PANEL DE EPILEPSIA

Si bien algunos tipos de convulsiones se clasifican fácilmente (es decir, parciales o generalizadas), otros no lo son o pueden convertirse más tarde en diferentes tipos (es decir, convulsiones parciales con generalización secundaria), lo que hace que las pruebas de panel específicas tengan menos probabilidades de lograr un diagnóstico. Nuestro **panel de epilepsia** es un panel dirigido por fenotipo que cubre diferentes tipos de síndromes convulsivos, que abarcan el síndrome de Dravet, la encefalopatía epiléptica infantil temprana, la epilepsia parcial, la epilepsia generalizada, la ausencia de epilepsia, el panel de epilepsia mioclónica y la hipomagnesemia. Este panel no incluye genes mitocondriales (es decir, genes que causan epilepsia mioclónica con fibras rojas irregulares -MERRF-).

Si la sospecha clínica se orienta hacia trastornos metabólicos o mitocondriales, solicite el panel Mito integral.

Nº de genes:	784
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis CNV incluido Análisis de Expansión de repetición: CSTB

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Síndrome de Aicardi-Goutieres

Síndromes de acumulación cerebral de hierro

Enfermedad de glicosilación congénita

Síndrome de Dravet

Encefalopatía epiléptica infantil temprana

Epilepsia

Epilepsia (ausencia) en la infancia

Epilepsia (generalizada) con convulsiones febriles

Epilepsia (parcial)

Encefalopatía epiléptica

Hipomagnesemia

síndrome de Leigh

Leucodistrofia y trastornos de la biogénesis de peroxisomas

Enfermedad de almacenamiento lisosomal

Agotamiento del ADN mitocondrial

Encefalomiopatía mitocondrial

Epilepsia mioclónica

Trastorno del ciclo de la urea

Gene	OMIM (Gene)	Associated diseases (OMIM)	Inheritance
AARS1	601065	Trichothiodystrophy 8, nonphotosensitive;Charcot-Marie-Tooth disease, axonal, type 2N;Developmental and epileptic encephalopathy 29;?Leukoencephalopathy, hereditary diffuse, with spheroids 2	AR, AD
AARS2	612035	Combined oxidative phosphorylation deficiency 8;Leukoencephalopathy, progressive, with ovarian failure	AR
ABAT	137150	GABA-transaminase deficiency	AR
ABCC8	600509	Diabetes mellitus, transient neonatal 2;Diabetes mellitus, noninsulin-dependent;Hyperinsulinemic hypoglycemia, familial, 1;Hypoglycemia of infancy, leucine-sensitive;Diabetes mellitus, permanent neonatal 3, with or without neurologic features	AD, AD, AR
ABCD1	300371	Adrenoleukodystrophy;Adrenomyeloneuropathy, adult	XLR
ABCD3	170995	?Bile acid synthesis defect, congenital, 5	AR
ACAD9	611103	Mi complex I deficiency, nuclear type 20	AR
ACADM	607008	Acyl-CoA dehydrogenase, medium chain, deficiency of	AR
ACADS	606885	Acyl-CoA dehydrogenase, short-chain, deficiency of	AR
ACADVL	609575	VLCAD deficiency	AR
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency;Mitchell syndrome	AR, AD
ACTL6B	612458	Developmental and epileptic encephalopathy 76;Intellectual developmental disorder with severe speech and ambulation defects	AR, AD
ACY1	104620	Aminoacylase 1 deficiency	AR
ADA	608958	Adenosine deaminase deficiency, partial;Severe combined immunodeficiency due to ADA deficiency	AR, SM
ADAM22	603709	?Developmental and epileptic encephalopathy 61	AR
ADAMTS10	608990	Weill-Marchesani syndrome 1, recessive	AR
ADAMTSL2	612277	Geleophysic dysplasia 1	AR
ADAR	146920	Aicardi-Goutieres syndrome 6;Dyschromatosis symmetrica hereditaria	AR, AD
ADGRG1	604110	Polymicrogyria, bilateral perisylvian;Polymicrogyria, bilateral frontoparietal	AR
ADSL	608222	Adenylosuccinase deficiency	AR
AFF3	601464	KINSSHIP syndrome	AD
AFG3L2	604581	Spinocerebellar ataxia 28;Spastic ataxia 5, AR;Optic atrophy 12	AD, AR

AGA	613228	Aspartylglucosaminuria	AR
AGK	610345	Sengers syndrome;Cataract 38, AR	AR
AGPS	603051	Rhizomelic chondrodysplasia punctata, type 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
AIMP2	600859	Leukodystrophy, hypomyelinating, 17	AR
ALDH3A2	609523	Sjogren-Larsson syndrome	AR
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency	AR
ALDH7A1	107323	Epilepsy, pyridoxine-dependent	AR
ALDOB	612724	Fructose intolerance, hereditary	AR
ALG1	605907	Congenital disorder of glycosylation, type Ik	AR
ALG11	613666	Congenital disorder of glycosylation, type Ip	AR
ALG12	607144	Congenital disorder of glycosylation, type Ig	AR
ALG13	300776	?Congenital disorder of glycosylation, type Is;Developmental and epileptic encephalopathy 36	XL
ALG2	607905	?Congenital disorder of glycosylation, type Ii;Myasthenic syndrome, congenital, 14, with tubular aggregates	AR
ALG3	608750	Congenital disorder of glycosylation, type Id	AR
ALG6	604566	Congenital disorder of glycosylation, type Ic	AR
ALG8	608103	Polycystic liver disease 3 with or without kidney cysts;Congenital disorder of glycosylation, type Ih	AD, AR
ALG9	606941	Congenital disorder of glycosylation, type Il;Gillessen-Kaesbach-Nishimura syndrome	AR
ALPL	171760	Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult	AR, AD, AR
AMPD2	102771	?Spastic paraplegia 63;Pontocerebellar hypoplasia, type 9	AR
AMT	238310	Glycine encephalopathy	AR
ANK3	600465	Mental retardation, AR, 37	AR
ANTXR2	608041	Hyaline fibromatosis syndrome	AR
AP2M1	601024	Intellectual developmental disorder 60 with seizures	AD
AP3B1	603401	Hermansky-Pudlak syndrome 2	AR
AP3B2	602166	Developmental and epileptic encephalopathy 48	AR
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

APP	104760	Alzheimer disease 1, familial;Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants	AD
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	AR
ARG1	608313	Argininemia	AR
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
ARHGEF9	300429	Developmental and epileptic encephalopathy 8	XL
ARSA	607574	Metachromatic leukodystrophy	AR
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR
ARV1	611647	Developmental and epileptic encephalopathy 38	AR
ARX	300382	Developmental and epileptic encephalopathy 1;Mental retardation, XL 29 and others;Hydranencephaly with abnormal genitalia;Partington syndrome;Lissencephaly, XL 2;Proud syndrome	XLR, XL
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy;Farber lipogranulomatosis	AR
ASL	608310	Argininosuccinic aciduria	AR
ASNS	108370	Asparagine synthetase deficiency	AR
ASPA	608034	Canavan disease	AR
ASS1	603470	Citrullinemia	AR
ASXL1	612990	Bohring-Opitz syndrome;Myelodysplastic syndrome, somatic	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
ATP6AP1	300197	Immunodeficiency 47	XLR
ATP6V0A2	611716	Cutis laxa, AR, type IIA;Wrinkly skin syndrome	AR
ATP6V1A	607027	Epileptic encephalopathy, infantile or early childhood, 3;Cutis laxa, AR, type IID	AD, AR
ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
ATP7B	606882	Wilson disease	AR
ATPAF2	608918	?Mi complex V (ATP synthase) deficiency, nuclear type 1	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

B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR
B3GLCT	610308	Peters-plus syndrome	AR
B4GALT1	137060	Congenital disorder of glycosylation, type IId	AR
BCAP31	300398	Deafness, dystonia, and cerebral hypomyelination	XLR
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
BEST1	607854	Macular dystrophy, vitelliform, 2;Retinitis pigmentosa-50;?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2;Vitreoretinochoroidopathy;Bestrophinopathy, AR;Retinitis pigmentosa, concentric	AD
BOLA3	613183	Multiple Mi dysfunctions syndrome 2 with hyperglycinemia	AR
BRAT1	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures;Rigidity and multifocal seizure syndrome, lethal neonatal	AR
BTD	609019	Biotinidase deficiency	AR
C12orf57	615140	Temtamy syndrome	AR
C19orf12	614297	Neurodegeneration with brain iron accumulation 4;?Spastic paraplegia 43, AR	AD, AR, AR
CA5A	114761	Hyperammonemia due to carbonic anhydrase VA deficiency	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
CACNA1E	601013	Developmental and epileptic encephalopathy 69	AD
CACNA1H	607904	Hyperaldosteronism, familial, type IV;Epilepsy, childhood absence, susceptibility to, 6;Epilepsy, idiopathic generalized, susceptibility to, 6	AD
CACNA2D2	607082	Cerebellar atrophy with seizures and variable developmental delay	AR
CACNB4	601949	Epilepsy, juvenile myoclonic, susceptibility to, 6;Epilepsy, idiopathic generalized, susceptibility to, 9;Episodic ataxia, type 5	AD
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
CAV1	601047	Pulmonary hypertension, primary, 3;Lipodystrophy, familial partial, type 7;?Lipodystrophy, congenital generalized, type 3	AD, AR

CBS	613381	Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types	AR
CCDC115	613734	Congenital disorder of glycosylation, type IIo	AR
CCDC88A	609736	?PEHO syndrome-like	AR
CDKL5	300203	Developmental and epileptic encephalopathy 2	XLD
CERS1	606919	?Epilepsy, progressive myoclonic, 8	AR
CHD2	602119	Epileptic encephalopathy, childhood-onset	AD
CHMP2B	609512	Frontotemporal dementia and/or amytrophic lateral sclerosis 7	AD
CHRNA2	118502	Epilepsy, nocturnal frontal lobe, type 4	AD
CHRNA4	118504	Nicotine addiction, susceptibility to;Epilepsy, nocturnal frontal lobe, 1	AD
CHRNA7	118511		
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3	
CIC	612082	Mental retardation, AD 45	AD
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
CLCN4	302910	Raynaud-Claes syndrome	XLD
CLDN16	603959	Hypomagnesemia 3, renal	AR
CLDN19	610036	Hypomagnesemia 5, renal, with ocular involvement	AR
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3	AR
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5	AR
CLN6	606725	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset;Ceroid lipofuscinosis, neuronal, 6	AR
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8;Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR
CLP1	608757	Pontocerebellar hypoplasia, type 10	AR
CLPP	601119	Perrault syndrome 3	AR
CLTC	118955	Mental retardation, AD 56	AD
CNNM2	607803	Hypomagnesemia 6, renal;Hypomagnesemia, seizures, and mental retardation	AD, AD, AR
CNPY3	610774	Developmental and epileptic encephalopathy 60	AR
CNTNAP1	602346	Lethal congenital contracture syndrome 7;Hypomyelinating neuropathy, congenital, 3	AR
CNTNAP2	604569	Cortical dysplasia-focal epilepsy syndrome;Pitt-Hopkins like syndrome 1;Autism susceptibility 15	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
COG1	606973	Congenital disorder of glycosylation, type IIg	AR

COG4	606976	Congenital disorder of glycosylation, type IIj;Saul-Wilson syndrome	AR, AD
COG5	606821	Congenital disorder of glycosylation, type IIi	AR
COG6	606977	Shaheen syndrome;Congenital disorder of glycosylation, type III	AR
COG7	606978	Congenital disorder of glycosylation, type IIe	AR
COG8	606979	Congenital disorder of glycosylation, type IIh	
COL11A2	120290	Deafness, AR 53;Otospondylomegaepiphyseal dysplasia, AR;Fibrochondrogenesis 2;Otospondylomegaepiphyseal dysplasia, AD;Deafness, AD 13	AR, AD, AR, AD
COL18A1	120328	Glaucoma, primary closed-angle;Knobloch syndrome, type 1	AD, AR
COL2A1	120140	Legg-Calve-Perthes disease;Stickler syndrome, type I;Osteoarthritis with mild chondrodysplasia;Platyspondylic skeletal dysplasia, Torrance type;Spondyloepiphyseal dysplasia, Stanescu type;Kniest dysplasia;Czech dysplasia;Stickler sydrome, type I, nonsyndromic ocular;?Vitreoretinopathy with phalangeal epiphyseal dysplasia;?Epiphyseal dysplasia, multiple, with myopia and deafness;Avascular necrosis of the femoral head;Spondyloperipheral dysplasia;Achondrogenesis, type II or hypochondrogenesis;SMED Strudwick type;SED congenita	AD
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
COLGALT1	617531	Brain small vessel disease 3	AR
COQ2	609825	Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to	AR, AD, AR
COQ8A	606980	Coenzyme Q10 deficiency, primary, 4	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
COX6B1	124089	Mi complex IV deficiency, nuclear type 7	AR
CP	117700	Cerebellar ataxia;Hemosiderosis, systemic, due to aceruloplasminemia;[Hypoceruloplasminemia, hereditary]	AR

CPA6	609562	Febrile seizures, familial, 11;Epilepsy, familial temporal lobe, 5	AR, AD, AR
CPLX1	605032	Developmental and epileptic encephalopathy 63	AR
CPS1	608307	Carbamoylphosphate synthetase I deficiency;Pulmonary hypertension, neonatal, susceptibility to	AR
CPT1A	600528	CPT deficiency, hepatic, type IA	AR
CPT2	600650	CPT II deficiency, myopathic, stress-induced;CPT II deficiency, infantile;Encephalopathy, acute, infection-induced, 4, susceptibility to;CPT II deficiency, lethal neonatal	AD, AR, AR
CSF1R	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis;Leukoencephalopathy, diffuse hereditary, with spheroids	AR, AD
CSNK2B	115441	Poirier-Bienvenu neurodevelopmental syndrome	AD
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CTNNA2	114025	Cortical dysplasia, complex, with other brain malformations 9	AR
CTNS	606272	Cystinosis, late-onset juvenile or adolescent nephropathic;Cystinosis, ocular nonnephropathic;Cystinosis, nephropathic;Cystinosis, atypical nephropathic	AR
CTSA	613111	Galactosialidosis	AR
CTSC	602365	Periodontitis 1, juvenile;Haim-Munk syndrome;Papillon-Lefevre syndrome	AR
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10	AR
CTSF	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type	AR
CTSK	601105	Pycnodysostosis	AR
CYFIP2	606323	Developmental and epileptic encephalopathy 65	AD
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
D2HGDH	609186	D-2-hydroxyglutaric aciduria	AR
DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	AR
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

DCAF17	612515	Woodhouse-Sakati syndrome	AR
DCX	300121	Lissencephaly, XL;Subcortical laminal heterotopia, XL	XL
DEAF1	602635	Vulto-van Silfout-de Vries syndrome;Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures	AD, AR
DEGS1	615843	Leukodystrophy, hypomyelinating, 18	AR
DENND5A	617278	Developmental and epileptic encephalopathy 49	AR
DEPDC5	614191	Epilepsy, familial focal, with variable foci 1	AD
DGUOK	601465	Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with Mi DNA deletions, AR 4;Mi DNA depletion syndrome 3 (hepatocerebral type)	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DHDDS	608172	Developmental delay and seizures with or without movement abnormalities;Retinitis pigmentosa 59;?Congenital disorder of glycosylation, type 1bb	AD, AR
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency	AR
DHX30	616423	Neurodevelopmental disorder with severe motor impairment and absent language	AD
DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome;Deafness, AD 1, with or without thrombocytopenia	AR, AD
DKC1	300126	Dyskeratosis congenita, XL	XLR
DLAT	608770	Pyruvate dehydrogenase E2 deficiency	AR
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	AR
DLL3	602768	Spondylocostal dysostosis 1, AR	AR
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type	AD
DNM1	602377	Developmental and epileptic encephalopathy 31	AD
DNM1L	603850	Optic atrophy 5;Encephalopathy, lethal, due to defective Mi peroxisomal fission 1	AD, AD, AR
DOCK6	614194	Adams-Oliver syndrome 2	AR
DOCK7	615730	Developmental and epileptic encephalopathy 23	AR
DOLK	610746	Congenital disorder of glycosylation, type Im	AR
DPAGT1	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates;Congenital disorder of glycosylation, type Ij	AR
DPM1	603503	Congenital disorder of glycosylation, type Ie	AR
DPM2	603564	Congenital disorder of glycosylation, type Iu	AR
DPM3	605951	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 15;?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15	AR
DPYD	612779	5-fluorouracil toxicity;Dihydropyrimidine dehydrogenase deficiency	AR

DPYS	613326	Dihydropyrimidinuria	AR
DYM	607461	Dyggve-Melchior-Clausen disease;Smith-McCort dysplasia	AR
DYRK1A	600855	Mental retardation, AD 7	AD
EARS2	612799	Combined oxidative phosphorylation deficiency 12	AR
ECHS1	602292	Mi short-chain enoyl-CoA hydratase 1 deficiency	AR
EEF1A2	602959	Mental retardation, AD 38;Developmental and epileptic encephalopathy 33	AD
EFHC1	608815	Epilepsy, juvenile absence, susceptibility to, 1;Myoclonic epilepsy, juvenile, susceptibility to, 1	AD
EGF	131530	Hypomagnesemia 4, renal	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
EIF3F	603914	Mental retardation, AR 67	AR
EMC10	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures	AR
EML1	602033	Band heterotopia	AR
EPG5	615068	Vici syndrome	AR
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora)	AR
EPRS1	138295	Leukodystrophy, hypomyelinating, 15	AR
ERCC6	609413	Lung cancer, susceptibility to;UV-sensitive syndrome 1;Premature ovarian failure 11;Macular degeneration, age-related, susceptibility to, 5;Cockayne syndrome, type B;De Sanctis-Cacchione syndrome;Cerebrooculofacioskeletal syndrome 1	AD, SM, AR, AD
ERCC8	609412	Cockayne syndrome, type A;UV-sensitive syndrome 2	AR
ETFA	608053	Glutaric acidemia IIA	AR
ETFB	130410	Glutaric acidemia IIB	AR
ETFDH	231675	Glutaric acidemia IIC	AR
ETHE1	608451	Ethylmalonic encephalopathy	AR
F2	176930	Dysprothrombinemia;Stroke, ischemic, susceptibility to;Pregnancy loss, recurrent, susceptibility to, 2;Hypoprothrombinemia;Thrombophilia due to thrombin defect	AR, mi, AD

F5	612309	Pregnancy loss, recurrent, susceptibility to, 1;Factor V deficiency;Stroke, ischemic, susceptibility to;Budd-Chiari syndrome;Thrombophilia, susceptibility to, due to factor V Leiden;Thrombophilia due to activated protein C resistance	AD, AR, mi
FA2H	611026	Spastic paraplegia 35, AR	AR
FAH	613871	Tyrosinemia, type I	AR
FAM126A	610531	Leukodystrophy, hypomyelinating, 5	AR
FARS2	611592	Combined oxidative phosphorylation deficiency 14;Spastic paraplegia 77, AR	AR
FARSB	609690	Rajab interstitial lung disease with brain calcifications 1	AR
FASTKD2	612322	Combined oxidative phosphorylation deficiency 44	AR
FBXL4	605654	Mi DNA depletion syndrome 13 (encephalomyopathic type)	AR
FDX2	614585	Mi myopathy, episodic, with optic atrophy and reversible leukoencephalopathy	AR
FGF12	601513	Developmental and epileptic encephalopathy 47	AD
FH	136850	Fumarase deficiency;Leiomyomatosis and renal cell cancer	AR, AD
FHL1	300163	Reducing body myopathy, XL 1b, with late childhood or adult onset;Scapuloperoneal myopathy, XLD;?Uruguay faciocardiomusculoskeletal syndrome;Myopathy, XL, with postural muscle atrophy;Reducing body myopathy, XL 1a, severe, infantile or early childhood onset;Emery-Dreifuss muscular dystrophy 6, XL	XL, XLD, XLR
FIG4	609390	Amyotrophic lateral sclerosis 11;Yunis-Varon syndrome;Charcot-Marie-Tooth disease, type 4J;?Polymicrogyria, bilateral temporooccipital	AD, AR
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR
FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	AR
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency	AR
FOXG1	164874	Rett syndrome, congenital variant	AD
FOXRED1	613622	Mi complex I deficiency, nuclear type 19	AR
FRRS1L	604574	Developmental and epileptic encephalopathy 37	AR
FTL	134790	Neurodegeneration with brain iron accumulation 3;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive	AD, AD, AR
FUCA1	612280	Fucosidosis	AR

FUT8	602589	Congenital disorder of glycosylation with defective fucosylation 1	AR
FXYD2	601814	Hypomagnesemia 2, renal	AD
GAA	606800	Glycogen storage disease II	AR
GABBR2	607340	Nicotine dependence, susceptibility to;Neurodevelopmental disorder with poor language and loss of hand skills;Developmental and epileptic encephalopathy 59;Nicotine dependence, protection against	AD
GABRA1	137160	Developmental and epileptic encephalopathy 19;Epilepsy, childhood absence, susceptibility to, 4;Epilepsy, juvenile myoclonic, susceptibility to, 5	AD
GABRA2	137140	Alcohol dependence, susceptibility to;Developmental and epileptic encephalopathy 78	mi, AD
GABRB1	137190	Developmental and epileptic encephalopathy 45	AD
GABRB2	600232	Epileptic encephalopathy, infantile or early childhood, 2	AD
GABRB3	137192	Epilepsy, childhood absence, susceptibility to, 5;Developmental and epileptic encephalopathy 43	AD
GABRD	137163	Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to;Epilepsy, juvenile myoclonic, susceptibility to;Epilepsy, idiopathic generalized, 10	AD
GABRG2	137164	Developmental and epileptic encephalopathy 74;Epilepsy, generalized, with febrile seizures plus, type 3;Febrile seizures, familial, 8	AD
GAD1	605363	?Cerebral palsy, spastic quadriplegic, 1;Developmental and epileptic encephalopathy 89	AR
GALC	606890	Krabbe disease	AR
GALNS	612222	Mucopolysaccharidosis IVA	AR
GALT	606999	Galactosemia	AR
GAMT	601240	Cerebral creatine deficiency syndrome 2	AR
GAN	605379	Giant axonal neuropathy-1	AR
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
GBE1	607839	Glycogen storage disease IV;Polyglucosan body disease, adult form	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
GFPT1	138292	Myasthenia, congenital, 12, with tubular aggregates	AR
GJA1	121014	Oculodentodigital dysplasia, AR;Atrioventricular septal defect 3;Syndactyly, type III;Craniometaphyseal dysplasia, AR;Palmoplantar keratoderma with congenital alopecia;Oculodentodigital dysplasia;Hypoplastic left heart syndrome 1;Erythrokeratodermia variabilis et progressiva 3	AR, AD
GJB1	304040	Charcot-Marie-Tooth neuropathy, XLD, 1	XLD
GJC2	608803	Leukodystrophy, hypomyelinating, 2;Spastic paraplegia 44, AR;Lymphatic malformation 3	AR, AD
GLA	300644	Fabry disease;Fabry disease, cardiac variant	XL
GLB1	611458	GM1-gangliosidosis, type II;GM1-gangliosidosis, type I;Mucopolysaccharidosis type IVB (Morquio);GM1-gangliosidosis, type III	AR
GLDC	238300	Glycine encephalopathy	AR
GLUD1	138130	Hyperinsulinism-hyperammonemia syndrome	AD
GLUL	138290	Glutamine deficiency, congenital	AR
GM2A	613109	GM2-gangliosidosis, AB variant	AR
GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome	AR
GNAO1	139311	Developmental and epileptic encephalopathy 17;Neurodevelopmental disorder with involuntary movements	AD
GNAQ	600998	Capillary malformations, congenital, 1, somatic, mosaic;Sturge-Weber syndrome, somatic, mosaic	
GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia;Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	AR
GNE	603824	Nonaka myopathy;Sialuria	AR, AD
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	607840	Mucolipidosis III alpha/beta;Mucolipidosis II alpha/beta	AR
GNPTG	607838	Mucolipidosis III gamma	AR
GNS	607664	Mucopolysaccharidosis type IIID	AR
GOSR2	604027	Epilepsy, progressive myoclonic 6	AR
GOT2	138150	Epileptic encephalopathy, early infantile, 82	AR
GPAA1	603048	Glycosylphosphatidylinositol biosynthesis defect 15	AR
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GRIA2	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities	AD

GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, AR;Neurodevelopmental disorder with or without hyperkinetic movements and seizures, AD	AR, AD
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without mental retardation	AD
GRIN2B	138252	Mental retardation, AD 6;Developmental and epileptic encephalopathy 27	AD
GRIN2D	602717	Developmental and epileptic encephalopathy 46	AD
GRN	138945	Ceroid lipofuscinosis, neuronal, 11;Frontotemporal lobar degeneration with ubiquitin-positive inclusions;Aphasia, primary progressive	AR, AD
GTPBP2	607434	Jaberi-Elahi syndrome	AR
GTPBP3	608536	Combined oxidative phosphorylation deficiency 23	AR
GUF1	617064	?Developmental and epileptic encephalopathy 40	AR
GUSB	611499	Mucopolysaccharidosis VII	AR
HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures	AR
HADHA	600890	HELLP syndrome, maternal, of pregnancy;LCHAD deficiency;Fatty liver, acute, of pregnancy;Mi trifunctional protein deficiency	AR
HADHB	143450	Trifunctional protein deficiency	AR
HCFC1	300019	Mental retardation, XL 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XLR
HCN1	602780	Generalized epilepsy with febrile seizures plus, type 10;Developmental and epileptic encephalopathy 24	AD
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
HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C);Retinitis pigmentosa 73	AR
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency	AR
HIKESHI	614908	Leukodystrophy, hypomyelinating, 13	AR
HLCS	609018	Holocarboxylase synthetase deficiency	AR
HMGCL	613898	HMG-CoA lyase deficiency	AR
HMGCS2	600234	HMG-CoA synthase-2 deficiency	AR
HNRNPR	607201		
HNRNPU	602869	Developmental and epileptic encephalopathy 54	AD

HRAS	190020	Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles	AD
HSD17B4	601860	Perrault syndrome 1;D-bifunctional protein deficiency	AR
HSPD1	118190	Leukodystrophy, hypomyelinating, 4;Spastic paraplegia 13, AD	AR, AD
HTRA1	602194	Macular degeneration, age-related, 7;CARASIL syndrome;Cerebral arteriopathy, AD, with subcortical infarcts and leukoencephalopathy, type 2;Macular degeneration, age-related, neovascular type	AR, AD
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
IDS	300823	Mucopolysaccharidosis II	XLR
IDUA	252800	Mucopolysaccharidosis Is;Mucopolysaccharidosis Ih/s;Mucopolysaccharidosis Ih	AR
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome	AR
IFIH1	606951	Aicardi-Goutieres syndrome 7;Singleton-Merten syndrome 1	AD
IQSEC2	300522	Mental retardation, XL 1/78	XLD
ISCA2	615317	Multiple Mi dysfunctions syndrome 4	AR
ITPA	147520	Developmental and epileptic encephalopathy 35;[Inosine triphosphatase deficiency]	AR
IVD	607036	Isovaleric acidemia	AR
JAG1	601920	Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot	AD
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	AR
KAT8	609912	Li-Ghorgani-Weisz-Hubshman syndrome	AD
KCNA1	176260	Episodic ataxia/myokymia syndrome	AD
KCNA2	176262	Developmental and epileptic encephalopathy 32	AD
KCNB1	600397	Developmental and epileptic encephalopathy 26	AD
KCNC1	176258	Epilepsy, progressive myoclonic 7	AD
KCNH1	603305	Zimmermann-Laband syndrome 1;Temple-Baraitser syndrome	AD
KCNJ10	602208	Enlarged vestibular aqueduct, digenic;SESAME syndrome	AR

KCNK4	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome	AD
KCNMA1	600150	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy;Epilepsy, idiopathic generalized, susceptibility to, 16;Liang-Wang syndrome;Cerebellar atrophy, developmental delay, and seizures	AD, AR
KCNQ2	602235	Seizures, benign neonatal, 1;Developmental and epileptic encephalopathy 7;Myokymia	AD
KCNQ3	602232	Seizures, benign neonatal, 2	AD
KCNT1	608167	Developmental and epileptic encephalopathy 14;Epilepsy nocturnal frontal lobe, 5	AD
KCNT2	610044	?Developmental and epileptic encephalopathy 57	AD
KCTD3	613272		
KCTD7	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	AR
KIAA1109	611565	Alkuraya-Kucinskas syndrome	AR
KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3	AD
KIF5A	602821	Amyotrophic lateral sclerosis, susceptibility to, 25;Myoclonus, intractable, neonatal;Spastic paraplegia 10, AD	AD
KMT2E	608444	O'Donnell-Luria-Rodan syndrome	AD
L2HGDH	609584	L-2-hydroxyglutaric aciduria	AR
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23	AR
LAMB1	150240	Lissencephaly 5	AR
LAMP2	309060	Danon disease	XLD
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR
LAT	602354	Immunodeficiency 52	AR
LDB3	605906	Cardiomyopathy, hypertrophic, 24;Myopathy, myofibrillar, 4;Cardiomyopathy, dilated, 1C, with or without LVNC;Left ventricular noncompaction 3	AD
LGI1	604619	Epilepsy, familial temporal lobe, 1	AD
LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures	AR
LIPA	613497	Wolman disease;Cholesteryl ester storage disease	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
MAF	177075	Ayme-Gripp syndrome;Cataract 21, multiple types	AD
MAGT1	300715	Congenital disorder of glycosylation, type Icc;Immunodeficiency, XL, with magnesium defect, Epstein-Barr virus infection and neoplasia	XLR
MAN1B1	604346	Rafiq syndrome	AR
MAN2B1	609458	Mannosidosis, alpha-, types I and II	AR
MANBA	609489	Mannosidosis, beta	AR
MAP2K1	176872	Cardiofaciocutaneous syndrome 3;Melorheostosis, isolated, somatic mosaic	AD
MAP2K2	601263	Cardiofaciocutaneous syndrome 4	AD
MARS2	609728	Spastic ataxia 3, AR;?Combined oxidative phosphorylation deficiency 25	AR
MBD5	611472	Mental retardation, AD 1	AD
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR
MCOLN1	605248	Mucolipidosis IV	AR
MDH2	154100	Developmental and epileptic encephalopathy 51	AR
MECP2	300005	Rett syndrome, preserved speech variant;Encephalopathy, neonatal severe;Mental retardation, XL, syndromic 13;Rett syndrome;Mental retardation, XL syndromic, Lubs type;Rett syndrome, atypical;Autism susceptibility, XL 3	XLD, XLR, XL
MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	AR
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR
MEF2C	600662	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations;Chromosome 5q14.3 deletion syndrome	AD
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
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7;Macular dystrophy with central cone involvement	AR
MGAT2	602616	Congenital disorder of glycosylation, type IIa	AR
MGME1	615076	Mi DNA depletion syndrome 11	AR
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MLPH	606526	Griscelli syndrome, type 3	AR

MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	AR
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	AR
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	AR
MMADHC	611935	Methylmalonic aciduria and homocystinuria, cblD type;Methylmalonic aciduria, cblD type, variant 2;Homocystinuria, cblD type, variant 1	AR
MMUT	609058	Methylmalonic aciduria, mut(0) type	AR
MOCS1	603707	Molybdenum cofactor deficiency A	AR
MOCS2	603708	Molybdenum cofactor deficiency B	AR
MOGS	601336	Congenital disorder of glycosylation, type IIb	AR
MPDU1	604041	Congenital disorder of glycosylation, type If	AR
MPI	154550	Congenital disorder of glycosylation, type Ib	AR
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE;Mi DNA depletion syndrome 6 (hepatocerebral type)	AR
MRPS22	605810	Ovarian dysgenesis 7;Combined oxidative phosphorylation deficiency 5	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
MTHFR	607093	Neural tube defects, susceptibility to;Homocystinuria due to MTHFR deficiency;Schizophrenia, susceptibility to;Thromboembolism, susceptibility to	AR, AD
MTHFS	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination	AR
MTOR	601231	Focal cortical dysplasia, type II, somatic;Smith-Kingsmore syndrome	AD
MTR	156570	Neural tube defects, folate-sensitive, susceptibility to;Homocystinuria-megaloblastic anemia, cblG complementation type	AR
MTRFR	613541	Spastic paraplegia 55, AR;Combined oxidative phosphorylation deficiency 7	AR
MYO5A	160777	Griscelli syndrome, type 1	AR
MYOT	604103	Myopathy, myofibrillar, 3;Myopathy, spheroid body	AD
NAGA	104170	Schindler disease, type III;Kanzaki disease;Schindler disease, type I	AR
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V;Mucopolysaccharidosis type IIIB (Sanfilippo B)	AD, AR
NAGS	608300	N-acetylglutamate synthase deficiency	AR
NARS2	612803	?Deafness, AR 94;Combined oxidative phosphorylation deficiency 24	AR
NAXD	615910	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 2	AR
NAXE	608862	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	AR
NBAS	608025	Short stature, optic nerve atrophy, and Pelger-Huet anomaly;Infantile liver failure syndrome 2	AR
NDE1	609449	Lissencephaly 4 (with microcephaly);?Microhydranencephaly	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
NECAP1	611623	Developmental and epileptic encephalopathy 21	AR
NEDD4L	606384	Periventricular nodular heterotopia 7	AD
NEU1	608272	Sialidosis, type I;Sialidosis, type II	AR
NEUROD2	601725	Developmental and epileptic encephalopathy 72	AD
NEXMIF	300524	Mental retardation, XL 98	XLD
NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NFE2L2	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia	AD
NFU1	608100	Multiple Mi dysfunctions syndrome 1	AR
NGLY1	610661	Congenital disorder of deglycosylation	AR
NHLRC1	608072	Epilepsy, progressive myoclonic 2B (Lafora)	AR
NOTCH1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
NOTCH3	600276	?Myofibromatosis, infantile 2;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1;Lateral meningocele syndrome	AD
NPC1	607623	Niemann-Pick disease, type C1;Niemann-Pick disease, type D	AR

NPC2	601015	Niemann-pick disease, type C2	AR
NPR2	108961	Acromesomelic dysplasia, Maroteaux type;Short stature with nonspecific skeletal abnormalities;Epiphyseal chondrodysplasia, Miura type	AR, AD
NPRL2	607072	Epilepsy, familial focal, with variable foci 2	AD
NPRL3	600928	Epilepsy, familial focal, with variable foci 3	AD
NRXN1	600565	Schizophrenia, susceptibility to, 17;Pitt-Hopkins-like syndrome 2	AR
NSD2	602952	Rauch-Steindl syndrome	AD
NUBPL	613621	Mi complex I deficiency, nuclear type 21	AR
NUS1	610463	?Congenital disorder of glycosylation, type 1aa;Mental retardation, AD 55, with seizures	AR, AD
OAT	613349	Gyrate atrophy of choroid and retina with or without ornithinemia	AR
OCLN	602876	Pseudo-TORCH syndrome 1	AR
OCRL	300535	Lowe syndrome;Dent disease 2	XLR
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
OSGEP	610107	Galloway-Mowat syndrome 3	AR
OTC	300461	Ornithine transcarbamylase deficiency	XL
OTUD6B	612021	Intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies	AR
P4HTM	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities	AR
PACS2	610423	Developmental and epileptic encephalopathy 66	AD
PAFAH1B1	601545	Subcortical laminar heterotopia;Lissencephaly 1	AD
PAH	612349	Phenylketonuria;[Hyperphenylalaninemia, non-PKU mild]	AR
PAK1	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay	AD
PANK2	606157	HARP syndrome;Neurodegeneration with brain iron accumulation 1	AR
PARS2	612036	Developmental and epileptic encephalopathy 75	AR
PC	608786	Pyruvate carboxylase deficiency	AR
PCCA	232000	Propionicacidemia	AR
PCCB	232050	Propionicacidemia	AR
PCDH12	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1	AR
PCDH19	300460	Developmental and epileptic encephalopathy 9	XL

PCYT2	602679	Spastic paraplegia 82, AR	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
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1	AR
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B	AR
PEX11B	603867	Peroxisome biogenesis disorder 14B	AR
PEX12	601758	Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (Zellweger)	AR
PEX13	601789	Peroxisome biogenesis disorder 11B;Peroxisome biogenesis disorder 11A (Zellweger)	AR
PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger)	AR
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger);Peroxisome biogenesis disorder 8B	AR
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger)	AR
PEX2	170993	Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger)	AR
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger);Peroxisome biogenesis disorder 7B	AR
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger);?Peroxisome biogenesis disorder 10B	AR
PEX5	600414	Rhizomelic chondrodysplasia punctata, type 5;Peroxisome biogenesis disorder 2B;Peroxisome biogenesis disorder 2A (Zellweger)	AR
PEX6	601498	Peroxisome biogenesis disorder 4B;Peroxisome biogenesis disorder 4A (Zellweger);Heimler syndrome 2	AD, AR, AR
PEX7	601757	Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1	AR
PGAP1	611655	Mental retardation, AR 42	AR
PGK1	311800	Phosphoglycerate kinase 1 deficiency	XLR
PGM1	171900	Congenital disorder of glycosylation, type It	AR
PHACTR1	608723	Developmental and epileptic encephalopathy 70	AD
PHGDH	606879	Phosphoglycerate dehydrogenase deficiency;Neu-Laxova syndrome 1	AR
PHYH	602026	Refsum disease	AR

PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic;Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR
PIGB	604122	Developmental and epileptic encephalopathy 80	AR
PIGH	600154	Glycosylphosphatidylinositol biosynthesis defect 17	AR
PIGO	614730	Hyperphosphatasia with mental retardation syndrome 2	AR
PIGP	605938	Developmental and epileptic encephalopathy 55	AR
PIGQ	605754	Developmental and epileptic encephalopathy 77	AR
PIGS	610271	Glycosylphosphatidylinositol biosynthesis defect 18	AR
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3;?Paroxysmal nocturnal hemoglobinuria 2	AR, AD, SM
PIGU	608528	Neurodevelopmental disorder with brain anomalies, seizures, and scoliosis	AR
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1	AR
PIGW	610275	Glycosylphosphatidylinositol biosynthesis defect 11	AR
PLA2G6	603604	Neurodegeneration with brain iron accumulation 2B;Parkinson disease 14, AR;Infantile neuroaxonal dystrophy 1	AR
PLAA	603873	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	AR
PLCB1	607120	Developmental and epileptic encephalopathy 12	AR
PLCG2	600220	Familial cold autoinflammatory syndrome 3;Autoinflammation, antibody deficiency, and immune dysregulation syndrome	AD
PLEKHG2	611893	Leukodystrophy and acquired microcephaly with or without dystonia	AR
PLK1	602098		
PLP1	300401	Pelizaeus-Merzbacher disease;Spastic paraplegia 2, XL	XLR
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR
PMP22	601097	Neuropathy, recurrent, with pressure palsies;Dejerine-Sottas disease;Roussy-Levy syndrome;?Neuropathy, inflammatory demyelinating;Charcot-Marie-Tooth disease, type 1E;Charcot-Marie-Tooth disease, type 1A	AD, AD, AR, ?AD
PMPCB	603131	Multiple Mi dysfunctions syndrome 6	AR
PNKP	605610	Ataxia-oculomotor apraxia 4;Microcephaly, seizures, and developmental delay;?Charcot-Marie-Tooth disease, type 2B2	AR
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	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
POLR1C	610060	Leukodystrophy, hypomyelinating, 11;Treacher Collins syndrome 3	AR
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
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12;?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12	AR
POMT1	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	AR
PPP2CA	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities	AD
PPP2R1A	605983	Mental retardation, AD 36	AD
PPP3CA	114105	Epileptic encephalopathy, infantile or early childhood, 1;Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development	AD
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1	AR
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2;Lymphoma, non-Hodgkin;Aplastic anemia	AR
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B	AR
PRICKLE2	608501		
PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures	AR
PRODH	606810	Schizophrenia, susceptibility to, 4;Hyperprolinemia, type I	AD, AR
PRPS1	311850	Charcot-Marie-Tooth disease, XLR, 5;Deafness, XL 1;Phosphoribosylpyrophosphate synthetase superactivity;Arts syndrome;Gout, PRPS-related	XLR, XL
PRRT2	614386	Episodic kinesigenic dyskinesia 1;Convulsions, familial infantile, with paroxysmal choreoathetosis;Seizures, benign familial infantile, 2	AD
PSAP	176801	Combined SAP deficiency;Gaucher disease, atypical;Krabbe disease, atypical;Parkinson disease 24, AD, susceptibility to;Metachromatic leukodystrophy due to SAP-b deficiency	AR, AD

PSEN1	104311	Cardiomyopathy, dilated, 1U;Pick disease;Alzheimer disease, type 3;Alzheimer disease, type 3, with spastic paraparesis and apraxia;?Acne inversa, familial, 3;Dementia, frontotemporal;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques	AD
PTEN	601728	Cowden syndrome 1;Lhermitte-Duclos syndrome;Macrocephaly/autism syndrome;Glioma susceptibility 2;Meningioma;Prostate cancer, somatic	AD
PTPN23	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	AR
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	AR
PUM1	607204	Spinocerebellar ataxia 47	AD
PURA	600473	Mental retardation, AD 31	AD
PYCR2	616406	Leukodystrophy, hypomyelinating, 10	AR
QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	AR
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C	AR
RAB11A	605570		
RAB11B	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter	AD
RAB27A	603868	Griscelli syndrome, type 2	AR
RAI1	607642	Smith-Magenis syndrome	AD, IC
RALA	179550	Hiatt-Neu-Cooper neurodevelopmental syndrome	AD
RALGAPA1	608884	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation	AR
RARS1	107820	Leukodystrophy, hypomyelinating, 9	AR
RARS2	611524	Pontocerebellar hypoplasia, type 6	AR
RELN	600514	Lissencephaly 2 (Norman-Roberts type);Epilepsy, familial temporal lobe, 7	AR, AD
RFT1	611908	Congenital disorder of glycosylation, type In	AR
RHOBTB2	607352	Developmental and epileptic encephalopathy 64	AD
RMND1	614917	Combined oxidative phosphorylation deficiency 11	AR
RNASEH2A	606034	Aicardi-Goutieres syndrome 4	AR
RNASEH2B	610326	Aicardi-Goutieres syndrome 2	AR
RNASEH2C	610330	Aicardi-Goutieres syndrome 3	AR
RNASET2	612944	Leukoencephalopathy, cystic, without megalencephaly	AR
RNF113A	300951	Trichothiodystrophy 5, nonphotosensitive	XL
RNF13	609247	Developmental and epileptic encephalopathy 73	AD
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism	AR
ROGDI	614574	Kohlschutter-Tonz syndrome	AR

RORA	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia	AD
RORB	601972	Epilepsy, idiopathic generalized, susceptibility to, 15	AD
RPIA	180430	Ribose 5-phosphate isomerase deficiency	AR
RRM2B	604712	Mi DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy);Progressive external ophthalmoplegia with Mi DNA deletions, AD 5;Mi DNA depletion syndrome 8B (MNGIE type)	AR, AD
RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	AR
SAMHD1	606754	?Chilblain lupus 2;Aicardi-Goutieres syndrome 5	AD, AR
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure	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
SCN1B	600235	Atrial fibrillation, familial, 13;Epilepsy, generalized, with febrile seizures plus, type 1;Brugada syndrome 5;Cardiac conduction defect, nonspecific;Developmental and epileptic encephalopathy 52	AD, AR
SCN2A	182390	Developmental and epileptic encephalopathy 11;Seizures, benign familial infantile, 3;Episodic ataxia, type 9	AD
SCN3A	182391	Epilepsy, familial focal, with variable foci 4;Developmental and epileptic encephalopathy 62	AD
SCN8A	600702	?Myoclonus, familial, 2;Cognitive impairment with or without cerebellar ataxia;Developmental and epileptic encephalopathy 13;Seizures, benign familial infantile, 5	AD
SCN9A	603415	Erythermalgia, primary;Generalized epilepsy with febrile seizures plus, type 7;Febrile seizures, familial, 3B;Neuropathy, hereditary sensory and autonomic, type IID;Insensitivity to pain, congenital;Paroxysmal extreme pain disorder;Small fiber neuropathy	AD, AR
SCO1	603644	Mi complex IV deficiency, nuclear type 4	AR
SCO2	604272	Myopia 6;Mi complex IV deficiency, nuclear type 2	AD, 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
SDHB	185470	Pheochromocytoma;Mi complex II deficiency, nuclear type 4;Paragangliomas 4;Paraganglioma and gastric stromal sarcoma;Gastrointestinal stromal tumor	AD, AR, AD, IC

SDHD	602690	Mi complex II deficiency;Paraganglioma and gastric stromal sarcoma;Paragangliomas 1, with or without deafness;Mi complex II deficiency, nuclear type 3;Pheochromocytoma	AR, AD
SEC23B	610512	Dyserythropoietic anemia, congenital, type II;?Cowden syndrome 7	AR, AD
SELENOI	607915	Spastic paraplegia 81, AR	AR
SEMA6B	608873	Epilepsy, progressive myoclonic, 11	AD
SEPSECS	613009	Pontocerebellar hypoplasia type 2D	AR
SERAC1	614725	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	AR
SERPINI1	602445	Encephalopathy, familial, with neuroserpin inclusion bodies	AD
SETD1A	611052	Neurodevelopmental disorder with speech impairment and dysmorphic facies;Epilepsy, early-onset, with or without developmental delay	AD
SGCE	604149	Dystonia-11, myoclonic	AD
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR
SIK1	605705	Developmental and epileptic encephalopathy 30	AD
SLC12A3	600968	Gitelman syndrome	AR
SLC12A5	606726	Epilepsy, idiopathic generalized, susceptibility to, 14;Developmental and epileptic encephalopathy 34	AD, AR
SLC13A3	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate	AR
SLC13A5	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta	AR
SLC16A2	300095	Allan-Herndon-Dudley syndrome	XL
SLC17A5	604322	Sialic acid storage disorder, infantile;Salla disease	AR
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	AR
SLC1A2	600300	Developmental and epileptic encephalopathy 41	AD
SLC1A4	600229	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	AR
SLC22A5	603377	Carnitine deficiency, systemic primary	AR
SLC25A1	190315	Myasthenic syndrome, congenital, 23, presynaptic;Combined D-2- and L-2-hydroxyglutaric aciduria	AR
SLC25A12	603667	Developmental and epileptic encephalopathy 39	AR
SLC25A13	603859	Citrullinemia, type II, neonatal-onset;Citrullinemia, adult-onset type II	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	AR
SLC25A22	609302	Developmental and epileptic encephalopathy 3	AR

SLC25A3	600370	Mi phosphate carrier deficiency	AR
SLC25A4	103220	Mi DNA depletion syndrome 12B (cardiomyopathic type) AR;Mi DNA depletion syndrome 12A (cardiomyopathic type) AD;Progressive external ophthalmoplegia with Mi DNA deletions, AD 2	AR, AD
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
SLC35A1	605634	Congenital disorder of glycosylation, type IIf	AR
SLC35A2	314375	Congenital disorder of glycosylation, type IIm	SM, XLD
SLC35C1	605881	Congenital disorder of glycosylation, type IIc	AR
SLC39A8	608732	Congenital disorder of glycosylation, type IIn	AR
SLC6A1	137165	Myoclonic-atonic epilepsy	AD
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	XLR
SLC7A7	603593	Lysinuric protein intolerance	AR
SLC9A6	300231	Mental retardation, XL syndromic, Christianson type	XL
SMC1A	300040	Developmental and epileptic encephalopathy 85, with or without midline brain defects;Cornelia de Lange syndrome 2	XLD
SMPD1	607608	Niemann-Pick disease, type A;Niemann-Pick disease, type B	AR
SNIP1	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	AR
SNTA1	601017	Long QT syndrome 12	AD
SNX27	611541		
SON	182465	ZTTK syndrome	AD
SOX10	602229	Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SPART	607111	Troyer syndrome	AR
SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome	AR
SPG11	610844	Spastic paraplegia 11, AR;Amyotrophic lateral sclerosis 5, juvenile;Charcot-Marie-Tooth disease, axonal, type 2X	AR
SPG7	602783	Spastic paraplegia 7, AR	AD, AR
SPTAN1	182810	Developmental and epileptic encephalopathy 5	AD
SRD5A3	611715	Congenital disorder of glycosylation, type Iq;Kahrizi syndrome	AR
SSR4	300090	Congenital disorder of glycosylation, type Iy	XLR

ST3GAL3	606494	?Developmental and epileptic encephalopathy 15;Mental retardation, AR 12	AR
ST3GAL5	604402	Salt and pepper developmental regression syndrome	AR
STAMBP	606247	Microcephaly-capillary malformation syndrome	AR
STAT1	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, AD;Immunodeficiency 31B, mycobacterial and viral infections, AR;Immunodeficiency 31A, mycobacteriosis, AD	AD, AR
STT3A	601134	Congenital disorder of glycosylation, type Iw, AD;Congenital disorder of glycosylation, type Iw	AD, AR
STX1B	601485	Generalized epilepsy with febrile seizures plus, type 9	AD
STXBP1	602926	Developmental and epileptic encephalopathy 4	AD
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
SUMF1	607939	Multiple sulfatase deficiency	AR
SUOX	606887	Sulfite oxidase deficiency	AR
SURF1	185620	Charcot-Marie-Tooth disease, type 4K;Mi complex IV deficiency, nuclear type 1	AR
SYN1	313440	?Intellectual developmental disorder, XL 50;Epilepsy, XL, with variable learning disabilities and behavior disorders	XL, XLD, XLR
SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8	AR, AD
SYNGAP1	603384	Mental retardation, AD 5	AD
SYNJ1	604297	Developmental and epileptic encephalopathy 53;Parkinson disease 20, early-onset	AR
SZT2	615463	Developmental and epileptic encephalopathy 18	AR
TACO1	612958	Mi complex IV deficiency, nuclear type 8	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
TBCD	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	AR
TBCE	604934	Hypoparathyroidism-retardation-dysmorphism syndrome;Encephalopathy, progressive, with amyotrophy and optic atrophy;Kenny-Caffey syndrome, type 1	AR
TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	AR

TCF4	602272	Pitt-Hopkins syndrome;Corneal dystrophy, Fuchs endothelial, 3	AD
TDP2	605764	Spinocerebellar ataxia, AR 23	AR
TGFB1	190180	Camurati-Engelmann disease;Cystic fibrosis lung disease, modifier of;Inflammatory bowel disease, immunodeficiency, and encephalopathy	AD, AR
TIMM50	607381	3-methylglutaconic aciduria, type IX	AR
TIMM8A	300356	Mohr-Tranebjaerg syndrome	XLR
TINF2	604319	Revesz syndrome;Dyskeratosis congenita, AD 3	AD
TK2	188250	Mi DNA depletion syndrome 2 (myopathic type);?Progressive external ophthalmoplegia with Mi DNA deletions, AR 3	AR
TMEM106B	613413	Leukodystrophy, hypomyelinating, 16	AD
TMEM126A	612988	Optic atrophy 7	AR
TMEM165	614726	Congenital disorder of glycosylation, type IIk	AR
TMEM199	616815	Congenital disorder of glycosylation, type IIp	AR
TMEM70	612418	Mi complex V (ATP synthase) deficiency, nuclear type 2	AR
TMX2	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	AR
TNK2	606994		
TPK1	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	AR
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2;Spinocerebellar ataxia, AR 7	AR
TRAK1	608112	Developmental and epileptic encephalopathy 68	AR
TRAPPC4	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	AR
TRAPPC9	611966	Mental retardation, AR 13	AR
TREM2	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2	AR
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive;Chilblain lupus;Vasculopathy, retinal, with cerebral leukodystrophy;Systemic lupus erythematosus, susceptibility to	AD, AR, AD
TRIM8	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome	AD
TRPM6	607009	Hypomagnesemia 1, intestinal	AR

TRPV4	605427	SED, Maroteaux type;Spondylometaphyseal dysplasia, Kozlowski type;Metatropic dysplasia;Brachyolmia type 3;Neuronopathy, distal hereditary motor, type VIII;[Sodium serum level QTL 1];?Avascular necrosis of femoral head, primary, 2;Scapuloperoneal spinal muscular atrophy;Parastremmatic dwarfism;Hereditary motor and sensory neuropathy, type IIc;Digital arthropathy-brachydactyly, familial	AD
TSC1	605284	Lymphangioleiomyomatosis;Focal cortical dysplasia, type II, somatic;Tuberous sclerosis-1	AD
TSC2	191092	?Focal cortical dysplasia, type II, somatic;Lymphangioleiomyomatosis, somatic;Tuberous sclerosis-2	AD
TSEN54	608755	Pontocerebellar hypoplasia type 2A;?Pontocerebellar hypoplasia type 5;Pontocerebellar hypoplasia type 4	AR
TSFM	604723	Combined oxidative phosphorylation deficiency 3	AR
TTC19	613814	Mi complex III deficiency, nuclear type 2	AR
TUBA1A	602529	Lissencephaly 3	AD
TUBB2A	615101	Cortical dysplasia, complex, with other brain malformations 5	AD
TUBB2B	612850	Cortical dysplasia, complex, with other brain malformations 7	AD
TUBB4A	602662	Dystonia 4, torsion, AD;Leukodystrophy, hypomyelinating, 6	AD
TUBG1	191135	Cortical dysplasia, complex, with other brain malformations 4	AD
TUFM	602389	Combined oxidative phosphorylation deficiency 4	AR
TUSC3	601385	Mental retardation, AR 7	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
TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1	AR
UBE2A	312180	Mental retardation, XL syndromic, Nascimento-type	XLR
UBE3A	601623	Angelman syndrome	AD
UFM1	610553	Leukodystrophy, hypomyelinating, 14	AR
UGP2	191760	Developmental and epileptic encephalopathy 83	AR
UMPS	613891	Orotic aciduria	AR
UPB1	606673	Beta-ureidopropionase deficiency	AR
UQCRQ	612080	Mi complex III deficiency, nuclear type 4	AR
VAMP2	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements	AD
VARS1	192150	Neurodevelopmental disorder with microcephaly, seizures, and cortical atrophy	AR

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
VPS11	608549	Leukodystrophy, hypomyelinating, 12;?Dystonia 32	AR
WARS2	604733	Neurodevelopmental disorder, Mi, with abnormal movements and lactic acidosis, with or without seizures	AR
WASF1	605035	Neurodevelopmental disorder with absent language and variable seizures	AD
WDR37	618586	Neurooculocardiogenitourinary syndrome	AD
WDR45	300526	Neurodegeneration with brain iron accumulation 5	XLD
WDR45B	609226	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	AR
WDR73	616144	Galloway-Mowat syndrome 1	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
YWHAG	605356	Developmental and epileptic encephalopathy 56	AD
ZEB2	605802	Mowat-Wilson syndrome	AD
ZFYVE26	612012	Spastic paraplegia 15, AR	AR
ZNF142	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements	AR
ZNF335	610827	Microcephaly 10, primary, AR	AR
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	AR