | 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 |
| *CAMTA1* | 611501 | Cerebellar ataxia, nonprogressive, with mental retardation, 614756 (3), Autosomal dominant |
| *CAPN1* | 114220 | Spastic paraplegia 76, autosomal recessive, 616907 (3), Autosomal recessive |
| *CC2D2A* | 612013 | Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive |
| *CCDC88C* | 611204 | ?Spinocerebellar ataxia 40, 616053 (3), Autosomal dominant; Hydrocephalus, congenital, 1, 236600 (3), Autosomal recessive |
| *CCT5* | 610150 | Neuropathy, hereditary sensory, with spastic paraplegia, 256840 (3), Autosomal recessive |
| *CDK16* | 311550 | No OMIM phenotype |
| *CEP290* | 610142 | ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive |
| *CHP1* | 606988 | ?Spastic ataxia 9, autosomal recessive, 618438 (3), Autosomal recessive |

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| *CLCN2* | 600570 | {Epilepsy, juvenile absence, susceptibility to, 2}, 607628 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 11}, 607628 (3), Autosomal dominant; {Epilepsy, juvenile myoclonic, susceptibility to, 8}, 607628 (3), Autosomal dominant; Leukoencephalopathy with ataxia, 615651 (3), Autosomal recessive; Hyperaldosteronism, familial, type II, 605635 (3), Autosomal dominant |
| *CNTNAP1* | 602346 | Lethal congenital contracture syndrome 7, 616286 (3), Autosomal recessive; Hypomyelinating neuropathy, congenital, 3, 618186 (3), Autosomal recessive |
| *COA7* | 615623 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3, 618387 (3), Autosomal recessive |
| *COQ2* | 609825 | {Multiple system atrophy, susceptibility to}, 146500 (3), Autosomal dominant, Autosomal recessive; Coenzyme Q10 deficiency, primary, 1, 607426 (3), Autosomal recessive |
| *COQ4* | 612898 | Coenzyme Q10 deficiency, primary, 7, 616276 (3), Autosomal recessive |
| *COQ8A* | 606980 | Coenzyme Q10 deficiency, primary, 4, 612016 (3), Autosomal recessive |
| *COQ9* | 612837 | Coenzyme Q10 deficiency, primary, 5, 614654 (3), Autosomal recessive |
| *COX10* | 602125 | Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial; Leigh syndrome due to mitochondrial COX4 deficiency, 256000 (3), Autosomal recessive, Mitochondrial |
| *COX15* | 603646 | Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119 (3), Autosomal recessive; Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3), Autosomal recessive, Mitochondrial |
| *COX20* | 614698 | Mitochondrial complex IV deficiency, 220110 (3), Autosomal recessive, Mitochondrial |
| *CP* | 117700 | [Hypoceruloplasminemia, hereditary], 604290 (3), Autosomal recessive; Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3), Autosomal recessive; Cerebellar ataxia, 604290 (3), Autosomal recessive |
| *CPT1C* | 608846 | ?Spastic paraplegia 73, autosomal dominant, 616282 (3), Autosomal dominant |
| *CSTB* | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg), 254800 (3), Autosomal recessive |
| *CTBP1* | 602618 | Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome, 617915 (3), Autosomal dominant |
| *CWF19L1* | 616120 | Spinocerebellar ataxia, autosomal recessive 17, 616127 (3), Autosomal recessive |
| *CYP27A1* | 606530 | Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive |
| *CYP2U1* | 610670 | Spastic paraplegia 56, autosomal recessive, 615030 (3), Autosomal recessive |

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| *CYP7B1* | 603711 | Spastic paraplegia 5A, autosomal recessive, 270800 (3), Autosomal recessive; Bile acid synthesis defect, congenital, 3, 613812 (3), Autosomal recessive |
| *DAB1* | 603448 | Spinocerebellar ataxia 37, 615945 (3), Autosomal dominant |
| *DARS1 (DARS)* | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281 (3), Autosomal recessive |
| *DARS2* | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105 (3), Autosomal recessive |
| *DBT* | 248610 | Maple syrup urine disease, type II, 248600 (3), Autosomal recessive |
| *DDHD1* | 614603 | Spastic paraplegia 28, autosomal recessive, 609340 (3), Autosomal recessive |
| *DDHD2* | 615003 | Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive |
| *DDX3X* | 300160 | Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958 (3), X-linked dominant, X-linked recessive |
| *DEGS1* | 615843 | Leukodystrophy, hypomyelinating, 18, 618404 (3), Autosomal recessive |
| *DLAT* | 608770 | Pyruvate dehydrogenase E2 deficiency, 245348 (3), Autosomal recessive |
| *DLD* | 238331 | Dihydrolipoamide dehydrogenase deficiency, 246900 (3), Autosomal recessive |
| *DNAJC19* | 608977 | 3-methylglutaconic aciduria, type V, 610198 (3), Autosomal recessive |
| *DNAJC3* | 601184 | ?Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192 (3), Autosomal recessive |
| *DNAJC5* | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350 (3), Autosomal dominant |
| *DNMT1* | 126375 | Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121 (3), Autosomal dominant; Neuropathy, hereditary sensory, type IE, 614116 (3), Autosomal dominant |
| *DOCK3* | 603123 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia, 618292 (3), Autosomal recessive |
| *EARS2* | 612799 | Combined oxidative phosphorylation deficiency 12, 614924 (3), Autosomal recessive |
| *EBF3* | 607407 | Hypotonia, ataxia, and delayed development syndrome, 617330 (3), Autosomal dominant |
| *ECHS1* | 602292 | Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277 (3), Autosomal recessive |
| *EEF2* | 130610 | ?Spinocerebellar ataxia 26, 609306 (3), Autosomal dominant |
| *EIF2AK1* | 613635 | ?Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome, 618878 (3) |
| *ELOVL1* | 611813 | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal dominant |

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| *ELOVL4* | 605512 | Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant; Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive |
| *ELOVL5* | 611805 | Spinocerebellar ataxia 38, 615957 (3), Autosomal dominant |
| *EMC1* | 616846 | Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875 (3), Autosomal recessive |
| *ENTPD1* | 601752 | Spastic paraplegia 64, autosomal recessive, 615683 (3), Autosomal recessive |
| *EPM2A* | 607566 | Epilepsy, progressive myoclonic 2A (Lafora), 254780 (3), Autosomal recessive |
| *ERCC1* | 126380 | Cerebrooculofacioskeletal syndrome 4, 610758 (3), Autosomal recessive |
| *ERCC4* | 133520 | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive |
| *ERCC8* | 609412 | Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive |
| *ERLIN1* | 611604 | Spastic paraplegia 62, 615681 (3), Autosomal recessive |
| *ERLIN2* | 611605 | Spastic paraplegia 18, autosomal recessive, 611225 (3), Autosomal recessive |
| *EXOSC3* | 606489 | Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive |
| *FA2H* | 611026 | Spastic paraplegia 35, autosomal recessive, 612319 (3), Autosomal recessive |
| *FARS2* | 611592 | Spastic paraplegia 77, autosomal recessive, 617046 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 14, 614946 (3), Autosomal recessive |
| *FASTKD2* | 612322 | Combined oxidative phosphorylation deficiency 44, 618855 (3), Autosomal recessive |
| *FAT2* | 604269 | Spinocerebellar ataxia 45, 617769 (3), Autosomal dominant |
| *FGF14* | 601515 | Spinocerebellar ataxia 27, 609307 (3), Autosomal dominant |
| *FITM2* | 612029 | Siddiqi syndrome, 618635 (3), Autosomal recessive |
| *FLRT1* | 604806 | No OMIM phenotype |
| *FLVCR1* | 609144 | Ataxia, posterior column, with retinitis pigmentosa, 609033 (3), Autosomal recessive |
| *FOLR1* | 136430 | Neurodegeneration due to cerebral folate transport deficiency, 613068 (3), Autosomal recessive |
| *FRMD4A* | 616305 | ?Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819 (3), Autosomal recessive |
| *FRMD7* | 300628 | Nystagmus 1, congenital, X-linked, 310700 (3), X-linked; Nystagmus, infantile periodic alternating, X-linked, 310700 (3), X-linked |
| *GALC* | 606890 | Krabbe disease, 245200 (3), Autosomal recessive |

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| *GAN* | 605379 | Giant axonal neuropathy-1, 256850 (3), Autosomal recessive |
| *GBA2* | 609471 | Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive |
| *GBE1* | 607839 | Polyglucosan body disease, adult form, 263570 (3), Autosomal recessive; Glycogen storage disease IV, 232500 (3), Autosomal recessive |
| *GDAP2* | 618128 | Spinocerebellar ataxia, autosomal recessive 27, 618369 (3), Autosomal recessive |
| *GFAP* | 137780 | Alexander disease, 203450 (3), Autosomal dominant |
| *GFM2* | 606544 | Combined oxidative phosphorylation deficiency 39, 618397 (3), Autosomal recessive |
| *GJC2* | 608803 | Spastic paraplegia 44, autosomal recessive, 613206 (3), Autosomal recessive; Lymphatic malformation 3, 613480 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 2, 608804 (3), Autosomal recessive |
| *GLB1* | 611458 | GM1-gangliosidosis, type III, 230650 (3), Autosomal recessive; GM1-gangliosidosis, type I, 230500 (3), Autosomal recessive; Mucopolysaccharidosis type IVB (Morquio), 253010 (3), Autosomal recessive; GM1-gangliosidosis, type II, 230600 (3), Autosomal recessive |
| *GLRX5* | 609588 | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 (3), Autosomal recessive; Spasticity, childhood-onset, with hyperglycinemia, 616859 (3), Autosomal recessive |
| *GLS* | 138280 | ?Infantile cataract, skin abnormalities, glutamate excess, and impaired intellectual development, 618339 (3), Autosomal dominant; Global developmental delay, progressive ataxia, and elevated glutamine, 618412 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 71, 618328 (3), Autosomal recessive |
| *GM2A* | 613109 | GM2-gangliosidosis, AB variant, 272750 (3), Autosomal recessive |
| *GOSR2* | 604027 | Epilepsy, progressive myoclonic 6, 614018 (3), Autosomal recessive |
| *GPR143* | 300808 | Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive |
| *GRID2* | 602368 | Spinocerebellar ataxia, autosomal recessive 18, 616204 (3), Autosomal recessive |
| *GRIN1* | 138249 | Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive, 617820 (3), Autosomal recessive; Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant, 614254 (3), Autosomal dominant |
| *GRM1* | 604473 | Spinocerebellar ataxia 44, 617691 (3), Autosomal dominant; Spinocerebellar ataxia, autosomal recessive 13, 614831 (3), Autosomal recessive |

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| *GRN* | 138945 | Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive; Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant |
| *GSX2* | 616253 | Diencephalic-mesencephalic junction dysplasia syndrome 2, 618646 (3), Autosomal recessive |
| *HACE1* | 610876 | Spastic paraplegia and psychomotor retardation with or without seizures, 616756 (3), Autosomal recessive |
| *HEXA* | 606869 | GM2-gangliosidosis, several forms, 272800 (3), Autosomal recessive; Tay-Sachs disease, 272800 (3), Autosomal recessive; [Hex A pseudodeficiency], 272800 (3), Autosomal recessive |
| *HEXB* | 606873 | Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3), Autosomal recessive |
| *HSD17B10* | 300256 | HSD10 mitochondrial disease, 300438 (3), X-linked dominant |
| *HSD17B4* | 601860 | D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive |
| *HSPD1* | 118190 | Spastic paraplegia 13, autosomal dominant, 605280 (3), Autosomal dominant; Leukodystrophy, hypomyelinating, 4, 612233 (3), Autosomal recessive |
| *IBA57* | 615316 | ?Spastic paraplegia 74, autosomal recessive, 616451 (3), Autosomal recessive; Multiple mitochondrial dysfunctions syndrome 3, 615330 (3), Autosomal recessive |
| *IFIH1* | 606951 | Aicardi-Goutieres syndrome 7, 615846 (3), Autosomal dominant; Singleton-Merten syndrome 1, 182250 (3), Autosomal dominant |
| *IFRD1* | 603502 | No OMIM phenotype |
| *IFT140* | 614620 | Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive |
| *INTS8* | 611351 | ?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity, 618572 (3), Autosomal recessive |
| *IRF2BPL* | 611720 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures, 618088 (3), Autosomal dominant |
| *ISCA1* | 611006 | Multiple mitochondrial dysfunctions syndrome 5, 617613 (3), Autosomal recessive |
| *ITM2B* | 603904 | Dementia, familial British, 176500 (3), Autosomal dominant; ?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079 (3), Autosomal dominant; Dementia, familial Danish, 117300 (3), Autosomal dominant |
| *ITPR1* | 147265 | Spinocerebellar ataxia 29, congenital nonprogressive, 117360 (3), Autosomal dominant; Spinocerebellar ataxia 15, 606658 (3), Autosomal dominant; Gillespie syndrome, 206700 (3), Autosomal dominant, Autosomal recessive |
| *JAM2* | 606870 | Basal ganglia calcification, idiopathic, 8, autosomal recessive, 618824 (3), Autosomal recessive |

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| *KANK1* | 607704 | Cerebral palsy, spastic quadriplegic, 2, 612900 (3) |
| *KCNA1* | 176260 | Episodic ataxia/myokymia syndrome, 160120 (3), Autosomal dominant |
| *KCNA2* | 176262 | Epileptic encephalopathy, early infantile, 32, 616366 (3), Autosomal dominant |
| *KCNA4* | 176266 | Microcephaly, cataracts, impaired intellectual development, and dystonia with abnormal striatum, 618284 (3), Autosomal recessive |
| *KCNC3* | 176264 | Spinocerebellar ataxia 13, 605259 (3), Autosomal dominant |
| *KCND3* | 605411 | Brugada syndrome 9, 616399 (3), Autosomal dominant; Spinocerebellar ataxia 19, 607346 (3), Autosomal dominant |
| *KCNJ10* | 602208 | Enlarged vestibular aqueduct, digenic, 600791 (3), Autosomal recessive; SESAME syndrome, 612780 (3), Autosomal recessive |
| *KCNJ6* | 600877 | Keppen-Lubinsky syndrome, 614098 (3), Autosomal dominant |
| *KCNMA1* | 600150 | Liang-Wang syndrome, 618729 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 16}, 618596 (3), Autosomal dominant; Cerebellar atrophy, developmental delay, and seizures, 617643 (3), Autosomal recessive; Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy, 609446 (3), Autosomal dominant |
| *KCNQ2* | 602235 | Epileptic encephalopathy, early infantile, 7, 613720 (3), Autosomal dominant; Seizures, benign neonatal, 1, 121200 (3), Autosomal dominant; Myokymia, 121200 (3), Autosomal dominant |
| *KCTD7* | 611725 | Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726 (3), Autosomal recessive |
| *KIDINS220* | 615759 | Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296 (3), Autosomal dominant |
| *KIF1A* | 601255 | NESCAV syndrome, 614255 (3), Autosomal dominant; Spastic paraplegia 30, autosomal dominant, 610357 (3), Autosomal dominant, Autosomal recessive; Neuropathy, hereditary sensory, type IIC, 614213 (3), Autosomal recessive; Spastic paraplegia 30, autosomal recessive, 610357 (3), Autosomal dominant, Autosomal recessive |
| *KIF1C* | 603060 | Spastic ataxia 2, autosomal recessive, 611302 (3), Autosomal recessive |
| *KIF5A* | 602821 | Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant |
| *KLC2* | 611729 | Spastic paraplegia, optic atrophy, and neuropathy, 609541 (3), Autosomal recessive |

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| *L1CAM* | 308840 | MASA syndrome, 303350 (3), X-linked recessive; Hydrocephalus with Hirschsprung disease, 307000 (3), X-linked recessive; Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3), X-linked recessive; Corpus callosum, partial agenesis of, 304100 (3), X-linked recessive; CRASH syndrome, 303350 (3), X-linked recessive; Hydrocephalus due to aqueductal stenosis, 307000 (3), X-linked recessive |
| *L2HGDH* | 609584 | L-2-hydroxyglutaric aciduria, 236792 (3), Autosomal recessive |
| *LAMA1* | 150320 | Poretti-Boltshauser syndrome, 615960 (3), Autosomal recessive |
| *LAMB1* | 150240 | Lissencephaly 5, 615191 (3), Autosomal recessive |
| *LIPT2* | 617659 | Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities, 617668 (3), Autosomal recessive |
| *LYRM7* | 615831 | Mitochondrial complex III deficiency, nuclear type 8, 615838 (3), Autosomal recessive |
| *LYST* | 606897 | Chediak-Higashi syndrome, 214500 (3), Autosomal recessive |
| *MAG* | 159460 | Spastic paraplegia 75, autosomal recessive, 616680 (3), Autosomal recessive |
| *MAN2B1* | 609458 | Mannosidosis, alpha-, types I and II, 248500 (3), Autosomal recessive |
| *MARS1 (MARS)* | 156560 | Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant; Interstitial lung and liver disease, 615486 (3), Autosomal recessive |
| *MARS2* | 609728 | Spastic ataxia 3, autosomal recessive, 611390 (3), Autosomal recessive; ?Combined oxidative phosphorylation deficiency 25, 616430 (3), Autosomal recessive |
| *MCOLN1* | 605248 | Mucolipidosis IV, 252650 (3), Autosomal recessive |
| *MECP2* | 300005 | Mental retardation, X-linked syndromic, Lubs type, 300260 (3), X-linked recessive; Encephalopathy, neonatal severe, 300673 (3), X-linked recessive; Mental retardation, X-linked, syndromic 13, 300055 (3), X-linked recessive; Rett syndrome, atypical, 312750 (3), X-linked dominant; {Autism susceptibility, X-linked 3}, 300496 (3), X-linked; Rett syndrome, 312750 (3), X-linked dominant; Rett syndrome, preserved speech variant, 312750 (3), X-linked dominant |
| *MLC1* | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3), Autosomal recessive |
| *MMADHC* | 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 |
| *MME* | 120520 | Charcot-Marie-Tooth disease, axonal, type 2T, 617017 (3), Autosomal dominant, Autosomal recessive; ?Spinocerebellar ataxia 43, 617018 (3), Autosomal dominant |
| *MPV17* | 137960 | Charcot-Marie-Tooth disease, axonal, type 2EE, 618400 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810 (3), Autosomal recessive |

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| *MRE11* | 600814 | Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive |
| *MRPS34* | 611994 | Combined oxidative phosphorylation deficiency 32, 617664 (3), Autosomal recessive |
| *MSTO1* | 617619 | Myopathy, mitochondrial, and ataxia, 617675 (3), Autosomal dominant, Autosomal recessive |
| *MTCL1* | 615766 | No OMIM phenotype |
| *MTPAP* | 613669 | ?Spastic ataxia 4, autosomal recessive, 613672 (3), Autosomal recessive |
| *MTTP* | 157147 | Abetalipoproteinemia, 200100 (3), Autosomal recessive; {Metabolic syndrome, protection against}, 605552 (3), Autosomal dominant |
| *MYORG* | 618255 | Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 (3), Autosomal recessive |
| *NANS* | 605202 | Spondyloepimetaphyseal dysplasia, Camera-Genevieve type, 610442 (3), Autosomal recessive |
| *NDUFA9* | 603834 | Mitochondrial complex I deficiency, nuclear type 26, 618247 (3), Autosomal recessive |
| *NDUFAF4* | 611776 | Mitochondrial complex I deficiency, nuclear type 15, 618237 (3), Autosomal recessive |
| *NDUFAF6* | 612392 | Fanconi renotubular syndrome 5, 618913 (3), Autosomal recessive; Mitochondrial complex I deficiency, nuclear type 17, 618239 (3), Autosomal recessive |
| *NDUFS1* | 157655 | Mitochondrial complex I deficiency, nuclear type 5, 618226 (3), Autosomal recessive |
| *NDUFS3* | 603846 | Mitochondrial complex I deficiency, nuclear type 8, 618230 (3), Autosomal recessive |
| *NDUFS7* | 601825 | Mitochondrial complex I deficiency, nuclear type 3, 618224 (3), Autosomal recessive |
| *NEFL* | 162280 | Charcot-Marie-Tooth disease, type 1F, 607734 (3), Autosomal dominant, Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate G, 617882 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2E, 607684 (3), Autosomal dominant |
| *NEU1* | 608272 | Sialidosis, type II, 256550 (3), Autosomal recessive; Sialidosis, type I, 256550 (3), Autosomal recessive |
| *NEXMIF* | 300524 | Mental retardation, X-linked 98, 300912 (3), X-linked dominant |
| *NF2* | 607379 | Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, somatic, 162091 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant |
| *NHLRC1* | 608072 | Epilepsy, progressive myoclonic 2B (Lafora), 254780 (3), Autosomal recessive |
| *NIPA1* | 608145 | Spastic paraplegia 6, autosomal dominant, 600363 (3), Autosomal dominant |
| *NKX6-2* | 605955 | Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy, 617560 (3), Autosomal recessive |

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| *NOL3* | 605235 | ?Myoclonus, familial, 1, 614937 (3), Autosomal dominant |
| *NPC1* | 607623 | Niemann-Pick disease, type D, 257220 (3), Autosomal recessive; Niemann-Pick disease, type C1, 257220 (3), Autosomal recessive |
| *NPC2* | 601015 | Niemann-pick disease, type C2, 607625 (3), Autosomal recessive |
| *NT5C2* | 600417 | Spastic paraplegia 45, autosomal recessive, 613162 (3), Autosomal recessive |
| *NTNG2* | 618689 | Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia, 618718 (3), Autosomal recessive |
| *NUP93* | 614351 | Nephrotic syndrome, type 12, 616892 (3), Autosomal recessive |
| *NUS1* | 610463 | Mental retardation, autosomal dominant 55, with seizures, 617831 (3), Autosomal dominant; ?Congenital disorder of glycosylation, type 1aa, 617082 (3), Autosomal recessive |
| *OCLN* | 602876 | Pseudo-TORCH syndrome 1, 251290 (3), Autosomal recessive |
| *OFD1* | 300170 | Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive |
| *OPA1* | 605290 | {Glaucoma, normal tension, susceptibility to}, 606657 (3); Behr syndrome, 210000 (3), Autosomal recessive; Optic atrophy 1, 165500 (3), Autosomal dominant; Optic atrophy plus syndrome, 125250 (3), Autosomal dominant; ?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896 (3), Autosomal recessive |
| *OPA3* | 606580 | 3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant |
| *OPHN1* | 300127 | Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486 (3), X-linked recessive |
| *OTC* | 300461 | Ornithine transcarbamylase deficiency, 311250 (3), X-linked recessive |
| *PARS2* | 612036 | Epileptic encephalopathy, early infantile, 75, 618437 (3), Autosomal recessive |
| *PC* | 608786 | Pyruvate carboxylase deficiency, 266150 (3), Autosomal recessive |
| *PCDH12* | 605622 | Diencephalic-mesencephalic junction dysplasia syndrome 1, 251280 (3), Autosomal recessive |
| *PCYT2* | 602679 | Spastic paraplegia 82, autosomal recessive, 618770 (3), Autosomal recessive |
| *PDHA1* | 300502 | Pyruvate dehydrogenase E1-alpha deficiency, 312170 (3), X-linked dominant |
| *PDHX* | 608769 | Lacticacidemia due to PDX1 deficiency, 245349 (3), Autosomal recessive |
| *PDYN* | 131340 | Spinocerebellar ataxia 23, 610245 (3), Autosomal dominant |
| *PEX10* | 602859 | Peroxisome biogenesis disorder 6B, 614871 (3), Autosomal recessive; Peroxisome biogenesis disorder 6A (Zellweger), 614870 (3), Autosomal recessive |

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| *PEX16* | 603360 | Peroxisome biogenesis disorder 8A (Zellweger), 614876 (3), Autosomal recessive; Peroxisome biogenesis disorder 8B, 614877 (3), Autosomal recessive |
| *PEX7* | 601757 | Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive |
| *PGAP1* | 611655 | Mental retardation, autosomal recessive 42, 615802 (3), Autosomal recessive |
| *PHYH* | 602026 | Refsum disease, 266500 (3), Autosomal recessive |
| *PIK3R5* | 611317 | Ataxia-oculomotor apraxia 3, 615217 (3), Autosomal recessive |
| *PLA2G6* | 603604 | Infantile neuroaxonal dystrophy 1, 256600 (3), Autosomal recessive; Parkinson disease 14, autosomal recessive, 612953 (3), Autosomal recessive; Neurodegeneration with brain iron accumulation 2B, 610217 (3), Autosomal recessive |
| *PLAA* | 603873 | Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies, 617527 (3), Autosomal recessive |
| *PLD3* | 615698 | ?Spinocerebellar ataxia 46, 617770 (3), Autosomal dominant |
| *PLP1* | 300401 | Pelizaeus-Merzbacher disease, 312080 (3), X-linked recessive; Spastic paraplegia 2, X-linked, 312920 (3), X-linked recessive |
| *PMM2* | 601785 | Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive |
| *PMPCA* | 613036 | Spinocerebellar ataxia, autosomal recessive 2, 213200 (3), Autosomal recessive |
| *PMPCB* | 603131 | Multiple mitochondrial dysfunctions syndrome 6, 617954 (3), Autosomal recessive |
| *PNKP* | 605610 | Microcephaly, seizures, and developmental delay, 613402 (3), Autosomal recessive; Ataxia-oculomotor apraxia 4, 616267 (3), Autosomal recessive; ?Charcot-Marie-Tooth disease, type 2B2, 605589 (3), Autosomal recessive |
| *PNPLA6* | 603197 | Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive |
| *POLG* | 174763 | Progressive external ophthalmoplegia, autosomal dominant 1, 157640 (3), Autosomal dominant; Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 (3), Autosomal recessive; Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 (3), Autosomal recessive; Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 (3), Autosomal recessive; Progressive external ophthalmoplegia, autosomal recessive 1, 258450 (3), Autosomal recessive |
| *POLR3A* | 614258 | Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (3), Autosomal recessive |

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| *POLR3B* | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 (3), Autosomal recessive |
| *PRF1* | 170280 | Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3), Autosomal recessive; Aplastic anemia, 609135 (3); Lymphoma, non-Hodgkin, 605027 (3) |
| *PRKCG* | 176980 | Spinocerebellar ataxia 14, 605361 (3), Autosomal dominant |
| *PRNP* | 176640 | Insomnia, fatal familial, 600072 (3), Autosomal dominant; {Kuru, susceptibility to}, 245300 (3); Huntington disease-like 1, 603218 (3), Autosomal dominant; Prion disease with protracted course, 606688 (3), Autosomal dominant; Cerebral amyloid angiopathy, PRNP-related, 137440 (3), Autosomal dominant; Creutzfeldt-Jakob disease, 123400 (3), Autosomal dominant; Gerstmann-Straussler disease, 137440 (3), Autosomal dominant |
| *PRPS1* | 311850 | Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070 (3), X-linked recessive; Phosphoribosylpyrophosphate synthetase superactivity, 300661 (3), X-linked recessive; Deafness, X-linked 1, 304500 (3), X-linked; Arts syndrome, 301835 (3), X-linked recessive; Gout, PRPS-related, 300661 (3), X-linked recessive |
| *PRRT2* | 614386 | Episodic kinesigenic dyskinesia 1, 128200 (3), Autosomal dominant; Seizures, benign familial infantile, 2, 605751 (3), Autosomal dominant; Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066 (3), Autosomal dominant |
| *PSAP* | 176801 | Gaucher disease, atypical, 610539 (3); Krabbe disease, atypical, 611722 (3), Autosomal recessive; Combined SAP deficiency, 611721 (3), Autosomal recessive; Metachromatic leukodystrophy due to SAP-b deficiency, 249900 (3), Autosomal recessive |
| *PSEN1* | 104311 | Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3), Autosomal dominant; Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3), Autosomal dominant; ?Acne inversa, familial, 3, 613737 (3), Autosomal dominant; Alzheimer disease, type 3, 607822 (3), Autosomal dominant; Dementia, frontotemporal, 600274 (3), Autosomal dominant; Pick disease, 172700 (3), Autosomal dominant; Cardiomyopathy, dilated, 1U, 613694 (3), Autosomal dominant |
| *PTPN23* | 606584 | Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity, 618890 (3), Autosomal recessive |
| *PTRH2* | 608625 | Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive |
| *PUM1* | 607204 | Spinocerebellar ataxia 47, 617931 (3), Autosomal dominant |
| *RAB11B* | 604198 | Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter, 617807 (3), Autosomal dominant |
| *RAB18* | 602207 | Warburg micro syndrome 3, 614222 (3), Autosomal recessive |
| *RAB3GAP1* | 602536 | Warburg micro syndrome 1, 600118 (3), Autosomal recessive |
| *RAB3GAP2* | 609275 | Warburg micro syndrome 2, 614225 (3), Autosomal recessive; Martsolf syndrome, 212720 (3), Autosomal recessive |

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| *REEP1* | 609139 | Spastic paraplegia 31, autosomal dominant, 610250 (3), Autosomal dominant; ?Neuronopathy, distal hereditary motor, type VB, 614751 (3), Autosomal dominant |
| *REEP2* | 609347 | ?Spastic paraplegia 72, autosomal dominant, 615625 (3), Autosomal dominant, Autosomal recessive; ?Spastic paraplegia 72, autosomal recessive, 615625 (3), Autosomal dominant, Autosomal recessive |
| *RETREG1* | 613114 | Neuropathy, hereditary sensory and autonomic, type IIB, 613115 (3), Autosomal recessive |
| *RNASEH2A* | 606034 | Aicardi-Goutieres syndrome 4, 610333 (3), Autosomal recessive |
| *RNASEH2B* | 610326 | Aicardi-Goutieres syndrome 2, 610181 (3), Autosomal recessive |
| *RNASEH2C* | 610330 | Aicardi-Goutieres syndrome 3, 610329 (3), Autosomal recessive |
| *RNF168* | 612688 | RIDDLE syndrome, 611943 (3), Autosomal recessive |
| *RNF170* | 614649 | Ataxia, sensory, 1, autosomal dominant, 608984 (3), Autosomal dominant |
| *RNF216* | 609948 | Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive |
| *RORA* | 600825 | Intellectual developmental disorder with or without epilepsy or cerebellar ataxia, 618060 (3), Autosomal dominant |
| *RPGRIP1L* | 610937 | COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal recessive |
| *RTN2* | 603183 | Spastic paraplegia 12, autosomal dominant, 604805 (3), Autosomal dominant |
| *RTN4IP1* | 610502 | Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732 (3), Autosomal recessive |
| *RUBCN* | 613516 | ?Spinocerebellar ataxia, autosomal recessive 15, 615705 (3), Autosomal recessive |
| *SACS* | 604490 | Spastic ataxia, Charlevoix-Saguenay type, 270550 (3), Autosomal recessive |
| *SAMD9L* | 611170 | Ataxia-pancytopenia syndrome, 159550 (3), Autosomal dominant |
| *SAMHD1* | 606754 | ?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive |
| *SARS1 (SARS)* | 607529 | ?Neurodevelopmental disorder with microcephaly, ataxia, and seizures, 617709 (3), Autosomal recessive |
| *SCN8A* | 600702 | Seizures, benign familial infantile, 5, 617080 (3), Autosomal dominant; Cognitive impairment with or without cerebellar ataxia, 614306 (3), Autosomal dominant; ?Myoclonus, familial, 2, 618364 (3), Autosomal dominant; Epileptic encephalopathy, early infantile, 13, 614558 (3), Autosomal dominant |
| *SCYL1* | 607982 | Spinocerebellar ataxia, autosomal recessive 21, 616719 (3), Autosomal recessive |

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| *SDHA* | 600857 | Cardiomyopathy, dilated, 1GG, 613642 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Paragangliomas 5, 614165 (3), Autosomal dominant; Mitochondrial respiratory chain complex II deficiency, 252011 (3), Autosomal recessive |
| *SDHAF1* | 612848 | Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive |
| *SDHD* | 602690 | Paragangliomas 1, with or without deafness, 168000 (3), Autosomal dominant; Mitochondrial complex II deficiency, 252011 (3), Autosomal recessive; Paraganglioma and gastric stromal sarcoma, 606864 (3); Pheochromocytoma, 171300 (3), Autosomal dominant |
| *SEPSECS* | 613009 | Pontocerebellar hypoplasia type 2D, 613811 (3), Autosomal recessive |
| *SETX* | 608465 | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant |
| *SIL1* | 608005 | Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive |
| *SLC13A5* | 608305 | Epileptic encephalopathy, early infantile, 25, 615905 (3), Autosomal recessive |
| *SLC16A2* | 300095 | Allan-Herndon-Dudley syndrome, 300523 (3), X-linked |
| *SLC17A5* | 604322 | Sialic acid storage disorder, infantile, 269920 (3), Autosomal recessive; Salla disease, 604369 (3), Autosomal recessive |
| *SLC19A3* | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483 (3), Autosomal recessive |
| *SLC1A3* | 600111 | Episodic ataxia, type 6, 612656 (3), Autosomal dominant |
| *SLC1A4* | 600229 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657 (3), Autosomal recessive |
| *SLC25A15* | 603861 | Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3), Autosomal recessive |
| *SLC25A46* | 610826 | Neuropathy, hereditary motor and sensory, type VIB, 616505 (3), Autosomal recessive |
| *SLC2A1* | 138140 | Dystonia 9, 601042 (3), Autosomal dominant; GLUT1 deficiency syndrome 1, infantile onset, severe, 606777 (3), Autosomal dominant, Autosomal recessive; Stomatin-deficient cryohydrocytosis with neurologic defects, 608885 (3), Autosomal dominant; GLUT1 deficiency syndrome 2, childhood onset, 612126 (3), Autosomal dominant; {Epilepsy, idiopathic generalized, susceptibility to, 12}, 614847 (3), Autosomal dominant |
| *SLC33A1* | 603690 | Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive |
| *SLC44A1* | 606105 | Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline, 618868 (3), Autosomal recessive |
| *SLC52A2* | 607882 | Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive |

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| *SLC52A3* | 613350 | Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive; ?Fazio-Londe disease, 211500 (3), Autosomal recessive |
| *SLC6A8* | 300036 | Cerebral creatine deficiency syndrome 1, 300352 (3), X-linked recessive |
| *SLC9A1* | 107310 | ?Lichtenstein-Knorr syndrome, 616291 (3), Autosomal recessive |
| *SLC9A6* | 300231 | Mental retardation, X-linked syndromic, Christianson type, 300243 (3), X-linked dominant |
| *SMPD1* | 607608 | Niemann-Pick disease, type A, 257200 (3), Autosomal recessive; Niemann-Pick disease, type B, 607616 (3), Autosomal recessive |
| *SNAP25* | 600322 | ?Myasthenic syndrome, congenital, 18, 616330 (3), Autosomal dominant |
| *SNX14* | 616105 | Spinocerebellar ataxia, autosomal recessive 20, 616354 (3), Autosomal recessive |
| *SOX10* | 602229 | Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant |
| *SPART* | 607111 | Troyer syndrome, 275900 (3), Autosomal recessive |
| *SPAST* | 604277 | Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant |
| *SPG11* | 610844 | Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive; Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive |
| *SPG21* | 608181 | Mast syndrome, 248900 (3), Autosomal recessive |
| *SPG7* | 602783 | Spastic paraplegia 7, autosomal recessive, 607259 (3), Autosomal dominant, Autosomal recessive |
| *SPR* | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716 (3), Autosomal recessive, ?Autosomal dominant |
| *SPTAN1* | 182810 | Epileptic encephalopathy, early infantile, 5, 613477 (3), Autosomal dominant |
| *SPTBN2* | 604985 | Spinocerebellar ataxia, autosomal recessive 14, 615386 (3), Autosomal recessive; Spinocerebellar ataxia 5, 600224 (3), Autosomal dominant |
| *SQSTM1* | 601530 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant |
| *SRD5A3* | 611715 | Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive |
| *STUB1* | 607207 | Spinocerebellar ataxia, autosomal recessive 16, 615768 (3), Autosomal recessive; ?Spinocerebellar ataxia 48, 618093 (3), Autosomal dominant |

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| *SUCLA2* | 603921 | Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073 (3), Autosomal recessive |
| *SUOX* | 606887 | Sulfite oxidase deficiency, 272300 (3), Autosomal recessive |
| *SURF1* | 185620 | Leigh syndrome, due to COX IV deficiency, 256000 (3), Autosomal recessive, Mitochondrial; Charcot-Marie-Tooth disease, type 4K, 616684 (3), Autosomal recessive |
| *SVBP* | 617853 | Neurodevelopmental disorder with ataxia, hypotonia, and microcephaly, 618569 (3), Autosomal recessive |
| *SYNE1* | 608441 | Arthrogryposis multiplex congenita 3, myogenic type, 618484 (3), Autosomal recessive; Spinocerebellar ataxia, autosomal recessive 8, 610743 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998 (3), Autosomal dominant |
| *SYT14* | 610949 | ?Spinocerebellar ataxia, autosomal recessive 11, 614229 (3), Autosomal recessive |
| *TANGO2* | 616830 | Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878 (3), Autosomal recessive |
| *TBC1D20* | 611663 | Warburg micro syndrome 4, 615663 (3), Autosomal recessive |
| *TDP1* | 607198 | ?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1, 607250 (3), Autosomal recessive |
| *TDP2* | 605764 | Spinocerebellar ataxia, autosomal recessive 23, 616949 (3), Autosomal recessive |
| *TECPR2* | 615000 | Spastic paraplegia 49, autosomal recessive, 615031 (3), Autosomal recessive |
| *TENM3* | 610083 | Microphthalmia, syndromic 15, 615145 (3), Autosomal recessive; ?Microphthalmia, isolated, with coloboma 9, 615145 (3), Autosomal recessive |
| *TFG* | 602498 | ?Spastic paraplegia 57, autosomal recessive, 615658 (3), Autosomal recessive; Hereditary motor and sensory neuropathy, Okinawa type, 604484 (3), Autosomal dominant |
| *TGM6* | 613900 | Spinocerebellar ataxia 35, 613908 (3), Autosomal dominant |
| *THG1L* | 618802 | Spinocerebellar ataxia, autosomal recessive 28, 618800 (3), Autosomal recessive |
| *TMEM216* | 613277 | Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive |
| *TMEM231* | 614949 | Meckel syndrome 11, 615397 (3), Autosomal recessive; Joubert syndrome 20, 614970 (3), Autosomal recessive |
| *TMEM240* | 616101 | Spinocerebellar ataxia 21, 607454 (3), Autosomal dominant |
| *TMEM67* | 609884 | Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, |

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| | | 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive |
| *TMX2* | 616715 | Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity, 618730 (3), Autosomal recessive |
| *TPP1* | 607998 | Spinocerebellar ataxia, autosomal recessive 7, 609270 (3), Autosomal recessive; Ceroid lipofuscinosis, neuronal, 2, 204500 (3), Autosomal recessive |
| *TRAK1* | 608112 | Epileptic encephalopathy, early infantile, 68, 618201 (3), Autosomal recessive |
| *TRAPPC12* | 614139 | Encephalopathy, progressive, early-onset, with brain atrophy and spasticity, 617669 (3), Autosomal recessive |
| *TRAPPC4* | 610971 | Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy, 618741 (3), Autosomal recessive |
| *TRIT1* | 617840 | Combined oxidative phosphorylation deficiency 35, 617873 (3), Autosomal recessive |
| *TRPC3* | 602345 | ?Spinocerebellar ataxia 41, 616410 (3), Autosomal dominant |
| *TSEN2* | 608753 | Pontocerebellar hypoplasia type 2B, 612389 (3), Autosomal recessive |
| *TSEN54* | 608755 | Pontocerebellar hypoplasia type 4, 225753 (3), Autosomal recessive; Pontocerebellar hypoplasia type 2A, 277470 (3), Autosomal recessive; ?Pontocerebellar hypoplasia type 5, 610204 (3), Autosomal recessive |
| *TTBK2* | 611695 | Spinocerebellar ataxia 11, 604432 (3), Autosomal dominant |
| *TTC19* | 613814 | Mitochondrial complex III deficiency, nuclear type 2, 615157 (3), Autosomal recessive |
| *TTPA* | 600415 | Ataxia with isolated vitamin E deficiency, 277460 (3), Autosomal recessive |
| *TUBA1A* | 602529 | Lissencephaly 3, 611603 (3), Autosomal dominant |
| *TUBB4A* | 602662 | Leukodystrophy, hypomyelinating, 6, 612438 (3), Autosomal dominant; Dystonia 4, torsion, autosomal dominant, 128101 (3), Autosomal dominant |
| *TUBG1* | 191135 | Cortical dysplasia, complex, with other brain malformations 4, 615412 (3), Autosomal dominant |
| *TWNK* | 606075 | Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive |
| *UBA5* | 610552 | ?Spinocerebellar ataxia, autosomal recessive 24, 617133 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 44, 617132 (3), Autosomal recessive |
| *UBAP1* | 609787 | Spastic paraplegia 80, autosomal dominant, 618418 (3), Autosomal dominant |

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| *UBTF* | 600673 | Neurodegeneration, childhood-onset, with brain atrophy, 617672 (3), Autosomal dominant |
| *UCHL1* | 191342 | Spastic paraplegia 79, autosomal recessive, 615491 (3), Autosomal recessive; {?Parkinson disease 5, susceptibility to}, 613643 (3), Autosomal dominant |
| *UFC1* | 610554 | Neurodevelopmental disorder with spasticity and poor growth, 618076 (3), Autosomal recessive |
| *UFM1* | 610553 | Leukodystrophy, hypomyelinating, 14, 617899 (3), Autosomal recessive |
| *UGP2* | 191760 | Epileptic encephalopathy, early infantile, 83, 618744 (3), Autosomal recessive |
| *USP8* | 603158 | Pituitary adenoma 4, ACTH-secreting, somatic, 219090 (3) |
| *VAMP1* | 185880 | Spastic ataxia 1, autosomal dominant, 108600 (3), Autosomal dominant; Myasthenic syndrome, congenital, 25, 618323 (3), Autosomal recessive |
| *VCP* | 601023 | Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954 (3) |
| *VLDLR* | 192977 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050 (3), Autosomal recessive |
| *VPS13B* | 607817 | Cohen syndrome, 216550 (3), Autosomal recessive |
| *VPS13D* | 608877 | Spinocerebellar ataxia, autosomal recessive 4, 607317 (3), Autosomal recessive |
| *VPS37A* | 609927 | Spastic paraplegia 53, autosomal recessive, 614898 (3), Autosomal recessive |
| *VRK1* | 602168 | Pontocerebellar hypoplasia type 1A, 607596 (3), Autosomal recessive |
| *VWA3B* | 614884 | ?Spinocerebellar ataxia, autosomal recessive 22, 616948 (3), Autosomal recessive |
| *WARS2* | 604733 | Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures, 617710 (3), Autosomal recessive |
| *WASHC5* | 610657 | Ritscher-Schinzel syndrome 1, 220210 (3), Autosomal recessive; Spastic paraplegia 8, autosomal dominant, 603563 (3), Autosomal dominant |
| *WDR45B* | 609226 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures, 617977 (3), Autosomal recessive |
| *WDR48* | 612167 | No OMIM phenotype |
| *WDR73* | 616144 | Galloway-Mowat syndrome 1, 251300 (3), Autosomal recessive |
| *WDR81* | 614218 | Hydrocephalus, congenital, 3, with brain anomalies, 617967 (3), Autosomal recessive; Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185 (3), Autosomal recessive |

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| *WWOX* | 605131 | Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive; Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3) |
| *XRCC1* | 194360 | ?Spinocerebellar ataxia, autosomal recessive 26, 617633 (3), Autosomal recessive |
| *ZC4H2* | 300897 | Wieacker-Wolff syndrome, 314580 (3), X-linked recessive; Wieacker-Wolff syndrome, female-restricted, 301041 (3), X-linked dominant |
| *ZFR* | 615635 | No OMIM phenotype |
| *ZFYVE26* | 612012 | Spastic paraplegia 15, autosomal recessive, 270700 (3), Autosomal recessive |
| *ZFYVE27* | 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: Aug 20, 2020

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.