

## 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.

<b>Nº de genes:</b>	481
<b>Entrega:</b>	25 días
<b>Cobertura:</b>	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
<b>Detalles:</b>	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

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	Protoporphyrin, 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

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

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

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;Developmental and epileptic encephalopathy 42	AD

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

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

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



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

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

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

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;Hyperphenylalaninemia, 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

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

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

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

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



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			

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			

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

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

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

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

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

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



		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

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

		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

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

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

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

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

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