## Panel de ataxia/paraplejía espástica

Nuestro panel de ataxia/paraplejía espástica incluye genes relevantes para trastornos neurológicos hereditarios caracterizados por ataxia, incluida la ataxia espinocerebelosa (dominante y recesiva), ataxia cerebelosa, ataxia episódica y ataxia pontocerebelosa. Estos trastornos normalmente comparten síntomas superpuestos y solo pueden diferenciarse claramente mediante pruebas genéticas moleculares. Nuestro panel de ataxia es la mejor opción para un paciente que muestra un desequilibrio en la marcha y una marcha descoordinada (ataxia). Las formas más comunes de ataxia hereditaria son causadas por expansión repetida.

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| **Nº de genes:** | 481 |
| **Entrega:** | 25 días |
| **Cobertura:** | ≥99.5% ≥20x<br>Cobertura media con profundidad ≥150 x |
| **Detalles:** | Secuenciación de nueva generación, análisis CNV incluido |

## Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel

| Genes | OMIM (Gene) | Associated diseases (OMIM) | Inheritance |
|---|---|---|---|
| AARS2 | 612035 | Combined oxidative phosphorylation deficiency 8;Leukoencephalopathy, progressive, with ovarian failure | AR |
| ABCB7 | 300135 | Anemia, sideroblastic, with ataxia | XLR |
| ABCD1 | 300371 | Adrenoleukodystrophy;Adrenomyeloneuropathy, adult | XLR |
| ABHD12 | 613599 | Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract | AR |
| ABHD5 | 604780 | Chanarin-Dorfman syndrome | AR |
| ACAD9 | 611103 | Mi complex I deficiency, nuclear type 20 | AR |

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| ACADVL | 609575 | VLCAD deficiency | AR |
| ACO2 | 100850 | ?Optic atrophy 9;Infantile cerebellar-retinal degeneration | AR |
| ADAR | 146920 | Aicardi-Goutieres syndrome 6;Dyschromatosis symmetrica hereditaria | AR, AD |
| ADPRS | 610624 | Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures | AR |
| AFG3L2 | 604581 | Spinocerebellar ataxia 28;Spastic ataxia 5, AR;Optic atrophy 12 | AD, AR |
| AGK | 610345 | Sengers syndrome;Cataract 38, AR | AR |
| AGTPBP1 | 606830 | Neurodegeneration, childhood-onset, with cerebellar atrophy | AR |
| AHI1 | 608894 | Joubert syndrome 3 | AR |
| AIFM1 | 300169 | Cowchock syndrome;Combined oxidative phosphorylation deficiency 6;Deafness, XL 5;Spondyloepimetaphyseal dysplasia, XL, with hypomyelinating leukodystrophy | XLR |
| AIMP1 | 603605 | Leukodystrophy, hypomyelinating, 3 | AR |
| ALAS2 | 301300 | Protoporphyria, erythropoietic, XL;Anemia, sideroblastic, 1 | XL, XLR |
| ALDH18A1 | 138250 | Cutis laxa, AD 3;Cutis laxa, AR, type IIIA;Spastic paraplegia 9B, AR;Spastic paraplegia 9A, AD | AD, AR |
| ALDH5A1 | 610045 | Succinic semialdehyde dehydrogenase deficiency | AR |
| ALS2 | 606352 | Amyotrophic lateral sclerosis 2, juvenile;Spastic paralysis, infantile onset ascending;Primary lateral sclerosis, juvenile | AR |
| AMACR | 604489 | Bile acid synthesis defect, congenital, 4;Alpha-methylacyl-CoA racemase deficiency | AR |
| AMPD2 | 102771 | ?Spastic paraplegia 63;Pontocerebellar hypoplasia, type 9 | AR |

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| ANO10 | 613726 | Spinocerebellar ataxia, AR 10 | AR |
| AP1S2 | 300629 | Mental retardation, XL syndromic 5 | XLR |
| AP4B1 | 607245 | Spastic paraplegia 47, AR | AR |
| AP4E1 | 607244 | Stuttering, familial persistent, 1;Spastic paraplegia 51, AR | AD, AR |
| AP4M1 | 602296 | Spastic paraplegia 50, AR | AR |
| AP4S1 | 607243 | Spastic paraplegia 52, AR | AR |
| AP5Z1 | 613653 | Spastic paraplegia 48, AR | AR |
| APTX | 606350 | Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia | AR |
| ARG1 | 608313 | Argininemia | AR |
| ARL13B | 608922 | Joubert syndrome 8 | AR |
| ARL6 | 608845 | Retinitis pigmentosa 55;Bardet-Biedl syndrome 1, modifier of;Bardet-Biedl syndrome 3 | AR, AR, DR |
| ARL6IP1 | 607669 | ?Spastic paraplegia 61, AR | AR |
| ARSA | 607574 | Metachromatic leukodystrophy | AR |
| ATCAY | 608179 | Ataxia, cerebellar, Cayman type | AR |
| ATL1 | 606439 | Spastic paraplegia 3A, AD;Neuropathy, hereditary sensory, type ID | AD |
| ATM | 607585 | Breast cancer, susceptibility to;Ataxia-telangiectasia | AD, SM, AR |
| ATP13A2 | 610513 | Spastic paraplegia 78, AR;Kufor-Rakeb syndrome | AR |

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| ATP1A2 | 182340 | Developmental and epileptic encephalopathy 98;Alternating hemiplegia of childhood 1;Migraine, familial basilar;Migraine, familial hemiplegic, 2;Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies | AD, AR |
| ATP1A3 | 182350 | Alternating hemiplegia of childhood 2;Dystonia-12;Developmental and epileptic encephalopathy 99;CAPOS syndrome | AD |
| ATP2B3 | 300014 | ?Spinocerebellar ataxia, XL 1 | XLR |
| ATP2B4 | 108732 | | |
| ATP7B | 606882 | Wilson disease | AR |
| ATP8A2 | 605870 | ?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 | AR |
| ATRX | 300032 | Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic | XLR, XLD |
| AUH | 600529 | 3-methylglutaconic aciduria, type I | AR |
| B4GALNT1 | 601873 | Spastic paraplegia 26, AR | AR |
| B9D1 | 614144 | ?Meckel syndrome 9;Joubert syndrome 27 | AR |
| BBS1 | 209901 | Bardet-Biedl syndrome 1 | AR, DR |
| BBS10 | 610148 | Bardet-Biedl syndrome 10 | AR |
| BBS12 | 610683 | Bardet-Biedl syndrome 12 | AR |
| BBS2 | 606151 | Bardet-Biedl syndrome 2;Retinitis pigmentosa 74 | AR |
| BBS4 | 600374 | Bardet-Biedl syndrome 4 | AR |

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| BBS5 | 603650 | Bardet-Biedl syndrome 5 | AR |
| BBS7 | 607590 | Bardet-Biedl syndrome 7 | AR |
| BBS9 | 607968 | Bardet-Biedl syndrome 9 | AR |
| BCKDHA | 608348 | Maple syrup urine disease, type Ia | AR |
| BCKDHB | 248611 | Maple syrup urine disease, type Ib | AR |
| BCS1L | 603647 | GRACILE syndrome;Bjornstad syndrome;Mi complex III deficiency, nuclear type 1 | AR |
| BICD2 | 609797 | Spinal muscular atrophy, lower extremity-predominant, 2A, AD;Spinal muscular atrophy, lower extremity-predominant, 2B, AD | AD |
| BLOC1S1 | 601444 | | |
| BOLA3 | 613183 | Multiple Mi dysfunctions syndrome 2 with hyperglycinemia | AR |
| BSCL2 | 606158 | Lipodystrophy, congenital generalized, type 2;Encephalopathy, progressive, with or without lipodystrophy;Silver spastic paraplegia syndrome;Neuropathy, distal hereditary motor, type VC | AR, AD |
| BTD | 609019 | Biotinidase deficiency | AR |
| C19orf12 | 614297 | Neurodegeneration with brain iron accumulation 4;?Spastic paraplegia 43, AR | AD, AR, AR |
| CA8 | 114815 | Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3 | AR |
| CACNA1A | 601011 | Episodic ataxia, type 2;Migraine, familial hemiplegic, 1;Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia;Spinocerebellar ataxia 6;Developemental and epileptic encephalopathy 42 | AD |

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| CACNA1G | 604065 | Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits;Spinocerebellar ataxia 42 | AD |
| CACNB4 | 601949 | Epilepsy, juvenile myoclonic, susceptibility to, 6;Epilepsy, idiopathic generalized, susceptibility to, 9;Episodic ataxia, type 5 | AD |
| CAMTA1 | 611501 | Cerebellar ataxia, nonprogressive, with mental retardation | AD |
| CAPN1 | 114220 | Spastic paraplegia 76, AR | AR |
| CARS2 | 612800 | Combined oxidative phosphorylation deficiency 27 | AR |
| CASK | 300172 | Mental retardation, with or without nystagmus;Mental retardation and microcephaly with pontine and cerebellar hypoplasia;FG syndrome 4 | XLD |
| CC2D2A | 612013 | Meckel syndrome 6;Joubert syndrome 9;COACH syndrome 2 | AR |
| CCDC88C | 611204 | Hydrocephalus, congenital, 1;?Spinocerebellar ataxia 40 | AR, AD |
| CCT5 | 610150 | Neuropathy, hereditary sensory, with spastic paraplegia | AR |
| CEP290 | 610142 | Leber congenital amaurosis 10;Meckel syndrome 4;?Bardet-Biedl syndrome 14;Senior-Loken syndrome 6;Joubert syndrome 5 | AR |
| CEP41 | 610523 | Joubert syndrome 15 | AR |
| CHMP1A | 164010 | Pontocerebellar hypoplasia, type 8 | AR |
| CLCN2 | 600570 | Epilepsy, juvenile absence, susceptibility to, 2;Hyperaldosteronism, familial, type II;Epilepsy, juvenile myoclonic, susceptibility to, 8;Epilepsy, idiopathic generalized, susceptibility to, 11;Leukoencephalopathy with ataxia | AD, AR |
| CLN5 | 608102 | Ceroid lipofuscinosis, neuronal, 5 | AR |

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| CLN6 | 606725 | Ceroid lipofuscinosis, neuronal, Kufs type, adult onset;Ceroid lipofuscinosis, neuronal, 6 | AR |
| CLPB | 616254 | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia | AR |
| CLPP | 601119 | Perrault syndrome 3 | AR |
| COA6 | 614772 | Mi complex IV deficiency, nuclear type 13 | AR |
| COA7 | 615623 | Spinocerebellar ataxia, AR, with axonal neuropathy 3 | AR |
| COA8 | 616003 | Mi complex IV deficiency, nuclear type 17 | AR |
| COASY | 609855 | Neurodegeneration with brain iron accumulation 6;Pontocerebellar hypoplasia, type 12 | AR |
| COL4A1 | 120130 | ?Retinal arteries, tortuosity of;Hemorrhage, intracerebral, susceptibility to;Microangiopathy and leukoencephalopathy, pontine, AD;Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps;Brain small vessel disease with or without ocular anomalies | AD |
| COL4A2 | 120090 | Hemorrhage, intracerebral, susceptibility to;Brain small vessel disease 2 | AD |
| COQ2 | 609825 | Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to | AR, AD, AR |
| COQ4 | 612898 | Coenzyme Q10 deficiency, primary, 7 | AR |
| COQ6 | 614647 | Coenzyme Q10 deficiency, primary, 6 | AR |
| COQ7 | 601683 | ?Coenzyme Q10 deficiency, primary, 8 | AR |
| COQ8A | 606980 | Coenzyme Q10 deficiency, primary, 4 | AR |

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| COQ8B | 615567 | Nephrotic syndrome, type 9 | AR |
| COQ9 | 612837 | Coenzyme Q10 deficiency, primary, 5 | AR |
| COX10 | 602125 | Mi complex IV deficiency, nuclear type 3 | AR |
| COX15 | 603646 | Mi complex IV deficiency, nuclear type 6 | AR |
| COX20 | 614698 | Mi complex IV deficiency, nuclear type 11 | AR |
| COX6A1 | 602072 | Charcot-Marie-Tooth disease, recessive intermediate D | AR |
| COX6B1 | 124089 | Mi complex IV deficiency, nuclear type 7 | AR |
| CP | 117700 | Cerebellar ataxia;Hemosiderosis, systemic, due to aceruloplasminemia;[Hypoceruloplasminemia, hereditary] | AR |
| CPT1C | 608846 | ?Spastic paraplegia 73, AD | AD |
| CSPP1 | 611654 | Joubert syndrome 21 | AR |
| CSTB | 601145 | Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) | AR |
| CWF19L1 | 616120 | Spinocerebellar ataxia, AR 17 | AR |
| CYC1 | 123980 | Mi complex III deficiency, nuclear type 6 | AR |
| CYP27A1 | 606530 | Cerebrotendinous xanthomatosis | AR |
| CYP2U1 | 610670 | Spastic paraplegia 56, AR | AR |
| CYP7B1 | 603711 | Spastic paraplegia 5A, AR;Bile acid synthesis defect, congenital, 3 | AR |
| DAB1 | 603448 | Spinocerebellar ataxia 37 | AD |

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| DARS1 | 603084 | Hypomyelination with brainstem and spinal cord involvement and leg spasticity | AR |
| DARS2 | 610956 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation | AR |
| DBT | 248610 | Maple syrup urine disease, type II | AR |
| DDHD1 | 614603 | Spastic paraplegia 28, AR | AR |
| DDHD2 | 615003 | Spastic paraplegia 54, AR | AR |
| DGUOK | 601465 | Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with Mi DNA deletions, AR 4;Mi DNA depletion syndrome 3 (hepatocerebral type) | AR |
| DHPS | 600944 | Neurodevelopmental disorder with seizures and speech and walking impairment | AR |
| DLAT | 608770 | Pyruvate dehydrogenase E2 deficiency | AR |
| DLD | 238331 | Dihydrolipoamide dehydrogenase deficiency | AR |
| DNA2 | 601810 | ?Seckel syndrome 8;Progressive external ophthalmoplegia with Mi DNA deletions, AD 6 | AR, AD |
| DNAJC19 | 608977 | 3-methylglutaconic aciduria, type V | AR |
| DNAJC5 | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type | AD |
| DNM1L | 603850 | Optic atrophy 5;Encephalopathy, lethal, due to defective Mi peroxisomal fission 1 | AD, AD, AR |
| DNMT1 | 126375 | Neuropathy, hereditary sensory, type IE;Cerebellar ataxia, deafness, and narcolepsy, AD | AD |
| DOCK3 | 603123 | Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia | AR |
| DSTYK | 612666 | Spastic paraplegia 23;Congenital anomalies of kidney and urinary tract 1 | AR, AD |

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| EARS2 | 612799 | Combined oxidative phosphorylation deficiency 12 | AR |
| EBF3 | 607407 | Hypotonia, ataxia, and delayed development syndrome | AD |
| ECHS1 | 602292 | Mi short-chain enoyl-CoA hydratase 1 deficiency | AR |
| EIF2B1 | 606686 | Leukoencephalopathy with vanishing white matter | AR |
| EIF2B2 | 606454 | Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter | AR |
| EIF2B3 | 606273 | Leukoencephalopathy with vanishing white matter | AR |
| EIF2B4 | 606687 | Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter | AR |
| EIF2B5 | 603945 | Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter | AR |
| ELAC2 | 605367 | Prostate cancer, hereditary, 2, susceptibility to;Combined oxidative phosphorylation deficiency 17 | AR |
| ELOVL4 | 605512 | Ichthyosis, spastic quadriplegia, and mental retardation;Stargardt disease 3;Spinocerebellar ataxia 34 | AR, AD |
| ELOVL5 | 611805 | Spinocerebellar ataxia 38 | AD |
| ENTPD1 | 601752 | Spastic paraplegia 64, AR | AR |
| ERLIN1 | 611604 | Spastic paraplegia 62 | AR |
| ERLIN2 | 611605 | Spastic paraplegia 18, AR | AR |
| ETFA | 608053 | Glutaric acidemia IIA | AR |
| ETFB | 130410 | Glutaric acidemia IIB | AR |
| ETFDH | 231675 | Glutaric acidemia IIC | AR |

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| ETHE1 | 608451 | Ethylmalonic encephalopathy | AR |
| EXOSC3 | 606489 | Pontocerebellar hypoplasia, type 1B | AR |
| FA2H | 611026 | Spastic paraplegia 35, AR | AR |
| FARS2 | 611592 | Combined oxidative phosphorylation deficiency 14;Spastic paraplegia 77, AR | AR |
| FASTKD2 | 612322 | Combined oxidative phosphorylation deficiency 44 | AR |
| FAT2 | 604269 | Spinocerebellar ataxia 45 | AD |
| FBXL4 | 605654 | Mi DNA depletion syndrome 13 (encephalomyopathic type) | AR |
| FDX2 | 614585 | Mi myopathy, episodic, with optic atrophy and reversible leukoencephalopathy | AR |
| FDXR | 103270 | Auditory neuropathy and optic atrophy | AR |
| FGF14 | 601515 | Spinocerebellar ataxia 27 | AD |
| FH | 136850 | Fumarase deficiency;Leiomyomatosis and renal cell cancer | AR, AD |
| FLAD1 | 610595 | Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency | AR |
| FLVCR1 | 609144 | Ataxia, posterior column, with retinitis pigmentosa | AR |
| FOXRED1 | 613622 | Mi complex I deficiency, nuclear type 19 | AR |
| FTL | 134790 | Neurodegeneration with brain iron accumulation 3;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive | AD, AD, AR |
| FXN | 606829 | Friedreich ataxia;Friedreich ataxia with retained reflexes | AR |

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| GAD1 | 605363 | ?Cerebral palsy, spastic quadriplegic, 1;Developmental and epileptic encephalopathy 89 | AR |
| GALC | 606890 | Krabbe disease | AR |
| GARS1 | 600287 | Spinal muscular atrophy, infantile, James type;Charcot-Marie-Tooth disease, type 2D;Neuronopathy, distal hereditary motor, type VA | AD |
| GBA | 606463 | Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I | AD, AR, AD, mi |
| GBA2 | 609471 | Spastic paraplegia 46, AR | AR |
| GCDH | 608801 | Glutaricaciduria, type I | AR |
| GCH1 | 600225 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia;Hyperphenylalanine mia, BH4-deficient, B | AD, AR, AR |
| GFAP | 137780 | Alexander disease | AD |
| GFER | 600924 | Myopathy, Mi progressive, with congenital cataract and developmental delay | AR |
| GFM1 | 606639 | Combined oxidative phosphorylation deficiency 1 | AR |
| GFM2 | 606544 | Combined oxidative phosphorylation deficiency 39 | AR |
| GJB1 | 304040 | Charcot-Marie-Tooth neuropathy, XLD, 1 | XLD |
| GJC2 | 608803 | Leukodystrophy, hypomyelinating, 2;Spastic paraplegia 44, AR;Lymphatic malformation 3 | AR, AD |
| GLRX5 | 609588 | Spasticity, childhood-onset, with hyperglycinemia;Anemia, sideroblastic, 3, pyridoxine-refractory | AR |

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| GOSR2 | 604027 | Epilepsy, progressive myoclonic 6 | AR |
| GRID2 | 602368 | Spinocerebellar ataxia, AR 18 | AR |
| GRM1 | 604473 | Spinocerebellar ataxia 44;Spinocerebellar ataxia, AR 13 | AD, AR |
| GSS | 601002 | Glutathione synthetase deficiency;Hemolytic anemia due to glutathione synthetase deficiency | AR |
| GTPBP3 | 608536 | Combined oxidative phosphorylation deficiency 23 | AR |
| HACE1 | 610876 | Spastic paraplegia and psychomotor retardation with or without seizures | AR |
| HARS2 | 600783 | Perrault syndrome 2 | AR |
| HEPACAM | 611642 | Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation;Megalencephalic leukoencephalopathy with subcortical cysts 2A | AD, AR |
| HEXA | 606869 | Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms | AR |
| HEXB | 606873 | Sandhoff disease, infantile, juvenile, and adult forms | AR |
| HIBCH | 610690 | 3-hydroxyisobutryl-CoA hydrolase deficiency | AR |
| HMGCL | 613898 | HMG-CoA lyase deficiency | AR |
| HSPD1 | 118190 | Leukodystrophy, hypomyelinating, 4;Spastic paraplegia 13, AD | AR, AD |
| HTRA2 | 606441 | Parkinson disease 13;3-methylglutaconic aciduria, type VIII | AR |
| IARS2 | 612801 | ?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia | AR |

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| IBA57 | 615316 | ?Spastic paraplegia 74, AR;Multiple Mi dysfunctions syndrome 3 | AR |
| INPP5E | 613037 | Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1 | AR |
| IRF2BPL | 611720 | Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures | AD |
| ISCA2 | 615317 | Multiple Mi dysfunctions syndrome 4 | AR |
| ISCU | 611911 | Myopathy with lactic acidosis, hereditary | AR |
| ITM2B | 603904 | Dementia, familial British;?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities;Dementia, familial Danish | AD |
| ITPR1 | 147265 | Spinocerebellar ataxia 15;Spinocerebellar ataxia 29, congenital nonprogressive;Gillespie syndrome | AD, AD, AR |
| KCNA1 | 176260 | Episodic ataxia/myokymia syndrome | AD |
| KCNA2 | 176262 | Developmental and epileptic encephalopathy 32 | AD |
| KCNC3 | 176264 | Spinocerebellar ataxia 13 | AD |
| KCND3 | 605411 | Brugada syndrome 9;Spinocerebellar ataxia 19 | AD |
| KCNJ10 | 602208 | Enlarged vestibular aqueduct, digenic;SESAME syndrome | AR |
| KDM5C | 314690 | Mental retardation, XL, syndromic, Claes-Jensen type | XLR |
| KIDINS220 | 615759 | Ventriculomegaly and arthrogryposis;Spastic paraplegia, intellectual disability, nystagmus, and obesity | AR, AD |
| KIF1A | 601255 | NESCAV syndrome;Spastic paraplegia 30, AR;Neuropathy, hereditary sensory, type IIC;Spastic paraplegia 30, AD | AD, AD, AR, AR |
| KIF1C | 603060 | Spastic ataxia 2, AR | AR |

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| KIF5A | 602821 | Amyotrophic lateral sclerosis, susceptibility to, 25;Myoclonus, intractable, neonatal;Spastic paraplegia 10, AD | AD |
| KIF7 | 611254 | Joubert syndrome 12;?Hydrolethalus syndrome 2;?Al-Gazali-Bakalinova syndrome;Acrocallosal syndrome | AR |
| L1CAM | 308840 | Hydrocephalus with congenital idiopathic intestinal pseudoobstruction;CRASH syndrome;Corpus callosum, partial agenesis of;Hydrocephalus with Hirschsprung disease;MASA syndrome;Hydrocephalus due to aqueductal stenosis | XLR |
| LAMA1 | 150320 | Poretti-Boltshauser syndrome | AR |
| LAMP2 | 309060 | Danon disease | XLD |
| LARS2 | 604544 | ?Hydrops, lactic acidosis, and sideroblastic anemia;Perrault syndrome 4 | AR |
| LIAS | 607031 | Hyperglycinemia, lactic acidosis, and seizures | AR |
| LIPT1 | 610284 | Lipoyltransferase 1 deficiency | AR |
| LMNB1 | 150340 | Leukodystrophy, adult-onset, AD;Microcephaly 26, primary, AD | AD |
| LRPPRC | 607544 | Mi complex IV deficiency, nuclear type 5, (French-Canadian) | AR |
| LYRM7 | 615831 | Mi complex III deficiency, nuclear type 8 | AR |
| LYST | 606897 | Chediak-Higashi syndrome | AR |
| MAG | 159460 | Spastic paraplegia 75, AR | AR |
| MARS1 | 156560 | Charcot-Marie-Tooth disease, axonal, type 2U;?Trichothiodystrophy 9, nonphotosensitive;Interstitial lung and liver disease | AD, AR |
| MARS2 | 609728 | Spastic ataxia 3, AR;?Combined oxidative phosphorylation deficiency 25 | AR |

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| MECR | 608205 | Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities | AR |
| MFF | 614785 | Encephalopathy due to defective Mi and peroxisomal fission 2 | AR |
| MFN2 | 608507 | Hereditary motor and sensory neuropathy VIA;Charcot-Marie-Tooth disease, axonal, type 2A2B;Charcot-Marie-Tooth disease, axonal, type 2A2A | AD, AR |
| MGME1 | 615076 | Mi DNA depletion syndrome 11 | AR |
| MICU1 | 605084 | Myopathy with extrapyramidal signs | AR |
| MKKS | 604896 | McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6 | AR |
| MKS1 | 609883 | Bardet-Biedl syndrome 13;Joubert syndrome 28;Meckel syndrome 1 | AR |
| MLC1 | 605908 | Megalencephalic leukoencephalopathy with subcortical cysts | AR |
| MPC1 | 614738 | Mi pyruvate carrier deficiency | AR |
| MPV17 | 137960 | Charcot-Marie-Tooth disease, axonal, type 2EE;Mi DNA depletion syndrome 6 (hepatocerebral type) | AR |
| MRE11 | 600814 | Ataxia-telangiectasia-like disorder 1 | AR |
| MRPL3 | 607118 | Combined oxidative phosphorylation deficiency 9 | AR |
| MRPL44 | 611849 | ?Combined oxidative phosphorylation deficiency 16 | AR |
| MRPS16 | 609204 | Combined oxidative phosphorylation deficiency 2 | AR |
| MRPS22 | 605810 | Ovarian dysgenesis 7;Combined oxidative phosphorylation deficiency 5 | AR |
| MSTO1 | 617619 | Myopathy, Mi, and ataxia | AD, AR |

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| MT-ATP6 | | | |
| MT-ATP8 | | | |
| MT-CO1 | | | |
| MT-CO2 | | | |
| MT-CO3 | | | |
| MT-CYB | | | |
| MT-ND1 | | | |
| MT-ND2 | | | |
| MT-ND3 | | | |
| MT-ND4 | | | |
| MT-ND4L | | | |
| MT-ND5 | | | |
| MT-ND6 | | | |
| MT-RNR1 | | | |
| MT-RNR2 | | | |
| MT-TA | | | |
| MT-TC | | | |

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| MT-TD | | | |
| MT-TE | | | |
| MT-TF | | | |
| MT-TG | | | |
| MT-TH | | | |
| MT-TI | | | |
| MT-TK | | | |
| MT-TL1 | | | |
| MT-TL2 | | | |
| MT-TM | | | |
| MT-TN | | | |
| MT-TP | | | |
| MT-TQ | | | |
| MT-TR | | | |
| MT-TS1 | | | |
| MT-TS2 | | | |
| MT-TT | | | |

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| MT-TV | | | |
| MT-TW | | | |
| MT-TY | | | |
| MTFMT | 611766 | Combined oxidative phosphorylation deficiency 15;Mi complex I deficiency, nuclear type 27 | AR |
| MTO1 | 614667 | Combined oxidative phosphorylation deficiency 10 | AR |
| MTPAP | 613669 | ?Spastic ataxia 4, AR | AR |
| MTRFR | 613541 | Spastic paraplegia 55, AR;Combined oxidative phosphorylation deficiency 7 | AR |
| MTTP | 157147 | Metabolic syndrome, protection against;Abetalipoproteinemia | AD, AR |
| NARS2 | 612803 | ?Deafness, AR 94;Combined oxidative phosphorylation deficiency 24 | AR |
| NDUFA1 | 300078 | Mi complex I deficiency, nuclear type 12 | XLR |
| NDUFA10 | 603835 | Mi complex I deficiency, nuclear type 22 | AR |
| NDUFA11 | 612638 | Mi complex I deficiency, nuclear type 14 | AR |
| NDUFA12 | 614530 | ?Mi complex I deficiency, nuclear type 23 | AR |
| NDUFA2 | 602137 | Mi complex I deficiency, nuclear type 13 | AR |
| NDUFA9 | 603834 | Mi complex I deficiency, nuclear type 26 | AR |
| NDUFAF1 | 606934 | Mi complex I deficiency, nuclear type 11 | AR |
| NDUFAF2 | 609653 | Mi complex I deficiency, nuclear type 10 | AR |

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| NDUFAF3 | 612911 | Mi complex I deficiency, nuclear type 18 | AR |
| NDUFAF4 | 611776 | Mi complex I deficiency, nuclear type 15 | AR |
| NDUFAF5 | 612360 | Mi complex I deficiency, nuclear type 16 | AR |
| NDUFAF6 | 612392 | Mi complex I deficiency, nuclear type 17;Fanconi renotubular syndrome 5 | AR |
| NDUFB3 | 603839 | Mi complex I deficiency, nuclear type 25 | AR |
| NDUFS1 | 157655 | Mi complex I deficiency, nuclear type 5 | AR |
| NDUFS2 | 602985 | Mi complex I deficiency, nuclear type 6 | AR |
| NDUFS3 | 603846 | Mi complex I deficiency, nuclear type 8 | AR |
| NDUFS4 | 602694 | Mi complex I deficiency, nuclear type 1 | AR |
| NDUFS6 | 603848 | Mi complex I deficiency, nuclear type 9 | AR |
| NDUFS7 | 601825 | Mi complex I deficiency, nuclear type 3 | AR |
| NDUFS8 | 602141 | Mi complex I deficiency, nuclear type 2 | AR |
| NDUFV1 | 161015 | Mi complex I deficiency, nuclear type 4 | AR |
| NDUFV2 | 600532 | Mi complex I deficiency, nuclear type 7 | AR |
| NFU1 | 608100 | Multiple Mi dysfunctions syndrome 1 | AR |
| NIPA1 | 608145 | Spastic paraplegia 6, AD | AD |
| NKX6-2 | 605955 | Spastic ataxia 8, AR, with hypomyelinating leukodystrophy | AR |

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| NPC1 | 607623 | Niemann-Pick disease, type C1;Niemann-Pick disease, type D | AR |
| NPC2 | 601015 | Niemann-pick disease, type C2 | AR |
| NPHP1 | 607100 | Joubert syndrome 4;Nephronophthisis 1, juvenile;Senior-Loken syndrome-1 | AR |
| NR2F1 | 132890 | Bosch-Boonstra-Schaaf optic atrophy syndrome | AD |
| NT5C2 | 600417 | Spastic paraplegia 45, AR | AR |
| NUBPL | 613621 | Mi complex I deficiency, nuclear type 21 | AR |
| OFD1 | 300170 | Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I | XLR, XLD |
| OPA1 | 605290 | Optic atrophy 1;?Mi DNA depletion syndrome 14 (encephalocardiomyopathic type);Glaucoma, normal tension, susceptibility to;Optic atrophy plus syndrome;Behr syndrome | AD, AR |
| OPA3 | 606580 | 3-methylglutaconic aciduria, type III;Optic atrophy 3 with cataract | AR, AD |
| OPHN1 | 300127 | Mental retardation, XL, with cerebellar hypoplasia and distinctive facial appearance | XLR |
| OTC | 300461 | Ornithine transcarbamylase deficiency | XL |
| PANK2 | 606157 | HARP syndrome;Neurodegeneration with brain iron accumulation 1 | AR |
| PARS2 | 612036 | Developmental and epileptic encephalopathy 75 | AR |
| PAX6 | 607108 | ?Coloboma, ocular;Aniridia;?Morning glory disc anomaly;Keratitis;Optic nerve hypoplasia;?Coloboma of optic nerve;Anterior segment dysgenesis 5, multiple subtypes;Cataract with late-onset corneal dystrophy;Foveal hypoplasia 1 | AD |

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| PC | 608786 | Pyruvate carboxylase deficiency | AR |
| PCCA | 232000 | Propionicacidemia | AR |
| PCCB | 232050 | Propionicacidemia | AR |
| PDHA1 | 300502 | Pyruvate dehydrogenase E1-alpha deficiency | XLD |
| PDHB | 179060 | Pyruvate dehydrogenase E1-beta deficiency | AR |
| PDHX | 608769 | Lacticacidemia due to PDX1 deficiency | AR |
| PDP1 | 605993 | Pyruvate dehydrogenase phosphatase deficiency | AR |
| PDSS1 | 607429 | Coenzyme Q10 deficiency, primary, 2 | AR |
| PDSS2 | 610564 | Coenzyme Q10 deficiency, primary, 3 | AR |
| PDYN | 131340 | Spinocerebellar ataxia 23 | AD |
| PET100 | 614770 | Mi complex IV deficiency, nuclear type 12 | AR |
| PEX10 | 602859 | Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B | AR |
| PEX2 | 170993 | Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger) | AR |
| PEX7 | 601757 | Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1 | AR |
| PGAP1 | 611655 | Mental retardation, AR 42 | AR |
| PHYH | 602026 | Refsum disease | AR |
| PIK3R5 | 611317 | Ataxia-oculomotor apraxia 3 | AR |

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| PLA2G6 | 603604 | Neurodegeneration with brain iron accumulation 2B;Parkinson disease 14, AR;Infantile neuroaxonal dystrophy 1 | AR |
| PLK1 | 602098 | | |
| PLP1 | 300401 | Pelizaeus-Merzbacher disease;Spastic paraplegia 2, XL | XLR |
| PMPCA | 613036 | Spinocerebellar ataxia, AR 2 | AR |
| PNKD | 609023 | Paroxysmal nonkinesigenic dyskinesia 1 | AD |
| PNKP | 605610 | Ataxia-oculomotor apraxia 4;Microcephaly, seizures, and developmental delay;?Charcot-Marie-Tooth disease, type 2B2 | AR |
| PNPLA6 | 603197 | Spastic paraplegia 39, AR;Boucher-Neuhauser syndrome;Oliver-McFarlane syndrome;?Laurence-Moon syndrome | AR |
| PNPT1 | 610316 | Deafness, AR 70;Combined oxidative phosphorylation deficiency 13 | AR |
| POLG | 174763 | Progressive external ophthalmoplegia, AR 1;Progressive external ophthalmoplegia, AD 1;Mi recessive ataxia syndrome (includes SANDO and SCAE);Mi DNA depletion syndrome 4B (MNGIE type);Mi DNA depletion syndrome 4A (Alpers type) | AR, AD |
| POLG2 | 604983 | Mi DNA depletion syndrome 16 (hepatic type);?Mi DNA depletion syndrome 16B (neuroophthalmic type);Progressive external ophthalmoplegia with Mi DNA deletions, AD 4 | AR, AD |
| POLR3A | 614258 | Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism;Wiedemann-Rautenstrauch syndrome | AR |
| POLR3B | 614366 | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism | AR |
| PRICKLE1 | 608500 | Epilepsy, progressive myoclonic 1B | AR |

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| PRKCG | 176980 | Spinocerebellar ataxia 14 | AD |
| PRRT2 | 614386 | Episodic kinesigenic dyskinesia 1;Convulsions, familial infantile, with paroxysmal choreoathetosis;Seizures, benign familial infantile, 2 | AD |
| PUM1 | 607204 | Spinocerebellar ataxia 47 | AD |
| QARS1 | 603727 | Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy | AR |
| RAB3GAP2 | 609275 | Warburg micro syndrome 2;Martsolf syndrome | AR |
| RAP1GDS1 | 179502 | | |
| RARS1 | 107820 | Leukodystrophy, hypomyelinating, 9 | AR |
| RARS2 | 611524 | Pontocerebellar hypoplasia, type 6 | AR |
| REEP1 | 609139 | Spastic paraplegia 31, AD;?Neuronopathy, distal hereditary motor, type VB | AD |
| REEP2 | 609347 | ?Spastic paraplegia 72, AR;?Spastic paraplegia 72, AD | AD, AR |
| RMND1 | 614917 | Combined oxidative phosphorylation deficiency 11 | AR |
| RNASEH1 | 604123 | Progressive external ophthalmoplegia with Mi DNA deletions, AR 2 | AR |
| RNASEH2B | 610326 | Aicardi-Goutieres syndrome 2 | AR |
| RNF216 | 609948 | Cerebellar ataxia and hypogonadotropic hypogonadism | AR |
| RPGRIP1L | 610937 | Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5 | AR |
| RRM2B | 604712 | Mi DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy);Progressive external ophthalmoplegia with Mi DNA deletions, AD | AR, AD |

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| | | 5;Mi DNA depletion syndrome 8B (MNGIE type) | |
| RTN2 | 603183 | Spastic paraplegia 12, AD | AD |
| RUBCN | 613516 | Spinocerebellar ataxia, AR 15 | AR |
| SACS | 604490 | Spastic ataxia, Charlevoix-Saguenay type | AR |
| SAMD9L | 611170 | Ataxia-pancytopenia syndrome;Monosomy 7 myelodysplasia and leukemia syndrome 1 | AD |
| SARS2 | 612804 | Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis | AR |
| SCN1A | 182389 | Epilepsy, generalized, with febrile seizures plus, type 2;Migraine, familial hemiplegic, 3;Developmental and epileptic encephalopathy 6B, non-Dravet;Febrile seizures, familial, 3A;Dravet syndrome | AD |
| SCN2A | 182390 | Developmental and epileptic encephalopathy 11;Seizures, benign familial infantile, 3;Episodic ataxia, type 9 | AD |
| SCO1 | 603644 | Mi complex IV deficiency, nuclear type 4 | AR |
| SCO2 | 604272 | Myopia 6;Mi complex IV deficiency, nuclear type 2 | AD, AR |
| SCYL1 | 607982 | Spinocerebellar ataxia, AR 21 | AR |
| SDHA | 600857 | Neurodegeneration with ataxia and late-onset optic atrophy;Cardiomyopathy, dilated, 1GG;Leigh syndrome;Mi respiratory chain complex II deficiency;Paragangliomas 5 | AD, AR, AR, Mi |
| SDHAF1 | 612848 | Mi complex II deficiency, nuclear type 2;Mi complex II deficiency | AR |
| SELENOI | 607915 | Spastic paraplegia 81, AR | AR |
| SERAC1 | 614725 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome | AR |

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| SETX | 608465 | Spinocerebellar ataxia, AR, with axonal neuropathy 2;Amyotrophic lateral sclerosis 4, juvenile | AR, AD |
| SFXN4 | 615564 | Combined oxidative phosphorylation deficiency 18 | AR |
| SIL1 | 608005 | Marinesco-Sjogren syndrome | AR |
| SLC16A2 | 300095 | Allan-Herndon-Dudley syndrome | XL |
| SLC17A5 | 604322 | Sialic acid storage disorder, infantile;Salla disease | AR |
| SLC19A2 | 603941 | Thiamine-responsive megaloblastic anemia syndrome | AR |
| SLC19A3 | 606152 | Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2) | AR |
| SLC1A3 | 600111 | Episodic ataxia, type 6 | AD |
| SLC1A4 | 600229 | Spastic tetraplegia, thin corpus callosum, and progressive microcephaly | AR |
| SLC20A2 | 158378 | Basal ganglia calcification, idiopathic, 1 | AD |
| SLC22A5 | 603377 | Carnitine deficiency, systemic primary | AR |
| SLC25A19 | 606521 | Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type);Microcephaly, Amish type | AR |
| SLC25A26 | 611037 | Combined oxidative phosphorylation deficiency 28 | AR |
| SLC25A3 | 600370 | Mi phosphate carrier deficiency | AR |
| SLC25A38 | 610819 | Anemia, sideroblastic, 2, pyridoxine-refractory | AR |
| SLC25A4 | 103220 | Mi DNA depletion syndrome 12B (cardiomyopathic type) AR;Mi DNA depletion syndrome 12A (cardiomyopathic type) | AR, AD |

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| | | AD;Progressive external ophthalmoplegia with Mi DNA deletions, AD 2 | |
| SLC25A46 | 610826 | Neuropathy, hereditary motor and sensory, type VIB;Pontocerebellar hypoplasia, type 1E | AR |
| SLC2A1 | 138140 | GLUT1 deficiency syndrome 2, childhood onset;GLUT1 deficiency syndrome 1, infantile onset, severe;Epilepsy, idiopathic generalized, susceptibility to, 12;Stomatin-deficient cryohydrocytosis with neurologic defects;Dystonia 9 | AD, AD, AR |
| SLC33A1 | 603690 | Congenital cataracts, hearing loss, and neurodegeneration;Spastic paraplegia 42, AD | AR, AD |
| SLC52A2 | 607882 | Brown-Vialetto-Van Laere syndrome 2 | AR |
| SLC52A3 | 613350 | ?Fazio-Londe disease;Brown-Vialetto-Van Laere syndrome 1 | AR |
| SLC9A6 | 300231 | Mental retardation, XL syndromic, Christianson type | XL |
| SNX14 | 616105 | Spinocerebellar ataxia, AR 20 | AR |
| SPART | 607111 | Troyer syndrome | AR |
| SPAST | 604277 | Spastic paraplegia 4, AD | AD |
| SPG11 | 610844 | Spastic paraplegia 11, AR;Amyotrophic lateral sclerosis 5, juvenile;Charcot-Marie-Tooth disease, axonal, type 2X | AR |
| SPG21 | 608181 | Mast syndrome | AR |
| SPG7 | 602783 | Spastic paraplegia 7, AR | AD, AR |
| SPR | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency | ?AD, AR |
| SPTBN2 | 604985 | Spinocerebellar ataxia, AR 14;Spinocerebellar ataxia 5 | AR, AD |

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| STUB1 | 607207 | ?Spinocerebellar ataxia 48;Spinocerebellar ataxia, AR 16 | AD, AR |
| SUCLA2 | 603921 | Mi DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) | AR |
| SUCLG1 | 611224 | Mi DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria) | AR |
| SURF1 | 185620 | Charcot-Marie-Tooth disease, type 4K;Mi complex IV deficiency, nuclear type 1 | AR |
| SYNE1 | 608441 | Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8 | AR, AD |
| TACO1 | 612958 | Mi complex IV deficiency, nuclear type 8 | AR |
| TARS2 | 612805 | ?Combined oxidative phosphorylation deficiency 21 | AR |
| TBC1D24 | 613577 | Deafness, AD 65;Deafness , AR 86;Myoclonic epilepsy, infantile, familial;Developmental and epileptic encephalopathy 16;DOORS syndrome;Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp | AD, AR |
| TCTN1 | 609863 | Joubert syndrome 13 | AR |
| TCTN2 | 613846 | ?Meckel syndrome 8;Joubert syndrome 24 | AR |
| TCTN3 | 613847 | Joubert syndrome 18;Orofaciodigital syndrome IV | AR |
| TDP1 | 607198 | ?Spinocerebellar ataxia, AR, with axonal neuropathy 1 | AR |
| TECPR2 | 615000 | Spastic paraplegia 49, AR | AR |
| TFG | 602498 | Hereditary motor and sensory neuropathy, Okinawa type;?Spastic paraplegia 57, AR | AD, AR |
| TGM6 | 613900 | Spinocerebellar ataxia 35 | AD |

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| TIMM8A | 300356 | Mohr-Tranebjaerg syndrome | XLR |
| TK2 | 188250 | Mi DNA depletion syndrome 2 (myopathic type);?Progressive external ophthalmoplegia with Mi DNA deletions, AR 3 | AR |
| TMEM126B | 615533 | Mi complex I deficiency, nuclear type 29 | AR |
| TMEM138 | 614459 | Joubert syndrome 16 | AR |
| TMEM216 | 613277 | Meckel syndrome 2;Joubert syndrome 2 | AR |
| TMEM231 | 614949 | Joubert syndrome 20;Meckel syndrome 11 | AR |
| TMEM237 | 614423 | Joubert syndrome 14 | AR |
| TMEM240 | 616101 | Spinocerebellar ataxia 21 | AD |
| TMEM67 | 609884 | COACH syndrome 1;?RHYNS syndrome;Meckel syndrome 3;Joubert syndrome 6;Bardet-Biedl syndrome 14, modifier of;Nephronophthisis 11 | AR |
| TMEM70 | 612418 | Mi complex V (ATP synthase) deficiency, nuclear type 2 | AR |
| TPK1 | 606370 | Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type) | AR |
| TPP1 | 607998 | Ceroid lipofuscinosis, neuronal, 2;Spinocerebellar ataxia, AR 7 | AR |
| TRIM32 | 602290 | Muscular dystrophy, limb-girdle, AR 8;?Bardet-Biedl syndrome 11 | AR |
| TRIT1 | 617840 | Combined oxidative phosphorylation deficiency 35 | AR |
| TRMT10C | 615423 | Combined oxidative phosphorylation deficiency 30 | AR |
| TRNT1 | 612907 | Retinitis pigmentosa and erythrocytic microcytosis;Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay | AR |

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| TSEN2 | 608753 | Pontocerebellar hypoplasia type 2B | AR |
| TSEN34 | 608754 | ?Pontocerebellar hypoplasia type 2C | AR |
| TSEN54 | 608755 | Pontocerebellar hypoplasia type 2A;?Pontocerebellar hypoplasia type 5;Pontocerebellar hypoplasia type 4 | AR |
| TSFM | 604723 | Combined oxidative phosphorylation deficiency 3 | AR |
| TTBK2 | 611695 | Spinocerebellar ataxia 11 | AD |
| TTC19 | 613814 | Mi complex III deficiency, nuclear type 2 | AR |
| TTC8 | 608132 | Bardet-Biedl syndrome 8;?Retinitis pigmentosa 51 | AR |
| TTPA | 600415 | Ataxia with isolated vitamin E deficiency | AR |
| TTR | 176300 | [Dystransthyretinemic hyperthyroxinemia];Carpal tunnel syndrome, familial;Amyloidosis, hereditary, transthyretin-related | AD |
| TUBB4A | 602662 | Dystonia 4, torsion, AD;Leukodystrophy, hypomyelinating, 6 | AD |
| TUFM | 602389 | Combined oxidative phosphorylation deficiency 4 | AR |
| TWNK | 606075 | Progressive external ophthalmoplegia with Mi DNA deletions, AD 3;Perrault syndrome 5;Mi DNA depletion syndrome 7 (hepatocerebral type) | AD, AR |
| TYMP | 131222 | Mi DNA depletion syndrome 1 (MNGIE type) | AR |
| UBA5 | 610552 | Developmental and epileptic encephalopathy 44;?Spinocerebellar ataxia, AR 24 | AR |
| UBAP1 | 609787 | Spastic paraplegia 80, AD | AD |
| UBE3A | 601623 | Angelman syndrome | AD |

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| UBTF | 600673 | Neurodegeneration, childhood-onset, with brain atrophy | AD |
| UCHL1 | 191342 | Parkinson disease 5, susceptibility to;Spastic paraplegia 79, AR | AD, AR |
| UNC80 | 612636 | Hypotonia, infantile, with psychomotor retardation and characteristic facies 2 | AR |
| UQCC2 | 614461 | Mi complex III deficiency, nuclear type 7 | AR |
| UQCRB | 191330 | Mi complex III deficiency, nuclear type 3 | AR |
| UQCRC2 | 191329 | Mi complex III deficiency, nuclear type 5 | AR |
| UQCRQ | 612080 | Mi complex III deficiency, nuclear type 4 | AR |
| USP8 | 603158 | Pituitary adenoma 4, ACTH-secreting, somatic | |
| VAMP1 | 185880 | Myasthenic syndrome, congenital, 25;Spastic ataxia 1, AD | AR, AD |
| VARS2 | 612802 | Combined oxidative phosphorylation deficiency 20 | AR |
| VCP | 601023 | Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1;Charcot-Marie-Tooth disease, type 2Y;Frontotemporal dementia and/or amyotrophic lateral sclerosis 6 | AD |
| VLDLR | 192977 | Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1 | AR |
| VPS37A | 609927 | Spastic paraplegia 53, AR | AR |
| VRK1 | 602168 | Pontocerebellar hypoplasia type 1A | AR |
| WASHC5 | 610657 | Spastic paraplegia 8, AD;Ritscher-Schinzel syndrome 1 | AD, AR |
| WDR45 | 300526 | Neurodegeneration with brain iron accumulation 5 | XLD |

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| WDR45B | 609226 | Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures | AR |
| WDR81 | 614218 | Hydrocephalus, congenital, 3, with brain anomalies;Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2 | AR |
| WFS1 | 606201 | ?Cataract 41;Wolfram-like syndrome, AD;Wolfram syndrome 1;Diabetes mellitus, noninsulin-dependent, association with;Deafness, AD 6/14/38 | AD, AR |
| WWOX | 605131 | Esophageal squamous cell carcinoma, somatic;Developmental and epileptic encephalopathy 28;Spinocerebellar ataxia, AR 12 | AR |
| YARS2 | 610957 | Myopathy, lactic acidosis, and sideroblastic anemia 2 | AR |
| ZFYVE26 | 612012 | Spastic paraplegia 15, AR | AR |
| ZFYVE27 | 610243 | Spastic paraplegia 33, AD | AD |
| ZNF423 | 604557 | Joubert syndrome 19;Nephronophthisis 14 | AD, AR |