Panel UCI: para el diagnóstico más temprano y rápido

Está diseñado para diagnosticar afecciones genéticas que se presentan en el período del recién nacido o la primera infancia. Muchos de estos tienen fenotipos superpuestos y el diagnóstico puede tener implicaciones inmediatas para el tratamiento de recién nacidos y niños.

¿Qué es Panel UCI?

Los recién nacidos y los niños menores de 24 meses que presentan afecciones potencialmente mortales necesitan un diagnóstico rápido y preciso para garantizarun diagnóstico y un inicio terapéutico rápidos y eficaces.

Hasta un tercio de todos los bebés y niños ingresados en la UCI tienen una enfermedad genética. Para muchos de ellos, la identificación temprana puede marcar la diferencia para su salud inmediata y posterior.

Panel UCI es un panel completo de NGS que incluye más de 800 genes, seleccionados explícitamente para las pruebas genéticas de recién nacidos en estado crítico y niños menores de 24 meses. Está diseñado para abordar múltiples afecciones genéticas que pueden presentarse en el recién nacido o en el períodode la primera infancia, muchas de ellas con fenotipos superpuestos e implicaciones inmediatas para el inicio del tratamiento. Permite a los médicos utilizar una sola prueba para proporcionar un diagnóstico preciso de las enfermedades relacionadas con el recién nacido.

No. de genes:	855
Entrega:	15 días / 10 días (opción rápida)
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Secuenciación de próxima generación.

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

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
AARS1	*601065*	*Trichothiodystrophy 8, nonphotosensitive;Charcot-Marie-Tooth disease, axonal, type 2N;Developmental and epileptic encephalopathy 29;?Leukoencephalopathy, hereditary diffuse, with spheroids 2*	*AR, AD*
AARS2	*612035*	*Combined oxidative phosphorylation deficiency 8;Leukoencephalopathy, progressive, with ovarian failure*	*AR*
AASS	*605113*	*Hyperlysinemia*	*AR*
ABAT	*137150*	*GABA-transaminase deficiency*	*AR*
ABCA12	*607800*	*Ichthyosis, congenital, AR 4A;Ichthyosis, congenital, AR 4B (harlequin)*	*AR*
ABCA3	*601615*	*Surfactant metabolism dysfunction, pulmonary, 3*	*AR*
ABCB11	*603201*	*Cholestasis, benign recurrent intrahepatic, 2;Cholestasis, progressive familial intrahepatic 2*	*AR*
ABCC8	*600509*	*Diabetes mellitus, transient neonatal 2;Diabetes mellitus, noninsulin-dependent;Hyperinsulinemic hypoglycemia, familial, 1;Hypoglycemia of infancy, leucine-sensitive;Diabetes mellitus, permanent neonatal 3, with or without neurologic features*	*AD, AD, AR*
ABCD1	*300371*	*Adrenoleukodystrophy;Adrenomyeloneuropathy, adult*	*XLR*
ABCD3	*170995*	*?Bile acid synthesis defect, congenital, 5*	*AR*
ABCD4	*603214*	*Methylmalonic aciduria and homocystinuria, cblJ type*	*AR*
ACAD8	*604773*	*Isobutyryl-CoA dehydrogenase deficiency*	*AR*
ACAD9	*611103*	*Mi complex I deficiency, nuclear type 20*	*AR*
ACADM	*607008*	*Acyl-CoA dehydrogenase, medium chain, deficiency of*	*AR*
ACADS	*606885*	*Acyl-CoA dehydrogenase, short-chain, deficiency of*	*AR*
ACADSB	*600301*	*2-methylbutyrylglycinuria*	*AR*
ACADVL	*609575*	*VLCAD deficiency*	*AR*
ACAT1	*607809*	*Alpha-methylacetoacetic aciduria*	*AR*

ACO2	*100850*	*?Optic atrophy 9;Infantile cerebellar-retinal degeneration*	*AR*
ACOX1	*609751*	*Peroxisomal acyl-CoA oxidase deficiency;Mitchell syndrome*	*AR, AD*
ACSF3	*614245*	*Combined malonic and methylmalonic aciduria*	
ACTA1	*102610*	*Nemaline myopathy 3, AD or recessive;?Myopathy, scapulohumeroperoneal;Myopathy, congenital, with fiber-type disproportion 1;Myopathy, actin, congenital, with cores;Myopathy, actin, congenital, with excess of thin myofilaments*	*AD, AR, AD*
ACY1	*104620*	*Aminoacylase 1 deficiency*	*AR*
ADA	*608958*	*Adenosine deaminase deficiency, partial;Severe combined immunodeficiency due to ADA deficiency*	*AR, SM*
ADAMTS13	*604134*	*Thrombotic thrombocytopenic purpura, hereditary*	*AR*
ADAMTSL2	*612277*	*Geleophysic dysplasia 1*	*AR*
ADAR	*146920*	*Aicardi-Goutieres syndrome 6;Dyschromatosis symmetrica hereditaria*	*AR, AD*
ADK	*102750*	*Hypermethioninemia due to adenosine kinase deficiency*	*AR*
ADNP	*611386*	*Helsmoortel-van der Aa syndrome*	*AD*
ADSL	*608222*	*Adenylosuccinase deficiency*	*AR*
AGA	*613228*	*Aspartylglucosaminuria*	*AR*
AGK	*610345*	*Sengers syndrome;Cataract 38, AR*	*AR*
AGL	*610860*	*Glycogen storage disease IIIb;Glycogen storage disease IIIa*	*AR*
AGPAT2	*603100*	*Lipodystrophy, congenital generalized, type 1*	*AR*
AGPS	*603051*	*Rhizomelic chondrodysplasia punctata, type 3*	*AR*
AGRN	*103320*	*Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects*	*AR*
AGXT	*604285*	*Hyperoxaluria, primary, type 1*	*AR*
AHCY	*180960*	*Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase*	*AR*
AICDA	*605257*	*Immunodeficiency with hyper-IgM, type 2*	*AR*

AIFM1	*300169*	*Cowchock syndrome;Combined oxidative phosphorylation deficiency 6;Deafness, XL 5;Spondyloepimetaphyseal dysplasia, XL, with hypomyelinating leukodystrophy*	*XLR*
AIMP1	*603605*	*Leukodystrophy, hypomyelinating, 3*	*AR*
AKAP9	*604001*	*?Long QT syndrome 11*	*AD*
AKR1D1	*604741*	*Bile acid synthesis defect, congenital, 2*	*AR*
AKT2	*164731*	*Diabetes mellitus, type II;Hypoinsulinemic hypoglycemia with hemihypertrophy*	*AD*
ALAD	*125270*	*Porphyria, acute hepatic;Lead poisoning, susceptibility to*	*AR*
ALAS2	*301300*	*Protoporphyria, erythropoietic, XL;Anemia, sideroblastic, 1*	*XL, XLR*
ALDH18A1	*138250*	*Cutis laxa, AD 3;Cutis laxa, AR, type IIIA;Spastic paraplegia 9B, AR;Spastic paraplegia 9A, AD*	*AD, AR*
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;Developmental and epileptic encephalopathy 36*	*XL*
ALG14	*612866*	*Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies;?Myasthenic syndrome, congenital, 15, without tubular aggregates;Myopathy, epilepsy, and progressive cerebral atrophy*	*AR*

ALG2	*607905*	*?Congenital disorder of glycosylation, type Ii;Myasthenic syndrome, congenital, 14, with tubular aggregates*	*AR*
ALG3	*608750*	*Congenital disorder of glycosylation, type Id*	*AR*
ALG6	*604566*	*Congenital disorder of glycosylation, type Ic*	*AR*
ALG8	*608103*	*Polycystic liver disease 3 with or without kidney cysts;Congenital disorder of glycosylation, type Ih*	*AD, AR*
ALG9	*606941*	*Congenital disorder of glycosylation, type Il;Gillessen-Kaesbach-Nishimura syndrome*	*AR*
ALOX12B	*603741*	*Ichthyosis, congenital, AR 2*	*AR*
ALOXE3	*607206*	*Ichthyosis, congenital, AR 3*	*AR*
ALPL	*171760*	*Hypophosphatasia, infantile;Odontohypophosphatasia;Hypophosphatasia, childhood;Hypophosphatasia, adult*	*AR, AD, AR*
ALS2	*606352*	*Amyotrophic lateral sclerosis 2, juvenile;Spastic paralysis, infantile onset ascending;Primary lateral sclerosis, juvenile*	*AR*
AMACR	*604489*	*Bile acid synthesis defect, congenital, 4;Alpha-methylacyl-CoA racemase deficiency*	*AR*
AMN	*605799*	*Imerslund-Grasbeck syndrome 2*	*AR*
AMPD1	*102770*	*Myopathy due to myoadenylate deaminase deficiency*	*AR*
AMT	*238310*	*Glycine encephalopathy*	*AR*
ANK1	*612641*	*Spherocytosis, type 1*	*AD, AR*
ANKRD26	*610855*	*Thrombocytopenia 2*	*AD*
ANKS6	*615370*	*Nephronophthisis 16*	*AR*
ANTXR1	*606410*	*?Hemangioma, capillary infantile, susceptibility to;GAPO syndrome*	*AD, AR*
ANTXR2	*608041*	*Hyaline fibromatosis syndrome*	*AR*
AP2S1	*602242*	*Hypocalciuric hypercalcemia, type III*	*AD*
AP4B1	*607245*	*Spastic paraplegia 47, AR*	*AR*
AP4E1	*607244*	*Stuttering, familial persistent, 1;Spastic paraplegia 51, AR*	*AD, AR*
AP4M1	*602296*	*Spastic paraplegia 50, AR*	*AR*
AP4S1	*607243*	*Spastic paraplegia 52, AR*	*AR*
APOB	*107730*	*Hypobetalipoproteinemia;Hypercholesterolemia, familial, 2*	*AR, AD*

APTX	*606350*	*Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia*	*AR*
ARG1	*608313*	*Argininemia*	*AR*
ARL6	*608845*	*Retinitis pigmentosa 55;Bardet-Biedl syndrome 1, modifier of;Bardet-Biedl syndrome 3*	*AR, AR, DR*
ARSA	*607574*	*Metachromatic leukodystrophy*	*AR*
ARSB	*611542*	*Mucopolysaccharidosis type VI (Maroteaux-Lamy)*	*AR*
ARX	*300382*	*Developmental and epileptic encephalopathy 1;Mental retardation, XL 29 and others;Hydranencephaly with abnormal genitalia;Partington syndrome;Lissencephaly, XL 2;Proud syndrome*	*XLR, XL*
ASAH1	*613468*	*Spinal muscular atrophy with progressive myoclonic epilepsy;Farber lipogranulomatosis*	*AR*
ASL	*608310*	*Argininosuccinic aciduria*	*AR*
ASNS	*108370*	*Asparagine synthetase deficiency*	*AR*
ASPA	*608034*	*Canavan disease*	*AR*
ASPM	*605481*	*Microcephaly 5, primary, AR*	*AR*
ASS1	*603470*	*Citrullinemia*	*AR*
ATIC	*601731*	*AICA-ribosiduria due to ATIC deficiency*	*AR*
ATP1A3	*182350*	*Alternating hemiplegia of childhood 2;Dystonia-12;Developmental and epileptic encephalopathy 99;CAPOS syndrome*	*AD*
ATP6V0A2	*611716*	*Cutis laxa, AR, type IIA;Wrinkly skin syndrome*	*AR*
ATP6V1B1	*192132*	*Distal renal tubular acidosis 2 with progressive sensorineural hearing loss*	*AR*
ATP7A	*300011*	*Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3*	*XLR*
ATP7B	*606882*	*Wilson disease*	*AR*
ATP8B1	*602397*	*Cholestasis, progressive familial intrahepatic 1;Cholestasis, intrahepatic, of pregnancy, 1;Cholestasis, benign recurrent intrahepatic*	*AR, AD*
ATPAF2	*608918*	*?Mi complex V (ATP synthase) deficiency, nuclear type 1*	*AR*

ATR	*601215*	*?Cutaneous telangiectasia and cancer syndrome, familial;Seckel syndrome 1*	*AD, AR*
ATRX	*300032*	*Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic*	*XLR, XLD*
AUH	*600529*	*3-methylglutaconic aciduria, type I*	*AR*
B3GLCT	*610308*	*Peters-plus syndrome*	*AR*
B4GALT1	*137060*	*Congenital disorder of glycosylation, type IId*	*AR*
BCAP31	*300398*	*Deafness, dystonia, and cerebral hypomyelination*	*XLR*
BCKDHA	*608348*	*Maple syrup urine disease, type Ia*	*AR*
BCKDHB	*248611*	*Maple syrup urine disease, type Ib*	*AR*
BCKDK	*614901*	*Branched-chain ketoacid dehydrogenase kinase deficiency*	
BCS1L	*603647*	*GRACILE syndrome;Bjornstad syndrome;Mi complex III deficiency, nuclear type 1*	*AR*
BICD2	*609797*	*Spinal muscular atrophy, lower extremity-predominant, 2A, AD;Spinal muscular atrophy, lower extremity-predominant, 2B, AD*	*AD*
BIN1	*601248*	*Centronuclear myopathy 2*	*AR*
BLNK	*604515*	*?Agammaglobulinemia 4*	*AR*
BOLA3	*613183*	*Multiple Mi dysfunctions syndrome 2 with hyperglycinemia*	*AR*
BRAF	*164757*	*Cardiofaciocutaneous syndrome;Adenocarcinoma of lung, somatic;Noonan syndrome 7;Colorectal cancer, somatic;Melanoma, malignant, somatic,;LEOPARD syndrome 3*	*AD*
BRAT1	*614506*	*Neurodevelopmental disorder with cerebellar atrophy and with or without seizures;Rigidity and multifocal seizure syndrome, lethal neonatal*	*AR*
BRCA2	*600185*	*Prostate cancer;Breast-ovarian cancer, familial, 2;Glioblastoma 3;Wilms tumor;Pancreatic cancer 2;Breast cancer, male, susceptibility to;Fanconi anemia, complementation group D1;Medulloblastoma*	*AD, SM, AD, AR, AD, AR, SM*

BSCL2	*606158*	*Lipodystrophy, congenital generalized, type 2;Encephalopathy, progressive, with or without lipodystrophy;Silver spastic paraplegia syndrome;Neuropathy, distal hereditary motor, type VC*	*AR, AD*
BSND	*606412*	*Bartter syndrome, type 4a;Sensorineural deafness with mild renal dysfunction*	*AR*
BTD	*609019*	*Biotinidase deficiency*	*AR*
BTK	*300300*	*Agammaglobulinemia, XL 1;Isolated growth hormone deficiency, type III, with agammaglobulinemia*	*XLR*
CA12	*603263*	*Hyperchlorhidrosis, isolated*	*AR*
CACNA1C	*114205*	*Long QT syndrome 8;Brugada syndrome 3;Timothy syndrome*	*AD*
CACNB2	*600003*	*Brugada syndrome 4*	
CALM1	*114180*	*Long QT syndrome 14;Ventricular tachycardia, catecholaminergic polymorphic, 4*	*AD*
CAMTA1	*611501*	*Cerebellar ataxia, nonprogressive, with mental retardation*	*AD*
CASK	*300172*	*Mental retardation, with or without nystagmus;Mental retardation and microcephaly with pontine and cerebellar hypoplasia;FG syndrome 4*	*XLD*
CASR	*601199*	*Epilepsy idiopathic generalized, susceptibility to, 8;Hypocalcemia, AD, with Bartter syndrome;Hypocalciuric hypercalcemia, type I;Hyperparathyroidism, neonatal;Hypocalcemia, AD*	*AD, AD, AR*
CAST	*114090*	*Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads*	*AR*
CAV1	*601047*	*Pulmonary hypertension, primary, 3;Lipodystrophy, familial partial, type 7;?Lipodystrophy, congenital generalized, type 3*	*AD, AR*

CAV3	*601253*	*Rippling muscle disease 2;Cardiomyopathy, familial hypertrophic;Creatine phosphokinase, elevated serum;Long QT syndrome 9;Myopathy, distal, Tateyama type*	*AD, AD, DD*
CAVIN1	*603198*	*Lipodystrophy, congenital generalized, type 4*	*AR*
CBS	*613381*	*Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types*	*AR*
CCDC103	*614677*	*Ciliary dyskinesia, primary, 17*	*AR*
CCDC78	*614666*	*?Centronuclear myopathy 4*	*AD*
CD19	*107265*	*Immunodeficiency, common variable, 3*	*AR*
CD247	*186780*	*?Immunodeficiency 25*	*AR*
CD320	*606475*	*Methylmalonic aciduria, transient, due to transcobalamin receptor defect*	
CD3D	*186790*	*Immunodeficiency 19*	*AR*
CD3E	*186830*	*Immunodeficiency 18, SCID variant;Immunodeficiency 18*	*AR*
CD3G	*186740*	*Immunodeficiency 17, CD3 gamma deficient*	*AR*
CD40	*109535*	*Immunodeficiency with hyper-IgM, type 3*	*AR*
CD40LG	*300386*	*Immunodeficiency, XL, with hyper-IgM*	*XLR*
CD59	*107271*	*Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy*	*AR*
CD79A	*112205*	*Agammaglobulinemia 3*	*AR*
CD79B	*147245*	*Agammaglobulinemia 6*	*AR*
CD81	*186845*	*Immunodeficiency, common variable, 6*	*AR*
CD96	*606037*	*C syndrome*	*AD*
CDAN1	*607465*	*Dyserythropoietic anemia, congenital, type Ia*	*AR*
CDCA8	*609977*		
CDK5RAP2	*608201*	*Microcephaly 3, primary, AR*	*AR*
CDKL5	*300203*	*Developmental and epileptic encephalopathy 2*	*XLD*
CDKN1C	*600856*	*IMAGE syndrome;Beckwith-Wiedemann syndrome*	*AD*

CENPJ	*609279*	*?Seckel syndrome 4;Microcephaly 6, primary, AR*	*AR*
CEP152	*613529*	*Microcephaly 9, primary, AR;Seckel syndrome 5*	*AR*
CEP290	*610142*	*Leber congenital amaurosis 10;Meckel syndrome 4;?Bardet-Biedl syndrome 14;Senior-Loken syndrome 6;Joubert syndrome 5*	*AR*
CERS3	*615276*	*Ichthyosis, congenital, AR 9*	*AR*
CFAP298	*615494*	*Ciliary dyskinesia, primary, 26*	*AR*
CFH	*134370*	*Basal laminar drusen;Macular degeneration, age-related, 4;Hemolytic uremic syndrome, atypical, susceptibility to, 1;Complement factor H deficiency*	*AD, AD, AR*
CFHR3	*605336*	*Macular degeneration, age-related, reduced risk of;Hemolytic uremic syndrome, atypical, susceptibility to*	*AD, AD, AR*
CFL2	*601443*	*Nemaline myopathy 7, AR*	*AR*
CFTR	*602421*	*Congenital bilateral absence of vas deferens;Pancreatitis, hereditary;Bronchiectasis with or without elevated sweat chloride 1, modifier of;Cystic fibrosis*	*AR, AD*
CHAT	*118490*	*Myasthenic syndrome, congenital, 6, presynaptic*	*AR*
CHD7	*608892*	*CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia*	*AD*
CHKB	*612395*	*Muscular dystrophy, congenital, megaconial type*	*AR*
CHM	*300390*	*Choroideremia*	*XL*
CHRNA1	*100690*	*Myasthenic syndrome, congenital, 1B, fast-channel;Myasthenic syndrome, congenital, 1A, slow-channel;Multiple pterygium syndrome, lethal type*	*AD, AR, AD, AR*
CHRNB1	*100710*	*Myasthenic syndrome, congenital, 2A, slow-channel;?Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency*	*AD, AR*

CHRND	*100720*	*?Myasthenic syndrome, congenital, 3A, slow-channel;Myasthenic syndrome, congenital, 3B, fast-channel;Multiple pterygium syndrome, lethal type;?Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency*	*AD, AR*
CHRNE	*100725*	*Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency;Myasthenic syndrome, congenital, 4A, slow-channel;Myasthenic syndrome, congenital, 4B, fast-channel*	*AR, AD, AR*
CLCN1	*118425*	*Myotonia congenita, dominant;Myotonia congenita, recessive*	*AD, AR*
CLCNKA	*602024*	*Bartter syndrome, type 4b, digenic*	*DR*
CLCNKB	*602023*	*Bartter syndrome, type 3;Bartter syndrome, type 4b, digenic*	*AR, DR*
CLDN16	*603959*	*Hypomagnesemia 3, renal*	*AR*
CLN3	*607042*	*Ceroid lipofuscinosis, neuronal, 3*	*AR*
CLN5	*608102*	*Ceroid lipofuscinosis, neuronal, 5*	*AR*
CLN6	*606725*	*Ceroid lipofuscinosis, neuronal, Kufs type, adult onset;Ceroid lipofuscinosis, neuronal, 6*	*AR*
CLN8	*607837*	*Ceroid lipofuscinosis, neuronal, 8;Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant*	*AR*
CLPB	*616254*	*3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia*	*AR*
CNTN1	*600016*	*?Myopathy, congenital, Compton-North*	*AR*
COA5	*613920*	*?Mi complex IV, deficiency, nuclear type 9*	*AR*
COG1	*606973*	*Congenital disorder of glycosylation, type IIg*	*AR*
COG6	*606977*	*Shaheen syndrome;Congenital disorder of glycosylation, type III*	*AR*
COG7	*606978*	*Congenital disorder of glycosylation, type IIe*	*AR*
COL11A1	*120280*	*Marshall syndrome;Fibrochondrogenesis 1;Lumbar disc herniation, susceptibility to;?Deafness, AD 37;Stickler syndrome, type II*	*AD, AR*

COL17A1	*113811*	*Epithelial recurrent erosion dystrophy;Epidermolysis bullosa, junctional, localisata variant;Epidermolysis bullosa, junctional, non-Herlitz type*	*AD, AR*
COL1A1	*120150*	*Ehlers-Danlos syndrome, arthrochalasia type, 1;Bone mineral density variation QTL, osteoporosis;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type I;Caffey disease;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1;Osteogenesis imperfecta, type II*	*AD*
COL1A2	*120160*	*Osteoporosis, postmenopausal;Ehlers-Danlos syndrome, cardiac valvular type;Ehlers-Danlos syndrome, arthrochalasia type, 2;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2;Osteogenesis imperfecta, type II*	*AD, AR*
COL2A1	*120140*	*Legg-Calve-Perthes disease;Stickler syndrome, type I;Osteoarthritis with mild chondrodysplasia;Platyspondylic skeletal dysplasia, Torrance type;Spondyloepiphyseal dysplasia, Stanescu type;Kniest dysplasia;Czech dysplasia;Stickler sydrome, type I, nonsyndromic ocular;?Vitreoretinopathy with phalangeal epiphyseal dysplasia;?Epiphyseal dysplasia, multiple, with myopia and deafness;Avascular necrosis of the femoral head;Spondyloperipheral dysplasia;Achondrogenesis, type II or hypochondrogenesis;SMED Strudwick type;SED congenita*	*AD*
COL3A1	*120180*	*Ehlers-Danlos syndrome, vascular type;Polymicrogyria with or without vascular-type EDS*	*AD, AR*

COL5A2	*120190*	*Ehlers-Danlos syndrome, classic type, 2*	*AD*
COL6A1	*120220*	*Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1*	*AD, AR*
COL6A2	*120240*	*Bethlem myopathy 1;Ullrich congenital muscular dystrophy 1;?Myosclerosis, congenital*	*AD, AR, AR*
COL6A3	*120250*	*Dystonia 27;Ullrich congenital muscular dystrophy 1;Bethlem myopathy 1*	*AR, AD, AR*
COL7A1	*120120*	*Epidermolysis bullosa dystrophica, AR;Epidermolysis bullosa dystrophica, AD;Epidermolysis bullosa pruriginosa;Transient bullous of the newborn;Epidermolysis bullosa, pretibial;Toenail dystrophy, isolated;EBD, Bart type;EBD inversa*	*AR, AD, AD, AR*
COLQ	*603033*	*Myasthenic syndrome, congenital, 5*	*AR*
COMP	*600310*	*Carpal tunnel syndrome 2;Epiphyseal dysplasia, multiple, 1;Pseudoachondroplasia*	*AD*
COQ2	*609825*	*Coenzyme Q10 deficiency, primary, 1;Multiple system atrophy, susceptibility to*	*AR, AD, AR*
COQ8A	*606980*	*Coenzyme Q10 deficiency, primary, 4*	*AR*
COQ9	*612837*	*Coenzyme Q10 deficiency, primary, 5*	*AR*
CORO1A	*605000*	*Immunodeficiency 8*	*AR*
COX10	*602125*	*Mi complex IV deficiency, nuclear type 3*	*AR*
COX15	*603646*	*Mi complex IV deficiency, nuclear type 6*	*AR*
COX20	*614698*	*Mi complex IV deficiency, nuclear type 11*	*AR*
COX6B1	*124089*	*Mi complex IV deficiency, nuclear type 7*	*AR*
CPS1	*608307*	*Carbamoylphosphate synthetase I deficiency;Pulmonary hypertension, neonatal, susceptibility to*	*AR*
CPT1A	*600528*	*CPT deficiency, hepatic, type IA*	*AR*

CPT2	*600650*	*CPT II deficiency, myopathic, stress-induced;CPT II deficiency, infantile;Encephalopathy, acute, infection-induced, 4, susceptibility to;CPT II deficiency, lethal neonatal*	*AD, AR, AR*
CR2	*120650*	*Immunodeficiency, common variable, 7;Systemic lupus erythematosus, susceptibility to, 9*	*AR*
CRPPA	*614631*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7*	*AR*
CRTAP	*605497*	*Osteogenesis imperfecta, type VII*	*AR*
CTNS	*606272*	*Cystinosis, late-onset juvenile or adolescent nephropathic;Cystinosis, ocular nonnephropathic;Cystinosis, nephropathic;Cystinosis, atypical nephropathic*	*AR*
CTPS1	*123860*	*Immunodeficiency 24*	*AR*
CTSA	*613111*	*Galactosialidosis*	*AR*
CTSD	*116840*	*Ceroid lipofuscinosis, neuronal, 10*	*AR*
CUL4B	*300304*	*Mental retardation, XL, syndromic 15 (Cabezas type)*	*XLR*
CXCR4	*162643*	*WHIM syndrome*	*AD*
CYP11B1	*610613*	*Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency;Aldosteronism, glucocorticoid-remediable*	*AR, AD*
CYP11B2	*124080*	*Hypoaldosteronism, congenital, due to CMO II deficiency;Hypoaldosteronism, congenital, due to CMO I deficiency*	*AR*
CYP17A1	*609300*	*17,20-lyase deficiency, isolated;17-alpha-hydroxylase/17,20-lyase deficiency*	*AR*
CYP4F22	*611495*	*Ichthyosis, congenital, AR 5*	*AR*
CYP7B1	*603711*	*Spastic paraplegia 5A, AR;Bile acid synthesis defect, congenital, 3*	*AR*
D2HGDH	*609186*	*D-2-hydroxyglutaric aciduria*	*AR*
DBT	*248610*	*Maple syrup urine disease, type II*	*AR*
DCLRE1C	*605988*	*Omenn syndrome;Severe combined immunodeficiency, Athabascan type*	*AR*

DDC	*107930*	*Aromatic L-amino acid decarboxylase deficiency*	*AR*
DDOST	*602202*	*?Congenital disorder of glycosylation, type Ir*	*AR*
DDR2	*191311*	*Spondylometaepiphyseal dysplasia, short limb-hand type;Warburg-Cinotti syndrome*	*AR, AD*
DEPDC5	*614191*	*Epilepsy, familial focal, with variable foci 1*	*AD*
DES	*125660*	*Cardiomyopathy, dilated, 1I;Myopathy, myofibrillar, 1;Scapuloperoneal syndrome, neurogenic, Kaeser type*	*AD, AD, AR*
DGUOK	*601465*	*Portal hypertension, noncirrhotic;Progressive external ophthalmoplegia with Mi DNA deletions, AR 4;Mi DNA depletion syndrome 3 (hepatocerebral type)*	*AR*
DHCR24	*606418*	*Desmosterolosis*	*AR*
DHCR7	*602858*	*Smith-Lemli-Opitz syndrome*	*AR*
DIAPH1	*602121*	*Seizures, cortical blindness, microcephaly syndrome;Deafness, AD 1, with or without thrombocytopenia*	*AR, AD*
DLAT	*608770*	*Pyruvate dehydrogenase E2 deficiency*	*AR*
DLD	*238331*	*Dihydrolipoamide dehydrogenase deficiency*	*AR*
DMD	*300377*	*Cardiomyopathy, dilated, 3B;Duchenne muscular dystrophy;Becker muscular dystrophy*	*XL, XLR*
DNA2	*601810*	*?Seckel syndrome 8;Progressive external ophthalmoplegia with Mi DNA deletions, AD 6*	*AR, AD*
DNAH11	*603339*	*Ciliary dyskinesia, primary, 7, with or without situs inversus*	*AR*
DNAH5	*603335*	*Ciliary dyskinesia, primary, 3, with or without situs inversus*	*AR*
DNAI1	*604366*	*Ciliary dyskinesia, primary, 1, with or without situs inversus*	*AR*
DNAI2	*605483*	*Ciliary dyskinesia, primary, 9, with or without situs inversus*	*AR*
DNAJC19	*608977*	*3-methylglutaconic aciduria, type V*	*AR*

DNM2	*602378*	*Lethal congenital contracture syndrome 5;Charcot-Marie-Tooth disease, axonal type 2M;Charcot-Marie-Tooth disease, dominant intermediate B;Centronuclear myopathy 1*	*AR, AD*
DOCK7	*615730*	*Developmental and epileptic encephalopathy 23*	*AR*
DOCK8	*611432*	*Hyper-IgE recurrent infection syndrome, AR*	*AR*
DOK7	*610285*	*Fetal akinesia deformation sequence 3;Myasthenic syndrome, congenital, 10*	*AR*
DOLK	*610746*	*Congenital disorder of glycosylation, type Im*	*AR*
DPAGT1	*191350*	*Myasthenic syndrome, congenital, 13, with tubular aggregates;Congenital disorder of glycosylation, type Ij*	*AR*
DPM2	*603564*	*Congenital disorder of glycosylation, type Iu*	*AR*
DPYD	*612779*	*5-fluorouracil toxicity;Dihydropyrimidine dehydrogenase deficiency*	*AR*
DRC1	*615288*	*Ciliary dyskinesia, primary, 21*	*AR*
DSP	*125647*	*Cardiomyopathy, dilated, with woolly hair and keratoderma;Arrhythmogenic right ventricular dysplasia 8;Keratosis palmoplantaris striata II;Skin fragility-woolly hair syndrome;Epidermolysis bullosa, lethal acantholytic;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis*	*AR, AD*
DST	*113810*	*?Neuropathy, hereditary sensory and autonomic, type VI;Epidermolysis bullosa simplex, AR 2*	*AR*
DUOX2	*606759*	*Thyroid dyshormonogenesis 6*	*AR*
DUOXA2	*612772*	*Thyroid dyshormonogenesis 5*	*AR*
DYSF	*603009*	*Muscular dystrophy, limb-girdle, AR 2;Myopathy, distal, with anterior tibial onset;Miyoshi muscular dystrophy 1*	*AR*

EDN3	*131242*	*Waardenburg syndrome, type 4B;Hirschsprung disease, susceptibility to, 4;Central hypoventilation syndrome, congenital*	*AD, AR, AD*
EEF1A2	*602959*	*Mental retardation, AD 38;Developmental and epileptic encephalopathy 33*	*AD*
EGR2	*129010*	*Dejerine-Sottas disease;Hypomyelinating neuropathy, congenital, 1;Charcot-Marie-Tooth disease, type 1D*	*AD, AR, AD*
EIF2AK3	*604032*	*Wolcott-Rallison syndrome*	*AR*
EIF2B1	*606686*	*Leukoencephalopathy with vanishing white matter*	*AR*
EIF2B2	*606454*	*Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter*	*AR*
EIF2B3	*606273*	*Leukoencephalopathy with vanishing white matter*	*AR*
EIF2B4	*606687*	*Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter*	*AR*
EIF2B5	*603945*	*Ovarioleukodystrophy;Leukoencephalopathy with vanishing white matter*	*AR*
ELAC2	*605367*	*Prostate cancer, hereditary, 2, susceptibility to;Combined oxidative phosphorylation deficiency 17*	*AR*
ELANE	*130130*	*Neutropenia, cyclic;Neutropenia, severe congenital 1, AD*	*AD*
ENPP1	*173335*	*Arterial calcification, generalized, of infancy, 1;Obesity, susceptibility to;Hypophosphatemic rickets, AR, 2;Cole disease;Diabetes mellitus, non-insulin-dependent, susceptibility to*	*AR, AD, AR, mi, AD*
EPB42	*177070*	*Spherocytosis, type 5*	
EPCAM	*185535*	*Colorectal cancer, hereditary nonpolyposis, type 8;Diarrhea 5, with tufting enteropathy, congenital*	*AR*
ETFA	*608053*	*Glutaric acidemia IIA*	*AR*
ETFB	*130410*	*Glutaric acidemia IIB*	*AR*
ETFDH	*231675*	*Glutaric acidemia IIC*	*AR*
ETHE1	*608451*	*Ethylmalonic encephalopathy*	*AR*

EVC	*604831*	*Ellis-van Creveld syndrome;?Weyers acrofacial dysostosis*	*AR, AD*
EVC2	*607261*	*Ellis-van Creveld syndrome;Weyers acrofacial dysostosis*	*AR, AD*
EXOSC3	*606489*	*Pontocerebellar hypoplasia, type 1B*	*AR*
EYA1	*601653*	*Anterior segment anomalies with or without cataract;Branchiootorenal syndrome 1, with or without cataracts;?Otofaciocervical syndrome;Branchiootic syndrome 1*	*AD*
EYA4	*603550*	*Deafness, AD 10;?Cardiomyopathy, dilated, 1J*	*AD*
F10	*613872*	*Factor X deficiency*	*AR*
F11	*264900*	*Factor XI deficiency, AD;Factor XI deficiency, AR*	
F13A1	*134570*	*Factor XIIIA deficiency;Venous thrombosis, protection against;Myocardial infarction, protection against*	*AR, AD*
F2	*176930*	*Dysprothrombinemia;Stroke, ischemic, susceptibility to;Pregnancy loss, recurrent, susceptibility to, 2;Hypoprothrombinemia;Thrombophilia due to thrombin defect*	*AR, mi, AD*
F5	*612309*	*Pregnancy loss, recurrent, susceptibility to, 1;Factor V deficiency;Stroke, ischemic, susceptibility to;Budd-Chiari syndrome;Thrombophilia, susceptibility to, due to factor V Leiden;Thrombophilia due to activated protein C resistance*	*AD, AR, mi*
F7	*613878*	*Myocardial infarction, decreased susceptibility to;Factor VII deficiency*	*AR*
F8	*300841*	*Hemophilia A*	*XLR*
F9	*300746*	*Warfarin sensitivity;Thrombophilia, XL, due to factor IX defect;Hemophilia B;Deep venous thrombosis, protection against*	*XL, XLR*

FADD	*602457*	*Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations*	*AR*
FAH	*613871*	*Tyrosinemia, type I*	*AR*
FANCA	*607139*	*Fanconi anemia, complementation group A*	*AR*
FANCB	*300515*	*Fanconi anemia, complementation group B*	*XLR*
FANCC	*613899*	*Fanconi anemia, complementation group C*	*AR*
FANCD2	*613984*	*Fanconi anemia, complementation group D2*	*AR*
FANCL	*608111*	*Fanconi anemia, complementation group L*	*AR*
FARS2	*611592*	*Combined oxidative phosphorylation deficiency 14;Spastic paraplegia 77, AR*	*AR*
FASTKD2	*612322*	*Combined oxidative phosphorylation deficiency 44*	*AR*
FBN1	*134797*	*Marfan lipodystrophy syndrome;Geleophysic dysplasia 2;Acromicric dysplasia;Marfan syndrome;Weill-Marchesani syndrome 2, dominant;Stiff skin syndrome;MASS syndrome;Ectopia lentis, familial*	*AD*
FBP1	*611570*	*Fructose-1,6-bisphosphatase deficiency*	*AR*
FBXL4	*605654*	*Mi DNA depletion syndrome 13 (encephalomyopathic type)*	*AR*
FGA	*134820*	*Hypodysfibrinogenemia, congenital;Afibrinogenemia, congenital;Amyloidosis, familial visceral;Dysfibrinogenemia, congenital*	*AR, AD*
FGB	*134830*	*Afibrinogenemia, congenital;Dysfibrinogenemia, congenital;Hypofibrinogenemia, congenital*	*AR*

FGFR2	*176943*	*Crouzon syndrome;Saethre-Chotzen syndrome;Craniofacial-skeletal-dermatologic dysplasia;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis;Gastric cancer, somatic;LADD syndrome;Beare-Stevenson cutis gyrata syndrome;Scaphocephaly, maxillary retrusion, and mental retardation;Apert syndrome;Bent bone dysplasia syndrome;Pfeiffer syndrome;Jackson-Weiss syndrome*	*AD*
FGFR3	*134934*	*Achondroplasia;CATSHL syndrome;Thanatophoric dysplasia, type I;Bladder cancer, somatic;Hypochondroplasia;SADDAN;Colorectal cancer, somatic;Crouzon syndrome with acanthosis nigricans;Cervical cancer, somatic;Nevus, epidermal, somatic;Thanatophoric dysplasia, type II;Spermatocytic seminoma, somatic;Muenke syndrome;LADD syndrome*	*AD, AD, AR*
FGG	*134850*	*Dysfibrinogenemia, congenital;Afibrinogenemia, congenital;Hypofibrinogenemia, congenital;Hypodysfibrinogenemia*	*AR*
FH	*136850*	*Fumarase deficiency;Leiomyomatosis and renal cell cancer*	*AR, AD*
FIG4	*609390*	*Amyotrophic lateral sclerosis 11;Yunis-Varon syndrome;Charcot-Marie-Tooth disease, type 4J;?Polymicrogyria, bilateral temporooccipital*	*AD, AR*
FKBP14	*614505*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 2*	*AR*

FKRP	*606596*	*Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5*	*AR*
FKTN	*607440*	*Cardiomyopathy, dilated, 1X;Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4*	*AR*
FOXC1	*601090*	*Anterior segment dysgenesis 3, multiple subtypes;Axenfeld-Rieger syndrome, type 3*	*AD*
FOXE1	*602617*	*Bamforth-Lazarus syndrome;Thyroid cancer, nonmedullary, 4*	*AR, AD*
FOXG1	*164874*	*Rett syndrome, congenital variant*	*AD*
FOXP3	*300292*	*Immunodysregulation, polyendocrinopathy, and enteropathy, XL*	*XLR*
FOXRED1	*613622*	*Mi complex I deficiency, nuclear type 19*	*AR*
FRAS1	*607830*	*Fraser syndrome 1*	*AR*
FUCA1	*612280*	*Fucosidosis*	*AR*
G6PD	*305900*	*Resistance to malaria due to G6PD deficiency;Hemolytic anemia, G6PD deficient (favism)*	*XLD*
GAA	*606800*	*Glycogen storage disease II*	*AR*
GALC	*606890*	*Krabbe disease*	*AR*
GALE	*606953*	*Galactose epimerase deficiency*	*AR*
GALK1	*604313*	*Galactokinase deficiency with cataracts*	*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;Charcot-Marie-Tooth disease, type 2D;Neuronopathy, distal hereditary motor, type VA*	*AD*
GATA1	*305371*	*Thrombocytopenia with beta-thalassemia, XL;Thrombocytopenia, XL, with or without dyserythropoietic anemia;Anemia, XL, with/without neutropenia and/or platelet abnormalities;Leukemia, megakaryoblastic, with or without Down syndrome, somatic*	*XLR*
GATM	*602360*	*Cerebral creatine deficiency syndrome 3;Fanconi renotubular syndrome 1*	*AR, AD*
GBA	*606463*	*Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I*	*AD, AR, AD, mi*
GBE1	*607839*	*Glycogen storage disease IV;Polyglucosan body disease, adult form*	*AR*
GCDH	*608801*	*Glutaricaciduria, type I*	*AR*
GCH1	*600225*	*Dystonia, DOPA-responsive, with or without hyperphenylalaninemia;Hyperphenylalaninemia, BH4-deficient, B*	*AD, AR, AR*
GCK	*138079*	*Diabetes mellitus, permanent neonatal 1;Diabetes mellitus, noninsulin-dependent, late onset;Hyperinsulinemic hypoglycemia, familial, 3;MODY, type II*	*AR, AD*
GCSH	*238330*	*?Glycine encephalopathy*	*AR*
GDAP1	*606598*	*Charcot-Marie-Tooth disease, axonal, type 2K;Charcot-Marie-Tooth disease, type 4A;Charcot-Marie-Tooth disease, axonal, with vocal cord paresis;Charcot-Marie-Tooth disease, recessive intermediate, A*	*AD, AR, AR*
GFAP	*137780*	*Alexander disease*	*AD*
GFM1	*606639*	*Combined oxidative phosphorylation deficiency 1*	*AR*

GFPT1	*138292*	*Myasthenia, congenital, 12, with tubular aggregates*	*AR*
GJA1	*121014*	*Oculodentodigital dysplasia, AR;Atrioventricular septal defect 3;Syndactyly, type III;Craniometaphyseal dysplasia, AR;Palmoplantar keratoderma with congenital alopecia;Oculodentodigital dysplasia;Hypoplastic left heart syndrome 1;Erythrokeratodermia variabilis et progressiva 3*	*AR, AD*
GJB2	*121011*	*Keratoderma, palmoplantar, with deafness;Keratitis-ichthyosis-deafness syndrome;Deafness, AD 3A;Hystrix-like ichthyosis with deafness;Bart-Pumphrey syndrome;Vohwinkel syndrome;Deafness, AR 1A*	*AD, AR, DD*
GJB4	*605425*	*Erythrokeratodermia variabilis et progressiva 2*	*AD*
GK	*300474*	*Glycerol kinase deficiency*	*XLR*
GLA	*300644*	*Fabry disease;Fabry disease, cardiac variant*	*XL*
GLB1	*611458*	*GM1-gangliosidosis, type II;GM1-gangliosidosis, type I;Mucopolysaccharidosis type IVB (Morquio);GM1-gangliosidosis, type III*	*AR*
GLDC	*238300*	*Glycine encephalopathy*	*AR*
GLIS3	*610192*	*Diabetes mellitus, neonatal, with congenital hypothyroidism*	*AR*
GLRA1	*138491*	*Hyperekplexia 1*	*AD, AR*
GLRB	*138492*	*Hyperekplexia 2*	*AR*
GLUD1	*138130*	*Hyperinsulinism-hyperammonemia syndrome*	*AD*
GLYCTK	*610516*	*D-glyceric aciduria*	*AR*
GMPPB	*615320*	*Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14*	*AR*

GNAS	*139320*	*Pseudohypoparathyroidism Ic;Osseous heteroplasia, progressive;Pseudopseudohypoparathyroidism;Pseudohypoparathyroidism Ia;Pseudohypoparathyroidism Ib;Pituitary adenoma 3, multiple types, somatic;ACTH-independent macronodular adrenal hyperplasia;McCune-Albright syndrome, somatic, mosaic*	*AD, SM*
GNE	*603824*	*Nonaka myopathy;Sialuria*	*AR, AD*
GNMT	*606628*	*Glycine N-methyltransferase deficiency*	*AR*
GNPAT	*602744*	*Rhizomelic chondrodysplasia punctata, type 2*	*AR*
GNPTAB	*607840*	*Mucolipidosis III alpha/beta;Mucolipidosis II alpha/beta*	*AR*
GP1BA	*606672*	*Bernard-Soulier syndrome, type A1 (recessive);von Willebrand disease, platelet-type;Bernard-Soulier syndrome, type A2 (dominant);Nonarteritic anterior ischemic optic neuropathy, susceptibility to*	*AR, AD*
GP1BB	*138720*	*Giant platelet disorder, isolated;Bernard-Soulier syndrome, type B*	*AR*
GP9	*173515*	*Bernard-Soulier syndrome, type C*	*AR*
GPC3	*300037*	*Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic*	*XLR*
GPHN	*603930*	*Molybdenum cofactor deficiency C*	*AR*
GPSM2	*609245*	*Chudley-McCullough syndrome*	*AR*
GSS	*601002*	*Glutathione synthetase deficiency;Hemolytic anemia due to glutathione synthetase deficiency*	*AR*
GUSB	*611499*	*Mucopolysaccharidosis VII*	*AR*
GYS2	*138571*	*Glycogen storage disease 0, liver*	*AR*
HADH	*601609*	*3-hydroxyacyl-CoA dehydrogenase deficiency;Hyperinsulinemic hypoglycemia, familial, 4*	*AR*

HADHA	*600890*	*HELLP syndrome, maternal, of pregnancy;LCHAD deficiency;Fatty liver, acute, of pregnancy;Mi trifunctional protein deficiency*	*AR*
HADHB	*143450*	*Trifunctional protein deficiency*	*AR*
HAMP	*606464*	*Hemochromatosis, type 2B*	*AR*
HAX1	*605998*	*Neutropenia, severe congenital 3, AR*	*AR*
HBA1	*141800*	*Methemoglobinemia, alpha type;Heinz body anemias, alpha-;Erythrocytosis 7;Thalassemias, alpha-;Hemoglobin H disease, nondeletional*	*AD*
HBA2	*141850*	*Thalassemia, alpha-;Erythrocytosis 7;Heinz body anemia;Hemoglobin H disease, deletional and nondeletional*	*AD*
HBB	*141900*	*Erythrocytosis 6;Methemoglobinemia, beta type;Delta-beta thalassemia;Thalassemia-beta, dominant inclusion-body;Sickle cell anemia;Hereditary persistence of fetal hemoglobin;Malaria, resistance to;Thalassemia, beta;Heinz body anemia*	*AD, AR*
HCFC1	*300019*	*Mental retardation, XL 3 (methylmalonic acidemia and homocysteinemia, cblX type)*	*XLR*
HESX1	*601802*	*Septooptic dysplasia;Pituitary hormone deficiency, combined, 5;Growth hormone deficiency with pituitary anomalies*	*AD, AR*
HEXA	*606869*	*Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms*	*AR*
HEXB	*606873*	*Sandhoff disease, infantile, juvenile, and adult forms*	*AR*
HGD	*607474*	*Alkaptonuria*	*AR*
HGF	*142409*	*Deafness, AR 39*	*AR*
HIBCH	*610690*	*3-hydroxyisobutryl-CoA hydrolase deficiency*	*AR*
HLCS	*609018*	*Holocarboxylase synthetase deficiency*	*AR*
HMGCL	*613898*	*HMG-CoA lyase deficiency*	*AR*
HMGCS2	*600234*	*HMG-CoA synthase-2 deficiency*	*AR*

HNF1A	*142410*	*Diabetes mellitus, insulin-dependent;Diabetes mellitus, insulin-dependent, 20;MODY, type III;Diabetes mellitus, noninsulin-dependent, 2;Hepatic adenoma, somatic;Renal cell carcinoma*	*AR, AD*
HNF1B	*189907*	*Renal cysts and diabetes syndrome;Diabetes mellitus, noninsulin-dependent;Renal cell carcinoma*	*AD*
HNF4A	*600281*	*Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young;Diabetes mellitus, noninsulin-dependent;MODY, type I*	*AD*
HPD	*609695*	*Hawkinsinuria;Tyrosinemia, type III*	*AD, AR*
HPGD	*601688*	*Cranioosteoarthropathy;?Digital clubbing, isolated congenital;Hypertrophic osteoarthropathy, primary, AR 1*	*AR*
HRAS	*190020*	*Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles*	*AD*
HSD17B10	*300256*	*HSD10 Mi disease*	*XLD*
HSD17B4	*601860*	*Perrault syndrome 1;D-bifunctional protein deficiency*	*AR*
HSD3B2	*613890*	*Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency*	*AR*
HSD3B7	*607764*	*Bile acid synthesis defect, congenital, 1*	*AR*
HSPA9	*600548*	*Even-plus syndrome;Anemia, sideroblastic, 4*	*AR, AD*
HSPD1	*118190*	*Leukodystrophy, hypomyelinating, 4;Spastic paraplegia 13, AD*	*AR, AD*
HSPG2	*142461*	*Schwartz-Jampel syndrome, type 1;Dyssegmental dysplasia, Silverman-Handmaker type*	*AR*
ICOS	*604558*	*Immunodeficiency, common variable, 1*	*AR*

IDUA	*252800*	*Mucopolysaccharidosis Is;Mucopolysaccharidosis Ih/s;Mucopolysaccharidosis Ih*	*AR*
IER3IP1	*609382*	*Microcephaly, epilepsy, and diabetes syndrome*	*AR*
IFIH1	*606951*	*Aicardi-Goutieres syndrome 7;Singleton-Merten syndrome 1*	*AD*
IFT172	*607386*	*Retinitis pigmentosa 71;Bardet-Biedl syndrome 20;Short-rib thoracic dysplasia 10 with or without polydactyly*	*AR*
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;Charcot-Marie-Tooth disease, axonal, type 2S*	*AR*
IGLL1	*146770*	*Agammaglobulinemia 2*	*AR*
IGSF1	*300137*	*Hypothyroidism, central, and testicular enlargement*	*XLR*
IKBKB	*603258*	*Immunodeficiency 15B;Immunodeficiency 15A*	*AR, AD*
IL12RB1	*601604*	*Immunodeficiency 30*	*AR*
IL2RA	*147730*	*Diabetes, mellitus, insulin-dependent, susceptibility to, 10;Immunodeficiency 41 with lymphoproliferation and autoimmunity*	*AR*
IL2RG	*308380*	*Severe combined immunodeficiency, XL;Combined immunodeficiency, XL, moderate*	*XLR*
IL7R	*146661*	*Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type*	*AR*
INS	*176730*	*Diabetes mellitus, insulin-dependent, 2;Maturity-onset diabetes of the young, type 10;Hyperproinsulinemia;Diabetes mellitus, permanent neonatal 4*	*AD, AD, AR*
INSR	*147670*	*Diabetes mellitus, insulin-resistant, with acanthosis nigricans;Rabson-Mendenhall syndrome;Hyperinsulinemic hypoglycemia, familial, 5;Leprechaunism*	*AR, AD*

INVS	*243305*	*Nephronophthisis 2, infantile*	*AR*
IRF8	*601565*	*Immunodeficiency 32B, monocyte and dendritic cell deficiency, AR;Immunodeficiency 32A, mycobacteriosis, AD*	*AR, AD*
IRS4	*300904*	*Hypothyroidism, congenital, nongoitrous, 9*	*XLR*
ITGA2B	*607759*	*Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD*	*AR, AD*
ITGA6	*147556*	*Epidermolysis bullosa, junctional, with pyloric stenosis*	*AR*
ITGA7	*600536*	*Muscular dystrophy, congenital, due to ITGA7 deficiency*	*AR*
ITGB3	*173470*	*Glanzmann thrombasthenia 2;Glanzmann thrombasthenia;Bleeding disorder, platelet-type, 16, AD;Myocardial infarction, susceptibility to;Bleeding disorder, platelet-type, 24, AD*	*AR, AD*
ITGB4	*147557*	*Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa of hands and feet;Epidermolysis bullosa, junctional, with pyloric atresia*	*AR, AD*
IVD	*607036*	*Isovaleric acidemia*	*AR*
IYD	*612025*	*Thyroid dyshormonogenesis 4*	*AR*
JAG1	*601920*	*Alagille syndrome 1;Charcot-Marie-Tooth disease, axonal, type 2HH;?Deafness, congenital heart defects, and posterior embryotoxon;Tetralogy of Fallot*	*AD*
JAGN1	*616012*	*Neutropenia, severe congenital, 6, AR*	*AR*
JAK3	*600173*	*SCID, AR, T-negative/B-positive type*	*AR*
JAM3	*606871*	*Hemorrhagic destruction of the brain, subependymal calcification, and cataracts*	*AR*
KAT6A	*601408*	*Arboleda-Tham syndrome*	*AD*
KAT6B	*605880*	*SBBYSS syndrome;Genitopatellar syndrome*	*AD*
KBTBD13	*613727*	*Nemaline myopathy 6, AD*	*AD*
KCNE1	*176261*	*Jervell and Lange-Nielsen syndrome 2;Long QT syndrome 5*	*AR, AD*
KCNH1	*603305*	*Zimmermann-Laband syndrome 1;Temple-Baraitser syndrome*	*AD*

KCNH2	*152427*	*Long QT syndrome 2;Short QT syndrome 1;Long QT syndrome 2, acquired, susceptibility to*	*AD*
KCNJ10	*602208*	*Enlarged vestibular aqueduct, digenic;SESAME syndrome*	*AR*
KCNJ11	*600937*	*Maturity-onset diabetes of the young, type 13;Diabetes mellitus, transient neonatal 3;Hyperinsulinemic hypoglycemia, familial, 2;Diabetes, permanent neonatal 2, with or without neurologic features;Diabetes mellitus, type 2, susceptibility to*	*AD, AD, AR*
KCNQ1	*607542*	*Long QT syndrome 1, acquired, susceptibility to;Jervell and Lange-Nielsen syndrome;Atrial fibrillation, familial, 3;Short QT syndrome 2;Long QT syndrome 1*	*AD, AR*
KCNQ2	*602235*	*Seizures, benign neonatal, 1;Developmental and epileptic encephalopathy 7;Myokymia*	*AD*
KCNQ3	*602232*	*Seizures, benign neonatal, 2*	*AD*
KCNT1	*608167*	*Developmental and epileptic encephalopathy 14;Epilepsy nocturnal frontal lobe, 5*	*AD*
KCTD7	*611725*	*Epilepsy, progressive myoclonic 3, with or without intracellular inclusions*	*AR*
KIF1B	*605995*	*?Charcot-Marie-Tooth disease, type 2A1;Pheochromocytoma;Neuroblastoma, susceptibility to, 1*	*AD, AD, SM*
KLF1	*600599*	*Dyserythropoietic anemia, congenital, type IV;Blood group--Lutheran inhibitor;[Hereditary persistence of fetal hemoglobin]*	*AD*
KLHL40	*615340*	*Nemaline myopathy 8, AR*	*AR*
KLHL41	*607701*	*Nemaline myopathy 9*	*AR*
KLHL7	*611119*	*PERCHING syndrome;Retinitis pigmentosa 42*	*AR, AD*

KRAS	*190070*	*Arteriovenous malformation of the brain, somatic;Gastric cancer, somatic;Oculoectodermal syndrome, somatic;RAS-associated autoimmune leukoproliferative disorder;Pancreatic carcinoma, somatic;Lung cancer, somatic;Cardiofaciocutaneous syndrome 2;Bladder cancer, somatic;Leukemia, acute myeloid, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Breast cancer, somatic;Noonan syndrome 3*	*AD*
KRT5	*148040*	*Epidermolysis bullosa simplex-MP;Epidermolysis bullosa simplex, Koebner type;Epidermolysis bullosa simplex, recessive 1;Epidermolysis bullosa simplex, Weber-Cockayne type;Epidermolysis bullosa simplex 2B, generalized intermediate;Epidermolysis bullosa simplex 2A, generalized severe;Epidermolysis bullosa simplex-MCR;Dowling-Degos disease 1;Epidermolysis bullosa simplex 2C, localized;Epidermolysis bullosa simplex, Dowling-Meara type;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, AR*	*AD, AR*
LAMA2	*156225*	*Muscular dystrophy, congenital, merosin deficient or partially deficient;Muscular dystrophy, limb-girdle, AR 23*	*AR*
LAMA3	*600805*	*Laryngoonychocutaneous syndrome;Epidermolysis bullosa, junctional, Herlitz type;Epidermolysis bullosa, generalized atrophic benign*	*AR*
LAMB3	*150310*	*Epidermolysis bullosa, junctional, non-Herlitz type;Amelogenesis imperfecta, type IA;Epidermolysis bullosa, junctional, Herlitz type*	*AR, AD*

LAMC2	*150292*	*Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa, junctional, Herlitz type*	*AR*
LAMP2	*309060*	*Danon disease*	*XLD*
LAMTOR2	*610389*	*Immunodeficiency due to defect in MAPBP-interacting protein*	*AR*
LARS2	*604544*	*?Hydrops, lactic acidosis, and sideroblastic anemia;Perrault syndrome 4*	*AR*
LAS1L	*300964*	*Wilson-Turner syndrome*	*XLR*
LCT	*603202*	*Lactase deficiency, congenital*	*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*
LIG4	*601837*	*Multiple myeloma, resistance to;LIG4 syndrome*	*SM, AR*
LIPA	*613497*	*Wolman disease;Cholesteryl ester storage disease*	*AR*
LIPN	*613924*	*Ichthyosis, congenital, AR 8*	*AR*
LIPT1	*610284*	*Lipoyltransferase 1 deficiency*	*AR*
LMBRD1	*612625*	*Methylmalonic aciduria and homocystinuria, cblF type*	*AR*
LMNA	*150330*	*Malouf syndrome;Emery-Dreifuss muscular dystrophy 3, AR;Emery-Dreifuss muscular dystrophy 2, AD;Hutchinson-Gilford progeria;Muscular dystrophy, congenital;Restrictive dermopathy, lethal;Lipodystrophy, familial partial, type 2;Charcot-Marie-Tooth disease, type 2B1;Mandibuloacral dysplasia;Cardiomyopathy, dilated, 1A;Heart-hand syndrome, Slovenian type*	*AD, AR*
LPIN1	*605518*	*Myoglobinuria, acute recurrent, AR*	*AR*
LRBA	*606453*	*Immunodeficiency, common variable, 8, with autoimmunity*	*AR*
LRPPRC	*607544*	*Mi complex IV deficiency, nuclear type 5, (French-Canadian)*	*AR*
LRRC8A	*608360*	*?Agammaglobulinemia 5*	*AD*
MAGEL2	*605283*	*Schaaf-Yang syndrome*	*AD*

MAGT1	*300715*	*Congenital disorder of glycosylation, type Icc;Immunodeficiency, XL, with magnesium defect, Epstein-Barr virus infection and neoplasia*	*XLR*
MALT1	*604860*	*Immunodeficiency 12*	*AR*
MAN2B1	*609458*	*Mannosidosis, alpha-, types I and II*	*AR*
MANBA	*609489*	*Mannosidosis, beta*	*AR*
MAP2K1	*176872*	*Cardiofaciocutaneous syndrome 3;Melorheostosis, isolated, somatic mosaic*	*AD*
MAP2K2	*601263*	*Cardiofaciocutaneous syndrome 4*	*AD*
MAT1A	*610550*	*Methionine adenosyltransferase deficiency, AR;Hypermethioninemia, persistent, AD, due to methionine adenosyltransferase I/III deficiency*	*AD, AR*
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*
MCPH1	*607117*	*Microcephaly 1, primary, AR*	*AR*
MECP2	*300005*	*Rett syndrome, preserved speech variant;Encephalopathy, neonatal severe;Mental retardation, XL, syndromic 13;Rett syndrome;Mental retardation, XL syndromic, Lubs type;Rett syndrome, atypical;Autism susceptibility, XL 3*	*XLD, XLR, XL*
MED12	*300188*	*Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome*	*XLR, XLD*
MEF2C	*600662*	*Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations;Chromosome 5q14.3 deletion syndrome*	*AD*
MEGF10	*612453*	*Myopathy, areflexia, respiratory distress, and dysphagia, early-onset;Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, mild variant*	*AR*

MFN2	*608507*	*Hereditary motor and sensory neuropathy VIA;Charcot-Marie-Tooth disease, axonal, type 2A2B;Charcot-Marie-Tooth disease, axonal, type 2A2A*	*AD, AR*
MFSD8	*611124*	*Ceroid lipofuscinosis, neuronal, 7;Macular dystrophy with central cone involvement*	*AR*
MITF	*156845*	*Melanoma, cutaneous malignant, susceptibility to, 8;Waardenburg syndrome, type 2A;Waardenburg syndrome/ocular albinism, digenic;Tietz albinism-deafness syndrome;COMMAD syndrome*	*AD, AR*
MKKS	*604896*	*McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6*	*AR*
MLC1	*605908*	*Megalencephalic leukoencephalopathy with subcortical cysts*	*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;Methylmalonic aciduria, cblD type, variant 2;Homocystinuria, cblD type, variant 1*	*AR*
MMUT	*609058*	*Methylmalonic aciduria, mut(0) type*	*AR*
MOCS1	*603707*	*Molybdenum cofactor deficiency A*	*AR*
MOCS2	*603708*	*Molybdenum cofactor deficiency B*	*AR*
MPC1	*614738*	*Mi pyruvate carrier deficiency*	*AR*
MPI	*154550*	*Congenital disorder of glycosylation, type Ib*	*AR*
MPL	*159530*	*Myelofibrosis with myeloid metaplasia, somatic;Thrombocytopenia, congenital amegakaryocytic;Thrombocythemia 2*	*AR, AD, SM*

MPV17	*137960*	*Charcot-Marie-Tooth disease, axonal, type 2EE;Mi DNA depletion syndrome 6 (hepatocerebral type)*	*AR*
MPZ	*159440*	*Charcot-Marie-Tooth disease, type 2I;Dejerine-Sottas disease;Hypomyelinating neuropathy, congenital, 2;Charcot-Marie-Tooth disease, type 2J;Charcot-Marie-Tooth disease, dominant intermediate D;Charcot-Marie-Tooth disease, type 1B;Roussy-Levy syndrome*	*AD, AD, AR*
MRPL3	*607118*	*Combined oxidative phosphorylation deficiency 9*	*AR*
MRPL44	*611849*	*?Combined oxidative phosphorylation deficiency 16*	*AR*
MSMO1	*607545*	*Microcephaly, congenital cataract, and psoriasiform dermatitis*	*AR*
MTHFR	*607093*	*Neural tube defects, susceptibility to;Homocystinuria due to MTHFR deficiency;Schizophrenia, susceptibility to;Thromboembolism, susceptibility to*	*AR, AD*
MTM1	*300415*	*Myotubular myopathy, XL*	*XLR*
MTMR14	*611089*	*Centronuclear myopathy, autosomal, modifier of*	*AD*
MTO1	*614667*	*Combined oxidative phosphorylation deficiency 10*	*AR*
MTR	*156570*	*Neural tube defects, folate-sensitive, susceptibility to;Homocystinuria-megaloblastic anemia, cblG complementation type*	*AR*
MTRFR	*613541*	*Spastic paraplegia 55, AR;Combined oxidative phosphorylation deficiency 7*	*AR*
MTRR	*602568*	*Homocystinuria-megaloblastic anemia, cbl E type;Neural tube defects, folate-sensitive, susceptibility to*	*AR*
MUSK	*601296*	*Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency;Fetal akinesia deformation sequence 1*	*AR*
MVK	*251170*	*Mevalonic aciduria;Hyper-IgD syndrome;Porokeratosis 3, multiple types*	*AR, AD*

MYCN	*164840*	*Feingold syndrome 1*	*AD*
MYH9	*160775*	*Deafness, AD 17;Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss*	*AD*
NAA10	*300013*	*Ogden syndrome;Microphthalmia, syndromic 1*	*XLD, XLR, XL*
NAGA	*104170*	*Schindler disease, type III;Kanzaki disease;Schindler disease, type I*	*AR*
NAGS	*608300*	*N-acetylglutamate synthase deficiency*	*AR*
NALCN	*611549*	*Congenital contractures of the limbs and face, hypotonia, and developmental delay;Hypotonia, infantile, with psychomotor retardation and characteristic facies 1*	*AD, AR*
NARS2	*612803*	*?Deafness, AR 94;Combined oxidative phosphorylation deficiency 24*	*AR*
NBAS	*608025*	*Short stature, optic nerve atrophy, and Pelger-Huet anomaly;Infantile liver failure syndrome 2*	*AR*
NDUFA1	*300078*	*Mi complex I deficiency, nuclear type 12*	*XLR*
NDUFA10	*603835*	*Mi complex I deficiency, nuclear type 22*	*AR*
NDUFA11	*612638*	*Mi complex I deficiency, nuclear type 14*	*AR*
NDUFA2	*602137*	*Mi complex I deficiency, nuclear type 13*	*AR*
NDUFA9	*603834*	*Mi complex I deficiency, nuclear type 26*	*AR*
NDUFAF1	*606934*	*Mi complex I deficiency, nuclear type 11*	*AR*
NDUFAF2	*609653*	*Mi complex I deficiency, nuclear type 10*	*AR*
NDUFAF3	*612911*	*Mi complex I deficiency, nuclear type 18*	*AR*
NDUFAF4	*611776*	*Mi complex I deficiency, nuclear type 15*	*AR*
NDUFAF5	*612360*	*Mi complex I deficiency, nuclear type 16*	*AR*
NDUFAF6	*612392*	*Mi complex I deficiency, nuclear type 17;Fanconi renotubular syndrome 5*	*AR*

NDUFB3	*603839*	*Mi complex I deficiency, nuclear type 25*	*AR*
NDUFB9	*601445*	*?Mi complex I deficiency, nuclear type 24*	*AR*
NDUFS1	*157655*	*Mi complex I deficiency, nuclear type 5*	*AR*
NDUFS2	*602985*	*Mi complex I deficiency, nuclear type 6*	*AR*
NDUFS3	*603846*	*Mi complex I deficiency, nuclear type 8*	*AR*
NDUFS4	*602694*	*Mi complex I deficiency, nuclear type 1*	*AR*
NDUFS6	*603848*	*Mi complex I deficiency, nuclear type 9*	*AR*
NDUFS7	*601825*	*Mi complex I deficiency, nuclear type 3*	*AR*
NDUFV1	*161015*	*Mi complex I deficiency, nuclear type 4*	*AR*
NDUFV2	*600532*	*Mi complex I deficiency, nuclear type 7*	*AR*
NEB	*161650*	*Nemaline myopathy 2, AR;Arthrogryposis multiplex congenita 6*	*AR*
NEU1	*608272*	*Sialidosis, type I;Sialidosis, type II*	*AR*
NEUROG3	*604882*	*Diarrhea 4, malabsorptive, congenital*	*AR*
NEXN	*613121*	*Cardiomyopathy, hypertrophic, 20;Cardiomyopathy, dilated, 1CC*	*AD*
NFKB2	*164012*	*Immunodeficiency, common variable, 10*	*AD*
NFU1	*608100*	*Multiple Mi 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*	
NIPAL4	*609383*	*Ichthyosis, congenital, AR 6*	*AR*
NIPBL	*608667*	*Cornelia de Lange syndrome 1*	*AD*
NKX2-1	*600635*	*Chorea, hereditary benign;Choreoathetosis, hypothyroidism, and neonatal respiratory distress;Thyroid cancer, nonmedullary, 1*	*AD*

NKX2-5	*600584*	*Hypoplastic left heart syndrome 2;Hypothyroidism, congenital nongoitrous, 5;Atrial septal defect 7, with or without AV conduction defects;Conotruncal heart malformations, variable;Ventricular septal defect 3;Tetralogy of Fallot*	*AD*
NLRC4	*606831*	*Autoinflammation with infantile enterocolitis;?Familial cold autoinflammatory syndrome 4*	*AD*
NLRP3	*606416*	*CINCA syndrome;Deafness, AD 34, with or without inflammation;Keratoendothelitis fugax hereditaria;Familial cold inflammatory syndrome 1;Muckle-Wells syndrome*	*AD*
NOTCH2	*600275*	*Alagille syndrome 2;Hajdu-Cheney syndrome*	*AD*
NPC1	*607623*	*Niemann-Pick disease, type C1;Niemann-Pick disease, type D*	*AR*
NPC2	*601015*	*Niemann-pick disease, type C2*	*AR*
NPHP1	*607100*	*Joubert syndrome 4;Nephronophthisis 1, juvenile;Senior-Loken syndrome-1*	*AR*
NPHP3	*608002*	*Nephronophthisis 3;Meckel syndrome 7;Renal-hepatic-pancreatic dysplasia 1*	*AR*
NR0B1	*300473*	*Adrenal hypoplasia, congenital;46XY sex reversal 2, dosage-sensitive*	*XLR, XL*
NR3C2	*600983*	*Hypertension, early-onset, AD, with exacerbation in pregnancy;Pseudohypoaldosteronism type I, AD*	*AD*
NRAS	*164790*	*Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Colorectal cancer, somatic;Neurocutaneous melanosis, somatic;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic;Noonan syndrome 6;Melanocytic nevus syndrome, congenital, somatic;Thyroid carcinoma, follicular, somatic;Epidermal nevus, somatic*	*AD*
NSD1	*606681*	*Sotos syndrome 1*	*AD*

NSDHL	*300275*	*CK syndrome;CHILD syndrome*	*XLR, XLD*
NUBPL	*613621*	*Mi complex I deficiency, nuclear type 21*	*AR*
OAT	*613349*	*Gyrate atrophy of choroid and retina with or without ornithinemia*	*AR*
OCLN	*602876*	*Pseudo-TORCH syndrome 1*	*AR*
OCRL	*300535*	*Lowe syndrome;Dent disease 2*	*XLR*
ODAD1	*615038*	*Ciliary dyskinesia, primary, 20*	*AR*
OPA3	*606580*	*3-methylglutaconic aciduria, type III;Optic atrophy 3 with cataract*	*AR, AD*
OPHN1	*300127*	*Mental retardation, XL, with cerebellar hypoplasia and distinctive facial appearance*	*XLR*
ORC1	*601902*	*Meier-Gorlin syndrome 1*	*AR*
ORC4	*603056*	*Meier-Gorlin syndrome 2*	*AR*
OTC	*300461*	*Ornithine transcarbamylase deficiency*	*XL*
OTX2	*600037*	*Retinal dystrophy, early-onset, with or without pituitary dysfunction;Microphthalmia, syndromic 5;Pituitary hormone deficiency, combined, 6*	*AD*
OXCT1	*601424*	*Succinyl CoA:3-oxoacid CoA transferase deficiency*	*AR*
PAFAH1B1	*601545*	*Subcortical laminar heterotopia;Lissencephaly 1*	*AD*
PAH	*612349*	*Phenylketonuria;[Hyperphenylalaninemia, non-PKU mild]*	*AR*
PAX2	*167409*	*Glomerulosclerosis, focal segmental, 7;Papillorenal syndrome*	*AD*
PAX3	*606597*	*Waardenburg syndrome, type 3;Rhabdomyosarcoma 2, alveolar;Craniofacial-deafness-hand syndrome;Waardenburg syndrome, type 1*	*AD, AR, SM, AD*
PAX6	*607108*	*?Coloboma, ocular;Aniridia;?Morning glory disc anomaly;Keratitis;Optic nerve hypoplasia;?Coloboma of optic nerve;Anterior segment dysgenesis 5, multiple subtypes;Cataract with late-onset corneal dystrophy;Foveal hypoplasia 1*	*AD*
PAX8	*167415*	*Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia*	*AD*

PC	*608786*	*Pyruvate carboxylase deficiency*	*AR*
PCBD1	*126090*	*Hyperphenylalaninemia, BH4-deficient, D*	*AR*
PCCA	*232000*	*Propionicacidemia*	*AR*
PCCB	*232050*	*Propionicacidemia*	*AR*
PCDH19	*300460*	*Developmental and epileptic encephalopathy 9*	*XL*
PCNT	*605925*	*Microcephalic osteodysplastic primordial dwarfism, type II*	*AR*
PDCD10	*609118*	*Cerebral cavernous malformations 3*	*AD*
PDE10A	*610652*	*Dyskinesia, limb and orofacial, infantile-onset;Striatal degeneration, AD*	*AR, 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*
PDP1	*605993*	*Pyruvate dehydrogenase phosphatase deficiency*	*AR*
PDSS2	*610564*	*Coenzyme Q10 deficiency, primary, 3*	*AR*
PDX1	*600733*	*Pancreatic agenesis 1;MODY, type IV;Diabetes mellitus, type II, susceptibility to*	*AR, AD*
PEPD	*613230*	*Prolidase deficiency*	*AR*
PEX1	*602136*	*Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1*	*AR*
PEX10	*602859*	*Peroxisome biogenesis disorder 6A (Zellweger);Peroxisome biogenesis disorder 6B*	*AR*
PEX12	*601758*	*Peroxisome biogenesis disorder 3B;Peroxisome biogenesis disorder 3A (Zellweger)*	*AR*
PEX13	*601789*	*Peroxisome biogenesis disorder 11B;Peroxisome biogenesis disorder 11A (Zellweger)*	*AR*
PEX14	*601791*	*Peroxisome biogenesis disorder 13A (Zellweger)*	*AR*
PEX16	*603360*	*Peroxisome biogenesis disorder 8A (Zellweger);Peroxisome biogenesis disorder 8B*	*AR*
PEX19	*600279*	*Peroxisome biogenesis disorder 12A (Zellweger)*	*AR*

PEX2	*170993*	*Peroxisome biogenesis disorder 5B;Peroxisome biogenesis disorder 5A (Zellweger)*	*AR*
PEX26	*608666*	*Peroxisome biogenesis disorder 7A (Zellweger);Peroxisome biogenesis disorder 7B*	*AR*
PEX3	*603164*	*Peroxisome biogenesis disorder 10A (Zellweger);?Peroxisome biogenesis disorder 10B*	*AR*
PEX5	*600414*	*Rhizomelic chondrodysplasia punctata, type 5;Peroxisome biogenesis disorder 2B;Peroxisome biogenesis disorder 2A (Zellweger)*	*AR*
PEX6	*601498*	*Peroxisome biogenesis disorder 4B;Peroxisome biogenesis disorder 4A (Zellweger);Heimler syndrome 2*	*AD, AR, AR*
PEX7	*601757*	*Peroxisome biogenesis disorder 9B;Rhizomelic chondrodysplasia punctata, type 1*	*AR*
PGAP1	*611655*	*Mental retardation, AR 42*	*AR*
PGM1	*171900*	*Congenital disorder of glycosylation, type It*	*AR*
PHGDH	*606879*	*Phosphoglycerate dehydrogenase deficiency;Neu-Laxova syndrome 1*	*AR*
PHKG2	*172471*	*Glycogen storage disease IXc*	*AR*
PHOX2B	*603851*	*Central hypoventilation syndrome, congenital, with or without Hirschsprung disease;Neuroblastoma, susceptibility to, 2;Neuroblastoma with Hirschsprung disease*	*AD*
PIGA	*311770*	*Paroxysmal nocturnal hemoglobinuria, somatic;Multiple congenital anomalies-hypotonia-seizures syndrome 2*	*XLR*
PIGN	*606097*	*Multiple congenital anomalies-hypotonia-seizures syndrome 1*	*AR*
PIGT	*610272*	*Multiple congenital anomalies-hypotonia-seizures syndrome 3;?Paroxysmal nocturnal hemoglobinuria 2*	*AR, AD, SM*
PIGV	*610274*	*Hyperphosphatasia with mental retardation syndrome 1*	*AR*

PIK3CD	*602839*	*Immunodeficiency 14;?Roifman-Chitayat syndrome, digenic;Immunodeficiency 14B, AR*	*AD, DR, AR*
PKD2	*173910*	*Polycystic kidney disease 2*	*AD*
PKHD1	*606702*	*Polycystic kidney disease 4, with or without hepatic disease*	*AR*
PKLR	*609712*	*Adenosine triphosphate, elevated, of erythrocytes;Pyruvate kinase deficiency*	*AD, AR*
PLCB4	*600810*	*Auriculocondylar syndrome 2*	*AD, AR*
PLEC	*601282*	*Epidermolysis bullosa simplex, Ogna type;Epidermolysis bullosa simplex with muscular dystrophy;?Epidermolysis bullosa simplex with nail dystrophy;Epidermolysis bullosa simplex with pyloric atresia;Muscular dystrophy, limb-girdle, AR 17*	*AD, AR*
PLOD1	*153454*	*Ehlers-Danlos syndrome, kyphoscoliotic type, 1*	*AR*
PLP1	*300401*	*Pelizaeus-Merzbacher disease;Spastic paraplegia 2, XL*	*XLR*
PMM2	*601785*	*Congenital disorder of glycosylation, type Ia*	*AR*
PMP22	*601097*	*Neuropathy, recurrent, with pressure palsies;Dejerine-Sottas disease;Roussy-Levy syndrome;?Neuropathy, inflammatory demyelinating;Charcot-Marie-Tooth disease, type 1E;Charcot-Marie-Tooth disease, type 1A*	*AD, AD, AR, ?AD*
PNKP	*605610*	*Ataxia-oculomotor apraxia 4;Microcephaly, seizures, and developmental delay;?Charcot-Marie-Tooth disease, type 2B2*	*AR*
PNP	*164050*	*Immunodeficiency due to purine nucleoside phosphorylase deficiency*	*AR*
PNPLA1	*612121*	*Ichthyosis, congenital, AR 10*	*AR*
PNPO	*603287*	*Pyridoxamine 5'-phosphate oxidase deficiency*	*AR*
PNPT1	*610316*	*Deafness, AR 70;Combined oxidative phosphorylation deficiency 13*	*AR*
POGZ	*614787*	*White-Sutton syndrome*	*AD*

POLG	*174763*	*Progressive external ophthalmoplegia, AR 1;Progressive external ophthalmoplegia, AD 1;Mi recessive ataxia syndrome (includes SANDO and SCAE);Mi DNA depletion syndrome 4B (MNGIE type);Mi DNA depletion syndrome 4A (Alpers type)*	*AR, AD*
POLG2	*604983*	*Mi DNA depletion syndrome 16 (hepatic type);?Mi DNA depletion syndrome 16B (neuroophthalmic type);Progressive external ophthalmoplegia with Mi DNA deletions, AD 4*	*AR, AD*
POMGNT1	*606822*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3;Retinitis pigmentosa 76*	*AR*
POMGNT2	*614828*	*Muscular dystrophy-dystroglycanopathy (limb-girdle) type C, 8;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;?Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12*	*AR*
POMT1	*607423*	*Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1;Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1*	*AR*

POMT2	*607439*	*Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2;Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2*	*AR*
POU1F1	*173110*	*Pituitary hormone deficiency, combined, 1*	*AD, AR*
PPT1	*600722*	*Ceroid lipofuscinosis, neuronal, 1*	*AR*
PRDM16	*605557*	*Cardiomyopathy, dilated, 1LL;Left ventricular noncompaction 8*	*AD*
PRKAG2	*602743*	*Glycogen storage disease of heart, lethal congenital;Cardiomyopathy, hypertrophic 6;Wolff-Parkinson-White syndrome*	*AD*
PRKAR1A	*188830*	*Myxoma, intracardiac;Pigmented nodular adrenocortical disease, primary, 1;Carney complex, type 1;Acrodysostosis 1, with or without hormone resistance*	*AD*
PROC	*612283*	*Thrombophilia due to protein C deficiency, AD;Thrombophilia due to protein C deficiency, AR*	*AD, AR*
PRODH	*606810*	*Schizophrenia, susceptibility to, 4;Hyperprolinemia, type I*	*AD, AR*
PROP1	*601538*	*Pituitary hormone deficiency, combined, 2*	*AR*
PROS1	*176880*	*Thrombophilia due to protein S deficiency, AR;Thrombophilia due to protein S deficiency, AD*	*AR, AD*
PRPS1	*311850*	*Charcot-Marie-Tooth disease, XLR, 5;Deafness, XL 1;Phosphoribosylpyrophosphate synthetase superactivity;Arts syndrome;Gout, PRPS-related*	*XLR, XL*
PRRT2	*614386*	*Episodic kinesigenic dyskinesia 1;Convulsions, familial infantile, with paroxysmal choreoathetosis;Seizures, benign familial infantile, 2*	*AD*

PSAP	*176801*	*Combined SAP deficiency;Gaucher disease, atypical;Krabbe disease, atypical;Parkinson disease 24, AD, susceptibility to;Metachromatic leukodystrophy due to SAP-b deficiency*	*AR, AD*
PSAT1	*610936*	*Neu-Laxova syndrome 2;?Phosphoserine aminotransferase deficiency*	*AR*
PSPH	*172480*	*Phosphoserine phosphatase deficiency*	*AR*
PTPN11	*176876*	*Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1*	*AD*
PTPRC	*151460*	*Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive;Hepatitis C virus, susceptibility to*	*AR*
PTRH2	*608625*	*Infantile-onset multisystem neurologic, endocrine, and pancreatic disease*	*AR*
PTS	*612719*	*Hyperphenylalaninemia, BH4-deficient, A*	*AR*
PURA	*600473*	*Mental retardation, AD 31*	*AD*
QDPR	*612676*	*Hyperphenylalaninemia, BH4-deficient, C*	*AR*
RAB18	*602207*	*Warburg micro syndrome 3*	*AR*
RAB3GAP1	*602536*	*Martsolf syndrome 2;Warburg micro syndrome 1*	*AR*
RAB3GAP2	*609275*	*Warburg micro syndrome 2;Martsolf syndrome*	*AR*
RAC2	*602049*	*?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia;Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis;Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia*	*AR, AD*
RAF1	*164760*	*Noonan syndrome 5;LEOPARD syndrome 2;Cardiomyopathy, dilated, 1NN*	*AD*

RAG1	*179615*	*Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity;Omenn syndrome;Severe combined immunodeficiency, B cell-negative;Combined cellular and humoral immune defects with granulomas*	*AR*
RAG2	*179616*	*Omenn syndrome;Combined cellular and humoral immune defects with granulomas;Severe combined immunodeficiency, B cell-negative*	*AR*
RANBP2	*601181*	*Encephalopathy, acute, infection-induced, 3, susceptibility to*	*AD*
RAPSN	*601592*	*Fetal akinesia deformation sequence 2;Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency*	*AR*
RARS2	*611524*	*Pontocerebellar hypoplasia, type 6*	*AR*
RB1	*614041*	*Bladder cancer, somatic;Retinoblastoma, trilateral;Small cell cancer of the lung, somatic;Osteosarcoma, somatic;Retinoblastoma*	*AD, SM*
RBBP8	*604124*	*Seckel syndrome 2;Jawad syndrome*	*AR*
RBM8A	*605313*	*Thrombocytopenia-absent radius syndrome*	*AR*
RET	*164761*	*Hirschsprung disease, susceptibility to, 1;Multiple endocrine neoplasia IIA;Medullary thyroid carcinoma;Pheochromocytoma;Multiple endocrine neoplasia IIB;Hirschsprung disease, protection against;Central hypoventilation syndrome, congenital*	*AD*
RFT1	*611908*	*Congenital disorder of glycosylation, type In*	*AR*
RFX5	*601863*	*Bare lymphocyte syndrome, type II, complementation group E;Bare lymphocyte syndrome, type II, complementation group C*	*AR*
RFX6	*612659*	*Mitchell-Riley syndrome*	*AR*
RIT1	*609591*	*Noonan syndrome 8*	*AD*

RMND1	*614917*	*Combined oxidative phosphorylation deficiency 11*	*AR*
RNASEH2C	*610330*	*Aicardi-Goutieres syndrome 3*	*AR*
RNASET2	*612944*	*Leukoencephalopathy, cystic, without megalencephaly*	*AR*
RORC	*602943*	*Immunodeficiency 42*	*AR*
RPS19	*603474*	*Diamond-Blackfan anemia 1*	*AD*
RRM2B	*604712*	*Mi DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy);Progressive external ophthalmoplegia with Mi DNA deletions, AD 5;Mi DNA depletion syndrome 8B (MNGIE type)*	*AR, AD*
RXYLT1	*605862*	*Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10*	*AR*
RYR1	*180901*	*Malignant hyperthermia susceptibility 1;King-Denborough syndrome;Central core disease;Neuromuscular disease, congenital, with uniform type 1 fiber;Minicore myopathy with external ophthalmoplegia*	*AD, AD, AR, AR*
SALL1	*602218*	*Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1*	*AD*
SATB2	*608148*	*Glass syndrome*	*AD*
SBDS	*607444*	*Aplastic anemia, susceptibility to;Shwachman-Diamond syndrome*	*AR*
SCN1A	*182389*	*Epilepsy, generalized, with febrile seizures plus, type 2;Migraine, familial hemiplegic, 3;Developmental and epileptic encephalopathy 6B, non-Dravet;Febrile seizures, familial, 3A;Dravet syndrome*	*AD*
SCN2A	*182390*	*Developmental and epileptic encephalopathy 11;Seizures, benign familial infantile, 3;Episodic ataxia, type 9*	*AD*

SCN4A	*603967*	*Paramyotonia congenita;Myotonia congenita, atypical, acetazolamide-responsive;Myasthenic syndrome, congenital, 16;Hyperkalemic periodic paralysis, type 2;Hypokalemic periodic paralysis, type 2*	*AD, AR*
SCN5A	*600163*	*Heart block, nonprogressive;Ventricular fibrillation, familial, 1;Sick sinus syndrome 1;Brugada syndrome 1;Heart block, progressive, type IA;Atrial fibrillation, familial, 10;Long QT syndrome 3;Cardiomyopathy, dilated, 1E;Sudden infant death syndrome, susceptibility to*	*AD, AR*
SCN9A	*603415*	*Erythermalgia, primary;Generalized epilepsy with febrile seizures plus, type 7;Febrile seizures, familial, 3B;Neuropathy, hereditary sensory and autonomic, type IID;Insensitivity to pain, congenital;Paroxysmal extreme pain disorder;Small fiber neuropathy*	*AD, AR*
SCO1	*603644*	*Mi complex IV deficiency, nuclear type 4*	*AR*
SCO2	*604272*	*Myopia 6;Mi complex IV deficiency, nuclear type 2*	*AD, AR*
SDHA	*600857*	*Neurodegeneration with ataxia and late-onset optic atrophy;Cardiomyopathy, dilated, 1GG;Leigh syndrome;Mi respiratory chain complex II deficiency;Paragangliomas 5*	*AD, AR, AR, Mi*
SDHAF1	*612848*	*Mi complex II deficiency, nuclear type 2;Mi complex II deficiency*	*AR*
SECISBP2	*607693*	*Thyroid hormone metabolism, abnormal*	*AR*
SELENON	*606210*	*Muscular dystrophy, rigid spine, 1;Myopathy, congenital, with fiber-type disproportion*	*AR, AD, AR*
SERAC1	*614725*	*3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome*	*AR*
SERPINC1	*107300*	*Thrombophilia due to antithrombin III deficiency*	*AD, AR*

SERPING1	*606860*	*Complement component 4, partial deficiency of;Angioedema, hereditary, types I and II*	*AD, AD, AR*
SFTPB	*178640*	*Surfactant metabolism dysfunction, pulmonary, 1*	*AR*
SFTPC	*178620*	*Surfactant metabolism dysfunction, pulmonary, 2*	*AD*
SHOC2	*602775*	*Noonan syndrome-like with loose anagen hair 1*	*AD*
SIL1	*608005*	*Marinesco-Sjogren syndrome*	*AR*
SIX3	*603714*	*Holoprosencephaly 2;Schizencephaly*	*AD*
SIX5	*600963*	*Branchiootorenal syndrome 2*	
SKI	*164780*	*Shprintzen-Goldberg syndrome*	*AD*
SLC12A6	*604878*	*Agenesis of the corpus callosum with peripheral neuropathy*	*AR*
SLC16A1	*600682*	*Hyperinsulinemic hypoglycemia, familial, 7;Erythrocyte lactate transporter defect;Monocarboxylate transporter 1 deficiency*	*AD, AD, AR*
SLC16A2	*300095*	*Allan-Herndon-Dudley syndrome*	*XL*
SLC17A5	*604322*	*Sialic acid storage disorder, infantile;Salla disease*	*AR*
SLC19A2	*603941*	*Thiamine-responsive megaloblastic anemia syndrome*	*AR*
SLC19A3	*606152*	*Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)*	*AR*
SLC22A5	*603377*	*Carnitine deficiency, systemic primary*	*AR*
SLC25A1	*190315*	*Myasthenic syndrome, congenital, 23, presynaptic;Combined D-2- and L-2-hydroxyglutaric aciduria*	*AR*
SLC25A12	*603667*	*Developmental and epileptic encephalopathy 39*	*AR*
SLC25A13	*603859*	*Citrullinemia, type II, neonatal-onset;Citrullinemia, adult-onset type II*	*AR*
SLC25A15	*603861*	*Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome*	*AR*
SLC25A19	*606521*	*Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type);Microcephaly, Amish type*	*AR*
SLC25A20	*613698*	*Carnitine-acylcarnitine translocase deficiency*	*AR*

SLC25A22	*609302*	*Developmental and epileptic encephalopathy 3*	*AR*
SLC25A3	*600370*	*Mi phosphate carrier deficiency*	*AR*
SLC26A2	*606718*	*Diastrophic dysplasia, broad bone-platyspondylic variant;Achondrogenesis Ib;Epiphyseal dysplasia, multiple, 4;De la Chapelle dysplasia;Diastrophic dysplasia;Atelosteogenesis, type II*	*AR*
SLC26A3	*126650*	*Diarrhea 1, secretory chloride, congenital*	*AR*
SLC26A4	*605646*	*Pendred syndrome;Deafness, AR 4, with enlarged vestibular aqueduct*	*AR*
SLC2A1	*138140*	*GLUT1 deficiency syndrome 2, childhood onset;GLUT1 deficiency syndrome 1, infantile onset, severe;Epilepsy, idiopathic generalized, susceptibility to, 12;Stomatin-deficient cryohydrocytosis with neurologic defects;Dystonia 9*	*AD, AD, AR*
SLC30A2	*609617*	*Zinc deficiency, transient neonatal*	*AD*
SLC33A1	*603690*	*Congenital cataracts, hearing loss, and neurodegeneration;Spastic paraplegia 42, AD*	*AR, AD*
SLC3A1	*104614*	*Cystinuria*	*AD, AR*
SLC4A1	*109270*	*[Blood group, Wright];Distal renal tubular acidosis 1;Ovalocytosis, SA type;[Malaria, resistance to];[Blood group, Swann];[Blood group, Froese];[Blood group, Waldner];Cryohydrocytosis;[Blood group, Diego];Distal renal tubular acidosis 4 with hemolytic anemia;Spherocytosis, type 4*	*AD, AR*
SLC52A1	*607883*	*Riboflavin deficiency*	*AD*
SLC52A3	*613350*	*?Fazio-Londe disease;Brown-Vialetto-Van Laere syndrome 1*	*AR*
SLC5A1	*182380*	*Glucose/galactose malabsorption*	*AR*
SLC5A5	*601843*	*Thyroid dyshormonogenesis 1*	*AR*
SLC6A1	*137165*	*Myoclonic-atonic epilepsy*	*AD*
SLC6A3	*126455*	*Nicotine dependence, protection against;Parkinsonism-dystonia, infantile, 1*	*AR*
SLC6A5	*604159*	*Hyperekplexia 3*	*AD, AR*

SLC7A7	*603593*	*Lysinuric protein intolerance*	*AR*
SLC7A9	*604144*	*Cystinuria*	*AD, AR*
SLCO1B1	*604843*	*Hyperbilirubinemia, Rotor type, digenic*	*DR*
SLCO1B3	*605495*	*Hyperbilirubinemia, Rotor type, digenic*	*DR*
SMPD1	*607608*	*Niemann-Pick disease, type A;Niemann-Pick disease, type B*	*AR*
SNAI2	*602150*	*Waardenburg syndrome, type 2D;Piebaldism*	*AR, AD*
SNX10	*614780*	*Osteopetrosis, AR 8*	*AR*
SOS1	*182530*	*Noonan syndrome 4;?Fibromatosis, gingival, 1*	*AD*
SOX10	*602229*	*Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement*	*AD*
SOX2	*184429*	*Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system*	*AD*
SOX9	*608160*	*Acampomelic campomelic dysplasia;Campomelic dysplasia;Campomelic dysplasia with autosomal sex reversal*	*AD*
SPAST	*604277*	*Spastic paraplegia 4, AD*	*AD*
SPEG	*615950*	*Centronuclear myopathy 5*	*AR*
SPINK5	*605010*	*Netherton syndrome*	*AR*
SPINT2	*605124*	*Diarrhea 3, secretory sodium, congenital, syndromic*	*AR*
SPR	*182125*	*Dystonia, dopa-responsive, due to sepiapterin reductase deficiency*	*?AD, AR*
SPRED1	*609291*	*Legius syndrome*	*AD*
SPTA1	*182860*	*Pyropoikilocytosis;Elliptocytosis-2;Spherocytosis, type 3*	*AR, AD*
SPTAN1	*182810*	*Developmental and epileptic encephalopathy 5*	*AD*
SPTB	*182870*	*Spherocytosis, type 2;Elliptocytosis-3;Anemia, neonatal hemolytic, fatal or near-fatal*	*AD*
SRD5A3	*611715*	*Congenital disorder of glycosylation, type Iq;Kahrizi syndrome*	*AR*
ST3GAL3	*606494*	*?Developmental and epileptic encephalopathy 15;Mental retardation, AR 12*	*AR*

ST3GAL5	*604402*	*Salt and pepper developmental regression syndrome*	*AR*
STAR	*600617*	*Lipoid adrenal hyperplasia*	*AR*
STAT1	*600555*	*Immunodeficiency 31C, chronic mucocutaneous candidiasis, AD;Immunodeficiency 31B, mycobacterial and viral infections, AR;Immunodeficiency 31A, mycobacteriosis, AD*	*AD, AR*
STAT3	*102582*	*Hyper-IgE recurrent infection syndrome;Autoimmune disease, multisystem, infantile-onset, 1*	*AD*
STIL	*181590*	*Microcephaly 7, primary, AR*	*AR*
STIM1	*605921*	*Myopathy, tubular aggregate, 1;Stormorken syndrome;Immunodeficiency 10*	*AD, AR*
STING1	*612374*	*STING-associated vasculopathy, infantile-onset*	*AD*
STS	*300747*	*Ichthyosis, XL*	*XLR*
STT3B	*608605*	*?Congenital disorder of glycosylation, type Ix*	*AR*
STXBP1	*602926*	*Developmental and epileptic encephalopathy 4*	*AD*
SUCLA2	*603921*	*Mi DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)*	*AR*
SUCLG1	*611224*	*Mi DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)*	*AR*
SUMF1	*607939*	*Multiple sulfatase deficiency*	*AR*
SUOX	*606887*	*Sulfite oxidase deficiency*	*AR*
SYNE1	*608441*	*Arthrogryposis multiplex congenita 3, myogenic type;Emery-Dreifuss muscular dystrophy 4, AD;Spinocerebellar ataxia, AR 8*	*AR, AD*
TACO1	*612958*	*Mi complex IV deficiency, nuclear type 8*	*AR*
TAFAZZIN	*300394*	*Barth syndrome*	*XLR*
TAT	*613018*	*Tyrosinemia, type II*	*AR*
TBC1D24	*613577*	*Deafness, AD 65;Deafness , AR 86;Myoclonic epilepsy, infantile, familial;Developmental and epileptic encephalopathy 16;DOORS syndrome;Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp*	*AD, AR*

TBCE	*604934*	*Hypoparathyroidism-retardation-dysmorphism syndrome;Encephalopathy, progressive, with amyotrophy and optic atrophy;Kenny-Caffey syndrome, type 1*	*AR*
TBL1X	*300196*	*Hypothyroidism, congenital, nongoitrous, 8*	*XL*
TBX19	*604614*	*Adrenocorticotropic hormone deficiency*	*AR*
TBX5	*601620*	*Holt-Oram syndrome*	*AD*
TCAP	*604488*	*Muscular dystrophy, limb-girdle, AR 7;Cardiomyopathy, hypertrophic, 25*	*AR, AD*
TCN2	*613441*	*Transcobalamin II deficiency*	*AR*
TFR2	*604720*	*Hemochromatosis, type 3*	*AR*
TG	*188450*	*Thyroid dyshormonogenesis 3;Autoimmune thyroid disease, susceptibility to, 3*	*AR*
TGM1	*190195*	*Ichthyosis, congenital, AR 1*	*AR*
TH	*191290*	*Segawa syndrome, recessive*	*AR*
THRA	*190120*	*Hypothyroidism, congenital, nongoitrous, 6*	*AD*
THRB	*190160*	*Thyroid hormone resistance, AR;Thyroid hormone resistance;Thyroid hormone resistance, selective pituitary*	*AR, AD*
TJP2	*607709*	*Hypercholanemia, familial;Cholestasis, progressive familial intrahepatic 4*	*AR*
TMCO1	*614123*	*Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome*	*AR*
TMEM165	*614726*	*Congenital disorder of glycosylation, type IIk*	*AR*
TMEM70	*612418*	*Mi complex V (ATP synthase) deficiency, nuclear type 2*	*AR*
TNFRSF13B	*604907*	*Immunodeficiency, common variable, 2;Immunoglobulin A deficiency 2*	*AD, AR*
TNFRSF13C	*606269*	*Immunodeficiency, common variable, 4*	*AR*
TNFSF4	*603594*	*Myocardial infarction, susceptibility to*	
TNNT1	*191041*	*Nemaline myopathy 5, Amish type*	*AR*

TP63	*603273*	*Rapp-Hodgkin syndrome;Orofacial cleft 8;Limb-mammary syndrome;Split-hand/foot malformation 4;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;Hay-Wells syndrome;ADULT syndrome*	*AD*
TPM2	*190990*	*Arthrogryposis, distal, type 2B4;Arthrogryposis, distal, type 1A;CAP myopathy 2;Nemaline myopathy 4, AD*	*AD*
TPM3	*191030*	*Myopathy, congenital, with fiber-type disproportion;Nemaline myopathy 1, AD or recessive;CAP myopathy 1*	*AD, AR*
TPO	*606765*	*Thyroid dyshormonogenesis 2A*	*AR*
TPP1	*607998*	*Ceroid lipofuscinosis, neuronal, 2;Spinocerebellar ataxia, AR 7*	*AR*
TRH	*613879*	*Thyrotropin-releasing hormone deficiency*	*AR*
TRHR	*188545*	*Hypothyroidism, congenital, nongoitrous, 7*	*AR*
TRIP11	*604505*	*Achondrogenesis, type IA;Osteochondrodysplasia*	*AR*
TRMU	*610230*	*Liver failure, transient infantile;Deafness, Mi, modifier of*	*AR, Mi*
TRPV4	*605427*	*SED, Maroteaux type;Spondylometaphyseal dysplasia, Kozlowski type;Metatropic dysplasia;Brachyolmia type 3;Neuronopathy, distal hereditary motor, type VIII;[Sodium serum level QTL 1];?Avascular necrosis of femoral head, primary, 2;Scapuloperoneal spinal muscular atrophy;Parastremmatic dwarfism;Hereditary motor and sensory neuropathy, type IIc;Digital arthropathy-brachydactyly, familial*	*AD*
TSC1	*605284*	*Lymphangioleiomyomatosis;Focal cortical dysplasia, type II, somatic;Tuberous sclerosis-1*	*AD*
TSC2	*191092*	*?Focal cortical dysplasia, type II, somatic;Lymphangioleiomyomatosis, somatic;Tuberous sclerosis-2*	*AD*
TSFM	*604723*	*Combined oxidative phosphorylation deficiency 3*	*AR*

TSHB	*188540*	*Hypothyroidism, congenital, nongoitrous 4*	*AR*
TSHR	*603372*	*Hyperthyroidism, nonautoimmune;Hypothyroidism, congenital, nongoitrous, 1;Hyperthyroidism, familial gestational*	*AD, AR*
TSPYL1	*604714*	*Sudden infant death with dysgenesis of the testes syndrome*	*AR*
TTC7A	*609332*	*Gastrointestinal defects and immunodeficiency syndrome*	*AR*
TTN	*188840*	*Cardiomyopathy, dilated, 1G;Muscular dystrophy, limb-girdle, AR 10;Tibial muscular dystrophy, tardive;Myopathy, myofibrillar, 9, with early respiratory failure;Salih myopathy;Cardiomyopathy, familial hypertrophic, 9*	*AR, AD*
TUBA8	*605742*	*Cortical dysplasia, complex, with other brain malformations 8*	*AR*
TUBB1	*612901*	*Macrothrombocytopenia, AD, TUBB1-related*	*AD*
TUBB2A	*615101*	*Cortical dysplasia, complex, with other brain malformations 5*	*AD*
TWNK	*606075*	*Progressive external ophthalmoplegia with Mi DNA deletions, AD 3;Perrault syndrome 5;Mi DNA depletion syndrome 7 (hepatocerebral type)*	*AD, AR*
UBA1	*314370*	*VEXAS syndrome, somatic;Spinal muscular atrophy, XL 2, infantile*	*XLR*
UBR1	*605981*	*Johanson-Blizzard syndrome*	*AR*
UGT1A1	*191740*	*[Gilbert syndrome];Crigler-Najjar syndrome, type II;Crigler-Najjar syndrome, type I;Hyperbilirubinemia, familial transient neonatal;[Bilirubin, serum level of, QTL1]*	*AR*
UMPS	*613891*	*Orotic aciduria*	*AR*
UNG	*191525*	*Immunodeficiency with hyper IgM, type 5*	*AR*
UPB1	*606673*	*Beta-ureidopropionase deficiency*	*AR*
UQCRC2	*191329*	*Mi complex III deficiency, nuclear type 5*	*AR*
UROD	*613521*	*Porphyria, hepatoerythropoietic;Porphyria cutanea tarda*	*AD, AR*

UROS	*606938*	*Porphyria, congenital erythropoietic*	*AR*
WAS	*300392*	*Wiskott-Aldrich syndrome;Thrombocytopenia, XL;Neutropenia, severe congenital, XL;Thrombocytopenia, XL, intermittent*	*XLR*
WDPCP	*613580*	*Congenital heart defects, hamartomas of tongue, and polysyndactyly;?Bardet-Biedl syndrome 15*	*AR*
WDR62	*613583*	*Microcephaly 2, primary, AR, with or without cortical malformations*	*AR*
WDR73	*616144*	*Galloway-Mowat syndrome 1*	*AR*
WFS1	*606201*	*?Cataract 41;Wolfram-like syndrome, AD;Wolfram syndrome 1;Diabetes mellitus, noninsulin-dependent, association with;Deafness, AD 6/14/38*	*AD, AR*
WNK1	*605232*	*Neuropathy, hereditary sensory and autonomic, type II;Pseudohypoaldosteronism, type IIC*	*AR, AD*
WT1	*607102*	*Denys-Drash syndrome;Mesothelioma, somatic;Frasier syndrome;Meacham syndrome;Wilms tumor, type 1;Nephrotic syndrome, type 4*	*AD, SM, AD*
ZAP70	*176947*	*Autoimmune disease, multisystem, infantile-onset, 2;Immunodeficiency 48*	*AR*
ZEB2	*605802*	*Mowat-Wilson syndrome*	*AD*
ZFP57	*612192*	*Diabetes mellitus, transient neonatal 1*	*AD*
ZNF423	*604557*	*Joubert syndrome 19;Nephronophthisis 14*	*AD, AR*