Panel Neurológico

Es nuestro panel más grande, diseñado para detectar una gran variedad de trastornos neurológicos, desde casos de UCI neonatal hasta demencia o trastornos del movimiento en adultos. Este panel incluye genes relacionados con enfermedades neurológicas, como esclerosis lateral amiotrófica, demencia, Parkinson, enfermedades neuromusculares, Charcot-Marie-Tooth, distonía, epilepsia, autismo, discapacidad intelectual, migraña, paraplejía espástica, ataxia, síndrome de Leigh, peroxisomal enfermedades, encefalopatías epilépticas y trastornos del movimiento, entre otras.

Limitaciones: *DMD* solo se analiza mediante NGS. Si existe una alta sospecha diagnóstica de distrofia muscular de Duchenne, recomendamos que el médico ordene un análisis de deleción/duplicación por MLPA dirigido al gen *DMD como un servicio adicional.*

Nº de genes: 1902
Entrega: 25 días
Cobertura: ≥99,5% ≥20x
Cobertura media con profundidad ≥ 150 x
Detalles: Análisis CNV incluido

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

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
AAAS	605378	Achalasia-addisonianism-alacrimia syndrome	AR
AARS1	601065	Trichothiodystrophy 8, nonphotosensitive	AR
		Charcot-Marie-Tooth disease, axonal, type 2N	AD
		Developmental and epileptic encephalopathy 29	AR
		?Leukoencephalopathy, hereditary diffuse, with spheroids 2	AD
AARS2	612035	Combined oxidative phosphorylation deficiency 8	AR

		Leukoencephalopathy, progressive, with ovarian failure	AR
AASS	605113	Hyperlysinemia	AR
ABAT	137150	GABA-transaminase deficiency	AR
ABCA1	600046	HDL deficiency, familial, 1	AD
		Tangier disease	AR
ABCA7	605414	Alzheimer disease 9, susceptibility to	AD
ABCB6	605452	Microphthalmia, isolated, with coloboma 7	AD
		[Blood group, Langereis system]	-
		Dyschromatosis universalis hereditaria 3	AD
		Pseudohyperkalemia, familial, 2, due to red cell leak	AD
ABCB7	300135	Anemia, sideroblastic, with ataxia	XLR
ABCC6	603234	Pseudoxanthoma elasticum, forme fruste	AD
		Arterial calcification, generalized, of infancy, 2	AR
		Pseudoxanthoma elasticum	AR
ABCC8	600509	Diabetes mellitus, transient neonatal 2	-
		Diabetes mellitus, noninsulin-dependent	AD
		Hyperinsulinemic hypoglycemia, familial, 1	AD, AR
		Hypoglycemia of infancy, leucine-sensitive	AD
		Diabetes mellitus, permanent neonatal 3, with or without neurologic features	AD, AR

ABCD1	300371	Adrenoleukodystrophy	XLR
		Adrenomyeloneuropathy, adult	XLR
ABCD3	170995	?Bile acid synthesis defect, congenital, 5	AR
ABCD4	603214	Methylmalonic aciduria and homocystinuria, cblJ type	AR
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract	AR
ABHD5	604780	Chanarin-Dorfman syndrome	AR
ACACA	200350	Acetyl-CoA carboxylase deficiency	AR
ACAD8	604773	Isobutyryl-CoA dehydrogenase deficiency	AR
ACAD9	611103	Mitochondrial 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
ACADSB	600301	2-methylbutyrylglycinuria	AR
ACADVL	609575	VLCAD deficiency	AR
ACAT1	607809	Alpha-methylacetoacetic aciduria	AR
ACE	106180	Microvascular complications of diabetes 3	-
		Stroke, hemorrhagic	-
		Renal tubular dysgenesis	AR
ACHE	100740	[Blood group, Yt system]	-
ACO2	100850	?Optic atrophy 9	AR

		Infantile cerebellar-retinal degeneration	AR
ACOX1	609751	Peroxisomal acyl-CoA oxidase deficiency	AR
		Mitchell syndrome	AD
ACSF3	614245	Combined malonic and methylmalonic aciduria	-
ACSL4	300157	Mental retardation, X-linked 63	XLD
ACTA1	102610	Nemaline myopathy 3, autosomal dominant or recessive	AD, AR
		?Myopathy, scapulohumeroperoneal	AD
		Myopathy, congenital, with fiber-type disproportion 1	AD, AR
		Myopathy, actin, congenital, with cores	AD, AR
		Myopathy, actin, congenital, with excess of thin myofilaments	AD, AR
ACTA2	102620	Aortic aneurysm, familial thoracic 6	AD
		Moyamoya disease 5	-
		Multisystemic smooth muscle dysfunction syndrome	AD
ACTB	102630	Baraitser-Winter syndrome 1	AD
		?Dystonia, juvenile-onset	AD
ACTG1	102560	Baraitser-Winter syndrome 2	AD
		Deafness, autosomal dominant 20/26	AD
ACTG2	102545	Visceral myopathy	AD
		Megacystis-microcolon-intestinal hypoperistalsis syndrome 5	AD

ACTL6B	612458	Developmental and epileptic encephalopathy 76	AR
		Intellectual developmental disorder with severe speech and ambulation defects	AD
ACTN4	604638	Glomerulosclerosis, focal segmental, 1	AD
ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2	AD
ACY1	104620	Aminoacylase 1 deficiency	AR
ADA	608958	Adenosine deaminase deficiency, partial	AR, Somatic mosaicism
		Severe combined immunodeficiency due to ADA deficiency	AR, Somatic mosaicism
ADAM10	602192	Alzheimer disease 18, susceptibility to	-
		Reticulate acropigmentation of Kitamura	AD
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	AR
		Dyschromatosis symmetrica hereditaria	AD
ADAT3	615302	Mental retardation, autosomal recessive 36	AR
ADCY5	600293	Dyskinesia with orofacial involvement, autosomal recessive	AR
		Neurodevelopmental disorder with hyperkinetic movements and dyskinesia	AR
		Dyskinesia, familial, with facial myokymia	AD
ADGRG1	604110	Polymicrogyria, bilateral perisylvian	-

		Polymicrogyria, bilateral frontoparietal	AR
ADGRG6	612243	Lethal congenital contracture syndrome 9	AR
ADGRV1	602851	Usher syndrome, type 2C	AR, DD
		?Febrile seizures, familial, 4	AD
		Usher syndrome, type 2C, GPR98/PDZD7 digenic	AR, DD
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	AR
ADNP	611386	Helsmoortel-van der Aa syndrome	AD
ADPRS	610624	Neurodegeneration, childhood-onset, stress-induced, with variable ataxia and seizures	AR
ADSL	608222	Adenylosuccinase deficiency	AR
AFF2	300806	Mental retardation, X-linked, FRAXE type	XLR
AFF3	601464	KINSSHIP syndrome	AD
AFG3L2	604581	Spinocerebellar ataxia 28	AD
		Spastic ataxia 5, autosomal recessive	AR
		Optic atrophy 12	AD
AGA	613228	Aspartylglucosaminuria	AR
AGK	610345	Sengers syndrome	AR
		Cataract 38, autosomal recessive	AR
AGL	610860	Glycogen storage disease IIIb	AR
		Glycogen storage disease IIIa	AR

AGPS	603051	Rhizomelic chondrodysplasia punctata, type 3	AR
AGRN	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects	AR
AGTPBP1	606830	Neurodegeneration, childhood-onset, with cerebellar atrophy	AR
AGXT	604285	Hyperoxaluria, primary, type 1	AR
AHCY	180960	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AR
AHDC1	615790	Xia-Gibbs syndrome	AD
AHI1	608894	Joubert syndrome 3	AR
AIFM1	300169	Cowchock syndrome	XLR
		Combined oxidative phosphorylation deficiency 6	XLR
		Deafness, X-linked 5	XLR
		Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy	XLR
AIMP1	603605	Leukodystrophy, hypomyelinating, 3	AR
AIMP2	600859	Leukodystrophy, hypomyelinating, 17	AR
AK2	103020	Reticular dysgenesis	AR
AKT3	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	AD
ALAD	125270	Porphyria, acute hepatic	AR
		Lead poisoning, susceptibility to	AR
ALAS2	301300	Protoporphyria, erythropoietic, X-linked	XL
		Anemia, sideroblastic, 1	XLR

ALDH18A1	138250	Cutis laxa, autosomal dominant 3	AD
		Cutis laxa, autosomal recessive, type IIIA	AR
		Spastic paraplegia 9B, autosomal recessive	AR
		Spastic paraplegia 9A, autosomal dominant	AD
ALDH2	100650	Alcohol sensitivity, acute	AD
		Hangover, susceptibility to	AD
ALDH3A2	609523	Sjogren-Larsson syndrome	AR
ALDH4A1	606811	Hyperprolinemia, type II	AR
ALDH5A1	610045	Succinic semialdehyde dehydrogenase deficiency	AR
ALDH6A1	603178	Methylmalonate semialdehyde dehydrogenase deficiency	AR
ALDH7A1	107323	Epilepsy, pyridoxine-dependent	AR
ALDOA	103850	Glycogen storage disease XII	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	XL
		Developmental and epileptic encephalopathy 36	XL
ALG14	612866	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies	AR

		?Myasthenic syndrome, congenital, 15, without tubular aggregates	AR
		Myopathy, epilepsy, and progressive cerebral atrophy	AR
ALG2	607905	?Congenital disorder of glycosylation, type Ii	AR
		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	AD
		Congenital disorder of glycosylation, type Ih	AR
ALG9	606941	Congenital disorder of glycosylation, type Il	AR
		Gillessen-Kaesbach-Nishimura syndrome	AR
ALPL	171760	Hypophosphatasia, infantile	AR
		Odontohypophosphatasia	AD, AR
		Hypophosphatasia, childhood	AR
		Hypophosphatasia, adult	AD, AR
ALS2	606352	Amyotrophic lateral sclerosis 2, juvenile	AR
		Spastic paralysis, infantile onset ascending	AR
		Primary lateral sclerosis, juvenile	AR
ALX1	601527	Frontonasal dysplasia 3	AR
ALX3	606014	Frontonasal dysplasia 1	AR

ALX4	605420	Craniosynostosis 5, susceptibility to	AD
		Parietal foramina 2	AD
		Frontonasal dysplasia 2	AR
AMACR	604489	Bile acid synthesis defect, congenital, 4	AR
		Alpha-methylacyl-CoA racemase deficiency	AR
AMMECR1	300195	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis	XLR
AMPD1	102770	Myopathy due to myoadenylate deaminase deficiency	AR
AMPD2	102771	?Spastic paraplegia 63	AR
		Pontocerebellar hypoplasia, type 9	AR
AMT	238310	Glycine encephalopathy	AR
ANG	105850	Amyotrophic lateral sclerosis 9	-
ANK2	106410	Long QT syndrome 4	AD
		Cardiac arrhythmia, ankyrin-B-related	AD
ANK3	600465	Mental retardation, autosomal recessive, 37	AR
ANKLE2	616062	Microcephaly 16, primary, autosomal recessive	AR
ANKRD11	611192	KBG syndrome	AD
ANO10	613726	Spinocerebellar ataxia, autosomal recessive 10	AR
ANO3	610110	Dystonia 24	AD
ANO5	608662	Gnathodiaphyseal dysplasia	AD

		Muscular dystrophy, limb-girdle, autosomal recessive 12	AR
		Miyoshi muscular dystrophy 3	AR
ANTXR2	608041	Hyaline fibromatosis syndrome	AR
ANXA11	602572	Amytrophic lateral sclerosis 23	AD
		Inclusion body myopathy and brain white matter abnormalities	AD
AP1S1	603531	MEDNIK syndrome	AR
AP1S2	300629	Mental retardation, X-linked syndromic 5	XLR
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, autosomal recessive	AR
AP4E1	607244	Stuttering, familial persistent, 1	AD
		Spastic paraplegia 51, autosomal recessive	AR
AP4M1	602296	Spastic paraplegia 50, autosomal recessive	AR
AP4S1	607243	Spastic paraplegia 52, autosomal recessive	AR
AP5Z1	613653	Spastic paraplegia 48, autosomal recessive	AR
APOE	107741	Sea-blue histiocyte disease	AR
		Lipoprotein glomerulopathy	-
		?Alzheimer disease, protection against, due to APOE3-Christchurch	AD

		Hyperlipoproteinemia, type III	-
		Coronary artery disease, severe, susceptibility to	-
		?Macular degeneration, age-related	AD
		Alzheimer disease 2	AD
APP	104760	Alzheimer disease 1, familial	AD
		Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants	AD
APTX	606350	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	AR
ARFGEF2	605371	Periventricular heterotopia with microcephaly	AR
ARG1	608313	Argininemia	AR
ARHGAP31	610911	Adams-Oliver syndrome 1	AD
ARHGEF10	608136	?Slowed nerve conduction velocity, AD	AD
ARHGEF9	300429	Developmental and epileptic encephalopathy 8	XL
ARID1A	603024	Coffin-Siris syndrome 2	AD
ARID1B	614556	Coffin-Siris syndrome 1	AD
ARID2	609539	Coffin-Siris syndrome 6	AD
ARL13B	608922	Joubert syndrome 8	AR
ARL6	608845	Retinitis pigmentosa 55	AR
		Bardet-Biedl syndrome 1, modifier of	AR, DR
		Bardet-Biedl syndrome 3	AR

ARL6IP1	607669	?Spastic paraplegia 61, autosomal recessive	AR
ARSA	607574	Metachromatic leukodystrophy	AR
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR
ARSL	300180	Chondrodysplasia punctata, X-linked recessive	XLR
ARV1	611647	Developmental and epileptic encephalopathy 38	AR
ARX	300382	Developmental and epileptic encephalopathy 1	XLR
		Mental retardation, X-linked 29 and others	XLR
		Hydranencephaly with abnormal genitalia	XL
		Partington syndrome	XLR
		Lissencephaly, X-linked 2	XL
		Proud syndrome	XL
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy	AR
		Farber lipogranulomatosis	AR
ASCC1	614215	Spinal muscular atrophy with congenital bone fractures 2	AR
		Barrett esophagus/esophageal adenocarcinoma	-
ASCL1	100790	Central hypoventilation syndrome, congenital	AD
		Haddad syndrome	AD
ASH1L	607999	Mental retardation, autosomal dominant 52	AD
ASL	608310	Argininosuccinic aciduria	AR

ASNS	108370	Asparagine synthetase deficiency	AR
ASPA	608034	Canavan disease	AR
ASPM	605481	Microcephaly 5, primary, autosomal recessive	AR
ASS1	603470	Citrullinemia	AR
ASXL1	612990	Bohring-Opitz syndrome	AD
		Myelodysplastic syndrome, somatic	-
ASXL3	615115	Bainbridge-Ropers syndrome	AD
ATAD1	614452	Hyperekplexia 4	AR
ATCAY	608179	Ataxia, cerebellar, Cayman type	AR
ATIC	601731	AICA-ribosiduria due to ATIC deficiency	AR
ATL1	606439	Spastic paraplegia 3A, autosomal dominant	AD
		Neuropathy, hereditary sensory, type ID	AD
ATM	607585	Breast cancer, susceptibility to	AD, SM
		Ataxia-telangiectasia	AR
ATN1	607462	Dentatorubral-pallidoluysian atrophy	AD
		Congenital hypotonia, epilepsy, developmental delay, and digital anomalies	AD
ATP13A2	610513	Spastic paraplegia 78, autosomal recessive	AR
		Kufor-Rakeb syndrome	AR
ATP1A1	182310	Hypomagnesemia, seizures, and mental retardation 2	AD

		Charcot-Marie-Tooth disease, axonal, type 2DD	AD
ATP1A2	182340	Developmental and epileptic encephalopathy 98	AD
		Alternating hemiplegia of childhood 1	AD
		Migraine, familial basilar	AD
		Migraine, familial hemiplegic, 2	AD
		Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies	AR
ATP1A3	182350	Alternating hemiplegia of childhood 2	AD
		Dystonia-12	AD
		Developmental and epileptic encephalopathy 99	AD
		CAPOS syndrome	AD
ATP2A1	108730	Brody myopathy	AR
ATP2A2	108740	Acrokeratosis verruciformis	AD
		Darier disease	AD
ATP2B3	300014	?Spinocerebellar ataxia, X-linked 1	XLR
ATP5F1A	164360	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 4	AR
		?Combined oxidative phosphorylation deficiency 22	AR
ATP5F1E	606153	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3	AR
ATP6AP1	300197	Immunodeficiency 47	XLR
ATP6AP2	300556	Mental retardation, X-linked, syndromic, Hedera type	XLR

		?Parkinsonism with spasticity, X-linked	XLR
		Congenital disorder of glycosylation, type IIr	XLR
ATP6V0A2	611716	Cutis laxa, autosomal recessive, type IIA	AR
		Wrinkly skin syndrome	AR
ATP6V1A	607027	Epileptic encephalopathy, infantile or early childhood, 3	AD
		Cutis laxa, autosomal recessive, type IID	AR
ATP7A	300011	Menkes disease	XLR
		Occipital horn syndrome	XLR
		Spinal muscular atrophy, distal, X-linked 3	XLR
ATP7B	606882	Wilson disease	AR
ATP8A2	605870	?Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4	AR
ATPAF2	608918	?Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1	AR
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial	AD
		Seckel syndrome 1	AR
ATRX	300032	Mental retardation-hypotonic facies syndrome, X-linked	XLR
		Alpha-thalassemia/mental retardation syndrome	XLD
		Alpha-thalassemia myelodysplasia syndrome, somatic	-
AUH	600529	3-methylglutaconic aciduria, type I	AR
AUTS2	607270	Mental retardation, autosomal dominant 26	AD

B3GALNT2	610194	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11	AR
B3GLCT	610308	Peters-plus syndrome	AR
B4GALNT1	601873	Spastic paraplegia 26, autosomal recessive	AR
B4GALT1	137060	Congenital disorder of glycosylation, type IId	AR
B4GAT1	605517	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13	AR
B9D1	614144	?Meckel syndrome 9	AR
		Joubert syndrome 27	AR
B9D2	611951	?Meckel syndrome 10	AR
		Joubert syndrome 34	AR
BAG3	603883	Cardiomyopathy, dilated, 1HH	AD
		Myopathy, myofibrillar, 6	AD
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	AR
		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
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
BCKDK	614901	Branched-chain ketoacid dehydrogenase kinase deficiency	-
BCL11A	606557	Dias-Logan syndrome	AD
BCOR	300485	Microphthalmia, syndromic 2	XLD
BCS1L	603647	GRACILE syndrome	AR
		Bjornstad syndrome	AR
		Mitochondrial complex III deficiency, nuclear type 1	AR
BEST1	607854	Macular dystrophy, vitelliform, 2	AD
		Retinitis pigmentosa-50	-
		?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2	AD
		Vitreoretinochoroidopathy	AD
		Bestrophinopathy, autosomal recessive	-
		Retinitis pigmentosa, concentric	-
BICD2	609797	Spinal muscular atrophy, lower extremity-predominant, 2A, autosomal dominant	AD
		Spinal muscular atrophy, lower extremity-predominant, 2B, autosomal dominant	AD
BIN1	601248	Centronuclear myopathy 2	AR

BLOC1S3	609762	Hermansky-Pudlak syndrome 8	AR
BLOC1S6	604310	?Hermansky-pudlak syndrome 9	AR
BOLA3	613183	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	AR
BRAF	164757	Cardiofaciocutaneous syndrome	AD
		Adenocarcinoma of lung, somatic	-
		Noonan syndrome 7	AD
		Colorectal cancer, somatic	-
		Melanoma, malignant, somatic,	-
		LEOPARD syndrome 3	AD
BRAT1	614506	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	AR
		Rigidity and multifocal seizure syndrome, lethal neonatal	AR
BRWD3	300553	Mental retardation, X-linked 93	XLR
BSCL2	606158	Lipodystrophy, congenital generalized, type 2	AR
		Encephalopathy, progressive, with or without lipodystrophy	AR
		Silver spastic paraplegia syndrome	AD
		Neuropathy, distal hereditary motor, type VC	AD
BSND	606412	Bartter syndrome, type 4a	AR
		Sensorineural deafness with mild renal dysfunction	AR
BTD	609019	Biotinidase deficiency	AR

BVES	604577	Muscular dystrophy, limb-girdle, autosomal recessive 25	AR
C12orf4	616082	Mental retardation, autosomal recessive 66	AR
C12orf57	615140	Temtamy syndrome	AR
C19orf12	614297	Neurodegeneration with brain iron accumulation 4	AD, AR
		?Spastic paraplegia 43, autosomal recessive	AR
C1QBP	601269	Combined oxidative phosphorylation deficiency 33	AR
CA2	611492	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	AR
CA5A	114761	Hyperammonemia due to carbonic anhydrase VA deficiency	AR
CA8	114815	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	AR
CACNA1A	601011	Episodic ataxia, type 2	AD
		Migraine, familial hemiplegic, 1	AD
		Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia	AD
		Spinocerebellar ataxia 6	AD
		Developemental and epileptic encephalopathy 42	AD
CACNA1B	601012	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements	AR
CACNA1C	114205	Long QT syndrome 8	AD
		Brugada syndrome 3	-
		Timothy syndrome	AD
CACNA1D	114206	Primary aldosteronism, seizures, and neurologic abnormalities	AD

		Sinoatrial node dysfunction and deafness	AR
CACNA1E	601013	Developmental and epileptic encephalopathy 69	AD
CACNA1F	300110	Aland Island eye disease	XL
		Night blindness, congenital stationary (incomplete), 2A, X-linked	XL
		Cone-rod dystrophy, X-linked, 3	XLR
CACNA1G	604065	Spinocerebellar ataxia 42, early-onset, severe, with neurodevelopmental deficits	AD
		Spinocerebellar ataxia 42	AD
CACNA1H	607904	Hyperaldosteronism, familial, type IV	AD
		Epilepsy, childhood absence, susceptibility to, 6	-
		Epilepsy, idiopathic generalized, susceptibility to, 6	-
CACNA1S	114208	Thyrotoxic periodic paralysis, susceptibility to, 1	AD
		Malignant hyperthermia susceptibility 5	AD
		Malignant hyperthermia, susceptibility to, 5	AD
		Hypokalemic periodic paralysis, type 1	AD
CACNA2D2	607082	Cerebellar atrophy with seizures and variable developmental delay	AR
CACNB2	600003	Brugada syndrome 4	-
CACNB4	601949	Epilepsy, juvenile myoclonic, susceptibility to, 6	AD
		Epilepsy, idiopathic generalized, susceptibility to, 9	AD
		Episodic ataxia, type 5	AD

CAD	114010	Developmental and epileptic encephalopathy 50	AR
CAMK2A	114078	?Mental retardation, autosomal recessive 63	AR
		Mental retardation, autosomal dominant 53	AD
CAMK2B	607707	Mental retardation, autosomal dominant 54	AD
CAMK2G	602123	Mental retardation, autosomal dominant 59	AD
CAMTA1	611501	Cerebellar ataxia, nonprogressive, with mental retardation	AD
CAPN1	114220	Spastic paraplegia 76, autosomal recessive	AR
CAPN3	114240	Muscular dystrophy, limb-girdle, autosomal dominant 4	AD
		Muscular dystrophy, limb-girdle, autosomal recessive 1	AR
CARD11	607210	Immunodeficiency 11A	AR
		Immunodeficiency 11B with atopic dermatitis	AD
		B-cell expansion with NFKB and T-cell anergy	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	XLD
		FG syndrome 4	-
CASQ1	114250	Myopathy, vacuolar, with CASQ1 aggregates	AD
CASR	601199	Epilepsy idiopathic generalized, susceptibility to, 8	-
		Hypocalcemia, autosomal dominant, with Bartter syndrome	AD

		Hypocalciuric hypercalcemia, type I	AD
		Hyperparathyroidism, neonatal	AD, AR
		Hypocalcemia, autosomal dominant	AD
CAT	115500	Acatalasemia	-
CAV1	601047	Pulmonary hypertension, primary, 3	AD
		Lipodystrophy, familial partial, type 7	AD
		?Lipodystrophy, congenital generalized, type 3	AR
CAV3	601253	Rippling muscle disease 2	AD
		Cardiomyopathy, familial hypertrophic	AD, DD
		Creatine phosphokinase, elevated serum	AD
		Long QT syndrome 9	AD
		Myopathy, distal, Tateyama type	AD
CAVIN1	603198	Lipodystrophy, congenital generalized, type 4	AR
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	AD
		?Juvenile myelomonocytic leukemia	AD, SM
CBS	613381	Thrombosis, hyperhomocysteinemic	AR
		Homocystinuria, B6-responsive and nonresponsive types	AR
CC2D1A	610055	Mental retardation, autosomal recessive 3	AR
CC2D2A	612013	Meckel syndrome 6	AR

		Retinitis pigmentosa 93	AR
		Joubert syndrome 9	AR
		COACH syndrome 2	AR
CCDC115	613734	Congenital disorder of glycosylation, type IIo	AR
CCDC22	300859	Ritscher-Schinzel syndrome 2	XLR
CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CCDC78	614666	?Centronuclear myopathy 4	AD
CCDC88A	609736	?PEHO syndrome-like	AR
CCDC88C	611204	Hydrocephalus, congenital, 1	AR
		?Spinocerebellar ataxia 40	AD
CCM2	607929	Cerebral cavernous malformations-2	AD
CCNF	600227	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5	AD
CCT5	610150	Neuropathy, hereditary sensory, with spastic paraplegia	AR
CD320	606475	Methylmalonic aciduria, transient, due to transcobalamin receptor defect	AR
CD59	107271	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	AR
CD96	606037	C syndrome	AD
CDH11	600023	Teebi hypertelorism syndrome 2	AD
		Elsahy-Waters syndrome	AR
CDH15	114019	Mental retardation, autosomal dominant 3	-

CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive	AR
CDKL5	300203	Developmental and epileptic encephalopathy 2	XLD
CDON	608707	Holoprosencephaly 11	AD
CEL	114840	Maturity-onset diabetes of the young, type VIII	AD
CENPF	600236	Stromme syndrome	AR
CENPJ	609279	?Seckel syndrome 4	AR
		Microcephaly 6, primary, autosomal recessive	AR
CEP135	611423	Microcephaly 8, primary, autosomal recessive	AR
CEP152	613529	Microcephaly 9, primary, autosomal recessive	AR
		Seckel syndrome 5	AR
CEP164	614848	Nephronophthisis 15	AR
CEP290	610142	Leber congenital amaurosis 10	-
		Meckel syndrome 4	AR
		?Bardet-Biedl syndrome 14	AR
		Senior-Loken syndrome 6	AR
		Joubert syndrome 5	AR
CEP41	610523	Joubert syndrome 15	AR
CEP63	614724	?Seckel syndrome 6	AR
CERS1	606919	?Epilepsy, progressive myoclonic, 8	AR

CERT1	604677	Mental retardation, autosomal dominant 34	AD
CFAP418	614477	Retinitis pigmentosa 64	AR
		Cone-rod dystrophy 16	AR
		Bardet-Biedl syndrome 21	AR
CFL2	601443	Nemaline myopathy 7, autosomal recessive	AR
CHAMP1	616327	Mental retardation, autosomal dominant 40	AD
CHAT	118490	Myasthenic syndrome, congenital, 6, presynaptic	AR
CHCHD10	615903	?Myopathy, isolated mitochondrial, autosomal dominant	AD
		Spinal muscular atrophy, Jokela type	AD
		Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	AD
CHCHD2	616244	Parkinson disease 22, autosomal dominant	AD
CHD1	602118	Pilarowski-Bjornsson syndrome	AD
CHD2	602119	Epileptic encephalopathy, childhood-onset	AD
CHD3	602120	Snijders Blok-Campeau syndrome	AD
CHD7	608892	CHARGE syndrome	AD
		Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHD8	610528	Autism, susceptibility to, 18	AD
CHKB	612395	Muscular dystrophy, congenital, megaconial type	AR
CHMP1A	164010	Pontocerebellar hypoplasia, type 8	AR

CHMP2B	609512	Frontotemporal dementia and/or amytrophic lateral sclerosis 7	AD
CHRNA1	100690	Myasthenic syndrome, congenital, 1B, fast-channel	AD, AR
		Myasthenic syndrome, congenital, 1A, slow-channel	AD
		Multiple pterygium syndrome, lethal type	AR
CHRNA2	118502	Epilepsy, nocturnal frontal lobe, type 4	AD
CHRNA4	118504	Nicotine addiction, susceptibility to	-
		Epilepsy, nocturnal frontal lobe, 1	AD
CHRNB1	100710	Myasthenic syndrome, congenital, 2A, slow-channel	AD
		?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency	AR
CHRNB2	118507	Epilepsy, nocturnal frontal lobe, 3	-
CHRND	100720	?Myasthenic syndrome, congenital, 3A, slow-channel	AD
		Myasthenic syndrome, congenital, 3B, fast-channel	AR
		Multiple pterygium syndrome, lethal type	AR
		?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency	AR
CHRNE	100725	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency	AR
		Myasthenic syndrome, congenital, 4A, slow-channel	AD, AR
		Myasthenic syndrome, congenital, 4B, fast-channel	AR
CHRNG	100730	Multiple pterygium syndrome, lethal type	AR

		Escobar syndrome	AR
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	AR
CHSY1	608183	Temtamy preaxial brachydactyly syndrome	AR
CIB2	605564	Usher syndrome, type IJ	AR
		Deafness, autosomal recessive 48	AR
CIC	612082	Mental retardation, autosomal dominant 45	AD
CILK1	612325	Endocrine-cerebroosteodysplasia	AR
		Epilepsy, juvenile myoclonic, susceptibility to, 10	AD
CISD2	611507	Wolfram syndrome 2	AR
CIT	605629	Microcephaly 17, primary, autosomal recessive	AR
CLCN1	118425	Myotonia congenita, dominant	AD
		Myotonia congenita, recessive	AR
CLCN2	600570	Epilepsy, juvenile absence, susceptibility to, 2	AD
		Hyperaldosteronism, familial, type II	AD
		Epilepsy, juvenile myoclonic, susceptibility to, 8	AD
		Epilepsy, idiopathic generalized, susceptibility to, 11	AD
		Leukoencephalopathy with ataxia	AR
CLCN4	302910	Raynaud-Claes syndrome	XLD
CLCNKA	602024	Bartter syndrome, type 4b, digenic	DR

CLCNKB	602023	Bartter syndrome, type 3	AR
		Bartter syndrome, type 4b, digenic	DR
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	AR
		Ceroid lipofuscinosis, neuronal, 6	AR
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8	AR
		Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR
CLP1	608757	Pontocerebellar hypoplasia, type 10	AR
CLPB	616254	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR
		3-methylglutaconic aciduria, type VIIA, autosomal dominant	AD
		Neutropenia, severe congenital, 9, autosomal dominant	AD
CLPP	601119	Perrault syndrome 3	AR
CLTC	118955	Mental retardation, autosomal dominant 56	AD
CNBP	116955	Myotonic dystrophy 2	AD
CNGB3	605080	Achromatopsia 3	AR
CNKSR2	300724	Mental retardation, X-linked, syndromic, Houge type	XL

CNNM2	607803	Hypomagnesemia 6, renal	AD
		Hypomagnesemia, seizures, and mental retardation	AD, AR
CNPY3	610774	Developmental and epileptic encephalopathy 60	AR
CNTNAP1	602346	Lethal congenital contracture syndrome 7	AR
		Hypomyelinating neuropathy, congenital, 3	AR
CNTNAP2	604569	Cortical dysplasia-focal epilepsy syndrome	AR
		Pitt-Hopkins like syndrome 1	AR
		Autism susceptibility 15	-
COA5	613920	?Mitochondrial complex IV, deficiency, nuclear type 9	AR
COA6	614772	Mitochondrial complex IV deficiency, nuclear type 13	AR
COA7	615623	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 3	AR
COA8	616003	Mitochondrial complex IV deficiency, nuclear type 17	AR
COASY	609855	Neurodegeneration with brain iron accumulation 6	AR
		Pontocerebellar hypoplasia, type 12	AR
COG1	606973	Congenital disorder of glycosylation, type IIg	AR
COG4	606976	Congenital disorder of glycosylation, type IIj	AR
		Saul-Wilson syndrome	AD
COG5	606821	Congenital disorder of glycosylation, type IIi	AR
COG6	606977	Shaheen syndrome	AR

		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, autosomal recessive 53	AR
		Otospondylomegaepiphyseal dysplasia, autosomal recessive	AR
		Fibrochondrogenesis 2	AD, AR
		Otospondylomegaepiphyseal dysplasia, autosomal dominant	AD
		Deafness, autosomal dominant 13	AD
COL12A1	120320	?Ullrich congenital muscular dystrophy 2	AR
		Bethlem myopathy 2	AD
COL13A1	120350	Myasthenic syndrome, congenital, 19	AR
COL2A1	120140	Legg-Calve-Perthes disease	AD
		Stickler syndrome, type I	AD
		Osteoarthritis with mild chondrodysplasia	AD
		Platyspondylic skeletal dysplasia, Torrance type	AD
		Spondyloepiphyseal dysplasia, Stanescu type	AD
		Kniest dysplasia	AD
		Czech dysplasia	AD
		Stickler sydrome, type I, nonsyndromic ocular	AD

		?Vitreoretinopathy with phalangeal epiphyseal dysplasia	AD
		?Epiphyseal dysplasia, multiple, with myopia and deafness	AD
		Avascular necrosis of the femoral head	AD
		Spondyloperipheral dysplasia	AD
		Achondrogenesis, type II or hypochondrogenesis	AD
		SMED Strudwick type	AD
		SED congenita	AD
COL4A1	120130	?Retinal arteries, tortuosity of	AD
		Hemorrhage, intracerebral, susceptibility to	-
		Microangiopathy and leukoencephalopathy, pontine, autosomal dominant	AD
		Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	AD
		Brain small vessel disease with or without ocular anomalies	AD
COL4A2	120090	Hemorrhage, intracerebral, susceptibility to	-
		Brain small vessel disease 2	AD
COL6A1	120220	Bethlem myopathy 1	AD, AR
		Ullrich congenital muscular dystrophy 1	AD, AR
COL6A2	120240	Bethlem myopathy 1	AD, AR
		Ullrich congenital muscular dystrophy 1	AD, AR
		?Myosclerosis, congenital	AR

COL6A3	120250	Dystonia 27	AR
		Ullrich congenital muscular dystrophy 1	AD, AR
		Bethlem myopathy 1	AD, AR
COLGALT1	617531	Brain small vessel disease 3	AR
COLQ	603033	Myasthenic syndrome, congenital, 5	AR
COMT	116790	Panic disorder, susceptibility to	?AD
		Schizophrenia, susceptibility to	AD
COQ2	609825	Coenzyme Q10 deficiency, primary, 1	AR
		Multiple system atrophy, susceptibility to	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	Mitochondrial complex IV deficiency, nuclear type 3	AR
COX14	614478	?Mitochondrial complex IV deficiency, nuclear type 10	AR
COX15	603646	Mitochondrial complex IV deficiency, nuclear type 6	AR
COX20	614698	Mitochondrial complex IV deficiency, nuclear type 11	AR

COX4I2	607976	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	AR
COX6A1	602072	Charcot-Marie-Tooth disease, recessive intermediate D	AR
COX6B1	124089	Mitochondrial complex IV deficiency, nuclear type 7	AR
COX7B	300885	Linear skin defects with multiple congenital anomalies 2	XLD
CP	117700	Cerebellar ataxia	AR
		Hemosiderosis, systemic, due to aceruloplasminemia	AR
		[Hypoceruloplasminemia, hereditary]	AR
CPA6	609562	Febrile seizures, familial, 11	AR
		Epilepsy, familial temporal lobe, 5	AD, AR
CPLANE1	614571	Orofaciodigital syndrome VI	AR
		Joubert syndrome 17	AR
CPLX1	605032	Developmental and epileptic encephalopathy 63	AR
CPOX	612732	Harderoporphyria	AR
		Coproporphyria	AD, AR
CPS1	608307	Carbamoylphosphate synthetase I deficiency	AR
		Pulmonary hypertension, neonatal, susceptibility to	-
CPT1A	600528	CPT deficiency, hepatic, type IA	AR
CPT1C	608846	?Spastic paraplegia 73, autosomal dominant	AD
CPT2	600650	CPT II deficiency, myopathic, stress-induced	AD, AR

		CPT II deficiency, infantile	AR
		Encephalopathy, acute, infection-induced, 4, susceptibility to	AD, AR
		CPT II deficiency, lethal neonatal	AR
CRADD	603454	Mental retardation, autosomal recessive 34, with variant lissencephaly	AR
CRBN	609262	Mental retardation, autosomal recessive 2	AR
CREBBP	600140	Menke-Hennekam syndrome 1	AD
		Rubinstein-Taybi syndrome 1	AD
CRIPT	604594	Short stature with microcephaly and distinctive facies	AR
CRLF1	604237	Cold-induced sweating syndrome 1	AR
CRPPA	614631	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7	AR
CRYAB	123590	Cataract 16, multiple types	AD, AR
		Cardiomyopathy, dilated, 1II	AD
		Myopathy, myofibrillar, 2	AD
		Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related	AR
CSF1R	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis	AR
		Leukoencephalopathy, diffuse hereditary, with spheroids	AD
CSNK2B	115441	Poirier-Bienvenu neurodevelopmental syndrome	AD
CSPP1	611654	Joubert syndrome 21	AR

CSRP3	600824	?Cardiomyopathy, dilated, 1M	-
		Cardiomyopathy, hypertrophic, 12	AD
CST3	604312	Macular degeneration, age-related, 11	-
		Cerebral amyloid angiopathy	AD
CSTB	601145	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg)	AR
CTC1	613129	Cerebroretinal microangiopathy with calcifications and cysts	AR
CTCF	604167	Mental retardation, autosomal dominant 21	AD
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy	AR
CTNNA2	114025	Cortical dysplasia, complex, with other brain malformations 9	AR
CTNNA3	607667	Arrhythmogenic right ventricular dysplasia, familial, 13	AD
CTNNB1	116806	Ovarian cancer, somatic	-
		Colorectal cancer, somatic	-
		Pilomatricoma, somatic	-
		Neurodevelopmental disorder with spastic diplegia and visual defects	AD
		Exudative vitreoretinopathy 7	AD
		Medulloblastoma, somatic	-
		Hepatocellular carcinoma, somatic	-
CTNS	606272	Cystinosis, late-onset juvenile or adolescent nephropathic	AR
		Cystinosis, ocular nonnephropathic	AR

		Cystinosis, nephropathic	AR
		Cystinosis, atypical nephropathic	AR
CTSA	613111	Galactosialidosis	AR
CTSC	602365	Periodontitis 1, juvenile	AR
		Haim-Munk syndrome	AR
		Papillon-Lefevre syndrome	AR
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10	AR
CTSF	603539	Ceroid lipofuscinosis, neuronal, 13, Kufs type	AR
CTSK	601105	Pycnodysostosis	AR
CUL3	603136	Pseudohypoaldosteronism, type IIE	AD
		Neurodevelopmental disorder with or without autism or seizures	AD
CUL4B	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type)	XLR
CUL7	609577	3-M syndrome 1	AR
CUX1	116896	Global developmental delay with or without impaired intellectual development	AD
CUX2	610648	Developmental and epileptic encephalopathy 67	AD
CWF19L1	616120	Spinocerebellar ataxia, autosomal recessive 17	AR
CX3CR1	601470	Macular degeneration, age-related, 12	-
		Coronary artery disease, resistance to	-
		Rapid progression to AIDS from HIV1 infection	-

CYB5A	613218	Methemoglobinemia and ambiguous genitalia	AR
CYB5R3	613213	Methemoglobinemia, type I	AR
		Methemoglobinemia, type II	AR
CYC1	123980	Mitochondrial complex III deficiency, nuclear type 6	AR
CYCS	123970	Thrombocytopenia 4	AD
CYFIP2	606323	Developmental and epileptic encephalopathy 65	AD
CYLD	605018	Trichoepithelioma, multiple familial, 1	AD
		?Frontotemporal dementia and/or amytrophic lateral sclerosis 8	-
		Cylindromatosis, familial	AD
		Brooke-Spiegler syndrome	AD
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	-
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	AR
		Aldosteronism, glucocorticoid-remediable	AD
CYP11B2	124080	Hypoaldosteronism, congenital, due to CMO II deficiency	AR
		Hypoaldosteronism, congenital, due to CMO I deficiency	AR
CYP24A1	126065	Hypercalcemia, infantile, 1	AR
CYP27A1	606530	Cerebrotendinous xanthomatosis	AR
CYP27B1	609506	Vitamin D-dependent rickets, type I	AR
CYP2U1	610670	Spastic paraplegia 56, autosomal recessive	AR

CYP7B1	603711	Spastic paraplegia 5A, autosomal recessive	AR
		Bile acid synthesis defect, congenital, 3	AR
D2HGDH	609186	D-2-hydroxyglutaric aciduria	AR
DAB1	603448	Spinocerebellar ataxia 37	AD
DAG1	128239	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9	AR
		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
DCTN1	601143	Perry syndrome	AD
		Neuronopathy, distal hereditary motor, type VIIB	AD
		Amyotrophic lateral sclerosis, susceptibility to	AD, AR
DCX	300121	Lissencephaly, X-linked	XL
		Subcortical laminal heterotopia, X-linked	XL
DDC	107930	Aromatic L-amino acid decarboxylase deficiency	AR
DDHD1	614603	Spastic paraplegia 28, autosomal recessive	AR
DDHD2	615003	Spastic paraplegia 54, autosomal recessive	AR
DDOST	602202	?Congenital disorder of glycosylation, type Ir	AR

DDX3X	300160	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type	XLD, XLR
DEAF1	602635	Vulto-van Silfout-de Vries syndrome	AD
		Neurodevelopmental disorder with hypotonia, impaired expressive language, and with or without seizures	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
DES	125660	Cardiomyopathy, dilated, 1I	AD
		Myopathy, myofibrillar, 1	AD, AR
		Scapuloperoneal syndrome, neurogenic, Kaeser type	AD
DGUOK	601465	Portal hypertension, noncirrhotic	AR
		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4	AR
		Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	AR
DHCR24	606418	Desmosterolosis	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DHDDS	608172	Developmental delay and seizures with or without movement abnormalities	AD
		Retinitis pigmentosa 59	AR
		?Congenital disorder of glycosylation, type 1bb	AR
DHFR	126060	Megaloblastic anemia due to dihydrofolate reductase deficiency	AR
DHH	605423	46XY gonadal dysgenesis with minifascicular neuropathy	AR

		46XY sex reversal 7	AR
DHODH	126064	Miller syndrome	AR
DHPS	600944	Neurodevelopmental disorder with seizures and speech and walking impairment	AR
DHTKD1	614984	2-aminoadipic 2-oxoadipic aciduria	AR
		?Charcot-Marie-Tooth disease, axonal, type 2Q	AD
DHX30	616423	Neurodevelopmental disorder with severe motor impairment and absent language	AD
DIABLO	605219	Deafness, autosomal dominant 64	AD
DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome	AR
		Deafness, autosomal dominant 1, with or without thrombocytopenia	AD
DIAPH3	614567	Auditory neuropathy, autosomal dominant, 1	AD
DIP2B	611379	Mental retardation, FRA12A type	AD
DKC1	300126	Dyskeratosis congenita, X-linked	XLR
DLAT	608770	Pyruvate dehydrogenase E2 deficiency	AR
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	AR
DLG3	300189	Mental retardation, X-linked 90	XLR
DLG4	602887	Intellectual developmental disorder 62	AD
DLL3	602768	Spondylocostal dysostosis 1, autosomal recessive	AR
DLX3	600525	Amelogenesis imperfecta, type IV	AD
		Trichodontoosseous syndrome	AD

DMD	300377	Cardiomyopathy, dilated, 3B	XL
		Duchenne muscular dystrophy	XLR
		Becker muscular dystrophy	XLR
DMGDH	605849	Dimethylglycine dehydrogenase deficiency	AR
DMPK	605377	Myotonic dystrophy 1	AD
DMXL2	612186	?Polyendocrine-polyneuropathy syndrome	AR
		Developmental and epileptic encephalopathy 81	AR
		?Deafness, autosomal dominant 71	AD
DNA2	601810	?Seckel syndrome 8	AR
		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6	AD
DNAJB2	604139	Spinal muscular atrophy, distal, autosomal recessive, 5	AR
DNAJB6	611332	Muscular dystrophy, limb-girdle, autosomal dominant 1	AD
DNAJC12	606060	Hyperphenylalaninemia, mild, non-BH4-deficient	AR
DNAJC19	608977	3-methylglutaconic aciduria, type V	AR
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type	AD
DNAJC6	608375	Parkinson disease 19b, early-onset	AR
		Parkinson disease 19a, juvenile-onset	AR
DNM1	602377	Developmental and epileptic encephalopathy 31	AD
DNM1L	603850	Optic atrophy 5	AD

		Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1	AD, AR
DNM2	602378	Lethal congenital contracture syndrome 5	AR
		Charcot-Marie-Tooth disease, axonal type 2M	AD
		Charcot-Marie-Tooth disease, dominant intermediate B	AD
		Centronuclear myopathy 1	AD
DNMT1	126375	Neuropathy, hereditary sensory, type IE	AD
		Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant	AD
DNMT3A	602769	Tatton-Brown-Rahman syndrome	AD
		Heyn-Sproul-Jackson syndrome	AD
		Acute myeloid leukemia, somatic	-
DOCK3	603123	Neurodevelopmental disorder with impaired intellectual development, hypotonia, and ataxia	AR
DOCK6	614194	Adams-Oliver syndrome 2	AR
DOCK7	615730	Developmental and epileptic encephalopathy 23	AR
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive	AR
DOK7	610285	Fetal akinesia deformation sequence 3	AR
		Myasthenic syndrome, congenital, 10	AR
DOLK	610746	Congenital disorder of glycosylation, type Im	AR
DPAGT1	191350	Myasthenic syndrome, congenital, 13, with tubular aggregates	AR
		Congenital disorder of glycosylation, type Ij	AR

DPF2	601671	Coffin-Siris syndrome 7	AD
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	AR
		?Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 15	AR
DPP6	126141	Ventricular fibrillation, paroxysmal familial, 2	AD
		Mental retardation, autosomal dominant 33	-
DPYD	612779	5-fluorouracil toxicity	AR
		Dihydropyrimidine dehydrogenase deficiency	AR
DPYS	613326	Dihydropyrimidinuria	AR
DRD3	126451	Schizophrenia, susceptibility to	AD
		Essential tremor, hereditary, 1	AD
DST	113810	?Neuropathy, hereditary sensory and autonomic, type VI	AR
		Epidermolysis bullosa simplex, autosomal recessive 2	AR
DSTYK	612666	Spastic paraplegia 23	AR
		Congenital anomalies of kidney and urinary tract 1	AD
DTNBP1	607145	Hermansky-Pudlak syndrome 7	AR
DVL3	601368	Robinow syndrome, autosomal dominant 3	AD
DYM	607461	Dyggve-Melchior-Clausen disease	AR

		Smith-McCort dysplasia	AR
DYNC1H1	600112	Spinal muscular atrophy, lower extremity-predominant 1, AD	AD
		Charcot-Marie-Tooth disease, axonal, type 20	AD
		Mental retardation, autosomal dominant 13	AD
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly	AR, DR
DYRK1A	600855	Mental retardation, autosomal dominant 7	AD
DYSF	603009	Muscular dystrophy, limb-girdle, autosomal recessive 2	AR
		Myopathy, distal, with anterior tibial onset	AR
		Miyoshi muscular dystrophy 1	AR
EARS2	612799	Combined oxidative phosphorylation deficiency 12	AR
EBF3	607407	Hypotonia, ataxia, and delayed development syndrome	AD
EBP	300205	MEND syndrome	XLR
		Chondrodysplasia punctata, X-linked dominant	XLD
ECEL1	605896	Arthrogryposis, distal, type 5D	AR
ECHS1	602292	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	AR
EDC3	609842	?Mental retardation, autosomal recessive 50	AR
EDN3	131242	Waardenburg syndrome, type 4B	AD, AR
		Hirschsprung disease, susceptibility to, 4	AD
		Central hypoventilation syndrome, congenital	AD

EDNRB	131244	Waardenburg syndrome, type 4A	AD, AR
		Hirschsprung disease, susceptibility to, 2	AD
		ABCD syndrome	AR
EEF1A2	602959	Mental retardation, autosomal dominant 38	AD
		Developmental and epileptic encephalopathy 33	AD
EFHC1	608815	Epilepsy, juvenile absence, susceptibility to, 1	AD
		Myoclonic epilepsy, juvenile, susceptibility to, 1	AD
EFTUD2	603892	Mandibulofacial dysostosis, Guion-Almeida type	AD
EGF	131530	Hypomagnesemia 4, renal	AR
EGR2	129010	Dejerine-Sottas disease	AD, AR
		Hypomyelinating neuropathy, congenital, 1	AD, AR
		Charcot-Marie-Tooth disease, type 1D	AD
EHMT1	607001	Kleefstra syndrome 1	AD
EIF2B1	606686	Leukoencephalopathy with vanishing white matter	AR
EIF2B2	606454	Ovarioleukodystrophy	AR
		Leukoencephalopathy with vanishing white matter	AR
EIF2B3	606273	Leukoencephalopathy with vanishing white matter	AR
EIF2B4	606687	Ovarioleukodystrophy	AR
		Leukoencephalopathy with vanishing white matter	AR

EIF2B5	603945	Ovarioleukodystrophy	AR
		Leukoencephalopathy with vanishing white matter	AR
EIF2S3	300161	MEHMO syndrome	XLR
EIF3F	603914	Mental retardation, autosomal recessive 67	AR
EIF4G1	600495	Parkinson disease 18	AD
ELAC2	605367	Prostate cancer, hereditary, 2, susceptibility to	-
		Combined oxidative phosphorylation deficiency 17	AR
ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation	AR
		Stargardt disease 3	AD
		Spinocerebellar ataxia 34	AD
ELOVL5	611805	Spinocerebellar ataxia 38	AD
ELP1	603722	Dysautonomia, familial	AR
		Medulloblastoma	AD, AR, SM
ELP2	616054	Mental retardation, autosomal recessive 58	AR
EMC10	614545	Neurodevelopmental disorder with dysmorphic facies and variable seizures	AR
EMD	300384	Emery-Dreifuss muscular dystrophy 1, X-linked	XLR
EML1	602033	Band heterotopia	AR
EMX2	600035	Schizencephaly	-
ENO3	131370	?Glycogen storage disease XIII	AR

ENTPD1	601752	Spastic paraplegia 64, autosomal recessive	AR
EP300	602700	Rubinstein-Taybi syndrome 2	AD
		Colorectal cancer, somatic	-
		Menke-Hennekam syndrome 2	AD
EPB41L1	602879	?Mental retardation, autosomal dominant 11	AD
EPG5	615068	Vici syndrome	AR
EPHX2	132811	Hypercholesterolemia, familial, due to LDLR defect, modifier of	AD, AR
EPM2A	607566	Epilepsy, progressive myoclonic 2A (Lafora)	AR
EPRS1	138295	Leukodystrophy, hypomyelinating, 15	AR
ERBB4	600543	Amyotrophic lateral sclerosis 19	AD
ERCC1	126380	Cerebrooculofacioskeletal syndrome 4	AR
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2	AR
		Xeroderma pigmentosum, group D	AR
		Trichothiodystrophy 1, photosensitive	AR
ERCC5	133530	Xeroderma pigmentosum, group G	AR
		Cerebrooculofacioskeletal syndrome 3	AR
		Xeroderma pigmentosum, group G/Cockayne syndrome	AR
ERCC6	609413	Lung cancer, susceptibility to	AD, SM
		UV-sensitive syndrome 1	AR

		Premature ovarian failure 11	AD
		Macular degeneration, age-related, susceptibility to, 5	-
		Cockayne syndrome, type B	AR
		De Sanctis-Cacchione syndrome	AR
		Cerebrooculofacioskeletal syndrome 1	AR
ERCC8	609412	Cockayne syndrome, type A	AR
		UV-sensitive syndrome 2	AR
ERLIN1	611604	Spastic paraplegia 62	AR
ERLIN2	611605	Spastic paraplegia 18, autosomal recessive	AR
ESCO2	609353	Roberts syndrome	AR
		Juberg-Hayward syndrome	AR
		SC phocomelia syndrome	AR
ETFA	608053	Glutaric acidemia IIA	AR
ETFB	130410	Glutaric acidemia IIB	AR
ETFDH	231675	Glutaric acidemia IIC	AR
ETHE1	608451	Ethylmalonic encephalopathy	AR
EWSR1	133450	Ewing sarcoma	-
		Neuroepithelioma	-
EXOC6B	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3	AR

EXOSC3	606489	Pontocerebellar hypoplasia, type 1B	AR
EXOSC8	606019	Pontocerebellar hypoplasia, type 1C	AR
EXOSC9	606180	Pontocerebellar hypoplasia, type 1D	AR
EXT1	608177	Chondrosarcoma	SM
		Exostoses, multiple, type 1	AD
EZH2	601573	Weaver syndrome	AD
F2	176930	Dysprothrombinemia	AR
		Stroke, ischemic, susceptibility to	MF
		Pregnancy loss, recurrent, susceptibility to, 2	AD
		Hypoprothrombinemia	AR
		Thrombophilia due to thrombin defect	AD
F5	612309	Pregnancy loss, recurrent, susceptibility to, 1	AD
		Factor V deficiency	AR
		Stroke, ischemic, susceptibility to	MF
		Budd-Chiari syndrome	AR
		Thrombophilia, susceptibility to, due to factor V Leiden	AD
		Thrombophilia due to activated protein C resistance	AD
FA2H	611026	Spastic paraplegia 35, autosomal recessive	AR
FADD	602457	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations	AR

FAH	613871	Tyrosinemia, type I	AR
FAM126A	610531	Leukodystrophy, hypomyelinating, 5	AR
FAN1	613534	Interstitial nephritis, karyomegalic	AR
FANCB	300515	Fanconi anemia, complementation group B	XLR
FARS2	611592	Combined oxidative phosphorylation deficiency 14	AR
		Spastic paraplegia 77, autosomal recessive	AR
FARSB	609690	Rajab interstitial lung disease with brain calcifications 1	AR
FASTKD2	612322	Combined oxidative phosphorylation deficiency 44	AR
FAT2	604269	Spinocerebellar ataxia 45	AD
FBLN5	604580	Cutis laxa, autosomal recessive, type IA	AR
		Neuropathy, hereditary, with or without age-related macular degeneration	AD
		Charcot-Marie-Tooth disease, demyelinating, type 1H	AD
		Macular degeneration, age-related, 3	AD
		?Cutis laxa, autosomal dominant 2	AD
FBN1	134797	Marfan lipodystrophy syndrome	AD
		Geleophysic dysplasia 2	AD
		Acromicric dysplasia	AD
		Marfan syndrome	AD
		Weill-Marchesani syndrome 2, dominant	AD

		Stiff skin syndrome	AD
		MASS syndrome	AD
		Ectopia lentis, familial	AD
FBN2	612570	Contractural arachnodactyly, congenital	AD
		Macular degeneration, early-onset	AD
FBXL4	605654	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	AR
FBXO11	607871	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities	AD
FBXO38	608533	Neuronopathy, distal hereditary motor, type IID	AD
FBXO7	605648	Parkinson disease 15, autosomal recessive	AR
FDX2	614585	Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy	AR
FDXR	103270	Auditory neuropathy and optic atrophy	AR
FECH	612386	Protoporphyria, erythropoietic, 1	AR
FEZF1	613301	Hypogonadotropic hypogonadism 22, with or without anosmia	AR
FGA	134820	Hypodysfibrinogenemia, congenital	-
		Afibrinogenemia, congenital	AR
		Amyloidosis, familial visceral	AD
		Dysfibrinogenemia, congenital	-
FGD1	300546	Aarskog-Scott syndrome	XLR
		Mental retardation, X-linked syndromic 16	XLR

FGD4	611104	Charcot-Marie-Tooth disease, type 4H	AR
FGF10	602115	Aplasia of lacrimal and salivary glands	AD
		LADD syndrome	AD
FGF12	601513	Developmental and epileptic encephalopathy 47	AD
FGF14	601515	Spinocerebellar ataxia 27	AD
FGFR2	176943	Crouzon syndrome	AD
		Saethre-Chotzen syndrome	AD
		Craniofacial-skeletal-dermatologic dysplasia	AD
		Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	AD
		Gastric cancer, somatic	-
		LADD syndrome	AD
		Beare-Stevenson cutis gyrata syndrome	AD
		Scaphocephaly, maxillary retrusion, and mental retardation	-
		Apert syndrome	AD
		Bent bone dysplasia syndrome	AD
		Pfeiffer syndrome	AD
		Jackson-Weiss syndrome	AD
FGFR3	134934	Achondroplasia	AD
		CATSHL syndrome	AD, AR

		Thanatophoric dysplasia, type I	AD
		Bladder cancer, somatic	-
		Hypochondroplasia	AD
		SADDAN	AD
		Colorectal cancer, somatic	-
		Crouzon syndrome with acanthosis nigricans	AD
		Cervical cancer, somatic	-
		Nevus, epidermal, somatic	-
		Thanatophoric dysplasia, type II	AD
		Spermatocytic seminoma, somatic	-
		Muenke syndrome	AD
		LADD syndrome	AD
FH	136850	Fumarase deficiency	AR
		Leiomyomatosis and renal cell cancer	AD
FHL1	300163	Reducing body myopathy, X-linked 1b, with late childhood or adult onset	XL
		Scapuloperoneal myopathy, X-linked dominant	XLD
		?Uruguay faciocardiomusculoskeletal syndrome	XLR
		Myopathy, X-linked, with postural muscle atrophy	XLR
		Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset	XLD

		Emery-Dreifuss muscular dystrophy 6, X-linked	XLR
FIG4	609390	Amyotrophic lateral sclerosis 11	AD
		Yunis-Varon syndrome	AR
		Charcot-Marie-Tooth disease, type 4J	AR
		?Polymicrogyria, bilateral temporooccipital	AR
FKBP10	607063	Osteogenesis imperfecta, type XI	AR
		Bruck syndrome 1	AR
FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	AR
FKRP	606596	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR
FKTN	607440	Cardiomyopathy, dilated, 1X	AR
		Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	AR
FLAD1	610595	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency	AR
FLNA	300017	Frontometaphyseal dysplasia 1	XLR
		Heterotopia, periventricular, 1	XLD

		Terminal osseous dysplasia	XLD
		Congenital short bowel syndrome	XLR
		Otopalatodigital syndrome, type II	XLD
		Melnick-Needles syndrome	XLD
		Cardiac valvular dysplasia, X-linked	XL
		Intestinal pseudoobstruction, neuronal	XLR
		?FG syndrome 2	XL
		Otopalatodigital syndrome, type I	XLD
FLNC	102565	Cardiomyopathy, familial restrictive 5	AD
		Myopathy, myofibrillar, 5	AD
		Myopathy, distal, 4	AD
		Cardiomyopathy, familial hypertrophic, 26	AD
FLVCR1	609144	Ataxia, posterior column, with retinitis pigmentosa	AR
FLVCR2	610865	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	AR
FMN2	606373	Mental retardation, autosomal recessive 47	AR
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency	AR
FOXC1	601090	Anterior segment dysgenesis 3, multiple subtypes	AD
		Axenfeld-Rieger syndrome, type 3	AD
FOXG1	164874	Rett syndrome, congenital variant	AD

FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1	AD, AR
		Blepharophimosis, epicanthus inversus, and ptosis, type 2	AD, AR
		Premature ovarian failure 3	AD
FOXP1	605515	Mental retardation with language impairment and with or without autistic features	AD
FOXP2	605317	Speech-language disorder-1	AD
FOXRED1	613622	Mitochondrial complex I deficiency, nuclear type 19	AR
FRMD7	300628	Nystagmus, infantile periodic alternating, X-linked	XL
		Nystagmus 1, congenital, X-linked	XL
FRMPD4	300838	Mental retardation, X-linked 104	XL
FRRS1L	604574	Developmental and epileptic encephalopathy 37	AR
FTL	134790	Neurodegeneration with brain iron accumulation 3	AD
		Hyperferritinemia-cataract syndrome	AD
		L-ferritin deficiency, dominant and recessive	AD, AR
FTO	610966	Growth retardation, developmental delay, facial dysmorphism	AR
		Obesity, susceptibility to, BMIQ14	AR
FTSJ1	300499	Mental retardation, X-linked 9/44	XLR
FUCA1	612280	Fucosidosis	AR
FUS	137070	Essential tremor, hereditary, 4	AD
		Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	-

FUT8	602589	Congenital disorder of glycosylation with defective fucosylation 1	AR
FXN	606829	Friedreich ataxia	AR
		Friedreich ataxia with retained reflexes	AR
FXR1	600819	?Myopathy, congenital, with respiratory insufficiency and bone fractures	AR
		?Myopathy, congenital proximal, with minicore lesions	AR
FXYD2	601814	Hypomagnesemia 2, renal	AD
G6PD	305900	Resistance to malaria due to G6PD deficiency	-
		Hemolytic anemia, G6PD deficient (favism)	XLD
GAA	606800	Glycogen storage disease II	AR
GABBR2	607340	Nicotine dependence, susceptibility to	-
		Neurodevelopmental disorder with poor language and loss of hand skills	AD
		Developmental and epileptic encephalopathy 59	AD
		Nicotine dependence, protection against	-
GABRA1	137160	Developmental and epileptic encephalopathy 19	AD
		Epilepsy, childhood absence, susceptibility to, 4	-
		Epilepsy, juvenile myoclonic, susceptibility to, 5	-
GABRA2	137140	Alcohol dependence, susceptibility to	MF
		Developmental and epileptic encephalopathy 78	AD
GABRA5	137142	Developmental and epileptic encephalopathy 79	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	AD
		Epilepsy, juvenile myoclonic, susceptibility to	AD
		Epilepsy, idiopathic generalized, 10	AD
GABRG2	137164	Developmental and epileptic encephalopathy 74	AD
		Epilepsy, generalized, with febrile seizures plus, type 3	AD
		Febrile seizures, familial, 8	AD
GAD1	605363	?Cerebral palsy, spastic quadriplegic, 1	AR
		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
GARS1	600287	Spinal muscular atrophy, infantile, James type	AD
		Charcot-Marie-Tooth disease, type 2D	AD

		Neuronopathy, distal hereditary motor, type VA	AD
GATAD2B	614998	GAND syndrome	AD
GATM	602360	Cerebral creatine deficiency syndrome 3	AR
		Fanconi renotubular syndrome 1	AD
GBA	606463	Lewy body dementia, susceptibility to	AD
		Gaucher disease, type IIIC	AR
		Parkinson disease, late-onset, susceptibility to	AD, MF
		Gaucher disease, type II	AR
		Gaucher disease, type III	AR
		Gaucher disease, perinatal lethal	AR
		Gaucher disease, type I	AR
GBA2	609471	Spastic paraplegia 46, autosomal recessive	AR
GBE1	607839	Glycogen storage disease IV	AR
		Polyglucosan body disease, adult form	AR
GCDH	608801	Glutaricaciduria, type I	AR
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	AD, AR
		Hyperphenylalaninemia, BH4-deficient, B	AR
GCK	138079	Diabetes mellitus, permanent neonatal 1	AR
		Diabetes mellitus, noninsulin-dependent, late onset	AD

		Hyperinsulinemic hypoglycemia, familial, 3	AD
		MODY, type II	AD
GCSH	238330	?Glycine encephalopathy	AR
GDAP1	606598	Charcot-Marie-Tooth disease, axonal, type 2K	AD, AR
		Charcot-Marie-Tooth disease, type 4A	AR
		Charcot-Marie-Tooth disease, axonal, with vocal cord paresis	AR
		Charcot-Marie-Tooth disease, recessive intermediate, A	AR
GDI1	300104	Mental retardation, X-linked 41	XLD
GDNF	600837	Pheochromocytoma, modifier of	AD
		Central hypoventilation syndrome	AD
		Hirschsprung disease, susceptibility to, 3	AD
GFAP	137780	Alexander disease	AD
GFER	600924	Myopathy, mitochondrial 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
GIGYF2	612003	Parkinson disease 11	-
GJA1	121014	Oculodentodigital dysplasia, autosomal recessive	AR
		Atrioventricular septal defect 3	AD

		Syndactyly, type III	AD
		Craniometaphyseal dysplasia, autosomal recessive	AR
		Palmoplantar keratoderma with congenital alopecia	AD
		Oculodentodigital dysplasia	AD
		Hypoplastic left heart syndrome 1	AR
		Erythrokeratodermia variabilis et progressiva 3	AD
GJB1	304040	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	XLD
GJB3	603324	Deafness, autosomal dominant 2B	AD
		Erythrokeratodermia variabilis et progressiva 1	AD, AR
		Deafness, digenic, GJB2/GJB3	AR, DD
GJC2	608803	Leukodystrophy, hypomyelinating, 2	AR
		Spastic paraplegia 44, autosomal recessive	AR
		Lymphatic malformation 3	AD
GK	300474	Glycerol kinase deficiency	XLR
GLA	300644	Fabry disease	XL
		Fabry disease, cardiac variant	XL
GLB1	611458	GM1-gangliosidosis, type II	AR
		GM1-gangliosidosis, type I	AR
		Mucopolysaccharidosis type IVB (Morquio)	AR

		GM1-gangliosidosis, type III	AR
GLDC	238300	Glycine encephalopathy	AR
GLDN	608603	Lethal congenital contracture syndrome 11	AR
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease	AR
		Lethal congenital contracture syndrome 1	AR
GLI2	165230	Holoprosencephaly 9	AD
		Culler-Jones syndrome	AD
GLI3	165240	Pallister-Hall syndrome	AD
		Polydactyly, preaxial, type IV	AD
		Polydactyly, postaxial, types A1 and B	AD
		Greig cephalopolysyndactyly syndrome	AD
GLRA1	138491	Hyperekplexia 1	AD, AR
GLRB	138492	Hyperekplexia 2	AR
GLRX5	609588	Spasticity, childhood-onset, with hyperglycinemia	AR
		Anemia, sideroblastic, 3, pyridoxine-refractory	AR
GLUD1	138130	Hyperinsulinism-hyperammonemia syndrome	AD
GLUL	138290	Glutamine deficiency, congenital	AR
GLYCTK	610516	D-glyceric aciduria	AR
GM2A	613109	GM2-gangliosidosis, AB variant	AR

GMPPA	615495	Alacrima, achalasia, and mental retardation syndrome	AR
GMPPB	615320	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	AR
GNAL	139312	Dystonia 25	AD
GNAO1	139311	Developmental and epileptic encephalopathy 17	AD
		Neurodevelopmental disorder with involuntary movements	AD
GNAQ	600998	Capillary malformations, congenital, 1, somatic, mosaic	-
		Sturge-Weber syndrome, somatic, mosaic	-
GNAS	139320	Pseudohypoparathyroidism Ic	AD
		Osseous heteroplasia, progressive	AD
		Pseudopseudohypoparathyroidism	AD
		Pseudohypoparathyroidism Ia	AD
		Pseudohypoparathyroidism Ib	AD
		Pituitary adenoma 3, multiple types, somatic	-
		ACTH-independent macronodular adrenal hyperplasia	SM
		McCune-Albright syndrome, somatic, mosaic	-
GNB1	139380	Leukemia, acute lymphoblastic, somatic	-
		Myelodysplastic syndrome, somatic	-

		Mental retardation, autosomal dominant 42	AD
GNB4	610863	Charcot-Marie-Tooth disease, dominant intermediate F	AD
GNB5	604447	Intellectual developmental disorder with cardiac arrhythmia	AR
		Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	AR
GNE	603824	Nonaka myopathy	AR
		Sialuria	AD
GNPAT	602744	Rhizomelic chondrodysplasia punctata, type 2	AR
GNPTAB	607840	Mucolipidosis III alpha/beta	AR
		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	XLR
		Wilms tumor, somatic	-
GPC4	300168	Keipert syndrome	XLR
GPC6	604404	Omodysplasia 1	AR
GPHN	603930	Molybdenum cofactor deficiency C	AR

GPI	172400	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency	AR
GPT2	138210	Neurodevelopmental disorder with microcephaly and spastic paraplegia	AR
GPX1	138320	Hemolytic anemia due to glutathione peroxidase deficiency	AR
GRHPR	604296	Hyperoxaluria, primary, type II	AR
GRIA1	138248	Intellectual developmental disorder, autosomal dominant 67	-
		Intellectual developmental disorder, autosomal recessive 76	-
GRIA2	138247	Neurodevelopmental disorder with language impairment and behavioral abnormalities	AD
GRIA3	305915	Intellectual developmental disorder, X-linked, syndromic, Wu type	XLR
GRIA4	138246	Neurodevelopmental disorder with or without seizures and gait abnormalities	AD
GRID2	602368	Spinocerebellar ataxia, autosomal recessive 18	AR
GRIK2	138244	Neurodevelopmental disorder with impaired language and ataxia and with or without seizures	AD
		Mental retardation, autosomal recessive, 6	AR
GRIN1	138249	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive	AR
		Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant	AD
		Developmental and epileptic encephalopathy 101	AR
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without mental retardation	AD
GRIN2B	138252	Mental retardation, autosomal dominant 6	AD
		Developmental and epileptic encephalopathy 27	AD

GRIN2D	602717	Developmental and epileptic encephalopathy 46	AD
GRIP1	604597	Fraser syndrome 3	AR
GRM1	604473	Spinocerebellar ataxia 44	AD
		Spinocerebellar ataxia, autosomal recessive 13	AR
GRN	138945	Ceroid lipofuscinosis, neuronal, 11	AR
		Frontotemporal lobar degeneration with ubiquitin-positive inclusions	AD
		Aphasia, primary progressive	AD
GSN	137350	Amyloidosis, Finnish type	AD
GSR	138300	Hemolytic anemia due to glutathione reductase deficiency	AR
GSS	601002	Glutathione synthetase deficiency	AR
		Hemolytic anemia due to glutathione synthetase deficiency	AR
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
GYG1	603942	Polyglucosan body myopathy 2	AR
		?Glycogen storage disease XV	AR
GYS1	138570	Glycogen storage disease 0, muscle	AR
HACE1	610876	Spastic paraplegia and psychomotor retardation with or without seizures	AR

HADH	601609	3-hydroxyacyl-CoA dehydrogenase deficiency	AR
		Hyperinsulinemic hypoglycemia, familial, 4	AR
HADHA	600890	HELLP syndrome, maternal, of pregnancy	AR
		LCHAD deficiency	AR
		Fatty liver, acute, of pregnancy	AR
		Mitochondrial trifunctional protein deficiency	AR
HADHB	143450	Trifunctional protein deficiency	AR
HAMP	606464	Hemochromatosis, type 2B	AR
HARS2	600783	Perrault syndrome 2	AR
HAX1	605998	Neutropenia, severe congenital 3, autosomal recessive	AR
HBB	141900	Erythrocytosis 6	AD
		Methemoglobinemia, beta type	AD
		Delta-beta thalassemia	AD
		Thalassemia-beta, dominant inclusion-body	AD
		Sickle cell anemia	AR
		Hereditary persistence of fetal hemoglobin	AD
		Malaria, resistance to	-
		Thalassemia, beta	-
		Heinz body anemia	AD

HCCS	300056	Linear skin defects with multiple congenital anomalies 1	XLD
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XLR
HCN1	602780	Generalized epilepsy with febrile seizures plus, type 10	AD
		Developmental and epileptic encephalopathy 24	AD
HDAC4	605314	Neurodevelopmental disorder with central hypotonia and dysmorphic facies	AD
HDAC8	300269	Cornelia de Lange syndrome 5	XLD
HECW2	617245	Neurodevelopmental disorder with hypotonia, seizures, and absent language	AD
HEPACAM	611642	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation	AD
		Megalencephalic leukoencephalopathy with subcortical cysts 2A	AR
HERC2	605837	[Skin/hair/eye pigmentation 1, blond/brown hair]	AR
		[Skin/hair/eye pigmentation 1, blue/nonblue eyes]	AR
		Mental retardation, autosomal recessive 38	AR
HESX1	601802	Septooptic dysplasia	AD, AR
		Pituitary hormone deficiency, combined, 5	AD, AR
		Growth hormone deficiency with pituitary anomalies	AD, AR
HEXA	606869	Tay-Sachs disease	AR
		[Hex A pseudodeficiency]	AR
		GM2-gangliosidosis, several forms	AR
HEXB	606873	Sandhoff disease, infantile, juvenile, and adult forms	AR

HGSNAT	610453	Mucopolysaccharidosis type IIIC (Sanfilippo C)	AR
		Retinitis pigmentosa 73	AR
HIBCH	610690	3-hydroxyisobutryl-CoA hydrolase deficiency	AR
HIKESHI	614908	Leukodystrophy, hypomyelinating, 13	AR
HINT1	601314	Neuromyotonia and axonal neuropathy, autosomal recessive	AR
HIVEP2	143054	Mental retardation, autosomal dominant 43	AD
HK1	142600	Retinitis pigmentosa 79	AD
		Hemolytic anemia due to hexokinase deficiency	AR
		Neurodevelopmental disorder with visual defects and brain anomalies	AD
		Neuropathy, hereditary motor and sensory, Russe type	AR
HLCS	609018	Holocarboxylase synthetase deficiency	AR
HMBS	609806	Porphyria, acute intermittent	AD
		Porphyria, acute intermittent, nonerythroid variant	AD
HMGCL	613898	HMG-CoA lyase deficiency	AR
HMGCS2	600234	HMG-CoA synthase-2 deficiency	AR
HNMT	605238	Asthma, susceptibility to	AD
		Mental retardation, autosomal recessive 51	AR
HNRNPA1	164017	Amyotrophic lateral sclerosis 20	AD
		?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3	AD

HNRNPA2B1	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	AD
HNRNPDL	607137	Muscular dystrophy, limb-girdle, autosomal dominant 3	AD
HNRNPH2	300610	Mental retardation, X-linked, syndromic, Bain type	XLD
HNRNPU	602869	Developmental and epileptic encephalopathy 54	AD
HOGA1	613597	Hyperoxaluria, primary, type III	AR
HOXA1	142955	Bosley-Salih-Alorainy syndrome	AR
		Athabaskan brainstem dysgenesis syndrome	AR
HOXD10	142984	Vertical talus, congenital	AD
		Charcot-Marie-Tooth disease, foot deformity of	AD
HPCA	142622	Dystonia 2, torsion, autosomal recessive	AR
HPD	609695	Hawkinsinuria	AD
		Tyrosinemia, type III	AR
HPRT1	308000	Hyperuricemia, HRPT-related	XLR
		Lesch-Nyhan syndrome	XLR
HPS1	604982	Hermansky-Pudlak syndrome 1	AR
HPS4	606682	Hermansky-Pudlak syndrome 4	AR
HPS5	607521	Hermansky-Pudlak syndrome 5	AR
HPS6	607522	Hermansky-Pudlak syndrome 6	AR
HRAS	190020	Thyroid carcinoma, follicular, somatic	-

		Spitz nevus or nevus spilus, somatic	-
		Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	-
		Bladder cancer, somatic	-
		Costello syndrome	AD
		Nevus sebaceous or woolly hair nevus, somatic	-
		Congenital myopathy with excess of muscle spindles	AD
HSD11B1	600713	Cortisone reductase deficiency 2	AD
HSD17B10	300256	HSD10 mitochondrial disease	XLD
HSD17B4	601860	Perrault syndrome 1	AR
		D-bifunctional protein deficiency	AR
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HSPA9	600548	Even-plus syndrome	AR
		Anemia, sideroblastic, 4	AD
HSPB1	602195	Neuronopathy, distal hereditary motor, type IIB	AD
		Charcot-Marie-Tooth disease, axonal, type 2F	AD
HSPB3	604624	?Neuronopathy, distal hereditary motor, type IIC	AD
HSPB8	608014	Neuronopathy, distal hereditary motor, type IIA	AD
		Charcot-Marie-Tooth disease, axonal, type 2L	AD
HSPD1	118190	Leukodystrophy, hypomyelinating, 4	AR

		Spastic paraplegia 13, autosomal dominant	AD
HSPG2	142461	Schwartz-Jampel syndrome, type 1	AR
		Dyssegmental dysplasia, Silverman-Handmaker type	AR
HTRA1	602194	Macular degeneration, age-related, 7	-
		CARASIL syndrome	AR
		Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2	AD
		Macular degeneration, age-related, neovascular type	-
HTRA2	606441	Parkinson disease 13	-
		3-methylglutaconic aciduria, type VIII	AR
HUWE1	300697	Mental retardation, X-linked syndromic, Turner type	XL
HYAL1	607071	?Mucopolysaccharidosis type IX	AR
HYDIN	610812	Ciliary dyskinesia, primary, 5	AR
IARS2	612801	?Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia	AR
IBA57	615316	?Spastic paraplegia 74, autosomal recessive	AR
		Multiple mitochondrial dysfunctions syndrome 3	AR
IDH2	147650	D-2-hydroxyglutaric aciduria 2	-
IDH3B	604526	Retinitis pigmentosa 46	AR
IDS	300823	Mucopolysaccharidosis II	XLR
IDUA	252800	Mucopolysaccharidosis Is	AR

		Mucopolysaccharidosis Ih/s	AR
		Mucopolysaccharidosis Ih	AR
IER3IP1	609382	Microcephaly, epilepsy, and diabetes syndrome	AR
IFIH1	606951	Aicardi-Goutieres syndrome 7	AD
		Singleton-Merten syndrome 1	AD
		Immunodeficiency 95	AR
IFT140	614620	Retinitis pigmentosa 80	AR
		Short-rib thoracic dysplasia 9 with or without polydactyly	AR
IFT172	607386	Retinitis pigmentosa 71	AR
		Bardet-Biedl syndrome 20	AR
		Short-rib thoracic dysplasia 10 with or without polydactyly	AR
IFT27	615870	?Bardet-Biedl syndrome 19	AR
IGBP1	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia	XLR
IGF1	147440	Growth retardation with deafness and mental retardation due to IGF1 deficiency	AR
IGF1R	147370	Insulin-like growth factor I, resistance to	AD, AR
IGHMBP2	600502	Neuronopathy, distal hereditary motor, type VI	AR
		Charcot-Marie-Tooth disease, axonal, type 2S	AR
IL1RAPL1	300206	Mental retardation, X-linked 21/34	XLR
IMPA1	602064	Mental retardation, autosomal recessive 59	AR

INF2	610982	Glomerulosclerosis, focal segmental, 5	-
		Charcot-Marie-Tooth disease, dominant intermediate E	AD
INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis	AR
		Joubert syndrome 1	AR
INVS	243305	Nephronophthisis 2, infantile	AR
IQSEC2	300522	Mental retardation, X-linked 1/78	XLD
IRF2BPL	611720	Neurodevelopmental disorder with regression, abnormal movements, loss of speech, and seizures	AD
IRX5	606195	Hamamy syndrome	AR
ISCA1	611006	Multiple mitochondrial dysfunctions syndrome 5	AR
ISCA2	615317	Multiple mitochondrial dysfunctions syndrome 4	AR
ISCU	611911	Myopathy with lactic acidosis, hereditary	AR
ITGA7	600536	Muscular dystrophy, congenital, due to ITGA7 deficiency	AR
ITGB3	173470	Glanzmann thrombasthenia 2	-
		Glanzmann thrombasthenia	AR
		Bleeding disorder, platelet-type, 16, autosomal dominant	AD
		Myocardial infarction, susceptibility to	-
		Bleeding disorder, platelet-type, 24, autosomal dominant	-
ITM2B	603904	Dementia, familial British	AD
		?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities	AD

		Dementia, familial Danish	AD
ITPA	147520	Developmental and epileptic encephalopathy 35	AR
		[Inosine triphosphatase deficiency]	-
ITPR1	147265	Spinocerebellar ataxia 15	AD
		Spinocerebellar ataxia 29, congenital nonprogressive	AD
		Gillespie syndrome	AD, AR
IVD	607036	Isovaleric acidemia	AR
JAG1	601920	Alagille syndrome 1	AD
		Charcot-Marie-Tooth disease, axonal, type 2HH	AD
		?Deafness, congenital heart defects, and posterior embryotoxon	AD
		Tetralogy of Fallot	AD
JAM2	606870	Basal ganglia calcification, idiopathic, 8, autosomal recessive	AR
JAM3	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts	AR
KANK1	607704	Cerebral palsy, spastic quadriplegic, 2	-
KANSL1	612452	Koolen-De Vries syndrome	AD
KARS1	601421	?Charcot-Marie-Tooth disease, recessive intermediate, B	AR
		Deafness, congenital, and adult-onset progressive leukoencephalopathy	AR
		Leukoencephalopathy, progressive, infantile-onset, with or without deafness	AR
		Deafness, autosomal recessive 89	AR

KAT6A	601408	Arboleda-Tham syndrome	AD
KAT6B	605880	SBBYSS syndrome	AD
		Genitopatellar syndrome	AD
KAT8	609912	Li-Ghorgani-Weisz-Hubshman syndrome	AD
KATNB1	602703	Lissencephaly 6, with microcephaly	AR
KBTBD13	613727	Nemaline myopathy 6, autosomal dominant	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
KCNC3	176264	Spinocerebellar ataxia 13	AD
KCND3	605411	Brugada syndrome 9	AD
		Spinocerebellar ataxia 19	AD
KCNE3	604433	?Brugada syndrome 6	-
KCNH1	603305	Zimmermann-Laband syndrome 1	AD
		Temple-Baraitser syndrome	AD
KCNJ1	600359	Bartter syndrome, type 2	AR
KCNJ10	602208	Enlarged vestibular aqueduct, digenic	AR
		SESAME syndrome	AR

KCNJ2	600681	Short QT syndrome 3	-
		Atrial fibrillation, familial, 9	AD
		Andersen syndrome	AD
KCNK18	613655	Migraine, with or without aura, susceptibility to, 13	AD
KCNK4	605720	Facial dysmorphism, hypertrichosis, epilepsy, intellectual/developmental delay, and gingival overgrowth syndrome	AD
KCNK9	605874	Birk-Barel syndrome	-
KCNMA1	600150	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy	AD
		Epilepsy, idiopathic generalized, susceptibility to, 16	AD
		Liang-Wang syndrome	AD
		Cerebellar atrophy, developmental delay, and seizures	AR
KCNQ2	602235	Seizures, benign neonatal, 1	AD
		Developmental and epileptic encephalopathy 7	AD
		Myokymia	AD
KCNQ3	602232	Seizures, benign neonatal, 2	AD
KCNQ5	607357	Mental retardation, autosomal dominant 46	AD
KCNT1	608167	Developmental and epileptic encephalopathy 14	AD
		Epilepsy nocturnal frontal lobe, 5	AD
KCNT2	610044	?Developmental and epileptic encephalopathy 57	AD
KCTD17	616386	Dystonia 26, myoclonic	AD

KCTD7	611725	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions	AR
KDM4B	609765	Intellectual developmental disorder, autosomal dominant 65	AD
KDM5B	605393	Mental retardation, autosomal recessive 65	AR
KDM5C	314690	Mental retardation, X-linked, syndromic, Claes-Jensen type	XLR
KDM6A	300128	Kabuki syndrome 2	XLD
KIAA1109	611565	Alkuraya-Kucinskas syndrome	AR
KIDINS220	615759	Ventriculomegaly and arthrogryposis	AR
		Spastic paraplegia, intellectual disability, nystagmus, and obesity	AD
KIF11	148760	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation	AD
KIF14	611279	?Meckel syndrome 12	AR
		Microcephaly 20, primary, autosomal recessive	AR
KIF1A	601255	NESCAV syndrome	AD
		Spastic paraplegia 30, autosomal recessive	AD, AR
		Neuropathy, hereditary sensory, type IIC	AR
		Spastic paraplegia 30, autosomal dominant	AD, AR
KIF1B	605995	?Charcot-Marie-Tooth disease, type 2A1	AD
		Pheochromocytoma	AD
		Neuroblastoma, susceptibility to, 1	AD, SM
KIF1C	603060	Spastic ataxia 2, autosomal recessive	AR

KIF21A	608283	Fibrosis of extraocular muscles, congenital, 3B	AD
		Fibrosis of extraocular muscles, congenital, 1	AD
KIF2A	602591	Cortical dysplasia, complex, with other brain malformations 3	AD
KIF5A	602821	Amyotrophic lateral sclerosis, susceptibility to, 25	AD
		Myoclonus, intractable, neonatal	AD
		Spastic paraplegia 10, autosomal dominant	AD
KIF5C	604593	Cortical dysplasia, complex, with other brain malformations 2	AD
KIF7	611254	Joubert syndrome 12	AR
		?Hydrolethalus syndrome 2	AR
		?Al-Gazali-Bakalinova syndrome	AR
		Acrocallosal syndrome	AR
KIFBP	609367	Goldberg-Shprintzen megacolon syndrome	AR
KLHL40	615340	Nemaline myopathy 8, autosomal recessive	AR
KLHL41	607701	Nemaline myopathy 9	AR
KLHL7	611119	PERCHING syndrome	AR
		Retinitis pigmentosa 42	AD
KMT2A	159555	Wiedemann-Steiner syndrome	AD
KMT2B	606834	Dystonia 28, childhood-onset	AD
		Intellectual developmental disorder, autosomal dominant 68	-

KMT2C	606833	Kleefstra syndrome 2	AD
KMT2D	602113	Kabuki syndrome 1	AD
KMT2E	608444	O'Donnell-Luria-Rodan syndrome	AD
KMT5B	610881	Mental retardation, autosomal dominant 51	AD
KNL1	609173	Microcephaly 4, primary, autosomal recessive	AR
KPTN	615620	Mental retardation, autosomal recessive 41	AR
KRAS	190070	Arteriovenous malformation of the brain, somatic	-
		Gastric cancer, somatic	-
		Oculoectodermal syndrome, somatic	-
		RAS-associated autoimmune leukoproliferative disorder	AD
		Gastric cancer, somatic	-
		Pancreatic carcinoma, somatic	-
		Lung cancer, somatic	-
		Cardiofaciocutaneous syndrome 2	AD
		Bladder cancer, somatic	-
		Leukemia, acute myeloid, somatic	-
		Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic	-
		Breast cancer, somatic	-
		Noonan syndrome 3	AD

KRIT1	604214	Cavernous malformations of CNS and retina	AD
		Cerebral cavernous malformations-1	AD
		Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations	AD
KRT5	148040	Epidermolysis bullosa simplex-MP	AD
		Epidermolysis bullosa simplex, Koebner type	AD
		Epidermolysis bullosa simplex, recessive 1	AR
		Epidermolysis bullosa simplex, Weber-Cockayne type	AD
		Epidermolysis bullosa simplex 2B, generalized intermediate	AD
		Epidermolysis bullosa simplex 2A, generalized severe	AD
		Epidermolysis bullosa simplex-MCR	-
		Dowling-Degos disease 1	AD
		Epidermolysis bullosa simplex 2C, localized	AD
		Epidermolysis bullosa simplex, Dowling-Meara type	AD
		Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive	AR
KRT8	148060	Cirrhosis, cryptogenic	AR
		Cirrhosis, noncryptogenic, susceptibility to	AR
KY	605739	Myopathy, myofibrillar, 7	AR
L1CAM	308840	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction	XLR
		CRASH syndrome	XLR

		Corpus callosum, partial agenesis of	XLR
		Hydrocephalus with Hirschsprung disease	XLR
		MASA syndrome	XLR
		Hydrocephalus due to aqueductal stenosis	XLR
L2HGDH	609584	L-2-hydroxyglutaric aciduria	AR
LAMA1	150320	Poretti-Boltshauser syndrome	AR
LAMA2	156225	Muscular dystrophy, congenital, merosin deficient or partially deficient	AR
		Muscular dystrophy, limb-girdle, autosomal recessive 23	AR
LAMB1	150240	Lissencephaly 5	AR
LAMB2	150325	Nephrotic syndrome, type 5, with or without ocular abnormalities	-
		Pierson syndrome	AR
LAMC3	604349	Cortical malformations, occipital	AR
LAMP2	309060	Danon disease	XLD
LARGE1	603590	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR
LARS2	604544	?Hydrops, lactic acidosis, and sideroblastic anemia	AR
		Perrault syndrome 4	AR
LAT	602354	Immunodeficiency 52	AR
LBR	600024	?Reynolds syndrome	AD

		Greenberg skeletal dysplasia	AR
		Pelger-Huet anomaly	AD
		Pelger-Huet anomaly with mild skeletal anomalies	-
LDB3	605906	Cardiomyopathy, hypertrophic, 24	AD
		Myopathy, myofibrillar, 4	AD
		Cardiomyopathy, dilated, 1C, with or without LVNC	AD
		Left ventricular noncompaction 3	AD
LDHA	150000	Glycogen storage disease XI	AR
LEP	164160	Obesity, morbid, due to leptin deficiency	AR
LGI1	604619	Epilepsy, familial temporal lobe, 1	AD
LGI4	608303	Arthrogryposis multiplex congenita 1, neurogenic, with myelin defect	AR
LHX3	600577	Pituitary hormone deficiency, combined, 3	AR
LHX4	602146	Pituitary hormone deficiency, combined, 4	AD
LIAS	607031	Hyperglycinemia, lactic acidosis, and seizures	AR
LIMS2	607908	?Muscular dystrophy, autosomal recessive, with cardiomyopathy and triangular tongue	AR
LINS1	610350	Mental retardation, autosomal recessive 27	AR
LIPA	613497	Wolman disease	AR
		Cholesteryl ester storage disease	AR
LIPT1	610284	Lipoyltransferase 1 deficiency	AR

LIPT2	617659	Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	AR
LITAF	603795	Charcot-Marie-Tooth disease, type 1C	AD
LMAN2L	609552	?Mental retardation, autosomal recessive, 52	AR
		?Intellectual developmental disorder, autosomal dominant 69	-
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	AR
LMNA	150330	Restrictive dermopathy 2	-
		Malouf syndrome	AD
		Emery-Dreifuss muscular dystrophy 3, autosomal recessive	AR
		Emery-Dreifuss muscular dystrophy 2, autosomal dominant	AD
		Hutchinson-Gilford progeria	AD
		Muscular dystrophy, congenital	AD
		Restrictive dermopathy, lethal	AR
		Lipodystrophy, familial partial, type 2	AD
		Charcot-Marie-Tooth disease, type 2B1	AR
		Mandibuloacral dysplasia	AR
		Cardiomyopathy, dilated, 1A	AD
		Heart-hand syndrome, Slovenian type	AD
LMNB1	150340	Leukodystrophy, adult-onset, autosomal dominant	AD
		Microcephaly 26, primary, autosomal dominant	AD

LMOD3	616112	Nemaline myopathy 10	AR
LMX1B	602575	Nail-patella syndrome	AD
		Focal segmental glomerulosclerosis 10	AD
LONP1	605490	CODAS syndrome	AR
LPIN1	605518	Myoglobinuria, acute recurrent, autosomal recessive	AR
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity	AR
LRP1	107770	?Keratosis pilaris atrophicans	AR
LRP2	600073	Donnai-Barrow syndrome	AR
LRP4	604270	?Myasthenic syndrome, congenital, 17	AR
		Cenani-Lenz syndactyly syndrome	AR
		Sclerosteosis 2	AD, AR
LRPPRC	607544	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	AR
LRRK2	609007	Parkinson disease 8	AD
LRSAM1	610933	Charcot-Marie-Tooth disease, axonal, type 2P	AD, AR
LYRM7	615831	Mitochondrial complex III deficiency, nuclear type 8	AR
LYST	606897	Chediak-Higashi syndrome	AR
LZTFL1	606568	Bardet-Biedl syndrome 17	AR
LZTR1	600574	Noonan syndrome 2	AR
		Schwannomatosis-2, susceptibility to	AD

		Noonan syndrome 10	AD
MACF1	608271	Lissencephaly 9 with complex brainstem malformation	AD
MAF	177075	Ayme-Gripp syndrome	AD
		Cataract 21, multiple types	AD
MAG	159460	Spastic paraplegia 75, autosomal recessive	AR
MAGEL2	605283	Schaaf-Yang syndrome	AD
MAGI2	606382	Nephrotic syndrome, type 15	AR
MAGT1	300715	Congenital disorder of glycosylation, type Icc	XLR
		Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	XLR
MAMLD1	300120	Hypospadias 2, X-linked	XLR
MAN1B1	604346	Rafiq syndrome	AR
MAN2B1	609458	Mannosidosis, alpha-, types I and II	AR
MANBA	609489	Mannosidosis, beta	AR
MAOA	309850	Antisocial behavior	XLR
		Brunner syndrome	XLR
MAP2K1	176872	Cardiofaciocutaneous syndrome 3	AD
		Melorheostosis, isolated, somatic mosaic	-
MAP2K2	601263	Cardiofaciocutaneous syndrome 4	AD
MAPT	157140	Parkinson disease, susceptibility to	AD, MF

		Pick disease	AD
		Dementia, frontotemporal, with or without parkinsonism	AD
		Supranuclear palsy, progressive	AD
		Supranuclear palsy, progressive atypical	AR
MARS1	156560	Charcot-Marie-Tooth disease, axonal, type 2U	AD
		?Trichothiodystrophy 9, nonphotosensitive	AR
		Interstitial lung and liver disease	AR
MARS2	609728	Spastic ataxia 3, autosomal recessive	AR
		?Combined oxidative phosphorylation deficiency 25	AR
MASP1	600521	3MC syndrome 1	AR
MATR3	164015	Amyotrophic lateral sclerosis 21	AD
MBD5	611472	Mental retardation, autosomal dominant 1	AD
MBOAT7	606048	Mental retardation, autosomal recessive 57	AR
MBTPS2	300294	Osteogenesis imperfecta, type XIX	XLR
		?Olmsted syndrome, X-linked	XLR
		Keratosis follicularis spinulosa decalvans, X-linked	XLR
		IFAP syndrome with or without BRESHECK syndrome	XLR
MCCC1	609010	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR
MCCC2	609014	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR

MCEE	608419	Methylmalonyl-CoA epimerase deficiency	AR
MCM4	602638	Immunodeficiency 54	AR
MCM6	601806	Lactase persistence/nonpersistence	AD
MCOLN1	605248	Mucolipidosis IV	AR
MCPH1	607117	Microcephaly 1, primary, autosomal recessive	AR
MDH2	154100	Developmental and epileptic encephalopathy 51	AR
MECP2	300005	Rett syndrome, preserved speech variant	XLD
		Encephalopathy, neonatal severe	XLR
		Mental retardation, X-linked, syndromic 13	XLR
		Rett syndrome	XLD
		Mental retardation, X-linked syndromic, Lubs type	XLR
		Rett syndrome, atypical	XLD
		Autism susceptibility, X-linked 3	XL
MECR	608205	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities	AR
MED12	300188	Opitz-Kaveggia syndrome	XLR
		Lujan-Fryns syndrome	XLR
		Ohdo syndrome, X-linked	XLR
		Hardikar syndrome	XLD
MED13	603808	Intellectual developmental disorder 61	AD

MED13L	608771	Transposition of the great arteries, dextro-looped 1	AD
		Mental retardation and distinctive facial features with or without cardiac defects	AD
MED17	603810	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR
MED23	605042	Mental retardation, autosomal recessive 18	AR
MED25	610197	Basel-Vanagait-Smirin-Yosef syndrome	AR
MEF2C	600662	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations	AD
		Chromosome 5q14.3 deletion syndrome	AD
MEGF10	612453	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	AR
		Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant	AR
MEIS2	601740	Cleft palate, cardiac defects, and mental retardation	AD
METTL23	615262	Mental retardation, autosomal recessive 44	AR
MFF	614785	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	AR
MFN2	608507	Hereditary motor and sensory neuropathy VIA	AD
		Charcot-Marie-Tooth disease, axonal, type 2A2B	AR
		Charcot-Marie-Tooth disease, axonal, type 2A2A	AD
MFRP	606227	Microphthalmia, isolated 5	AR
		Nanophthalmos 2	-
MFSD2A	614397	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain abnormalities	AR
MFSD8	611124	Ceroid lipofuscinosis, neuronal, 7	AR

		Macular dystrophy with central cone involvement	AR
MGAT2	602616	Congenital disorder of glycosylation, type IIa	AR
MGME1	615076	Mitochondrial DNA depletion syndrome 11	AR
MIB1	608677	Left ventricular noncompaction 7	AD
MICU1	605084	Myopathy with extrapyramidal signs	AR
MID1	300552	Opitz GBBB syndrome, type I	XLR
MIPEP	602241	Combined oxidative phosphorylation deficiency 31	AR
MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8	-
		Waardenburg syndrome, type 2A	AD
		Waardenburg syndrome/ocular albinism, digenic	-
		Tietz albinism-deafness syndrome	AD
		COMMAD syndrome	AR
MKKS	604896	McKusick-Kaufman syndrome	AR
		Bardet-Biedl syndrome 6	AR
MKS1	609883	Bardet-Biedl syndrome 13	AR
		Joubert syndrome 28	AR
		Meckel syndrome 1	AR
MLC1	605908	Megalencephalic leukoencephalopathy with subcortical cysts	AR
MLPH	606526	Griscelli syndrome, type 3	AR

MLYCD	606761	Malonyl-CoA decarboxylase deficiency	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	AR
		Methylmalonic aciduria, cblD type, variant 2	AR
		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
MPC1	614738	Mitochondrial pyruvate carrier deficiency	AR
MPDU1	604041	Congenital disorder of glycosylation, type If	AR
MPDZ	603785	Hydrocephalus, congenital, 2, with or without brain or eye anomalies	AR
MPI	154550	Congenital disorder of glycosylation, type Ib	AR
MPV17	137960	Charcot-Marie-Tooth disease, axonal, type 2EE	AR
		Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	AR
MPZ	159440	Charcot-Marie-Tooth disease, type 2I	AD

		Dejerine-Sottas disease	AD, AR
		Hypomyelinating neuropathy, congenital, 2	AD
		Charcot-Marie-Tooth disease, type 2J	AD
		Charcot-Marie-Tooth disease, dominant intermediate D	AD
		Charcot-Marie-Tooth disease, type 1B	AD
		Roussy-Levy syndrome	AD
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
MRPS2	611971	Combined oxidative phosphorylation deficiency 36	AR
MRPS22	605810	Ovarian dysgenesis 7	AR
		Combined oxidative phosphorylation deficiency 5	AR
MRPS34	611994	Combined oxidative phosphorylation deficiency 32	AR
MSMO1	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis	AR
MSR1	153622	Barrett esophagus/esophageal adenocarcinoma	-
MSRB3	613719	Deafness, autosomal recessive 74	AR
MSTO1	617619	Myopathy, mitochondrial, and ataxia	AD, AR
MSX1	142983	Orofacial cleft 5	AD

		Tooth agenesis, selective, 1, with or without orofacial cleft	AD
		Ectodermal dysplasia 3, Witkop type	AD
MSX2	123101	Parietal foramina with cleidocranial dysplasia	AD
		Parietal foramina 1	AD
		Craniosynostosis 2	AD
MTFMT	611766	Combined oxidative phosphorylation deficiency 15	AR
		Mitochondrial complex I deficiency, nuclear type 27	AR
MTHFD1	172460	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	AR
		Neural tube defects, folate-sensitive, susceptibility to	AR
MTHFR	607093	Neural tube defects, susceptibility to	AR
		Homocystinuria due to MTHFR deficiency	AR
		Schizophrenia, susceptibility to	AD
		Thromboembolism, susceptibility to	AD
MTHFS	604197	Neurodevelopmental disorder with microcephaly, epilepsy, and hypomyelination	AR
MTM1	300415	Myotubular myopathy, X-linked	XLR
MTMR14	611089	Centronuclear myopathy, autosomal, modifier of	AD
MTMR2	603557	Charcot-Marie-Tooth disease, type 4B1	AR
MTO1	614667	Combined oxidative phosphorylation deficiency 10	AR
MTOR	601231	Focal cortical dysplasia, type II, somatic	-

		Smith-Kingsmore syndrome	AD
MTPAP	613669	?Spastic ataxia 4, autosomal recessive	AR
MTR	156570	Neural tube defects, folate-sensitive, susceptibility to	AR
		Homocystinuria-megaloblastic anemia, cblG complementation type	AR
MTRFR	613541	Spastic paraplegia 55, autosomal recessive	AR
		Combined oxidative phosphorylation deficiency 7	AR
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	AR
		Neural tube defects, folate-sensitive, susceptibility to	AR
MTTP	157147	Metabolic syndrome, protection against	AD
		Abetalipoproteinemia	AR
MUSK	601296	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency	AR
		Fetal akinesia deformation sequence 1	AR
MYBPC1	160794	Arthrogryposis, distal, type 1B	AD
		Lethal congenital contracture syndrome 4	AR
		Myopathy, congenital, with tremor	AD
MYBPC3	600958	Cardiomyopathy, hypertrophic, 4	AD, AR
		Cardiomyopathy, dilated, 1MM	AD
		Left ventricular noncompaction 10	AD
MYCN	164840	Feingold syndrome 1	AD

MYH2	160740	Proximal myopathy and ophthalmoplegia	AD, AR
MYH3	160720	Arthrogryposis, distal, type 2B3 (Sheldon-Hall)	AD
		Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B	AR
		Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A	AD
		Arthrogryposis, distal, type 2A (Freeman-Sheldon)	AD
MYH7	160760	Myopathy, myosin storage, autosomal recessive	AR
		Scapuloperoneal syndrome, myopathic type	AD
		Left ventricular noncompaction 5	AD
		Cardiomyopathy, dilated, 1S	AD
		Cardiomyopathy, hypertrophic, 1	AD, DD
		Myopathy, myosin storage, autosomal dominant	AD
		Laing distal myopathy	AD
MYH8	160741	Trismus-pseudocamptodactyly syndrome	AD
		Carney complex variant	-
MYL1	160780	Myopathy, congenital, with fast-twitch (type II) fiber atrophy	AR
MYL2	160781	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy	AR
		Cardiomyopathy, hypertrophic, 10	AD
MYMK	615345	Carey-Fineman-Ziter syndrome	AR
MYO18B	607295	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism	AR

MYO1E	601479	Glomerulosclerosis, focal segmental, 6	AR
MYO5A	160777	Griscelli syndrome, type 1	AR
MYO9A	604875	Myasthenic syndrome, congenital, 24, presynaptic	AR
MYO9B	602129	Celiac disease, susceptibility to, 4	-
MYORG	618255	Basal ganglia calcification, idiopathic, 7, autosomal recessive	AR
MYOT	604103	Myopathy, myofibrillar, 3	AD
		Myopathy, spheroid body	AD
MYPN	608517	Cardiomyopathy, hypertrophic, 22	AD
		Cardiomyopathy, dilated, 1KK	AD
		Cardiomyopathy, familial restrictive, 4	AD
		Nemaline myopathy 11, autosomal recessive	AR
MYT1L	613084	Mental retardation, autosomal dominant 39	AD
NAA10	300013	Ogden syndrome	XLD, XLR
		Microphthalmia, syndromic 1	XL
NAA15	608000	Mental retardation, autosomal dominant 50	AD
NACC1	610672	Neurodevelopmental disorder with epilepsy, cataracts, feeding difficulties, and delayed brain myelination	AD
NADK2	615787	2, 4-dienoyl-CoA reductase deficiency	AR
NAGA	104170	Schindler disease, type III	AR
		Kanzaki disease	AR

		Schindler disease, type I	AR
NAGLU	609701	?Charcot-Marie-Tooth disease, axonal, type 2V	AD
		Mucopolysaccharidosis type IIIB (Sanfilippo B)	AR
NAGS	608300	N-acetylglutamate synthase deficiency	AR
NALCN	611549	Congenital contractures of the limbs and face, hypotonia, and developmental delay	AD
		Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	AR
NARS2	612803	?Deafness, autosomal recessive 94	AR
		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	AR
		Infantile liver failure syndrome 2	AR
NBEA	604889	Neurodevelopmental disorder with or without early-onset generalized epilepsy	AD
NCAPD3	609276	Microcephaly 22, primary, autosomal recessive	AR
NDE1	609449	Lissencephaly 4 (with microcephaly)	AR
		?Microhydranencephaly	AR
NDP	300658	Norrie disease	XLR
		Exudative vitreoretinopathy 2, X-linked	XLD, XLR
NDRG1	605262	Charcot-Marie-Tooth disease, type 4D	AR

NDST1	600853	Mental retardation, autosomal recessive 46	AR
NDUFA1	300078	Mitochondrial complex I deficiency, nuclear type 12	XLR
NDUFA10	603835	Mitochondrial complex I deficiency, nuclear type 22	AR
NDUFA11	612638	Mitochondrial complex I deficiency, nuclear type 14	AR
NDUFA12	614530	?Mitochondrial complex I deficiency, nuclear type 23	AR
NDUFA2	602137	Mitochondrial complex I deficiency, nuclear type 13	AR
NDUFA6	602138	Mitochondrial complex I deficiency, nuclear type 33	AR
NDUFA9	603834	Mitochondrial complex I deficiency, nuclear type 26	AR
NDUFAF1	606934	Mitochondrial complex I deficiency, nuclear type 11	AR
NDUFAF2	609653	Mitochondrial complex I deficiency, nuclear type 10	AR
NDUFAF3	612911	Mitochondrial complex I deficiency, nuclear type 18	AR
NDUFAF4	611776	Mitochondrial complex I deficiency, nuclear type 15	AR
NDUFAF5	612360	Mitochondrial complex I deficiency, nuclear type 16	AR
NDUFAF6	612392	Mitochondrial complex I deficiency, nuclear type 17	AR
		Fanconi renotubular syndrome 5	AR
NDUFB11	300403	Linear skin defects with multiple congenital anomalies 3	XLD
		?Mitochondrial complex I deficiency, nuclear type 30	XL
NDUFB3	603839	Mitochondrial complex I deficiency, nuclear type 25	AR
NDUFB8	602140	Mitochondrial complex I deficiency, nuclear type 32	AR

NDUFB9	601445	?Mitochondrial complex I deficiency, nuclear type 24	AR
NDUFS1	157655	Mitochondrial complex I deficiency, nuclear type 5	AR
NDUFS2	602985	Mitochondrial complex I deficiency, nuclear type 6	AR
NDUFS3	603846	Mitochondrial complex I deficiency, nuclear type 8	AR
NDUFS4	602694	Mitochondrial complex I deficiency, nuclear type 1	AR
NDUFS6	603848	Mitochondrial complex I deficiency, nuclear type 9	AR
NDUFS7	601825	Mitochondrial complex I deficiency, nuclear type 3	AR
NDUFS8	602141	Mitochondrial complex I deficiency, nuclear type 2	AR
NDUFV1	161015	Mitochondrial complex I deficiency, nuclear type 4	AR
NDUFV2	600532	Mitochondrial complex I deficiency, nuclear type 7	AR
NEB	161650	Nemaline myopathy 2, autosomal recessive	AR
		Arthrogryposis multiplex congenita 6	AR
NECAP1	611623	Developmental and epileptic encephalopathy 21	AR
NECTIN1	600644	Orofacial cleft 7	AR
		Cleft lip/palate-ectodermal dysplasia syndrome	AR
NEDD4L	606384	Periventricular nodular heterotopia 7	AD
NEFH	162230	Charcot-Marie-Tooth disease, axonal, type 2CC	AD
		?{Amyotrophic lateral sclerosis, susceptibility to	AD, AR
NEK1	604588	Short-rib thoracic dysplasia 6 with or without polydactyly	AR, DR

		Amyotrophic lateral sclerosis, susceptibility to, 24	AD
NEK10	618726	Ciliary dyskinesia, primary, 44	AR
NEU1	608272	Sialidosis, type I	AR
		Sialidosis, type II	AR
NEUROD2	601725	Developmental and epileptic encephalopathy 72	AD
NEXMIF	300524	Mental retardation, X-linked 98	XLD
NF1	613113	Watson syndrome	AD
		Leukemia, juvenile myelomonocytic	AD, SM
		Neurofibromatosis, type 1	AD
		Neurofibromatosis, familial spinal	AD
		Neurofibromatosis-Noonan syndrome	AD
NFE2L2	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia	AD
NFIA	600727	Brain malformations with or without urinary tract defects	AD
NFIB	600728	Macrocephaly, acquired, with impaired intellectual development	AD
NFIX	164005	Marshall-Smith syndrome	AD
		Sotos syndrome 2	AD
NFU1	608100	Multiple mitochondrial dysfunctions syndrome 1	AR
NGF	162030	Neuropathy, hereditary sensory and autonomic, type V	AR
NGLY1	610661	Congenital disorder of deglycosylation	AR

NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	-
NHLRC1	608072	Epilepsy, progressive myoclonic 2B (Lafora)	AR
NHS	300457	Nance-Horan syndrome	XLD
		Cataract 40, X-linked	XL
NIPA1	608145	Spastic paraplegia 6, autosomal dominant	AD
NIPBL	608667	Cornelia de Lange syndrome 1	AD
NKX6-2	605955	Spastic ataxia 8, autosomal recessive, with hypomyelinating leukodystrophy	AR
NLGN3	300336	Autism susceptibility, X-linked 1	XL
		Asperger syndrome susceptibility, X-linked 1	IC, MF, XL
NLGN4X	300427	Asperger syndrome susceptibility, X-linked 2	XL
		Autism susceptibility, X-linked 2	IC, MF, XL
		Mental retardation, X-linked	IC, MF, XL
NLRP12	609648	Familial cold autoinflammatory syndrome 2	AD
NLRP3	606416	CINCA syndrome	AD
		Deafness, autosomal dominant 34, with or without inflammation	AD
		Keratoendothelitis fugax hereditaria	AD
		Familial cold inflammatory syndrome 1	AD
		Muckle-Wells syndrome	AD
NNT	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency	AR

NOTCH1	190198	Adams-Oliver syndrome 5	AD
		Aortic valve disease 1	AD
NOTCH2	600275	Alagille syndrome 2	AD
		Hajdu-Cheney syndrome	AD
NOTCH3	600276	?Myofibromatosis, infantile 2	AD
		Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1	AD
		Lateral meningocele syndrome	AD
NPC1	607623	Niemann-Pick disease, type C1	AR
		Niemann-Pick disease, type D	AR
NPC2	601015	Niemann-pick disease, type C2	AR
NPHP1	607100	Joubert syndrome 4	AR
		Nephronophthisis 1, juvenile	AR
		Senior-Loken syndrome-1	AR
NPHP3	608002	Nephronophthisis 3	AR
		Meckel syndrome 7	AR
		Renal-hepatic-pancreatic dysplasia 1	AR
NPR2	108961	Acromesomelic dysplasia, Maroteaux type	AR
		Short stature with nonspecific skeletal abnormalities	AD
		Epiphyseal chondrodysplasia, Miura type	AD

NPRL2	607072	Epilepsy, familial focal, with variable foci 2	AD
NPRL3	600928	Epilepsy, familial focal, with variable foci 3	AD
NR2F1	132890	Bosch-Boonstra-Schaaf optic atrophy syndrome	AD
NR3C2	600983	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy	-
		Pseudohypoaldosteronism type I, autosomal dominant	AD
NRXN1	600565	Schizophrenia, susceptibility to, 17	-
		Pitt-Hopkins-like syndrome 2	AR
NSD1	606681	Sotos syndrome 1	AD
NSD2	602952	Rauch-Steindl syndrome	AD
NSDHL	300275	CK syndrome	XLR
		CHILD syndrome	XLD
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome	AR
NSUN2	610916	Mental retardation, autosomal recessive 5	AR
NT5C2	600417	Spastic paraplegia 45, autosomal recessive	AR
NTHL1	602656	Familial adenomatous polyposis 3	AR
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	AR
NTRK2	600456	Developmental and epileptic encephalopathy 58	AD
		Obesity, hyperphagia, and developmental delay	AD
NUBPL	613621	Mitochondrial complex I deficiency, nuclear type 21	AR

NUP133	607613	Nephrotic syndrome, type 18	AR
		?Galloway-Mowat syndrome 8	AR
NUP62	605815	Striatonigral degeneration, infantile	AR
NUS1	610463	?Congenital disorder of glycosylation, type 1aa	AR
		Mental retardation, autosomal dominant 55, with seizures	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	XLR
		Dent disease 2	XLR
ODAD4	617095	Ciliary dyskinesia, primary, 35	AR
OFD1	300170	Joubert syndrome 10	XLR
		Simpson-Golabi-Behmel syndrome, type 2	XLR
		?Retinitis pigmentosa 23	XLR
		Orofaciodigital syndrome I	XLD
OGDH	613022	Alpha-ketoglutarate dehydrogenase deficiency	AR
OPA1	605290	Optic atrophy 1	AD
		?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type)	AR
		Glaucoma, normal tension, susceptibility to	-
		Optic atrophy plus syndrome	AD

		Behr syndrome	AR
OPA3	606580	3-methylglutaconic aciduria, type III	AR
		Optic atrophy 3 with cataract	AD
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance	XLR
OPTN	602432	Glaucoma, normal tension, susceptibility to	-
		Glaucoma 1, open angle, E	AD
		Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia	AD, AR
ORC1	601902	Meier-Gorlin syndrome 1	AR
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
OXCT1	601424	Succinyl CoA:3-oxoacid CoA transferase deficiency	AR
P4HA2	600608	Myopia 25, autosomal dominant	AD
P4HB	176790	Cole-Carpenter syndrome 1	AD
P4HTM	614584	Hypotonia, hypoventilation, impaired intellectual development, dysautonomia, epilepsy, and eye abnormalities	AR
PACS1	607492	Schuurs-Hoeijmakers syndrome	AD
PACS2	610423	Developmental and epileptic encephalopathy 66	AD
PAFAH1B1	601545	Subcortical laminar heterotopia	AD
		Lissencephaly 1	AD

PAH	612349	Phenylketonuria	AR
		[Hyperphenylalaninemia, non-PKU mild]	AR
PAK1	602590	Intellectual developmental disorder with macrocephaly, seizures, and speech delay	AD
PAK3	300142	Mental retardation, X-linked 30/47	XLR
PAM16	614336	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type	AR
PANK2	606157	HARP syndrome	AR
		Neurodegeneration with brain iron accumulation 1	AR
PARK7	602533	Parkinson disease 7, autosomal recessive early-onset	AR
PARS2	612036	Developmental and epileptic encephalopathy 75	AR
PAX3	606597	Waardenburg syndrome, type 3	AD, AR
		Rhabdomyosarcoma 2, alveolar	SM
		Craniofacial-deafness-hand syndrome	AD
		Waardenburg syndrome, type 1	AD
PAX6	607108	?Coloboma, ocular	AD
		Aniridia	AD
		?Morning glory disc anomaly	AD
		Keratitis	AD
		Optic nerve hypoplasia	AD
		?Coloboma of optic nerve	AD

		Anterior segment dysgenesis 5, multiple subtypes	AD
		Cataract with late-onset corneal dystrophy	AD
		Foveal hypoplasia 1	AD
PAX7	167410	Myopathy, congenital, progressive, with scoliosis	AR
		Rhabdomyosarcoma 2, alveolar	SM
PBX1	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay	AD
PC	608786	Pyruvate carboxylase deficiency	AR
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, D	AR
PCCA	232000	Propionicacidemia	AR
PCCB	232050	Propionicacidemia	AR
PCDH12	605622	Diencephalic-mesencephalic junction dysplasia syndrome 1	AR
PCDH15	605514	Usher syndrome, type 1D/F digenic	AR, DR
		Deafness, autosomal recessive 23	AR
		Usher syndrome, type 1F	AR
PCDH19	300460	Developmental and epileptic encephalopathy 9	XL
PCK2	614095	PEPCK deficiency, mitochondrial	AR
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PCYT2	602679	Spastic paraplegia 82, autosomal recessive	AR
PDCD1	600244	Systemic lupus erythematosus, susceptibility to, 2	-

		Multiple sclerosis, disease progression, modifier of	MF
PDCD10	609118	Cerebral cavernous malformations 3	AD
PDE10A	610652	Dyskinesia, limb and orofacial, infantile-onset	AR
		Striatal degeneration, autosomal dominant	AD
PDE6D	602676	Joubert syndrome 22	AR
PDE8B	603390	Striatal degeneration, autosomal dominant	AD
		Pigmented nodular adrenocortical disease, primary, 3	-
PDGFB	190040	Meningioma, SIS-related	AD
		Basal ganglia calcification, idiopathic, 5	AD
		Dermatofibrosarcoma protuberans	-
PDGFRB	173410	Myeloproliferative disorder with eosinophilia	AD
		Myofibromatosis, infantile, 1	AD
		Premature aging syndrome, Penttinen type	AD
		Basal ganglia calcification, idiopathic, 4	AD
		Kosaki overgrowth syndrome	AD
PDHA1	300502	Pyruvate dehydrogenase E1-alpha deficiency	XLD
PDHB	179060	Pyruvate dehydrogenase E1-beta deficiency	AR
PDHX	608769	Lacticacidemia due to PDX1 deficiency	AR
PDK3	300906	?Charcot-Marie-Tooth disease, X-linked dominant, 6	XLD

PDP1	605993	Pyruvate dehydrogenase phosphatase deficiency	AR
PDSS1	607429	Coenzyme Q10 deficiency, primary, 2	AR
PDSS2	610564	Coenzyme Q10 deficiency, primary, 3	AR
PDX1	600733	Pancreatic agenesis 1	AR
		MODY, type IV	-
		Diabetes mellitus, type II, susceptibility to	AD
PDYN	131340	Spinocerebellar ataxia 23	AD
PER2	603426	?Advanced sleep phase syndrome, familial, 1	AD
PET100	614770	Mitochondrial complex IV deficiency, nuclear type 12	AR
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger)	AR
		Peroxisome biogenesis disorder 1B (NALD/IRD)	AR
		Heimler syndrome 1	AR
PEX10	602859	Peroxisome biogenesis disorder 6A (Zellweger)	AR
		Peroxisome biogenesis disorder 6B	AR
PEX11B	603867	Peroxisome biogenesis disorder 14B	AR
PEX12	601758	Peroxisome biogenesis disorder 3B	AR
		Peroxisome biogenesis disorder 3A (Zellweger)	AR
PEX13	601789	Peroxisome biogenesis disorder 11B	AR
		Peroxisome biogenesis disorder 11A (Zellweger)	AR

PEX14	601791	Peroxisome biogenesis disorder 13A (Zellweger)	AR
PEX16	603360	Peroxisome biogenesis disorder 8A (Zellweger)	AR
		Peroxisome biogenesis disorder 8B	AR
PEX19	600279	Peroxisome biogenesis disorder 12A (Zellweger)	AR
PEX2	170993	Peroxisome biogenesis disorder 5B	AR
		Peroxisome biogenesis disorder 5A (Zellweger)	AR
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger)	AR
		Peroxisome biogenesis disorder 7B	AR
PEX3	603164	Peroxisome biogenesis disorder 10A (Zellweger)	AR
		?Peroxisome biogenesis disorder 10B	AR
PEX5	600414	Rhizomelic chondrodysplasia punctata, type 5	AR
		Peroxisome biogenesis disorder 2B	AR
		Peroxisome biogenesis disorder 2A (Zellweger)	AR
PEX6	601498	Peroxisome biogenesis disorder 4B	AD, AR
		Peroxisome biogenesis disorder 4A (Zellweger)	AR
		Heimler syndrome 2	AR
PEX7	601757	Peroxisome biogenesis disorder 9B	AR
		Rhizomelic chondrodysplasia punctata, type 1	AR
PFKM	610681	Glycogen storage disease VII	AR

PFN1	176610	Amyotrophic lateral sclerosis 18	-
PGAM2	612931	Glycogen storage disease X	AR
PGAP1	611655	Mental retardation, autosomal recessive 42	AR
PGAP2	615187	Hyperphosphatasia with mental retardation syndrome 3	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
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	XLR
PHF8	300560	Mental retardation syndrome, X-linked, Siderius type	XLR
PHGDH	606879	Phosphoglycerate dehydrogenase deficiency	AR
		Neu-Laxova syndrome 1	AR
PHIP	612870	Chung-Jansen syndrome	AD
PHKA1	311870	Muscle glycogenosis	XLR
PHOX2B	603851	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease	AD
		Neuroblastoma, susceptibility to, 2	-
		Neuroblastoma with Hirschsprung disease	-
PHYH	602026	Refsum disease	AR
PIEZO2	613629	Arthrogryposis, distal, type 3	AD
		?Marden-Walker syndrome	AD

		Arthrogryposis, distal, type 5	AD
		Arthrogryposis, distal, with impaired proprioception and touch	AR
PIGA	311770	Paroxysmal nocturnal hemoglobinuria, somatic	-
		Multiple congenital anomalies-hypotonia-seizures syndrome 2	XLR
		Neurodevelopmental disorder with epilepsy and hemochromatosis	-
PIGB	604122	Developmental and epileptic encephalopathy 80	AR
PIGC	601730	Glycosylphosphatidylinositol biosynthesis defect 16	AR
PIGG	616918	[Blood group, EMM system]	AR
		Mental retardation, autosomal recessive 53	AR
PIGH	600154	Glycosylphosphatidylinositol biosynthesis defect 17	AR
PIGL	605947	CHIME syndrome	AR
PIGN	606097	Multiple congenital anomalies-hypotonia-seizures syndrome 1	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	AR
		?Paroxysmal nocturnal hemoglobinuria 2	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
PIK3CA	171834	Keratosis, seborrheic, somatic	-
		Ovarian cancer, somatic	-
		CLAPO syndrome, somatic	-
		Macrodactyly, somatic	-
		CLOVE syndrome, somatic	-
		Gastric cancer, somatic	-
		Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic	-
		Colorectal cancer, somatic	-
		Nevus, epidermal, somatic	-
		Breast cancer, somatic	-
		Cerebral cavernous malformations 4, somatic	-
		Cowden syndrome 5	-
		Hepatocellular carcinoma, somatic	-
		Nonsmall cell lung cancer, somatic	-
PIK3R2	603157	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1	AD
PIK3R5	611317	Ataxia-oculomotor apraxia 3	AR
PINK1	608309	Parkinson disease 6, early onset	AR

PIP5K1C	606102	Lethal congenital contractural syndrome 3	AR
PITX1	602149	Liebenberg syndrome	AD
		Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly	AD
PITX2	601542	Anterior segment dysgenesis 4	AD
		Ring dermoid of cornea	AD
		Axenfeld-Rieger syndrome, type 1	AD
PKLR	609712	Adenosine triphosphate, elevated, of erythrocytes	AD
		Pyruvate kinase deficiency	AR
PLA2G6	603604	Neurodegeneration with brain iron accumulation 2B	AR
		Parkinson disease 14, autosomal recessive	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	AD
		Autoinflammation, antibody deficiency, and immune dysregulation syndrome	AD
PLEC	601282	Epidermolysis bullosa simplex, Ogna type	AD
		Epidermolysis bullosa simplex with muscular dystrophy	AR
		?Epidermolysis bullosa simplex with nail dystrophy	AR
		Epidermolysis bullosa simplex with pyloric atresia	AR

		Muscular dystrophy, limb-girdle, autosomal recessive 17	AR
PLEKHG2	611893	Leukodystrophy and acquired microcephaly with or without dystonia	AR
PLEKHG5	611101	Charcot-Marie-Tooth disease, recessive intermediate C	AR
		Spinal muscular atrophy, distal, autosomal recessive, 4	AR
PLK4	605031	Microcephaly and chorioretinopathy, autosomal recessive, 2	AR
PLN	172405	Cardiomyopathy, dilated, 1P	-
		Cardiomyopathy, hypertrophic, 18	AD
PLOD2	601865	Bruck syndrome 2	AR
PLP1	300401	Pelizaeus-Merzbacher disease	XLR
		Spastic paraplegia 2, X-linked	XLR
PLPBP	604436	Epilepsy, early-onset, vitamin B6-dependent	AR
PMM2	601785	Congenital disorder of glycosylation, type Ia	AR
PMP22	601097	Neuropathy, recurrent, with pressure palsies	AD
		Dejerine-Sottas disease	AD, AR
		Roussy-Levy syndrome	AD
		?Neuropathy, inflammatory demyelinating	?AD
		Charcot-Marie-Tooth disease, type 1E	AD
		Charcot-Marie-Tooth disease, type 1A	AD
PMPCA	613036	Spinocerebellar ataxia, autosomal recessive 2	AR

PMPCB	603131	Multiple mitochondrial dysfunctions syndrome 6	AR
PNKD	609023	Paroxysmal nonkinesigenic dyskinesia 1	AD
PNKP	605610	Ataxia-oculomotor apraxia 4	AR
		Microcephaly, seizures, and developmental delay	AR
		?Charcot-Marie-Tooth disease, type 2B2	AR
PNPLA2	609059	Neutral lipid storage disease with myopathy	AR
PNPLA6	603197	Spastic paraplegia 39, autosomal recessive	AR
		Boucher-Neuhauser syndrome	AR
		Oliver-McFarlane syndrome	AR
		?Laurence-Moon syndrome	AR
PNPLA8	612123	?Mitochondrial myopathy with lactic acidosis	AR
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	AR
PNPT1	610316	Deafness, autosomal recessive 70	AR
		Combined oxidative phosphorylation deficiency 13	AR
		Spinocerebellar ataxia 25	-
POGLUT1	615618	Dowling-Degos disease 4	AD
		?Muscular dystrophy, limb-girdle, autosomal recessive 21	AR
POGZ	614787	White-Sutton syndrome	AD
POLA1	312040	Van Esch-O'Driscoll syndrome	XLR

		Pigmentary disorder, reticulate, with systemic manifestations, X-linked	XLR
POLG	174763	Progressive external ophthalmoplegia, autosomal recessive 1	AR
		Progressive external ophthalmoplegia, autosomal dominant 1	AD
		Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	AR
		Mitochondrial DNA depletion syndrome 4B (MNGIE type)	AR
		Mitochondrial DNA depletion syndrome 4A (Alpers type)	AR
POLG2	604983	Mitochondrial DNA depletion syndrome 16 (hepatic type)	AR
		?Mitochondrial DNA depletion syndrome 16B (neuroophthalmic type)	AR
		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4	AD
POLR1C	610060	Leukodystrophy, hypomyelinating, 11	AR
		Treacher Collins syndrome 3	AR
POLR1D	613715	Treacher Collins syndrome 2	AD, AR
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism	AR
		Wiedemann-Rautenstrauch syndrome	AR
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	AR
		Charcot-Marie-Tooth disease, demyelinating, type 1I	AD
POMGNT1	606822	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	AR
		Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3	AR

		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3	AR
		Retinitis pigmentosa 76	AR
POMGNT2	614828	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8	AR
POMK	615247	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12	AR
		?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12	AR
POMT1	607423	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	AR
		Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1	AR
POMT2	607439	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2	AR
		Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	AR
		Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	AR
PON1	168820	Microvascular complications of diabetes 5	-
POP1	602486	Anauxetic dysplasia 2	AR
PORCN	300651	Focal dermal hypoplasia	XLD
POT1	606478	Melanoma, cutaneous malignant, susceptibility to, 10	AD
		Glioma susceptibility 9	AD
POU1F1	173110	Pituitary hormone deficiency, combined, 1	AD, AR

PPM1D	605100	Jansen de Vries syndrome	AD
		Breast cancer, somatic	-
PPOX	600923	Porphyria variegata	AD
PPP2CA	176915	Neurodevelopmental disorder and language delay with or without structural brain abnormalities	AD
PPP2R1A	605983	Mental retardation, autosomal dominant 36	AD
PPP2R5D	601646	Mental retardation, autosomal dominant 35	AD
PPP3CA	114105	Epileptic encephalopathy, infantile or early childhood, 1	AD
		Arthrogryposis, cleft palate, craniosynostosis, and impaired intellectual development	AD
PPT1	600722	Ceroid lipofuscinosis, neuronal, 1	AR
PQBP1	300463	Renpenning syndrome	XLR
PREPL	609557	Myasthenic syndrome, congenital, 22	AR
PRF1	170280	Hemophagocytic lymphohistiocytosis, familial, 2	AR
		Lymphoma, non-Hodgkin	-
		Aplastic anemia	-
PRICKLE1	608500	Epilepsy, progressive myoclonic 1B	AR
PRKAG2	602743	Glycogen storage disease of heart, lethal congenital	AD
		Cardiomyopathy, hypertrophic 6	AD
		Wolff-Parkinson-White syndrome	AD
PRKCG	176980	Spinocerebellar ataxia 14	AD

PRKN	602544	Parkinson disease, juvenile, type 2	AR
		Ovarian cancer, somatic	-
		Adenocarcinoma of lung, somatic	-
PRKRA	603424	Dystonia 16	AR
PRMT7	610087	Short stature, brachydactyly, intellectual developmental disability, and seizures	AR
PRNP	176640	Insomnia, fatal familial	AD
		Creutzfeldt-Jakob disease	AD
		Gerstmann-Straussler disease	AD
		Prion disease with protracted course	AD
		Kuru, susceptibility to	-
		Cerebral amyloid angiopathy, PRNP-related	AD
		Huntington disease-like 1	AD
PRODH	606810	Schizophrenia, susceptibility to, 4	AD
		Hyperprolinemia, type I	AR
PROP1	601538	Pituitary hormone deficiency, combined, 2	AR
PRPH	170710	Amyotrophic lateral sclerosis, susceptibility to	AD, AR
PRPS1	311850	Charcot-Marie-Tooth disease, X-linked recessive, 5	XLR
		Deafness, X-linked 1	XL
		Phosphoribosylpyrophosphate synthetase superactivity	XLR

		Arts syndrome	XLR
		Gout, PRPS-related	XLR
PRRT2	614386	Episodic kinesigenic dyskinesia 1	AD
		Convulsions, familial infantile, with paroxysmal choreoathetosis	AD
		Seizures, benign familial infantile, 2	AD
PRRX1	167420	Agnathia-otocephaly complex	AD, AR
PRSS12	606709	Mental retardation, autosomal recessive 1	AR
PRX	605725	Dejerine-Sottas disease	AD, AR
		Charcot-Marie-Tooth disease, type 4F	AR
PSAP	176801	Combined SAP deficiency	AR
		Gaucher disease, atypical	-
		Krabbe disease, atypical	AR
		Parkinson disease 24, autosomal dominant, susceptibility to	AD
		Metachromatic leukodystrophy due to SAP-b deficiency	AR
PSAT1	610936	Neu-Laxova syndrome 2	AR
		?Phosphoserine aminotransferase deficiency	AR
PSEN1	104311	Cardiomyopathy, dilated, 1U	AD
		Pick disease	AD
		Alzheimer disease, type 3	AD

		Alzheimer disease, type 3, with spastic paraparesis and apraxia	AD
		?Acne inversa, familial, 3	AD
		Dementia, frontotemporal	AD
		Alzheimer disease, type 3, with spastic paraparesis and unusual plaques	AD
PSEN2	600759	Alzheimer disease-4	AD
		Cardiomyopathy, dilated, 1V	AD
PSMD12	604450	Stankiewicz-Isidor syndrome	AD
PSPH	172480	Phosphoserine phosphatase deficiency	AR
PTCH1	601309	Basal cell carcinoma, somatic	-
		Holoprosencephaly 7	AD
		Basal cell nevus syndrome	AD
PTCHD1	300828	Autism, susceptibility to, X-linked 4	XLR
PTEN	601728	Cowden syndrome 1	AD
		Lhermitte-Duclos syndrome	AD
		Macrocephaly/autism syndrome	AD
		Glioma susceptibility 2	-
		Meningioma	AD
		Prostate cancer, somatic	-
PTF1A	607194	Pancreatic and cerebellar agenesis	AR

		Pancreatic agenesis 2	AR
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic	-
		LEOPARD syndrome 1	AD
		Metachondromatosis	AD
		Noonan syndrome 1	AD
PTPN23	606584	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	AR
PTPRC	151460	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive	AR
		Hepatitis C virus, susceptibility to	-
		Immunodeficiency 105, severe combined	-
PTRH2	608625	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease	AR
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	AR
PUM1	607204	Spinocerebellar ataxia 47	AD
PURA	600473	Mental retardation, autosomal dominant 31	AD
PUS1	608109	Myopathy, lactic acidosis, and sideroblastic anemia 1	AR
PUS3	616283	Neurodevelopmental disorder with microcephaly and gray sclerae	AR
PXDN	605158	Anterior segment dysgenesis 7, with sclerocornea	AR
PYCR1	179035	Cutis laxa, autosomal recessive, type IIB	AR
		Cutis laxa, autosomal recessive, type IIIB	AR
PYCR2	616406	Leukodystrophy, hypomyelinating, 10	AR

PYGM	608455	McArdle disease	AR
PYROXD1	617220	Myopathy, myofibrillar, 8	AR
QARS1	603727	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	AR
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C	AR
QRICH1	617387	Ververi-Brady syndrome	AD
RAB11B	604198	Neurodevelopmental disorder with ataxic gait, absent speech, and decreased cortical white matter	AD
RAB18	602207	Warburg micro syndrome 3	AR
RAB27A	603868	Griscelli syndrome, type 2	AR
RAB39B	300774	Mental retardation, X-linked 72	XLR
		Waisman syndrome	XLR
RAB3GAP1	602536	Martsolf syndrome 2	AR
		Warburg micro syndrome 1	AR
RAB3GAP2	609275	Warburg micro syndrome 2	AR
		Martsolf syndrome	AR
RAB7A	602298	Charcot-Marie-Tooth disease, type 2B	AD
RAC1	602048	Mental retardation, autosomal dominant 48	AD
RAD21	606462	Cornelia de Lange syndrome 4	AD
		?Mungan syndrome	AR
RAD50	604040	Nijmegen breakage syndrome-like disorder	AR

RAF1	164760	Noonan syndrome 5	AD
		LEOPARD syndrome 2	-
		Cardiomyopathy, dilated, 1NN	AD
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
RAPSN	601592	Fetal akinesia deformation sequence 2	AR
		Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR
RARS1	107820	Leukodystrophy, hypomyelinating, 9	AR
RARS2	611524	Pontocerebellar hypoplasia, type 6	AR
RBBP8	604124	Seckel syndrome 2	AR
		Jawad syndrome	AR
RBCK1	610924	Polyglucosan body myopathy 1 with or without immunodeficiency	AR
RBM10	300080	TARP syndrome	XLR
RBM8A	605313	Thrombocytopenia-absent radius syndrome	AR
RDH11	607849	?Retinal dystrophy, juvenile cataracts, and short stature syndrome	AR
REEP1	609139	Spastic paraplegia 31, autosomal dominant	AD
		?Neuronopathy, distal hereditary motor, type VB	AD
REEP2	609347	?Spastic paraplegia 72, autosomal recessive	AD, AR

		?Spastic paraplegia 72, autosomal dominant	AD, AR
RELN	600514	Lissencephaly 2 (Norman-Roberts type)	AR
		Epilepsy, familial temporal lobe, 7	AD
RERE	605226	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart	AD
REST	600571	Wilms tumor 6, susceptibility to	AD
		Fibromatosis, gingival, 5	AD
		?Deafness, autosomal dominant 27	AD
RET	164761	Hirschsprung disease, susceptibility to, 1	AD
		Multiple endocrine neoplasia IIA	AD
		Medullary thyroid carcinoma	AD
		Pheochromocytoma	AD
		Multiple endocrine neoplasia IIB	AD
		Hirschsprung disease, protection against	AD
		Central hypoventilation syndrome, congenital	AD
RETREG1	613114	Neuropathy, hereditary sensory and autonomic, type IIB	AR
RFT1	611908	Congenital disorder of glycosylation, type In	AR
RHOBTB2	607352	Developmental and epileptic encephalopathy 64	AD
RIMS1	606629	Cone-rod dystrophy 7	AD
RIN2	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis	AR

RMND1	614917	Combined oxidative phosphorylation deficiency 11	AR
RNASEH1	604123	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2	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
RNF168	612688	RIDDLE syndrome	AR
RNF170	614649	Ataxia, sensory, 1, autosomal dominant	AD
		Spastic paraplegia 85, autosomal recessive	AR
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism	AR
ROBO2	602431	Vesicoureteral reflux 2	AD
ROGDI	614574	Kohlschutter-Tonz syndrome	AR
ROR2	602337	Brachydactyly, type B1	AD
		Robinow syndrome, autosomal recessive	AR
RORA	600825	Intellectual developmental disorder with or without epilepsy or cerebellar ataxia	AD
RORB	601972	Epilepsy, idiopathic generalized, susceptibility to, 15	AD
RPGRIP1L	610937	Joubert syndrome 7	AR

		?COACH syndrome 3	AR
		Meckel syndrome 5	AR
RPIA	180430	Ribose 5-phosphate isomerase deficiency	AR
RPL10	312173	Autism, susceptibility to, X-linked 5	-
		Mental retardation, X-linked, syndromic, 35	XLR
RPL35A	180468	Diamond-Blackfan anemia 5	AD
RPS14	130620	Macrocytic anemia, refractory, due to 5q deletion, somatic	-
RPS6KA3	300075	Coffin-Lowry syndrome	XLD
		Mental retardation, X-linked 19	XLD
RRM2B	604712	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	AR
		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5	AD
		Mitochondrial DNA depletion syndrome 8B (MNGIE type)	AR
		Rod-cone dystrophy, sensorineural deafness, and Fanconi-type renal dysfunction	AR
RTN2	603183	Spastic paraplegia 12, autosomal dominant	AD
RTN4IP1	610502	Optic atrophy 10 with or without ataxia, mental retardation, and seizures	AR
RTTN	610436	Microcephaly, short stature, and polymicrogyria with seizures	AR
RUBCN	613516	Spinocerebellar ataxia, autosomal recessive 15	AR
RUSC2	611053	Mental retardation, autosomal recessive 61	AR

RXYLT1	605862	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10	AR
RYR1	180901	Malignant hyperthermia susceptibility 1	AD
		King-Denborough syndrome	AD
		King-Denborough syndrome	AD
		Central core disease	AD, AR
		Neuromuscular disease, congenital, with uniform type 1 fiber	AD, AR
		Minicore myopathy with external ophthalmoplegia	AR
SACS	604490	Spastic ataxia, Charlevoix-Saguenay type	AR
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome	AD
		Townes-Brocks syndrome 1	AD
SAMD9L	611170	Ataxia-pancytopenia syndrome	AD
		Monosomy 7 myelodysplasia and leukemia syndrome 1	AD
		Spinocerebellar ataxia 49	AD
SAMHD1	606754	?Chilblain lupus 2	AD
		Aicardi-Goutieres syndrome 5	AR
SARS2	612804	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	AR
SASH1	607955	Dyschromatosis universalis hereditaria 1	AD
		?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma	AR
SASS6	609321	?Microcephaly 14, primary, autosomal recessive	AR

SATB2	608148	Glass syndrome	AD
SBDS	607444	Aplastic anemia, susceptibility to	-
		Shwachman-Diamond syndrome	AR
SBF1	603560	Charcot-Marie-Tooth disease, type 4B3	AR
SBF2	607697	Charcot-Marie-Tooth disease, type 4B2	AR
SC5D	602286	Lathosterolosis	AR
SCARB2	602257	Epilepsy, progressive myoclonic 4, with or without renal failure	AR
SCN10A	604427	Episodic pain syndrome, familial, 2	AD
SCN1A	182389	Epilepsy, generalized, with febrile seizures plus, type 2	AD
		Migraine, familial hemiplegic, 3	AD
		Developmental and epileptic encephalopathy 6B, non-Dravet	AD
		Febrile seizures, familial, 3A	AD
		Dravet syndrome	AD
SCN1B	600235	Atrial fibrillation, familial, 13	AD
		Epilepsy, generalized, with febrile seizures plus, type 1	AD
		Brugada syndrome 5	-
		Cardiac conduction defect, nonspecific	-
		Developmental and epileptic encephalopathy 52	AR
SCN2A	182390	Developmental and epileptic encephalopathy 11	AD

		Seizures, benign familial infantile, 3	AD
		Episodic ataxia, type 9	AD
SCN3A	182391	Epilepsy, familial focal, with variable foci 4	AD
		Developmental and epileptic encephalopathy 62	AD
SCN4A	603967	Paramyotonia congenita	AD
		Myotonia congenita, atypical, acetazolamide-responsive	AD
		Myasthenic syndrome, congenital, 16	AR
		Hyperkalemic periodic paralysis, type 2	AD
		Hypokalemic periodic paralysis, type 2	AD
SCN8A	600702	?Myoclonus, familial, 2	AD
		Cognitive impairment with or without cerebellar ataxia	AD
		Developmental and epileptic encephalopathy 13	AD
		Seizures, benign familial infantile, 5	AD
SCN9A	603415	Erythermalgia, primary	AD
		Generalized epilepsy with febrile seizures plus, type 7	AD
		Febrile seizures, familial, 3B	AD
		Neuropathy, hereditary sensory and autonomic, type IID	AR
		Insensitivity to pain, congenital	AR
		Paroxysmal extreme pain disorder	AD

		Small fiber neuropathy	AD
SCO1	603644	Mitochondrial complex IV deficiency, nuclear type 4	AR
SCO2	604272	Myopia 6	AD
		Mitochondrial complex IV deficiency, nuclear type 2	AR
SCYL1	607982	Spinocerebellar ataxia, autosomal recessive 21	AR
SDCCAG8	613524	Bardet-Biedl syndrome 16	AR
		Senior-Loken syndrome 7	AR
SDHA	600857	Neurodegeneration with ataxia and late-onset optic atrophy	AD
		Cardiomyopathy, dilated, 1GG	AR
		Leigh syndrome	AR, Mitochondrial
		Mitochondrial respiratory chain complex II deficiency	AR
		Paragangliomas 5	AD
SDHAF1	612848	Mitochondrial complex II deficiency, nuclear type 2	AR
		Mitochondrial complex II deficiency	AR
SDHAF2	613019	Paragangliomas 2	AD
SDHB	185470	Pheochromocytoma	AD
		Mitochondrial complex II deficiency, nuclear type 4	AR
		Paragangliomas 4	AD
		Paraganglioma and gastric stromal sarcoma	-

		Gastrointestinal stromal tumor	AD, IC
SDHD	602690	Mitochondrial complex II deficiency	AR
		Paraganglioma and gastric stromal sarcoma	-
		Paragangliomas 1, with or without deafness	AD
		Mitochondrial complex II deficiency, nuclear type 3	AR
		Pheochromocytoma	AD
SEC23B	610512	Dyserythropoietic anemia, congenital, type II	AR
		?Cowden syndrome 7	AD
SECISBP2	607693	Thyroid hormone metabolism, abnormal	AR
SELENOI	607915	Spastic paraplegia 81, autosomal recessive	AR
SELENON	606210	Muscular dystrophy, rigid spine, 1	AR
		Myopathy, congenital, with fiber-type disproportion	AD, 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
SET	600960	Mental retardation, autosomal dominant 58	AD
SETBP1	611060	Schinzel-Giedion midface retraction syndrome	AD
		Mental retardation, autosomal dominant 29	AD

SETD1A	611052	Neurodevelopmental disorder with speech impairment and dysmorphic facies	AD
		Epilepsy, early-onset, with or without developmental delay	AD
SETD2	612778	Luscan-Lumish syndrome	AD
SETD5	615743	Mental retardation, autosomal dominant 23	AD
SETX	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2	AR
		Amyotrophic lateral sclerosis 4, juvenile	AD
SF3B1	605590	Myelodysplastic syndrome, somatic	-
SFXN4	615564	Combined oxidative phosphorylation deficiency 18	AR
SGCA	600119	Muscular dystrophy, limb-girdle, autosomal recessive 3	AR
SGCB	600900	Muscular dystrophy, limb-girdle, autosomal recessive 4	AR
SGCD	601411	Muscular dystrophy, limb-girdle, autosomal recessive 6	AR
		Cardiomyopathy, dilated, 1L	-
SGCE	604149	Dystonia-11, myoclonic	AD
SGCG	608896	Muscular dystrophy, limb-girdle, autosomal recessive 5	AR
SGSH	605270	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR
SH3TC2	608206	Mononeuropathy of the median nerve, mild	AD
		Charcot-Marie-Tooth disease, type 4C	AR
SHANK2	603290	Autism susceptibility 17	-
SHH	600725	Schizencephaly	-

		Microphthalmia with coloboma 5	AD
		Single median maxillary central incisor	AD
		Holoprosencephaly 3	AD
SHOC2	602775	Noonan syndrome-like with loose anagen hair 1	AD
SHROOM4	300579	Stocco dos Santos X-linked mental retardation syndrome	XL
SIGMAR1	601978	?Amyotrophic lateral sclerosis 16, juvenile	AR
		?Spinal muscular atrophy, distal, autosomal recessive, 2	AR
SIK1	605705	Developmental and epileptic encephalopathy 30	AD
SIL1	608005	Marinesco-Sjogren syndrome	AR
SIN3A	607776	Witteveen-Kolk syndrome	AD
SIX3	603714	Holoprosencephaly 2	AD
		Schizencephaly	-
SKI	164780	Shprintzen-Goldberg syndrome	AD
SLC12A3	600968	Gitelman syndrome	AR
SLC12A5	606726	Epilepsy, idiopathic generalized, susceptibility to, 14	AD
		Developmental and epileptic encephalopathy 34	AR
SLC12A6	604878	Agenesis of the corpus callosum with peripheral neuropathy	AR
SLC13A3	606411	Leukoencephalopathy, acute reversible, with increased urinary alpha-ketoglutarate	AR
SLC13A5	608305	Developmental and epileptic encephalopathy 25, with amelogenesis imperfecta	AR

SLC16A1	600682	Hyperinsulinemic hypoglycemia, familial, 7	AD
		Erythrocyte lactate transporter defect	AD
		Monocarboxylate transporter 1 deficiency	AD, AR
SLC16A2	300095	Allan-Herndon-Dudley syndrome	XL
SLC17A5	604322	Sialic acid storage disorder, infantile	AR
		Salla disease	AR
SLC18A3	600336	Myasthenic syndrome, congenital, 21, presynaptic	AR
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	AR
SLC1A1	133550	?Schizophrenia susceptibility 18	-
		Dicarboxylic aminoaciduria	AR
SLC1A2	600300	Developmental and epileptic encephalopathy 41	AD
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
SLC25A1	190315	Myasthenic syndrome, congenital, 23, presynaptic	AR
		Combined D-2- and L-2-hydroxyglutaric aciduria	AR
SLC25A12	603667	Developmental and epileptic encephalopathy 39	AR

SLC25A13	603859	Citrullinemia, type II, neonatal-onset	AR
		Citrullinemia, adult-onset type II	AR
SLC25A15	603861	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR
SLC25A19	606521	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	AR
		Microcephaly, Amish type	AR
SLC25A20	613698	Carnitine-acylcarnitine translocase deficiency	AR
SLC25A22	609302	Developmental and epileptic encephalopathy 3	AR
SLC25A26	611037	Combined oxidative phosphorylation deficiency 28	AR
SLC25A3	600370	Mitochondrial phosphate carrier deficiency	AR
SLC25A38	610819	Anemia, sideroblastic, 2, pyridoxine-refractory	AR
SLC25A4	103220	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR	AR
		Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD	AD
		Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2	AD
SLC25A42	610823	Metabolic crises, recurrent, with variable encephalomyopathic features and neurologic regression	AR
SLC25A46	610826	Neuropathy, hereditary motor and sensory, type VIB	AR
		Pontocerebellar hypoplasia, type 1E	AR
SLC27A4	604194	Ichthyosis prematurity syndrome	AR
SLC2A1	138140	GLUT1 deficiency syndrome 2, childhood onset	AD
		GLUT1 deficiency syndrome 1, infantile onset, severe	AD, AR

		Epilepsy, idiopathic generalized, susceptibility to, 12	AD
		Stomatin-deficient cryohydrocytosis with neurologic defects	AD
		Dystonia 9	AD
SLC2A10	606145	Arterial tortuosity syndrome	AR
SLC30A10	611146	Hypermanganesemia with dystonia 1	AR
SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration	AR
		Spastic paraplegia 42, autosomal dominant	AD
SLC35A1	605634	Congenital disorder of glycosylation, type IIf	AR
SLC35A2	314375	Congenital disorder of glycosylation, type IIm	Somatic mosaicism, XLD
SLC35A3	605632	?Arthrogryposis, mental retardation, and seizures	AR
SLC35C1	605881	Congenital disorder of glycosylation, type IIc	AR
SLC39A14	608736	?Hyperostosis cranalis interna	AD
		Hypermanganesemia with dystonia 2	AR
SLC39A8	608732	Congenital disorder of glycosylation, type IIn	AR
SLC3A1	104614	Cystinuria	AD, AR
SLC4A4	603345	Renal tubular acidosis, proximal, with ocular abnormalities	AR
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2	AR
SLC52A3	613350	?Fazio-Londe disease	AR
		Brown-Vialetto-Van Laere syndrome 1	AR

SLC5A7	608761	Neuronopathy, distal hereditary motor, type VIIA	AD
		Myasthenic syndrome, congenital, 20, presynaptic	AR
SLC6A1	137165	Myoclonic-atonic epilepsy	AD
SLC6A17	610299	Mental retardation, autosomal recessive 48	AR
SLC6A19	608893	Iminoglycinuria, digenic	AR, DR
		Hyperglycinuria	AD
		Hartnup disorder	AR
SLC6A3	126455	Nicotine dependence, protection against	-
		Parkinsonism-dystonia, infantile, 1	AR
SLC6A4	182138	Obsessive-compulsive disorder	AD
		Anxiety-related personality traits	-
SLC6A5	604159	Hyperekplexia 3	AD, AR
SLC6A8	300036	Cerebral creatine deficiency syndrome 1	XLR
SLC6A9	601019	Glycine encephalopathy with normal serum glycine	AR
SLC7A7	603593	Lysinuric protein intolerance	AR
SLC9A6	300231	Mental retardation, X-linked syndromic, Christianson type	XL
SLC9A9	608396	?Autism susceptibility 16	-
SLCO1B3	605495	Hyperbilirubinemia, Rotor type, digenic	DR
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	AD

		Pancreatic cancer, somatic	-
		Myhre syndrome	AD
		Polyposis, juvenile intestinal	AD
SMARCA2	600014	Blepharophimosis-impaired intellectual development syndrome	AD
		Nicolaides-Baraitser syndrome	AD
SMARCA4	603254	Coffin-Siris syndrome 4	AD
		Rhabdoid tumor predisposition syndrome 2	AD
SMARCB1	601607	Coffin-Siris syndrome 3	AD
		Rhabdoid tumor predisposition syndrome 1	AD
		Schwannomatosis-1, susceptibility to	AD
		Rhabdoid tumors, somatic	-
SMARCC2	601734	Coffin-Siris syndrome 8	AD
SMARCE1	603111	Coffin-Siris syndrome 5	AD
		Meningioma, familial, susceptibility to	AD
SMC1A	300040	Developmental and epileptic encephalopathy 85, with or without midline brain defects	XLD
		Cornelia de Lange syndrome 2	XLD
SMC3	606062	Cornelia de Lange syndrome 3	AD
SMCHD1	614982	Bosma arhinia microphthalmia syndrome	AD
		Fascioscapulohumeral muscular dystrophy 2, digenic	DD

SMPD1	607608	Niemann-Pick disease, type A	AR
		Niemann-Pick disease, type B	AR
SMPD4	610457	Neurodevelopmental disorder with microcephaly, arthrogryposis, and structural brain anomalies	AR
SMS	300105	Mental retardation, X-linked, Snyder-Robinson type	XLR
SNAI2	602150	Waardenburg syndrome, type 2D	AR
		Piebaldism	AD
SNAP25	600322	?Myasthenic syndrome, congenital, 18	AD
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	AR
SNCA	163890	Parkinson disease 1	AD
		Dementia, Lewy body	AD
		Parkinson disease 4	AD
SNCB	602569	Dementia, Lewy body	AD
SNIP1	608241	Psychomotor retardation, epilepsy, and craniofacial dysmorphism	AR
SNTA1	601017	Long QT syndrome 12	AD
SNX14	616105	Spinocerebellar ataxia, autosomal recessive 20	AR
SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus	AR
SOD1	147450	Spastic tetraplegia and axial hypotonia, progressive	AR
		Amyotrophic lateral sclerosis 1	AD, AR
SOD2	147460	Microvascular complications of diabetes 6	-

SON	182465	ZTTK syndrome	AD
SOS1	182530	Noonan syndrome 4	AD
		?Fibromatosis, gingival, 1	AD
SOX10	602229	Waardenburg syndrome, type 4C	AD
		PCWH syndrome	AD
		Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SOX11	600898	Coffin-Siris syndrome 9	AD
SOX2	184429	Microphthalmia, syndromic 3	AD
		Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SOX3	313430	Panhypopituitarism, X-linked	XL
		Mental retardation, X-linked, with isolated growth hormone deficiency	-
SOX5	604975	Lamb-Shaffer syndrome	AD
SPART	607111	Troyer syndrome	AR
SPAST	604277	Spastic paraplegia 4, autosomal dominant	AD
SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome	AR
SPEG	615950	Centronuclear myopathy 5	AR
SPG11	610844	Spastic paraplegia 11, autosomal recessive	AR
		Amyotrophic lateral sclerosis 5, juvenile	AR
		Charcot-Marie-Tooth disease, axonal, type 2X	AR

SPG21	608181	Mast syndrome	AR
SPG7	602783	Spastic paraplegia 7, autosomal recessive	AD, AR
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	?AD, AR
SPTAN1	182810	Developmental and epileptic encephalopathy 5	AD
SPTBN2	604985	Spinocerebellar ataxia, autosomal recessive 14	AR
		Spinocerebellar ataxia 5	AD
SPTBN4	606214	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness	AR
SPTLC1	605712	Neuropathy, hereditary sensory and autonomic, type IA	AD
SPTLC2	605713	Neuropathy, hereditary sensory and autonomic, type IC	AD
SQSTM1	601530	Paget disease of bone 3	AD
		Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	AR
		Myopathy, distal, with rimmed vacuoles	AD
		Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	AD
SRCAP	611421	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities	AD
		Floating-Harbor syndrome	AD
SRD5A3	611715	Congenital disorder of glycosylation, type Iq	AR
		Kahrizi syndrome	AR
SSR4	300090	Congenital disorder of glycosylation, type Iy	XLR
ST3GAL3	606494	?Developmental and epileptic encephalopathy 15	AR

		Mental retardation, autosomal recessive 12	AR
ST3GAL5	604402	Salt and pepper developmental regression syndrome	AR
STAC3	615521	Myopathy, congenital, Baily-Bloch	AR
STAG1	604358	Mental retardation, autosomal dominant 47	AD
STAMBP	606247	Microcephaly-capillary malformation syndrome	AR
STAR	600617	Lipoid adrenal hyperplasia	AR
STAT1	600555	Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant	AD
		Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive	AR
		Immunodeficiency 31A, mycobacteriosis, autosomal dominant	AD
STAT2	600556	Pseudo-TORCH syndrome 3	AR
		Immunodeficiency 44	AR
STIL	181590	Microcephaly 7, primary, autosomal recessive	AR
STIM1	605921	Myopathy, tubular aggregate, 1	AD
		Stormorken syndrome	AD
		Immunodeficiency 10	AR
STRA6	610745	Microphthalmia, isolated, with coloboma 8	AR
		Microphthalmia, syndromic 9	AR
STRADA	608626	Polyhydramnios, megalencephaly, and symptomatic epilepsy	AR
STT3A	601134	Congenital disorder of glycosylation, type Iw, autosomal dominant	AD

		Congenital disorder of glycosylation, type Iw	AR
STUB1	607207	?Spinocerebellar ataxia 48	AD
		Spinocerebellar ataxia, autosomal recessive 16	AR
STX1B	601485	Generalized epilepsy with febrile seizures plus, type 9	AD
STXBP1	602926	Developmental and epileptic encephalopathy 4	AD
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	AR
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	AR
SUGCT	609187	Glutaric aciduria III	AR
SUMF1	607939	Multiple sulfatase deficiency	AR
SUOX	606887	Sulfite oxidase deficiency	AR
SURF1	185620	Charcot-Marie-Tooth disease, type 4K	AR
		Mitochondrial complex IV deficiency, nuclear type 1	AR
SYN1	313440	?Intellectual developmental disorder, X-linked 50	XL
		Epilepsy, X-linked, with variable learning disabilities and behavior disorders	XLD, XLR
SYN2	600755	Schizophrenia, susceptibility to	AD
SYNE1	608441	Arthrogryposis multiplex congenita 3, myogenic type	AR
		Emery-Dreifuss muscular dystrophy 4, autosomal dominant	AD
		Spinocerebellar ataxia, autosomal recessive 8	AR
SYNE2	608442	Emery-Dreifuss muscular dystrophy 5, autosomal dominant	AD

SYNGAP1	603384	Mental retardation, autosomal dominant 5	AD
SYNJ1	604297	Developmental and epileptic encephalopathy 53	AR
		Parkinson disease 20, early-onset	AR
SYP	313475	Mental retardation, X-linked 96	XLR
SYT2	600104	Myasthenic syndrome, congenital, 7B, presynaptic, autosomal recessive	AR
		Myasthenic syndrome, congenital, 7, presynaptic	AD
SZT2	615463	Developmental and epileptic encephalopathy 18	AR
TACO1	612958	Mitochondrial complex IV deficiency, nuclear type 8	AR
TAF1	313650	Mental retardation, X-linked, syndromic 33	XLR
		Dystonia-Parkinsonism, X-linked	XLR
TAF13	600774	Mental retardation, autosomal recessive 60	AR
TAF15	601574	Chondrosarcoma, extraskeletal myxoid	-
TAF2	604912	Mental retardation, autosomal recessive 40	AR
TAF6	602955	Alazami-Yuan syndrome	AR
TAFAZZIN	300394	Barth syndrome	XLR
TANGO2	616830	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	AR
TAOK1	610266	Developmental delay with or without intellectual impairment or behavioral abnormalities	AD
TARDBP	605078	Frontotemporal lobar degeneration, TARDBP-related	AD
		Amyotrophic lateral sclerosis 10, with or without FTD	AD

TARS2	612805	?Combined oxidative phosphorylation deficiency 21	AR
TBC1D20	611663	Warburg micro syndrome 4	AR
TBC1D23	617687	Pontocerebellar hypoplasia, type 11	AR
TBC1D24	613577	Deafness, autosomal dominant 65	AD
		Deafness, autosomal recessive 86	AR
		Myoclonic epilepsy, infantile, familial	AR
		Developmental and epileptic encephalopathy 16	AR
		DOORS syndrome	AR
		Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp	AR
TBCD	604649	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum	AR
TBCE	604934	Hypoparathyroidism-retardation-dysmorphism syndrome	AR
		Encephalopathy, progressive, with amyotrophy and optic atrophy	AR
		Kenny-Caffey syndrome, type 1	AR
TBCK	616899	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	AR
TBK1	604834	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	AD
		Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8	AD
TBL1XR1	608628	Pierpont syndrome	AD
		Mental retardation, autosomal dominant 41	AD
TBR1	604616	Intellectual developmental disorder with autism and speech delay	AD

TBX1	602054	Velocardiofacial syndrome	AD
		Tetralogy of Fallot	AD
		DiGeorge syndrome	AD
		Conotruncal anomaly face syndrome	-
TBX3	601621	Ulnar-mammary syndrome	AD
TCAP	604488	Muscular dystrophy, limb-girdle, autosomal recessive 7	AR
		Cardiomyopathy, hypertrophic, 25	AD
TCF20	603107	Developmental delay with variable intellectual impairment and behavioral abnormalities	AD
TCF4	602272	Pitt-Hopkins syndrome	AD
		Corneal dystrophy, Fuchs endothelial, 3	AD
TCIRG1	604592	Osteopetrosis, autosomal recessive 1	AR
TCOF1	606847	Treacher Collins syndrome 1	AD
TCTN1	609863	Joubert syndrome 13	AR
TCTN2	613846	?Meckel syndrome 8	AR
		Joubert syndrome 24	AR
TCTN3	613847	Joubert syndrome 18	AR
		Orofaciodigital syndrome IV	AR
TDP1	607198	?Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 1	AR
TDP2	605764	Spinocerebellar ataxia, autosomal recessive 23	AR

TECPR2	615000	Spastic paraplegia 49, autosomal recessive	AR
TECR	610057	Mental retardation, autosomal recessive 14	AR
TECTA	602574	Deafness, autosomal dominant 8/12	AD
		Deafness, autosomal recessive 21	AR
TENM4	610084	Essential tremor, hereditary, 5	AD
TET2	612839	Myelodysplastic syndrome, somatic	-
		Immunodeficiency 75	AR
TFAP2A	107580	Branchiooculofacial syndrome	AD
TFAP2B	601601	Patent ductus arteriosus 2	AD
		Char syndrome	AD
TFG	602498	Hereditary motor and sensory neuropathy, Okinawa type	AD
		?Spastic paraplegia 57, autosomal recessive	AR
TFR2	604720	Hemochromatosis, type 3	AR
TG	188450	Thyroid dyshormonogenesis 3	AR
		Autoimmune thyroid disease, susceptibility to, 3	-
TGFB1	190180	Camurati-Engelmann disease	AD
		Cystic fibrosis lung disease, modifier of	AR
		Inflammatory bowel disease, immunodeficiency, and encephalopathy	AR
TGFB3	190230	Loeys-Dietz syndrome 5	AD

		Arrhythmogenic right ventricular dysplasia 1	AD
TGIF1	602630	Holoprosencephaly 4	AD
TGM6	613900	Spinocerebellar ataxia 35	AD
TH	191290	Segawa syndrome, recessive	AR
THAP1	609520	Dystonia 6, torsion	AD
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6	AD
THRB	190160	Thyroid hormone resistance, autosomal recessive	AR
		Thyroid hormone resistance	AD
		Thyroid hormone resistance, selective pituitary	AD
TIA1	603518	Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia	AD
		Welander distal myopathy	AD, AR
TIMM50	607381	3-methylglutaconic aciduria, type IX	AR
TIMM8A	300356	Mohr-Tranebjaerg syndrome	XLR
TIMMDC1	615534	Mitochondrial complex I deficiency, nuclear type 31	AR
TINF2	604319	Revesz syndrome	AD
		Dyskeratosis congenita, autosomal dominant 3	AD
TK2	188250	Mitochondrial DNA depletion syndrome 2 (myopathic type)	AR
		?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3	AR
TLK2	608439	Mental retardation, autosomal dominant 57	AD

TMCO1	614123	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome	AR
TMEM106B	613413	Leukodystrophy, hypomyelinating, 16	AD
TMEM126A	612988	Optic atrophy 7	AR
TMEM126B	615533	Mitochondrial complex I deficiency, nuclear type 29	AR
TMEM138	614459	Joubert syndrome 16	AR
TMEM165	614726	Congenital disorder of glycosylation, type IIk	AR
TMEM199	616815	Congenital disorder of glycosylation, type IIp	AR
TMEM216	613277	Meckel syndrome 2	AR
		Joubert syndrome 2	AR
TMEM231	614949	Joubert syndrome 20	AR
		Meckel syndrome 11	AR
TMEM237	614423	Joubert syndrome 14	AR
TMEM240	616101	Spinocerebellar ataxia 21	AD
TMEM43	612048	Auditory neuropathy, autosomal dominant 3	AD
		Arrhythmogenic right ventricular dysplasia 5	AD
		Emery-Dreifuss muscular dystrophy 7, AD	AD
TMEM67	609884	COACH syndrome 1	AR
		?RHYNS syndrome	AR
		Meckel syndrome 3	AR

		Joubert syndrome 6	AR
		Bardet-Biedl syndrome 14, modifier of	AR
		Nephronophthisis 11	AR
TMEM70	612418	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	AR
TMLHE	300777	Autism, susceptibility to, X-linked 6	XLR
TMTC3	617218	Lissencephaly 8	AR
TMX2	616715	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	AR
TNIK	610005	Mental retardation, autosomal recessive 54	AR
TNNI2	191043	Arthrogryposis, distal, type 2B1	AD
TNNT1	191041	Nemaline myopathy 5, Amish type	AR
TNNT3	600692	Arthrogryposis, distal, type 2B2	AD
TNPO3	610032	Muscular dystrophy, limb-girdle, autosomal dominant 2	AD
TOE1	613931	Pontocerebellar hypoplasia, type 7	AR
TOP3A	601243	Microcephaly, growth restriction, and increased sister chromatid exchange 2	AR
		?Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 5	AR
TOR1A	605204	Dystonia-1, torsion	AD
		Arthrogryposis multiplex congenita 5	AR
TOR1AIP1	614512	?Muscular dystrophy, autosomal recessive, with rigid spine and distal joint contractures	AR
TPI1	190450	Hemolytic anemia due to triosephosphate isomerase deficiency	AR

TPK1	606370	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	AR
TPM2	190990	Arthrogryposis, distal, type 2B4	AD
		Arthrogryposis, distal, type 1A	AD
		CAP myopathy 2	AD
		Nemaline myopathy 4, autosomal dominant	AD
TPM3	191030	Myopathy, congenital, with fiber-type disproportion	AD, AR
		Nemaline myopathy 1, autosomal dominant or recessive	AD, AR
		CAP myopathy 1	AD, AR
TPO	606765	Thyroid dyshormonogenesis 2A	AR
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2	AR
		Spinocerebellar ataxia, autosomal recessive 7	AR
TRAF3IP1	607380	Senior-Loken syndrome 9	AR
TRAF7	606692	Cardiac, facial, and digital anomalies with developmental delay	AD
TRAK1	608112	Developmental and epileptic encephalopathy 68	AR
TRAPPC11	614138	Muscular dystrophy, limb-girdle, autosomal recessive 18	AR
TRAPPC4	610971	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	AR
TRAPPC9	611966	Mental retardation, autosomal recessive 13	AR
TREM2	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2	AR
TREX1	606609	Aicardi-Goutieres syndrome 1, dominant and recessive	AD, AR

		Chilblain lupus	AD
		Vasculopathy, retinal, with cerebral leukodystrophy	AD
		Systemic lupus erythematosus, susceptibility to	AD
TRIM2	614141	Charcot-Marie-Tooth disease, type 2R	AR
TRIM32	602290	Muscular dystrophy, limb-girdle, autosomal recessive 8	AR
		?Bardet-Biedl syndrome 11	AR
TRIM8	606125	Focal segmental glomerulosclerosis and neurodevelopmental syndrome	AD
TRIO	601893	Intellectual developmental disorder, autosomal dominant 44, with microcephaly	AD
		Intellectual developmental disorder, autosomal dominant 63, with macrocephaly	AD
TRIP12	604506	Mental retardation, autosomal dominant 49	AD
TRIP4	604501	Spinal muscular atrophy with congenital bone fractures 1	AR
		?Muscular dystrophy, congenital, Davignon-Chauveau type	AR
TRIT1	617840	Combined oxidative phosphorylation deficiency 35	AR
TRMT10A	616013	Microcephaly, short stature, and impaired glucose metabolism 1	AR
TRMT10C	615423	Combined oxidative phosphorylation deficiency 30	AR
TRMT5	611023	Combined oxidative phosphorylation deficiency 26	AR
TRMU	610230	Liver failure, transient infantile	AR
		Deafness, mitochondrial, modifier of	Mitochondrial
TRNT1	612907	Retinitis pigmentosa and erythrocytic microcytosis	AR

		Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay	AR
TRPC6	603652	Glomerulosclerosis, focal segmental, 2	AD
TRPM1	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive	-
TRPM6	607009	Hypomagnesemia 1, intestinal	AR
TRPS1	604386	Trichorhinophalangeal syndrome, type I	AD
		Trichorhinophalangeal syndrome, type III	AD
TRPV4	605427	SED, Maroteaux type	AD
		Spondylometaphyseal dysplasia, Kozlowski type	AD
		Metatropic dysplasia	AD
		Brachyolmia type 3	AD
		Neuronopathy, distal hereditary motor, type VIII	AD
		[Sodium serum level QTL 1]	-
		?Avascular necrosis of femoral head, primary, 2	AD
		Scapuloperoneal spinal muscular atrophy	AD
		Parastremmatic dwarfism	AD
		Hereditary motor and sensory neuropathy, type IIc	AD
		Digital arthropathy-brachydactyly, familial	AD
TRRAP	603015	Developmental delay with or without dysmorphic facies and autism	AD
		?Deafness, autosomal dominant 75	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
TSEN15	608756	Pontocerebellar hypoplasia, type 2F	AR
TSEN2	608753	Pontocerebellar hypoplasia type 2B	AR
TSEN34	608754	?Pontocerebellar hypoplasia type 2C	AR
TSEN54	608755	Pontocerebellar hypoplasia type 2A	AR
		?Pontocerebellar hypoplasia type 5	AR
		Pontocerebellar hypoplasia type 4	AR
TSFM	604723	Combined oxidative phosphorylation deficiency 3	AR
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4	AR
TSHR	603372	Hyperthyroidism, nonautoimmune	AD
		Hypothyroidism, congenital, nongoitrous, 1	AR
		Hyperthyroidism, familial gestational	AD
TSPAN7	300096	Mental retardation, X-linked 58	XLR
TTBK2	611695	Spinocerebellar ataxia 11	AD

TTC19	613814	Mitochondrial complex III deficiency, nuclear type 2	AR
TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly	AR
		Nephronophthisis 12	AD, AR
TTC8	608132	Bardet-Biedl syndrome 8	AR
		?Retinitis pigmentosa 51	AR
TTI2	614426	Mental retardation, autosomal recessive 39	AR
TTN	188840	Cardiomyopathy, dilated, 1G	-
		Muscular dystrophy, limb-girdle, autosomal recessive 10	AR
		Tibial muscular dystrophy, tardive	AD
		Myopathy, myofibrillar, 9, with early respiratory failure	AD
		Salih myopathy	AR
		Cardiomyopathy, familial hypertrophic, 9	AD
TTPA	600415	Ataxia with isolated vitamin E deficiency	AR
TTR	176300	[Dystransthyretinemic hyperthyroxinemia]	AD
		Carpal tunnel syndrome, familial	AD
		Amyloidosis, hereditary, transthyretin-related	AD
TUBA1A	602529	Lissencephaly 3	AD
TUBA4A	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	AD
TUBA8	605742	Macrothrombocytopenia, isolated, 2, autosomal dominant	-

		Cortical dysplasia, complex, with other brain malformations 8	AR
TUBB2A	615101	Cortical dysplasia, complex, with other brain malformations 5	AD
TUBB2B	612850	Cortical dysplasia, complex, with other brain malformations 7	AD
TUBB3	602661	Cortical dysplasia, complex, with other brain malformations 1	AD
		Fibrosis of extraocular muscles, congenital, 3A	AD
TUBB4A	602662	Dystonia 4, torsion, autosomal dominant	AD
		Leukodystrophy, hypomyelinating, 6	AD
TUBG1	191135	Cortical dysplasia, complex, with other brain malformations 4	AD
TUBGCP4	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3	AR
TUBGCP6	610053	Microcephaly and chorioretinopathy, autosomal recessive, 1	AR
TUFM	602389	Combined oxidative phosphorylation deficiency 4	AR
TUSC3	601385	Mental retardation, autosomal recessive 7	AR
TWIST1	601622	Craniosynostosis 1	AD
		Robinow-Sorauf syndrome	AD
		Saethre-Chotzen syndrome with or without eyelid anomalies	AD
		Sweeney-Cox syndrome	AD
TWNK	606075	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	AD
		Perrault syndrome 5	AR
		Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	AR

TYMP	131222	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	AR
TYR	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin]	AD
		Waardenburg syndrome/albinism, digenic	-
		[Skin/hair/eye pigmentation 3, blue/green eyes]	AD
		Melanoma, cutaneous malignant, susceptibility to, 8	AD
		Albinism, oculocutaneous, type IA	AR
		Albinism, oculocutaneous, type IB	AR
TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1	AR
UBA1	314370	VEXAS syndrome, somatic	-
		Spinal muscular atrophy, X-linked 2, infantile	XLR
UBA5	610552	Developmental and epileptic encephalopathy 44	AR
		?Spinocerebellar ataxia, autosomal recessive 24	AR
UBAP1	609787	Spastic paraplegia 80, autosomal dominant	AD
UBE2A	312180	Mental retardation, X-linked syndromic, Nascimento-type	XLR
UBE3A	601623	Angelman syndrome	AD
UBE3B	608047	Kaufman oculocerebrofacial syndrome	AR
UBQLN2	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	XLD
UBR1	605981	Johanson-Blizzard syndrome	AR
UBTF	600673	Neurodegeneration, childhood-onset, with brain atrophy	AD

UCHL1	191342	?Parkinson disease 5, susceptibility to	AD
		Spastic paraplegia 79, autosomal recessive	AR
UFM1	610553	Leukodystrophy, hypomyelinating, 14	AR
UGP2	191760	Developmental and epileptic encephalopathy 83	AR
UMPS	613891	Orotic aciduria	AR
UNC80	612636	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	AR
UNG	191525	Immunodeficiency with hyper IgM, type 5	AR
UPB1	606673	Beta-ureidopropionase deficiency	AR
UPF3B	300298	Mental retardation, X-linked, syndromic 14	XLR
UQCC2	614461	Mitochondrial complex III deficiency, nuclear type 7	AR
UQCRB	191330	Mitochondrial complex III deficiency, nuclear type 3	AR
UQCRC2	191329	Mitochondrial complex III deficiency, nuclear type 5	AR
UQCRQ	612080	Mitochondrial complex III deficiency, nuclear type 4	AR
UROC1	613012	?Urocanase deficiency	AR
USH2A	608400	Usher syndrome, type 2A	AR
		Retinitis pigmentosa 39	-
USP8	603158	Pituitary adenoma 4, ACTH-secreting, somatic	-
USP9X	300072	Mental retardation, X-linked 99	XLR
		Mental retardation, X-linked 99, syndromic, female-restricted	XLD

USP9Y	400005	Spermatogenic failure, Y-linked, 2	Y-linked
VAMP1	185880	Myasthenic syndrome, congenital, 25	AR
		Spastic ataxia 1, autosomal dominant	AD
VAMP2	185881	Neurodevelopmental disorder with hypotonia and autistic features with or without hyperkinetic movements	AD
VANGL1	610132	Caudal regression syndrome	AD
		Neural tube defects, susceptibility to	AD
VAPB	605704	Spinal muscular atrophy, late-onset, Finkel type	AD
		Amyotrophic lateral sclerosis 8	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	AD
		Charcot-Marie-Tooth disease, type 2Y	AD
		Frontotemporal dementia and/or amyotrophic lateral sclerosis 6	AD
VDR	601769	Rickets, vitamin D-resistant, type IIA	AR
VHL	608537	Pheochromocytoma	AD
		von Hippel-Lindau syndrome	AD
		Renal cell carcinoma, somatic	-
		Erythrocytosis, familial, 2	AR
VIPAS39	613401	Arthrogryposis, renal dysfunction, and cholestasis 2	AR

VLDLR	192977	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	AR
VMA21	300913	Myopathy, X-linked, with excessive autophagy	XLR
VPS11	608549	Leukodystrophy, hypomyelinating, 12	AR
		?Dystonia 32	AR
VPS13A	605978	Choreoacanthocytosis	AR
VPS13B	607817	Cohen syndrome	AR
VPS13C	608879	Parkinson disease 23, autosomal recessive, early onset	AR
VPS13D	608877	Spinocerebellar ataxia, autosomal recessive 4	AR
VPS33B	608552	Arthrogryposis, renal dysfunction, and cholestasis 1	AR
VPS35	601501	Parkinson disease 17	AD
VPS37A	609927	Spastic paraplegia 53, autosomal recessive	AR
VPS53	615850	Pontocerebellar hypoplasia, type 2E	AR
VRK1	602168	Pontocerebellar hypoplasia type 1A	AR
WAC	615049	Desanto-Shinawi syndrome	AD
WARS2	604733	Neurodevelopmental disorder, mitochondrial, with abnormal movements and lactic acidosis, with or without seizures	AR
		Parkinsonism-dystonia 3, childhood-onset	AR
WASF1	605035	Neurodevelopmental disorder with absent language and variable seizures	AD
WASHC4	615748	?Mental retardation, autosomal recessive 43	AR
WASHC5	610657	Spastic paraplegia 8, autosomal dominant	AD

		Ritscher-Schinzel syndrome 1	AR
WDFY3	617485	?Microcephaly 18, primary, autosomal dominant	AD
WDR26	617424	Skraban-Deardorff syndrome	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
WDR62	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	AR
WDR73	616144	Galloway-Mowat syndrome 1	AR
WDR81	614218	Hydrocephalus, congenital, 3, with brain anomalies	AR
		Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2	AR
WFS1	606201	?Cataract 41	AD
		Wolfram-like syndrome, autosomal dominant	AD
		Wolfram syndrome 1	AR
		Diabetes mellitus, noninsulin-dependent, association with	AD
		Deafness, autosomal dominant 6/14/38	AD
WNK1	605232	Neuropathy, hereditary sensory and autonomic, type II	AR
		Pseudohypoaldosteronism, type IIC	AD
WNT1	164820	Osteogenesis imperfecta, type XV	AR
		Osteoporosis, early-onset, susceptibility to, autosomal dominant	-

WNT5A	164975	Robinow syndrome, autosomal dominant 1	AD
WNT7A	601570	Fuhrmann syndrome	AR
		Ulna and fibula, absence of, with severe limb deficiency	AR
WWOX	605131	Esophageal squamous cell carcinoma, somatic	-
		Developmental and epileptic encephalopathy 28	AR
		Spinocerebellar ataxia, autosomal recessive 12	AR
XK	314850	McLeod syndrome with or without chronic granulomatous disease	XL
XPNPEP3	613553	Nephronophthisis-like nephropathy 1	AR
XPR1	605237	Basal ganglia calcification, idiopathic, 6	AD
YAP1	606608	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation	AD
YARS1	603623	Charcot-Marie-Tooth disease, dominant intermediate C	AD
		Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2	AR
YARS2	610957	Myopathy, lactic acidosis, and sideroblastic anemia 2	AR
YWHAG	605356	Developmental and epileptic encephalopathy 56	AD
YY1	600013	Gabriele-de Vries syndrome	AD
ZBTB16	176797	Skeletal defects, genital hypoplasia, and mental retardation	AR
ZBTB18	608433	Mental retardation, autosomal dominant 22	AD
ZBTB20	606025	Primrose syndrome	AD
ZBTB24	614064	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	AR

ZC3H14	613279	Mental retardation, autosomal recessive 56	AR
ZC4H2	300897	Wieacker-Wolff syndrome	XLR
		Wieacker-Wolff syndrome, female-restricted	XLD
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type	XL
ZEB2	605802	Mowat-Wilson syndrome	AD
ZFYVE26	612012	Spastic paraplegia 15, autosomal recessive	AR
ZFYVE27	610243	Spastic paraplegia 33, autosomal dominant	AD
ZIC1	600470	Structural brain anomalies with impaired intellectual development and craniosynostosis	AD
		?Craniosynostosis 6	AD
ZIC2	603073	Holoprosencephaly 5	AD
ZIC3	300265	VACTERL association, X-linked	XLR
		Congenital heart defects, nonsyndromic, 1, X-linked	XLR
		Heterotaxy, visceral, 1, X-linked	XLR
ZMYND11	608668	Mental retardation, autosomal dominant 30	AD
ZNF142	604083	Neurodevelopmental disorder with impaired speech and hyperkinetic movements	AR
ZNF292	616213	Intellectual developmental disorder, autosomal dominant 64	AD
ZNF335	610827	Microcephaly 10, primary, autosomal recessive	AR
ZNF423	604557	Joubert syndrome 19	AD, AR
		Nephronophthisis 14	AD, AR

ZNF699	609571	DEGCAGS syndrome	AR
ZNF711	314990	Mental retardation, X-linked 97	XL
Close			
COMMON SYNDROMES AND DISORDERS COVERED			
Amyotrophic lateral sclerosis			
Arthrogryposis multiplex congenita			
Ataxia			
Dementia			
Dolichoectasia			
Dystonia			
Epilepsy			
Familial hemiplegic migraine			
Frontotemporal dementia			
Hypogonadotropic hypogonadism			
Intellectual disability			
Joubert syndrome			
Kallman syndrome			

Leigh syndrome			
Leukodystrophy and peroxisome biogenesis disorders			
Meckel syndrome			
Mitochondrial encephalomyopathy			
Neonatal mitochondrial hepatopathies			