COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

GENE	CHROMOSOME	LOCUS	DISEASE	OMIM
AAAS	chr12	12q13	Achalasia-addisonianism-alacrimia syndrome	231550
AARS2	chr6	6p21.1	Combined oxidative phosphorylation deficiency 8	614096
AARS2	chr6	6p21.1	Leukoencephalopathy progressive with ovarian failure	615889
AASS	chr7	7q31.3	Hyperlysinemia	238700
ABAT	chr16	16p13.3	GABA-transaminase deficiency	613163
ABCA1	chr9	9q22-q31	Tangier disease	205400
ABCA12	chr2	2q34	Ichthyosis congenital autosomal recessive 4B (harlequin)	242500
ABCA12	chr2	2q34	Ichthyosis congenital autosomal recessive 4A	601277
ABCA2	chr9	9q34	Intellectual developmental disorder with poor growth and with or without seizures or ataxia	618808
ABCA3	chr16	16p13.3	Surfactant metabolism dysfunction pulmonary 3	610921
ABCA4	chr1	1p22.1	Retinal dystrophy early-onset severe	248200
ABCA4	chr1	1p22.1	Stargardt disease 1	248200
ABCA4	chr1	1p22.1	Fundus flavimaculatus	248200
ABCA4	chr1	1p22.1	Retinitis pigmentosa 19	601718
ABCA5	chr17	17q24.2	Hypertrichosis congenital generalized with gingival hyperplasia	135400
ABCB11	chr2	2q24	Cholestasis progressive familial intrahepatic 2	601847
ABCB11	chr2	2q24	Cholestasis benign recurrent intrahepatic 2	605479
ABCB4	chr7	7q21.1	Gallbladder disease 1	600803
ABCB4	chr7	7q21.1	Cholestasis intrahepatic of pregnancy 3	614972
ABCB4	chr7	7q21.1	Cholestasis progressive familial intrahepatic 3	602347
ABCB7	chrX	Xq13.1-q13.3	Anemia sideroblastic with ataxia	301310
ABCC2	chr10	10q24	Dubin-Johnson syndrome	237500
ABCC6	chr16	16p13.1	Pseudoxanthoma elasticum	264800
ABCC6	chr16	16p13.1	Arterial calcification generalized of infancy 2	614473
ABCC8	chr11	11p15.1	Diabetes mellitus permanent neonatal 3 with or without neurologic features	618857
ABCC8	chr11	11p15.1	Hyperinsulinemic hypoglycemia familial 1	256450
ABCD1	chrX	Xq28	Adrenomyeloneuropathy adult	300100
ABCD1	chrX	Xq28	Adrenoleukodystrophy	300100
ABCD3	chr1	1p22-p21	Bile acid synthesis defect congenital 5	616278
ABCD4	chr14	14q24.3	Methylmalonic aciduria and homocystinuria cblJ type	614857
ABCG8	chr2	2p21	Sitosterolemia 1	210250
ABHD12	chr20	20p11.21	Polyneuropathy hearing loss ataxia retinitis pigmentosa and cataract	612674
ABHD5	chr3	3p21	Chanarin-Dorfman syndrome	275630
ACACA	chr17	17q21	Acetyl-CoA carboxylase deficiency	613933
ACAD8	chr11	11q25	Isobutyryl-CoA dehydrogenase deficiency	611283
ACAD9	chr3	3q26	Mitochondrial complex I deficiency nuclear type 20	611126
ACADM	chr1	1p31	Acyl-CoA dehydrogenase medium chain deficiency of	201450
ACADS	chr12	12q22-qter	Acyl-CoA dehydrogenase short-chain deficiency of	201470
ACADSB	chr10	10q25-q26	2-methylbutyrylglycinuria	610006

ACADVL	chr17	17p13	VLCAD deficiency	201475
ACAN	chr15	15q26.1	Spondyloepimetaphyseal dysplasia aggrecan type	612813
ACAT1	chr11	11q22.3-q23.1	Alpha-methylacetoacetic aciduria	203750
ACBD5	chr10	10p12.1	Retinal dystrophy with leukodystrophy	618863
ACE	chr17	17q23	Renal tubular dysgenesis	267430
ACKR1	chr1	1q23.2	Blood group Duffy system	110700
ACKR1	chr1	1q23.2	White blood cell count QTL	611862
ACO2	chr22	22q13.2	Infantile cerebellar-retinal degeneration	614559
ACO2	chr22	22q13.2	Optic atrophy 9	616289
ACOX1	chr17	17q25.1	Peroxisomal acyl-CoA oxidase deficiency	264470
ACP2	chr11	11p11.2	Lysosomal acid phosphatase deficiency	200950
ACP5	chr19	19p13.3-p13.1	Spondyloenchondrodysplasia with immune dysregulation	607944
ACTA1	chr1	1q42.1	Myopathy actin congenital with cores	161800
ACTA1	chr1	1q42.1	Nemaline myopathy 3 autosomal dominant or recessive	161800
ACTA1	chr1	1q42.1	Myopathy congenital with fiber-type disproportion 1	255310
ACTA1	chr1	1q42.1	Myopathy actin congenital with excess of thin myofilaments	161800
ACY1	chr3	3p21.1	Aminoacylase 1 deficiency	609924
ADA	chr20	20q13.11	Adenosine deaminase deficiency partial	102700
ADA	chr20	20q13.11	Severe combined immunodeficiency due to ADA deficiency	102700
ADA2	chr22	22q11.2	Vasculitis autoinflammation immunodeficiency and hematologic defects syndrome	615688
ADA2	chr22	22q11.2	Sneddon syndrome	182410
ADAM17	chr2	2p25	Inflammatory skin and bowel disease neonatal 1	614328
ADAM22	chr7	7q21	Epileptic encephalopathy early infantile 61	617933
ADAM9	chr8	8p11.23	Cone-rod dystrophy 9	612775
ADAMTS10	chr19	19p13.3-p13.2	Weill-Marchesani syndrome 1 recessive	277600
ADAMTS13	chr9	9q34	Thrombotic thrombocytopenic purpura hereditary	274150
ADAMTS17	chr15	15q24	Weill-Marchesani 4 syndrome recessive	613195
ADAMTS18	chr16	16q23	Microcornea myopic chorioretinal atrophy and telecanthus	615458
ADAMTS2	chr5	5q23	Ehlers-Danlos syndrome dermatosparaxis type	225410
ADAMTSL2	chr9	9q34.2	Geleophysic dysplasia 1	231050
ADAMTSL4	chr1	1q21	Ectopia lentis et pupillae	225200
ADAMTSL4	chr1	1q21	Ectopia lentis isolated autosomal recessive	225100
ADAR	chr1	1q21.3	Dyschromatosis symmetrica hereditaria	127400
ADAR	chr1	1q21.3	Aicardi-Goutieres syndrome 6	615010
ADARB1	chr21	21q22.3	Neurodevelopmental disorder with hypotonia microcephaly and seizures	618862
ADCY1	chr7	7p13-p12	Deafness autosomal recessive 44	610154
ADCY3	chr2	2p24-p22	Obesity susceptibility to BMIQ19	617885
ADCY6	chr12	12q12-q13	Lethal congenital contracture syndrome 8	616287
ADGRG1	chr16	16q13	Polymicrogyria bilateral frontoparietal	606854
ADGRV1	chr5	5q14.3	Usher syndrome type 2C	605472

ADGRV1	chr5	5q14.3	Usher syndrome type 2C GPR98/PDZD7 digenic	605472
ADK	chr10	10q11-q24	Hypermethioninemia due to adenosine kinase deficiency	614300
ADRB2	chr5	5q32-q34	Obesity susceptibility to	601665
ADRB3	chr8	8p12-p11.2	Obesity susceptibility to	601665
ADSL	chr22	22q13.1	Adenylosuccinase deficiency	103050
AEBP1	chr7	7p13	Ehlers-Danlos syndrome classic-like 2	618000
AFF2	chrX	Xq28	Mental retardation X-linked FRAXE type	309548
AFG3L2	chr18	18p11	Spastic ataxia 5 autosomal recessive	614487
AFP	chr4	4q11-q13	Alpha-fetoprotein deficiency	615969
AGA	chr4	4q32-q33	Aspartylglucosaminuria	208400
AGK	chr7	7q34	Sengers syndrome	212350
AGK	chr7	7q34	Cataract 38 autosomal recessive	614691
AGL	chr1	1p21	Glycogen storage disease IIIb	232400
AGL	chr1	1p21	Glycogen storage disease IIIa	232400
AGPAT2	chr9	9q34.3	Lipodystrophy congenital generalized type 1	608594
AGPS	chr2	2q31	Rhizomelic chondrodysplasia punctata type 3	600121
AGRN	chr1	1pter-p32	Myasthenic syndrome congenital 8 with pre- and postsynaptic defects	615120
AGRP	chr16	16q22	Obesity late-onset	601665
AGRP	chr16	16q22	Leanness inherited	601665
AGT	chr1	1q42-q43	Renal tubular dysgenesis	267430
AGTPBP1	chr9	9q21.33	Neurodegeneration childhood-onset with cerebellar atrophy	618276
AGTR1	chr3	3q21-q25	Renal tubular dysgenesis	267430
AGXT	chr2	2q36-q37	Hyperoxaluria primary type 1	259900
AGXT2	chr5	5p13	Beta-aminoisobutyric acid urinary excretion of	210100
AHCY	chr20	20cen-q13.1	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	613752
AHI1	chr6	6q23.3	Joubert syndrome 3	608629
AHR	chr7	7p15	Retinitis pigmentosa 85	618345
AHSG	chr3	3q27	Alopecia-mental retardation syndrome 1	203650
AICDA	chr12	12p13	Immunodeficiency with hyper-IgM type 2	605258
AIFM1	chrX	Xq26.1	Cowchock syndrome	310490
AIFM1	chrX	Xq26.1	Spondyloepimetaphyseal dysplasia X-linked with hypomyelinating leukodystrophy	300232
AIFM1	chrX	Xq26.1	Combined oxidative phosphorylation deficiency 6	300816
AIFM1	chrX	Xq26.1	Deafness X-linked 5	300614
AIMP1	chr4	4q24	Leukodystrophy hypomyelinating 3	260600
AIPL1	chr17	17p13.1	Leber congenital amaurosis 4	604393
AIPL1	chr17	17p13.1	Retinitis pigmentosa juvenile	604393
AIPL1	chr17	17p13.1	Cone-rod dystrophy	604393
AIRE	chr21	21q22.3	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia	240300
AK1	chr9	9q34.1	Hemolytic anemia due to adenylate kinase deficiency	612631
AK2	chr1	1p34	Reticular dysgenesis	267500

AK7	chr14	14q32.2	Spermatogenic failure 27	617965
AKR1C2	chr10	10p15-p14	46XY sex reversal 8	614279
AKR1C4	chr10	10p15-p14	46XY sex reversal 8 modifier of	614279
AKR1D1	chr7	7q32-q33	Bile acid synthesis defect congenital 2	235555
ALAD	chr9	9q34	Porphyria acute hepatic	612740
ALAD	chr9	9q34	Lead poisoning susceptibility to	612740
ALAS2	chrX	Xp11.21	Protoporphyria erythropoietic X-linked	300752
ALAS2	chrX	Xp11.21	Anemia sideroblastic 1	300751
ALDH18A1	chr10	10q24.3	Cutis laxa autosomal recessive type IIIA	219150
ALDH18A1	chr10	10q24.3	Spastic paraplegia 9B autosomal recessive	616586
ALDH1A3	chr15	15q26	Microphthalmia isolated 8	615113
ALDH3A2	chr17	17p11.2	Sjogren-Larsson syndrome	270200
ALDH4A1	chr1	1p36	Hyperprolinemia type II	239510
ALDH5A1	chr6	6p22	Succinic semialdehyde dehydrogenase deficiency	271980
ALDH6A1	chr14	14q24.3	Methylmalonate semialdehyde dehydrogenase deficiency	614105
ALDH7A1	chr5	5q31	Epilepsy pyridoxine-dependent	266100
ALDOA	chr16	16p11.2	Glycogen storage disease XII	611881
ALDOB	chr9	9q22.3	Fructose intolerance hereditary	229600
ALG1	chr16	16p13.3	Congenital disorder of glycosylation type Ik	608540
ALG11	chr13	13q14.3	Congenital disorder of glycosylation type Ip	613661
ALG12	chr22	22q13.33	Congenital disorder of glycosylation type Ig	607143
ALG14	chr1	1p21.3	Myasthenic syndrome congenital 15 without tubular aggregates	616227
ALG2	chr9	9q22	Myasthenic syndrome congenital 14 with tubular aggregates	616228
ALG2	chr9	9q22	Congenital disorder of glycosylation type Ii	607906
ALG3	chr3	3q27	Congenital disorder of glycosylation type Id	601110
ALG6	chr1	1p22.3	Congenital disorder of glycosylation type Ic	603147
ALG8	chr11	11pter-p15.5	Congenital disorder of glycosylation type Ih	608104
ALG8	chr11	11pter-p15.5	Polycystic liver disease 3 with or without kidney cysts	617874
ALG9	chr11	11q23	Gillessen-Kaesbach-Nishimura syndrome	263210
ALG9	chr11	11q23	Congenital disorder of glycosylation type Il	608776
ALMS1	chr2	2p13	Alstrom syndrome	203800
ALOX12B	chr17	17p13.1	Ichthyosis congenital autosomal recessive 2	242100
ALOXE3	chr17	17p13.1	Ichthyosis congenital autosomal recessive 3	606545
ALPL	chr1	1p36.1-p34	Hypophosphatasia adult	146300
ALPL	chr1	1p36.1-p34	Odontohypophosphatasia	146300
ALPL	chr1	1p36.1-p34	Hypophosphatasia childhood	241510
ALPL	chr1	1p36.1-p34	Hypophosphatasia infantile	241500
ALS2	chr2	2q33	Primary lateral sclerosis juvenile	606353
ALS2	chr2	2q33	Amyotrophic lateral sclerosis 2 juvenile	205100
ALS2	chr2	2q33	Spastic paralysis infantile onset ascending	607225
ALX1	chr12	12q21.3-q22	Frontonasal dysplasia 3	613456
ALX3	chr1	1p21-p13	Frontonasal dysplasia 1	136760
ALX4	chr11	11p11.2	Frontonasal dysplasia 2	613451
AMACR	chr5	5p13.2-q11.1	Bile acid synthesis defect congenital 4	214950

AMACR	chr5	5p13.2-q11.1	Alpha-methylacyl-CoA racemase deficiency	614307
AMBN	chr4	4q21	Amelogenesis imperfecta type IF	616270
AMH	chr19	19p13.3-p13.2	Persistent Mullerian duct syndrome type I	261550
AMHR2	chr12	12q13	Persistent Mullerian duct syndrome type II	261550
AMN	chr14	14q32	Imerslund-Grasbeck syndrome 2	618882
AMPD1	chr1	1p21-p13	Myopathy due to myoadenylate deaminase deficiency	615511
AMPD2	chr1	1p13.3	Spastic paraplegia 63	615686
AMPD2	chr1	1p13.3	Pontocerebellar hypoplasia type 9	615809
AMPD3	chr11	11pter-p13	AMP deaminase deficiency erythrocytic	612874
AMT	chr3	3p21.2-p21.1	Glycine encephalopathy	605899
ANAPC1	chr2	2q13	Rothmund-Thomson syndrome type 1	618625
ANGPTL3	chr1	1p31	Hypobetalipoproteinemia familial 2	605019
ANK1	chr8	8p11.2	Spherocytosis type 1	182900
ANK3	chr10	10q21	Mental retardation autosomal recessive 37	615493
ANKLE2	chr12	12q24.33	Microcephaly 16 primary autosomal recessive	616681
ANKS6	chr9	9q22.33	Nephronophthisis 16	615382
ANO10	chr3	3p22.1	Spinocerebellar ataxia autosomal recessive 10	613728
ANO5	chr11	11p14.3	Miyoshi muscular dystrophy 3	613319
ANO5	chr11	11p14.3	Muscular dystrophy limb-girdle autosomal recessive 12	611307
ANO6	chr12	12q12	Scott syndrome	262890
ANOS1	chrX	Xp22.31	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	308700
ANTXR1	chr2	2p13.1	GAPO syndrome	230740
ANTXR2	chr4	4q21	Hyaline fibromatosis syndrome	228600
AP1S1	chr7	7q22.1	MEDNIK syndrome	609313
AP1S2	chrX	Xp22	Mental retardation X-linked syndromic 5	304340
AP3B1	chr5	5q14.1	Hermansky-Pudlak syndrome 2	608233
AP3B2	chr15	15q25.2	Epileptic encephalopathy early infantile 48	617276
AP3D1	chr19	19p13.3	Hermansky-Pudlak syndrome 10	617050
AP4B1	chr1	1p13.2	Spastic paraplegia 47 autosomal recessive	614066
AP4E1	chr15	15q21.2	Spastic paraplegia 51 autosomal recessive	613744
AP4M1	chr7	7q22.1	Spastic paraplegia 50 autosomal recessive	612936
AP4S1	chr14	14q12	Spastic paraplegia 52 autosomal recessive	614067
AP5Z1	chr7	7p22.1	Spastic paraplegia 48 autosomal recessive	613647
APC2	chr19	19p13.3	Sotos syndrome 3	617169
APC2	chr19	19p13.3	Cortical dysplasia complex with other brain malformations 10	618677
APOB	chr2	2p24	Hypobetalipoproteinemia	615558
APOC2	chr19	19q13.2	Hyperlipoproteinemia type Ib	207750
APOE	chr19	19q13.2	Sea-blue histiocyte disease	269600
APRT	chr16	16q24.3	Adenine phosphoribosyltransferase deficiency	614723
APTX	chr9	9p13.3	Ataxia early-onset with oculomotor apraxia and hypoalbuminemia	208920
AQP2	chr12	12q13	Diabetes insipidus nephrogenic	125800
AQP7	chr9	9p13	Glycerol quantitative trait locus	614411

AR	chrX	Xq11-q12	Hypospadias 1 X-linked	300633
AR	chrX	Xq11-q12	Androgen insensitivity	300068
AR	chrX	Xq11-q12	Androgen insensitivity partial with or without breast cancer	312300
AR	chrX	Xq11-q12	Spinal and bulbar muscular atrophy of Kennedy	313200
ARFGEF2	chr20	20q13.13	Periventricular heterotopia with microcephaly	608097
ARG1	chr6	6q23	Argininemia	207800
ARHGDIA	chr17	17q25.3	Nephrotic syndrome type 8	615244
ARHGEF9	chrX	Xq22.1	Epileptic encephalopathy early infantile 8	300607
ARL13B	chr3	3q11.2	Joubert syndrome 8	612291
ARL2BP	chr16	16q13	Retinitis pigmentosa with or without situs inversus	615434
ARL3	chr10	10q23.3	Joubert syndrome 35	618161
ARL6	chr3	3p12-q13	Bardet-Biedl syndrome 3	600151
ARL6	chr3	3p12-q13	Bardet-Biedl syndrome 1 modifier of	209900
ARL6IP1	chr16	16p12-p11.2	Spastic paraplegia 61 autosomal recessive	615685
ARMC4	chr10	10p12.1	Ciliary dyskinesia primary 23	615451
ARNT2	chr15	15q24	Webb-Dattani syndrome	615926
ARSA	chr22	22q13.31-qter	Metachromatic leukodystrophy	250100
ARSB	chr5	5q11-q13	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200
ARV1	chr1	1q42.2	Epileptic encephalopathy early infantile 38	617020
ARX	chrX	Xp22.13	Epileptic encephalopathy early infantile 1	308350
ARX	chrX	Xp22.13	Lissencephaly X-linked 2	300215
ARX	chrX	Xp22.13	Proud syndrome	300004
ARX	chrX	Xp22.13	Mental retardation X-linked 29 and others	300419
ARX	chrX	Xp22.13	Partington syndrome	309510
ARX	chrX	Xp22.13	Hydranencephaly with abnormal genitalia	300215
ASAH1	chr8	8p22-p21.3	Farber lipogranulomatosis	228000
ASAH1	chr8	8p22-p21.3	Spinal muscular atrophy with progressive myoclonic epilepsy	159950
ASCC1	chr10	10q22.1	Spinal muscular atrophy with congenital bone fractures 2	616867
ASL	chr7	7cen-q11.2	Argininosuccinic aciduria	207900
ASNS	chr7	7q21-q31	Asparagine synthetase deficiency	615574
ASPA	chr17	17pter-p13	Canavan disease	271900
ASPH	chr8	8q12.1	Traboulsi syndrome	601552
ASPM	chr1	1q31	Microcephaly 5 primary autosomal recessive	608716
ASS1	chr9	9q34.1	Citrullinemia	215700
ATAD3A	chr1	1p36.33	Harel-Yoon syndrome	617183
ATAD3A	chr1	1p36.33	Pontocerebellar hypoplasia hypotonia and respiratory insufficiency syndrome neonatal lethal	618810
ATCAY	chr19	19p13.3	Ataxia cerebellar Cayman type	601238
ATF6	chr1	1q22-q23	Achromatopsia 7	616517
ATIC	chr2	2q35	AICA-ribosiduria due to ATIC deficiency	608688
ATM	chr11	11q22.3	Ataxia-telangiectasia	208900
ATOH7	chr10	10q21.3-q22.1	Persistent hyperplastic primary vitreous autosomal recessive	221900
ATP13A2	chr1	1p36	Kufor-Rakeb syndrome	606693
ATP13A2	chr1	1p36	Spastic paraplegia 78 autosomal recessive	617225

ATP2A1	chr16	16p12	Brody myopathy	601003
ATP2B2	chr3	3p26-p25	Deafness autosomal recessive 12 modifier of	601386
ATP2B3	chrX	Xq28	Spinocerebellar ataxia X-linked 1	302500
ATP6AP2	chrX	Xp11.4	Congenital disorder of glycosylation type IIr	301045
ATP6AP2	chrX	Xp11.4	Mental retardation X-linked syndromic Hedera type	300423
ATP6AP2	chrX	Xp11.4	Parkinsonism with spasticity X-linked	300911
ATP6V0A2	chr12	12q24.3	Wrinkly skin syndrome	278250
ATP6V0A2	chr12	12q24.3	Cutis laxa autosomal recessive type IIA	219200
ATP6V1B1	chr2	2cen-q13	Renal tubular acidosis with deafness	267300
ATP7A	chrX	Xq12-q13	Occipital horn syndrome	304150
ATP7A	chrX	Xq12-q13	Menkes disease	309400
ATP7A	chrX	Xq12-q13	Spinal muscular atrophy distal X-linked 3	300489
ATP7B	chr13	13q14.3-q21.1	Wilson disease	277900
ATP8A2	chr13	13q12	Cerebellar ataxia mental retardation and dysequilibrium syndrome 4	615268
ATP8B1	chr18	18q21	Cholestasis progressive familial intrahepatic 1	211600
ATP8B1	chr18	18q21	Cholestasis benign recurrent intrahepatic	243300
ATPAF2	chr17	17p11.2	Mitochondrial complex V (ATP synthase) deficiency nuclear type 1	604273
ATR	chr3	3q22-q24	Seckel syndrome 1	210600
ATRX	chrX	Xq13	Mental retardation-hypotonic facies syndrome X-linked	309580
AUH	chr9	9q22.31	3-methylglutaconic aciduria type I	250950
AURKC	chr19	19q13.43	Spermatogenic failure 5	243060
AVPR2	chrX	Xq28	Nephrogenic syndrome of inappropriate antidiuresis	300539
AVPR2	chrX	Xq28	Diabetes insipidus nephrogenic	304800
B2M	chr15	15q21-q22	Immunodeficiency 43	241600
B3GALNT2	chr1	1q42.3	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11	615181
B3GALT6	chr1	1p36.3	Ehlers-Danlos syndrome spondylodysplastic type 2	615349
B3GALT6	chr1	1p36.3	Spondyloepimetaphyseal dysplasia with joint laxity type 1 with or without fractures	271640
B3GAT3	chr11	11q12-q13	Multiple joint dislocations short stature craniofacial dysmorphism with or without congenital heart defects	245600
B3GLCT	chr13	13q12.3	Peters-plus syndrome	261540
B4GALNT1	chr12	12q13.3	Spastic paraplegia 26 autosomal recessive	609195
B4GALT1	chr9	9p13	Congenital disorder of glycosylation type IId	607091
B4GALT7	chr5	5q35.2-q35.3	Ehlers-Danlos syndrome spondylodysplastic type 1	130070
B4GAT1	chr11	11q13.2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 13	615287
B9D1	chr17	17p11.2	Meckel syndrome 9	614209
B9D1	chr17	17p11.2	Joubert syndrome 27	617120
B9D2	chr19	19q13.2	Joubert syndrome 34	614175

B9D2	chr19	19q13.2	Meckel syndrome 10	614175
BAAT	chr9	9q22.3	Hypercholanemia familial	607748
BANF1	chr11	11q13.1	Nestor-Guillermo progeria syndrome	614008
BBIP1	chr10	10q25.2	Bardet-Biedl syndrome 18	615995
BBS1	chr11	11q13	Bardet-Biedl syndrome 1	209900
BBS10	chr12	12q21.2	Bardet-Biedl syndrome 10	615987
BBS12	chr4	4q27	Bardet-Biedl syndrome 12	615989
BBS2	chr16	16q21	Bardet-Biedl syndrome 2	615981
BBS2	chr16	16q21	Retinitis pigmentosa 74	616562
BBS4	chr15	15q22.3-q23	Bardet-Biedl syndrome 4	615982
BBS5	chr2	2q31	Bardet-Biedl syndrome 5	615983
BBS7	chr4	4q27	Bardet-Biedl syndrome 7	615984
BBS9	chr7	7p14	Bardet-Biedl syndrome 9	615986
BCAM	chr19	19q13.2	Blood group Lutheran null	247420
BCAP31	chrX	Xq28	Deafness dystonia and cerebral hypomyelination	300475
BCAT2	chr19	19q13	Hypervalinemia or hyperleucine-isoleucinemia	618850
BCKDHA	chr19	19q13.1-q13.2	Maple syrup urine disease type Ia	248600
BCKDHB	chr6	6q14	Maple syrup urine disease type Ib	248600
BCL10	chr1	1p22	Immunodeficiency 37	616098
BCORL1	chrX	Xq25-q26.1	Shukla-Vernon syndrome	301029
BCS1L	chr2	2q33	Leigh syndrome	256000
BCS1L	chr2	2q33	GRACILE syndrome	603358
BCS1L	chr2	2q33	Bjornstad syndrome	262000
BCS1L	chr2	2q33	Mitochondrial complex III deficiency nuclear type 1	124000
BDP1	chr5	5q13	Deafness autosomal recessive 112	618257
BEST1	chr11	11q13	Bestrophinopathy autosomal recessive	611809
BFSP1	chr20	20p12.1-p11.23	Cataract 33 multiple types	611391
BGN	chrX	Xq28	Meester-Loeys syndrome	300989
BGN	chrX	Xq28	Spondyloepimetaphyseal dysplasia X-linked	300106
BHLHA9	chr17	17p13.3	Syndactyly mesoaxial synostotic with phalangeal reduction	609432
BHLHA9	chr17	17p13.3	Camptosynpolydactyly complex	607539
BIN1	chr2	2q14	Centronuclear myopathy 2	255200
BLM	chr15	15q26.1	Bloom syndrome	210900
BLNK	chr10	10q23.2	Agammaglobulinemia 4	613502
BLOC1S3	chr19	19q13	Hermansky-Pudlak syndrome 8	614077
BLOC1S6	chr15	15q15	Hermansky-pudlak syndrome 9	614171
BLVRA	chr7	7p13	Hyperbiliverdinemia	614156
BMP1	chr8	8p21	Osteogenesis imperfecta type XIII	614856
BMP2	chr20	20p12	HFE hemochromatosis modifier of	235200
BMPER	chr7	7p14.3	Diaphanospondylodysostosis	608022
BMPR1B	chr4	4q23-q24	Acromesomelic dysplasia Demirhan type	609441
BOLA3	chr2	2p13.1	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia	614299
BPGM	chr7	7q31-q34	Erythrocytosis familial 8	222800
BRAT1	chr7	7p22.3	Rigidity and multifocal seizure syndrome lethal neonatal	614498

BRAT1	chr7	7p22.3	Neurodevelopmental disorder with cerebellar atrophy and with or without seizures	618056
BRF1	chr14	14q32.33	Cerebellofaciodental syndrome	616202
BRWD3	chrX	Xq13	Mental retardation X-linked 93	300659
BSCL2	chr11	11q13	Lipodystrophy congenital generalized type 2	269700
BSCL2	chr11	11q13	Encephalopathy progressive with or without lipodystrophy	615924
BSND	chr1	1p31	Sensorineural deafness with mild renal dysfunction	602522
BSND	chr1	1p31	Bartter syndrome type 4a	602522
BTD	chr3	3p25	Biotinidase deficiency	253260
BTK	chrX	Xq21.3-q22	Isolated growth hormone deficiency type III with agammaglobulinemia	307200
BTK	chrX	Xq21.3-q22	Agammaglobulinemia X-linked 1	300755
BUB1B	chr15	15q15	Mosaic variegated aneuploidy syndrome 1	257300
BVES	chr6	6q21	Muscular dystrophy limb-girdle autosomal recessive 25	616812
C12orf4	chr12	12p13.3	Mental retardation autosomal recessive 66	618221
C12orf57	chr12	12p13	Temtamy syndrome	218340
C12orf65	chr12	12q24.31	Spastic paraplegia 55 autosomal recessive	615035
C12orf65	chr12	12q24.31	Combined oxidative phosphorylation deficiency 7	613559
C19orf12	chr19	19q12	Neurodegeneration with brain iron accumulation 4	614298
C19orf12	chr19	19q12	Spastic paraplegia 43 autosomal recessive	615043
C1QA	chr1	1p36.3-p34.1	C1q deficiency	613652
C1QB	chr1	1p36.3-p34.1	C1q deficiency	613652
C1QC	chr1	1p36.3-p34.1	C1q deficiency	613652
C2	chr6	6p21.3	C2 deficiency	217000
C2CD3	chr11	11q13.4	Orofaciodigital syndrome XIV	615948
C3	chr19	19p13.3-p13.2	Hemolytic uremic syndrome atypical susceptibility to 5	612925
C3	chr19	19p13.3-p13.2	C3 deficiency	613779
C4A	chr6	6p21.3	C4a deficiency	614380
C8A	chr1	1p32	C8 deficiency type I	613790
C8B	chr1	1p32	C8 deficiency type II	613789
C8orf37	chr8	8q22.1	Retinitis pigmentosa 64	614500
C8orf37	chr8	8q22.1	Bardet-Biedl syndrome 21	617406
C8orf37	chr8	8q22.1	Cone-rod dystrophy 16	614500
CA12	chr15	15q22	Hyperchlorhidrosis isolated	143860
CA2	chr8	8q22	Osteopetrosis autosomal recessive 3 with renal tubular acidosis	259730
CA5A	chr16	16q24.3	Hyperammonemia due to carbonic anhydrase VA deficiency	615751
CA8	chr8	8q11-q12	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3	613227
CABP2	chr11	11q13.1	Deafness autosomal recessive 93	614899
CABP4	chr11	11q13.1	Cone-rod synaptic disorder congenital nonprogressive	610427
CACNA1B	chr9	9q34	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements	618497
CACNA1D	chr3	3p21.1	Sinoatrial node dysfunction and deafness	614896
CACNA1F	chrX	Xp11.23	Cone-rod dystrophy X-linked 3	300476

CACNA1F	chrX	Xp11.23	Night blindness congenital stationary (incomplete) 2A X-linked	300071
CACNA1F	chrX	Xp11.23	Aland Island eye disease	300600
CACNA2D2	chr3	3p21.3	Cerebellar atrophy with seizures and variable developmental delay	618501
CACNA2D4	chr12	12p13.3	Retinal cone dystrophy 4	610478
CALCRL	chr2	2q31-q32	Lymphatic malformation 8	618773
CANT1	chr17	17q25.3	Desbuquois dysplasia 1	251450
CANT1	chr17	17q25.3	Epiphyseal dysplasia multiple 7	617719
CAPN1	chr11	11q13	Spastic paraplegia 76 autosomal recessive	616907
CAPN3	chr15	15q15.1-q21.1	Muscular dystrophy limb-girdle autosomal recessive 1	253600
CARD11	chr7	7p22	Immunodeficiency 11A	615206
CARD9	chr9	9q34.3	Candidiasis familial 2 autosomal recessive	212050
CARS2	chr13	13q34	Combined oxidative phosphorylation deficiency 27	616672
CARTPT	chr5	5q13.2	Obesity susceptibility to	601665
CASP8	chr2	2q33	Autoimmune lymphoproliferative syndrome type IIB	607271
CASQ2	chr1	1p13.3-p11	Ventricular tachycardia catecholaminergic polymorphic 2	611938
CASR	chr3	3q21.1	Hyperparathyroidism neonatal	239200
CAST	chr5	5q15-q21	Peeling skin with leukonychia acral punctate keratoses cheilitis and knuckle pads	616295
CATSPER1	chr11	11q13.1	Spermatogenic failure 7	612997
CAV1	chr7	7q31.1	Lipodystrophy congenital generalized type 3	612526
CAVIN1	chr17	17q21	Lipodystrophy congenital generalized type 4	613327
CBS	chr21	21q22.3	Homocystinuria B6-responsive and nonresponsive types	236200
CBS	chr21	21q22.3	Thrombosis hyperhomocysteinemic	236200
CBX2	chr17	17q25	46XY sex reversal 5	613080
CC2D1A	chr19	19p13.12	Mental retardation autosomal recessive 3	608443
CC2D2A	chr4	4p15.3	Meckel syndrome 6	612284
CC2D2A	chr4	4p15.3	Joubert syndrome 9	612285
CC2D2A	chr4	4p15.3	COACH syndrome	216360
CCBE1	chr18	18q21.32	Hennekam lymphangiectasia-lymphedema syndrome 1	235510
CCDC103	chr17	17q21.31	Ciliary dyskinesia primary 17	614679
CCDC114	chr19	19q13.3	Ciliary dyskinesia primary 20	615067
CCDC151	chr19	19p13.2	Ciliary dyskinesia primary 30	616037
CCDC22	chrX	Xp11.23	Ritscher-Schinzel syndrome 2	300963
CCDC28B	chr1	1p35.1	Bardet-Biedl syndrome 1 modifier of	209900
CCDC65	chr12	12q13.12	Ciliary dyskinesia primary 27	615504
CCDC8	chr19	19q13.2-q13.32	3-M syndrome 3	614205
CCDC88C	chr14	14q32.11	Hydrocephalus congenital 1	236600
CCNO	chr5	5q11.2	Ciliary dyskinesia primary 29	615872
CCT5	chr5	5p15.2	Neuropathy hereditary sensory with spastic paraplegia	256840
CD19	chr16	16p11.2	Immunodeficiency common variable 3	613493
CD247	chr1	1q22-q23	Immunodeficiency 25	610163

CD27	chr12	12p13	Lymphoproliferative syndrome 2	615122
CD36	chr7	7q21.11	Platelet glycoprotein IV deficiency	608404
CD3D	chr11	11q23	Immunodeficiency 19	615617
CD3E	chr11	11q23	Immunodeficiency 18 SCID variant	615615
CD3E	chr11	11q23	Immunodeficiency 18	615615
CD3G	chr11	11q23	Immunodeficiency 17 CD3 gamma deficient	615607
CD40	chr20	20q12-q13.2	Immunodeficiency with hyper-IgM type 3	606843
CD40LG	chrX	Xq26	Immunodeficiency X-linked with hyper-IgM	308230
CD46	chr1	1q32	Hemolytic uremic syndrome atypical susceptibility to 2	612922
CD55	chr1	1q32	Blood group Cromer	613793
CD55	chr1	1q32	Complement hyperactivation angiopathic thrombosis and protein-losing enteropathy	226300
CD59	chr11	11p13	Hemolytic anemia CD59-mediated with or without immune-mediated polyneuropathy	612300
CD79A	chr19	19q13.2	Agammaglobulinemia 3	613501
CD79B	chr17	17q23	Agammaglobulinemia 6	612692
CD81	chr11	11p	Immunodeficiency common variable 6	613496
CD8A	chr2	2p12	CD8 deficiency familial	608957
CDAN1	chr15	15q15	Dyserythropoietic anemia congenital type Ia	224120
CDC6	chr17	17q21.2	Meier-Gorlin syndrome 5	613805
CDH23	chr10	10q21-q22	Pituitary adenoma 5 multiple types	617540
CDH23	chr10	10q21-q22	Deafness autosomal recessive 12	601386
CDH23	chr10	10q21-q22	Usher syndrome type 1D/F digenic	601067
CDH23	chr10	10q21-q22	Usher syndrome type 1D	601067
CDH3	chr16	16q22.1	Ectodermal dysplasia ectrodactyly and macular dystrophy	225280
CDH3	chr16	16q22.1	Hypotrichosis congenital with juvenile macular dystrophy	601553
CDHR1	chr10	10q23.1	Cone-rod dystrophy 15	613660
CDHR1	chr10	10q23.1	Retinitis pigmentosa 65	613660
CDK5	chr7	7q36	Lissencephaly 7 with cerebellar hypoplasia	616342
CDK5RAP2	chr9	9q33.3	Microcephaly 3 primary autosomal recessive	604804
CDK6	chr7	7q21-q22	Microcephaly 12 primary autosomal recessive	616080
CDSN	chr6	6p21.3	Peeling skin syndrome 1	270300
CDT1	chr16	16q24.3	Meier-Gorlin syndrome 4	613804
CEACAM16	chr19	19q13.31	Deafness autosomal recessive 113	618410
CEBPE	chr14	14q11.2	Specific granule deficiency	245480
CENPE	chr4	4q24-q25	Microcephaly 13 primary autosomal recessive	616051
CENPJ	chr13	13q12.2	Microcephaly 6 primary autosomal recessive	608393
CENPJ	chr13	13q12.2	Seckel syndrome 4	613676
CEP120	chr5	5q23.2	Joubert syndrome 31	617761
CEP120	chr5	5q23.2	Short-rib thoracic dysplasia 13 with or without polydactyly	616300
CEP135	chr4	4q12	Microcephaly 8 primary autosomal recessive	614673
CEP152	chr15	15q21.1	Microcephaly 9 primary autosomal recessive	614852
CEP152	chr15	15q21.1	Seckel syndrome 5	613823
CEP164	chr11	11q23.3	Nephronophthisis 15	614845

CEP19	chr3	3q29	Morbid obesity and spermatogenic failure	615703
CEP250	chr20	20q11.2	Cone-rod dystrophy and hearing loss 2	618358
CEP290	chr12	12q21.3	Bardet-Biedl syndrome 14	615991
CEP290	chr12	12q21.3	Senior-Loken syndrome 6	610189
CEP290	chr12	12q21.3	Meckel syndrome 4	611134
CEP290	chr12	12q21.3	Joubert syndrome 5	610188
CEP41	chr7	7q32	Joubert syndrome 15	614464
CEP57	chr11	11q21	Mosaic variegated aneuploidy syndrome 2	614114
CEP63	chr3	3q22.2	Seckel syndrome 6	614728
CEP83	chr12	12q22	Nephronophthisis 18	615862
CERS3	chr15	15q26.3	Ichthyosis congenital autosomal recessive 9	615023
CFAP53	chr18	18q21.1	Heterotaxy visceral 6 autosomal recessive	614779
CFB	chr6	6p21.3	Complement factor B deficiency	615561
CFD	chr19	19p13.3	Complement factor D deficiency	613912
CFH	chr1	1q32	Complement factor H deficiency	609814
CFH	chr1	1q32	Hemolytic uremic syndrome atypical susceptibility to 1	235400
CFHR1	chr1	1q31-q32.1	Hemolytic uremic syndrome atypical susceptibility to	235400
CFHR3	chr1	1q31-q32.1	Hemolytic uremic syndrome atypical susceptibility to	235400
CFI	chr4	4q25	Complement factor I deficiency	610984
CFL2	chr14	14q12	Nemaline myopathy 7 autosomal recessive	610687
CFP	chrX	Xp11.4-p11.23	Properdin deficiency X-linked	312060
CFTR	chr7	7q31.2	Cystic fibrosis	219700
CFTR	chr7	7q31.2	Congenital bilateral absence of vas deferens	277180
CHAT	chr10	10q11.2	Myasthenic syndrome congenital 6 presynaptic	254210
CHIT1	chr1	1q31-q32	Chitotriosidase deficiency	614122
CHKB	chr22	22q13	Muscular dystrophy congenital megaconial type	602541
CHMP1A	chr16	16q24.3	Pontocerebellar hypoplasia type 8	614961
CHRDL1	chrX	Xq22.1-q23	Megalocornea 1 X-linked	309300
CHRM3	chr1	1q43	Prune belly syndrome	100100
CHRNA1	chr2	2q24-q32	Myasthenic syndrome congenital 1B fast-channel	608930
CHRNA1	chr2	2q24-q32	Multiple pterygium syndrome lethal type	253290
CHRNA3	chr15	15q25.1	Bladder dysfunction autonomic with impaired pupillary reflex and secondary CAKUT	191800
CHRNB1	chr17	17p12-p11	Myasthenic syndrome congenital 2C associated with acetylcholine receptor deficiency	616314
CHRND	chr2	2q33-q34	Myasthenic syndrome congenital 3C associated with acetylcholine receptor deficiency	616323
CHRND	chr2	2q33-q34	Myasthenic syndrome congenital 3B fast-channel	616322
CHRND	chr2	2q33-q34	Multiple pterygium syndrome lethal type	253290
CHRNE	chr17	17p13-p12	Myasthenic syndrome congenital 4A slow-channel	605809
CHRNE	chr17	17p13-p12	Myasthenic syndrome congenital 4C associated with acetylcholine receptor deficiency	608931
CHRNE	chr17	17p13-p12	Myasthenic syndrome congenital 4B fast-channel	616324
CHRNG	chr2	2q33-q34	Escobar syndrome	265000
CHRNG	chr2	2q33-q34	Multiple pterygium syndrome lethal type	253290

CHST14	chr15	15q14	Ehlers-Danlos syndrome musculocontractural type 1	601776
CHST3	chr10	10q22.1	Spondyloepiphyseal dysplasia with congenital joint dislocations	143095
CHST6	chr16	16q22	Macular corneal dystrophy	217800
CHST8	chr19	19q13.1	Peeling skin syndrome 3	616265
CHSY1	chr15	15q26.3	Temtamy preaxial brachydactyly syndrome	605282
CIB2	chr15	15q24	Deafness autosomal recessive 48	609439
CIB2	chr15	15q24	Usher syndrome type IJ	614869
CIDEC	chr3	3p25	Lipodystrophy familial partial type 5	615238
CIITA	chr16	16p13	Bare lymphocyte syndrome type II complementation group A	209920
CISD2	chr4	4q22-q24	Wolfram syndrome 2	604928
CKAP2L	chr2	2q13	Filippi syndrome	272440
CLCF1	chr11	11q13.3	Cold-induced sweating syndrome 2	610313
CLCN1	chr7	7q35	Myotonia congenita recessive	255700
CLCN2	chr3	3q26	Leukoencephalopathy with ataxia	615651
CLCN5	chrX	Xp11.22	Proteinuria low molecular weight with hypercalciuric nephrocalcinosis	308990
CLCN5	chrX	Xp11.22	Dent disease	300009
CLCN5	chrX	Xp11.22	Hypophosphatemic rickets	300554
CLCN5	chrX	Xp11.22	Nephrolithiasis type I	310468
CLCN7	chr16	16p13	Osteopetrosis autosomal recessive 4	611490
CLCNKA	chr1	1p36	Bartter syndrome type 4b digenic	613090
CLCNKB	chr1	1p36	Bartter syndrome type 3	607364
CLCNKB	chr1	1p36	Bartter syndrome type 4b digenic	613090
CLDN1	chr3	3q28-q29	Ichthyosis leukocyte vacuoles alopecia and sclerosing cholangitis	607626
CLDN14	chr21	21q22.3	Deafness autosomal recessive 29	614035
CLDN16	chr3	3q27	Hypomagnesemia 3 renal	248250
CLDN19	chr1	1p34.2	Hypomagnesemia 5 renal with ocular involvement	248190
CLEC7A	chr12	12p13.2-p12.3	Candidiasis familial 4 autosomal recessive	613108
CLIC2	chrX	Xq28	Mental retardation X-linked syndromic 32	300886
CLIC5	chr6	6p21.1-p12.1	Deafness autosomal recessive 103	616042
CLMP	chr11	11q24.1	Congenital short bowel syndrome	615237
CLN3	chr16	16p12.1	Ceroid lipofuscinosis neuronal 3	204200
CLN5	chr13	13q22.3	Ceroid lipofuscinosis neuronal 5	256731
CLN6	chr15	15q21-q23	Ceroid lipofuscinosis neuronal Kufs type adult onset	204300
CLN6	chr15	15q21-q23	Ceroid lipofuscinosis neuronal 6	601780
CLN8	chr8	8p23	Ceroid lipofuscinosis neuronal 8	600143
CLN8	chr8	8p23	Ceroid lipofuscinosis neuronal 8 Northern epilepsy variant	610003
CLP1	chr11	11q12.1	Pontocerebellar hypoplasia type 10	615803
CLPB	chr11	11q13.4	3-methylglutaconic aciduria type VII with cataracts neurologic involvement and neutropenia	616271
CLPP	chr19	19p13.3	Perrault syndrome 3	614129
CLRN1	chr3	3q21-q25	Usher syndrome type 3A	276902
CNGA3	chr2	2q11	Achromatopsia 2	216900

CNGB1	chr16	16q21	Retinitis pigmentosa 45	613767
CNGB3	chr8	8q21-q22	Achromatopsia 3	262300
CNNM2	chr10	10q24.33	Hypomagnesemia seizures and mental retardation	616418
CNNM4	chr2	2q11.2	Jalili syndrome	217080
CNPY3	chr6	6pter-p12	Epileptic encephalopathy early infantile 60	617929
CNTN1	chr12	12q11-q12	Myopathy congenital Compton-North	612540
CNTN2	chr1	1q32.1	Epilepsy myoclonic familial adult 5	615400
CNTNAP1	chr17	17q21	Lethal congenital contracture syndrome 7	616286
CNTNAP1	chr17	17q21	Hypomyelinating neuropathy congenital 3	618186
CNTNAP2	chr7	7q35-q36	Pitt-Hopkins like syndrome 1	610042
CNTNAP2	chr7	7q35-q36	Cortical dysplasia-focal epilepsy syndrome	610042
COA5	chr2	2q11.2	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 3	616500
COA6	chr1	1q42.2	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 4	616501
COASY	chr17	17q12-q21	Neurodegeneration with brain iron accumulation 6	615643
COASY	chr17	17q12-q21	Pontocerebellar hypoplasia type 12	618266
COCH	chr14	14q12-q13	Deafness autosomal recessive 110	618094
COG1	chr17	17q25.1	Congenital disorder of glycosylation type IIg	611209
COG2	chr1	1q42.2	Congenital disorder of glycosylation type IIq	617395
COG4	chr16	16q22.1	Congenital disorder of glycosylation type IIj	613489
COG5	chr7	7q31	Congenital disorder of glycosylation type IIi	613612
COG6	chr13	13q13.3	Congenital disorder of glycosylation type III	614576
COG6	chr13	13q13.3	Shaheen syndrome	615328
COG7	chr16	16p	Congenital disorder of glycosylation type IIe	608779
COL11A1	chr1	1p21	Fibrochondrogenesis 1	228520
COL11A2	chr6	6p21.3	Otospondylomegaepiphyseal dysplasia autosomal recessive	215150
COL11A2	chr6	6p21.3	Fibrochondrogenesis 2	614524
COL11A2	chr6	6p21.3	Deafness autosomal recessive 53	609706
COL17A1	chr10	10q24.3	Epidermolysis bullosa junctional non-Herlitz type	226650
COL17A1	chr10	10q24.3	Epidermolysis bullosa junctional localisata variant	226650
COL18A1	chr21	21q22.3	Knobloch syndrome type 1	267750
COL1A2	chr7	7q22.1	Ehlers-Danlos syndrome cardiac valvular type	225320
COL25A1	chr4	4q25	Fibrosis of extraocular muscles congenital 5	616219
COL27A1	chr9	9q32	Steel syndrome	615155
COL3A1	chr2	2q32.2	Ehlers-Danlos syndrome vascular type	130050
COL3A1	chr2	2q32.2	Polymicrogyria with or without vascular-type EDS	618343
COL4A3	chr2	2q36-q37	Alport syndrome 2 autosomal recessive	203780
COL4A4	chr2	2q36-q37	Alport syndrome 2 autosomal recessive	203780
COL4A6	chrX	Xq22.3	Deafness X-linked 6	300914
COL6A1	chr21	21q22.3	Ullrich congenital muscular dystrophy 1	254090
COL6A1	chr21	21q22.3	Bethlem myopathy 1	158810
COL6A2	chr21	21q22.3	Bethlem myopathy 1	158810
COL6A2	chr21	21q22.3	Ullrich congenital muscular dystrophy 1	254090
COL6A2	chr21	21q22.3	Myosclerosis congenital	255600

COL6A3	chr2	2q37	Bethlem myopathy 1	158810
COL6A3	chr2	2q37	Dystonia 27	616411
COL6A3	chr2	2q37	Ullrich congenital muscular dystrophy 1	254090
COL7A1	chr3	3p21.3	EBD inversa	226600
COL7A1	chr3	3p21.3	Epidermolysis bullosa dystrophica AR	226600
COL7A1	chr3	3p21.3	Transient bullous of the newborn	131705
COL7A1	chr3	3p21.3	Epidermolysis bullosa pruriginosa	604129
COL7A1	chr3	3p21.3	Epidermolysis bullosa pretibial	131850
COL9A2	chr1	1p34.2	Stickler syndrome type V	614284
COLEC11	chr2	2p25.3	3MC syndrome 2	265050
COLQ	chr3	3p25	Myasthenic syndrome congenital 5	603034
COQ2	chr4	4q21-q22	Multiple system atrophy susceptibility to	146500
COQ2	chr4	4q21-q22	Coenzyme Q10 deficiency primary 1	607426
COQ4	chr9	9q34.13	Coenzyme Q10 deficiency primary 7	616276
COQ6	chr14	14q24.3	Coenzyme Q10 deficiency primary 6	614650
COQ8A	chr1	1q42.2	Coenzyme Q10 deficiency primary 4	612016
COQ8B	chr19	19q13.1	Nephrotic syndrome type 9	615573
COQ9	chr16	16q13	Coenzyme Q10 deficiency primary 5	614654
CORO1A	chr16	16p11.2	Immunodeficiency 8	615401
COX10	chr17	17p12-p11.2	Mitochondrial complex IV deficiency	220110
COX10	chr17	17p12-p11.2	Leigh syndrome due to mitochondrial COX4 deficiency	256000
COX14	chr12	12q13.12	Mitochondrial complex IV deficiency	220110
COX15	chr10	10q24	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 2	615119
COX15	chr10	10q24	Leigh syndrome due to cytochrome c oxidase deficiency	256000
COX20	chr1	1q44	Mitochondrial complex IV deficiency	220110
COX4I2	chr20	20q11.21	Exocrine pancreatic insufficiency dyserythropoietic anemia and calvarial hyperostosis	612714
COX6A1	chr12	12q24.2	Charcot-Marie-Tooth disease recessive intermediate D	616039
COX6B1	chr19	19q13.1	Mitochondrial complex IV deficiency	220110
CP	chr3	3q23-q24	Hypoceruloplasminemia hereditary	604290
CP	chr3	3q23-q24	Hemosiderosis systemic due to aceruloplasminemia	604290
CP	chr3	3q23-q24	Cerebellar ataxia	604290
CPA6	chr8	8q13	Febrile seizures familial 11	614418
CPA6	chr8	8q13	Epilepsy familial temporal lobe 5	614417
CPN1	chr10	10q24.2	Carboxypeptidase N deficiency	212070
CPOX	chr3	3q12	Harderoporphyria	618892
CPOX	chr3	3q12	Coproporphyria	121300
CPS1	chr2	2q35	Carbamoylphosphate synthetase I deficiency	237300
CPT1A	chr11	11q13	CPT deficiency hepatic type IA	255120
CPT2	chr1	1p32	CPT II deficiency myopathic stress-induced	255110
CPT2	chr1	1p32	CPT II deficiency infantile	600649
CPT2	chr1	1p32	CPT II deficiency lethal neonatal	608836
CPT2	chr1	1p32	Encephalopathy acute infection-induced 4 susceptibility to	614212

CR2	chr1	1q32	Immunodeficiency common variable 7	614699
CRADD	chr12	12q21.33-q23.1	Mental retardation autosomal recessive 34 with variant lissencephaly	614499
CRAT	chr9	9q34.1	Neurodegeneration with brain iron accumulation 8	617917
CRB1	chr1	1q31-q32.1	Retinitis pigmentosa-12	600105
CRB1	chr1	1q31-q32.1	Leber congenital amaurosis 8	613835
CRB2	chr9	9q33.3	Ventriculomegaly with cystic kidney disease	219730
CRB2	chr9	9q33.3	Focal segmental glomerulosclerosis 9	616220
CRBN	chr3	3p26.2	Mental retardation autosomal recessive 2	607417
CREB3L1	chr11	11p11.2	Osteogenesis imperfecta type XVI	616229
CRIPT	chr2	2p21	Short stature with microcephaly and distinctive facies	615789
CRLF1	chr19	19p12	Cold-induced sweating syndrome 1	272430
CRTAP	chr3	3p22	Osteogenesis imperfecta type VII	610682
CRYAA	chr21	21q22.3	Cataract 9 multiple types	604219
CRYAB	chr11	11q22.3-q23.1	Myopathy myofibrillar 2	608810
CRYAB	chr11	11q22.3-q23.1	Cardiomyopathy dilated 1II	615184
CRYAB	chr11	11q22.3-q23.1	Myopathy myofibrillar fatal infantile hypertonic alpha-B crystallin-related	613869
CRYAB	chr11	11q22.3-q23.1	Cataract 16 multiple types	613763
CRYBB1	chr22	22q11.2-q12.1	Cataract 17 multiple types	611544
CRYBB3	chr22	22q11.2-q12.2	Cataract 22	609741
CSF1R	chr5	5q32	Brain abnormalities neurodegeneration and dysosteosclerosis	618476
CSF2RB	chr22	22q12.2-q13.1	Surfactant metabolism dysfunction pulmonary 5	614370
CSF3R	chr1	1p35-p34.3	Neutropenia severe congenital 7 autosomal recessive	617014
CSGALNACT1	chr8	8p21.3	Skeletal dysplasia mild with joint laxity and advanced bone age	618870
CSPP1	chr8	8q13.2	Joubert syndrome 21	615636
CST6	chr11	11q13	Ectodermal dysplasia 15 hypohidrotic/hair type	618535
CSTA	chr3	3q21	Peeling skin syndrome 4	607936
CSTB	chr21	21q22.3	Epilepsy progressive myoclonic 1A (Unverricht and Lundborg)	254800
CTC1	chr17	17p13.1	Cerebroretinal microangiopathy with calcifications and cysts	612199
CTDP1	chr18	18q23	Congenital cataracts facial dysmorphism and neuropathy	604168
CTH	chr1	1p31.1	Cystathioninuria	219500
CTNNA2	chr2	2p12-p11.1	Cortical dysplasia complex with other brain malformations 9	618174
CTNS	chr17	17p13	Cystinosis nephropathic	219800
CTNS	chr17	17p13	Cystinosis ocular nonnephropathic	219750
CTNS	chr17	17p13	Cystinosis late-onset juvenile or adolescent nephropathic	219900
CTNS	chr17	17p13	Cystinosis atypical nephropathic	219800
CTPS1	chr1	1p34.1	Immunodeficiency 24	615897
CTSA	chr20	20q13.1	Galactosialidosis	256540
CTSC	chr11	11q14.1-q14.3	Periodontitis 1 juvenile	170650

CTSC	chr11	11q14.1-q14.3	Papillon-Lefevre syndrome	245000
CTSC	chr11	11q14.1-q14.3	Haim-Munk syndrome	245010
CTSD	chr11	11p15.5	Ceroid lipofuscinosis neuronal 10	610127
CTSF	chr11	11q13.1	Ceroid lipofuscinosis neuronal 13 Kufs type	615362
CTSK	chr1	1q21	Pycnodysostosis	265800
CUBN	chr10	10p12.1	Proteinuria chronic benign	618884
CUBN	chr10	10p12.1	Imerslund-Grasbeck syndrome 1	261100
CUL4B	chrX	Xq23	Mental retardation X-linked syndromic 15 (Cabezas type)	300354
CUL7	chr6	6p21.1	3-M syndrome 1	273750
CWF19L1	chr10	10q24.31	Spinocerebellar ataxia autosomal recessive 17	616127
CYB5A	chr18	18q23	Methemoglobinemia and ambiguous genitalia	250790
CYB5R3	chr22	22q13.31-qter	Methemoglobinemia type I	250800
CYB5R3	chr22	22q13.31-qter	Methemoglobinemia type II	250800
CYBA	chr16	16q24	Chronic granulomatous disease 4 autosomal recessive	233690
CYBB	chrX	Xp21.1	Immunodeficiency 34 mycobacteriosis X-linked	300645
CYBB	chrX	Xp21.1	Chronic granulomatous disease X-linked	306400
CYC1	chr8	8q24.3	Mitochondrial complex III deficiency nuclear type 6	615453
CYP11B1	chr8	8q21	Adrenal hyperplasia congenital due to 11-beta-hydroxylase deficiency	202010
CYP11B2	chr8	8q21	Hypoaldosteronism congenital due to CMO II deficiency	610600
CYP11B2	chr8	8q21	Hypoaldosteronism congenital due to CMO I deficiency	203400
CYP17A1	chr10	10q24.3	17-alpha-hydroxylase/1720-lyase deficiency	202110
CYP17A1	chr10	10q24.3	1720-lyase deficiency isolated	202110
CYP1B1	chr2	2p22-p21	Glaucoma 3A primary open angle congenital juvenile or adult onset	231300
CYP21A2	chr6	6p21.3	Adrenal hyperplasia congenital due to 21-hydroxylase deficiency	201910
CYP21A2	chr6	6p21.3	Hyperandrogenism nonclassic type due to 21-hydroxylase deficiency	201910
CYP24A1	chr20	20q13.2-q13.3	Hypercalcemia infantile 1	143880
CYP26C1	chr10	10q23.3	Focal facial dermal dysplasia 4	614974
CYP27A1	chr2	2q33-qter	Cerebrotendinous xanthomatosis	213700
CYP27B1	chr12	12q13.1-q13.3	Vitamin D-dependent rickets type I	264700
CYP2C19	chr10	10q24.1-q24.3	Clopidogrel impaired responsiveness to	609535
CYP2C19	chr10	10q24.1-q24.3	Mephenytoin poor metabolizer	609535
CYP2C19	chr10	10q24.1-q24.3	Proguanil poor metabolizer	609535
CYP2C19	chr10	10q24.1-q24.3	Omeprazole poor metabolizer	609535
CYP2D6	chr22	22q13.1	Debrisoquine sensitivity	608902
CYP2D6	chr22	22q13.1	Codeine sensitivity	608902
CYP2R1	chr11	11p15.2	Rickets due to defect in vitamin D 25-hydroxylation	600081
CYP2U1	chr4	4q25	Spastic paraplegia 56 autosomal recessive	615030
CYP4F22	chr19	19p13.12	Ichthyosis congenital autosomal recessive 5	604777
CYP4V2	chr4	4q35.1	Bietti crystalline corneoretinal dystrophy	210370
CYP7B1	chr8	8q21.3	Spastic paraplegia 5A autosomal recessive	270800

CYP7B1	chr8	8q21.3	Bile acid synthesis defect congenital 3	613812
D2HGDH	chr2	2q37.3	D-2-hydroxyglutaric aciduria	600721
DAG1	chr3	3p21	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 9	613818
DAG1	chr3	3p21	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 9	616538
DARS2	chr1	1q25.1	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation	611105
DBH	chr9	9q34	Orthostatic hypotension 1 due to DBH deficiency	223360
DBT	chr1	1p31	Maple syrup urine disease type II	248600
DCAF17	chr2	2q22.3-q35	Woodhouse-Sakati syndrome	241080
DCC	chr18	18q21.3	Gaze palsy familial horizontal with progressive scoliosis 2	617542
DCDC2	chr6	6p22.1	Sclerosing cholangitis neonatal	617394
DCDC2	chr6	6p22.1	Nephronophthisis 19	616217
DCDC2	chr6	6p22.1	Deafness autosomal recessive 66	610212
DCHS1	chr11	11p15.4	Van Maldergem syndrome 1	601390
DCLRE1C	chr10	10p	Omenn syndrome	603554
DCLRE1C	chr10	10p	Severe combined immunodeficiency Athabascan type	602450
DCPS	chr11	11q24.2	Al-Raqad syndrome	616459
DCTN1	chr2	2p13	Amyotrophic lateral sclerosis susceptibility to	105400
DCXR	chr17	17q25.3	Pentosuria	260800
DDB2	chr11	11p12-p11	Xeroderma pigmentosum group E DDB-negative subtype	278740
DDC	chr7	7p11	Aromatic L-amino acid decarboxylase deficiency	608643
DDHD1	chr14	14q22.1	Spastic paraplegia 28 autosomal recessive	609340
DDHD2	chr8	8p11.23	Spastic paraplegia 54 autosomal recessive	615033
DDOST	chr1	1p36.1	Congenital disorder of glycosylation type Ir	614507
DDR2	chr1	1q12-qter	Spondylometaepiphyseal dysplasia short limb-hand type	271665
DDRGK1	chr20	20p13	Spondyloepimetaphyseal dysplasia Shohat type	602557
DDX11	chr12	12p11	Warsaw breakage syndrome	613398
DDX3X	chrX	Xp11.3-p11.23	Intellectual developmental disorder X-linked syndrome Snijders Blok type	300958
DDX59	chr1	1q32.1	Orofaciodigital syndrome V	174300
DEAF1	chr11	11p15.5	Neurodevelopmental disorder with hypotonia impaired expressive language and with or without seizures	617171
DENND5A	chr11	11p15.4	Epileptic encephalopathy early infantile 49	617281
DES	chr2	2q35	Myopathy myofibrillar 1	601419
DGAT1	chr8	8q24.3	Diarrhea 7 protein-losing enteropathy type	615863
DGKE	chr17	17q22	Hemolytic uremic syndrome atypical susceptibility to 7	615008
DGKE	chr17	17q22	Nephrotic syndrome type 7	615008
DGUOK	chr2	2p13	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive 4	617070
DGUOK	chr2	2p13	Portal hypertension noncirrhotic	617068

DGUOK	chr2	2p13	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	251880
DHCR24	chr1	1p33-p31.1	Desmosterolosis	602398
DHCR7	chr11	11q12-q13	Smith-Lemli-Opitz syndrome	270400
DHDDS	chr1	1p36.11	Retinitis pigmentosa 59	613861
DHDDS	chr1	1p36.11	Congenital disorder of glycosylation type 1bb	613861
DHFR	chr5	5q11.2-q13.2	Megaloblastic anemia due to dihydrofolate reductase deficiency	613839
DHH	chr12	12q13.1	46XY sex reversal 7	233420
DHODH	chr16	16q22	Miller syndrome	263750
DHTKD1	chr10	10p14	2-aminoadipic 2-oxoadipic aciduria	204750
DHX37	chr12	12q24.31	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies	618731
DHX38	chr16	16q21-q22	Retinitis pigmentosa 84	618220
DIAPH1	chr5	5q31	Seizures cortical blindness microcephaly syndrome	616632
DIS3L2	chr2	2q37.2	Perlman syndrome	267000
DKC1	chrX	Xq28	Dyskeratosis congenita X-linked	305000
DLAT	chr11	11q23.1	Pyruvate dehydrogenase E2 deficiency	245348
DLD	chr7	7q31-q32	Dihydrolipoamide dehydrogenase deficiency	246900
DLG3	chrX	Xq13.1	Mental retardation X-linked 90	300850
DLL3	chr19	19q13	Spondylocostal dysostosis 1 autosomal recessive	277300
DLX5	chr7	7q22	Split-hand/foot malformation 1 with sensorineural hearing loss	220600
DMD	chrX	Xp21.2	Cardiomyopathy dilated 3B	302045
DMD	chrX	Xp21.2	Becker muscular dystrophy	300376
DMD	chrX	Xp21.2	Duchenne muscular dystrophy	310200
DMGDH	chr5	5q12.2-q12.3	Dimethylglycine dehydrogenase deficiency	605850
DMP1	chr4	4q21	Hypophosphatemic rickets AR	241520
DMXL2	chr15	15q21.2	Polyendocrine-polyneuropathy syndrome	616113
DMXL2	chr15	15q21.2	Epileptic encephalopathy early infantile 81	618663
DNA2	chr10	10q21.3-q22.1	Seckel syndrome 8	615807
DNAAF1	chr16	16q24.1	Ciliary dyskinesia primary 13	613193
DNAAF3	chr19	19q13.4	Ciliary dyskinesia primary 2	606763
DNAAF4	chr15	15q21	Ciliary dyskinesia primary 25	615482
DNAAF5	chr7	7p22.3	Ciliary dyskinesia primary 18	614874
DNAH1	chr3	3p21.3	Ciliary dyskinesia primary 37	617577
DNAH1	chr3	3p21.3	Spermatogenic failure 18	617576
DNAH11	chr7	7p21	Ciliary dyskinesia primary 7 with or without situs inversus	611884
DNAH9	chr17	17p12	Ciliary dyskinesia primary 40	618300
DNAI1	chr9	9p13.3	Ciliary dyskinesia primary 1 with or without situs inversus	244400
DNAJB2	chr2	2q35	Spinal muscular atrophy distal autosomal recessive 5	614881
DNAJC19	chr3	3q26.3	3-methylglutaconic aciduria type V	610198
DNAJC3	chr13	13q32	Ataxia combined cerebellar and peripheral with hearing loss and diabetes mellitus	616192
DNAJC6	chr1	1p32.1-p31.3	Parkinson disease 19b early-onset	615528

DNAJC6	chr1	1p32.1-p31.3	Parkinson disease 19a juvenile-onset	615528
DNAL1	chr14	14q24.3	Ciliary dyskinesia primary 16	614017
DNAL4	chr22	22q13.1	Mirror movements 3	616059
DNASE1L3	chr3	3p21.1-p14.3	Systemic lupus erythematosus 16	614420
DNM1L	chr12	12p11.21	Encephalopathy lethal due to defective mitochondrial peroxisomal fission 1	614388
DNM2	chr19	19p13.2	Lethal congenital contracture syndrome 5	615368
DNMT3B	chr20	20q11.2	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860
DOCK3	chr3	3p14	Neurodevelopmental disorder with impaired intellectual development hypotonia and ataxia	618292
DOCK6	chr19	19p13.2	Adams-Oliver syndrome 2	614219
DOCK7	chr1	1p31.3	Epileptic encephalopathy early infantile 23	615859
DOCK8	chr9	9p24	Hyper-IgE recurrent infection syndrome autosomal recessive	243700
DOK7	chr4	4p16.2	Fetal akinesia deformation sequence 3	618389
DOK7	chr4	4p16.2	Myasthenic syndrome congenital 10	254300
DOLK	chr9	9q34.11	Congenital disorder of glycosylation type Im	610768
DPAGT1	chr11	11q23.3	Congenital disorder of glycosylation type Ij	608093
DPAGT1	chr11	11q23.3	Myasthenic syndrome congenital 13 with tubular aggregates	614750
DPH1	chr17	17p13.3	Developmental delay with short stature dysmorphic facial features and sparse hair	616901
DPM1	chr20	20q13.13	Congenital disorder of glycosylation type Ie	608799
DPM2	chr9	9q34.11	Congenital disorder of glycosylation type Iu	615042
DPM3	chr1	1q12-q21	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 15	612937
DPY19L2	chr12	12q14.2	Spermatogenic failure 9	613958
DPYD	chr1	1p22	Dihydropyrimidine dehydrogenase deficiency	274270
DPYD	chr1	1p22	5-fluorouracil toxicity	274270
DPYS	chr8	8q22	Dihydropyrimidinuria	222748
DRC1	chr2	2p23.3	Ciliary dyskinesia primary 21	615294
DSC2	chr18	18q12.1	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair	610476
DSC2	chr18	18q12.1	Arrhythmogenic right ventricular dysplasia 11	610476
DSC3	chr18	18q12.1	Hypotrichosis and recurrent skin vesicles	613102
DSE	chr6	6q22	Ehlers-Danlos syndrome musculocontractural type 2	615539
DSG1	chr18	18q12.1-q12.2	Erythroderma congenital with palmoplantar keratoderma hypotrichosis and hyper IgE	615508
DSG4	chr18	18q12	Hypotrichosis 6	607903
DSP	chr6	6p24	Epidermolysis bullosa lethal acantholytic	609638
DSP	chr6	6p24	Cardiomyopathy dilated with woolly hair and keratoderma	605676
DSP	chr6	6p24	Skin fragility-woolly hair syndrome	607655
DST	chr6	6p12-p11	Neuropathy hereditary sensory and autonomic type VI	614653
DST	chr6	6p12-p11	Epidermolysis bullosa simplex autosomal recessive 2	615425
DSTYK	chr1	1q32.1	Spastic paraplegia 23	270750

DTNBP1	chr6	6p22.3	Hermansky-Pudlak syndrome 7	614076
DUOX2	chr15	15q15.3	Thyroid dyshormonogenesis 6	607200
DUOXA2	chr15	15q15	Thyroid dyshormonogenesis 5	274900
DYM	chr18	18q12-q21.1	Smith-McCort dysplasia	607326
DYM	chr18	18q12-q21.1	Dyggve-Melchior-Clausen disease	223800
DYNC2H1	chr11	11q22.3	Short-rib thoracic dysplasia 3 with or without polydactyly	613091
DYSF	chr2	2p13.3-p13.1	Miyoshi muscular dystrophy 1	254130
DYSF	chr2	2p13.3-p13.1	Muscular dystrophy limb-girdle autosomal recessive 2	253601
DYSF	chr2	2p13.3-p13.1	Myopathy distal with anterior tibial onset	606768
EARS2	chr16	16p13.1-p11.2	Combined oxidative phosphorylation deficiency 12	614924
EBP	chrX	Xp11.23-p11.22	MEND syndrome	300960
ECEL1	chr2	2q36-q37	Arthrogryposis distal type 5D	615065
ECHS1	chr10	10q26.2-q26.3	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency	616277
ECM1	chr1	1q21	Urbach-Wiethe disease	247100
EDA	chrX	Xq12-q13.1	Ectodermal dysplasia 1 hypohidrotic X-linked	305100
EDAR	chr2	2q11-q13	Ectodermal dysplasia 10B hypohidrotic/hair/tooth type autosomal recessive	224900
EDARADD	chr1	1q42.2-q43	Ectodermal dysplasia 11B hypohidrotic/hair/tooth type autosomal recessive	614941
EDN1	chr6	6p24-p23	Auriculocondylar syndrome 3	615706
EDN3	chr20	20q13.2-q13.3	Waardenburg syndrome type 4B	613265
EDNRB	chr13	13q22	Waardenburg syndrome type 4A	277580
EDNRB	chr13	13q22	ABCD syndrome	600501
EFEMP2	chr11	11q13	Cutis laxa autosomal recessive type IB	614437
EGFR	chr7	7p12.3-p12.1	Inflammatory skin and bowel disease neonatal 2	616069
EGR2	chr10	10q21.1-q22.1	Dejerine-Sottas disease	145900
EGR2	chr10	10q21.1-q22.1	Hypomyelinating neuropathy congenital 1	605253
EIF2AK3	chr2	2p12	Wolcott-Rallison syndrome	226980
EIF2AK4	chr15	15q15.1	Pulmonary venoocclusive disease 2	234810
EIF2B1	chr12	12q24.31	Leukoencephalopathy with vanishing white matter	603896
EIF2B2	chr14	14q24	Ovarioleukodystrophy	603896
EIF2B2	chr14	14q24	Leukoencephalopathy with vanishing white matter	603896
EIF2B3	chr1	1p34.1	Leukoencephalopathy with vanishing white matter	603896
EIF2B4	chr2	2p23.3	Leukoencephalopathy with vanishing white matter	603896
EIF2B4	chr2	2p23.3	Ovarioleukodystrophy	603896
EIF2B5	chr3	3q27	Leukoencephalopathy with vanishing white matter	603896
EIF2B5	chr3	3q27	Ovarioleukodystrophy	603896
EIF2S3	chrX	Xp22.2-p22.1	MEHMO syndrome	300148
EIF4A3	chr17	17q25.3	Robin sequence with cleft mandible and limb anomalies	268305
ELAC2	chr17	17p11	Combined oxidative phosphorylation deficiency 17	615440
ELMOD3	chr2	2p11.3	Deafness autosomal recessive 88	615429
ELOVL4	chr6	6q14	Ichthyosis spastic quadriplegia and mental retardation	614457
ELP1	chr9	9q31	Dysautonomia familial	223900
ELP2	chr18	18q12.2	Mental retardation autosomal recessive 58	617270

EMC1	chr1	1p36.13	Cerebellar atrophy visual impairment and psychomotor retardation	616875
EMD	chrX	Xq28	Emery-Dreifuss muscular dystrophy 1 X-linked	310300
EMG1	chr12	12p13	Bowen-Conradi syndrome	211180
EML1	chr14	14q32	Band heterotopia	600348
EMP2	chr16	16p13.2	Nephrotic syndrome type 10	615861
ENAM	chr4	4q21	Amelogenesis imperfecta type IC	204650
ENO3	chr17	17pter-p12	Glycogen storage disease XIII	612932
ENPP1	chr6	6q22-q23	Hypophosphatemic rickets autosomal recessive 2	613312
ENPP1	chr6	6q22-q23	Obesity susceptibility to	601665
ENPP1	chr6	6q22-q23	Arterial calcification generalized of infancy 1	208000
ENTPD1	chr10	10q24	Spastic paraplegia 64 autosomal recessive	615683
EOGT	chr3	3p14.1	Adams-Oliver syndrome 4	615297
EPB41	chr1	1p36.2-p34	Elliptocytosis-1	611804
EPCAM	chr2	2p21	Diarrhea 5 with tufting enteropathy congenital	613217
EPG5	chr18	18q12.3-q21.1	Vici syndrome	242840
EPHB2	chr1	1p36.1-p35	Bleeding disorder platelet-type 22	618462
EPHX1	chr1	1q42.1	Hypercholanemia familial	607748
EPM2A	chr6	6q24	Epilepsy progressive myoclonic 2A (Lafora)	254780
EPO	chr7	7q21	Diamond-Blackfan anemia-like	617911
EPS8	chr12	12p12.3	Deafness autosomal recessive 102	615974
EPX	chr17	17q23.1	Eosinophil peroxidase deficiency	261500
ERBB3	chr12	12q13	Lethal congenital contractural syndrome 2	607598
ERCC1	chr19	19q13.2-q13.3	Cerebrooculofacioskeletal syndrome 4	610758
ERCC2	chr19	19q13.2-q13.3	Trichothiodystrophy 1 photosensitive	601675
ERCC2	chr19	19q13.2-q13.3	Cerebrooculofacioskeletal syndrome 2	610756
ERCC2	chr19	19q13.2-q13.3	Xeroderma pigmentosum group D	278730
ERCC3	chr2	2q21	Xeroderma pigmentosum group B	610651
ERCC3	chr2	2q21	Trichothiodystrophy 2 photosensitive	616390
ERCC4	chr16	16p13.3-p13.13	Xeroderma pigmentosum type F/Cockayne syndrome	278760
ERCC4	chr16	16p13.3-p13.13	Fanconi anemia complementation group Q	615272
ERCC4	chr16	16p13.3-p13.13	XFE progeroid syndrome	610965
ERCC4	chr16	16p13.3-p13.13	Xeroderma pigmentosum group F	278760
ERCC5	chr13	13q33	Xeroderma pigmentosum group G/Cockayne syndrome	278780
ERCC5	chr13	13q33	Xeroderma pigmentosum group G	278780
ERCC5	chr13	13q33	Cerebrooculofacioskeletal syndrome 3	616570
ERCC6	chr10	10q11	Cerebrooculofacioskeletal syndrome 1	214150
ERCC6	chr10	10q11	Cockayne syndrome type B	133540
ERCC6	chr10	10q11	UV-sensitive syndrome 1	600630
ERCC6	chr10	10q11	De Sanctis-Cacchione syndrome	278800
ERCC6L2	chr9	9q22.32	Bone marrow failure syndrome 2	615715
ERCC8	chr5	5q12	Cockayne syndrome type A	216400
ERCC8	chr5	5q12	UV-sensitive syndrome 2	614621
ERLIN1	chr10	10q24.31	Spastic paraplegia 62	615681
ERLIN2	chr8	8p11.2	Spastic paraplegia 18 autosomal recessive	611225

ESCO2	chr8	8p21.1	Roberts syndrome	268300
ESCO2	chr8	8p21.1	SC phocomelia syndrome	269000
ESPN	chr1	1p36.3-p36.1	Usher syndrome type 1M	618632
ESPN	chr1	1p36.3-p36.1	Deafness autosomal recessive 36	609006
ESPN	chr1	1p36.3-p36.1	Deafness neurosensory without vestibular involvement autosomal dominant	609006
ESR1	chr6	6q25.1	Estrogen resistance	615363
ESRRB	chr14	14q24.3	Deafness autosomal recessive 35	608565
ETFA	chr15	15q23-q25	Glutaric acidemia IIA	231680
ETFB	chr19	19q13.3	Glutaric acidemia IIB	231680
ETFDH	chr4	4q32-qter	Glutaric acidemia IIC	231680
ETHE1	chr19	19q13.32	Ethylmalonic encephalopathy	602473
EVC	chr4	4p16	Ellis-van Creveld syndrome	225500
EVC2	chr4	4p16	Ellis-van Creveld syndrome	225500
EXOC6B	chr2	2p13.2	Spondyloepimetaphyseal dysplasia with joint laxity type 3	618395
EXOSC3	chr9	9p13.2	Pontocerebellar hypoplasia type 1B	614678
EXOSC8	chr13	13q13.1	Pontocerebellar hypoplasia type 1C	616081
EXPH5	chr11	11q22.3	Epidermolysis bullosa nonspecific autosomal recessive	615028
EXT2	chr11	11p12-p11	Seizures scoliosis and macrocephaly syndrome	616682
EXTL3	chr8	8p21	Immunoskeletal dysplasia with neurodevelopmental abnormalities	617425
EYS	chr6	6q12	Retinitis pigmentosa 25	602772
F10	chr13	13q34	Factor X deficiency	227600
F11	chr4	4q35	Factor XI deficiency autosomal recessive	612416
F12	chr5	5q33-qter	Factor XII deficiency	234000
F13A1	chr6	6p25-p24	Factor XIIIA deficiency	613225
F13B	chr1	1q31-q32.1	Factor XIIIB deficiency	613235
F2	chr11	11p11-q12	Hypoprothrombinemia	613679
F2	chr11	11p11-q12	Dysprothrombinemia	613679
F5	chr1	1q23	Factor V deficiency	227400
F5	chr1	1q23	Budd-Chiari syndrome	600880
F7	chr13	13q34	Factor VII deficiency	227500
F8	chrX	Xq28	Hemophilia A	306700
F9	chrX	Xq27.1-q27.2	Hemophilia B	306900
F9	chrX	Xq27.1-q27.2	Warfarin sensitivity	301052
FA2H	chr16	16q23	Spastic paraplegia 35 autosomal recessive	612319
FADD	chr11	11q13.3	Infections recurrent with encephalopathy hepatic dysfunction and cardiovascular malformations	613759
FAH	chr15	15q23-q25	Tyrosinemia type I	276700
FAM126A	chr7	7p15.3	Leukodystrophy hypomyelinating 5	610532
FAM20A	chr17	17q24.2	Amelogenesis imperfecta type IG (enamel-renal syndrome)	204690
FAM20C	chr7	7p22	Raine syndrome	259775
FAN1	chr15	15q13.2-q13.3	Interstitial nephritis karyomegalic	614817
FANCA	chr16	16q24.3	Fanconi anemia complementation group A	227650
FANCB	chrX	Xp22.31	Fanconi anemia complementation group B	300514
FANCC	chr9	9q22.3	Fanconi anemia complementation group C	227645

FANCD2	chr3	3p25.3	Fanconi anemia complementation group D2	227646
FANCE	chr6	6p22-p21	Fanconi anemia complementation group E	600901
FANCI	chr15	15q25-q26	Fanconi anemia complementation group I	609053
FANCL	chr2	2p16.1	Fanconi anemia complementation group L	614083
FANCM	chr14	14q21.3	Spermatogenic failure 28	618086
FANCM	chr14	14q21.3	Premature ovarian failure 15	618096
FAR1	chr11	11p15.2	Peroxisomal fatty acyl-CoA reductase 1 disorder	616154
FARS2	chr6	6p25.1	Spastic paraplegia 77 autosomal recessive	617046
FARS2	chr6	6p25.1	Combined oxidative phosphorylation deficiency 14	614946
FASTKD2	chr2	2q33.3	Combined oxidative phosphorylation deficiency 44	618855
FAT4	chr4	4q28.1	Van Maldergem syndrome 2	615546
FAT4	chr4	4q28.1	Hennekam lymphangiectasia-lymphedema syndrome 2	616006
FBLN5	chr14	14q32.1	Macular degeneration age-related 3	608895
FBLN5	chr14	14q32.1	Cutis laxa autosomal dominant 2	614434
FBLN5	chr14	14q32.1	Neuropathy hereditary with or without age-related macular degeneration	608895
FBLN5	chr14	14q32.1	Cutis laxa autosomal recessive type IA	219100
FBP1	chr9	9q22.2-q22.3	Fructose-16-bisphosphatase deficiency	229700
FBXL4	chr6	6q16.1-q16.3	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)	615471
FBXO31	chr16	16q24.3	Mental retardation autosomal recessive 45	615979
FBXO7	chr22	22q12-q13	Parkinson disease 15 autosomal recessive	260300
FCGR2A	chr1	1q21-q23	Pseudomonas aeruginosa susceptibility to chronic infection by in cystic fibrosis	219700
FCGR3A	chr1	1q23	Immunodeficiency 20	615707
FCN3	chr1	1p36.11	Immunodeficiency due to ficolin 3 deficiency	613860
FDFT1	chr8	8p23.1-p22	Squalene synthase deficiency	618156
FECH	chr18	18q21.3	Protoporphyria erythropoietic 1	177000
FERMT1	chr20	20p13	Kindler syndrome	173650
FERMT3	chr11	11q12	Leukocyte adhesion deficiency type III	612840
FEZF1	chr7	7q31.32	Hypogonadotropic hypogonadism 22 with or without anosmia	616030
FGA	chr4	4q28	Afibrinogenemia congenital	202400
FGB	chr4	4q28	Afibrinogenemia congenital	202400
FGB	chr4	4q28	Hypofibrinogenemia congenital	202400
FGD1	chrX	Xp11.21	Mental retardation X-linked syndromic 16	305400
FGD1	chrX	Xp11.21	Aarskog-Scott syndrome	305400
FGD4	chr12	12p11.2	Charcot-Marie-Tooth disease type 4H	609311
FGF16	chrX	Xq21.1	Metacarpal 4-5 fusion	309630
FGF20	chr8	8p22-p21.3	Renal hypodysplasia/aplasia 2	615721
FGF23	chr12	12p13.3	Tumoral calcinosis hyperphosphatemic familial 2	617993
FGF3	chr11	11q13	Deafness congenital with inner ear agenesis microtia and microdontia	610706
FGF5	chr4	4q21	Trichomegaly	190330
FGFR3	chr4	4p16.3	CATSHL syndrome	610474
FGG	chr4	4q28	Hypofibrinogenemia congenital	202400
FH	chr1	1q42.1	Fumarase deficiency	606812

FHL1	chrX	Xq27.2	Reducing body myopathy X-linked 1b with late childhood or adult onset	300718
FHL1	chrX	Xq27.2	Uruguay faciocardiomusculoskeletal syndrome	300280
FHL1	chrX	Xq27.2	Emery-Dreifuss muscular dystrophy 6 X-linked	300696
FHL1	chrX	Xq27.2	Myopathy X-linked with postural muscle atrophy	300696
FIG4	chr6	6q21	Yunis-Varon syndrome	216340
FIG4	chr6	6q21	Polymicrogyria bilateral temporooccipital	612691
FIG4	chr6	6q21	Charcot-Marie-Tooth disease type 4J	611228
FKBP10	chr17	17q21.2	Bruck syndrome 1	259450
FKBP10	chr17	17q21.2	Osteogenesis imperfecta type XI	610968
FKBP14	chr7	7p15.1	Ehlers-Danlos syndrome kyphoscoliotic type 2	614557
FKRP	chr19	19q13.3	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 5	607155
FKRP	chr19	19q13.3	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation) type B 5	606612
FKRP	chr19	19q13.3	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 5	613153
FKTN	chr9	9q31	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 4	611588
FKTN	chr9	9q31	Cardiomyopathy dilated 1X	611615
FKTN	chr9	9q31	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation) type B 4	613152
FKTN	chr9	9q31	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 4	253800
FLG	chr1	1q21	Ichthyosis vulgaris	146700
FLI1	chr11	11q24	Bleeding disorder platelet-type 21	617443
FLNA	chrX	Xq28	Congenital short bowel syndrome	300048
FLNA	chrX	Xq28	Intestinal pseudoobstruction neuronal	300048
FLNA	chrX	Xq28	Cardiac valvular dysplasia X-linked	314400
FLNA	chrX	Xq28	FG syndrome 2	300321
FLNA	chrX	Xq28	Frontometaphyseal dysplasia 1	305620
FLNB	chr3	3p14.3	Spondylocarpotarsal synostosis syndrome	272460
FLVCR1	chr1	1q31.3	Ataxia posterior column with retinitis pigmentosa	609033
FLVCR2	chr14	14q24.3	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome	225790
FMN2	chr1	1q43	Mental retardation autosomal recessive 47	616193
FMO3	chr1	1q23-q25	Trimethylaminuria	602079
FOLR1	chr11	11q13.3-q13.5	Neurodegeneration due to cerebral folate transport deficiency	613068
FOXE1	chr9	9q22	Bamforth-Lazarus syndrome	241850
FOXE3	chr1	1p32	Anterior segment dysgenesis 2 multiple subtypes	610256
FOXI1	chr5	5q34	Enlarged vestibular aqueduct	600791
FOXL2	chr3	3q23	Blepharophimosis epicanthus inversus and ptosis type 2	110100
FOXL2	chr3	3q23	Blepharophimosis epicanthus inversus and ptosis type 1	110100
FOXN1	chr17	17q11-q12	T-cell immunodeficiency congenital alopecia and nail dystrophy	601705

FOXP3	chrX	Xp11.23-q13.3	Immunodysregulation polyendocrinopathy and enteropathy X-linked	304790
FOXRED1	chr11	11q24.2	Mitochondrial complex I deficiency nuclear type 19	618241
FRAS1	chr4	4q21	Fraser syndrome 1	219000
FREM1	chr9	9p22.3	Manitoba oculotrichoanal syndrome	248450
FREM2	chr13	13q13.3	Fraser syndrome 2	617666
FREM2	chr13	13q13.3	Cryptophthalmos unilateral or bilateral isolated	123570
FRRS1L	chr9	9q31.3	Epileptic encephalopathy early infantile 37	616981
FSHB	chr11	11p13	Hypogonadotropic hypogonadism 24 without anosmia	229070
FSHR	chr2	2p21-p16	Ovarian dysgenesis 1	233300
FSHR	chr2	2p21-p16	Ovarian response to FSH stimulation	276400
FTCD	chr21	21q22.3	Glutamate formiminotransferase deficiency	229100
FTL	chr19	19q13.3-q13.4	Hyperferritinemia-cataract syndrome	600886
FTL	chr19	19q13.3-q13.4	Neurodegeneration with brain iron accumulation 3	606159
FTL	chr19	19q13.3-q13.4	L-ferritin deficiency dominant and recessive	615604
FTO	chr16	16q12.2	Growth retardation developmental delay facial dysmorphism	612938
FTO	chr16	16q12.2	Obesity susceptibility to BMIQ14	612460
FTSJ1	chrX	Xp11.23	Mental retardation X-linked 9/44	309549
FUCA1	chr1	1p34	Fucosidosis	230000
FUT1	chr19	19q13.3	Bombay phenotype	616754
FUT2	chr19	19q13.3	Bombay phenotype digenic	616754
FUT8	chr14	14q23	Congenital disorder of glycosylation with defective fucosylation 1	618005
FXN	chr9	9q13	Friedreich ataxia with retained reflexes	229300
FXN	chr9	9q13	Friedreich ataxia	229300
FYCO1	chr3	3p21.3	Cataract 18 autosomal recessive	610019
FZD6	chr8	8q22.3-q23.1	Nail disorder nonsyndromic congenital 1	161050
G6PC	chr17	17q21	Glycogen storage disease Ia	232200
G6PC3	chr17	17q21	Dursun syndrome	612541
G6PC3	chr17	17q21	Neutropenia severe congenital 4 autosomal recessive	612541
GAA	chr17	17q25.2-q25.3	Glycogen storage disease II	232300
GAD1	chr2	2q31	Cerebral palsy spastic quadriplegic 1	603513
GALC	chr14	14q31	Krabbe disease	245200
GALE	chr1	1p36-p35	Galactose epimerase deficiency	230350
GALK1	chr17	17q24	Galactokinase deficiency with cataracts	230200
GALNS	chr16	16q24.3	Mucopolysaccharidosis IVA	253000
GALNT2	chr1	1q41-q42	Congenital disorder of glycosylation type IIt	618885
GALNT3	chr2	2q24-q31	Tumoral calcinosis hyperphosphatemic familial 1	211900
GALT	chr9	9p13	Galactosemia	230400
GAMT	chr19	19p13.3	Cerebral creatine deficiency syndrome 2	612736
GAN	chr16	16q24.1	Giant axonal neuropathy-1	256850
GAS2L2	chr17	17q12	Ciliary dyskinesia primary 41	618449

GATA1	chrX	Xp11.23	Anemia X-linked with/without neutropenia and/or platelet abnormalities	300835
GATA1	chrX	Xp11.23	Thrombocytopenia X-linked with or without dyserythropoietic anemia	300367
GATA1	chrX	Xp11.23	Thrombocytopenia with beta-thalassemia X-linked	314050
GATA5	chr20	20q13.2-q13.3	Congenital heart defects multiple types 5	617912
GATAD1	chr7	7q21.2	Cardiomyopathy dilated 2B	614672
GATM	chr15	15q21.1	Cerebral creatine deficiency syndrome 3	612718
GBA	chr1	1q21	Gaucher disease type III	231000
GBA	chr1	1q21	Gaucher disease type IIIC	231005
GBA	chr1	1q21	Gaucher disease type I	230800
GBA	chr1	1q21	Gaucher disease perinatal lethal	608013
GBA	chr1	1q21	Gaucher disease type II	230900
GBA2	chr9	9p13.3	Spastic paraplegia 46 autosomal recessive	614409
GBE1	chr3	3p12	Polyglucosan body disease adult form	263570
GBE1	chr3	3p12	Glycogen storage disease IV	232500
GCDH	chr19	19p13.2	Glutaricaciduria type I	231670
GCH1	chr14	14q22.1-q22.2	Hyperphenylalaninemia BH4-deficient B	233910
GCH1	chr14	14q22.1-q22.2	Dystonia DOPA-responsive with or without hyperphenylalaninemia	128230
GCK	chr7	7p15-p13	Diabetes mellitus permanent neonatal 1	606176
GCLC	chr6	6p12	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency	230450
GCNT2	chr6	6p24-p23	Cataract 13 with adult i phenotype	116700
GCSH	chr16	16q24	Glycine encephalopathy	605899
GDAP1	chr8	8q21.11	Charcot-Marie-Tooth disease recessive intermediate A	608340
GDAP1	chr8	8q21.11	Charcot-Marie-Tooth disease type 4A	214400
GDAP1	chr8	8q21.11	Charcot-Marie-Tooth disease axonal type 2K	607831
GDAP1	chr8	8q21.11	Charcot-Marie-Tooth disease axonal with vocal cord paresis	607706
GDF1	chr19	19p12	Right atrial isomerism (Ivemark)	208530
GDF5	chr20	20q11.2	Acromesomelic dysplasia Hunter-Thompson type	201250
GDF5	chr20	20q11.2	Brachydactyly type A1 C	615072
GDF5	chr20	20q11.2	Chondrodysplasia Grebe type	200700
GDF5	chr20	20q11.2	Du Pan syndrome	228900
GDF6	chr8	8q22.1	Leber congenital amaurosis 17	615360
GDF9	chr5	5q31.1	Premature ovarian failure 14	618014
GEMIN4	chr17	17p13.3	Neurodevelopmental disorder with microcephaly cataracts and renal abnormalities	617913
GFI1B	chr9	9q34.13	Bleeding disorder platelet-type 17	187900
GFM1	chr3	3q25.32	Combined oxidative phosphorylation deficiency 1	609060
GFM2	chr5	5q13	Combined oxidative phosphorylation deficiency 39	618397
GFPT1	chr2	2p13	Myasthenia congenital 12 with tubular aggregates	610542
GGCX	chr2	2p12	Vitamin K-dependent clotting factors combined deficiency of 1	277450
GH1	chr17	17q22-q24	Kowarski syndrome	262650

GH1	chr17	17q22-q24	Growth hormone deficiency isolated type IA	262400
GHR	chr5	5p13-p12	Laron dwarfism	262500
GHRHR	chr7	7p15-p14	Growth hormone deficiency isolated type IV	618157
GHRL	chr3	3p26-p25	Obesity susceptibility to	601665
GHSR	chr3	3q26.3	Growth hormone deficiency isolated partial	615925
GIPC3	chr19	19p13.3	Deafness autosomal recessive 15	601869
GJA1	chr6	5q22.31	Craniometaphyseal dysplasia autosomal recessive	218400
GJA1	chr6	5q22.31	Oculodentodigital dysplasia autosomal recessive	257850
GJA1	chr6	5q22.31	Hypoplastic left heart syndrome 1	241550
GJB2	chr13	13q11-q12	Deafness autosomal recessive 1A	220290
GJB3	chr1	1p35.1	Deafness autosomal recessive	
GJB3	chr1	1p35.1	Deafness digenic GJB2/GJB3	220290
GJB3	chr1	1p35.1	Erythrokeratodermia variabilis et progressiva 1	133200
GJB6	chr13	13q12	Deafness autosomal recessive 1B	612645
GJB6	chr13	13q12	Deafness digenic GJB2/GJB6	220290
GJC2	chr1	1q42.13	Spastic paraplegia 44 autosomal recessive	613206
GJC2	chr1	1q42.13	Leukodystrophy hypomyelinating 2	608804
GK	chrX	Xp21.3-p21.2	Glycerol kinase deficiency	307030
GLB1	chr3	3p21.33	GM1-gangliosidosis type III	230650
GLB1	chr3	3p21.33	GM1-gangliosidosis type I	230500
GLB1	chr3	3p21.33	Mucopolysaccharidosis type IVB (Morquio)	253010
GLB1	chr3	3p21.33	GM1-gangliosidosis type II	230600
GLDC	chr9	9p22	Glycine encephalopathy	605899
GLE1	chr9	9q34	Congenital arthrogryposis with anterior horn cell disease	611890
GLE1	chr9	9q34	Lethal congenital contracture syndrome 1	253310
GLI1	chr12	12q13.2-q13.3	Polydactyly postaxial type A8	618123
GLI1	chr12	12q13.2-q13.3	Polydactyly preaxial I	174400
GLIS3	chr9	9p24.3-p23	Diabetes mellitus neonatal with congenital hypothyroidism	610199
GLRA1	chr5	5q32	Hyperekplexia 1	149400
GLRB	chr4	4q31.3	Hyperekplexia 2	614619
GLRX5	chr14	14q32	Anemia sideroblastic 3 pyridoxine-refractory	616860
GLRX5	chr14	14q32	Spasticity childhood-onset with hyperglycinemia	616859
GLS	chr2	2q32-q34	Global developmental delay progressive ataxia and elevated glutamine	618412
GLS	chr2	2q32-q34	Epileptic encephalopathy early infantile 71	618328
GLUL	chr1	1q31	Glutamine deficiency congenital	610015
GLYCTK	chr3	3p21	D-glyceric aciduria	220120
GM2A	chr5	5q31.3-q33.1	GM2-gangliosidosis AB variant	272750
GMPPA	chr2	2q35	Alacrima achalasia and mental retardation syndrome	615510
GMPPB	chr3	3p21.31	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 14	615350
GMPPB	chr3	3p21.31	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 14	615351

GMPPB	chr3	3p21.31	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 14	615352
GNAT1	chr3	3p21	Night blindness congenital stationary type 1G	616389
GNB3	chr12	12p13	Night blindness congenital stationary type 1H	617024
GNB5	chr15	15q21.2	Intellectual developmental disorder with cardiac arrhythmia	617173
GNB5	chr15	15q21.2	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia	617182
GNE	chr9	9p13.3	Nonaka myopathy	605820
GNMT	chr6	6p12	Glycine N-methyltransferase deficiency	606664
GNPAT	chr1	1q42	Rhizomelic chondrodysplasia punctata type 2	222765
GNPTAB	chr12	12q23.3	Mucolipidosis II alpha/beta	252500
GNPTAB	chr12	12q23.3	Mucolipidosis III alpha/beta	252600
GNPTG	chr16	16p	Mucolipidosis III gamma	252605
GNRH1	chr8	8p21-p11.2	Hypogonadotropic hypogonadism 12 with or without anosmia	614841
GNRHR	chr4	4q21.2	Hypogonadotropic hypogonadism 7 without anosmia	146110
GNS	chr12	12q14	Mucopolysaccharidosis type IIID	252940
GORAB	chr1	1q24.2	Geroderma osteodysplasticum	231070
GOSR2	chr17	17q21	Epilepsy progressive myoclonic 6	614018
GP1BA	chr17	17pter-p12	Bernard-Soulier syndrome type A1 (recessive)	231200
GP1BA	chr17	17pter-p12	Nonarteritic anterior ischemic optic neuropathy susceptibility to	258660
GP1BB	chr22	22q11.2	Giant platelet disorder isolated	231200
GP1BB	chr22	22q11.2	Bernard-Soulier syndrome type B	231200
GP6	chr19	19q13.4	Bleeding disorder platelet-type 11	614201
GP9	chr3	3q21	Bernard-Soulier syndrome type C	231200
GPC3	chrX	Xq26	Simpson-Golabi-Behmel syndrome type 1	312870
GPC4	chrX	Xq26	Keipert syndrome	301026
GPC6	chr13	13q32	Omodysplasia 1	258315
GPD1	chr12	12q12-q13	Hypertriglyceridemia transient infantile	614480
GPHN	chr14	14q24	Molybdenum cofactor deficiency C	615501
GPI	chr19	19q13.1	Hemolytic anemia nonspherocytic due to glucose phosphate isomerase deficiency	613470
GPIHBP1	chr8	8q24.3	Hyperlipoproteinemia type 1D	615947
GPNMB	chr7	7p15.3	Amyloidosis primary localized cutaneous 3	617920
GPR143	chrX	Xp22.3	Ocular albinism type I Nettleship-Falls type	300500
GPR143	chrX	Xp22.3	Nystagmus 6 congenital X-linked	300814
GPR179	chr17	17q12	Night blindness congenital stationary (complete) 1E autosomal recessive	614565
GPR68	chr14	14q31	Amelogenesis imperfecta hypomaturation type IIA6	617217
GPSM2	chr1	1p13.1	Chudley-McCullough syndrome	604213
GPT2	chr16	16q12.1	Mental retardation autosomal recessive 49	616281
GPX1	chr3	3p21.3	Hemolytic anemia due to glutathione peroxidase deficiency	614164
GPX4	chr19	19p13.3	Spondylometaphyseal dysplasia Sedaghatian type	250220
GRHL2	chr8	8q22	Ectodermal dysplasia/short stature syndrome	616029
GRHPR	chr9	9cen	Hyperoxaluria primary type II	260000

GRIA3	chrX	Xq25-q26	Mental retardation X-linked 94	300699
GRID2	chr4	4q22	Spinocerebellar ataxia autosomal recessive 18	616204
GRIK2	chr6	6q21	Mental retardation autosomal recessive 6	611092
GRIN1	chr9	9q34.3	Neurodevelopmental disorder with or without hyperkinetic movements and seizures autosomal recessive	617820
GRIP1	chr12	12q14.3	Fraser syndrome 3	617667
GRM1	chr6	6q24	Spinocerebellar ataxia autosomal recessive 13	614831
GRM6	chr5	5q35	Night blindness congenital stationary (complete) 1B autosomal recessive	257270
GRM7	chr3	3p26.1	Neurodevelopmental disorder with seizures hypotonia and brain imaging abnormalities	618922
GRN	chr17	17q21.32	Ceroid lipofuscinosis neuronal 11	614706
GRXCR1	chr4	4p13	Deafness autosomal recessive 25	613285
GRXCR2	chr5	5q32	Deafness autosomal recessive 101	615837
GSC	chr14	14q32.1	Short stature auditory canal atresia mandibular hypoplasia skeletal abnormalities	602471
GSR	chr8	8p21.1	Hemolytic anemia due to glutathione reductase deficiency	618660
GSS	chr20	20q11.2	Glutathione synthetase deficiency	266130
GSS	chr20	20q11.2	Hemolytic anemia due to glutathione synthetase deficiency	231900
GSTZ1	chr14	14q24.3	Maleylacetoacetate isomerase deficiency	617596
GTPBP3	chr19	19p13.11	Combined oxidative phosphorylation deficiency 23	616198
GUCY2C	chr12	12p12	Meconium ileus	614665
GUCY2D	chr17	17p13.1	Cone-rod dystrophy 6	601777
GUCY2D	chr17	17p13.1	Leber congenital amaurosis 1	204000
GUCY2D	chr17	17p13.1	Night blindness congenital stationary type 1I	618555
GUSB	chr7	7q21.11	Mucopolysaccharidosis VII	253220
GYG1	chr3	3q24-q25.1	Glycogen storage disease XV	613507
GYG1	chr3	3q24-q25.1	Polyglucosan body myopathy 2	616199
GYS1	chr19	19q13.3	Glycogen storage disease 0 muscle	611556
GYS2	chr12	12p12.2	Glycogen storage disease 0 liver	240600
H6PD	chr1	1p36	Cortisone reductase deficiency 1	604931
HACE1	chr6	6q21	Spastic paraplegia and psychomotor retardation with or without seizures	616756
HADH	chr4	4q22-q26	3-hydroxyacyl-CoA dehydrogenase deficiency	231530
HADH	chr4	4q22-q26	Hyperinsulinemic hypoglycemia familial 4	609975
HADHA	chr2	2p23	LCHAD deficiency	609016
HADHA	chr2	2p23	HELLP syndrome maternal of pregnancy	609016
HADHA	chr2	2p23	Mitochondrial trifunctional protein deficiency	609015
HADHA	chr2	2p23	Fatty liver acute of pregnancy	609016
HADHB	chr2	2p23	Trifunctional protein deficiency	609015
HAL	chr12	12q22-q23	Histidinemia	235800
HAMP	chr19	19q13	Hemochromatosis type 2B	613313
HARS2	chr5	5q31.3	Perrault syndrome 2	614926
HAX1	chr1	1q21.3	Neutropenia severe congenital 3 autosomal recessive	610738
HBB	chr11	11p15.5	Thalassemia beta	613985

HBB	chr11	11p15.5	Sickle cell anemia	603903
HCFC1	chrX	Xq28	Mental retardation X-linked 3 (methylmalonic acidemia and homocysteinemia cblX type)	309541
HEPACAM	chr11	11q24	Megalencephalic leukoencephalopathy with subcortical cysts 2A	613925
HERC1	chr15	15q22.31	Macrocephaly dysmorphic facies and psychomotor retardation	617011
HERC2	chr15	15q13.1	Skin/hair/eye pigmentation 1 blue/nonblue eyes	227220
HERC2	chr15	15q13.1	Mental retardation autosomal recessive 38	615516
HERC2	chr15	15q13.1	Skin/hair/eye pigmentation 1 blond/brown hair	227220
HES7	chr17	17p13.2	Spondylocostal dysostosis 4 autosomal recessive	613686
HESX1	chr3	3p21.2-p21.1	Pituitary hormone deficiency combined 5	182230
HESX1	chr3	3p21.2-p21.1	Septooptic dysplasia	182230
HESX1	chr3	3p21.2-p21.1	Growth hormone deficiency with pituitary anomalies	182230
HEXA	chr15	15q23-q24	GM2-gangliosidosis several forms	272800
HEXA	chr15	15q23-q24	Tay-Sachs disease	272800
HEXA	chr15	15q23-q24	Hex A pseudodeficiency	272800
HEXB	chr5	5q13	Sandhoff disease infantile juvenile and adult forms	268800
HFE	chr6	6p21.3	Porphyria cutanea tarda susceptibility to	176100
HFE	chr6	6p21.3	Hemochromatosis	235200
HFM1	chr1	1p22.2	Premature ovarian failure 9	615724
HGD	chr3	3q13.33	Alkaptonuria	203500
HGF	chr7	7q21.1	Deafness autosomal recessive 39	608265
HGSNAT	chr8	8p11.1	Retinitis pigmentosa 73	616544
HGSNAT	chr8	8p11.1	Mucopolysaccharidosis type IIIC (Sanfilippo C)	252930
HIBCH	chr2	2q32.2	3-hydroxyisobutryl-CoA hydrolase deficiency	250620
HINT1	chr5	5q31.2	Neuromyotonia and axonal neuropathy autosomal recessive	137200
HK1	chr10	10q22	Hemolytic anemia due to hexokinase deficiency	235700
HK1	chr10	10q22	Neuropathy hereditary motor and sensory Russe type	605285
HK1	chr10	10q22	Neurodevelopmental disorder with visual defects and brain anomalies	618547
HK1	chr10	10q22	Retinitis pigmentosa 79	617460
HLA-DQA1	chr6	6p21.3	Celiac disease susceptibility to	212750
HLA-DQB1	chr6	6p21.3	Celiac disease susceptibility to	212750
HLCS	chr21	21q22.1	Holocarboxylase synthetase deficiency	253270
HMGCL	chr1	1pter-p33	HMG-CoA lyase deficiency	246450
HMGCS2	chr1	1p13-p12	HMG-CoA synthase-2 deficiency	605911
HMX1	chr4	4p16.1	Oculoauricular syndrome	612109
HNF1A	chr12	12q24.2	Diabetes mellitus insulin-dependent	222100
HNMT	chr2	2q22	Mental retardation autosomal recessive 51	616739
HOGA1	chr10	10q24.2	Hyperoxaluria primary type III	613616
HOXA2	chr7	7p15-p14	Microtia with or without hearing impairment (AD)	612290
HOXA2	chr7	7p15-p14	Microtia hearing impairment and cleft palate (AR)	612290
HOXB1	chr17	17q21-q22	Facial paresis hereditary congenital 3	614744
HOXC13	chr12	12q13	Ectodermal dysplasia 9 hair/nail type	614931
HPCA	chr1	1p35-p34.2	Dystonia 2 torsion autosomal recessive	224500

HPD	chr12	12q24-qter	Tyrosinemia type III	276710
HPGD	chr4	4q34-q35	Hypertrophic osteoarthropathy primary autosomal recessive 1	259100
HPGD	chr4	4q34-q35	Digital clubbing isolated congenital	119900
HPGD	chr4	4q34-q35	Cranioosteoarthropathy	259100
HPRT1	chrX	Xq26.2	HPRT-related gout	300323
HPRT1	chrX	Xq26.2	Lesch-Nyhan syndrome	300322
HPS1	chr10	10q23.1	Hermansky-Pudlak syndrome 1	203300
HPS3	chr3	3q24	Hermansky-Pudlak syndrome 3	614072
HPS4	chr22	22q11.2-q12.2	Hermansky-Pudlak syndrome 4	614073
HPS5	chr11	11p15-p13	Hermansky-Pudlak syndrome 5	614074
HPS6	chr10	10q24.32	Hermansky-Pudlak syndrome 6	614075
HPSE2	chr10	10q23-q24	Urofacial syndrome 1	236730
HR	chr8	8p21.2	Alopecia universalis	203655
HR	chr8	8p21.2	Atrichia with papular lesions	209500
HSD11B2	chr16	16q22	Apparent mineralocorticoid excess	218030
HSD17B3	chr9	9q22	Pseudohermaphroditism male with gynecomastia	264300
HSD17B4	chr5	5q23.1	D-bifunctional protein deficiency	261515
HSD17B4	chr5	5q23.1	Perrault syndrome 1	233400
HSD3B2	chr1	1p13.1	Adrenal hyperplasia congenital due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	201810
HSD3B7	chr16	16p12-p11.2	Bile acid synthesis defect congenital 1	607765
HSPA9	chr5	5q31.2	Even-plus syndrome	616854
HSPD1	chr2	2q33.1	Leukodystrophy hypomyelinating 4	612233
HSPG2	chr1	1p36.1	Dyssegmental dysplasia Silverman-Handmaker type	224410
HTRA1	chr10	10q25.3-q26.2	CARASIL syndrome	600142
HTRA2	chr2	2p12	3-methylglutaconic aciduria type VIII	617248
HTT	chr4	4p16.3	Lopes-Maciel-Rodan syndrome	617435
HYAL1	chr3	3p21.3-p21.2	Mucopolysaccharidosis type IX	601492
HYDIN	chr16	16q22.2	Ciliary dyskinesia primary 5	608647
HYLS1	chr11	11q24.2	Hydrolethalus syndrome	236680
IARS2	chr1	1q41	Cataracts growth hormone deficiency sensory neuropathy sensorineural hearing loss and skeletal dysplasia	616007
IBA57	chr1	1q42.13	Spastic paraplegia 74 autosomal recessive	616451
IBA57	chr1	1q42.13	Multiple mitochondrial dysfunctions syndrome 3	615330
ICOS	chr2	2q33	Immunodeficiency common variable 1	607594
IDS	chrX	Xq28	Mucopolysaccharidosis II	309900
IDUA	chr4	4p16.3	Mucopolysaccharidosis Ih/s	607015
IDUA	chr4	4p16.3	Mucopolysaccharidosis Ih	607014
IDUA	chr4	4p16.3	Mucopolysaccharidosis Is	607016
IER3IP1	chr18	18q21.1	Microcephaly epilepsy and diabetes syndrome	614231
IFNAR2	chr21	21q22.1	Immunodeficiency 45	616669
IFNG	chr12	12q14	Immunodeficiency 69 mycobacteriosis	618963
IFNGR1	chr6	6q23.3	Immunodeficiency 27A mycobacteriosis AR	209950

IFNGR2	chr21	21q22.1-q22.2	Immunodeficiency 28 mycobacteriosis	614889
IFT122	chr3	3q21	Cranioectodermal dysplasia 1	218330
IFT140	chr16	16p13.3	Retinitis pigmentosa 80	617781
IFT140	chr16	16p13.3	Short-rib thoracic dysplasia 9 with or without polydactyly	266920
IFT172	chr2	2p23.3	Retinitis pigmentosa 71	616394
IFT172	chr2	2p23.3	Short-rib thoracic dysplasia 10 with or without polydactyly	615630
IFT27	chr22	22q12.3	Bardet-Biedl syndrome 19	615996
IFT43	chr14	14q24.3	Cranioectodermal dysplasia 3	614099
IFT43	chr14	14q24.3	Short-rib thoracic dysplasia 18 with polydactyly	617866
IFT43	chr14	14q24.3	Retinitis pigmentosa 81	617871
IFT80	chr3	3q25.33	Short-rib thoracic dysplasia 2 with or without polydactyly	611263
IGBP1	chrX	Xq13.1-q13.3	Corpus callosum agenesis of with mental retardation ocular coloboma and micrognathia	300472
IGF1	chr12	12q22-q24.1	Growth retardation with deafness and mental retardation due to IGF1 deficiency	608747
IGF1R	chr15	15q25-q26	Insulin-like growth factor I resistance to	270450
IGFBP7	chr4	4q12	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis	614224
IGHM	chr14	14q32.33	Agammaglobulinemia 1	601495
IGHMBP2	chr11	11q13.2-q13.4	Neuronopathy distal hereditary motor type VI	604320
IGHMBP2	chr11	11q13.2-q13.4	Charcot-Marie-Tooth disease axonal type 2S	616155
IGLL1	chr22	22q11.21	Agammaglobulinemia 2	613500
IGSF1	chrX	Xq25	Hypothyroidism central and testicular enlargement	300888
IGSF3	chr1	1p13	Lacrimal duct defect	149700
IHH	chr2	2q33-q35	Acrocapitofemoral dysplasia	607778
IKBKB	chr8	8p11.2	Immunodeficiency 15B	615592
IKBKG	chrX	Xq28	Immunodeficiency 33	300636
IKBKG	chrX	Xq28	Ectodermal dysplasia and immunodeficiency 1	300291
IL10RA	chr11	11q23.3	Inflammatory bowel disease 28 early onset autosomal recessive	613148
IL10RB	chr21	21q22.1	Inflammatory bowel disease 25 early onset autosomal recessive	612567
IL11RA	chr9	9p13	Craniosynostosis and dental anomalies	614188
IL12B	chr5	5q33.3	Immunodeficiency 29 mycobacteriosis	614890
IL12RB1	chr19	19p13.1	Immunodeficiency 30	614891
IL17RA	chr22	22q11.22-q11.23	Immunodeficiency 51	613953
IL17RD	chr3	3p14.3-p14.2	Hypogonadotropic hypogonadism 18 with or without anosmia	615267
IL1RAPL1	chrX	Xp22.1-p21.3	Mental retardation X-linked 21/34	300143
IL1RN	chr2	2q14.2	Interleukin 1 receptor antagonist deficiency	612852
IL21	chr4	4q26-q27	Immunodeficiency common variable 11	615767
IL21R	chr16	16p11	Immunodeficiency 56	615207

IL2RA	chr10	10p15.1	Immunodeficiency 41 with lymphoproliferation and autoimmunity	606367
IL2RG	chrX	Xq13	Severe combined immunodeficiency X-linked	300400
IL2RG	chrX	Xq13	Combined immunodeficiency X-linked moderate	312863
IL36RN	chr2	2q13	Psoriasis 14 pustular	614204
IL6	chr7	7p21	Diabetes susceptibility to	222100
IL6R	chr1	1q21.3	Hyper-IgE recurrent infection syndrome 5 autosomal recessive	618944
IL6ST	chr5	5q11	Hyper-IgE recurrent infection syndrome 4 autosomal recessive	618523
IL7	chr8	8q21.13	Epidermodysplasia verruciformis susceptibility to 5	618309
IL7R	chr5	5p13	Severe combined immunodeficiency T-cell negative B-cell/natural killer cell-positive type	608971
ILDR1	chr3	3q21.1	Deafness autosomal recessive 42	609646
IMPG2	chr3	3q11.2	Retinitis pigmentosa 56	613581
INPP5K	chr17	17p13.3	Muscular dystrophy congenital with cataracts and intellectual disability	617404
INPPL1	chr11	11q23	Opsismodysplasia	258480
INS	chr11	11p15.5	Maturity-onset diabetes of the young type 10	613370
INS	chr11	11p15.5	Hyperproinsulinemia	616214
INS	chr11	11p15.5	Diabetes mellitus insulin-dependent 2	125852
INS	chr11	11p15.5	Diabetes mellitus permanent neonatal	618858
INSR	chr19	19p13.2	Rabson-Mendenhall syndrome	262190
INSR	chr19	19p13.2	Leprechaunism	246200
INTU	chr4	4q28	Short-rib thoracic dysplasia 20 with polydactyly	617925
INTU	chr4	4q28	Orofaciodigital syndrome XVII	617926
INVS	chr9	9q31	Nephronophthisis 2 infantile	602088
IQCB1	chr3	3q13.33	Senior-Loken syndrome 5	609254
IQCE	chr7	7p22.3	Polydactyly postaxial type A7	617642
IRAK4	chr12	12q12	Immunodeficiency 67	607676
IRF7	chr11	11p15.5	Immunodeficiency 39	616345
IRF8	chr16	16q24.1	Immunodeficiency 32B monocyte and dendritic cell deficiency autosomal recessive	226990
IRF9	chr14	14q11.2	Immunodeficiency 65 susceptibility to viral infections	618648
IRS4	chrX	Xq22.3	Hypothyroidism congenital nongoitrous 9	301035
IRX5	chr16	16q11.2-q13	Hamamy syndrome	611174
ISCA2	chr14	14q24.3	Multiple mitochondrial dysfunctions syndrome 4	616370
ISCU	chr12	12q24.1	Myopathy with lactic acidosis hereditary	255125
ISG15	chr1	1p36.33	Immunodeficiency 38	616126
ITCH	chr20	20q11.22-q11.23	Autoimmune disease multisystem with facial dysmorphism	613385
ITGA2B	chr17	17q21.32	Glanzmann thrombasthenia	273800
ITGA3	chr17	17q21.33	Interstitial lung disease nephrotic syndrome and epidermolysis bullosa congenital	614748
ITGA6	chr2	2q31.1	Epidermolysis bullosa junctional with pyloric stenosis	226730
ITGA7	chr12	12q13	Muscular dystrophy congenital due to ITGA7 deficiency	613204
ITGA8	chr10	10p13	Renal hypodysplasia/aplasia 1	191830

ITGB2	chr21	21q22.3	Leukocyte adhesion deficiency	116920
ITGB3	chr17	17q21.32	Glanzmann thrombasthenia	273800
ITGB4	chr17	17q11-qter	Epidermolysis bullosa junctional with pyloric atresia	226730
ITGB4	chr17	17q11-qter	Epidermolysis bullosa junctional non-Herlitz type	226650
ITGB6	chr2	2q24.2	Amelogenesis imperfecta type IH	616221
ITK	chr5	5q32	Lymphoproliferative syndrome 1	613011
ITPA	chr20	20p	Epileptic encephalopathy early infantile 35	616647
ITPR1	chr3	3p26.1	Gillespie syndrome	206700
ITPR2	chr12	12p11	Anhidrosis isolated with normal sweat glands	106190
ITPR3	chr6	6pter-p21	Diabetes type 1 susceptibility to	222100
IVD	chr15	15q14-q15	Isovaleric acidemia	243500
IYD	chr6	6q24-q25	Thyroid dyshormonogenesis 4	274800
JAGN1	chr3	3p25.3	Neutropenia severe congenital 6 autosomal recessive	616022
JAK3	chr19	19p13.1	SCID autosomal recessive T-negative/B-positive type	600802
JAM3	chr11	11q25	Hemorrhagic destruction of the brain subependymal calcification and cataracts	613730
JUP	chr17	17q21	Naxos disease	601214
KANK2	chr19	19p13.2	Palmoplantar keratoderma and woolly hair	616099
KANK2	chr19	19p13.2	Nephrotic syndrome type 16	617783
KATNB1	chr16	16q21	Lissencephaly 6 with microcephaly	616212
KCNA4	chr11	11q13.4-q14.1	Microcephaly cataracts impaired intellectual development and dystonia with abnormal striatum	618284
KCNE1	chr21	21q22.1-q22.2	Jervell and Lange-Nielsen syndrome 2	612347
KCNJ1	chr11	11q24	Bartter syndrome type 2	241200
KCNJ10	chr1	1q23.2	Enlarged vestibular aqueduct digenic	600791
KCNJ10	chr1	1q23.2	SESAME syndrome	612780
KCNJ11	chr11	11p15.1	Hyperinsulinemic hypoglycemia familial 2	601820
KCNJ13	chr2	2q37	Leber congenital amaurosis 16	614186
KCNMA1	chr10	10q22.3	Cerebellar atrophy developmental delay and seizures	617643
KCNQ1	chr11	11p15.5	Jervell and Lange-Nielsen syndrome	220400
KCNV2	chr9	9p24.2	Retinal cone dystrophy 3B	610356
KCTD7	chr7	7q11.21	Epilepsy progressive myoclonic 3 with or without intracellular inclusions	611726
KDM5B	chr1	1q32	Mental retardation autosomal recessive 65	618109
KDM5C	chrX	Xp11.22-p11.21	Mental retardation X-linked syndromic Claes-Jensen type	300534
KDSR	chr18	18q21.3	Erythrokeratodermia variabilis et progressiva 4	617526
KERA	chr12	12q22	Cornea plana 2 autosomal recessive	217300
KHDC3L	chr6	6q13	Hydatidiform mole recurrent 2	614293
KHK	chr2	2p23.3-p23.2	Fructosuria	229800
KIAA1109	chr4	4q27	Alkuraya-Kucinskas syndrome	617822
KIAA1549	chr7	7q34	Retinitis pigmentosa 86	618613
KIF14	chr1	1q31	Microcephaly 20 primary autosomal recessive	617914
KIF14	chr1	1q31	Meckel syndrome 12	616258

KIF1A	chr2	2q37	Spastic paraplegia 30 autosomal dominant	610357
KIF1A	chr2	2q37	Neuropathy hereditary sensory type IIC	614213
KIF1A	chr2	2q37	Spastic paraplegia 30 autosomal recessive	610357
KIF1C	chr17	17p13.2	Spastic ataxia 2 autosomal recessive	611302
KIF4A	chrX	Xq13.1	Mental retardation X-linked 100	300923
KIF7	chr15	15q26.1	Hydrolethalus syndrome 2	614120
KIF7	chr15	15q26.1	Acrocallosal syndrome	200990
KIF7	chr15	15q26.1	Joubert syndrome 12	200990
KIF7	chr15	15q26.1	Al-Gazali-Bakalinova syndrome	607131
KISS1	chr1	1q32	Hypogonadotropic hypogonadism 13 with or without anosmia	614842
KISS1R	chr19	19p13.3	Hypogonadotropic hypogonadism 8 with or without anosmia	614837
KIZ	chr20	20p11.23	Retinitis pigmentosa 69	615780
KL	chr13	13q12	Tumoral calcinosis hyperphosphatemic familial 3	617994
KLHDC8B	chr3	3p21.31	Hodgkin lymphoma susceptibility to	236000
KLHL15	chrX	Xp22.1	Mental retardation X-linked 103	300982
KLHL3	chr5	5q31	Pseudohypoaldosteronism type IID	614495
KLHL40	chr3	3p22.1	Nemaline myopathy 8 autosomal recessive	615348
KLHL41	chr2	2q31.1	Nemaline myopathy 9	615731
KLHL7	chr7	7p15.3	PERCHING syndrome	617055
KLK4	chr19	19q13.4	Amelogenesis imperfecta type IIA1	204700
KLKB1	chr4	4q35	Fletcher factor (prekallikrein) deficiency	612423
KNG1	chr3	3q27	Kininogen deficiency	228960
KNG1	chr3	3q27	High molecular weight kininogen deficiency	228960
KNL1	chr15	15q15.1	Microcephaly 4 primary autosomal recessive	604321
KPTN	chr19	19q13.4	Mental retardation autosomal recessive 41	615637
KRT1	chr12	12q13	Epidermolytic hyperkeratosis	113800
KRT10	chr17	17q21-q22	Epidermolytic hyperkeratosis	113800
KRT14	chr17	17q12-q21	Epidermolysis bullosa simplex recessive 1	601001
KRT18	chr12	12q13	Cirrhosis noncryptogenic susceptibility to	215600
KRT18	chr12	12q13	Cirrhosis cryptogenic	215600
KRT5	chr12	12q13	Epidermolysis bullosa simplex recessive 1	601001
KRT74	chr12	12q13	Ectodermal dysplasia 7 hair/nail type	614929
KRT8	chr12	12q13	Cirrhosis noncryptogenic susceptibility to	215600
KRT8	chr12	12q13	Cirrhosis cryptogenic	215600
KRT83	chr12	12q13	Erythrokeratodermia variabilis et progressiva 5	617756
KRT85	chr12	12q13	Ectodermal dysplasia 4 hair/nail type	602032
KYNU	chr2	2q22.2	Vertebral cardiac renal and limb defects syndrome 2	617661
KYNU	chr2	2q22.2	Hydroxykynureninuria	236800
L1CAM	chrX	Xq28	MASA syndrome	303350
L1CAM	chrX	Xq28	Hydrocephalus with Hirschsprung disease	307000
L1CAM	chrX	Xq28	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction	307000
L1CAM	chrX	Xq28	Corpus callosum partial agenesis of	304100
L1CAM	chrX	Xq28	CRASH syndrome	303350
L1CAM	chrX	Xq28	Hydrocephalus due to aqueductal stenosis	307000

L2HGDH	chr14	14q22.1	L-2-hydroxyglutaric aciduria	236792
LAMA1	chr18	18p11.31	Poretti-Boltshauser syndrome	615960
LAMA2	chr6	6q22-q23	Muscular dystrophy limb-girdle autosomal recessive 23	618138
LAMA2	chr6	6q22-q23	Muscular dystrophy congenital merosin deficient or partially deficient	607855
LAMA3	chr18	18q11.2	Epidermolysis bullosa junctional Herlitz type	226700
LAMA3	chr18	18q11.2	Laryngoonychocutaneous syndrome	245660
LAMA3	chr18	18q11.2	Epidermolysis bullosa generalized atrophic benign	226650
LAMB1	chr7	7q31.1-q31.3	Lissencephaly 5	615191
LAMB2	chr3	3p21	Pierson syndrome	609049
LAMB3	chr1	1q32	Epidermolysis bullosa junctional Herlitz type	226700
LAMB3	chr1	1q32	Epidermolysis bullosa junctional non-Herlitz type	226650
LAMC2	chr1	1q25-q31	Epidermolysis bullosa junctional non-Herlitz type	226650
LAMC2	chr1	1q25-q31	Epidermolysis bullosa junctional Herlitz type	226700
LAMC3	chr9	9q33-q34	Cortical malformations occipital	614115
LAMTOR2	chr1	1q22	Immunodeficiency due to defect in MAPBP-interacting protein	610798
LARGE1	chr22	22q12.3-q13.1	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 6	608840
LARGE1	chr22	22q12.3-q13.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 6	613154
LARP7	chr4	4q25	Alazami syndrome	615071
LARS2	chr3	3p21.3	Perrault syndrome 4	615300
LARS2	chr3	3p21.3	Hydrops lactic acidosis and sideroblastic anemia	617021
LAS1L	chrX	Xq12	Wilson-Turner syndrome	309585
LBR	chr1	1q42.1	Greenberg skeletal dysplasia	215140
LCA5	chr6	6q14.1	Leber congenital amaurosis 5	604537
LCAT	chr16	16q22.1	Norum disease	245900
LCAT	chr16	16q22.1	Fish-eye disease	136120
LCK	chr1	1p35-p34.3	Immunodeficiency 22	615758
LCT	chr2	2q21	Lactase deficiency congenital	223000
LDHA	chr11	11p15.4	Glycogen storage disease XI	612933
LDLRAP1	chr1	1p36-p35	Hypercholesterolemia familial 4	603813
LEP	chr7	7q31.3	Obesity morbid due to leptin deficiency	614962
LEPR	chr1	1p31	Obesity morbid due to leptin receptor deficiency	614963
P3H1	chr1	1p34	Osteogenesis imperfecta type VIII	610915
LFNG	chr7	7p22	Spondylocostal dysostosis 3 autosomal recessive	609813
LGI4	chr19	19q13.11	Arthrogryposis multiplex congenita 1 neurogenic with myelin defect	617468
LHB	chr19	19q13.32	Hypogonadotropic hypogonadism 23 with or without anosmia	228300
LHCGR	chr2	2p21	Luteinizing hormone resistance female	238320
LHCGR	chr2	2p21	Leydig cell hypoplasia with pseudohermaphroditism	238320
LHCGR	chr2	2p21	Leydig cell hypoplasia with hypergonadotropic hypogonadism	238320
LHFPL5	chr6	6p21.3	Deafness autosomal recessive 67	610265
LHX3	chr9	9q34.3	Pituitary hormone deficiency combined 3	221750

LIAS	chr4	4p14	Hyperglycinemia lactic acidosis and seizures	614462
LIFR	chr5	5p13.1	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	601559
LIG4	chr13	13q22-q34	LIG4 syndrome	606593
LIM2	chr19	19q13.4	Cataract 19 multiple types	615277
LIMS2	chr2	2q14.3	Muscular dystrophy autosomal recessive with cardiomyopathy and triangular tongue	616827
LINS1	chr15	15q26	Mental retardation autosomal recessive 27	614340
LIPA	chr10	10q23.31	Wolman disease	278000
LIPA	chr10	10q23.31	Cholesteryl ester storage disease	278000
LIPC	chr15	15q21.3	Hepatic lipase deficiency	614025
LIPE	chr19	19q13.1-q13.2	Lipodystrophy familial partial type 6	615980
LIPH	chr3	3q27-q28	Woolly hair autosomal recessive 2 with or without hypotrichosis	604379
LIPH	chr3	3q27-q28	Hypotrichosis 7	604379
LIPN	chr10	10q23.31	Ichthyosis congenital autosomal recessive 8	613943
LIPT1	chr2	2q11.2	Lipoyltransferase 1 deficiency	616299
LMAN1	chr18	18q21.3-q22	Combined factor V and VIII deficiency	227300
LMAN2L	chr2	2q11.2	Mental retardation autosomal recessive 52	616887
LMBR1	chr7	7q36	Acheiropody	200500
LMBRD1	chr6	6q13	Methylmalonic aciduria and homocystinuria cblF type	277380
LMF1	chr16	16p13.3	Lipase deficiency combined	246650
LMNA	chr1	1q21.2	Charcot-Marie-Tooth disease type 2B1	605588
LMNA	chr1	1q21.2	Hutchinson-Gilford progeria	176670
LMNA	chr1	1q21.2	Restrictive dermopathy lethal	275210
LMNA	chr1	1q21.2	Mandibuloacral dysplasia	248370
LMNA	chr1	1q21.2	Emery-Dreifuss muscular dystrophy 3 autosomal recessive	616516
LMNB2	chr19	19p13.3	Epilepsy progressive myoclonic 9	616540
LMOD3	chr3	3p14.1	Nemaline myopathy 10	616165
LONP1	chr19	19p13.2	CODAS syndrome	600373
LOXHD1	chr18	18q12-q21	Deafness autosomal recessive 77	613079
LPAR6	chr13	13q14.12-q14.2	Hypotrichosis 8	278150
LPAR6	chr13	13q14.12-q14.2	Woolly hair autosomal recessive 1 with or without hypotrichosis	278150
LPIN1	chr2	2p21	Myoglobinuria acute recurrent autosomal recessive	268200
LPL	chr8	8p22	Lipoprotein lipase deficiency	238600
LPL	chr8	8p22	High density lipoprotein cholesterol level QTL 11	238600
LRAT	chr4	4q31	Retinal dystrophy early-onset severe	613341
LRAT	chr4	4q31	Leber congenital amaurosis 14	613341
LRAT	chr4	4q31	Retinitis pigmentosa juvenile	613341
LRBA	chr4	4q31.3	Immunodeficiency common variable 8 with autoimmunity	614700
LRIG2	chr1	1p13	Urofacial syndrome 2	615112
LRIT3	chr4	4q25	Night blindness congenital stationary (complete) 1F autosomal recessive	615058

LRMDA	chr10	10q22.3	Albinism oculocutaneous type VII	615179
LRP1	chr12	12q13.1-q13.3	Keratosis pilaris atrophicans	604093
LRP2	chr2	2q24-q31	Donnai-Barrow syndrome	222448
LRP4	chr11	11p12-p11.2	Myasthenic syndrome congenital 17	616304
LRP4	chr11	11p12-p11.2	Sclerosteosis 2	614305
LRP4	chr11	11p12-p11.2	Cenani-Lenz syndactyly syndrome	212780
LRP5	chr11	11q13.4	Exudative vitreoretinopathy 4	601813
LRP5	chr11	11q13.4	Osteoporosis-pseudoglioma syndrome	259770
LRPAP1	chr4	4p16.3	Myopia 23 autosomal recessive	615431
LRPPRC	chr2	2p21	Leigh syndrome French-Canadian type	220111
LRRC6	chr8	8q24.22	Ciliary dyskinesia primary 19	614935
LRSAM1	chr9	9q33.3-q34.11	Charcot-Marie-Tooth disease axonal type 2P	614436
LRTOMT	chr11	11q13.3-q13.4	Deafness autosomal recessive 63	611451
LTBP2	chr14	14q24	Microspherophakia and/or megalocornea with ectopia lentis and with or without secondary glaucoma	251750
LTBP2	chr14	14q24	Weill-Marchesani syndrome 3 recessive	614819
LTBP3	chr11	11q12	Dental anomalies and short stature	601216
LTBP4	chr19	19q13.1-q13.2	Cutis laxa autosomal recessive type IC	613177
LTC4S	chr5	5q35	Leukotriene C4 synthase deficiency	614037
LYRM4	chr6	6p25.1	Combined oxidative phosphorylation deficiency 19	615595
LYRM7	chr5	5q23.3	Mitochondrial complex III deficiency nuclear type 8	615838
LYST	chr1	1q42.3	Chediak-Higashi syndrome	214500
LZTFL1	chr3	3p21.3	Bardet-Biedl syndrome 17	615994
LZTR1	chr22	22q11.1-q11.2	Noonan syndrome 2	605275
MAB21L2	chr4	4q31.3	Microphthalmia/coloboma and skeletal dysplasia syndrome	615877
MAG	chr19	19q13.1	Spastic paraplegia 75 autosomal recessive	616680
MAGI2	chr7	7q21	Nephrotic syndrome type 15	617609
MAGT1	chrX	Xq13.1-q13.2	Immunodeficiency X-linked with magnesium defect Epstein-Barr virus infection and neoplasia	300853
MAGT1	chrX	Xq13.1-q13.2	Congenital disorder of glycosylation type Icc	301031
MAK	chr6	6p24.2	Retinitis pigmentosa 62	614181
MALT1	chr18	18q21	Immunodeficiency 12	615468
MAMLD1	chrX	Xq28	Hypospadias 2 X-linked	300758
MAN1B1	chr9	9q34.3	Mental retardation autosomal recessive 15	614202
MAN2B1	chr19	19cen-q12	Mannosidosis alpha- types I and II	248500
MANBA	chr4	4q22-q25	Mannosidosis beta	248510
MAOA	chrX	Xp11.23	Brunner syndrome	300615
MAOA	chrX	Xp11.23	Antisocial behavior	300615
MAPT	chr17	17q21.1	Supranuclear palsy progressive atypical	260540
MARS2	chr2	2q33.1	Spastic ataxia 3 autosomal recessive	611390
MARS2	chr2	2q33.1	Combined oxidative phosphorylation deficiency 25	616430
MARVELD2	chr5	5q13.1	Deafness autosomal recessive 49	610153

MASP1	chr3	3q27-q28	3MC syndrome 1	257920
MASP2	chr1	1p36.3-p36.2	MASP2 deficiency	613791
MAT1A	chr10	10q22	Methionine adenosyltransferase deficiency autosomal recessive	250850
MAT1A	chr10	10q22	Hypermethioninemia persistent autosomal dominant due to methionine adenosyltransferase I/III deficiency	250850
MATN3	chr2	2p24-p23	Spondyloepimetaphyseal dysplasia	608728
MBOAT7	chr19	19q13.4	Mental retardation autosomal recessive 57	617188
MBTPS2	chrX	Xp22.12-p22.11	IFAP syndrome with or without BRESHECK syndrome	308205
MBTPS2	chrX	Xp22.12-p22.11	Keratosis follicularis spinulosa decalvans X-linked	308800
MBTPS2	chrX	Xp22.12-p22.11	Osteogenesis imperfecta type XIX	301014
MBTPS2	chrX	Xp22.12-p22.11	?Olmsted syndrome X-linked	300918
MC1R	chr16	16q24.3	Skin/hair/eye pigmentation 2 blond hair/fair skin	266300
MC1R	chr16	16q24.3	Albinism oculocutaneous type II modifier of	203200
MC1R	chr16	16q24.3	UV-induced skin damage	266300
MC1R	chr16	16q24.3	Skin/hair/eye pigmentation 2 red hair/fair skin	266300
MC2R	chr18	18p11.2	Glucocorticoid deficiency due to ACTH unresponsiveness	202200
MC4R	chr18	18q22	Obesity (BMIQ20)	618406
MCCC1	chr3	3q25-q27	3-Methylcrotonyl-CoA carboxylase 1 deficiency	210200
MCCC2	chr5	5q12-q13	3-Methylcrotonyl-CoA carboxylase 2 deficiency	210210
MCEE	chr2	2p13.3	Methylmalonyl-CoA epimerase deficiency	251120
MCM3AP	chr21	21q22.3	Peripheral neuropathy autosomal recessive with or without impaired intellectual development	618124
MCM4	chr8	8q11.2	Immunodeficiency 54	609981
MCM5	chr22	22q13.1	Meier-Gorlin syndrome 8	617564
MCM8	chr20	20p13-p12.3	Premature ovarian failure 10	612885
MCM9	chr6	6q22.1-q22.33	Ovarian dysgenesis 4	616185
MCOLN1	chr19	19p13.3-p13.2	Mucolipidosis IV	252650
MCPH1	chr8	8p23	Microcephaly 1 primary autosomal recessive	251200
MDH2	chr7	7q11.23	Epileptic encephalopathy early infantile 51	617339
MDM2	chr12	12q14.3-q15	Lessel-Kubisch syndrome	618681
MECP2	chrX	Xq28	Mental retardation X-linked syndromic Lubs type	300260
MECP2	chrX	Xq28	Encephalopathy neonatal severe	300673
MECP2	chrX	Xq28	Mental retardation X-linked syndromic 13	300055
MECP2	chrX	Xq28	Autism susceptibility X-linked 3	300496
MED12	chrX	Xq13	Ohdo syndrome X-linked	300895
MED12	chrX	Xq13	Lujan-Fryns syndrome	309520
MED12	chrX	Xq13	Opitz-Kaveggia syndrome	305450
MED17	chr11	11q21	Microcephaly postnatal progressive with seizures and brain atrophy	613668
MED23	chr6	6q23.2	Mental retardation autosomal recessive 18	614249
MED25	chr19	19q13.3	Basel-Vanagait-Smirin-Yosef syndrome	616449
MEFV	chr16	16p13	Familial Mediterranean fever AR	249100

MEGF10	chr5	5q23.2	Myopathy areflexia respiratory distress and dysphagia early-onset mild variant	614399
MEGF10	chr5	5q23.2	Myopathy areflexia respiratory distress and dysphagia early-onset	614399
MEGF8	chr19	19q12	Carpenter syndrome 2	614976
MEOX1	chr17	17q21	Klippel-Feil syndrome 2	214300
MERTK	chr2	2q14.1	Retinitis pigmentosa 38	613862
MESD	chr15	15q25.1	Osteogenesis imperfecta type XX	618644
MESP2	chr15	15q26.1	Spondylocostal dysostosis 2 autosomal recessive	608681
MET	chr7	7q31	Deafness autosomal recessive 97	616705
METTL23	chr17	17q25.1	Mental retardation autosomal recessive 44	615942
MFF	chr2	2q36.3	Encephalopathy due to defective mitochondrial and peroxisomal fission 2	617086
MFN2	chr1	1p36.2	Charcot-Marie-Tooth disease axonal type 2A2B	617087
MFRP	chr11	11q23	Microphthalmia isolated 5	611040
MFSD2A	chr1	1p34.2	Neurodevelopmental disorder with progressive microcephaly spasticity and brain imaging abnormalities	616486
MFSD8	chr4	4q28.1-q28.2	Macular dystrophy with central cone involvement	616170
MFSD8	chr4	4q28.1-q28.2	Ceroid lipofuscinosis neuronal 7	610951
MGAT2	chr14	14q21	Congenital disorder of glycosylation type IIa	212066
MGME1	chr20	20p11.23	Mitochondrial DNA depletion syndrome 11	615084
MGP	chr12	12p13.1-p12.3	Keutel syndrome	245150
MICU1	chr10	10q22.1	Myopathy with extrapyramidal signs	615673
MID1	chrX	Xp22	Opitz GBBB syndrome type I	300000
MID2	chrX	Xq22	Mental retardation X-linked 101	300928
MIR2861	chr9	9q34.11	Bone mineral density QTL 15	613418
MITF	chr3	3p14.1-p12.3	COMMAD syndrome	617306
MKKS	chr20	20p12	Bardet-Biedl syndrome 6	605231
MKKS	chr20	20p12	McKusick-Kaufman syndrome	236700
MKS1	chr17	17q23	Bardet-Biedl syndrome 13	615990
MKS1	chr17	17q23	Joubert syndrome 28	617121
MKS1	chr17	17q23	Meckel syndrome 1	249000
MLC1	chr22	22q13.33	Megalencephalic leukoencephalopathy with subcortical cysts	604004
MLH1	chr3	3p21.3	Mismatch repair cancer syndrome	276300
MLPH	chr2	2q37	Griscelli syndrome type 3	609227
MLYCD	chr16	16q24	Malonyl-CoA decarboxylase deficiency	248360
MMAA	chr4	4q31.1-q31.2	Methylmalonic aciduria vitamin B12-responsive	251100
MMAB	chr12	12q24	Methylmalonic aciduria vitamin B12-responsive due to defect in synthesis of adenosylcobalamin cblB complementation type	251110
MMACHC	chr1	1p34.1	Methylmalonic aciduria and homocystinuria cblC type	277400
MMADHC	chr2	2q23.2	Homocystinuria cblD type variant 1	277410
MMADHC	chr2	2q23.2	Methylmalonic aciduria and homocystinuria cblD type	277410
MMADHC	chr2	2q23.2	Methylmalonic aciduria cblD type variant 2	277410
MME	chr3	3q21-q27	Charcot-Marie-Tooth disease axonal type 2T	617017

MMP1	chr11	11q22-q23	Epidermolysis bullosa dystrophica autosomal recessive modifier of	226600
MMP13	chr11	11q22.3	Metaphyseal dysplasia Spahr type	250400
MMP2	chr16	16q13	Multicentric osteolysis nodulosis and arthropathy	259600
MMP20	chr11	11q22.3-q23	Amelogenesis imperfecta type IIA2	612529
MOCOS	chr18	18q12.2	Xanthinuria type II	603592
MOCS1	chr6	6p21.3	Molybdenum cofactor deficiency A	252150
MOCS2	chr5	5q11	Molybdenum cofactor deficiency B	252160
MOGS	chr2	2p13.1	Congenital disorder of glycosylation type IIb	606056
MPC1	chr6	6q27	Mitochondrial pyruvate carrier deficiency	614741
MPDU1	chr17	17p13.1-p12	Congenital disorder of glycosylation type If	609180
MPDZ	chr9	9p23	Hydrocephalus congenital 2 with or without brain or eye anomalies	615219
MPI	chr15	15q22-qter	Congenital disorder of glycosylation type Ib	602579
MPL	chr1	1p34	Thrombocytopenia congenital amegakaryocytic	604498
MPLKIP	chr7	7p14	Trichothiodystrophy 4 nonphotosensitive	234050
MPO	chr17	17q23.1	Myeloperoxidase deficiency	254600
MPV17	chr2	2p23-p21	Charcot-Marie-Tooth disease axonal type 2EE	618400
MPV17	chr2	2p23-p21	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	256810
MPZ	chr1	1q22	Dejerine-Sottas disease	145900
MRAP	chr21	21q22.1	Glucocorticoid deficiency 2	607398
MRPL12	chr17	17q25-qter	Combined oxidative phosphorylation deficiency 45	618951
MRPL3	chr3	3q22.1	Combined oxidative phosphorylation deficiency 9	614582
MRPL44	chr2	2q36	Combined oxidative phosphorylation deficiency 16	615395
MRPS16	chr10	10q22.1	Combined oxidative phosphorylation deficiency 2	610498
MRPS2	chr9	9q34	Combined oxidative phosphorylation deficiency 36	617950
MRPS22	chr3	3q23	Combined oxidative phosphorylation deficiency 5	611719
MRPS22	chr3	3q23	Ovarian dysgenesis 7	618117
MS4A1	chr11	11q13	Immunodeficiency common variable 5	613495
MSH2	chr2	2p22-p21	Mismatch repair cancer syndrome	276300
MSH3	chr5	5q11-q12	Familial adenomatous polyposis 4	617100
MSH5	chr6	6p21.33	Premature ovarian failure 13	617442
MSH6	chr2	2p16	Mismatch repair cancer syndrome	276300
MSMO1	chr4	4q32-q34	Microcephaly congenital cataract and psoriasiform dermatitis	616834
MSRB3	chr12	12q14.3	Deafness autosomal recessive 74	613718
MTFMT	chr15	15q22.31	Combined oxidative phosphorylation deficiency 15	614947
MTFMT	chr15	15q22.31	Mitochondrial complex I deficiency nuclear type 27	618248
MTHFD1	chr14	14q24	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	617780
MTHFD1	chr14	14q24	Neural tube defects folate-sensitive susceptibility to	601634
MTHFR	chr1	1p36.3	Homocystinuria due to MTHFR deficiency	236250
MTHFR	chr1	1p36.3	Neural tube defects susceptibility to	601634
MTHFS	chr15	15q25.1	Neurodevelopmental disorder with microcephaly epilepsy and hypomyelination	618367

MTM1	chrX	Xq28	Myotubular myopathy X-linked	310400
MTMR2	chr11	11q22	Charcot-Marie-Tooth disease type 4B1	601382
MTO1	chr6	6q13	Combined oxidative phosphorylation deficiency 10	614702
MTPAP	chr10	10p11.23	Spastic ataxia 4 autosomal recessive	613672
MTR	chr1	1q43	Neural tube defects folate-sensitive susceptibility to	601634
MTR	chr1	1q43	Homocystinuria-megaloblastic anemia cblG complementation type	250940
MTRR	chr5	5p15.3-p15.2	Neural tube defects folate-sensitive susceptibility to	601634
MTRR	chr5	5p15.3-p15.2	Homocystinuria-megaloblastic anemia cbl E type	236270
MTTP	chr4	4q22-q24	Abetalipoproteinemia	200100
MUSK	chr9	9q31.3-q32	Fetal akinesia deformation sequence 1	208150
MUSK	chr9	9q31.3-q32	Myasthenic syndrome congenital 9 associated with acetylcholine receptor deficiency	616325
MUTYH	chr1	1p34.3-p32.1	Adenomas multiple colorectal	608456
MVK	chr12	12q24	Hyper-IgD syndrome	260920
MVK	chr12	12q24	Mevalonic aciduria	610377
MYBPC1	chr12	12q23.2	Lethal congenital contracture syndrome 4	614915
MYBPC3	chr11	11p11.2	Cardiomyopathy hypertrophic 4	115197
MYD88	chr3	3p22-p21.3	Immunodeficiency 68	612260
MYF5	chr12	12q21	Ophthalmoplegia external with rib and vertebral anomalies	618155
MYH2	chr17	17p13.1	Proximal myopathy and ophthalmoplegia	605637
MYH3	chr17	17p13.1	Contractures pterygia and spondylocarpotarsal fusion syndrome 1B	618469
MYH7	chr14	14q12	Myopathy myosin storage autosomal recessive	255160
MYL1	chr2	2q32.1-qter	Myopathy congenital with fast-twitch (type II) fiber atrophy	618414
MYL3	chr3	3p	Cardiomyopathy hypertrophic 8	608751
MYLK	chr3	3q21	Megacystis-microcolon-intestinal hypoperistalsis syndrome	249210
MYO15A	chr17	17p11.2	Deafness autosomal recessive 3	600316
MYO18B	chr22	22q12.1	Klippel-Feil syndrome 4 autosomal recessive with myopathy and facial dysmorphism	616549
MYO1E	chr15	15q21-q22	Glomerulosclerosis focal segmental 6	614131
MYO3A	chr10	10p11.1	Deafness autosomal recessive 30	607101
MYO5A	chr15	15q21	Griscelli syndrome type 1	214450
MYO5B	chr18	18q21	Microvillus inclusion disease	251850
MYO6	chr6	6q13	Deafness autosomal recessive 37	607821
MYO7A	chr11	11q13.5	Deafness autosomal recessive 2	600060
MYO7A	chr11	11q13.5	Usher syndrome type 1B	276900
MYPN	chr10	10q21.1	Nemaline myopathy 11 autosomal recessive	617336
NAA10	chrX	Xq28	Ogden syndrome	300855
NAA10	chrX	Xq28	Microphthalmia syndromic 1	309800
NADK2	chr5	5p13.2	24-dienoyl-CoA reductase deficiency	616034
NAGA	chr22	22q11	Kanzaki disease	609242
NAGA	chr22	22q11	Schindler disease type I	609241
NAGA	chr22	22q11	Schindler disease type III	609241
NAGLU	chr17	17q21	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920

NAGS	chr17	17q21.31	N-acetylglutamate synthase deficiency	237310
NALCN	chr13	13q33.11	Hypotonia infantile with psychomotor retardation and characteristic facies 1	615419
NARS2	chr11	11q14.1	Deafness autosomal recessive 94	618434
NARS2	chr11	11q14.1	Combined oxidative phosphorylation deficiency 24	616239
NAT2	chr8	8p23.1-p21.3	Acetylation slow	243400
NAT8L	chr4	4p16.3	N-acetylaspartate deficiency	614063
NBAS	chr2	2p24-p23	Infantile liver failure syndrome 2	616483
NBAS	chr2	2p24-p23	Short stature optic nerve atrophy and Pelger-Huet anomaly	614800
NBEAL2	chr3	3p22.1-p21.1	Gray platelet syndrome	139090
NBN	chr8	8q21	Nijmegen breakage syndrome	251260
NCAPD2	chr12	12p13.31	Microcephaly 21 primary autosomal recessive	617983
NCAPG2	chr7	7q36.3	Khan-Khan-Katsanis syndrome	618460
NCF1	chr7	7q11.23	Chronic granulomatous disease 1 autosomal recessive	233700
NCF2	chr1	1q25	Chronic granulomatous disease due to deficiency of NCF-2	233710
NCF4	chr22	22q13.1	Chronic granulomatous disease 3 autosomal recessive	613960
NDE1	chr16	16p13.1	Lissencephaly 4 (with microcephaly)	614019
NDE1	chr16	16p13.1	Microhydranencephaly	605013
NDP	chrX	Xp11.4	Exudative vitreoretinopathy 2 X-linked	305390
NDP	chrX	Xp11.4	Norrie disease	310600
NDRG1	chr8	8q24.3	Charcot-Marie-Tooth disease type 4D	601455
NDST1	chr5	5q32-q33.3	Mental retardation autosomal recessive 46	616116
NDUFA1	chrX	Xq24	Mitochondrial complex I deficiency nuclear type 12	301020
NDUFA10	chr2	2q37.3	Mitochondrial complex I deficiency nuclear type 22	618243
NDUFA11	chr19	19p13.3	Mitochondrial complex I deficiency nuclear type 14	618236
NDUFA12	chr12	12q22	Mitochondrial complex I deficiency nuclear type 23	618244
NDUFA13	chr19	19p13.2-p13.1	Mitochondrial complex I deficiency nuclear type 28	618249
NDUFA2	chr5	5q31.2	Mitochondrial complex I deficiency nuclear type 13	618235
NDUFA6	chr22	22q13.1	Mitochondrial complex I deficiency nuclear type 33	618253
NDUFA9	chr12	12p	Mitochondrial complex I deficiency nuclear type 26	618247
NDUFAF1	chr15	15q13.3	Mitochondrial complex I deficiency nuclear type 11	618234
NDUFAF2	chr5	5q12.1	Mitochondrial complex I deficiency nuclear type 10	618233
NDUFAF3	chr3	3p21.31	Mitochondrial complex I deficiency nuclear type 18	618240
NDUFAF4	chr6	6q16.1	Mitochondrial complex I deficiency nuclear type 15	618237
NDUFAF5	chr20	20p12.1	Mitochondrial complex I deficiency nuclear type 16	618238
NDUFAF6	chr8	8q22.1	Fanconi renotubular syndrome 5	618913
NDUFAF6	chr8	8q22.1	Mitochondrial complex I deficiency nuclear type 17	618239
NDUFB3	chr2	2q31.3	Mitochondrial complex I deficiency nuclear type 25	618246
NDUFB8	chr10	10q23.2-q23.33	Mitochondrial complex I deficiency nuclear type 32	618252

NDUFB9	chr8	8q13.3	Mitochondrial complex I deficiency nuclear type 24	618245
NDUFS1	chr2	2q33-q34	Mitochondrial complex I deficiency nuclear type 5	618226
NDUFS2	chr1	1q23	Mitochondrial complex I deficiency nuclear type 6	618228
NDUFS3	chr11	11p11.11	Mitochondrial complex I deficiency nuclear type 8	618230
NDUFS4	chr5	5q11.1	Mitochondrial complex I deficiency nuclear type 1	252010
NDUFS6	chr5	5pter-p15.33	Mitochondrial complex I deficiency nuclear type 9	618232
NDUFS7	chr19	19p13	Mitochondrial complex I deficiency nuclear type 3	618224
NDUFS8	chr11	11q13	Mitochondrial complex I deficiency nuclear type 2	618222
NDUFV1	chr11	11q13	Mitochondrial complex I deficiency nuclear type 4	618225
NDUFV2	chr18	18p11.31-p11.2	Mitochondrial complex I deficiency nuclear type 7	618229
NEB	chr2	2q22	Nemaline myopathy 2 autosomal recessive	256030
NECAP1	chr12	12p13.31	Epileptic encephalopathy early infantile 21	615833
NECTIN1	chr11	11q23-q24	Orofacial cleft 7	225060
NECTIN1	chr11	11q23-q24	Cleft lip/palate-ectodermal dysplasia syndrome	225060
NECTIN4	chr1	1q23.3	Ectodermal dysplasia-syndactyly syndrome 1	613573
NEFH	chr22	22q12.2	Charcot-Marie-Tooth disease axonal type 2CC	616924
NEFH	chr22	22q12.2	Amyotrophic lateral sclerosis susceptibility to	105400
NEFL	chr8	8p21	Charcot-Marie-Tooth disease type 1F	607734
NEK1	chr4	4q33	Short-rib thoracic dysplasia 6 with or without polydactyly	263520
NEK2	chr1	1q32.2-q41	Retinitis pigmentosa 67	615565
NEK8	chr17	17q11.1	Renal-hepatic-pancreatic dysplasia 2	615415
NEU1	chr6	6p21.3	Sialidosis type II	256550
NEU1	chr6	6p21.3	Sialidosis type I	256550
NEUROG3	chr10	10q21.3	Diarrhea 4 malabsorptive congenital	610370
NFU1	chr2	2p15-p13	Multiple mitochondrial dysfunctions syndrome 1	605711
NGF	chr1	1p13.1	Neuropathy hereditary sensory and autonomic type V	608654
NGLY1	chr3	3p24	Congenital disorder of deglycosylation	615273
NHLRC1	chr6	6p22.3	Epilepsy progressive myoclonic 2B (Lafora)	254780
NHP2	chr5	5q35.3	Dyskeratosis congenita autosomal recessive 2	613987
NIN	chr14	14q22.1	Seckel syndrome 7	614851
NIPAL4	chr5	5q33	Ichthyosis congenital autosomal recessive 6	612281
NKX3-2	chr4	4p16.1	Spondylo-megaepiphyseal-metaphyseal dysplasia	613330
NLRP1	chr17	17p13	Autoinflammation with arthritis and dyskeratosis	617388
NLRP1	chr17	17p13	Respiratory papillomatosis juvenile recurrent congenital	618803
NLRP7	chr19	19q13.4	Hydatidiform mole recurrent 1	231090
NME8	chr7	7p14.1	Ciliary dyskinesia primary 6	610852
NMNAT1	chr1	1p36.22	Leber congenital amaurosis 9	608553
NNT	chr5	5p12	Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency	614736
NOP10	chr15	15q14-q15	Dyskeratosis congenita autosomal recessive 1	224230
NPC1	chr18	18q11-q12	Niemann-Pick disease type D	257220
NPC1	chr18	18q11-q12	Niemann-Pick disease type C1	257220
NPC2	chr14	14q24.3	Niemann-pick disease type C2	607625
NPHP1	chr2	2q13	Nephronophthisis 1 juvenile	256100

NPHP1	chr2	2q13	Senior-Loken syndrome-1	266900
NPHP1	chr2	2q13	Joubert syndrome 4	609583
NPHP3	chr3	3q22	Meckel syndrome 7	267010
NPHP3	chr3	3q22	Renal-hepatic-pancreatic dysplasia 1	208540
NPHP3	chr3	3q22	Nephronophthisis 3	604387
NPHP4	chr1	1p36	Nephronophthisis 4	606966
NPHP4	chr1	1p36	Senior-Loken syndrome 4	606996
NPHS1	chr19	19q13.1	Nephrotic syndrome type 1	256300
NPHS2	chr1	1q25-q31	Nephrotic syndrome type 2	600995
NPPA	chr1	1p36.2	Atrial standstill 2	615745
NPR2	chr9	9p21-p12	Acromesomelic dysplasia Maroteaux type	602875
NR0B1	chrX	Xp21.3-p21.2	Adrenal hypoplasia congenital	300200
NR0B1	chrX	Xp21.3-p21.2	46XY sex reversal 2 dosage-sensitive	300018
NR0B2	chr1	1p36.1	Obesity mild early-onset	601665
NR1H4	chr12	12q23.1	Cholestasis progressive familial intrahepatic 5	617049
NR2E3	chr15	15q23	Enhanced S-cone syndrome	268100
NR2E3	chr15	15q23	Retinitis pigmentosa 37	611131
NRL	chr14	14q11.1-q11.2	Retinal degeneration autosomal recessive clumped pigment type	
NRXN1	chr2	2p16.3	Pitt-Hopkins-like syndrome 2	614325
NSDHL	chrX	Xq28	CK syndrome	300831
NSMCE3	chr15	15q13.1	Lung disease immunodeficiency and chromosome breakage syndrome	617241
NSUN2	chr5	5p15.31	Mental retardation autosomal recessive 5	611091
NT5C2	chr10	10q24.3	Spastic paraplegia 45 autosomal recessive	613162
NT5C3A	chr7	7p15-p14	Anemia hemolytic due to UMPH1 deficiency	266120
NT5E	chr6	6q14-q21	Calcification of joints and arteries	211800
NTHL1	chr16	16p13.3-p13.2	Familial adenomatous polyposis 3	616415
NTRK1	chr1	1q21-q22	Insensitivity to pain congenital with anhidrosis	256800
NUBPL	chr14	14q12	Mitochondrial complex I deficiency nuclear type 21	618242
NUP107	chr12	12q15	Galloway-Mowat syndrome 7	618348
NUP107	chr12	12q15	Ovarian dysgenesis 6	618078
NUP107	chr12	12q15	Nephrotic syndrome type 11	616730
NUP155	chr5	5p13	Atrial fibrillation 15	615770
NUP214	chr9	9q34.1	Encephalopathy acute infection-induced susceptibility to 9	618426
NUP62	chr19	19q13.33	Striatonigral degeneration infantile	271930
NXN	chr17	17p13.3	Robinow syndrome autosomal recessive 2	618529
NYX	chrX	Xp11.4	Night blindness congenital stationary (complete) 1A X-linked	310500
OAT	chr10	10q26	Gyrate atrophy of choroid and retina with or without ornithinemia	258870
OBSL1	chr2	2q35	3-M syndrome 2	612921
OCA2	chr15	15q11.2-q12	Skin/hair/eye pigmentation 1 blue/nonblue eyes	227220
OCA2	chr15	15q11.2-q12	Skin/hair/eye pigmentation 1 blond/brown hair	227220
OCA2	chr15	15q11.2-q12	Albinism oculocutaneous type II	203200
OCA2	chr15	15q11.2-q12	Albinism brown oculocutaneous	203200

OCLN	chr5	5q13.1	Pseudo-TORCH syndrome 1	251290
OCRL	chrX	Xq26.1	Lowe syndrome	309000
OCRL	chrX	Xq26.1	Dent disease 2	300555
ODAPH	chr4	4q21.1	Amelogenesis imperfecta type IIA4	614832
OFD1	chrX	Xp22.3-p22.2	Retinitis pigmentosa 23	300424
OFD1	chrX	Xp22.3-p22.2	Joubert syndrome 10	300804
OFD1	chrX	Xp22.3-p22.2	Simpson-Golabi-Behmel syndrome type 2	300209
OGDH	chr7	7p14-p13	Alpha-ketoglutarate dehydrogenase deficiency	203740
OGT	chrX	Xq13	Mental retardation X-linked 106	300997
OPA1	chr3	3q28-q29	Behr syndrome	210000
OPA1	chr3	3q28-q29	Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type)	616896
OPA3	chr19	19q13.2-q13.3	3-methylglutaconic aciduria type III	258501
OPHN1	chrX	Xq12	Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance	300486
OPLAH	chr8	8q24.3	5-oxoprolinase deficiency	260005
OPN1LW	chrX	Xq28	Blue cone monochromacy	303700
OPN1LW	chrX	Xq28	Colorblindness protan	303900
OPN1MW	chrX	Xq28	Colorblindness deutan	303800
OPN1MW	chrX	Xq28	Blue cone monochromacy	303700
ORAI1	chr12	12q24	Immunodeficiency 9	612782
ORC1	chr1	1p32	Meier-Gorlin syndrome 1	224690
ORC4	chr2	2q22-q23	Meier-Gorlin syndrome 2	613800
ORC6	chr16	16q12	Meier-Gorlin syndrome 3	613803
OSTM1	chr6	6q21	Osteopetrosis autosomal recessive 5	259720
OTC	chrX	Xp21.1	Ornithine transcarbamylase deficiency	311250
OTOA	chr16	16p12.2	Deafness autosomal recessive 22	607039
OTOF	chr2	2p23-p22	Auditory neuropathy autosomal recessive 1	601071
OTOF	chr2	2p23-p22	Deafness autosomal recessive 9	601071
OTOG	chr11	11p14.3	Deafness autosomal recessive 18B	614945
OTOGL	chr12	12q21.31	Deafness autosomal recessive 84B	614944
P2RY12	chr3	3q24-q25	Bleeding disorder platelet-type 8	609821
P3H2	chr3	3q29	Myopia high with cataract and vitreoretinal degeneration	614292
PADI6	chr1	1p36.13	Preimplantation embryonic lethality 2	617234
PAH	chr12	12q24.1	Hyperphenylalaninemia non-PKU mild	261600
PAH	chr12	12q24.1	Phenylketonuria	261600
PAK3	chrX	Xq23	Mental retardation X-linked 30/47	300558
PAM16	chr16	16p13.3	Spondylometaphyseal dysplasia Megarbane-Dagher-Melike type	613320
PANK2	chr20	20p13-p12.3	HARP syndrome	607236
PANK2	chr20	20p13-p12.3	Neurodegeneration with brain iron accumulation 1	234200
PAPSS2	chr10	10q22-q24	Brachyolmia 4 with mild epiphyseal and metaphyseal changes	612847
PARK7	chr1	1p36	Parkinson disease 7 autosomal recessive early-onset	606324
PARN	chr16	16p13.12	Dyskeratosis congenita autosomal recessive 6	616353
PARS2	chr1	1p32.2	Epileptic encephalopathy early infantile 75	618437

PAX1	chr20	20p11.2	Otofaciocervical syndrome 2	615560
PAX3	chr2	2q36.1	Waardenburg syndrome type 3	148820
PAX4	chr7	7q32	Diabetes mellitus ketosis-prone susceptibility to	612227
PAX7	chr1	1p36.2-p36.12	Myopathy congenital progressive with scoliosis	618578
PC	chr11	11q13.4-q13.5	Pyruvate carboxylase deficiency	266150
PCBD1	chr10	10q22	Hyperphenylalaninemia BH4-deficient D	264070
PCCA	chr13	13q32	Propionicacidemia	606054
PCCB	chr3	3q21-q22	Propionicacidemia	606054
PCDH15	chr10	10q21-q22	Usher syndrome type 1D/F digenic	601067
PCDH15	chr10	10q21-q22	Usher syndrome type 1F	602083
PCDH15	chr10	10q21-q22	Deafness autosomal recessive 23	609533
PCK1	chr20	20q13.31	Phosphoenolpyruvate carboxykinase deficiency cytosolic	261680
PCK2	chr14	14q11.2-q12	PEPCK deficiency mitochondrial	261650
PCLO	chr7	7q11.23-q21.1	Pontocerebellar hypoplasia type 3	608027
PCNA	chr20	20p12	Ataxia-telangiectasia-like disorder 2	615919
PCNT	chr21	21q22.3	Microcephalic osteodysplastic primordial dwarfism type II	210720
PCSK1	chr5	5q15-q21	Obesity with impaired prohormone processing	600955
PCYT1A	chr3	3q29	Spondylometaphyseal dysplasia with cone-rod dystrophy	608940
PDE10A	chr6	6q27	Dyskinesia limb and orofacial infantile-onset	616921
PDE6B	chr4	4p16.3	Retinitis pigmentosa-40	613801
PDE6C	chr10	10q24	Cone dystrophy 4	613093
PDE6D	chr2	2q36-q37	Joubert syndrome 22	615665
PDE6G	chr17	17q25	Retinitis pigmentosa 57	613582
PDE6H	chr12	12p13	Retinal cone dystrophy 3	610024
PDE6H	chr12	12p13	Achromatopsia 6	610024
PDHX	chr11	11p13	Lacticacidemia due to PDX1 deficiency	245349
PDP1	chr8	8q22.1	Pyruvate dehydrogenase phosphatase deficiency	608782
PDSS1	chr10	10p12.1	Coenzyme Q10 deficiency primary 2	614651
PDSS2	chr6	6q21	Coenzyme Q10 deficiency primary 3	614652
PDX1	chr13	13q12.1	Pancreatic agenesis 1	260370
PDXK	chr21	21q22.3	Neuropathy hereditary motor and sensory type VIC with optic atrophy	618511
PDZD7	chr10	10q24.3	Usher syndrome type IIC GPR98/PDZD7 digenic	605472
PDZD7	chr10	10q24.3	Deafness autosomal recessive 57	618003
PDZD7	chr10	10q24.3	Retinal disease in Usher syndrome type IIA modifier of	276901
PEPD	chr19	19cen-q13.11	Prolidase deficiency	170100
PET100	chr19	19p13.2	Mitochondrial complex IV deficiency	220110
PEX1	chr7	7q21-q22	Heimler syndrome 1	234580
PEX1	chr7	7q21-q22	Peroxisome biogenesis disorder 1B (NALD/IRD)	601539
PEX1	chr7	7q21-q22	Peroxisome biogenesis disorder 1A (Zellweger)	214100
PEX10	chr1	1p36.32	Peroxisome biogenesis disorder 6B	614871
PEX10	chr1	1p36.32	Peroxisome biogenesis disorder 6A (Zellweger)	614870
PEX11B	chr1	1q21.1	Peroxisome biogenesis disorder 14B	614920

PEX12	chr17	17q12	Peroxisome biogenesis disorder 3A (Zellweger)	614859
PEX12	chr17	17q12	Peroxisome biogenesis disorder 3B	266510
PEX13	chr2	2p15	Peroxisome biogenesis disorder 11A (Zellweger)	614883
PEX13	chr2	2p15	Peroxisome biogenesis disorder 11B	614885
PEX14	chr1	1p36.2	Peroxisome biogenesis disorder 13A (Zellweger)	614887
PEX16	chr11	11p12-p11.2	Peroxisome biogenesis disorder 8A (Zellweger)	614876
PEX16	chr11	11p12-p11.2	Peroxisome biogenesis disorder 8B	614877
PEX19	chr1	1q22	Peroxisome biogenesis disorder 12A (Zellweger)	614886
PEX2	chr8	8q21.1	Peroxisome biogenesis disorder 5A (Zellweger)	614866
PEX2	chr8	8q21.1	Peroxisome biogenesis disorder 5B	614867
PEX26	chr22	22q11.21	Peroxisome biogenesis disorder 7A (Zellweger)	614872
PEX26	chr22	22q11.21	Peroxisome biogenesis disorder 7B	614873
PEX3	chr6	6q24.2	Peroxisome biogenesis disorder 10A (Zellweger)	614882
PEX3	chr6	6q24.2	Peroxisome biogenesis disorder 10B	617370
PEX5	chr12	12p13.3	Peroxisome biogenesis disorder 2B	202370
PEX5	chr12	12p13.3	Rhizomelic chondrodysplasia punctata type 5	616716
PEX5	chr12	12p13.3	Peroxisome biogenesis disorder 2A (Zellweger)	214110
PEX6	chr6	6p21.1	Peroxisome biogenesis disorder 4B	614863
PEX6	chr6	6p21.1	Heimler syndrome 2	616617
PEX6	chr6	6p21.1	Peroxisome biogenesis disorder 4A (Zellweger)	614862
PEX7	chr6	6q23.3	Peroxisome biogenesis disorder 9B	614879
PEX7	chr6	6q23.3	Rhizomelic chondrodysplasia punctata type 1	215100
PFKM	chr12	12q13.3	Glycogen storage disease VII	232800
PGAM2	chr7	7p13-p12.3	Glycogen storage disease X	261670
PGAP1	chr2	2q33.1	Mental retardation autosomal recessive 42	615802
PGAP2	chr11	11p15.5	Hyperphosphatasia with mental retardation syndrome 3	614207
PGAP3	chr17	17q12	Hyperphosphatasia with mental retardation syndrome 4	615716
PGK1	chrX	Xq13	Phosphoglycerate kinase 1 deficiency	300653
PGM1	chr1	1p31	Congenital disorder of glycosylation type It	614921
PGM3	chr6	6q12	Immunodeficiency 23	615816
PGR	chr11	11q22	Progesterone resistance	264080
PHC1	chr12	12p13.31	Microcephaly 11 primary autosomal recessive	615414
PHF6	chrX	Xq26.3	Borjeson-Forssman-Lehmann syndrome	301900
PHF8	chrX	Xp11.2	Mental retardation syndrome X-linked Siderius type	300263
PHGDH	chr1	1p12	Neu-Laxova syndrome 1	256520
PHGDH	chr1	1p12	Phosphoglycerate dehydrogenase deficiency	601815
PHKA1	chrX	Xq13	Muscle glycogenosis	300559
PHKA2	chrX	Xp22.2-p22.1	Glycogen storage disease type IXa2	306000
PHKA2	chrX	Xp22.2-p22.1	Glycogen storage disease type IXa1	306000
PHKB	chr16	16q12-q13	Phosphorylase kinase deficiency of liver and muscle autosomal recessive	261750
PHKG2	chr16	16p12.1-p11.2	Glycogen storage disease IXc	613027
PHOX2A	chr11	11q13.3-q13.4	Fibrosis of extraocular muscles congenital 2	602078
PHYH	chr10	10pter-p11.2	Refsum disease	266500

PI4KA	chr22	22q11.21	Polymicrogyria perisylvian with cerebellar hypoplasia and arthrogryposis	616531
PIBF1	chr13	13q21-q22	Joubert syndrome 33	617767
PIEZO1	chr16	16q24.3	Lymphatic malformation 6	616843
PIEZO2	chr18	18p11.22	Arthrogryposis distal with impaired proprioception and touch	617146
PIGA	chrX	Xp22.1	Multiple congenital anomalies-hypotonia-seizures syndrome 2	300868
PIGL	chr17	17p12-p11.2	CHIME syndrome	280000
PIGM	chr1	1q23.2	Glycosylphosphatidylinositol deficiency	610293
PIGN	chr18	18q21.33	Multiple congenital anomalies-hypotonia-seizures syndrome 1	614080
PIGO	chr9	9p13	Hyperphosphatasia with mental retardation syndrome 2	614749
PIGQ	chr16	16p13.3	Epileptic encephalopathy early infantile 77	618548
PIGT	chr20	20q13.12	Multiple congenital anomalies-hypotonia-seizures syndrome 3	615398
PIGV	chr1	1p36.11	Hyperphosphatasia with mental retardation syndrome 1	239300
PIGW	chr17	17q12	Glycosylphosphatidylinositol biosynthesis defect 11	616025
PIK3R1	chr5	5q13	Agammaglobulinemia 7 autosomal recessive	615214
PIK3R5	chr17	17p13.1	Ataxia-oculomotor apraxia 3	615217
PINK1	chr1	1p36	Parkinson disease 6 early onset	605909
PIP5K1C	chr19	19p13.3	Lethal congenital contractural syndrome 3	611369
PITX3	chr10	10q25	Cataract 11 syndromic autosomal recessive	610623
PITX3	chr10	10q25	Cataract 11 multiple types	610623
PJVK	chr2	2q31.2	Deafness autosomal recessive 59	610220
PKD1L1	chr7	7p13-p12	Heterotaxy visceral 8 autosomal	617205
PKHD1	chr6	6p12.3-p12.2	Polycystic kidney disease 4 with or without hepatic disease	263200
PKLR	chr1	1q21	Pyruvate kinase deficiency	266200
PKP1	chr1	1q32	Ectodermal dysplasia/skin fragility syndrome	604536
PLA2G4A	chr1	1q25	Gastrointestinal ulceration recurrent with dysfunctional platelets	618372
PLA2G5	chr1	1p36-p34	Fleck retina familial benign	228980
PLA2G6	chr22	22q13.1	Infantile neuroaxonal dystrophy 1	256600
PLA2G6	chr22	22q13.1	Parkinson disease 14 autosomal recessive	612953
PLA2G6	chr22	22q13.1	Neurodegeneration with brain iron accumulation 2B	610217
PLA2G7	chr6	6p21.2-p12	Platelet-activating factor acetylhydrolase deficiency	614278
PLCB1	chr20	20p12	Epileptic encephalopathy early infantile 12	613722
PLCB4	chr20	20p12	Auriculocondylar syndrome 2	614669
PLCD1	chr3	3p22-p21.3	Nail disorder nonsyndromic congenital 3 (leukonychia)	151600
PLCE1	chr10	10q23	Nephrotic syndrome type 3	610725
PLCZ1	chr12	12p12.3	Spermatogenic failure 17	617214
PLEC	chr8	8q24	Muscular dystrophy limb-girdle autosomal recessive 17	613723
PLEC	chr8	8q24	Epidermolysis bullosa simplex with pyloric atresia	612138

PLEC	chr8	8q24	Epidermolysis bullosa simplex with muscular dystrophy	226670
PLEC	chr8	8q24	Epidermolysis bullosa simplex with nail dystrophy	616487
PLEKHG5	chr1	1p36	Spinal muscular atrophy distal autosomal recessive 4	611067
PLEKHG5	chr1	1p36	Charcot-Marie-Tooth disease recessive intermediate C	615376
PLEKHM1	chr17	17q21.3	Osteopetrosis autosomal recessive 6	611497
PLG	chr6	6q26	Dysplasminogenemia	217090
PLG	chr6	6q26	Plasminogen deficiency type I	217090
PLK4	chr4	4q27-q28	Microcephaly and chorioretinopathy autosomal recessive 2	616171
PLOD1	chr1	1p36.3-p36.2	Ehlers-Danlos syndrome kyphoscoliotic type 1	225400
PLOD2	chr3	3q23-q24	Bruck syndrome 2	609220
PLOD3	chr7	7q22	Lysyl hydroxylase 3 deficiency	612394
PLP1	chrX	Xq22	Pelizaeus-Merzbacher disease	312080
PLP1	chrX	Xq22	Spastic paraplegia 2 X-linked	312920
PMM2	chr16	16p13.3-p13.2	Congenital disorder of glycosylation type Ia	212065
PMP22	chr17	17p11.2	Dejerine-Sottas disease	145900
PMPCA	chr9	9q34.3	Spinocerebellar ataxia autosomal recessive 2	213200
PMS2	chr7	7p22	Mismatch repair cancer syndrome	276300
PNKP	chr19	19q13.4	Microcephaly seizures and developmental delay	613402
PNKP	chr19	19q13.4	Ataxia-oculomotor apraxia 4	616267
PNKP	chr19	19q13.4	Charcot-Marie-Tooth disease type 2B2	605589
PNLIP	chr10	10q26.1	Pancreatic lipase deficiency	614338
PNP	chr14	14q13.1	Immunodeficiency due to purine nucleoside phosphorylase deficiency	613179
PNPLA1	chr6	6p21.31	Ichthyosis congenital autosomal recessive 10	615024
PNPLA2	chr11	11p15.5	Neutral lipid storage disease with myopathy	610717
PNPLA6	chr19	19p13.3	Spastic paraplegia 39 autosomal recessive	612020
PNPLA6	chr19	19p13.3	Boucher-Neuhauser syndrome	215470
PNPLA6	chr19	19p13.3	Oliver-McFarlane syndrome	275400
PNPLA6	chr19	19p13.3	Laurence-Moon syndrome	245800
PNPO	chr17	17q21.32	Pyridoxamine 5'-phosphate oxidase deficiency	610090
PNPT1	chr2	2p16.1	Deafness autosomal recessive 70	614934
PNPT1	chr2	2p16.1	Combined oxidative phosphorylation deficiency 13	614932
POC1A	chr3	3p21.2	Short stature onychodysplasia facial dysmorphism and hypotrichosis	614813
POC1B	chr12	12q21.33	Cone-rod dystrophy 20	615973
POF1B	chrX	Xq21	Premature ovarian failure 2B	300604
POGLUT1	chr3	3q13.33	Muscular dystrophy limb-girdle autosomal recessive 21	617232
POLA1	chrX	Xp22.3-p21.1	Pigmentary disorder reticulate with systemic manifestations X-linked	301220
POLA1	chrX	Xp22.3-p21.1	Van Esch-O'Driscoll syndrome	301030
POLE	chr12	12q24.3	FILS syndrome	615139
POLE	chr12	12q24.3	IMAGE-I syndrome	618336
POLG	chr15	15q25	Mitochondrial DNA depletion syndrome 4B (MNGIE type)	613662

POLG	chr15	15q25	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	607459
POLG	chr15	15q25	Mitochondrial DNA depletion syndrome 4A (Alpers type)	203700
POLG	chr15	15q25	Progressive external ophthalmoplegia autosomal recessive 1	258450
POLG2	chr17	17q23-q24	Mitochondrial DNA depletion syndrome 16 (hepatic type)	618528
POLH	chr6	6p21.1-p12	Xeroderma pigmentosum variant type	278750
POLR1C	chr6	6p22.3	Treacher Collins syndrome 3	248390
POLR1C	chr6	6p22.3	Leukodystrophy hypomyelinating 11	616494
POLR1D	chr13	13q12.2	Treacher Collins syndrome 2	613717
POLR3A	chr10	10q22.3	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism	607694
POLR3A	chr10	10q22.3	Wiedemann-Rautenstrauch syndrome	264090
POLR3B	chr12	12q23.3	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism	614381
POMC	chr2	2p23.3	Obesity early-onset susceptibility to	601665
POMC	chr2	2p23.3	Obesity adrenal insufficiency and red hair due to POMC deficiency	609734
POMGNT1	chr1	1p34-p33	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 3	613151
POMGNT1	chr1	1p34-p33	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 3	613157
POMGNT1	chr1	1p34-p33	Retinitis pigmentosa 76	617123
POMGNT1	chr1	1p34-p33	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 3	253280
POMGNT2	chr3	3p22.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 8	614830
POMGNT2	chr3	3p22.1	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 8	618135
POMK	chr8	8p11	?Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 12	616094
POMK	chr8	8p11	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 12	615249
POMP	chr13	13q12.3	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	601952
POMT1	chr9	9q34.1	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 1	609308
POMT1	chr9	9q34.1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 1	236670
POMT1	chr9	9q34.1	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 1	613155
POMT2	chr14	14q24.3	Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 2	613158
POMT2	chr14	14q24.3	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 2	613150

POMT2	chr14	14q24.3	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 2	613156
POP1	chr8	8q22	Anauxetic dysplasia 2	617396
POR	chr7	7q11.2	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	201750
POU1F1	chr3	3p11	Pituitary hormone deficiency combined 1	613038
POU3F4	chrX	Xq21.1	Deafness X-linked 2	304400
PPARG	chr3	3p25	Obesity severe	601665
PPIB	chr15	15q21-q22	Osteogenesis imperfecta type IX	259440
PPP1R15B	chr1	1q32.1	Microcephaly short stature and impaired glucose metabolism 2	616817
PPT1	chr1	1p32	Ceroid lipofuscinosis neuronal 1	256730
PQBP1	chrX	Xp11.23	Renpenning syndrome	309500
PRDM5	chr4	4q27	Brittle cornea syndrome 2	614170
PREPL	chr2	2p21	Myasthenic syndrome congenital 22	616224
PRF1	chr10	10q22	Hemophagocytic lymphohistiocytosis familial 2	603553
PRG4	chr1	1q24-q25	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome	208250
PRICKLE1	chr12	12q12	Epilepsy progressive myoclonic 1B	612437
PRKACG	chr9	9q21.11	Bleeding disorder platelet-type 19	616176
PRKCD	chr3	3p	Autoimmune lymphoproliferative syndrome type III	615559
PRKDC	chr8	8q11	Immunodeficiency 26 with or without neurologic abnormalities	615966
PRKN	chr6	6q25.2-q27	Parkinson disease juvenile type 2	600116
PRKRA	chr2	2q31.3	Dystonia 16	612067
PRLR	chr5	5p13.2	Hyperprolactinemia	615555
PRMT7	chr16	16q22.1	Short stature brachydactyly intellectual developmental disability and seizures	617157
PROC	chr2	2q13-q14	Thrombophilia due to protein C deficiency autosomal recessive	612304
PRODH	chr22	22q11.2	Hyperprolinemia type I	239500
PROM1	chr4	4p15.3	Retinitis pigmentosa 41	612095
PROM1	chr4	4p15.3	Cone-rod dystrophy 12	612657
PROP1	chr5	5q	Pituitary hormone deficiency combined 2	262600
PROS1	chr3	3q11.2	Thrombophilia due to protein S deficiency autosomal recessive	614514
PRPH	chr12	12q12-q13	Amyotrophic lateral sclerosis susceptibility to	105400
PRPH2	chr6	6p21.1-cen	Retinitis punctata albescens	136880
PRPH2	chr6	6p21.1-cen	Retinitis pigmentosa 7 and digenic form	608133
PRPH2	chr6	6p21.1-cen	Leber congenital amaurosis 18	608133
PRPS1	chrX	Xq22-q24	Charcot-Marie-Tooth disease X-linked recessive 5	311070
PRPS1	chrX	Xq22-q24	Phosphoribosylpyrophosphate synthetase superactivity	300661
PRPS1	chrX	Xq22-q24	Deafness X-linked 1	304500
PRPS1	chrX	Xq22-q24	Arts syndrome	301835
PRPS1	chrX	Xq22-q24	Gout PRPS-related	300661
PRRX1	chr1	1q24	Agnathia-otocephaly complex	202650
PRSS12	chr4	4q25-q26	Mental retardation autosomal recessive 1	249500
PRSS56	chr2	2q37.1	Microphthalmia isolated 6	613517

PRX	chr19	19q13.1-q13.2	Charcot-Marie-Tooth disease type 4F	614895
PRX	chr19	19q13.1-q13.2	Dejerine-Sottas disease	145900
PSAP	chr10	10q22.1	Krabbe disease atypical	611722
PSAP	chr10	10q22.1	Combined SAP deficiency	611721
PSAP	chr10	10q22.1	Metachromatic leukodystrophy due to SAP-b deficiency	249900
PSAT1	chr9	9q21.31	Neu-Laxova syndrome 2	616038
PSAT1	chr9	9q21.31	Phosphoserine aminotransferase deficiency	610992
PSMB8	chr6	6p21.3	Proteasome-associated autoinflammatory syndrome 1 and digenic forms	256040
PSMB9	chr6	6p21.3	Proteasome-associated autoinflammatory syndrome 3 digenic	617591
PSMC3IP	chr17	17q21.2	Ovarian dysgenesis 3	614324
PSPH	chr7	7p11.2	Phosphoserine phosphatase deficiency	614023
PTCHD1	chrX	Xp22.11	Autism susceptibility to X-linked 4	300830
PTF1A	chr10	10p12.3	Pancreatic and cerebellar agenesis	609069
PTF1A	chr10	10p12.3	Pancreatic agenesis 2	615935
PTGER2	chr14	14q22	Asthma aspirin-induced susceptibility to	208550
PTH	chr11	11p15.3-p15.1	Hypoparathyroidism familial isolated 1	146200
PTH1R	chr3	3p22-p21.1	Eiken syndrome	600002
PTH1R	chr3	3p22-p21.1	Chondrodysplasia Blomstrand type	215045
PTPN14	chr1	1q32	Choanal atresia and lymphedema	613611
PTPN22	chr1	1p13	Diabetes type 1 susceptibility to	222100
PTPN23	chr3	3p21.3	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	618890
PTPRC	chr1	1q31-q32	Severe combined immunodeficiency T cell-negative B-cell/natural killer-cell positive	608971
PTPRF	chr1	1p32	Breasts and/or nipples aplasia or hypoplasia of 2	616001
PTPRO	chr12	12p13-p12	Nephrotic syndrome type 6	614196
PTPRQ	chr12	12q21.2	Deafness autosomal recessive 84A	613391
PTRH2	chr17	17q23.1	Infantile-onset multisystem neurologic endocrine and pancreatic disease	616263
PTS	chr11	11q22.3-q23.3	Hyperphenylalaninemia BH4-deficient A	261640
PUS1	chr12	12q24.33	Myopathy lactic acidosis and sideroblastic anemia 1	600462
PXDN	chr2	2p25.3	Anterior segment dysgenesis 7 with sclerocornea	269400
PYCR1	chr17	17q25.3	Cutis laxa autosomal recessive type IIB	612940
PYCR2	chr1	1q42.13	Leukodystrophy hypomyelinating 10	616420
PYGL	chr14	14q22.1	Glycogen storage disease VI	232700
PYGM	chr11	11q13	McArdle disease	232600
QDPR	chr4	4p15.31	Hyperphenylalaninemia BH4-deficient C	261630
RAB18	chr10	10p12.1	Warburg micro syndrome 3	614222
RAB23	chr6	6p11	Carpenter syndrome	201000
RAB27A	chr15	15q21	Griscelli syndrome type 2	607624
RAB28	chr4	4p15.33	Cone-rod dystrophy 18	615374
RAB33B	chr4	4q31.1	Smith-McCort dysplasia 2	615222

RAB39B	chrX	Xq28	Waisman syndrome	311510
RAB39B	chrX	Xq28	Mental retardation X-linked 72	300271
RAB3GAP1	chr2	2q21.3	Warburg micro syndrome 1	600118
RAB3GAP2	chr1	1q41	Warburg micro syndrome 2	614225
RAB3GAP2	chr1	1q41	Martsolf syndrome	212720
RAD21	chr8	8q24	Mungan syndrome	611376
RAD51C	chr17	17q22	Fanconi anemia complementation group O	613390
RAG1	chr11	11p13	Omenn syndrome	603554
RAG1	chr11	11p13	Severe combined immunodeficiency B cell-negative	601457
RAG1	chr11	11p13	Combined cellular and humoral immune defects with granulomas	233650
RAG2	chr11	11p13	Severe combined immunodeficiency B cell-negative	601457
RAG2	chr11	11p13	Combined cellular and humoral immune defects with granulomas	233650
RAG2	chr11	11p13	Omenn syndrome	603554
RALGAPA1	chr14	14q13.2	Neurodevelopmental disorder with hypotonia neonatal respiratory insufficiency and thermodysregulation	618797
RAPSN	chr11	11p11.2-p11.1	Fetal akinesia deformation sequence 2	618388
RAPSN	chr11	11p11.2-p11.1	Myasthenic syndrome congenital 11 associated with acetylcholine receptor deficiency	616326
RARB	chr3	3p24	Microphthalmia syndromic 12	615524
RARS2	chr6	6q16.1	Pontocerebellar hypoplasia type 6	611523
RASGRP1	chr15	15q15	Immunodeficiency 64	618534
RASGRP2	chr11	11q13	Bleeding disorder platelet-type 18	615888
RAX	chr18	18q21.3	Microphthalmia isolated 3	611038
RBBP8	chr18	18q11.2	Jawad syndrome	251255
RBBP8	chr18	18q11.2	Seckel syndrome 2	606744
RBCK1	chr20	20p13	Polyglucosan body myopathy 1 with or without immunodeficiency	615895
RBM10	chrX	Xp11.23	TARP syndrome	311900
RBM28	chr7	7q32.1	Alopecia neurologic defects and endocrinopathy syndrome	612079
RBM8A	chr1	1q12	Thrombocytopenia-absent radius syndrome	274000
RBP3	chr10	10q11.2	Retinitis pigmentosa 66	615233
RBP4	chr10	10q24	Retinal dystrophy iris coloboma and comedogenic acne syndrome	615147
RCBTB1	chr13	13q14	Retinal dystrophy with or without extraocular anomalies	617175
RD3	chr1	1q32.3	Leber congenital amaurosis 12	610612
RDH11	chr14	14q23.3	Retinal dystrophy juvenile cataracts and short stature syndrome	616108
RDH12	chr14	14q23.3	Leber congenital amaurosis 13	612712
RDH5	chr12	12q13-q14	Fundus albipunctatus	136880
RDX	chr11	11q23	Deafness autosomal recessive 24	611022
RECQL4	chr8	8q24.3	RAPADILINO syndrome	266280
RECQL4	chr8	8q24.3	Baller-Gerold syndrome	218600
RECQL4	chr8	8q24.3	Rothmund-Thomson syndrome type 2	268400
REEP2	chr5	5q31	Spastic paraplegia 72 autosomal dominant	615625

REEP2	chr5	5q31	Spastic paraplegia 72 autosomal recessive	615625
RELN	chr7	7q22	Lissencephaly 2 (Norman-Roberts type)	257320
REN	chr1	1q32	Renal tubular dysgenesis	267430
RETREG1	chr5	5p15.1	Neuropathy hereditary sensory and autonomic type IIB	613115
RFC1	chr4	4p14-p13	Cerebellar ataxia neuropathy and vestibular areflexia syndrome	614575
RFT1	chr3	3p21.1	Congenital disorder of glycosylation type In	612015
RFX5	chr1	1q21.1-q21.3	Bare lymphocyte syndrome type II complementation group C	209920
RFX5	chr1	1q21.1-q21.3	Bare lymphocyte syndrome type II complementation group E	209920
RFX6	chr6	6q22.2	Mitchell-Riley syndrome	615710
RFXANK	chr19	19p12	MHC class II deficiency complementation group B	209920
RFXAP	chr13	13q14	Bare lymphocyte syndrome type II complementation group D	209920
RHO	chr3	3q21-q24	Retinitis punctata albescens	136880
RHO	chr3	3q21-q24	Retinitis pigmentosa 4 autosomal dominant or recessive	613731
RHOH	chr4	4p13	Epidermodysplasia verruciformis susceptibility to 4	618307
RIC1	chr9	9p24.1	CATIFA syndrome	618761
RIMS2	chr8	8q22.3	Cone-rod synaptic disorder syndrome congenital nonprogressive	618970
RIN2	chr20	20p11.22	Macrocephaly alopecia cutis laxa and scoliosis	613075
RINT1	chr7	7q22.3	Infantile liver failure syndrome 3	618641
RIPK4	chr21	21q22.3	Popliteal pterygium syndrome Bartsocas-Papas type	263650
RIPK4	chr21	21q22.3	CHAND syndrome	214350
RIPOR2	chr6	6p22.3-p21.32	Deafness autosomal recessive 104	616515
RLBP1	chr15	15q26	Retinitis punctata albescens	136880
RLBP1	chr15	15q26	Bothnia retinal dystrophy	607475
RLBP1	chr15	15q26	Fundus albipunctatus	136880
RMND1	chr6	6q25	Combined oxidative phosphorylation deficiency 11	614922
RMRP	chr9	9p21-p12	Anauxetic dysplasia 1	607095
RMRP	chr9	9p21-p12	Cartilage-hair hypoplasia	250250
RMRP	chr9	9p21-p12	Metaphyseal dysplasia without hypotrichosis	250460
RNASEH2A	chr19	19p13.13	Aicardi-Goutieres syndrome 4	610333
RNASEH2B	chr13	13q14.1	Aicardi-Goutieres syndrome 2	610181
RNASEH2C	chr11	11q13.2	Aicardi-Goutieres syndrome 3	610329
RNASET2	chr6	6q27	Leukoencephalopathy cystic without megalencephaly	612951
RNF168	chr3	3q29	RIDDLE syndrome	611943
RNF216	chr7	7p22	Cerebellar ataxia and hypogonadotropic hypogonadism	212840
RNU4ATAC	chr2	2q14.2	Microcephalic osteodysplastic primordial dwarfism type I	210710
RNU4ATAC	chr2	2q14.2	Roifman syndrome	616651
ROBO3	chr11	11q23-q25	Gaze palsy familial horizontal with progressive scoliosis 1	607313

ROGDI	chr16	16p13.3	Kohlschutter-Tonz syndrome	226750
ROM1	chr11	11q13	Retinitis pigmentosa 7 digenic form	608133
ROR2	chr9	9q22	Brachydactyly type B1	113000
ROR2	chr9	9q22	Robinow syndrome autosomal recessive	268310
RORC	chr1	1q21	Immunodeficiency 42	616622
RP1	chr8	8q11-q13	Retinitis pigmentosa 1	180100
RP1L1	chr8	8p23	Retinitis pigmentosa 88	618826
RP1L1	chr8	8p23	Occult macular dystrophy	613587
RPE65	chr1	1p31	Leber congenital amaurosis 2	204100
RPE65	chr1	1p31	Retinitis pigmentosa 87 with choroidal involvement	618697
RPE65	chr1	1p31	Retinitis pigmentosa 20	613794
RPGR	chrX	Xp11.4	Cone-rod dystrophy X-linked 1	304020
RPGR	chrX	Xp11.4	Macular degeneration X-linked atrophic	300834
RPGRIP1	chr14	14q11	Leber congenital amaurosis 6	613826
RPGRIP1L	chr16	16q12.2	COACH syndrome	216360
RPGRIP1L	chr16	16q12.2	Meckel syndrome 5	611561
RPGRIP1L	chr16	16q12.2	Joubert syndrome 7	611560
RPIA	chr2	2p11.2	Ribose 5-phosphate isomerase deficiency	608611
RPL10	chrX	Xq28	Mental retardation X-linked syndromic 35	300998
RRM2B	chr8	8q23.1	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)	612075
RRM2B	chr8	8q23.1	Mitochondrial DNA depletion syndrome 8B (MNGIE type)	612075
RS1	chrX	Xp22.2-p22.1	Retinoschisis	312700
RSPH1	chr21	21q22.3	Ciliary dyskinesia primary 24	615481
RSPO1	chr1	1p34.3	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal	610644
RSPO1	chr1	1p34.3	Palmoplantar hyperkeratosis and true hermaphroditism	610644
RSPO4	chr20	20p13	Anonychia congenita	206800
RSRC1	chr3	3q25.32	Intellectual developmental disorder autosomal recessive 70	618402
RTEL1	chr20	20q13.3	Dyskeratosis congenita autosomal recessive 5	615190
RTEL1	chr20	20q13.3	Dyskeratosis congenita autosomal dominant 4	615190
RTTN	chr18	18q22.2	Microcephaly short stature and polymicrogyria with seizures	614833
RUBCN	chr3	3q29	Spinocerebellar ataxia autosomal recessive 15	615705
RYR1	chr19	19q13.1	Central core disease	117000
RYR1	chr19	19q13.1	Minicore myopathy with external ophthalmoplegia	255320
RYR1	chr19	19q13.1	Neuromuscular disease congenital with uniform type 1 fiber	117000
SACS	chr13	13q12	Spastic ataxia Charlevoix-Saguenay type	270550
SAG	chr2	2q37.1	Oguchi disease-1	258100
SALL2	chr14	14q11.1-q12	Coloboma ocular autosomal recessive	216820
SAMD9	chr7	7q21	Tumoral calcinosis familial normophosphatemic	610455
SAMHD1	chr20	20q11.2	Aicardi-Goutieres syndrome 5	612952
SAR1B	chr5	5q31.1	Chylomicron retention disease	246700
SARDH	chr9	9q34.2	Sarcosinemia	268900

SARS2	chr19	19q13.2	Hyperuricemia pulmonary hypertension renal failure and alkalosis	613845
SASH1	chr6	6q24.3	Cancer alopecia pigment dyscrasia onychodystrophy and keratoderma	618373
SASS6	chr1	1p21.2	Microcephaly 14 primary autosomal recessive	616402
SBDS	chr7	7q11	Shwachman-Diamond syndrome	260400
SBF1	chr22	22q13.33	Charcot-Marie-Tooth disease type 4B3	615284
SBF2	chr11	11p15	Charcot-Marie-Tooth disease type 4B2	604563
SC5D	chr11	11q23.3	Lathosterolosis	607330
SCAPER	chr15	15q24	Intellectual developmental disorder and retinitis pigmentosa	618195
SCARB2	chr4	4q13-q21	Epilepsy progressive myoclonic 4 with or without renal failure	254900
SCARF2	chr22	22q11	Van den Ende-Gupta syndrome	600920
SCN1B	chr19	19q13.1	Epileptic encephalopathy early infantile 52	617350
SCN4A	chr17	17q23.1-q25.3	Myasthenic syndrome congenital 16	614198
SCN5A	chr3	3p21	Sick sinus syndrome 1	608567
SCN5A	chr3	3p21	Sudden infant death syndrome susceptibility to	272120
SCN9A	chr2	2q24	Neuropathy hereditary sensory and autonomic type IID	243000
SCN9A	chr2	2q24	Insensitivity to pain congenital	243000
SCNN1A	chr12	12p13	Pseudohypoaldosteronism type I	264350
SCNN1B	chr16	16p13-p12	Pseudohypoaldosteronism type I	264350
SCNN1G	chr16	16p13-p12	Pseudohypoaldosteronism type I	264350
SCO1	chr17	17p13-p12	Mitochondrial complex IV deficiency	220110
SCO2	chr22	22q13	Cardioencephalomyopathy fatal infantile due to cytochrome c oxidase deficiency 1	604377
SCP2	chr1	1p32	Leukoencephalopathy with dystonia and motor neuropathy	613724
SDC3	chr1	1pter-p22.3	Obesity association with	601665
SDCCAG8	chr1	1q43-q44	Bardet-Biedl syndrome 16	615993
SDHA	chr5	5p15	Cardiomyopathy dilated 1GG	613642
SDHA	chr5	5p15	Leigh syndrome	256000
SDHA	chr5	5p15	Mitochondrial respiratory chain complex II deficiency	252011
SDHAF1	chr19	19q12-q13.2	Mitochondrial complex II deficiency	252011
SDHD	chr11	11q23	Mitochondrial complex II deficiency	252011
SEC23A	chr14	14q21.1	Craniolenticulosutural dysplasia	607812
SEC23B	chr20	20p11.2	Dyserythropoietic anemia congenital type II	224100
SEC24D	chr4	4q26	Cole-Carpenter syndrome 2	616294
SECISBP2	chr9	9q22.2	Thyroid hormone metabolism abnormal	609698
SELENON	chr1	1p36-p35	Muscular dystrophy rigid spine 1	602771
SELENON	chr1	1p36-p35	Myopathy congenital with fiber-type disproportion	255310
SEMA4A	chr1	1q22	Cone-rod dystrophy 10	610283
SEMA4A	chr1	1q22	Retinitis pigmentosa 35	610282
SEPSECS	chr4	4p15.2	Pontocerebellar hypoplasia type 2D	613811
SERAC1	chr6	6q25.3	3-methylglutaconic aciduria with deafness encephalopathy and Leigh-like syndrome	614739

SERPINA1	chr14	14q32.1	Hemorrhagic diathesis due to antithrombin Pittsburgh	613490
SERPINA1	chr14	14q32.1	Emphysema-cirrhosis due to AAT deficiency	613490
SERPINA1	chr14	14q32.1	Emphysema due to AAT deficiency	613490
SERPINA6	chr14	14q32.1	Corticosteroid-binding globulin deficiency	611489
SERPINB6	chr6	6p25	Deafness autosomal recessive 91	613453
SERPINB7	chr18	18q21.3	Palmoplantar keratoderma Nagashima type	615598
SERPINC1	chr1	1q23-q25	Thrombophilia due to antithrombin III deficiency	613118
SERPINE1	chr7	7q21.3-q22	Plasminogen activator inhibitor-1 deficiency	613329
SERPINF1	chr17	17p13.3	Osteogenesis imperfecta type VI	613982
SERPINF2	chr17	17pter-p12	Alpha-2-plasmin inhibitor deficiency	262850
SERPING1	chr11	11q11-q13.1	Angioedema hereditary types I and II	106100
SERPINH1	chr11	11q13.5	Osteogenesis imperfecta type X	613848
SERPINH1	chr11	11q13.5	Preterm premature rupture of the membranes susceptibility to	610504
SETX	chr9	9q34	Spinocerebellar ataxia autosomal recessive with axonal neuropathy 2	606002
SFTPB	chr2	2p12-p11.2	Surfactant metabolism dysfunction pulmonary 1	265120
SFXN4	chr10	10q26.11	Combined oxidative phosphorylation deficiency 18	615578
SGCA	chr17	17q12-q21.33	Muscular dystrophy limb-girdle autosomal recessive 3	608099
SGCB	chr4	4q12	Muscular dystrophy limb-girdle autosomal recessive 4	604286
SGCD	chr5	5q33	Muscular dystrophy limb-girdle autosomal recessive 6	601287
SGCG	chr13	13q12	Muscular dystrophy limb-girdle autosomal recessive 5	253700
SGO1	chr3	3p24.3	Chronic atrial and intestinal dysrhythmia	616201
SGSH	chr17	17q25.3	Mucopolysaccharidosis type IIIA (Sanfilippo A)	252900
SH2D1A	chrX	Xq25	Lymphoproliferative syndrome X-linked 1	308240
SH3PXD2B	chr5	5q35.1	Frank-ter Haar syndrome	249420
SH3TC2	chr5	5q32	Charcot-Marie-Tooth disease type 4C	601596
SHOX	chrX	Xpter-p22.32	Langer mesomelic dysplasia	249700
SI	chr3	3q25-q26	Sucrase-isomaltase deficiency congenital	222900
SIGMAR1	chr9	9p13	Amyotrophic lateral sclerosis 16 juvenile	614373
SIGMAR1	chr9	9p13	Spinal muscular atrophy distal autosomal recessive 2	605726
SIK3	chr11	11q23.3	Spondyloepimetaphyseal dysplasia Krakow type	618162
SIL1	chr5	5q31	Marinesco-Sjogren syndrome	248800
SIX6	chr14	14q23	Optic disc anomalies with retinal and/or macular dystrophy	212550
SKIV2L	chr6	6p21	Trichohepatoenteric syndrome 2	614602
SLC10A2	chr13	13q33	Bile acid malabsorption primary	613291
SLC11A2	chr12	12q13	Anemia hypochromic microcytic with iron overload 1	206100
SLC12A1	chr15	15q15-q21.1	Bartter syndrome type 1	601678
SLC12A3	chr16	16q13	Gitelman syndrome	263800
SLC12A5	chr20	20q12-q13.1	Epileptic encephalopathy early infantile 34	616645
SLC12A6	chr15	15q13-q14	Agenesis of the corpus callosum with peripheral neuropathy	218000

SLC13A5	chr17	17p13-p12	Epileptic encephalopathy early infantile 25	615905
SLC16A1	chr1	1p13.2-p12	Monocarboxylate transporter 1 deficiency	616095
SLC17A5	chr6	6q14-q15	Sialic acid storage disorder infantile	269920
SLC17A5	chr6	6q14-q15	Salla disease	604369
SLC19A2	chr1	1q23.3	Thiamine-responsive megaloblastic anemia syndrome	249270
SLC19A3	chr2	2q36.3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	607483
SLC1A1	chr9	9p24	Dicarboxylic aminoaciduria	222730
SLC22A12	chr11	11q13	Hypouricemia renal	220150
SLC22A5	chr5	5q31.1	Carnitine deficiency systemic primary	212140
SLC24A1	chr15	15q22	Night blindness congenital stationary (complete) 1D autosomal recessive	613830
SLC24A4	chr14	14q32	Skin/hair/eye pigmentation 6 blue/green eyes	210750
SLC24A4	chr14	14q32	Amelogenesis imperfecta type IIA5	615887
SLC24A4	chr14	14q32	Skin/hair/eye pigmentation 6 blond/brown hair	210750
SLC24A5	chr15	15q21.1	Skin/hair/eye pigmentation 4 fair/dark skin	113750
SLC24A5	chr15	15q21.1	Albinism oculocutaneous type VI	113750
SLC25A1	chr22	22q11	Myasthenic syndrome congenital 23 presynaptic	618197
SLC25A1	chr22	22q11	Combined D-2- and L-2-hydroxyglutaric aciduria	615182
SLC25A12	chr2	2q24	Epileptic encephalopathy early infantile 39	612949
SLC25A13	chr7	7q21.3	Citrullinemia adult-onset type II	603471
SLC25A13	chr7	7q21.3	Citrullinemia type II neonatal-onset	605814
SLC25A15	chr13	13q14	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970
SLC25A19	chr17	17q25.3	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)	613710
SLC25A19	chr17	17q25.3	Microcephaly Amish type	607196
SLC25A20	chr3	3p21.31	Carnitine-acylcarnitine translocase deficiency	212138
SLC25A21	chr14	14q11.2	Mitochondrial DNA depletion syndrome 18	618811
SLC25A22	chr11	11p15.5	Epileptic encephalopathy early infantile 3	609304
SLC25A38	chr3	3p22.1	Anemia sideroblastic 2 pyridoxine-refractory	205950
SLC25A4	chr4	4q35	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR	615418
SLC25A46	chr5	5q22.1	Neuropathy hereditary motor and sensory type VIB	616505
SLC26A1	chr4	4p16.3	Nephrolithiasis calcium oxalate	167030
SLC26A2	chr5	5q32-q33.1	De la Chapelle dysplasia	256050
SLC26A2	chr5	5q32-q33.1	Atelosteogenesis type II	256050
SLC26A2	chr5	5q32-q33.1	Diastrophic dysplasia broad bone-platyspondylic variant	222600
SLC26A2	chr5	5q32-q33.1	Diastrophic dysplasia	222600
SLC26A2	chr5	5q32-q33.1	Achondrogenesis Ib	600972
SLC26A2	chr5	5q32-q33.1	Epiphyseal dysplasia multiple 4	226900
SLC26A3	chr7	7q22-q31.1	Diarrhea 1 secretory chloride congenital	214700
SLC26A4	chr7	7q31	Deafness autosomal recessive 4 with enlarged vestibular aqueduct	600791
SLC26A4	chr7	7q31	Pendred syndrome	274600
SLC26A5	chr7	7q22.1	Deafness autosomal recessive 61	613865

SLC28A1	chr15	15q25-q26	Uridine-cytidineuria	618477
SLC29A3	chr10	10q22.1	Histiocytosis-lymphadenopathy plus syndrome	602782
SLC2A1	chr1	1p34.2	GLUT1 deficiency syndrome 1 infantile onset severe	606777
SLC2A10	chr20	20q13.1	Arterial tortuosity syndrome	208050
SLC2A2	chr3	3q26.1-q26.3	Fanconi-Bickel syndrome	227810
SLC2A9	chr4	4p16-p15.3	Uric acid concentration serum QTL 2	612076
SLC2A9	chr4	4p16-p15.3	Hypouricemia renal 2	612076
SLC30A10	chr1	1q41	Hypermanganesemia with dystonia 1	613280
SLC33A1	chr3	3q25.31	Congenital cataracts hearing loss and neurodegeneration	614482
SLC34A1	chr5	5q35	Hypercalcemia infantile 2	616963
SLC34A1	chr5	5q35	Fanconi renotubular syndrome 2	613388
SLC34A2	chr4	4p15.31-p15.2	Pulmonary alveolar microlithiasis	265100
SLC34A3	chr9	9q34	Hypophosphatemic rickets with hypercalciuria	241530
SLC35A1	chr6	6q15	Congenital disorder of glycosylation type IIf	603585
SLC35A3	chr1	1p21	Arthrogryposis mental retardation and seizures	615553
SLC35C1	chr11	11p11.2	Congenital disorder of glycosylation type IIc	266265
SLC35D1	chr1	1p32-p31	Schneckenbecken dysplasia	269250
SLC36A2	chr5	5q33.1	Iminoglycinuria digenic	242600
SLC36A2	chr5	5q33.1	Hyperglycinuria	138500
SLC37A4	chr11	11q23	Glycogen storage disease Ib	232220
SLC38A8	chr16	16q23.3	Foveal hypoplasia 2 with or without optic nerve misrouting and/or anterior segment dysgenesis	609218
SLC39A13	chr11	11p11.2	Ehlers-Danlos syndrome spondylodysplastic type 3	612350
SLC39A4	chr8	8q24.3	Acrodermatitis enteropathica	201100
SLC3A1	chr2	2p16.3	Cystinuria	220100
SLC45A2	chr5	5p13.3	Skin/hair/eye pigmentation 5 dark/fair skin	227240
SLC45A2	chr5	5p13.3	Skin/hair/eye pigmentation 5 black/nonblack hair	227240
SLC45A2	chr5	5p13.3	Albinism oculocutaneous type IV	606574
SLC45A2	chr5	5p13.3	Skin/hair/eye pigmentation 5 dark/light eyes	227240
SLC46A1	chr17	17q11.1	Folate malabsorption hereditary	229050
SLC4A1	chr17	17q21-q22	Renal tubular acidosis distal AR	611590
SLC4A11	chr20	20p13-p12	Corneal endothelial dystrophy autosomal recessive	217700
SLC4A11	chr20	20p13-p12	Corneal endothelial dystrophy and perceptive deafness	217400
SLC4A4	chr4	4q21	Renal tubular acidosis proximal with ocular abnormalities	604278
SLC52A2	chr8	8q24.3	Brown-Vialetto-Van Laere syndrome 2	614707
SLC52A3	chr20	20p13	Brown-Vialetto-Van Laere syndrome 1	211530
SLC52A3	chr20	20p13	Fazio-Londe disease	211500
SLC5A1	chr22	22q13.1	Glucose/galactose malabsorption	606824
SLC5A2	chr16	16p11.2	Renal glucosuria	233100
SLC5A5	chr19	19p13.2-p12	Thyroid dyshormonogenesis 1	274400
SLC5A7	chr2	2q12.3	Myasthenic syndrome congenital 20 presynaptic	617143
SLC6A17	chr1	1p13.3	Mental retardation autosomal recessive 48	616269

SLC6A19	chr5	5p15.33	Iminoglycinuria digenic	242600
SLC6A19	chr5	5p15.33	Hartnup disorder	234500
SLC6A20	chr3	3p21.3	Iminoglycinuria digenic	242600
SLC6A3	chr5	5p15.3	Parkinsonism-dystonia infantile 1	613135
SLC6A5	chr11	11p15.2-p15.1	Hyperekplexia 3	614618
SLC6A8	chrX	Xq28	Cerebral creatine deficiency syndrome 1	300352
SLC6A9	chr1	1p33	Glycine encephalopathy with normal serum glycine	617301
SLC7A14	chr3	3q26.2	Retinitis pigmentosa 68	615725
SLC7A7	chr14	14q11.2	Lysinuric protein intolerance	222700
SLC7A9	chr19	19q13.1	Cystinuria	220100
SLC9A1	chr1	1p36.1-p35	Lichtenstein-Knorr syndrome	616291
SLC9A3	chr5	5p15.3	Diarrhea 8 secretory sodium congenital	616868
SLCO1B1	chr12	12p12	Hyperbilirubinemia Rotor type digenic	237450
SLCO1B3	chr12	12p12.2	Hyperbilirubinemia Rotor type digenic	237450
SLCO2A1	chr3	3q21	Hypertrophic osteoarthropathy primary autosomal recessive 2	614441
SLITRK6	chr13	13q31	Deafness and myopia	221200
SLURP1	chr8	8qter	Meleda disease	248300
SLX4	chr16	16p13.3	Fanconi anemia complementation group P	613951
SMARCAL1	chr2	2q34-q36	Schimke immunoosseous dysplasia	242900
SMIM1	chr1	1p36	Blood group Vel system	615264
SMN1	chr5	5q12.2-q13.3	Spinal muscular atrophy-2	253550
SMN1	chr5	5q12.2-q13.3	Spinal muscular atrophy-3	253400
SMN1	chr5	5q12.2-q13.3	Spinal muscular atrophy-1	253300
SMN1	chr5	5q12.2-q13.3	Spinal muscular atrophy-4	271150
SMN2	chr5	5q12.2-q13.3	Spinal muscular atrophy type III modifier of	253400
SMO	chr7	7q31-q32	Pallister-Hall-like syndrome	241800
SMOC1	chr14	14q24.1	Microphthalmia with limb anomalies	206920
SMOC2	chr6	6q27	Dentin dysplasia type I with microdontia and misshapen teeth	125400
SMPD1	chr11	11p15.4-p15.1	Niemann-Pick disease type A	257200
SMPD1	chr11	11p15.4-p15.1	Niemann-Pick disease type B	607616
SMS	chrX	Xp22.1	Mental retardation X-linked Snyder-Robinson type	309583
SNAI2	chr8	8q11	Waardenburg syndrome type 2D	608890
SNAP29	chr22	22q11.2	Cerebral dysgenesis neuropathy ichthyosis and palmoplantar keratoderma syndrome	609528
SNIP1	chr1	1p34.3	Psychomotor retardation epilepsy and craniofacial dysmorphism	614501
SNX10	chr7	7p15.2	Osteopetrosis autosomal recessive 8	615085
SNX14	chr6	6q14.3	Spinocerebellar ataxia autosomal recessive 20	616354
SOBP	chr6	6q21	Mental retardation anterior maxillary protrusion and strabismus	613671
SOD1	chr21	21q22.1	Spastic tetraplegia and axial hypotonia progressive	618598
SOD1	chr21	21q22.1	Amyotrophic lateral sclerosis 1	105400
SOHLH1	chr9	9q34.3	Ovarian dysgenesis 5	617690

SORD	chr15	15q15.3	Sorbitol dehydrogenase deficiency with peripheral neuropathy	618912
SOST	chr17	17q12-q21	Sclerosteosis 1	269500
SOST	chr17	17q12-q21	Van Buchem disease	239100
SOX18	chr20	20q13.33	Hypotrichosis-lymphedema-telangiectasia syndrome	607823
SP110	chr2	2q37.1	Hepatic venoocclusive disease with immunodeficiency	235550
SP7	chr12	12q13.13	Osteogenesis imperfecta type XII	613849
SPAG1	chr8	8q22	Ciliary dyskinesia primary 28	615505
SPART	chr13	13q12.3	Troyer syndrome	275900
SPATA16	chr3	3q26.3	Spermatogenic failure 6	102530
SPATA7	chr14	14q31.3	Retinitis pigmentosa juvenile autosomal recessive	604232
SPEF2	chr5	5p13.2	Spermatogenic failure 43	618751
SPEG	chr2	2q35	Centronuclear myopathy 5	615959
SPG11	chr15	15q21.1	Charcot-Marie-Tooth disease axonal type 2X	616668
SPG11	chr15	15q21.1	Spastic paraplegia 11 autosomal recessive	604360
SPG11	chr15	15q21.1	Amyotrophic lateral sclerosis 5 juvenile	602099
SPG21	chr15	15q21-q22	Mast syndrome	248900
SPG7	chr16	16q24.3	Spastic paraplegia 7 autosomal recessive	607259
SPINK1	chr5	5q32	Fibrocalculous pancreatic diabetes susceptibility to	608189
SPINK1	chr5	5q32	Tropical calcific pancreatitis	608189
SPINK5	chr5	5q32	Netherton syndrome	256500
SPINT2	chr19	19q13.1	Diarrhea 3 secretory sodium congenital syndromic	270420
SPR	chr2	2p14-p12	Dystonia dopa-responsive due to sepiapterin reductase deficiency	612716
SPRTN	chr1	1q42.2	Ruijs-Aalfs syndrome	616200
SPTA1	chr1	1q21	Pyropoikilocytosis	266140
SPTA1	chr1	1q21	Spherocytosis type 3	270970
SPTBN2	chr11	11q13	Spinocerebellar ataxia autosomal recessive 14	615386
SQSTM1	chr5	5q35	Neurodegeneration with ataxia dystonia and gaze palsy childhood-onset	617145
SRD5A2	chr2	2p23	Pseudovaginal perineoscrotal hypospadias	264600
SRD5A3	chr4	4q12	Kahrizi syndrome	612713
SRD5A3	chr4	4q12	Congenital disorder of glycosylation type Iq	612379
SSR4	chrX	Xq28	Congenital disorder of glycosylation type Iy	300934
ST14	chr11	11q24-q25	Ichthyosis congenital autosomal recessive 11	602400
ST3GAL3	chr1	1p34.1	Mental retardation autosomal recessive 12	611090
ST3GAL3	chr1	1p34.1	Epileptic encephalopathy early infantile 15	615006
ST3GAL5	chr2	2p11.2	Salt and pepper developmental regression syndrome	609056
STAC3	chr12	12q13.3	Myopathy congenital Baily-Bloch	255995
STAG2	chrX	Xq25	Mullegama-Klein-Martinez syndrome	301022
STAG2	chrX	Xq25	Holoprosencephaly 13 X-linked	301043
STAG3	chr7	7q22	Premature ovarian failure 8	615723
STAMBP	chr2	2p13.1	Microcephaly-capillary malformation syndrome	614261
STAR	chr8	8p11.2	Lipoid adrenal hyperplasia	201710
STAT1	chr2	2q32.2-q32.3	Immunodeficiency 31B mycobacterial and viral infections autosomal recessive	613796

STAT2	chr12	12q13.2	Immunodeficiency 44	616636
STAT2	chr12	12q13.2	Pseudo-TORCH syndrome 3	618886
STIL	chr1	1p33	Microcephaly 7 primary autosomal recessive	612703
STIM1	chr11	11p15.5	Immunodeficiency 10	612783
STRA6	chr15	15q24.1	Microphthalmia isolated with coloboma 8	601186
STRA6	chr15	15q24.1	Microphthalmia syndromic 9	601186
STRADA	chr17	17q23.3	Polyhydramnios megalencephaly and symptomatic epilepsy	611087
STRC	chr15	15q15	Deafness autosomal recessive 16	603720
STS	chrX	Xp22.32	Ichthyosis X-linked	308100
STT3A	chr11	11q23.3	Congenital disorder of glycosylation type Iw	615596
STT3B	chr3	3p23	Congenital disorder of glycosylation type Ix	615597
STUB1	chr16	16p13.3	Spinocerebellar ataxia autosomal recessive 16	615768
STX11	chr6	6q24	Hemophagocytic lymphohistiocytosis familial 4	603552
SUCLA2	chr13	13q14.2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	612073
SUCLG1	chr2	2p11.2	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	245400
SUFU	chr10	10q24-q25	Medulloblastoma desmoplastic	155255
SUFU	chr10	10q24-q25	Joubert syndrome 32	617757
SUGCT	chr7	7p14	Glutaric aciduria III	231690
SULT2B1	chr19	19q13.3	Ichthyosis congenital autosomal recessive 14	617571
SUMF1	chr3	3p26	Multiple sulfatase deficiency	272200
SUOX	chr12	Chr.12	Sulfite oxidase deficiency	272300
SURF1	chr9	9q34	Leigh syndrome due to COX IV deficiency	256000
SURF1	chr9	9q34	Charcot-Marie-Tooth disease type 4K	616684
SYN1	chrX	Xp11.4-p11.2	Epilepsy X-linked with variable learning disabilities and behavior disorders	300491
SYNE1	chr6	6q25	Arthrogryposis multiplex congenita 3 myogenic type	618484
SYNE1	chr6	6q25	Spinocerebellar ataxia autosomal recessive 8	610743
SYNE4	chr19	19q13.12	Deafness autosomal recessive 76	615540
SYNJ1	chr21	21q22.2	Epileptic encephalopathy early infantile 53	617389
SYNJ1	chr21	21q22.2	Parkinson disease 20 early-onset	615530
SYP	chrX	Xp11.23-p11.22	Mental retardation X-linked 96	300802
SYT14	chr1	1q32.2	Spinocerebellar ataxia autosomal recessive 11	614229
SZT2	chr1	1p34.2	Epileptic encephalopathy early infantile 18	615476
TAC3	chr12	12q13-q21	Hypogonadotropic hypogonadism 10 with or without anosmia	614839
TACO1	chr17	17q22-q24.2	Mitochondrial complex IV deficiency	220110
TACR3	chr4	4q25	Hypogonadotropic hypogonadism 11 with or without anosmia	614840
TACSTD2	chr1	1p32	Corneal dystrophy gelatinous drop-like	204870
TAF1	chrX	Xq13	Dystonia-Parkinsonism X-linked	314250
TAF1	chrX	Xq13	Mental retardation X-linked syndromic 33	300966
TAF2	chr8	8q24.12	Mental retardation autosomal recessive 40	615599
TAF4B	chr18	18q11.2	Spermatogenic failure 13	615841

TAF6	chr7	7q22.1	Alazami-Yuan syndrome	617126
TALDO1	chr11	11p15.5-p15.4	Transaldolase deficiency	606003
TAP1	chr6	6p21.3	Bare lymphocyte syndrome type I	604571
TAP2	chr6	6p21.3	Bare lymphocyte syndrome type I due to TAP2 deficiency	604571
TAPBP	chr6	6p21.3	Bare lymphocyte syndrome type I	604571
TARS2	chr1	1q21.2	Combined oxidative phosphorylation deficiency 21	615918
TAT	chr16	16q22.1-q22.3	Tyrosinemia type II	276600
TAZ	chrX	Xq28	Barth syndrome	302060
TBC1D20	chr20	20p13	Warburg micro syndrome 4	615663
TBC1D23	chr3	3q12.1-q12.2	Pontocerebellar hypoplasia type 11	617695
TBC1D24	chr16	16p13.3	Epilepsy rolandic with proxysmal exercise-induce dystonia and writer's cramp	608105
TBC1D24	chr16	16p13.3	DOORS syndrome	220500
TBC1D24	chr16	16p13.3	Epileptic encephalopathy early infantile 16	615338
TBC1D24	chr16	16p13.3	Myoclonic epilepsy infantile familial	605021
TBC1D24	chr16	16p13.3	Deafness autosomal recessive 86	614617
TBC1D7	chr6	6p24.1	Macrocephaly/megalencephaly syndrome autosomal recessive	248000
TBCE	chr1	1q42-q43	Kenny-Caffey syndrome type 1	244460
TBCE	chr1	1q42-q43	Hypoparathyroidism-retardation-dysmorphism syndrome	241410
TBCE	chr1	1q42-q43	Encephalopathy progressive with amyotrophy and optic atrophy	617207
TBCK	chr4	4q24	Hypotonia infantile with psychomotor retardation and characteristic facies 3	616900
TBX15	chr1	1p13	Cousin syndrome	260660
TBX19	chr1	1q23-q24	Adrenocorticotropic hormone deficiency	201400
TBX21	chr17	17q21.3	Asthma aspirin-induced susceptibility to	208550
TBX21	chr17	17q21.3	Asthma and nasal polyps	208550
TBX4	chr17	17q21-q22	Amelia posterior with pelvic and pulmonary hypoplasia syndrome	601360
TBX4	chr17	17q21-q22	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension	147891
TBX6	chr16	16p11.2	Spondylocostal dysostosis 5	122600
TBXAS1	chr7	7q34	Ghosal hematodiaphyseal syndrome	231095
TCAP	chr17	17q12	Muscular dystrophy limb-girdle autosomal recessive 7	601954
TCIRG1	chr11	11q13.4-q13.5	Osteopetrosis autosomal recessive 1	259700
TCN2	chr22	22q11.2-qter	Transcobalamin II deficiency	275350
TCTN1	chr12	12q24.1	Joubert syndrome 13	614173
TCTN2	chr12	12q24.31	Joubert syndrome 24	616654
TCTN2	chr12	12q24.31	Meckel syndrome 8	613885
TCTN3	chr10	10q24.1	Orofaciodigital syndrome IV	258860
TCTN3	chr10	10q24.1	Joubert syndrome 18	614815
TDO2	chr4	4q31-q32	Hypertryptophanemia	600627

TDP1	chr14	14q31-q32	Spinocerebellar ataxia autosomal recessive with axonal neuropathy 1	607250
TDP2	chr6	6p22.3-p22.1	Spinocerebellar ataxia autosomal recessive 23	616949
TDRD7	chr9	9q22.33	Cataract 36	613887
TECPR2	chr14	14q32.31	Spastic paraplegia 49 autosomal recessive	615031
TECR	chr19	19p13.12	Mental retardation autosomal recessive 14	614020
TECTA	chr11	11q22-q24	Deafness autosomal recessive 21	603629
TELO2	chr16	16p13.3	You-Hoover-Fong syndrome	616954
TENM3	chr4	4q35.1	Microphthalmia syndromic 15	615145
TENM3	chr4	4q35.1	Microphthalmia isolated with coloboma 9	615145
TERT	chr5	5p15.33	Dyskeratosis congenita autosomal recessive 4	613989
TERT	chr5	5p15.33	Dyskeratosis congenita autosomal dominant 2	613989
TEX14	chr17	17q22	Spermatogenic failure 23	617707
TF	chr3	3q21	Atransferrinemia	209300
TFAM	chr10	10q21	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)	617156
TFG	chr3	3q11-q12	Spastic paraplegia 57 autosomal recessive	615658
TFR2	chr7	7q22	Hemochromatosis type 3	604250
TFRC	chr3	3q29	Immunodeficiency 46	616740
TG	chr8	8q24.2-q24.3	Thyroid dyshormonogenesis 3	274700
TGDS	chr13	13q32.1	Catel-Manzke syndrome	616145
TGFB1	chr19	19q13.1	Cystic fibrosis lung disease modifier of	219700
TGFB1	chr19	19q13.1	Inflammatory bowel disease immunodeficiency and encephalopathy	618213
TGM1	chr14	14q11.2	Ichthyosis congenital autosomal recessive 1	242300
TGM5	chr15	15q15.2	Peeling skin syndrome 2	609796
TH	chr11	11p15.5	Segawa syndrome recessive	605407
THOC2	chrX	Xq25	Mental retardation X-linked 12/35	300957
THOC6	chr16	16p13.3	Beaulieu-Boycott-Innes syndrome	613680
THRB	chr3	3p24.3	Thyroid hormone resistance autosomal recessive	274300
TIA1	chr2	2p13	Welander distal myopathy	604454
TICAM1	chr19	19p13.3	Encephalopathy acute infection-induced (herpes-specific) susceptibility to 6	614850
TIMM8A	chrX	Xq22	Mohr-Tranebjaerg syndrome	304700
TJP2	chr9	9q12-q13	Cholestasis progressive familial intrahepatic 4	615878
TJP2	chr9	9q12-q13	Hypercholanemia familial	607748
TK2	chr16	16q22	Mitochondrial DNA depletion syndrome 2 (myopathic type)	609560
TK2	chr16	16q22	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive 3	617069
TLE6	chr19	19p13.3	Preimplantation embryonic lethality	616814
TLR3	chr4	4q35	Encephalopathy acute infection-induced (herpes-specific) susceptibility to 2	613002
TMC1	chr9	9q13-q21	Deafness autosomal recessive 7	600974
TMC6	chr17	17q25	Epidermodysplasia verruciformis	226400
TMC8	chr17	17q25	Epidermodysplasia verruciformis 2	618231
TMCO1	chr1	1q24.1	Craniofacial dysmorphism skeletal anomalies and mental retardation syndrome	213980

TMEM126A	chr11	11q14.1-q21	Optic atrophy 7	612989
TMEM138	chr11	11q12.2	Joubert syndrome 16	614465
TMEM165	chr4	4q12	Congenital disorder of glycosylation type IIk	614727
TMEM216	chr11	11q12.2	Meckel syndrome 2	603194
TMEM216	chr11	11q12.2	Joubert syndrome 2	608091
TMEM231	chr16	16q23.1	Meckel syndrome 11	615397
TMEM231	chr16	16q23.1	Joubert syndrome 20	614970
TMEM237	chr2	2q33.2	Joubert syndrome 14	614424
TMEM67	chr8	8q21.13-q22.1	Meckel syndrome 3	607361
TMEM67	chr8	8q21.13-q22.1	RHYNS syndrome	602152
TMEM67	chr8	8q21.13-q22.1	Nephronophthisis 11	613550
TMEM67	chr8	8q21.13-q22.1	Bardet-Biedl syndrome 14 modifier of	615991
TMEM67	chr8	8q21.13-q22.1	COACH syndrome	216360
TMEM67	chr8	8q21.13-q22.1	Joubert syndrome 6	610688
TMEM70	chr8	8q21.11	Mitochondrial complex V (ATP synthase) deficiency nuclear type 2	614052
TMIE	chr3	3p21	Deafness autosomal recessive 6	600971
TMLHE	chrX	Xq28	Autism susceptibility to X-linked 6	300872
TMPRSS15	chr21	21q21	Enterokinase deficiency	226200
TMPRSS3	chr21	21q22.3	Deafness autosomal recessive 8/10	601072
TMPRSS6	chr22	22q12-q13	Iron-refractory iron deficiency anemia	206200
TMTC3	chr12	12q21.32	Lissencephaly 8	617255
TNFRSF10B	chr8	8p22-p21	Squamous cell carcinoma head and neck	275355
TNFRSF11A	chr18	18q22.1	Osteopetrosis autosomal recessive 7	612301
TNFRSF11B	chr8	8q24	Paget disease of bone 5 juvenile-onset	239000
TNFRSF13B	chr17	17p11.2	Immunodeficiency common variable 2	240500
TNFRSF13C	chr22	22q13.1-q13.31	Immunodeficiency common variable 4	613494
TNFRSF4	chr1	1p36	Immunodeficiency 16	615593
TNFSF11	chr13	13q14	Osteopetrosis autosomal recessive 2	259710
TNNI3	chr19	19q13.4	Cardiomyopathy dilated 2A	611880
TNNT1	chr19	19q13.4	Nemaline myopathy 5 Amish type	605355
TNXB	chr6	6p21.3	Ehlers-Danlos syndrome classic-like 1	606408
TOP3A	chr17	17p12-p11.2	Microcephaly growth restriction and increased sister chromatid exchange 2	618097
TOP3A	chr17	17p12-p11.2	Progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive 5	618098
TOR1AIP1	chr1	1q24	Muscular dystrophy autosomal recessive with rigid spine and distal joint contractures	617072
TP53RK	chr20	20q13.12	Galloway-Mowat syndrome 4	617730
TPI1	chr12	12p13	Hemolytic anemia due to triosephosphate isomerase deficiency	615512
TPK1	chr7	7q34	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)	614458

TPM3	chr1	1q22-q23	CAP myopathy 1	609284
TPM3	chr1	1q22-q23	Nemaline myopathy 1 autosomal dominant or recessive	609284
TPM3	chr1	1q22-q23	Myopathy congenital with fiber-type disproportion	255310
TPMT	chr6	6p22.3	Thiopurines poor metabolism of 1	610460
TPO	chr2	2p25	Thyroid dyshormonogenesis 2A	274500
TPP1	chr11	11p15.5	Spinocerebellar ataxia autosomal recessive 7	609270
TPP1	chr11	11p15.5	Ceroid lipofuscinosis neuronal 2	204500
TPRN	chr9	9q34.3	Deafness autosomal recessive 79	613307
TRAC	chr14	14q11.2	Immunodeficiency 7 TCR-alpha/beta deficient	615387
TRAF3IP1	chr2	2q37.3	Senior-Loken syndrome 9	616629
TRAF3IP2	chr6	6q21	Candidiasis familial 8	615527
TRAK1	chr3	3p25.3-p24.1	Epileptic encephalopathy early infantile 68	618201
TRAPPC11	chr4	4q35.1	Muscular dystrophy limb-girdle autosomal recessive 18	615356
TRAPPC2	chrX	Xp22.2-p22.1	Spondyloepiphyseal dysplasia tarda	313400
TRAPPC9	chr8	8q24.3	Mental retardation autosomal recessive 13	613192
TRDN	chr6	6q22-q23	Ventricular tachycardia catecholaminergic polymorphic 5 with or without muscle weakness	615441
TREH	chr11	11q23	Trehalase deficiency	612119
TREX1	chr3	3p21.3-p21.2	Systemic lupus erythematosus susceptibility to	152700
TREX1	chr3	3p21.3-p21.2	Vasculopathy retinal with cerebral leukodystrophy	192315
TREX1	chr3	3p21.3-p21.2	Aicardi-Goutieres syndrome 1 dominant and recessive	225750
TRH	chr3	3q13.3-q21	Thyrotropin-releasing hormone deficiency	275120
TRHR	chr8	8q23	Hypothyroidism congenital nongoitrous 7	618573
TRIM2	chr4	4q31.3	Charcot-Marie-Tooth disease type 2R	615490
TRIM32	chr9	9q31-q34.1	Bardet-Biedl syndrome 11	615988
TRIM32	chr9	9q31-q34.1	Muscular dystrophy limb-girdle autosomal recessive 8	254110
TRIM37	chr17	17q22-q23	Mulibrey nanism	253250
TRIOBP	chr22	22q13.1	Deafness autosomal recessive 28	609823
TRIP11	chr14	14q31-q32	Osteochondrodysplasia	184260
TRIP11	chr14	14q31-q32	Achondrogenesis type IA	200600
TRIP13	chr5	5p15.33	Mosaic variegated aneuploidy syndrome 3	617598
TRIT1	chr1	1p34.2	Combined oxidative phosphorylation deficiency 35	617873
TRMT1	chr19	19p13.3	Mental retardation autosomal recessive 68	618302
TRMT10A	chr4	4q23	Microcephaly short stature and impaired glucose metabolism 1	616033
TRMU	chr22	22q13	Liver failure transient infantile	613070
TRNT1	chr3	3p25.1	Sideroblastic anemia with B-cell immunodeficiency periodic fevers and developmental delay	616084
TRNT1	chr3	3p25.1	Retinitis pigmentosa and erythrocytic microcytosis	616959
TRPM1	chr15	15q13-q14	Night blindness congenital stationary (complete) 1C autosomal recessive	613216
TRPM6	chr9	9q22	Hypomagnesemia 1 intestinal	602014
TSEN15	chr1	1q25	Pontocerebellar hypoplasia type 2F	617026
TSEN2	chr3	3p25.1	Pontocerebellar hypoplasia type 2B	612389
TSEN34	chr19	19q13.4	Pontocerebellar hypoplasia type 2C	612390

TSEN54	chr17	17q25.1	Pontocerebellar hypoplasia type 4	225753
TSEN54	chr17	17q25.1	Pontocerebellar hypoplasia type 2A	277470
TSEN54	chr17	17q25.1	Pontocerebellar hypoplasia type 5	610204
TSFM	chr12	12q13-q14	Combined oxidative phosphorylation deficiency 3	610505
TSHB	chr1	1p13	Hypothyroidism congenital nongoitrous 4	275100
TSHR	chr14	14q31	Hypothyroidism congenital nongoitrous 1	275200
TSPAN7	chrX	Xq11	Mental retardation X-linked 58	300210
TSPEAR	chr21	21q22.3	Ectodermal dysplasia 14 hair/tooth type with or without hypohidrosis	618180
TSPEAR	chr21	21q22.3	Deafness autosomal recessive 98	614861
TSPYL1	chr6	6q22-q23	Sudden infant death with dysgenesis of the testes syndrome	608800
TSR2	chrX	Xp11.22	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis	300946
TTC19	chr17	17p12	Mitochondrial complex III deficiency nuclear type 2	615157
TTC21B	chr2	2q24.3	Nephronophthisis 12	613820
TTC21B	chr2	2q24.3	Short-rib thoracic dysplasia 4 with or without polydactyly	613819
TTC37	chr5	5q15	Trichohepatoenteric syndrome 1	222470
TTC7A	chr2	2p21	Gastrointestinal defects and immunodeficiency syndrome	243150
TTC8	chr14	14q32.1	Bardet-Biedl syndrome 8	615985
TTC8	chr14	14q32.1	Retinitis pigmentosa 51	613464
TTI2	chr8	8p12	Mental retardation autosomal recessive 39	615541
TTLL5	chr14	14q24.3	Cone-rod dystrophy 19	615860
TTN	chr2	2q31	Muscular dystrophy limb-girdle autosomal recessive 10	608807
TTN	chr2	2q31	Salih myopathy	611705
TTPA	chr8	8q13.1-q13.3	Ataxia with isolated vitamin E deficiency	277460
TUB	chr11	11p15.4	Retinal dystrophy and obesity	616188
TUBA8	chr22	22q11	Cortical dysplasia complex with other brain malformations 8	613180
TUBGCP4	chr15	15q15	Microcephaly and chorioretinopathy autosomal recessive 3	616335
TUBGCP6	chr22	22q13.33	Microcephaly and chorioretinopathy autosomal recessive 1	251270
TUFM	chr16	16p11.2	Combined oxidative phosphorylation deficiency 4	610678
TULP1	chr6	6p21.3	Retinitis pigmentosa 14	600132
TULP1	chr6	6p21.3	Leber congenital amaurosis 15	613843
TUSC3	chr8	8p22	Mental retardation autosomal recessive 7	611093
TWIST2	chr2	2q37.3	Focal facial dermal dysplasia 3 Setleis type	227260
TWNK	chr10	10q24	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	271245
TWNK	chr10	10q24	Perrault syndrome 5	616138
TXN2	chr22	22q12.3	Combined oxidative phosphorylation deficiency 29	616811
TXNL4A	chr18	18q23	Burn-McKeown syndrome	608572
TXNRD2	chr22	22q11.2	Glucocorticoid deficiency 5	617825
TYK2	chr19	19p13.2	Immunodeficiency 35	611521
TYMP	chr22	22q13.32-qter	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	603041

TYR	chr11	11q14-q21	Albinism oculocutaneous type IB	606952
TYR	chr11	11q14-q21	Albinism oculocutaneous type IA	203100
TYROBP	chr19	19q13.1	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1	221770
TYRP1	chr9	9p23	Albinism oculocutaneous type III	203290
UBA1	chrX	Xp11.23	Spinal muscular atrophy X-linked 2 infantile	301830
UBE2A	chrX	Xq24	Mental retardation X-linked syndromic Nascimento-type	300860
UBE2T	chr1	1q31	Fanconi anemia complementation group T	616435
UBE3B	chr12	12q24.11	Kaufman oculocerebrofacial syndrome	244450
UBR1	chr15	15q15-q21.1	Johanson-Blizzard syndrome	243800
UCHL1	chr4	4p14	Spastic paraplegia 79 autosomal recessive	615491
UCHL1	chr4	4p14	Parkinson disease 5 susceptibility to	613643
UCP3	chr11	11q13	Obesity severe and type II diabetes	601665
UGT1A1	chr2	2q37	Gilbert syndrome	143500
UGT1A1	chr2	2q37	Hyperbilirubinemia familial transient neonatal	237900
UGT1A1	chr2	2q37	Crigler-Najjar syndrome type I	218800
UGT1A1	chr2	2q37	Crigler-Najjar syndrome type II	606785
UMPS	chr3	3q13	Orotic aciduria	258900
UNC13D	chr17	17q25.1	Hemophagocytic lymphohistiocytosis familial 3	608898
UNC80	chr2	2q35	Hypotonia infantile with psychomotor retardation and characteristic facies 2	616801
UNG	chr12	12q23-q24.1	Immunodeficiency with hyper IgM type 5	608106
UPB1	chr22	22q11.2	Beta-ureidopropionase deficiency	613161
UPF3B	chrX	Xq25-q26	Mental retardation X-linked syndromic 14	300676
UQCRB	chr8	8q22	Mitochondrial complex III deficiency nuclear type 3	615158
UQCRC2	chr16	16p12	Mitochondrial complex III deficiency nuclear type 5	615160
UQCRFS1	chr19	19q12	Mitochondrial complex III deficiency nuclear type 10	618775
UQCRQ	chr5	5q31.1	Mitochondrial complex III deficiency nuclear type 4	615159
UROC1	chr3	3q21.3	Urocanase deficiency	276880
UROD	chr1	1p34	Porphyria hepatoerythropoietic	176100
UROD	chr1	1p34	Porphyria cutanea tarda	176100
UROS	chr10	10q25.2-q26.3	Porphyria congenital erythropoietic	263700
USB1	chr16	16q13	Poikiloderma with neutropenia	604173
USH1C	chr11	11p15.1	Deafness autosomal recessive 18A	602092
USH1C	chr11	11p15.1	Usher syndrome type 1C	276904
USH1G	chr17	17q24-q25	Usher syndrome type 1G	606943
USH2A	chr1	1q41	Usher syndrome type 2A	276901
USP27X	chrX	Xp11.2	Mental retardation X-linked 105	300984
USP9X	chrX	Xp11.4	Mental retardation X-linked 99	300919
USP9X	chrX	Xp11.4	Mental retardation X-linked 99 syndromic female-restricted	300968
UVSSA	chr4	4p16.3	UV-sensitive syndrome 3	614640
VAMP1	chr12	12p13.31	Myasthenic syndrome congenital 25	618323
VARS2	chr6	6p21.3	Combined oxidative phosphorylation deficiency 20	615917
VAX1	chr10	10q26.1	Microphthalmia syndromic 11	614402
VDR	chr12	12q12-q14	Rickets vitamin D-resistant type IIA	277440

VHL	chr3	3p26-p25	Erythrocytosis familial 2	263400
VIPAS39	chr14	14q24.3	Arthrogryposis renal dysfunction and cholestasis 2	613404
VLDLR	chr9	9p24	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	224050
VMA21	chrX	Xq28	Myopathy X-linked with excessive autophagy	310440
VPS13A	chr9	9q21	Choreoacanthocytosis	200150
VPS13B	chr8	8q22-q23	Cohen syndrome	216550
VPS33B	chr15	15q26.1	Arthrogryposis renal dysfunction and cholestasis 1	208085
VPS37A	chr8	8p23-p21	Spastic paraplegia 53 autosomal recessive	614898
VPS45	chr1	1q21-q22	Neutropenia severe congenital 5 autosomal recessive	615285
VPS53	chr17	17p13.3	Pontocerebellar hypoplasia type 2E	615851
VRK1	chr14	14q32	Pontocerebellar hypoplasia type 1A	607596
VWA3B	chr2	2q11.2	Spinocerebellar ataxia autosomal recessive 22	616948
VWF	chr12	12p13.3	von Willebrand disease type 1	193400
VWF	chr12	12p13.3	von Willebrand disease types 2A 2B 2M and 2N	613554
VWF	chr12	12p13.3	von Willibrand disease type 3	277480
WAS	chrX	Xp11.23-p11.22	Thrombocytopenia X-linked intermittent	313900
WAS	chrX	Xp11.23-p11.22	Thrombocytopenia X-linked	313900
WAS	chrX	Xp11.23-p11.22	Wiskott-Aldrich syndrome	301000
WAS	chrX	Xp11.23-p11.22	Neutropenia severe congenital X-linked	300299
WASHC4	chr12	12q23.3	Mental retardation autosomal recessive 43	615817
WASHC5	chr8	8q24.13	Ritscher-Schinzel syndrome 1	220210
WDPCP	chr2	2p15	Bardet-Biedl syndrome 15	615992
WDPCP	chr2	2p15	Congenital heart defects hamartomas of tongue and polysyndactyly	217085
WDR19	chr4	4p14-p11	Nephronophthisis 13	614377
WDR19	chr4	4p14-p11	Senior-Loken syndrome 8	616307
WDR19	chr4	4p14-p11	Cranioectodermal dysplasia 4	614378
WDR19	chr4	4p14-p11	Short-rib thoracic dysplasia 5 with or without polydactyly	614376
WDR35	chr2	2p24.1	Short-rib thoracic dysplasia 7 with or without polydactyly	614091
WDR35	chr2	2p24.1	Cranioectodermal dysplasia 2	613610
WDR4	chr21	21q22.3	Microcephaly growth deficiency seizures and brain malformations	618346
WDR4	chr21	21q22.3	Galloway-Mowat syndrome 6	618347
WDR45B	chr17	17q25.3	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures	617977
WDR62	chr19	19q13.12	Microcephaly 2 primary autosomal recessive with or without cortical malformations	604317
WDR72	chr15	15q21.3	Amelogenesis imperfecta type IIA3	613211
WDR73	chr15	15q25.2	Galloway-Mowat syndrome 1	251300
WDR81	chr17	17p13.3	Hydrocephalus congenital 3 with brain anomalies	617967
WDR81	chr17	17p13.3	Cerebellar ataxia mental retardation and dysequilibrium syndrome 2	610185

WFS1	chr4	4p16.1	Wolfram syndrome 1	222300
WHRN	chr9	9q32-q34	Deafness autosomal recessive 31	607084
WHRN	chr9	9q32-q34	Usher syndrome type 2D	611383
WNK1	chr12	12p13	Neuropathy hereditary sensory and autonomic type II	201300
WNT1	chr12	12q12-q13	Osteogenesis imperfecta type XV	615220
WNT10A	chr2	2q35	Schopf-Schulz-Passarge syndrome	224750
WNT10A	chr2	2q35	Tooth agenesis selective 4	150400
WNT10A	chr2	2q35	Odontoonychodermal dysplasia	257980
WNT10B	chr12	12q13	Split-hand/foot malformation 6	225300
WNT3	chr17	17q21	Tetra-amelia syndrome 1	273395
WNT4	chr1	1p35	SERKAL syndrome	611812
WNT7A	chr3	3p25	Fuhrmann syndrome	228930
WNT7A	chr3	3p25	Ulna and fibula absence of with severe limb deficiency	276820
WRAP53	chr17	17p13	Dyskeratosis congenita autosomal recessive 3	613988
WRN	chr8	8p12-p11.2	Werner syndrome	277700
WWOX	chr16	16q23.3-q24.1	Spinocerebellar ataxia autosomal recessive 12	614322
WWOX	chr16	16q23.3-q24.1	Epileptic encephalopathy early infantile 28	616211
XDH	chr2	2p23-p22	Xanthinuria type I	278300
XIAP	chrX	Xq25	Lymphoproliferative syndrome X-linked 2	300635
XPA	chr9	9q22.3	Xeroderma pigmentosum group A	278700
XPC	chr3	3p25	Xeroderma pigmentosum group C	278720
XPNPEP3	chr22	22q13.31-q13.33	Nephronophthisis-like nephropathy 1	613159
XRCC1	chr19	19q13.2	Spinocerebellar ataxia autosomal recessive 26	617633
XRCC2	chr7	7q36.1	Fanconi anemia complementation group U	617247
XRCC4	chr5	5q13-q14	Short stature microcephaly and endocrine dysfunction	616541
XYLT1	chr16	16p13.1	Pseudoxanthoma elasticum modifier of severity of	264800
XYLT1	chr16	16p13.1	Desbuquois dysplasia 2	615777
XYLT2	chr17	17q21.3-q22	Pseudoxanthoma elasticum modifier of severity of	264800
XYLT2	chr17	17q21.3-q22	Spondyloocular syndrome	605822
YARS2	chr12	12p11.21	Myopathy lactic acidosis and sideroblastic anemia 2	613561
ZAP70	chr2	2q12	Autoimmune disease multisystem infantile-onset 2	617006
ZAP70	chr2	2q12	Immunodeficiency 48	269840
ZBTB11	chr3	3q12.3	Intellectual developmental disorder autosomal recessive 69	618383
ZBTB16	chr11	11q23.1	Skeletal defects genital hypoplasia and mental retardation	612447
ZBTB24	chr6	6q21	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	614069
ZBTB42	chr14	14q32.33	Lethal congenital contracture syndrome 6	616248
ZC3H14	chr14	14q31.3	Mental retardation autosomal recessive 56	617125
ZC4H2	chrX	Xq11.2	Wieacker-Wolff syndrome	314580
ZFYVE26	chr14	14q24.1	Spastic paraplegia 15 autosomal recessive	270700
ZIC3	chrX	Xq26.2	Congenital heart defects nonsyndromic 1 X-linked	306955

ZIC3	chrX	Xq26.2	Heterotaxy visceral 1 X-linked	306955
ZIC3	chrX	Xq26.2	VACTERL association X-linked	314390
ZMPSTE24	chr1	1p34	Restrictive dermopathy lethal	275210
ZMPSTE24	chr1	1p34	Mandibuloacral dysplasia with type B lipodystrophy	608612
ZMYND10	chr3	3p21.3	Ciliary dyskinesia primary 22	615444
ZMYND15	chr17	17p13.2	Spermatogenic failure 14	615842
ZNF141	chr4	4p16.3	Polydactyly postaxial type A6	615226
ZNF335	chr20	20q11.2-q13.1	Microcephaly 10 primary autosomal recessive	615095
ZNF408	chr11	11p11.2	Exudative vitreoretinopathy 6	616468
ZNF408	chr11	11p11.2	Retinitis pigmentosa 72	616469
ZNF423	chr16	16q12	Nephronophthisis 14	614844
ZNF423	chr16	16q12	Joubert syndrome 19	614844
ZNF469	chr16	16q24	Brittle cornea syndrome 1	229200
ZNF513	chr2	2p24.1-p22.3	Retinitis pigmentosa 58	613617
ZP1	chr11	11q12.2	Oocyte maturation defect 1	615774
AGTR2	chrX	Xq23	Mental retardation, X-linked 88	300852
CASP10	chr2	2q33-q34	Autoimmune lymphoproliferative syndrome type II	603909
ZNF41	chrX	Xp22.1-cen	Mental retardation, X-linked	314995
CDKL5	chrX	Xp22	Epileptic encephalopathy early infantile 2	300672
PTEN	chr10	10q23.31	Lhermitte-Duclos syndrome	158350
SFTPC	chr8	8p21	Surfactant metabolism dysfunction pulmonary 2	610913
ARHGEF6	chrX	Xq26	X-linked non-syndromic intellectual disability	
UBE3A	chr15	15q11-q13	Angelman syndrome	105830
CASK	chrX	Xp11.4	Mental retardation with or without nystagmus	300422
CASK	chrX	Xp11.4	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	300749
CASK	chrX	Xp11.4	FG syndrome 4	300422
PDHA1	chrX	Xp22.2-p22.1	Leigh syndrome with leukodystrophy	312170
TCF4	chr18	18q21.2	Corneal dystrophy Fuchs endothelial 3	613267
SHROOM4	chrX	Xp11.2	Stocco dos Santos X-linked mental retardation syndrome	300434
OXCT1	chr5	5p13	Succinyl CoA:3-oxoacid CoA transferase deficiency	245050
GLA	chrX	Xq22	Fabry disease	301500
GLA	chrX	Xq22	Fabry disease cardiac variant	301500
NHEJ1	chr2	2q35	Cernunnos-XLF deficiency	611291
ZEB2	chr2	2q22	Mowat-Wilson syndrome	235730
NSD1	chr5	5q35	Sotos syndrome 1	117550
NLGN4X	chrX	Xp22.33	Mental retardation X-linked	300495
NLGN4X	chrX	Xp22.33	Asperger syndrome susceptibility X-linked 2	300497
NLGN4X	chrX	Xp22.33	Autism susceptibility X-linked 2	300495
BCOR	chrX	Xp11.4	Microphthalmia syndromic 2	300166
RPS6KA3	chrX	Xp22.2-p22.1	Mental retardation X-linked 19	300844
RPS6KA3	chrX	Xp22.2-p22.1	Coffin-Lowry syndrome	303600
STXBP2	chr19	19p13.3-p13.2	Hemophagocytic lymphohistiocytosis familial 5	613101
CHMP1B	chr18	18p11	Spastic paraplegia	606486

COMPATIBILITY
GENETIC TEST

INSTITUTO BERNABEU

EFNB1	chrX	Xq12	Craniofrontonasal dysplasia	304110
HJV	chr1	1q21	Hemochromatosis type 2A	602390
DCX	chrX	Xq22.3-q23	Subcortical laminal heterotopia X-linked	300067
DCX	chrX	Xq22.3-q23	Lissencephaly X-linked	300067
NXF5	chrX	Xq22.1	Focal segmental glomerulosclerosis	300319
COL1A1	chr17	17q21.31-q22	Osteogenesis imperfecta type I	166200
COL1A1	chr17	17q21.31-q22	Osteogenesis imperfecta type IV	166220
COL1A1	chr17	17q21.31-q22	Osteogenesis imperfecta type II	166210
COL1A1	chr17	17q21.31-q22	Bone mineral density variation QTL osteoporosis	166710
COL1A1	chr17	17q21.31-q22	Caffey disease	114000
COL1A1	chr17	17q21.31-q22	Ehlers-Danlos syndrome arthrochalasia type 1	130060
COL1A1	chr17	17q21.31-q22	Osteogenesis imperfecta type III	259420
TUBA1A	chr12	12q12-q14	Lissencephaly 3	611603
SLC16A2	chrX	Xq13.2	Allan-Herndon-Dudley syndrome	300523
SLC9A6	chrX	Xq26.3	Mental retardation X-linked syndromic Christianson type	300243
ZNF674	chrX	Xp11	X-linked mental retardation	300573
FASLG	chr1	1q23	Autoimmune lymphoproliferative syndrome type IB	601859
HSD17B10	chrX	Xp11.2	HSD10 mitochondrial disease	300438
NDUFA7	chr19	19p13.2	Disease associated to NDUFA7	602139
ZNF711	chrX	Xq21.1-q21.3	Mental retardation X-linked 97	300803
FOXG1	chr14	14q13	Rett syndrome congenital variant	613454
PCDH19	chrX	Xq22	Epileptic encephalopathy early infantile 9	300088
ARSL	chrX	Xp22.3	Chondrodysplasia punctata X-linked recessive	302950
ACADL	chr2	2q34-q35	Pulmonary surfactant dysfunction	609576
HBA1	chr16	16pter-p13.3	Hemoglobin H disease nondeletional	613978
HBA1	chr16	16pter-p13.3	Thalassemias alpha-	604131
HBA1	chr16	16pter-p13.3	Erythrocytosis 7	617981
HBA1	chr16	16pter-p13.3	Methemoglobinemia alpha type	617973
HBA1	chr16	16pter-p13.3	Heinz body anemias alpha-	140700
NR5A1	chr9	9q33	Adrenocortical insufficiency	612964
NR5A1	chr9	9q33	46 XX sex reversal 4	617480
NR5A1	chr9	9q33	Premature ovarian failure 7	612964
NR5A1	chr9	9q33	Spermatogenic failure 8	613957
NR5A1	chr9	9q33	46XY sex reversal 3	612965
HUWE1	chrX	Xp11.2	Mental retardation X-linked syndromic Turner type	309590
ZDHHC9	chrX	Xq26.1	Mental retardation X-linked syndromic Raymond type	300799
G6PD	chrX	Xq28	Hemolytic anemia G6PD deficient (favism)	300908
G6PD	chrX	Xq28	Resistance to malaria due to G6PD deficiency	611162
CYP11A1	chr15	15q24.1	Adrenal insufficiency congenital with 46XY sex reversal partial or complete	613743
NDUFS5	chr1	1p34.2-p33	Complex I-deficiency	603847
MMUT	chr6	6p21	Methylmalonic aciduria mut(0) type	251000
GTF2H5	chr6	6q25.3	Trichothiodystrophy 3 photosensitive	616395

UNC93B1	chr11	11q13	Encephalopathy acute infection-induced (herpes-specific) susceptibility to 1	610551
NHS	chrX	Xp22.13	Nance-Horan syndrome	302350
NHS	chrX	Xp22.13	Cataract 40 X-linked	302200
SOX3	chrX	Xq26.3	Mental retardation X-linked with isolated growth hormone deficiency	300123
SOX3	chrX	Xq26.3	Panhypopituitarism X-linked	312000
ACSL4	chrX	Xq22.3	Mental retardation X-linked 63	300387
GDI1	chrX	Xq28	Mental retardation X-linked 41	300849
FAS	chr10	10q24.1	Autoimmune lymphoproliferative syndrome type IA	601859
FAS	chr10	10q24.1	Autoimmune lymphoproliferative syndrome	601859
COL4A5	chrX	Xq22.3	Alport syndrome 1 X-linked	301050