Gene (2430 genes)	Associated Phenotype description and OMIM disease ID
NUP107	Nephrotic syndrome,type 11,616730
TFRC	Immunodeficiency 46,616740
POP1	Anauxetic dysplasia 2
IFT172	Retinitis pigmentosa 71,616394 Short-rib thoracic dysplasia 10 with or without polydactyly,615630
SPINT2	Diarrhea 3,secretory sodium,congenital,syndromic 270420
ALKBH8	Intellectual developmental disorder,autosomal recessive 71
NDST1	Mental retardation,autosomal recessive 46,616116
SCP2	Leukoencephalopathy with dystonia and motor neuropathy,613724
MLYCD	Malonyl-CoA decarboxylase deficiency,248360
WLS	-
SECISBP2	Thyroid hormone metabolism,abnormal,609698
CACNA2D2	Cerebellar atrophy with seizures and variable developmental delay
CD2AP	Glomerulosclerosis,focal segmental,3,607832
LARS2	Perrault syndrome 4,615300 ?Hydrops,lactic acidosis,and sideroblastic anemia,617021
MTHFD1	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia
SCO1	Mitochondrial complex IV deficiency,220110
SCO2	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 1,604377 Myopia 6,608908
TFR2	Hemochromatosis,type 3,604250
LARS1	?Infantile liver failure syndrome 1
AGTR1	Renal tubular dysgenesis,267430
TMEM126A	Optic atrophy 7,612989
ORAI1	Immunodeficiency 9 612782
FAR1	Peroxisomal fatty acyl-CoA reductase 1 disorder,616154
TMEM126B	Mitochondrial complex I deficiency,252010
ATF6	Achromatopsia 7,616517
COL13A1	Myasthenic syndrome,congenital,19,616720
SAR1B	Chylomicron retention disease,246700
B3GLCT	Peters-plus syndrome,261540
CDCA7	Immunodeficiency-centromeric instability-facial anomalies syndrome 3,616910
PLA2G6	Infantile neuroaxonal dystrophy 1,256600 Neurodegeneration with brain iron accumulation 2B,610217 Parkinson disease 14,autosomal recessive,612953
PLA2G7	Platelet-activating factor acetylhydrolase deficiency,614278
ADAMTS10	Weill-Marchesani syndrome 1,recessive,277600
ADAMTS15	-
ADAMTS13	Thrombotic thrombocytopenic purpura,familial,274150
RDH12	Leber congenital amaurosis 13,612712
RNF216	Cerebellar ataxia and hypogonadotropic hypogonadism,212840
HLCS	Holocarboxylase synthetase deficiency,253270
RDH11	?Retinal dystrophyjuvenile cataractsand short stature syndrome

ADAMTS18	Microcornea,myopic chorioretinal atrophy,and telecanthus,615458
ADAMTS17	Weill-Marchesani-like syndrome,613195
PIP5K1C	Lethal congenital contractural syndrome 3,611369
ATG7	-
ATG5	?Spinocerebellar ataxiaautosomal recessive 25
TOE1	Pontocerebellar hypoplasia,type 7
JUP	Naxos disease,601214
FUCA1	Fucosidosis,230000
EIF2AK3	Wolcott-Rallison syndrome,226980
LARP7	Alazami syndrome,615071
EIF2AK4	Pulmonary venoocclusive disease 2,234810
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency,613839
PPFIBP1	-
B4GAT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,13
STRC	Deafness,autosomal recessive 16,603720
SFXN4	Combined oxidative phosphorylation deficiency 18,615578
F13B	Factor XIIIB deficiency,613235
FAT4	Hennekam lymphangiectasia-lymphedema syndrome 2,616006 Van Maldergem syndrome 2,615546
RNF220	-
DPYS	Dihydropyrimidinuria,222748
NCAPG2	Khan-Khan-Katsanis syndrome
SNX10	Osteopetrosis,autosomal recessive 8,615085
HOXC13	Ectodermal dysplasia 9,hair/nail type,614931
MMP21	Heterotaxy,visceral,7,autosomal,616749
MED17	Microcephaly,postnatal progressive,with seizures and brain atrophy,613668
MMP20	Amelogenesis imperfecta,type IIA2,612529
CASP14	Ichthyosiscongenitalautosomal recessive 12
ATIC	AICA-ribosiduria due to ATIC deficiency,608688
SMPD4	Neurodevelopmental disorder with microcephaly,arthrogryposis,and structural brain anomalies
SMPD1	Niemann-Pick disease,type A,257200 Niemann-Pick disease,type B,607616
JAK3	SCID,autosomal recessive,T-negative/B-positive type,600802
SLC18A2	?Parkinsonism-dystoniainfantile2
SLC18A3	Myasthenic syndromecongenital21presynaptic
APC2	Sotos syndrome 3
CHST8	?Peeling skin syndrome 3
CHST6	Macular corneal dystrophy,217800
MYBPC1	Lethal congenital contracture syndrome 4,614915
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,7,614643 Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,7,616052
IFT140	Short-rib thoracic dysplasia 9 with or without polydactyly,266920
SLC39A13	Ehlers-Danlos syndrome-like,612350

MED27	-
SLC39A14	Hypermanganesemia with dystonia 2,617013
DDB2	Xeroderma pigmentosum,group E,DDB-negative subtype,278740
MED23	Mental retardation,autosomal recessive 18,614249
MED25	Basel-Vanagait-Smirin-Yosef syndrome,616449 ?Charcot-Marie-Tooth disease,type 2B2,605589
MMP14	?Winchester syndrome
MMP13	Metaphyseal dysplasia,Spahr type,250400
SLC7A7	Lysinuric protein intolerance,222700
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency
SLC7A9	Cystinuria,220100
PTRH2	Infantile-onset multisystem neurologic,endocrine,and pancreatic disease,616263
AGPS	Rhizomelic chondrodysplasia punctata,type 3,600121
DPYD	5-fluorouracil toxicity,274270 Dihydropyrimidine dehydrogenase deficiency,274270
PXDN	Corneal opacification and other ocular anomalies,269400
SDHAF1	Mitochondrial complex II deficiency,252011
L2HGDH	L-2-hydroxyglutaric aciduria,236792
CRYAA	Cataract 9,multiple types,604219
CRYAB	Myopathy,myofibrillar,fatal infantile hypertonic,alpha-B crystallin-related
CHST3	Spondyloepiphyseal dysplasia with congenital joint dislocations,143095
CRB2	Focal segmental glomerulosclerosis 9,616220 Ventriculomegaly with cystic kidney disease,219730
CRB1	Retinitis pigmentosa-12,autosomal recessive,600105
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency,616277
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia,258870
SAMD9	Tumoral calcinosis,familial,normophosphatemic,610455
ADAM22	Epileptic encephalopathy,early infantile,61
ABCB11	Cholestasis,benign recurrent intrahepatic,2,605479 Cholestasis,progressive familial intrahepatic 2,601847
EGFR	Inflammatory skin and bowel disease,neonatal,2,616069
WRN	Werner syndrome,277700
MFN2	Charcot-Marie-Tooth disease,axonal,type 2A2B,617087
ZNF469	Brittle cornea syndrome 1,229200
IL12RB1	Immunodeficiency 30,614891
P4HTM	Hypotonia,hypoventilation,impaired intellectual development,dysautonomia,epilepsy,and eye abnormalities
JAM2	Basal ganglia calcification, idiopathic, 8, autosomal recessiveutosomal recessive
JAM3	Hemorrhagic destruction of the brain,subependymal calcification,and cataracts,613730
ATAD1	Hyperekplexia 4

CRBN	Mental retardation,autosomal recessive 2,607417
PKLR	Pyruvate kinase deficiency,266200
WNT7A	Fuhrmann syndrome,228930 Ulna and fibula,absence of,with severe limb deficiency,276820
CENPE	?Microcephaly 13primaryautosomal recessive
CENPF	Stromme syndrome,243605
COL3A1	Polymicrogyria with or without vascular-type EDS
CCDC8	3-M syndrome 3,614205
CENPJ	Microcephaly 6,primary,autosomal recessive,608393 ?Seckel syndrome 4,613676
SNX14	Spinocerebellar ataxia,autosomal recessive 20,616354
NCAPD2	?Microcephaly 21primaryautosomal recessive
NCAPD3	Microcephaly 22primaryautosomal recessive
AGRN	Myasthenic syndrome,congenital,8,with pre- and postsynaptic defects,615120
PTF1A	Pancreatic agenesis 2,615935 Pancreatic and cerebellar agenesis,609069
CRAT	?Neurodegeneration with brain iron accumulation 8
CRLF1	Cold-induced sweating syndrome 1,272430
LIMS2	Muscular dystrophy,limb-girdle,type 2W,616827
GZF1	Joint laxityshort statureand myopia
HYCC1	Leukodystrophy,hypomyelinating,5,610532
PLOD3	Lysyl hydroxylase 3 deficiency,612394
HIKESHI	Leukodystrophy,hypomyelinating,13,616881
PLOD2	Bruck syndrome 2,609220
PLOD1	Ehlers-Danlos syndrome,type VI,225400
EFEMP2	Cutis laxa,autosomal recessive,type IB,614437
UROD	Porphyria cutanea tarda,176100 Porphyria,hepatoerythropoietic,176100
NEK1	Short-rib thoracic dysplasia 6 with or without polydactyly,263520
NEK2	?Retinitis pigmentosa 67
MTPAP	Ataxia,spastic,4,613672
TNFRSF4	?Immunodeficiency 16
PCYT2	-
DGAT1	?Diarrhea 7,protein-losing enteropathy type
CNPY3	Epileptic encephalopathy,early infantile,60,617929
MSH6	Mismatch repair cancer syndrome,276300
RINT1	Infantile liver failure syndrome 3
MSH2	Mismatch repair cancer syndrome,276300
SMO	Pallister-Hall-like syndrome
MSH3	Familial adenomatous polyposis 4
PKP1	Ectodermal dysplasia/skin fragility syndrome,604536
IL6ST	Hyper-IgE recurrent infection syndrome 4,autosomal recessive
SLC27A4	Ichthyosis prematurity syndrome,608649
COL17A1	Epidermolysis bullosa,junctional,localisata variant,226650 Epidermolysis bullosa,junctional,non-Herlitz type,226650

FTO	Growth retardation,developmental delay,facial dysmorphism,612938 {Obesity,susceptibility to,BMIQ14},612460
BLM	Bloom syndrome,210900
KCNE1	Jervell and Lange-Nielsen syndrome 2,612347 Long QT syndrome 5,613695
UQCRB	Mitochondrial complex III deficiency,nuclear type 3,615158
DLX5	?Split-hand/foot malformation 1 with sensorineural hearing loss
IREB2	Neurodegeneration,early-onset,with choreoathetoid movements and microcytic anemia
KMT2B	Intellectual disability
CORO1A	Immunodeficiency 8,615401
SPATA7	Leber congenital amaurosis 3,604232 Retinitis pigmentosa,juvenile,autosomal recessive,604232
LOXHD1	Deafness,autosomal recessive 77,613079
AP4S1	Spastic paraplegia 52,autosomal recessive,614067
KDSR	-
ZNF423	Joubert syndrome 19,614844 Nephronophthisis 14,614844
NEK8	Renal-hepatic-pancreatic dysplasia 2 615415
NEK9	Lethal congenital contracture syndrome 10,617022 Nevus comedonicus,somatic,617025 ?Arthrogryposis,Perthes disease,and upward gaze palsy,614262
UROS	Porphyria,congenital erythropoietic,263700
HPS1	Hermansky-Pudlak syndrome 1,203300
HPS4	Hermansky-Pudlak syndrome 4,614073
HPS3	Hermansky-Pudlak syndrome 3,614072
HPS6	Hermansky-Pudlak syndrome 6,614075
RFXANK	MHC class II deficiency,complementation group B,209920
HPS5	Hermansky-Pudlak syndrome 5,614074
G6PC1	Glycogen storage disease Ia,232200
FBXO31	Mental retardation,autosomal recessive 45
TMPRSS15	Enterokinase deficiency,226200
YRDC	-
BFSP2	Cataract 12,multiple types,611597
BFSP1	Cataract 33,611391
G6PC3	Dursun syndrome,612541 Neutropenia,severe congenital 4,autosomal recessive,612541
UQCRQ	Mitochondrial complex III deficiency,nuclear type 4,615159
DMXL2	?Polyendocrine-polyneuropathy syndrome
TBXAS1	Ghosal hematodiaphyseal syndrome,231095 ?Thromboxane synthase deficiency,614158
ADSS1	Myopathy,distal,5,617030
ACO2	Infantile cerebellar-retinal degeneration,614559 ?Optic atrophy 9,616289
SP7	?Osteogenesis imperfectatype XII
NOP10	Dyskeratosis congenita,autosomal recessive 1,224230
MIPEP	Combined oxidative phosphorylation deficiency 31
CIITA	Bare lymphocyte syndrome,type II,complementation group A,209920 {Rheumatoid arthritis,susceptibility to},180300

MCFD2	Factor V and factor VIII,combined deficiency of,613625
ITGB4	Epidermolysis bullosa of hands and feet,131800 Epidermolysis bullosa,junctional,non-Herlitz type,226650 Epidermolysis bullosa,junctional,with pyloric atresia,226730
ITGB3	Glanzmann thrombasthenia,273800 Purpura,posttransfusion Thrombocytopenia,neonatal alloimmune {Myocardial infarction,susceptibility to},608446
ITGB2	Leukocyte adhesion deficiency,116920
RMND1	Combined oxidative phosphorylation deficiency 11,614922
DYSF	Miyoshi muscular dystrophy 1,254130 Muscular dystrophy,limb-girdle,type 2B,253601 Myopathy,distal,with anterior tibial onset,606768
ETFA	Glutaric acidemia IIA,231680
ETFB	Glutaric acidemia IIB,231680
TSPAN12	Exudative vitreoretinopathy 5,613310
RLBP1	Bothnia retinal dystrophy,607475 Fundus albipunctatus,136880 Newfoundland rod-cone dystrophy,607476 Retinitis punctata albescens,136880
ALAD	Porphyria,acute hepatic,612740
AP4M1	Spastic paraplegia 50,autosomal recessive,612936
UNC80	Hypotonia,infantile,with psychomotor retardation and characteristic facies 2,616801
KIAA1549	Retinitis pigmentosa 86
SPR	Dystonia,dopa-responsive,due to sepiapterin reductase deficiency,612716
ZNF408	Retinitis pigmentosa 72,616469 ?Exudative vitreoretinopathy 6,616468
PGM3	Immunodeficiency 23,615816
ACP5	Spondyloenchondrodysplasia with immune dysregulation,607944
ITGB6	Amelogenesis imperfecta,type IH,616221
CEP290	Joubert syndrome 5,610188 Leber congenital amaurosis 10,611755 Meckel syndrome 4,611134 Senior-Loken syndrome 6,610189 ?Bardet-Biedl syndrome 14,615991
SNAP29	Cerebral dysgenesis,neuropathy,ichthyosis,and palmoplantar keratoderma syndrome,609528
NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia,613330
PGM1	Congenital disorder of glycosylation,type It,614921
RMRP	Anauxetic dysplasia,607095 Cartilage-hair hypoplasia,250250 Metaphyseal dysplasia without hypotrichosis,250460
CERS3	Ichthyosis,congenital,autosomal recessive 9,615023
BCAS3	Hengel-Maroofian-Schols syndrome
GRID2	Spinocerebellar ataxia,autosomal recessive 18,616204
ADGRV1	Usher syndrome,type 2C,605472
ITGA3	Interstitial lung disease,nephrotic syndrome,and epidermolysis bullosa,congenital,614748
RNASET2	Leukoencephalopathy,cystic,without megalencephaly,612951
VPS13C	Parkinson disease 23,autosomal recessive,early onset,616840

DHPS	Neurodevelopmental disorder with seizures and speech and walking impairment
VPS13D	Spinocerebellar ataxia,autosomal recessive 4
VPS13A	Choreoacanthocytosis,200150
VPS13B	Cohen syndrome,216550
SPTB	Anemia,neonatal hemolytic,fatal and near-fatal Elliptocytosis-3 Spherocytosis,type 2,616649
LRTOMT	Deafness,autosomal recessive 63,611451
MFRP	Microphthalmia,isolated 5,611040 Nanophthalmos 2,609549
NEMF	-
PCCA	Propionicacidemia,606054
RRM2B	Mitochondrial DNA depletion syndrome 8B (MNGIE type) ,612075
MYO3A	Deafness,autosomal recessive 30,607101
PCCB	Propionicacidemia,606054
ITGA8	Renal hypodysplasia/aplasia 1,191830
ITGA7	Muscular dystrophy,congenital,due to ITGA7 deficiency,613204
ITGA6	Epidermolysis bullosa,junctional,with pyloric stenosis,226730
AGXT	Hyperoxaluria,primary,type 1,259900
FXN	Friedreich ataxia with retained reflexes,229300 Friedreich ataxia,229300
CERS1	?Epilepsyprogressive myoclonic8
CANT1	Desbuquois dysplasia 1,251450
C1S	C1s deficiency,613783
TBXT	Sacral agenesis with vertebral anomalies
FOXRED1	Leigh syndrome due to mitochondrial complex I deficiency,256000 Mitochondrial complex I deficiency,252010
MALT1	Immunodeficiency 12,615468
CDC45	Meier-Gorlin syndrome 7,617063
ARL13B	Joubert syndrome 8,612291
PHKG2	Glycogen storage disease IXc,613027
BSND	Bartter syndrome,type 4a,602522 Sensorineural deafness with mild renal dysfunction,602522
DRAM2	Cone-rod dystrophy 21,616502
KCNJ1	Bartter syndrome,type 2,241200
NTRK1	Insensitivity to pain,congenital,with anhidrosis,256800 Medullary thyroid carcinoma,familial,155240
NAGA	Kanzaki disease,609242 Schindler disease,type I,609241 Schindler disease,type III,609241
SYT14	Spinocerebellar ataxia,autosomal recessive 11,614229
TBX6	Spondylocostal dysostosis 5
MYO1E	Glomerulosclerosis,focal segmental,6,614131
MCM3AP	Peripheral neuropathy,autosomal recessive,with or without impaired intellectual development
PINK1	Parkinson disease 6,early onset,605909
NECAP1	Epileptic encephalopathy,early infantile,21

SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria),612073
UROC1	Urocanase deficiency
IL2RA	Immunodeficiency 41 with lymphoproliferation and autoimmunity,606367
IL2RB	Immunodeficiency 63 with lymphoproliferation and autoimmunity
STUB1	Spinocerebellar ataxia,autosomal recessive 16,615768
TRIM37	Mulibrey nanism,253250
CD247	Immunodeficiency 25 610163
EIF3F	Mental retardation,autosomal recessive 67
NAGS	N-acetylglutamate synthase deficiency,237310
TRIM32	Muscular dystrophy,limb-girdle,type 2H,254110 ?Bardet-Biedl syndrome 11,615988
DMGDH	Dimethylglycine dehydrogenase deficiency,605850
ODAD1	Ciliary dyskinesia,primary,20,615067
SMARCAL1	Schimke immunoosseous dysplasia,242900
ODAD3	Ciliary dyskinesia,primary,30,616037
ODAD2	Ciliary dyskinesia,primary,23,615451
ODAD4	Ciliary dyskinesia,primary,35,617092
NUDT2	-
RCBTB1	Retinal dystrophy with or without extraocular anomalies
PEX26	Peroxisome biogenesis disorder 7A (Zellweger),614872 Peroxisome biogenesis disorder 7B,614873
C4B	C4B deficiency,614379
C4A	C4a deficiency 614380
HERC2	Mental retardation,autosomal recessive 38,615516 [Skin/hair/eye pigmentation 1,blond/brown hair],227220[Skin/hair/eye pigmentation 1,blue/nonblue eyes],227220
HERC1	Macrocephaly,dysmorphic facies,and psychomotor retardation,617011
MCIDAS	-
PIEZO2	Arthrogryposis,distal,with impaired proprioception and touch,617146
PIEZO1	Lymphatic malformation 6
TNFSF11	Osteopetrosis,autosomal recessive 2,259710
ATP6V1E1	Cutis laxaautosomal recessivetype IIC
FBXO7	Parkinson disease 15,autosomal recessive,260300
SCAPER	Retinitis Pigmentosa with intellectual disability
MFSD2A	Microcephaly 15,primary,autosomal recessive,616486
ALG8	Congenital disorder of glycosylation,type Ih,608104
ALG9	Congenital disorder of glycosylation,type Il,608776 Gillessen-Kaesbach-Nishimura syndrome,263210
ALG6	Congenital disorder of glycosylation,type Ic,603147
ALG2	Myasthenic syndrome,congenital,14,with tubular aggregates,616228 ?Congenital disorder of glycosylation,type Ii,607906
ALG3	Congenital disorder of glycosylation,type Id,601110

TALDO1	Transaldolase deficiency,606003
MOGS	Congenital disorder of glycosylation,type IIb,606056
ALG1	Congenital disorder of glycosylation,type Ik,608540
MYO7A	Deafness,autosomal recessive 2,600060 Usher syndrome,type 1B,276900
BCS1L	Bjornstad syndrome,262000 GRACILE syndrome,603358 Leigh syndrome,256000 Mitochondrial complex III deficiency,nuclear type 1,124000
BTD	Biotinidase deficiency,253260
PPA2	?Sudden cardiac failure,alcohol-induced 617223,Sudden cardiac failure,infantile 617222
KCNQ1	Jervell and Lange-Nielsen syndrome,220400
RIN2	Macrocephaly,alopecia,cutis laxa,and scoliosis,613075
CERKL	Retinitis pigmentosa 26,608380
AARS1	Epileptic encephalopathy,early infantile,29
SLC45A2	Albinism,oculocutaneous,type IV,606574
AARS2	Combined oxidative phosphorylation deficiency 8,614096Leukoencephalopathy,progressive,with ovarian failure,615889
EPB42	Spherocytosis,type 5,612690
EPB41	Elliptocytosis-1,611804
SLC45A1	Intellectual developmental disorder with neuropsychiatric features
KALRN	Intellectual disability and short stature
PKHD1	Polycystic kidney and hepatic disease,263200
BRIP1	Fanconi anemia,complementation group J
MC2R	Glucocorticoid deficiency,due to ACTH unresponsiveness,202200
PPCS	Cardiomyopathy,dilated,2C
NEU1	Sialidosis,type I,256550 Sialidosis,type II,256550
B4GALNT1	Spastic paraplegia 26,autosomal recessive,609195
APOE	Sea-blue histiocyte disease,269600
PCK1	Phosphoenolpyruvate carboxykinase deficiency,cytosolic 261680
PCK2	PEPCK deficiency,mitochondrial 261650
BBS2	Bardet-Biedl syndrome 2,615981 Retinitis pigmentosa 74,616562
BBS1	Bardet-Biedl syndrome 1,209900
NTNG2	Neurodevelopmental disorder with behavioral abnormalities, absent speech, and hypotonia
STAT5B	Growth hormone insensitivity with immunodeficiency,245590
EGR2	Dejerine-Sottas disease,145900 Neuropathy,congenital hypomyelinating,1,605253
HARS1	Usher syndrome type 3B,614504
SLC10A7	Short stature,amelogenesis imperfecta,and skeletal dysplasia with scoliosis
TGDS	Catel-Manzke syndrome,616145
MYO5A	Griscelli syndrome,type 1,214450
RAB27A	Griscelli syndrome,type 2,607624

CPLANE1	Joubert syndrome 17,614615 Orofaciodigital syndrome VI,277170
NBEAL2	Gray platelet syndrome,139090
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency
HARS2	?Perrault syndrome 2
AIMP1	Leukodystrophy,hypomyelinating,3,260600
AIMP2	Leukodystrophyhypomyelinating17
EXT2	?Seizures,scoliosis,and macrocephaly syndrome
ALDH6A1	Methylmalonate semialdehyde dehydrogenase deficiency,614105
EPRS1	Leukodystrophy,hypomyelinating,15 ,617951
NDUFAF6	Leigh syndrome due to mitochondrial complex I deficiency,256000
LPAR6	Hypotrichosis 8,278150 Woolly hair,autosomal recessive 1,with or without hypotrichosis,278150
MYO5B	Microvillus inclusion disease,251850
NDUFAF4	Mitochondrial complex I deficiency,252010
CWF19L1	Spinocerebellar ataxia,autosomal recessive 17
NDUFAF5	Mitochondrial complex 1 deficiency,252010
NDUFAF2	Leigh syndrome,256000 Mitochondrial complex I deficiency,252010
NDUFAF3	Mitochondrial complex I deficiency,252010
FBXL3	Intellectual developmental disorder with short stature,facial anomalies,and speech defects
GRAP	Deafness,autosomal recessive 114
NDUFAF1	Mitochondrial complex I deficiency,252010
FBXL4	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type),615471
TP73	Ciliary dyskinesia, primary, 47, and lissencephaly
IGHM	Agammaglobulinemia 1,601495
ECM1	Urbach-Wiethe disease,247100
MRPS16	Combined oxidative phosphorylation deficiency 2,610498
ACY1	Aminoacylase 1 deficiency,609924
PYROXD1	Myopathymyofibrillar8
AP4E1	Spastic paraplegia 51,autosomal recessive,613744 Stuttering,familial persistent,1,184450
GRIK2	Mental retardation,autosomal recessive,6,611092
C8B	C8 deficiency,type II,613789
C8A	C8 deficiency,type I,613790
PCLO	Pontocerebellar hypoplasia,type 3
OTUD6B	Intellectual developmental disorder with dysmorphic facies,seizures,and distal limb anomalies
SUFU	Joubert syndrome 32,617757
GNRHR	Hypogonadotropic hypogonadism 7 without anosmia,146110
BBS9	Bardet-Biedl syndrome 9,615986
EPG5	Vici syndrome,242840
PHGDH	Neu-Laxova syndrome 1,256520 Phosphoglycerate dehydrogenase deficiency,601815
BBS7	Bardet-Biedl syndrome 7,615984

NANS	Sponyloepimetaphyseal dysplasia,Genevieve type,610442
BBS5	Bardet-Biedl syndrome 5,615983
BBS4	Bardet-Biedl syndrome 4,615982
ATPAF2	Mitochondrial complex V (ATP synthase) deficiency,nuclear type 1,604273
COX8A	?Mitochondrial complex IV deficiency
SFTPB	Surfactant metabolism dysfunction,pulmonary,1,265120
PCYT1A	Spondylometaphyseal dysplasia with cone-rod dystrophy,608940
FSHB	Hypogonadotropic hypogonadism 24 without anosmia,229070
GAA	Glycogen storage disease II,232300
MRPS22	Combined oxidative phosphorylation deficiency 5,611719
KIF7	Acrocallosal syndrome,200990 Joubert syndrome 12,200990 ?Al-Gazali-Bakalinova syndrome,607131 ?Hydrolethalus syndrome 2,614120
APRT	Adenine phosphoribosyltransferase deficiency,614723
GAN	Giant axonal neuropathy-1,256850
FSHR	Ovarian dysgenesis 1,233300 Ovarian hyperstimulation syndrome,608115 Ovarian response to FSH stimulation,276400
UFC1	Neurodevelopmental disorder with spasticity and poor growth
ARHGEF2	?Neurodevelopmental disorder with midbrain and hindbrain malformations
MICU1	Myopathy with extrapyramidal signs,615673
KIZ	Retinitis pigmentosa 69,615780
DDR2	Spondylometaepiphyseal dysplasia,short limb-hand type,271665
DYNC1I2	Neurodevelopmental disorder with microcephaly and structural brain anomalies
NAPB	-
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase,613752
MRPS34	Combined oxidative phosphorylation deficiency 32,617664
HSD17B3	Pseudohermaphroditism,male,with gynecomastia,264300
HSD17B4	D-bifunctional protein deficiency,261515 Perrault syndrome 1,233400
FITM2	Siddiqi syndrome
PRICKLE1	Epilepsy,progressive myoclonic 1B,612437
CYP17A1	17,20-lyase deficiency,isolated,202110 17-alpha-hydroxylase/17,20-lyase deficiency,202110
RELB	?Immunodeficiency 53
RELN	Lissencephaly 2 (Norman-Roberts type),257320 {Epilepsy,familial temporal lobe,7},616436
SLX4	Fanconi anemia,complementation group P,613951
IGKC	Kappa light chain deficiency,614102
MGAT2	Congenital disorder of glycosylation,type IIa,212066
PCNT	Microcephalic osteodysplastic primordial dwarfism,type II,210720

XDH	Xanthinuria,type I,278300
PEX16	Peroxisome biogenesis disorder 8A,(Zellweger),614876 Peroxisome biogenesis disorder 8B,614877
PEX19	Peroxisome biogenesis disorder 12A (Zellweger),614886
PNPLA8	?Mitochondrial myopathy with lactic acidosis
GAD1	Cerebral palsy,spastic quadriplegic,1
KLHL3	Pseudohypoaldosteronism,type IID,614495
PEX10	Peroxisome biogenesis disorder 6A (Zellweger),614870 Peroxisome biogenesis disorder 6B,614871
PEX12	Peroxisome biogenesis disorder 3A (Zellweger),614859 Peroxisome biogenesis disorder 3B,266510
APTX	Ataxia,early-onset,with oculomotor apraxia and hypoalbuminemia,208920
MYO9A	Myasthenic syndrome,congenital,24,presynaptic
PEX13	Peroxisome biogenesis disorder 11A (Zellweger),614883 Peroxisome biogenesis disorder 11B,614885
GCK	Diabetes mellitus,permanent neonatal,606176
PEX14	Peroxisome biogenesis disorder 13A (Zellweger),614887
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures,autosomal recessive
YIF1B	Kaya-Barakat-Masson syndrome
RP1L1	Retinitis pigmentosa 88
ITCH	Autoimmune disease,multisystem,with facial dysmorphism,613385
TTC8	Bardet-Biedl syndrome 8,615985 ?Retinitis pigmentosa 51,613464
KLHL7	PERCHING syndrome
OTULIN	Autoinflammation,panniculitis,and dermatosis syndrome,617099
TCAP	Muscular dystrophy,limb-girdle,type 2G,601954
PNPLA6	Boucher-Neuhauser syndrome,215470 Oliver-McFarlane syndrome,275400 Spastic paraplegia 39,autosomal recessive,612020 ?Laurence-Moon syndrome,245800
RTN4IP1	Optic atrophy 10 with or without ataxia,mental retardation,and seizures,616732
PNPLA1	Ichthyosis,congenital,autosomal recessive 10,615024
PNPLA2	Neutral lipid storage disease with myopathy,610717
NUP188	Sandestig-Stefanova syndrome
TACO1	Mitochondrial complex IV deficiency,220110
PRDM5	Brittle cornea syndrome 2,614170
INPPL1	Opsismodysplasia,258480
BUB1B	Mosaic variegated aneuploidy syndrome 1
MSMO1	Microcephaly,congenital cataract,and psoriasiform dermatitis,616834
TMEM260	Structural heart defects and renal anomalies syndrome
UBE3B	Kaufman oculocerebrofacial syndrome,244450
COX6A1	Charcot-Marie-Tooth disease,recessive intermediate D,616039
YARS1	-
COX6A2	Mitochondrial complex IV deficiency

YARS2	Myopathy,lactic acidosis,and sideroblastic anemia 2,613561
IGF1R	Insulin-like growth factor I,resistance to,270450
TGM1	Ichthyosis,congenital,autosomal recessive 1,242300
GRIP1	Fraser syndrome,617667
GNPTAB	Mucolipidosis II alpha/beta,252500 Mucolipidosis III alpha/beta,252600
PPIP5K2	Deafness,autosomal recessive 100
ENPP1	Arterial calcification,generalized,of infancy,1,208000
RXYLT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,10,615041
DENND5A	Epileptic encephalopathy,early infantile,49
RAG2	Combined cellular and humoral immune defects with granulomas,233650 Omenn syndrome,603554 Severe combined immunodeficiency,B cell-negative,601457
TGM5	Peeling skin syndrome 2,609796
RAG1	Severe combined immunodeficiency,B cell-negative,601457
CAST	Peeling skin with leukonychia,acral punctate keratoses,cheilitis,and knuckle pads,616295
CASR	Hyperparathyroidism,neonatal,239200
SPINK5	Atopy,147050 Netherton syndrome,256500
SDHD	Mitochondrial complex II deficiency
TTC7A	Gastrointestinal defects and immunodeficiency syndrome,243150
SDHA	Mitochondrial respiratory chain complex II deficiency,252011 Paragangliomas 5,614165
SDHB	Mitochondrial complex II deficiency, nuclear type 4
COX6B1	Mitochondrial complex IV deficiency,220110
DHDDS	Retinitis pigmentosa 59,613861
SCNN1G	Pseudohypoaldosteronism,type I,264350
ADAM17	?Inflammatory skin and bowel diseaseneonatal1
SHQ1	-
TYROBP	Nasu-Hakola disease,221770
TBC1D20	Warburg micro syndrome 4,615663
SCNN1B	Pseudohypoaldosteronism,type I,264350
SCNN1A	Pseudohypoaldosteronism,type I,264350
TBC1D24	Deafness ,autosomal recessive 86,614617 Deafness,autosomal dominant 65,616044 DOOR syndrome,220500 Epileptic encephalopathy,early infantile,16,615338 Myoclonic epilepsy,infantile,familial,605021
ANKS6	Nephronophthisis 16,615382
PLAA	Neurodevelopmental disorder with progressive microcephalyspasticityand brain anomalies
TBC1D23	Pontocerebellar hypoplasia,type 11,617695
PPIB	Osteogenesis imperfecta,type IX,259440
CARS2	Combined oxidative phosphorylation deficiency 27,616672
CARD11	Immunodeficiency 11,615206
PKDCC	-
COX16	-

COX15	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 2,615119 Leigh syndrome due to cytochrome c oxidase deficiency,256000
NDUFB11	Mitochondrial complex I deficiency,252010
UBA5	Epileptic encephalopathy,early infantile,44
CACNA1B	Neurodevelopmental disorder with seizures and nonepileptic hyperkinetic movements
HTRA1	CARASIL syndrome,600142 Cerebral arteriopathy,autosomal dominant,with subcortical infarcts and leukoencephalopathy,type 2,616779 {Macular degeneration,age-related,7},610149 {Macular degeneration,age-related,neovascular type},610149
HTRA2	3-methylglutaconic aciduriatype VIII
CACNA1D	Sinoatrial node dysfunction and deafness
PLD1	Cardiac valvular defectdevelopmental
NUP160	?Nephrotic syndrome,type 19
TIMM50	3-methylglutaconic aciduriatype IX
GRK1	Oguchi disease-2,613411
ADGRG1	Polymicrogyria,bilateral frontoparietal
KCNV2	Retinal cone dystrophy 3B,610356
RNASEH1	Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal recessive 2,616479
ADGRG6	Lethal congenital contracture syndrome 9,616503
FGF20	?Renal hypodysplasia/aplasia 2
COX14	?Mitochondrial complex IV deficiency
IL36RN	Psoriasis 14,pustular,614204
CACNA1S	-
FGF23	Tumoral calcinosis,hyperphosphatemic,familial,211900
PIBF1	Joubert syndrome 33
COX10	Leigh syndrome due to mitochondrial COX4 deficiency,256000 Mitochondrial complex IV deficiency,220110
BHLHA9	Syndactyly,mesoaxial synostotic,with phalangeal reduction,609432 ?Camptosynpolydactyly,complex,607539
CRTAP	Osteogenesis imperfecta,type VII,610682
SYT2	-
CAV1	Lipodystrophy,congenital generalized,type 3,612526
ADA2	Polyarteritis nodosa,childhood-onset,615688 ?Sneddon syndrome,182410
USB1	Poikiloderma with neutropenia,604173
AP4B1	Spastic paraplegia 47,autosomal recessive,614066
BLTP1	Alkuraya-Kucinskas syndrome,617822
TMEM231	Joubert syndrome 20,614970 Meckel syndrome 11,615397
AHI1	Joubert syndrome-3,608629
GH1	Growth hormone deficiency,isolated,type IA,262400 Growth hormone deficiency,isolated,type IB,612781 Growth hormone deficiency,isolated,type II,173100 Kowarski syndrome,262650
TMEM237	Joubert syndrome 14,614424
UBE2T	Fanconi anemia,complementation group T,616435

UVSSA	UV-sensitive syndrome 3,614640
CNTN1	Myopathy,congenital,Compton-North 612540
ALPL	Hypophosphatasia,adult,146300 Hypophosphatasia,childhood,241510 Hypophosphatasia,infantile,241500
CNTN2	?Epilepsymyoclonicfamilial adult5
GNB3	Night blindness,congenital stationary,type 1H,617024 {Hypertension,essential,susceptibility to},145500
IFT27	?Bardet-Biedl syndrome 19
DEGS1	Leukodystrophy,hypomyelinating,18
GNB5	Intellectual developmental disorder with cardiac arrhythmia
IL7R	Severe combined immunodeficiency,T-cell negative,B-cell/natural killer cell-positive type,608971
DNAL1	Ciliary dyskinesia,primary,16,614017
COX20	Mitochondrial complex IV deficiency,220110
GALK1	Galactokinase deficiency with cataracts,230200
C12orf4	Mental retardation,autosomal recessive 66
TAT	Tyrosinemia,type II,276600
NGLY1	Congenital disorder of deglycosylation,615273
ADAR	Aicardi-Goutieres syndrome 6,615010
MYSM1	Bone marrow failure syndrome 4
GRM1	Spinocerebellar ataxia,autosomal recessive 13,614831
LMNB2	?Epilepsyprogressive myoclonic9
GHR	Growth hormone insensitivity,partial,604271 Increased responsiveness to growth hormone Laron dwarfism,262500{Hypercholesterolemia,familial,modifier of},143890
FYCO1	Cataract 18,autosomal recessive,610019
IFT52	Short-rib thoracic dysplasia 16 with or without polydactyly
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies,614416
GRM7	-
CA2	Osteopetrosis,autosomal recessive 3,with renal tubular acidosis,259730
GRM6	Night blindness,congenital stationary (complete),1B,autosomal recessive,257270
MYO18B	Klippel-Feil syndrome 4,autosomal recessive,with myopathy and facial dysmorphism,616549
CA8	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3,613227
IFT57	?Orofaciodigital syndrome XVIII
DNAI2	Ciliary dyskinesia,primary,9,with or without situs inversus,612444
MKKS	Bardet-Biedl syndrome 6,605231 McKusick-Kaufman syndrome,236700
NUP133	Nephrotic syndrome,type 18 ,?Galloway-Mowat syndrome 8
CAD	Epileptic encephalopathy,early infantile,50
IRAK4	Invasive pneumococcal disease,recurrent isolated,1,610799 IRAK4 deficiency,607676
TENT5A	Osteogenesis imperfecta,type XVIII

TTI2	Mental retardation,autosomal recessive 39,615541
TAPBP	Bare lymphocyte syndrome,type I,604571
ACTA1	Myopathy,actin,congenital,with cores,161800
CYP26C1	Focal facial dermal dysplasia 4,614974
IFT43	Cranioectodermal dysplasia 3,614099
ACOX2	Bile acid synthesis defectcongenital6
ALS2	Amyotrophic lateral sclerosis 2,juvenile,205100 Primary lateral sclerosis,juvenile,606353 Spastic paralysis,infantile onset ascending,607225
ACOX1	Peroxisomal acyl-CoA oxidase deficiency,264470
TMEM216	Joubert syndrome 2,608091 Meckel syndrome 2,603194
CAT	Acatalasemia,614097
TMEM218	-
MAN1B1	Mental retardation,autosomal recessive 15,614202
ZMYND10	Ciliary dyskinesia,primary,22,615444
NAT8L	?N-acetylaspartate deficiency
FBP1	Fructose-1,6-bisphosphatase deficiency,229700
MS4A1	Immunodeficiency,common variable,5,613495
ATP6V1B1	Renal tubular acidosis with deafness,267300
CLRN1	Retinitis pigmentosa 61,614180 Usher syndrome,type 3A,276902
PLEC	Epidermolysis bullosa simplex with pyloric atresia,612138
DNAH1	Ciliary dyskinesia,primary,37
GALT	Galactosemia,230400
NAXE	Encephalopathyprogressiveearly-onsetwith brain edema and/or leukoencephalopathy
DNAH5	Ciliary dyskinesia,primary,3,with or without situs inversus,608644
PKD1L1	Heterotaxyvisceral8autosomal
SLC3A1	Cystinuria,220100
DNAH9	Ciliary dyskinesia,primary,40
RSPH9	Ciliary dyskinesia,primary,12,612650
ADD3	Cerebral palsy,spastic quadriplegic,3,617008
NT5C2	Spastic paraplegia 45,autosomal recessive,613162
PPOX	-
AGPAT2	Lipodystrophy,congenital generalized,type 1,608594
IFT74	?Bardet-Biedl syndrome 20
CBS	Homocystinuria,B6-responsive and nonresponsive types,236200 Thrombosis,hyperhomocysteinemic,236200
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency,245050
DNAI1	Ciliary dyskinesia,primary,1,with or without situs inversus,244400
ARFGEF2	Periventricular heterotopia with microcephaly,608097
HSD3B7	Bile acid synthesis defect,congenital,1,607765
HSD3B2	3-beta-hydroxysteroid dehydrogenase,type II,deficiency,201810
SUOX	Sulfite oxidase deficiency,272300
TBX19	Adrenocorticotropic hormone deficiency,201400
TTC19	Mitochondrial complex III deficiency,nuclear type 2,615157
YY1AP1	Grange syndrome,602531

WARS2	Neurodevelopmental disordermitochondrialwith abnormal movements and lactic acidosiswith or without seizures
GALC	Krabbe disease,245200
GALNS	Mucopolysaccharidosis IVA,253000
RSPH3	Ciliary dyskinesia,primary,32,616481
TBX15	Cousin syndrome,260660
GALE	Galactose epimerase deficiency,230350
RSPH1	Ciliary dyskinesia,primary,24,615481
UFM1	Leukodystrophyhypomyelinating14
ERCC3	Trichothiodystrophy 2,photosensitive,616390 Xeroderma pigmentosum,group B,610651
ERCC4	Fanconi anemia,complementation group Q,615272 Xeroderma pigmentosum,group F,278760 Xeroderma pigmentosum,type F/Cockayne syndrome,278760 ?XFE progeroid syndrome,610965
MYO15A	Deafness,autosomal recessive 3,600316
COL7A1	Epidermolysis bullosa dystrophica,AR,226600
ERCC1	Cerebrooculofacioskeletal syndrome 4,610758
ERCC2	Cerebrooculofacioskeletal syndrome 2,610756 Trichothiodystrophy 1,photosensitive,601675 Xeroderma pigmentosum,group D,278730
ERCC8	Cockayne syndrome,type A,216400 UV-sensitive syndrome 2,614621
ERCC5	Cerebrooculofacioskeletal syndrome 3,616570 Xeroderma pigmentosum,group G,278780 Xeroderma pigmentosum,group G/Cockayne syndrome,278780
ERCC6	Cerebrooculofacioskeletal syndrome 1 ,214150
NAXD	Encephalopathy,progressive,early-onset,with brain edema and/or leukoencephalopathy,2
ANKLE2	Microcephaly 16,primary,autosomal recessive
WIPF1	?Wiskott-Aldrich syndrome 2
XYLT2	Spondyloocular syndrome,605822 {Pseudoxanthoma elasticum,modifier of severity of},264800
XYLT1	Desbuquois dysplasia 2,615777 {Pseudoxanthoma elasticum,modifier of severity of},264800
DMP1	Hypophosphatemic rickets,AR,241520
SLC4A1	Renal tubular acidosis,distal,611590
ADARB1	Neurodevelopmental disorder with hypotonia, microcephaly, and seizures,PMID: 32220291
SLC4A4	Renal tubular acidosis,proximal,with ocular abnormalities,604278
CAPN15	-
GLS	Global developmental delay,progressive ataxia,and elevated glutamine ,Epileptic encephalopathy,early infantile,71
GJA1	Craniometaphyseal dysplasia,autosomal recessive,218400
TFG	?Spastic paraplegia 57,autosomal recessive
CSPP1	Joubert syndrome 21,615636
AP1S1	MEDNIK syndrome,609313
CDK5RAP2	Microcephaly 3,primary,autosomal recessive,604804

GAMT	Cerebral creatine deficiency syndrome 2,612736
NUP214	Encephalopathy,acute,infection-induced,susceptibility to,9}
DNAAF3	Ciliary dyskinesia,primary,2,606763
DNAAF2	Ciliary dyskinesia,primary,10,612518
DDX11	Warsaw breakage syndrome,613398
DNAAF1	Ciliary dyskinesia,primary,13,613193
PRKCD	Autoimmune lymphoproliferative syndrome,type III,615559
ARMC9	Joubert syndrome 30
DNAAF5	Ciliary dyskinesia,primary,18,614874
WDR72	Amelogenesis imperfecta,type IIA3,613211
DNAAF4	Ciliary dyskinesia,primary,25,615482 {Dyslexia,susceptibility to,1},127700
WDR73	Galloway-Mowat syndrome,251300
GTPBP2	Jaberi-Elahi syndrome
GTPBP3	Combined oxidative phosphorylation deficiency 23,616198
NUP93	Nephrotic syndrome,type 12,616892
WDR81	Cerebellar ataxia,mental retardation,and dysequilibrium syndrome 2,610185
ADPRS	Neurodegeneration,childhood-onset,stress-induced,with variable ataxia and seizures
TARS2	?Combined oxidative phosphorylation deficiency 21
FREM1	Manitoba oculotrichoanal syndrome,248450
FREM2	Fraser syndrome,219000
GNE	Nonaka myopathy,605820
CFD	Complement factor D deficiency,613912
LIPT2	Encephalopathy,neonatal severe,with lactic acidosis and brain abnormalities,617668
LIPT1	Lipoyltransferase 1 deficiency,616299
LRPAP1	Myopia 23,autosomal recessive,615431
NUP205	?Nephrotic syndrometype 13
CFH	Complement factor H deficiency,609814
PRKDC	Immunodeficiency 26,with or without neurologic abnormalities,615966
CFI	Complement factor I deficiency,610984
XPA	Xeroderma pigmentosum,group A,278700
PRDM13	Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism
XPC	Xeroderma pigmentosum,group C,278720
BCL10	?Immunodeficiency 37
PRDM12	Neuropathy,hereditary sensory and autonomic,type VIII,616488
WDR62	Microcephaly 2,primary,autosomal recessive,with or without cortical malformations,604317
RNPC3	?Growth hormone deficiency,isolated,type V
GNS	Mucopolysaccharidosis type IIID,252940
BLOC1S6	Hermansky-pudlak syndrome 9,614171
LDHA	Glycogen storage disease XI,612933
GJC2	Leukodystrophy,hypomyelinating,2,608804 Lymphedema,hereditary,IC,613480 Spastic paraplegia 44,autosomal recessive,613206

NTHL1	Familial adenomatous polyposis 3,616415
NUP85	Nephrotic syndrome,type 17
TTPA	Ataxia with isolated vitamin E deficiency,277460
BLOC1S3	Hermansky-Pudlak syndrome 8,614077
LDHD	D-lactic aciduria
ALX3	Frontonasal dysplasia 1,136760
VPS11	Leukodystrophy,hypomyelinating,12,616683
ALX4	Frontonasal dysplasia 2,613451
HMOX1	Heme oxygenase-1 deficiency,614034 {Pulmonary disease,chronic obstructive,susceptibility to},606963
ST3GAL5	Amish infantile epilepsy syndrome,609056
ALX1	Frontonasal dysplasia 3
NCKAP1L	-
NUP88	Fetal akinesia deformation sequence 4
ICOS	Immunodeficiency,common variable,1,607594
PROM1	Retinitis pigmentosa 41,612095
NHEJ1	Severe combined immunodeficiency with microcephaly,growth retardation,and sensitivity to ionizing radiation,611291
CEP78	Cone-rod dystrophy and hearing loss
FDFT1	Squalene synthase deficiency
ST3GAL3	Epileptic encephalopathy,early infantile,15,615006 Mental retardation,autosomal recessive 12,611090
PLK4	Microcephaly and chorioretinopathy,autosomal recessive,2,616171
GP6	Bleeding disorder,platelet-type,11,614201
GNPTG	Mucolipidosis III gamma,252605
GP9	Bernard-Soulier syndrome,type C,231200
GJB2	Deafness,autosomal recessive 1A,220290
ARL2BP	Retinitis pigmentosa with or without situs inversus,615434
TCN2	Transcobalamin II deficiency,275350
GJB3	-
GJB6	Deafness,autosomal recessive 1B,612645
PAH	Phenylketonuria,261600 [Hyperphenylalaninemia,non-PKU mild],261600
MKS1	Bardet-Biedl syndrome 13,615990 Meckel syndrome 1,249000
PPT1	Ceroid lipofuscinosis,neuronal,1,256730
CEP83	Nephronophthisis 18,615862
SNAI2	Waardenburg syndrome,type 2D,608890
ITPA	Epileptic encephalopathy,early infantile,35,616647 [Inosine triphosphatase deficiency],613850
FARSB	Rajab syndrome
CEP57	Mosaic variegated aneuploidy syndrome 2,614114
MESP2	Spondylocostal dysostosis 2,autosomal recessive,608681
GPI	Hemolytic anemia,nonspherocytic,due to glucose phosphate isomerase deficiency,613470
MOCS2	Molybdenum cofactor deficiency B,252160
ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease

PYGM	McArdle disease,232600
SMG9	Heart and brain malformation syndrome,616920
PYGL	Glycogen storage disease VI,232700
LYST	Chediak-Higashi syndrome,214500
SMG8	-
FARS2	Combined oxidative phosphorylation deficiency 14,614946 ?Spastic paraplegia 77,autosomal recessive,617046
MCEE	Methylmalonyl-CoA epimerase deficiency,251120
ADAMTSL4	Ectopia lentis et pupillae,225200 Ectopia lentis,isolated,autosomal recessive,225100
TCTN3	Joubert syndrome 18,614815 Orofaciodigital syndrome IV,258860
TCTN2	Joubert syndrome 24,616654 ?Meckel syndrome 8,613885
ADAMTSL2	Geleophysic dysplasia 1,231050
TCTN1	Joubert syndrome 13,614173
TK2	Mitochondrial DNA depletion syndrome 2 (myopathic type),609560
CEP55	Multinucleated neuronsanhydramniosrenal dysplasiacerebellar hypoplasiaand hydranencephaly
EDARADD	Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type,autosomal recessive,614941
HELLS	Immunodeficiency-centromeric instability-facial anomalies syndrome 4,616911
TPI1	Hemolytic anemia due to triosephosphate isomerase deficiency,615512
WDR35	Cranioectodermal dysplasia 2,613610 Short-rib thoracic dysplasia 7 with or without polydactyly,614091
CIT	Microcephaly 17,primary,autosomal recessive
HADHB	Trifunctional protein deficiency,609015
HADHA	Fatty liver,acute,of pregnancy,609016 HELLP syndrome,maternal,of pregnancy,609016 LCHAD deficiency,609016 Trifunctional protein deficiency,609015
KIFBP	Goldberg-Shprintzen megacolon syndrome,609460
ANGPTL3	Hypobetalipoproteinemia,familial,2,605019
RARB	Microphthalmia,syndromic 12,615524
CEP63	?Seckel syndrome 6
TKT	Short stature,developmental delay,and congenital heart defects,617044
MOCS1	Molybdenum cofactor deficiency A,252150
GAS8	Ciliary dyskinesia,primary,33,616726
GRN	Ceroid lipofuscinosis,neuronal,11,614706
NBAS	Infantile liver failure syndrome 2,616483 Short stature,optic nerve atrophy,and Pelger-Huet anomaly,614800
AP5Z1	Spastic paraplegia 48,autosomal recessive,613647
WDR45B	Neurodevelopmental disorder with spastic quadriplegia and brain abnormalities with or without seizures
PRUNE1	Neurodevelopmental disorder with microcephalyhypotoniaand variable brain anomalies
ADCY6	?Lethal congenital contracture syndrome 8

PAPSS2	Brachyolmia 4 with mild epiphyseal and metaphyseal changes,612847
ADCY5	-
FGD4	Charcot-Marie-Tooth disease,type 4H,609311
GPNMB	Amyloidosis,primary localized cutaneous,3,617920
EPCAM	Diarrhea 5,with tufting enteropathy,congenital,613217
GSC	Short stature,auditory canal atresia,mandibular hypoplasia,skeletal abnormalities,602471
SLC17A5	Salla disease,604369 Sialic acid storage disorder,infantile,269920
ZNF341	Hyper-IgE recurrent infection syndrome 3,autosomal recessive
RSPH4A	Ciliary dyskinesia,primary,11,612649
IL10RB	Inflammatory bowel disease 25,early onset,autosomal recessive,612567 {Hepatitis B virus,susceptibility to},610424
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type),614458
WHRN	Deafness,autosomal recessive 31 607084,Usher syndrome,type 2D 611383
GLYCTK	D-glyceric aciduria,220120
GSS	Glutathione synthetase deficiency,266130 Hemolytic anemia due to glutathione synthetase deficiency,231900
IL10RA	Inflammatory bowel disease 28,early onset,autosomal recessive,613148
ECEL1	Arthrogryposis,distal,type 5D,615065
MOCOS	Xanthinuria,type II,603592
WDR19	?Short-rib thoracic dysplasia 5 with or without polydactyly,?Cranioectodermal dysplasia 4,Nephronophthisis 13,Senior-Loken syndrome 8
ELP1	Dysautonomia,familial,223900
NR1H4	Cholestasis,progressive familial intrahepatic 5,617049
ELP2	Mental retardation,autosomal recessive 58
GNMT	Glycine N-methyltransferase deficiency,606664
SMOC2	Dentin dysplasia,type I,with microdontia and misshapen teeth,125400
GATM	Cerebral creatine deficiency syndrome 3,612718
OSTM1	Osteopetrosis,autosomal recessive 5,259720
IGLL1	Agammaglobulinemia 2,613500
CLCF1	Cold-induced sweating syndrome 2,610313
SMOC1	Microphthalmia with limb anomalies,206920
COQ8B	Nephrotic syndrome,type 9,615573
CEP41	Joubert syndrome 15,614464
COQ8A	Coenzyme Q10 deficiency,primary,4
ZNF335	?Microcephaly 10primaryautosomal recessive
KLKB1	Fletcher factor (prekallikrein) deficiency,612423
MTFMT	Combined oxidative phosphorylation deficiency 15,614947
MRE11	Ataxia-telangiectasia-like disorder,604391
POGLUT1	?Muscular dystrophy,limb-girdle,autosomal recessive 21
CEP19	Morbid obesity and spermatogenic failure,615703
PRORP	Combined oxidative phosphorylation deficiency 54

PROS1	Thrombophilia due to protein S deficiency,autosomal recessive,614514
CEP120	Short-rib thoracic dysplasia 13 with or without polydactyly,616300
PARK7	Parkinson disease 7,autosomal recessive early-onset,606324
CDC14A	Deafness,autosomal recessive 105,616958
FGF3	Deafness,congenital with inner ear agenesis,microtia,and microdontia,610706
EDC3	Mental retardation,autosomal recessive 50
RIMS2	Cone-rod synaptic disorder syndrome, congenital nonprogressive,PMID: 32470375
IPO8	VISS syndrome
MPC1	Mitochondrial pyruvate carrier deficiency,614741
CFL2	Nemaline myopathy 7,autosomal recessive,610687
TNR	-
SLC39A8	Congenital disorder of glycosylation,type IIn,616721
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,6,613154 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation),type B,6,608840
TXNL4A	Burn-McKeown syndrome,608572
SLC39A4	Acrodermatitis enteropathica,201100
IFNAR2	?Immunodeficiency 45
RARS2	Pontocerebellar hypoplasia,type 6,611523
CEP135	Microcephaly 8,primary,autosomal recessive
COL27A1	Steel syndrome
RARS1	Leukodystrophy,hypomyelinating,9,616140
RALGAPA1	Neurodevelopmental disorder with hypotonia, neonatal respiratory insufficiency, and thermodysregulation
TPM3	Nemaline myopathy 1,autosomal dominant or recessive
MAGI2	Nephrotic syndrometype 15
KERA	Cornea plana congenita,recessive,217300
VRK1	Pontocerebellar hypoplasia type 1A,607596
LEMD2	Cataract 46,juvenile-onset,212500
TLE6	Preimplantation embryonic lethality,616814
THOC6	Beaulieu-Boycott-Innes syndrome,613680
EDAR	Ectodermal dysplasia 10B,hypohidrotic/hair/tooth type,autosomal recessive,224900 [Hair morphology 1,hair thickness],612630
SLCO2A1	Hypertrophic osteoarthropathy,primary,autosomal recessive 2,614441
STAMBP	Microcephaly-capillary malformation syndrome,614261
IFNAR1	-
CEP104	Joubert syndrome 25,616781
SLC26A2	Achondrogenesis Ib,600972 Atelosteogenesis II,256050 De la Chapelle dysplasia,256050 Diastrophic dysplasia,222600 Diastrophic dysplasia,broad bone-platyspondylic variant,222600 Epiphyseal dysplasia,multiple,4,226900
SLC26A1	Nephrolithiasis,calcium oxalate 167030
AEBP1	Ehlers-Danlos syndromeclassic-like2

UBR1	Johanson-Blizzard syndrome,243800
ORC4	Meier-Gorlin syndrome 2,613800
TPO	Thyroid dyshormonogenesis 2A,274500
ORC6	Meier-Gorlin syndrome 3,613803
ORC1	Meier-Gorlin syndrome 1,224690
DDRGK1	Spondyloepimetaphyseal dysplasiaShohat type
TTC21B	Nephronophthisis 12,613820 Short-rib thoracic dysplasia 4 with or without polydactyly,613819
MAP3K20	Split-foot malformation with mesoaxial polydactyly,Centronuclear myopathy 6 with fiber-type disproportion
SMN1	Spinal muscular atrophy-1,253300 Spinal muscular atrophy-2,253550 Spinal muscular atrophy-3,253400 Spinal muscular atrophy-4,271150
MPDZ	Hydrocephalus,nonsyndromic,autosomal recessive 2,615219
EIF2B5	Leukoencephalopathy with vanishing white matter,603896 Ovarioleukodystrophy,603896
FANCI	Fanconi anemia,complementation group I,609053
EIF2B4	Leukoencephaly with vanishing white matter,603896 Ovarioleukodystrophy,603896
EIF2B3	Leukoencephalopathy with vanishing white matter,603896
EIF2B2	Leukoencephalopathy with vanishing white matter,603896 Ovarioleukodystrophy,603896
TMEM132E	?Deafness,autosomal recessive 99
FANCL	Fanconi anemia,complementation group L,614083
AMBN	Amelogenesis imperfecta,type IF,616270
TRAPPC4	-
FANCA	Fanconi anemia,complementation group A,227650
SLC4A11	Corneal endothelial dystrophy and perceptive deafness,217400
FANCC	Fanconi anemia,complementation group C,227645
PHKB	Phosphorylase kinase deficiency of liver and muscle,autosomal recessive,261750
FANCB	Fanconi anemia,complementation group B,300514
TRAPPC9	Mental retardation,autosomal recessive 13,613192
FANCE	Fanconi anemia,complementation group E,600901
FANCG	Fanconi anemia,complementation group G,614082
FANCF	Fanconi anemia,complementation group F,603467
LHX3	Pituitary hormone deficiency,combined,3,221750
SYNJ1	Parkinson disease 20,early-onset,615530
REEP2	?Spastic paraplegia 72,autosomal recessive
PLEKHM1	Osteopetrosis,autosomal recessive 6,611497
CPE	-
TPP1	Ceroid lipofuscinosis,neuronal,2,204500 Spinocerebellar ataxia,autosomal recessive 7,609270
REEP6	Retinitis pigmentosa 77
DRC1	Ciliary dyskinesia,primary,21,615294
SLC26A4	Deafness,autosomal recessive 4,with enlarged vestibular aqueduct,600791 Pendred syndrome,274600
SLC26A3	Diarrhea 1,secretory chloride,congenital,214700

LAT	Immunodeficiency 52
CCDC47	Trichohepatoneurodevelopmental syndrome
MCM9	Ovarian dysgenesis 4,616185
CEP164	Nephronophthisis 15,614845
WDR4	Microcephaly,growth deficiency,seizures,and brain malformations ,Galloway-Mowat syndrome 6
TCIRG1	Osteopetrosis,autosomal recessive 1,259700
TRH	Thyrotropin-releasing hormone deficiency 275120
PIK3C2A	Oculoskeletodental syndrome
SYNE4	Deafness,autosomal recessive 76,615540
MTO1	Combined oxidative phosphorylation deficiency 10,614702
FTCD	Glutamate formiminotransferase deficiency,229100
SYNE1	Spinocerebellar ataxia,autosomal recessive 8,610743
ADAMTS2	Ehlers-Danlos syndrome,type VIIC,225410
ADAMTS3	Lymfoedeem,618154
IDH3B	Retinitis pigmentosa 46,612572
PRKG2	-
HMGCS2	HMG-CoA synthase-2 deficiency,605911
SERAC1	3-methylglutaconic aciduria with deafness,encephalopathy,and Leigh-like syndrome,614739
COA8	Mitochondrial complex IV deficiency,220110
EIF2B1	Leukoencephalopathy with vanishing white matter,603896
COA7	Spinocerebellar ataxia,autosomal recessive,with axonal neuropathy 3
LBR	Greenberg skeletal dysplasia 215140
COA6	Cardioencephalomyopathy,fatal infantile,due to cytochrome c oxidase deficiency 4,616501
COA5	?Cardioencephalomyopathyfatal infantiledue to cytochrome c oxidase deficiency 3
IDH3A	-
TPRN	Deafness,autosomal recessive 79,613307
CCDC39	Ciliary dyskinesia,primary,14,613807
ADSL	Adenylosuccinase deficiency,103050
CR2	Immunodeficiency,common variable,7,614699
SLC13A3	Leukoencephalopathy,acute reversible,with increased urinary alpha-ketoglutarate
PLEKHG2	Leukodystrophy and acquired microcephaly with or without dystonia,616763
PROP1	Pituitary hormone deficiency,combined,2,262600
GGPS1	-
RFC1	Cerebellar ataxia,neuropathy,and vestibular areflexia syndrome
UGT1A1	Crigler-Najjar syndrome,type I,218800 Crigler-Najjar syndrome,type II,606785 Hyperbilirubinemia,familial transient neonatal,237900 [Bilirubin,serum level of,QTL1],601816 [Gilbert syndrome],143500
CKAP2L	Filippi syndrome,272440
PLEKHG5	Charcot-Marie-Tooth disease,recessive intermediate C,615376 Spinal muscular atrophy,distal,autosomal recessive,4,611067

BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency,222800
WIPI2	?Intellectual developmental disorder with short stature and variable skeletal anomalies
DNAJC3	?Ataxiacombined cerebellar and peripheralwith hearing loss and diabetes mellitus
SKIC2	Trichohepatoenteric syndrome 2,614602
DNAJC6	Parkinson disease 19,juvenile-onset,615528
LCK	?Immunodeficiency 22
SKIC3	Trichohepatoenteric syndrome 1,222470
POLR1C	Leukodystrophy,hypomyelinating,11,616494 Treacher Collins syndrome 3,248390
POLR1D	Treacher Collins syndrome 2
MCM4	Natural killer cell and glucocorticoid deficiency with DNA repair defect,609981
MCM5	?Meier-Gorlin syndrome 8
LCT	Lactase deficiency,congenital,223000
NSRP1	-
CCDC40	Ciliary dyskinesia,primary,15,613808
CATSPER1	Spermatogenic failure 7,612997
CEBPE	Specific granule deficiency,245480
PLG	Dysplasminogenemia,217090 Plasminogen deficiency,type I,217090
PDHB	Pyruvate dehydrogenase E1-beta deficiency,614111
TULP1	Leber congenital amaurosis 15,613843 Retinitis pigmentosa 14,600132
THSD1	-
COCH	?Deafness,autosomal recessive 110
LMF1	Lipase deficiency,combined,246650
DNAJB2	Spinal muscular atrophy,distal,autosomal recessive,5,614881
MSTO1	Myopathy,mitochondrial,and ataxia
VPS51	Pontocerebellar hypoplasia,type 13
VPS50	-
RAB23	Carpenter syndrome,201000
SVBP	Neurodevelopmental disorder with ataxia,hypotonia,and microcephaly
VPS53	Pontocerebellar hypoplasia,type 2E,615851
IL21R	Immunodeficiency,primary,autosomal recessive,IL21R-related,615207
RAB28	Cone-rod dystrophy 18,615374
SLC13A5	Epileptic encephalopathy,early infantile,25,615905
ALOXE3	Ichthyosis,congenital,autosomal recessive 3,606545
FKTN	Cardiomyopathy,dilated,1X,611615
TUB	?Retinal dystrophy and obesity
MGME1	Mitochondrial DNA depletion syndrome 11,615084
SLC35A1	Congenital disorder of glycosylation,type IIf,603585
PPIL1	Pontocerebellar hypoplasia, type 14
CEACAM16	Deafness,autosomal recessive 113
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,5,613153

SLC35A3	?Arthrogryposismental retardationand seizures
ASCC1	?Spinal muscular atrophy with congenital bone fractures 2
AMFR	-
CEP152	Microcephaly 9,primary,autosomal recessive,614852 Seckel syndrome 5,613823
TPRKB	Galloway-Mowat syndrome 5
GFER	Myopathy,mitochondrial progressive,with congenital cataract,hearing loss,and developmental delay,613076
OTOGL	Deafness,autosomal recessive 84B,614944
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency,614923
FRAS1	Fraser syndrome,219000
POLR3A	Leukodystrophy,hypomyelinating,7,with or without oligodontia and/or hypogonadotropic hypogonadism,607694
POLR3B	Leukodystrophy,hypomyelinating,8,with or without oligodontia and/or hypogonadotropic hypogonadism,614381
KIAA0586	Joubert syndrome 23,616490 Short-rib thoracic dysplasia 14 with polydactyly,616546
PSAT1	Neu-Laxova syndrome 2,616038 ?Phosphoserine aminotransferase deficiency,610992
APOC2	Hyperlipoproteinemia,type Ib,207750
LEP	Obesity,morbid,due to leptin deficiency,614962
MAB21L1	Cerebellar,ocular,craniofacial,and genital syndrome
MAB21L2	Microphthalmia,syndromic 14,615877
RAB18	Warburg micro syndrome 3,614222
CCDC65	Ciliary dyskinesia,primary,27,615504
VPS45	Neutropenia,severe congenital,5,autosomal recessive,615285
NECTIN4	Ectodermal dysplasia-syndactyly syndrome 1,613573
NECTIN1	Cleft lip/palate-ectodermal dysplasia syndrome
DYNC2I2	Short-rib thoracic dysplasia 11 with or without polydactyly,615633
DYNC2I1	Short-rib thoracic dysplasia 8 with or without polydactyly,615503
ISCA2	Multiple mitochondrial dysfunctions syndrome 4,616370
C12orf57	Temtamy syndrome,218340
IL1RN	Interleukin 1 receptor antagonist deficiency,612852 {Gastric cancer risk after H. pylori infection},137215 {Microvascular complications of diabetes 4},612628
ISCA1	Multiple mitochondrial dysfunctions syndrome 5
PANK2	HARP syndrome,607236 Neurodegeneration with brain iron accumulation 1,234200
MTRR	Homocystinuria-megaloblastic anemia,cbl E type,236270 {Neural tube defects,folate-sensitive,susceptibility to},601634
TRAF3IP1	Senior-Loken syndrome 9,616629
PARS2	Epileptic encephalopathy,early infantile,75
CISD2	Wolfram syndrome 2,604928
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts,604004

GBA1	Gaucher disease,perinatal lethal,608013
AQP2	Diabetes insipidus,nephrogenic,125800
GBA2	Spastic paraplegia 46,autosomal recessive,614409
NFU1	Multiple mitochondrial dysfunctions syndrome 1,605711
NIPAL4	Ichthyosis,congenital,autosomal recessive 6,612281
PNP	Immunodeficiency due to purine nucleoside phosphorylase deficiency,613179
FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency,255100
DAG1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,9,616538 Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,9,613818
DZIP1L	Polycystic kidney disease 5
LONP1	CODAS syndrome,600373
SIL1	Marinesco-Sjogren syndrome,248800
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly,613091
PDHX	Lacticacidemia due to PDX1 deficiency,245349
COG8	Congenital disorder of glycosylation,type IIh,611182
CPT1A	CPT deficiency,hepatic,type IA,255120
COG7	Congenital disorder of glycosylation,type IIe,608779
COG6	Congenital disorder of glycosylation,type III,614576 Shaheen syndrome,615328
COG5	Congenital disorder of glycosylation,type IIi,613612
COG4	Congenital disorder of glycosylation,type IIj,613489
TANGO2	Metabolic encephalomyopathic crises,recurrent,with rhabdomyolysis,cardiac arrhythmias and neurodegeneration,616878
SLC35C1	Congenital disorder of glycosylation,type IIc,266265
AP1B1	Keratitis-ichthyosis-deafness syndrome, autosomal recessive
KRT8	Cirrhosis,cryptogenic,215600 {Cirrhosis,noncryptogenic,susceptibility to},215600
P3H2	Myopia,high,with cataract and vitreoretinal degeneration,614292
COG1	Congenital disorder of glycosylation,type IIg,611209
P3H1	Osteogenesis imperfecta,type VIII,610915
ESCO2	Roberts syndrome,268300 SC phocomelia syndrome,269000
KNL1	Microcephaly 4,primary,autosomal recessive
KRT5	Dowling-Degos disease 1,Epidermolysis bullosa simplexrecessive 1,Epidermolysis bullosa simplexDowling-Meara type,Epidermolysis bullosa simplex-MCR,Epidermolysis bullosa simplexKoebner type,Epidermolysis bullosa simplex-MP,Epidermolysis bullosa simplexWeber-Cockayne type
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis,201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase,613571
CYP2U1	Spastic paraplegia 56,autosomal recessive,615030

SIK3	?Spondyloepimetaphyseal dysplasia,Krakow type
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria),245400
DNA2	?Seckel syndrome 8
OBSL1	3-M syndrome 2,612921
CFTR	Cystic fibrosis,219700
SPTBN4	?Myopathycongenitalwith neuropathy and deafness
RTEL1	Dyskeratosis congenita,autosomal dominant 4,615190 Dyskeratosis congenita,autosomal recessive 5,615190 Pulmonary fibrosis and/or bone marrow failure,telomere-related,3,616373
SLC22A5	Carnitine deficiency,systemic primary,212140
SLC35D1	Schneckenbecken dysplasia,269250
LHB	Hypogonadotropic hypogonadism 23 with or without anosmia,228300
MTTP	Abetalipoproteinemia,200100
STXBP2	Hemophagocytic lymphohistiocytosis,familial,5,613101
TWIST2	Focal facial dermal dysplasia 3,Setleis type,227260
SNORD118	Leukoencephalopathybrain calcificationsand cysts
LIAS	Hyperglycinemia,lactic acidosis,and seizures,614462
CD79B	Agammaglobulinemia 6,612692
TRPM1	Night blindness,congenital stationary (complete),1C,autosomal recessive,613216
CD79A	Agammaglobulinemia 3,613501
ZNHIT3	PEHO syndrome
ATP6V0A2	Cutis laxa,autosomal recessive,type IIA,219200 Wrinkly skin syndrome,278250
ATP6V0A4	Renal tubular acidosis,distal,autosomal recessive,602722
TRPM6	Hypomagnesemia 1,intestinal,602014
SPTBN2	Spinocerebellar ataxia,autosomal recessive 14
PYCR1	Cutis laxa,autosomal recessive,type IIB,612940 Cutis laxa,autosomal recessive,type IIIB,614438
PYCR2	Leukodystrophy,hypomyelinating,10,616420
TYR	Albinism,oculocutaneous,type IA,203100
TECPR2	Spastic paraplegia 49,autosomal recessive,615031
CD320	Methylmalonic aciduria due to transcobalamin receptor defect,613646
COL18A1	Knobloch syndrome,type 1,267750
ROBO3	Gaze palsy,horizontal,with progressive scoliosis,607313
GFM1	Combined oxidative phosphorylation deficiency 1,609060
TENM3	Microphthalmia,isolated,with coloboma 9,615145
C2CD3	Orofaciodigital syndrome XIV
SLC44A1	Neurodegeneration, childhood-onset, with ataxia, tremor, optic atrophy, and cognitive decline
GFM2	Combined oxidative phosphorylation deficiency 39
GBE1	Glycogen storage disease IV,232500 Polyglucosan body disease,adult form,263570
HBB	Sickle cell anemia,603903,Thalassemia-beta,dominant inclusion-body,603902,Thalassemias,beta-,613985

MEOX1	Klippel-Feil syndrome 2,214300
ROBO1	-
SLC22A12	Hypouricemia,renal,220150
C1QBP	Combined oxidative phosphorylation deficiency 33,617713
CHMP1A	Pontocerebellar hypoplasia,type 8,614961
PRX	Charcot-Marie-Tooth disease,type 4F,614895 Dejerine-Sottas disease,145900
SH3PXD2B	Frank-ter Haar syndrome,249420
FEZF1	Hypogonadotropic hypogonadism 22,with or without anosmia,616030
EDN1	Auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798 {High density lipoprotein cholesterol level QTL 7}
EDN3	Waardenburg syndrome,type 4B,613265
GP1BB	Bernard-Soulier syndrome,type B,231200 Giant platelet disorder,isolated,231200
STRADA	Polyhydramnios,megalencephaly,and symptomatic epilepsy,611087
TOP3A	?Progressive external ophthalmoplegia with mitochondrial DNA deletions,autosomal recessive 5 ,Microcephaly,growth restriction,and increased sister chromatid exchange 2
GP1BA	Bernard-Soulier syndrome,type A1 (recessive),231200 Bernard-Soulier syndrome,type A2 (dominant),153670 von Willebrand disease,platelet-type,177820 {Nonarteritic anterior ischemic optic neuropathy,susceptibility to},258660
TYK2	Immunodeficiency 35,611521
CYP7B1	Bile acid synthesis defect,congenital,3,613812 Spastic paraplegia 5A,autosomal recessive,270800
CLDN10	HELIX syndrome
UGDH	Developmental and epileptic encephalopathy 84
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency,271980
CLDN14	Deafness,autosomal recessive 29,614035
CLDN19	Hypomagnesemia 5,renal,with ocular involvement,248190
VWA1	Neuropathy, hereditary motor, with myopathic features
CLDN16	Hypomagnesemia 3,renal,248250
MATN3	Spondyloepimetaphyseal dysplasia,608728 {Osteoarthritis susceptibility 2},140600
HMX1	Oculoauricular syndrome 612109
SDCCAG8	Bardet-Biedl syndrome 16,615993 Senior-Loken syndrome 7,613615
C1QB	C1q deficiency,613652
GUCY2C	Meconium ileus,614665
C1QA	C1q deficiency,613652
CSTB	Epilepsy,progressive myoclonic 1A (Unverricht and Lundborg),254800
CSTA	Peeling skin syndrome 4,607936
DTYMK	Neurodegeneration, childhood-onset, with progressive microcephaly
GUCY2D	Cone-rod dystrophy 6,215500
HAAO	Vertebralcardiacrenaland limb defects syndrome 1

PTS	Hyperphenylalaninemia,BH4-deficient,A,261640
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type),603041
PSMC3IP	Ovarian dysgenesis 3,614324
CFAP298	Ciliary dyskinesia,primary,26,615500
NT5E	Calcification of joints and arteries,211800
EMC1	Cerebellar atrophy,visual impairment,and psychomotor retardation,616875
LMNA	Charcot-Marie-Tooth disease,type 2B1,605588
KISS1R	Hypogonadotropic hypogonadism 8 with or without anosmia,614837 ?Precocious puberty,central,1,176400
DUOX2	Thryoid dyshormonogenesis 6,607200
GINS1	Immunodeficiency 55
LHFPL5	Deafness,autosomal recessive 67,610265
MLH1	Mismatch repair cancer syndrome,276300
VAX1	?Microphthalmiasyndromic 11
COLQ	Myasthenic syndrome,congenital,5,603034
SLURP1	Meleda disease,248300
PDP1	Pyruvate dehydrogenase phosphatase deficiency,608782
PRKRA	Dystonia 16,612067
ATP5PO	-
MEGF8	Carpenter syndrome 2,614976
C1QC	C1q deficiency,613652
RIPOR2	?Deafnessautosomal recessive 104
FCN3	Immunodeficiency due to ficolin 3 deficiency,613860
NDUFA13	?Mitochondrial complex I deficiency,nuclear type 28
NDUFA11	Mitochondrial complex I deficiency,252010
NDUFA12	Leigh syndrome due to mitochondrial complex 1 deficiency,256000
NDUFA10	Leigh syndrome
ROGDI	Kohlschutter-Tonz syndrome,226750
CWC27	Retinitis pigmentosa with or without skeletal anomalies
BMPER	Diaphanospondylodysostosis,608022
NARS1	-
ARHGDIA	Nephrotic syndrome,type 8,615244
NARS2	Combined oxidative phosphorylation deficiency 24,616239
SACS	Spastic ataxia,Charlevoix-Saguenay type,270550
TRAPPC12	Encephalopathyprogressiveearly-onsetwith brain atrophy and spasticity
TRAPPC11	Muscular dystrophy,limb-girdle,type 2S,615356
NRCAM	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities
TRAPPC14	?Microcephaly 25,primary,autosomal recessive
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency,231530 Hyperinsulinemic hypoglycemia,familial,4,609975
TMEM38B	Osteogenesis imperfecta,type XIV,615066
PUS3	Mental retardation,autosomal recessive 55
CHRNB1	?Myasthenic syndrome,congenital,2C,associated with acetylcholine receptor deficiency,616314
GUCY1A1	Moyamoya 6 with achalasia,615750

MLIP	-
PHYH	Refsum disease,266500
SZT2	Epileptic encephalopathy,early infantile,18,615476
TRAPPC2L	Encephalopathy,progressive,early-onset,with episodic rhabdomyolysis
HGD	Alkaptonuria,203500
LIG1	-
IL11RA	Craniosynostosis and dental anomalies,614188
HGF	Deafness,autosomal recessive 39,608265
PUS1	Myopathy,lactic acidosis,and sideroblastic anemia 1,600462
ALG14	?Myasthenic syndromecongenital15without tubular aggregates
LIG4	LIG4 syndrome,606593
ALG12	Congenital disorder of glycosylation,type Ig,607143
LIFR	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome,601559
LIG3	-
ALG11	Congenital disorder of glycosylation,type Ip,613661
PUS7	Intellectual developmental disorder with abnormal behavior,microcephaly,and short stature
LZTFL1	Bardet-Biedl syndrome 17,615994
TUFM	Combined oxidative phosphorylation deficiency 4,610678
FAM161A	Retinitis pigmentosa 28,606068
FANCD2	Fanconi anemia,complementation group D2,227646
PNLIP	Pancreatic lipase deficiency 614338
CHRNA1	Multiple pterygium syndrome,lethal type,253290
COPB2	?Microcephaly 19primaryautosomal recessive
CABP4	Cone-rod synaptic disorder,congenital nonprogressive,610427
CABP2	Deafness,autosomal recessive 93,614899
LPL	Lipoprotein lipase deficiency,238600
CAMSAP1	-
FBLN5	Cutis laxa,autosomal recessive,type IA,219100
SCUBE3	-
EMG1	Bowen-Conradi syndrome,211180
THPO	-
NLRP7	Hydatidiform mole,recurrent,1,231090
CDH23	Deafness,autosomal recessive 12,601386 Usher syndrome,type 1D,601067 Usher syndrome,type 1D/F digenic,601067
NLRP1	Autoinflammation with arthritis and dyskeratosis
PNPT1	Combined oxidative phosphorylation deficiency 13,614932 Deafness,autosomal recessive 70,614934
UQCC3	?Mitochondrial complex III deficiencynuclear type 9
UQCC2	?Mitochondrial complex III deficiencynuclear type 7
COQ9	Coenzyme Q10 deficiency,primary,5,614654
COQ6	Coenzyme Q10 deficiency,primary,6,614650
COQ4	Coenzyme Q10 deficiency,primary,7,616276
DIAPH1	Seizures,cortical blindness,microcephaly syndrome,616632
GRHPR	Hyperoxaluria,primary,type II,260000

SEPSECS	Pontocerebellar hypoplasia type 2D,613811
COQ2	Coenzyme Q10 deficiency,primary,1,607426{Multiple system atrophy,susceptibility to},146500
AMACR	Alpha-methylacyl-CoA racemase deficiency,614307 Bile acid synthesis defect,congenital,4,214950
PI4KA	Polymicrogyria,perisylvian,with cerebellar hypoplasia and arthrogryposis,616531
BMPR1B	Acromesomelic dysplasia,Demirhan type,609441
MYF5	Ophthalmoplegia,external,with rib and vertebral anomalies
GPSM2	Chudley-McCullough syndrome,604213
KDM5B	Mental retardation,autosomal recessive 65
THRB	Thyroid hormone resistance,autosomal recessive,274300
WNT2B	-
MEGF10	Myopathy,areflexia,respiratory distress,and dysphagia,early-onset,614399 Myopathy,areflexia,respiratory distress,and dysphagia,early-onset,mild variant,614399
DBH	Dopamine beta-hydroxylase deficiency,223360 [Dopamine-beta-hydroxylase activity levels,plasma]
OPLAH	5-oxoprolinase deficiency,260005
NR2E3	Enhanced S-cone syndrome,268100 Retinitis pigmentosa 37,611131
HK1	Hemolytic anemia due to hexokinase deficiency,235700 Neuropathy,hereditary motor and sensory,Russe type,605285
MFSD8	Ceroid lipofuscinosis,neuronal,7,610951 Macular dystrophy with central cone involvement,616170
SPRED2	Noonan syndrome 14
SIX6	Optic disc anomalies with retinal and/or macular dystrophy 212550
CASP8	Autoimmune lymphoproliferative syndrome,type IIB,607271
HJV	Hemochromatosis type 2A,602390
CYP2R1	Rickets due to defect in vitamin D 25-hydroxylation,600081
DBT	Maple syrup urine disease,type II,248600
COLGALT1	Brain small vessel disease 3
RNU4ATAC	-
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C),252930 Retinitis pigmentosa 73,616544
DCC	Gaze palsy,familial horizontal,with progressive scoliosis,2 ,617542
POMGNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,8,614830
LSS	Cataract 44,616509
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,3,253280
DNM1	-
DNM2	Lethal congenital contracture syndrome 5
ALDH1A3	Microphthalmia,isolated 8,615113
ZAP70	Autoimmune disease,multisystem,infantile-onset,2,617006 Immunodeficiency 48,269840
MYH2	Proximal myopathy and ophthalmoplegia,605637

MYH3	Contractures,pterygia,and spondylocarpotarsal fusion syndrome 1B
TRAPPC6B	Neurodevelopmental disorder with microcephalyepilepsyand brain atrophy
ALDH1A2	-
COL6A2	Ullrich congenital muscular dystrophy 1,254090 ?Myosclerosis,congenital,255600
CDH11	Elsahy-Waters syndrome
COL6A1	Ullrich congenital muscular dystrophy 1,254090
RFT1	Congenital disorder of glycosylation,type In,612015
CASQ2	Ventricular tachycardia,catecholaminergic polymorphic,2,611938
COL6A3	Dystonia 27,616411 Ullrich congenital muscular dystrophy 1,254090
COLEC11	3MC syndrome 2,265050
VAC14	Striatonigral degeneration,childhood-onset,617054
COLEC10	3MC syndrome 3
DDC	Aromatic L-amino acid decarboxylase deficiency,608643
PDXK	Neuropathy,hereditary motor and sensory,type VIC,with optic atrophy
BRAT1	Rigidity and multifocal seizure syndrome,lethal neonatal,614498
NRXN1	Pitt-Hopkins-like syndrome 2,614325 {Schizophrenia,susceptibility to,17},614332
ETFDH	Glutaric acidemia IIC,231680
CHSY1	Temtamy preaxial brachydactyly syndrome,605282
PPP2R3C	Gonadal dysgenesis,dysmorphic facies,retinal dystrophy,and myopathy
POC1A	Short stature,onychodysplasia,facial dysmorphism,and hypotrichosis,614813
PDSS2	Coenzyme Q10 deficiency,primary,3,614652
POC1B	Cone-rod dystrophy 20,615973
PDSS1	Coenzyme Q10 deficiency,primary,2,614651
LIM2	Cataract 19,multiple types,615277
PDX1	Pancreatic agenesis 1,260370
DES	Myopathy,myofibrillar,1
MLPH	Griscelli syndrometype 3
PDE10A	Dyskinesia,limb and orofacial,infantile-onset,616921
WEE2	Oocyte maturation defect 5,617996
CAPN10	intellectual disability
FCSK	Congenital disorder of glycosylation with defective fucosylation 2
ATP13A2	Kufor-Rakeb syndrome,606693 ?Ceroid lipofuscinosis,neuronal,12,606693
LPIN1	Myoglobinuria,acute recurrent,autosomal recessive,268200
LPIN2	Majeed syndrome,609628
MAPKBP1	Nephronophthisis 20
PGAP1	Mental retardation,autosomal recessive 42,615802
PGAP2	Hyperphosphatasia with mental retardation syndrome 3,614207

PGAP3	Hyperphosphatasia with mental retardation syndrome 4,615716
AVIL	-
TMEM94	Intellectual developmental disorder with cardiac defects and dysmorphic facies
CLDN1	Ichthyosis,leukocyte vacuoles,alopecia,and sclerosing cholangitis,607626
LIPA	Cholesteryl ester storage disease,278000 Wolman disease,278000
TSEN54	Pontocerebellar hypoplasia type 2A,277470 Pontocerebellar hypoplasia type 4,225753 ?Pontocerebellar hypoplasia type 5,610204
MYLK	Megacystis-microcolon-intestinal hypoperistalsis syndrome
LIPE	Lipodystrophy,familial partial,type 6,615980
ZFYVE26	Spastic paraplegia 15,autosomal recessive,270700
LIPH	Hypotrichosis 7,604379 Woolly hair,autosomal recessive 2 with or without hypotrichosis,604379
LIPN	Ichthyosis,congenital,autosomal recessive 8,613943
LRAT	Leber congenital amaurosis 14,613341 Retinal dystrophy,early-onset severe,613341 Retinitis pigmentosa,juvenile,613341
HES7	Spondylocostal dysostosis 4,autosomal recessive,613686
CCBE1	Hennekam lymphangiectasia-lymphedema syndrome 1,235510
EML1	Band heterotopia
HSPG2	Dyssegmental dysplasia,Silverman-Handmaker type,224410 Schwartz-Jampel syndrome,type 1,255800
UPB1	Beta-ureidopropionase deficiency,613161
SLC5A7	Myasthenic syndromecongenital20presynaptic,Neuronopathydistal hereditary motortype VIIA
RBP4	Retinal dystrophy,iris coloboma,and comedogenic acne syndrome,615147
RFX6	Mitchell-Riley syndrome,615710
MYL1	Myopathy,congenital,with fast-twitch (type II) fiber atrophy
RBP3	?Retinitis pigmentosa 66
ETHE1	Ethylmalonic encephalopathy,602473
MYL2	Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy
MYL3	Cardiomyopathy,hypertrophic,8,608751
RFX5	Bare lymphocyte syndrome,type II,complementation group C,209920 Bare lymphocyte syndrome,type II,complementation group E,209920
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency,615895
UQCRC2	Mitochondrial complex III deficiency,nuclear type 5,615160
HPD	Tyrosinemia,type III,276710
HACD1	-
MTMR2	Charcot-Marie-Tooth disease,type 4B1,601382

AIRE	Autoimmune polyendocrinopathy syndrome ,type I,with or without reversible metaphyseal dysplasia,240300
DHH	46XY sex reversal 7,233420
ABCB4	Cholestasis,intrahepatic,of pregnancy,3,614972 Cholestasis,progressive familial intrahepatic 3,602347 Gallbladder disease 1,600803
CLCNKB	Bartter syndrome,type 3,607364
SLC5A1	Glucose/galactose malabsorption,606824
AAAS	Achalasia-addisonianism-alacrimia syndrome,231550
SLC30A10	Hypermanganesemia with dystonia 1,613280
SLC5A2	Renal glucosuria,233100
NCAPH	?Microcephaly 23primaryautosomal recessive
SLC5A5	Thyroid dyshormonogenesis 1,274400
C3	C3 deficiency,613779
DYNC2LI1	Short-rib thoracic dysplasia 15 with polydactyly,617088
GHRHR	Growth hormone deficiency,isolated,type IB,612781
HSD11B2	Apparent mineralocorticoid excess,218030
C5	C5 deficiency,609536
CYB5R3	Methemoglobinemia,type I,250800 Methemoglobinemia,type II,250800
C9	C9 deficiency,613825
MYO6	Deafness,autosomal recessive 37,607821
HESX1	Growth hormone deficiency with pituitary anomalies,182230 Pituitary hormone deficiency,combined,5,182230 Septooptic dysplasia,182230
EXPH5	Epidermolysis bullosa,nonspecific,autosomal recessive,615028
GPIHBP1	Hyperlipoproteinemia,type 1D,615947
ABCA1	HDL deficiency,type 2,604091 Tangier disease,205400 {Coronary artery disease in familial hypercholesterolemia,protection against},143890
B3GALNT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies,type A,11,615181
CTC1	Cerebroretinal microangiopathy with calcifications and cysts,612199
EDEM3	Congenital disorder of glycosylation, type 2V
HACE1	Spastic paraplegia and psychomotor retardation with or without seizures,616756
ABCA3	Surfactant metabolism dysfunction,pulmonary,3,610921
LRBA	Immunodeficiency,common variable,8,with autoimmunity,614700
ABCA4	Cone-rod dystrophy 3,604116 Fundus flavimaculatus,248200 Retinal dystrophy,early-onset severe,248200 Retinitis pigmentosa 19,601718 Stargardt disease 1,248200 {Macular degeneration,age-related,2},153800
PMM2	Congenital disorder of glycosylation,type Ia,212065
ERLIN1	Spastic Paraplegia 62,615681
EXOC6B	Spondyloepimetaphyseal dysplasia with joint laxity,type 3
ERLIN2	Spastic paraplegia 18,autosomal recessive,611225

CP	Cerebellar ataxia,604290 Hemosiderosis,systemic,due to aceruloplasminemia,604290 [Hypoceruloplasminemia,hereditary],604290
NR0B2	Obesity,mild,early-onset,601665
MYMK	Carey-Fineman-Ziter syndrome ,254940
SETX	Spinocerebellar ataxia,autosomal recessive 1,606002
FERMT1	Kindler syndrome,173650
CYP2C8	Rhabdomyolysis,cerivastatin-induced,601129
COL1A2	Ehlers-Danlos syndrome,cardiac valvular type,225320
TDP2	Spinocerebellar ataxia,autosomal recessive,616949
TDP1	Spinocerebellar ataxia,autosomal recessive with axonal neuropathy,607250
SASS6	?Microcephaly 14primaryautosomal recessive
DCHS1	Van Maldergem syndrome 1,601390
CDIN1	Dyserythropoietic anemia,congenital,type Ib,615631
HAMP	Hemochromatosis,type 2B,613313
FERMT3	Leukocyte adhesion deficiency,type III,612840
PIGS	Glycosylphosphatidylinositol biosynthesis defect 18
LRSAM1	Charcot-Marie-Toothe disease,axonal,type 2P,614436
PIGU	Glycosylphosphatidylinositol biosynthesis defect 21
PIGT	Multiple congenital anomalies-hypotonia-seizures syndrome 3,615398
PIGO	Hyperphosphatasia with mental retardation syndrome 2,614749
PIGN	Multiple congenital anomalies-hypotonia-seizures syndrome 1,614080
HEXB	Sandhoff disease,infantile,juvenile,and adult forms,268800
PIGQ	Epileptic encephalopathy,early infantile,77
HEXA	GM2-gangliosidosis,several forms,272800 Tay-Sachs disease,272800 [Hex A pseudodeficiency],272800
PIGP	?Epileptic encephalopathyearly infantile55
PTH1R	Chondrodysplasia,Blomstrand type,215045Eiken syndrome,600002Failure of tooth eruption,primary,125350 Metaphyseal chondrodysplasia,Murk Jansen type,156400
TMEM53	-
PIGW	Glycosylphosphatidylinositol biosynthesis defect 11
PIGV	Hyperphosphatasia with mental retardation syndrome 1,239300
MYPN	Nemaline myopathy 11,autosomal recessive,617336
PIGY	Hyperphosphatasia with mental retardation syndrome 6,616809
TRIOBP	Deafness,autosomal recessive 28,609823
IKBKB	Immunodeficiency 15,615592
LMBR1	Acheiropody,200500
FLVCR1	Ataxia,posterior column,with retinitis pigmentosa,609033
FLVCR2	Proliferative vasculopathy and hydraencephaly-hydrocephaly syndrome,225790
TMC1	Deafness,autosomal recessive 7,600974
ACTL6B	Epileptic encephalopathy,early infantile,76

ELOVL4	Ichthyosis,spastic quadriplegia,and mental retardation,614457
ARG1	Argininemia,207800
NSUN2	Mental retardation,autosomal recessive 5,611091
PGAM2	Glycogen storage disease X,261670
RIPK4	Popliteal pterygium syndrome,Bartsocas-Papas type,263650
TMC8	Epidermodysplasia verruciformis,226400
EMP2	Nephrotic syndrome,type 10,615861
TMC6	Epidermodysplasia verruciformis,226400
F2	Dysprothrombinemia,613679
F5	Factor V deficiency,227400
MRPL44	?Combined oxidative phosphorylation deficiency 16
SUMF1	Multiple sulfatase deficiency,272200
GON7	-
PIGC	Glycosylphosphatidylinositol biosynthesis defect 16
F7	Factor VII deficiency,227500 {Myocardial infarction,decreased susceptibility to},608446
PIGB	Epileptic encephalopathy,early infantile,80
KCNMA1	Cerebellar atrophy,developmental delay,and seizures
PIGK	Neurodevelopmental disorder with hypotonia and cerebellar atrophy, with or without seizures,PMID: 32220290
VAMP1	Myasthenic syndrome,congenital,25
PIGM	Glycosylphosphatidylinositol deficiency,610293
PIGL	CHIME syndrome,280000
PIGG	Mental retardation,autosomal recessive 53,616917
DLD	Dihydrolipoamide dehydrogenase deficiency,246900
SLC29A3	Histiocytosis-lymphadenopathy plus syndrome,602782
PIGH	Glycosylphosphatidylinositol biosynthesis defect 17
KANK2	Palmoplantar keratoderma and woolly hair,616099
KATNIP	Joubert syndrome 26
FH	Fumarase deficiency,606812
IDUA	Mucopolysaccharidosis Ih,607014 Mucopolysaccharidosis Ih/s,607015 Mucopolysaccharidosis Is,607016
KARS1	Deafness,autosomal recessive 89,613916 ?Charcot-Marie-Tooth disease,recessive intermediate,B,613641
COL11A1	Fibrochondrogenesis 1,228520
COL11A2	Deafness,autosomal recessive 53,609706 Fibrochondrogenesis 2,614524 Otospondylomegaepiphyseal dysplasia,215150 Stickler syndrome,type III,184840 Weissenbacher-Zweymuller syndrome,277610
TMEM70	Mitochondrial complex V (ATP synthase) deficiency,nuclear type 2,614052
UNG	Immunodeficiency with hyper IgM,type 5 608106
RECQL4	Baller-Gerold syndrome,218600 RAPADILINO syndrome,266280 Rothmund-Thomson syndrome,268400
GLRA1	Hyperekplexia,hereditary 1,autosomal dominant or recessive,149400
PSMB4	?Proteasome-associated autoinflammatory syndrome 3 and digenic forms

TMEM67	COACH syndrome,216360 Joubert syndrome 6,610688 Meckel syndrome 3,607361 Nephronophthisis 11,613550 {Bardet-Biedl syndrome 14,modifier of},615991
ANO10	Spinocerebellar ataxia,autosomal recessive 10,613728
PMS2	Mismatch repair cancer syndrome,276300
FLNB	Spondylocarpotarsal synostosis syndrome,272460
EXTL3	Immunoskeletal dysplasia with neurodevelopmental abnormalities
EVC2	Ellis-van Creveld syndrome,225500
P2RY12	Bleeding disorder,platelet-type,8,609821
TRMT10A	Microcephaly,short stature,and impaired glucose metabolism 1,616033
SLC16A1	Monocarboxylate transporter 1 deficiency
TRMT10C	Combined oxidative phosphorylation deficiency 30,616974
PET100	Mitochondrial complex IV deficiency,220110
MDH2	Epileptic encephalopathy,early infantile,51
STAT1	Immunodeficiency 31A,mycobacteriosis,autosomal dominant,614892 ncy 31B,mycobacterial and viral infections,autosomal recessive,613796 Immunodeficiency 31C,autosomal dominant,614162
STAT2	Immunodeficiency 44,616636
GSX2	Diencephalic-mesencephalic junction dysplasia syndrome 2
VWA3B	Spinocerebellar ataxia,autosomal recessive 22
PSMB8	Autoinflammation,lipodystrophy,and dermatosis syndrome,256040
PSMB9	?Proteasome-associated autoinflammatory syndrome 3digenic
TMCO1	Craniofacial dysmorphism,skeletal anomalies,and mental retardation syndrome,213980
TTLL5	Cone-rod dystrophy 19,615860
LYRM7	Mitochondrial complex III deficiency,nuclear type 8,615838
MPLKIP	Trichothiodystrophy 4,nonphotosensitive,234050
NRROS	Seizures, early-onset, with neurodegeneration and brain calcification,PMID: 32197075
NCF1	Chronic granulomatous disease due to deficiency of NCF-1,233700
NCF2	Chronic granulomatous disease due to deficiency of NCF-2,233710
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome-2,614069
NCF4	Granulomatous disease,chronic,autosomal recessive,cytochrome b-positive,type III,613960
RORC	Immunodeficiency 42,616622
F13A1	Factor XIIIA deficiency,613225
HR	Alopecia universalis,203655 Atrichia with papular lesions,209500 Hypotrichosis 4,146550
HNMT	Mental retardation,autosomal recessive 51,616739 {Asthma,susceptibility to},600807
TSEN2	Pontocerebellar hypoplasia type 2B,612389
ATCAY	Ataxia,cerebellar,Cayman type,601238

LEPR	Obesity,morbid,due to leptin receptor deficiency,614963
PMPCB	Multiple mitochondrial dysfunctions syndrome 6 ,617954
PMPCA	Spinocerebellar ataxia,autosomal recessive 2,213200
SLC25A42	Metabolic crises,recurrent,with variable encephalomyopathic features and neurologic regression
NEPRO	Anauxetic dysplasia 3
SLC25A46	Neuropathy,hereditary motor and sensory,type VIB,616505
EPM2A	Epilepsy,progressive myoclonic 2A (Lafora),254780
AGTPBP1	Neurodegeneration,childhood-onset,with cerebellar atrophy
ANAPC7	-
AFG2A	Epilepsy,hearing loss,and mental retardation syndrome,616577
AFG2B	Deafness, autosomal recessive 119,Neurodevelopmental disorder with hearing loss and spasticity
VPS33A	Mucopolysaccharidosis-plus syndrome
VPS33B	Arthrogryposis,renal dysfunction,and cholestasis 1,208085
SLC9A3	Diarrhea 8,secretory sodium,congenital,616868
MAG	Spastic paraplegia 75,autosomal recessive,616680
MAK	Retinitis pigmentosa 62,614181
IGFALS	Acid-labile subunit,deficiency of,615961
ROR2	Robinow syndrome,autosomal recessive,268310
SQSTM1	Neurodegeneration with ataxia,dystonia,and gaze palsy,childhood-onset
ANAPC1	Rothmund-Thomson syndrome,type 1
DSC2	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair,610476 Arrhythmogenic right ventricular dysplasia 11,610476
DSC3	Hypotrichosis and recurrent skin vesicles 613102
RNASEH2C	Aicardi-Goutieres syndrome 3,610329
LRRC56	Ciliary dyskinesia,primary,39
RNASEH2B	Aicardi-Goutieres syndrome 2,610181
RNASEH2A	Aicardi-Goutieres syndrome 4,610333
ARL6	Bardet-Biedl syndrome 3
TUBGCP2	Pachygyria, microcephaly, developmental delay, and dysmorphic facies, with or without seizures
ARL3	Joubert syndrome 35
GLIS3	Diabetes mellitus,neonatal,with congenital hypothyroidism,610199
TACSTD2	Corneal dystrophy,gelatinous drop-like,204870
TNFRSF11B	Paget disease of bone 5,juvenile-onset,239000
TNFRSF11A	Osteopetrosis,autosomal recessive 7
CYP19A1	Aromatase deficiency,613546
ACAT1	Alpha-methylacetoacetic aciduria,203750
GLIS2	Nephronophthisis 7,611498
SLC9A1	?Lichtenstein-Knorr syndrome
CPAMD8	Anterior segment dysgenesis 8
SLC38A8	Foveal hypoplasia 2,with or without optic nerve misrouting and/or anterior segment dysgenesis,609218
CPA6	Febrile seizures,familial,11,614418

KL	Tumoral calcinosis,hyperphosphatemic,617994
SRD5A2	Pseudovaginal perineoscrotal hypospadias,264600
ZBTB16	Skeletal defects,genital hypoplasia,and mental retardation,612447
GFPT1	Myasthenia,congenital,12,with tubular aggregates,610542
SRD5A3	Congenital disorder of glycosylation,type Iq,612379 Kahrizi syndrome,612713
DCDC2	Nephronophthisis 19,616217 ?Deafness,autosomal recessive 66,610212
B3GALT6	Ehlers-Danlos syndrome,progeroid type,2,615349 Spondyloepimetaphyseal dysplasia with joint laxity,type 1,with or without fractures,271640
ZBTB11	Intellectual developmental disorder,autosomal recessive 69
FOXN1	T-cell immunodeficiency,congenital alopecia,and nail dystrophy,601705
AICDA	Immunodeficiency with hyper-IgM,type 2,605258
KY	Myopathymyofibrillar7
TMIE	Deafness,autosomal recessive 6,600971
PTPRC	Severe combined immunodeficiency,T cell-negative,B-cell/natural killer-cell positive,608971 {Hepatitic C virus,susceptibility to},609532
GGCX	Vitamin K-dependent clotting factors,combined deficiency of,1,277450
LRIT3	Night blindness,congenital stationary (complete),1F,autosomal recessive,615058
TUBGCP6	Microcephaly and chorioretinopathy,autosomal recessive,1,251270
TUBGCP4	Microcephaly and chorioretinopathy,autosomal recessive,3,616335
MYD88	Pyogenic bacterial infections,recurrent,due to MYD88 deficiency,612260
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,2,613150
SLC25A1	Combined D-2- and L-2-hydroxyglutaric aciduria,615182
CNTNAP2	Cortical dysplasia-focal epilepsy syndrome,610042 Pitt-Hopkins like syndrome 1,610042 {Autism susceptibility 15},612100
CNTNAP1	Lethal congenital contracture syndrome 7,616286
SLC25A3	Mitochondrial phosphate carrier deficiency,610773
COX4I2	Exocrine pancreatic insufficiency,dyserythropoietic anemia,and calvarial hyperostosis,612714
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,1,236670
PIK3CD	-
MRPL3	Combined oxidative phosphorylation deficiency 9,614582
NADK2	?2,4-dienoyl-CoA reductase deficiency
FCGR3A	Immunodeficiency 20,615707
ATP7B	Wilson disease,277900
DSE	Ehlers-Danlos syndromemusculocontractural type 2

DNM1L	Encephalopahty,lethal,due to defective mitochondrial peroxisomal fission,614388
SPG11	Amyotrophic lateral sclerosis 5,juvenile,602099 Charcot-Marie-Tooth disease,axonal,type 2X,616668 Spastic paraplegia 11,autosomal recessive,604360
DIS3L2	Perlman syndrome,267000
DSP	Epidermolysis bullosa,lethal acantholytic,609638
DST	Epidermolysis bullosa simplex,autosomal recessive 2,615425 ?Neuropathy,hereditary sensory and autonomic,type VI,614653
SIGMAR1	?Spinal muscular atrophydistalautosomal recessive2,?Amyotrophic lateral sclerosis 16juvenile
VPS37A	Spastic paraplegia 53,autosomal recessive,614898
DDOST	?Congenital disorder of glycosylationtype Ir
NT5C3A	Anemia,hemolytic,due to UMPH1 deficiency,266120
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome,238970
HAX1	Neutropenia,severe congenital 3,autosomal recessive,610738
NPC1	Niemann-Pick disease,type C1,257220 Niemann-Pick disease,type D,257220 {Nasopharyngeal carcinoma 1}
NPC2	Niemann-pick disease,type C2,607625
SLC25A19	Microcephaly,Amish type,607196 Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type),613710
DSG4	Hypotrichosis 6,607903
SLC25A12	Epileptic encephalopathy,early infantile,39,612949
MET	?Deafnessautosomal recessive 97
SLC25A4	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR
SLC25A13	Citrullinemia,adult-onset type II,603471 Citrullinemia,type II,neonatal-onset,605814
NDUFB9	?Mitochondrial complex I deficiency,nuclear type 24
PPP1R15B	Microcephaly,short stature,and impaired glucose metabolism 2,616817
NDUFB8	Mitochondrial complex I deficiency,nuclear type 32
SEC23A	Craniolenticulosutural dysplasia,607812
LRP1	?Keratosis pilaris atrophicans
PRCD	Retinitis pigmentosa 36,610599
NDUFB3	Mitochondrial complex I deficiency,252010
LRP5	Osteoporosis-pseudoglioma syndrome,259770
USH1C	Deafness,autosomal recessive 18A,602092 Usher syndrome,type 1C,276904
KATNB1	Lissencephaly 6,with microcephaly,616212
LRP4	Cenani-Lenz syndactyly syndrome,212780 Sclerosteosis 2,614305 ?Myasthenic syndrome,congenital,17,616304
ZBTB42	?Lethal congenital contracture syndrome 6
LRP2	Donnai-Barrow syndrome,222448
PPM1K	?Maple syrup urine disease,mild variant

MFF	Encephalopathy due to defective mitochondrial and peroxisomal fission 2
SLC25A26	Combined oxidative phosphorylation deficiency 28,616794
SLITRK6	Deafness and myopia,221200
D2HGDH	D-2-hydroxyglutaric aciduria,600721
IARS2	Cataracts,growth hormone deficiency,sensory neuropathy,sensorineural hearing loss,and skeletal dysplasia
PCBD1	Hyperphenylalaninemia,BH4-deficient,D,264070
IARS1	Growth retardation,intellectual developmental disorder,hypotonia and hepatopathy
SLC25A20	Carnitine-acylcarnitine translocase deficiency,212138
CNNM2	Hypomagnesemia,seizures,and mental retardation,616418
SEC23B	Cowden syndrome 7,616858 Dyserythropoietic anemia,congenital,type II,224100
SLC25A22	Epileptic encephalopathy,early infantile,3,609304
ATP9A	-
CNNM4	Jalili syndrome,217080
NDUFA9	Leigh syndrome due to mitochondrial complex I deficiency,256000
SLC12A2	Kilquist syndrome
CYB5A	Methemoglobinemia,type IV,250790
LINGO1	Mental retardation,autosomal recessive 64
NDUFA8	-
SLC12A3	Gitelman syndrome,263800
NDUFA6	Mitochondrial complex I deficiency,nuclear type 33
SLC12A5	Epileptic encephalopathy,early infantile,34,616645 {Epilepsy,idiopathic generalized,susceptibility to,14},616685
MMAA	Methylmalonic aciduria,vitamin B12-responsive,251100
MMAB	Methylmalonic aciduria,vitamin B12-responsive,due to defect in synthesis of adenosylcobalamin,cblB complementation type,251110
MBOAT7	Mental retardation,autosomal recessive 57
NDUFA2	Leigh syndrome due to mitochondrial complex I deficiency,256000
SLC12A1	Bartter syndrome,type 1,601678
DONSON	Microcephaly,short stature,and limb abnormalities
CTNS	Cystinosis,atypical nephropathic,219800 Cystinosis,late-onset juvenile or adolescent nephropathic,219900 Cystinosis,nephropathic,219800 Cystinosis,ocular nonnephropathic,219750
MCOLN1	Mucolipidosis IV,252650
MERTK	Retinitis pigmentosa 38,613862
USH2A	Retinitis pigmentosa 39,613809 Usher syndrome,type 2A,276901
DHODH	Miller syndrome,263750
SLC25A38	Anemia,sideroblastic,2,pyridoxine-refractory,205950
MTRFR	Combined oxidative phosphorylation deficiency 7,613559 Spastic paraplegia 55,autosomal recessive,615035
PC	Pyruvate carboxylase deficiency,266150

USH1G	Usher syndrome,type 1G,606943
MGP	Keutel syndrome,245150
VIPAS39	Arthrogryposis,renal dysfunction,and cholestasis 2,613404
SEC24D	Cole-Carpenter syndrome 2,616294
SLC25A36	-
SPG21	Mast syndrome,248900
KHDC3L	Hydatidiform mole,recurrent,2,614293
EIF4A3	Robin sequence with cleft mandible and limb abnormalities,268305
FOXI1	Enlarged vestibular aqueduct 600791
PRF1	Hemophagocytic lymphohistiocytosis,familial,2,603553
TONSL	Spondyloepimetaphyseal dysplasia,sponastrime type
THUMPD1	Intellectual disability and dysmorphic facies
C2orf69	Combined oxidative phosphorylation deficiency 53
CCN6	Arthropathy,progressive pseudorheumatoid,of childhood,208230 Spondyloepiphyseal dysplasia tarda with progressive arthropathy,208230
IL12B	Immunodeficiency 29,mycobacteriosis,614890
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy,218000
SCN1B	Epileptic encephalopathy,early infantile,52
SLC34A3	Hypophosphatemic rickets with hypercalciuria,241530
ARSA	Metachromatic leukodystrophy,250100
SLC34A2	Pulmonary alveolar microlithiasis,265100
ENTPD1	Spastic paraplegia 64,autosomal recessive,615683
MUSK	Fetal akinesia deformation sequence,208150 Myasthenic syndrome,congenital,9,associated with acetylcholine receptor deficiency,616325
NSMCE3	Lung diseaseimmunodeficiencyand chromosome breakage syndrome
SLC34A1	Fanconi renotubular syndrome 2,613388 Hypercalcemia,infantile 2,616963 Nephrolithiasis/osteoporosis,hypophosphatemic,1,612286
NSMCE2	Seckel syndrome 10
ANK3	Mental retardation,autosomal recessive,37
ANK1	Spherocytosis,type 1
GNPAT	Rhizomelic chondrodysplasia punctata,type 2,222765
SFRP4	Pyle disease,265900
ALPK3	Cardiomyopathyfamilial hypertrophic 27
KDELR2	-
CCNO	Ciliary dyskinesia,primary,29,615872
MMADHC	Homocystinuria,cblD type,variant 1,277410 Methylmalonic aciduria and homocystinuria,cblD type,277410 Methylmalonic aciduria,cblD type,variant 2,277410
GCDH	Glutaricaciduria,type I,231670
PEX11B	Peroxisome biogenesis disorder 14B,614920
PCDH15	Deafness,autosomal recessive 23,609533
PCDH12	Microcephalyseizuresspasticityand brain calcification
PIK3R1	Agammaglobulinemia 7,autosomal recessive,615214
OSGEP	Galloway-Mowat syndrome 3

PIK3R5	Ataxia-oculomotor apraxia 3,615217
ZMPSTE24	Restrictive dermopathy,lethal,275210
ANKH	mental retardation,deafness,ankylosis,and mild hypophosphatemia
CD19	Immunodeficiency,common variable,3,613493
SI	Sucrase-isomaltase deficiency,congenital,222900
DHX38	Retinitis pigmentosa 84
DYM	Dyggve-Melchior-Clausen disease,223800 Smith-McCort dysplasia,607326
ARSG	Usher syndrome,type IV
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy),253200
SEC31A	?Neurodevelopmental disorder with spastic quadriplegia,optic atrophy,seizures,and structural brain anomalies
CTSA	Galactosialidosis,256540
SURF1	Charcot-Marie-Tooth disease,type 4K
CRADD	Mental retardation,autosomal recessive 34,614499
INTS1	Neurodevelopmental disorder with cataracts,poor growth,and dysmorphic facies
XRCC4	Short stature,microcephaly,and endocrine dysfunction,616541
IQSEC1	Intellectual developmental disorder with short stature and behavioral abnormalities
PPP1R21	Neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities
XRCC2	?Fanconi anemiacomplementation group U
XRCC1	?Spinocerebellar ataxiaautosomal recessive 26
PRG4	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome,208250
ERAL1	Perrault syndrome 6
RPE65	Leber congenital amaurosis 2,204100 Retinitis pigmentosa 20,613794
TF	Atransferrinemia,209300
TG	Thyroid dyshormonogenesis 3,274700 {Autoimmune thyroid disease,susceptibility to,3},608175
TH	Segawa syndrome,recessive,605407
WNK1	Neuropathy,hereditary sensory and autonomic,type II,201300 Pseudohypoaldosteronism,type IIC,614492
ACER3	Leukodystrophy,progressive,early childhood-onset
COL9A1	Stickler syndrome,type IV,614134
CD27	Lymphoproliferative syndrome 2,615122
COL9A3	-
COL9A2	Stickler syndrome,type V,614284
INTS8	?Neurodevelopmental disorder with cerebellar hypoplasia and spasticity
RETREG1	Neuropathy,hereditary sensory and autonomic,type IIB,613115
VSX2	Microphthalmia with coloboma 3,610092 Microphthalmia,isolated 2,610093

FKBP10	Bruck syndrome 1,259450 Osteogenesis imperfecta,type XI,610968
PCSK1	Obesity with impaired prohormone processing 600955
CD40	Immunodeficiency with hyper-IgM,type 3,606843
FOXE3	Anterior segment dysgenesis 2multiple subtypes
FOXE1	Bamforth-Lazarus syndrome,241850
TNFRSF13B	Immunodeficiency,common variable,2,240500 Immunoglobulin A deficiency 2,609529
FKBP14	Ehlers-Danlos syndrome with progressive kyphoscoliosis,myopathy,and hearing loss,614557
GPAA1	Glycosylphosphatidylinositol biosynthesis defect 15 ,617810
PTPRQ	Deafness,autosomal recessive 84A,613391
PTPRO	Nephrotic syndrome,type 6,614196
TECR	Mental retardation,autosomal recessive 14,614020
KIF14	?Meckel syndrome 12,Microcephaly 20primaryautosomal recessive
SLC7A14	Retinitis pigmentosa 68,615725
CD3G	Immunodeficiency 17,CD3 gamma deficient,615607
KIF12	-
BRCA1	Fanconi anemiacomplementation group S
TNFRSF13C	Immunodeficiency,common variable,4,613494
BRCA2	Fanconi anemiacomplementation group D1
CD3E	Immunodeficiency 18,615615 Immunodeficiency 18,SCID variant,615615
IER3IP1	Microcephaly,epilepsy,and diabetes syndrome,614231
PEPD	Prolidase deficiency,170100
CD3D	Immunodeficiency 19,615617
ARV1	Epileptic encephalopathy,early infantile,38,617020
HYAL1	Mucopolysaccharidosis type IX ,601492
RSRC1	Intellectual developmental disorder,autosomal recessive 70
CTSK	Pycnodysostosis,265800
KIF1C	Spastic ataxia 2,autosomal recessive,611302
ACADM	Acyl-CoA dehydrogenase,medium chain,deficiency of,201450
CTSF	Ceroid lipofuscinosis,neuronal,13,Kufs type,615362
KIF1A	Neuropathy,hereditary sensory,type IIC,614213 Spastic paraplegia 30,autosomal recessive,610357
ACADS	Acyl-CoA dehydrogenase,short-chain,deficiency of,201470
CTSD	Ceroid lipofuscinosis,neuronal,10,610127
AASS	Hyperlysinemia 238700,Saccharopinuria 268700
CTSC	Haim-Munk syndrome,245010 Papillon-Lefevre syndrome,245000 Periodontitis 1,juvenile,170650
ACAD8	Isobutyryl-CoA dehydrogenase deficiency,611283
VKORC1	Vitamin K-dependent clotting factors,combined deficiency of,2,607473 Warfarin resistance,122700
MME	Charcot-Marie-Tooth disease,axonal,type 2T,617017 ?Spinocerebellar ataxia 43,617018
CCDC115	Congenital disorder of glycosylation,type IIo,616828
ACAD9	Mitochondrial complex I deficiency due to ACAD9 deficiency,611126

MRPS2	Combined oxidative phosphorylation deficiency 36 ,617950
ANO6	Scott syndrome,262890
ANO5	Miyoshi muscular dystrophy 3,613319 Muscular dystrophy,limb-girdle,type 2L,611307
CTU2	Microcephaly,facial dysmorphism,renal agenesis,and ambiguous genitalia syndrome
ENAM	Amelogenesis imperfecta,type IC,204650
PRKN	Parkinson disease,juvenile,type 2 600116
TMPRSS6	Iron-refractory iron deficiency anemia,206200
CCDC103	Ciliary dyskinesia,primary,17,614679
TMPRSS3	Deafness,autosomal recessive 8/10,601072
AKR1D1	Bile acid synthesis defect,congenital,2,235555
DYNLT2B	Short-rib thoracic dysplasia 17 with or without polydactyly
IGHMBP2	Charcot-Marie-Tooth disease,axonal,type 2S,616155 Neuronopathy,distal hereditary motor,type VI,604320
IBA57	?Spastic paraplegia 74autosomal recessive,Multiple mitochondrial dysfunctions syndrome 3
STX11	Hemophagocytic lymphohistiocytosis,familial,4,603552
CPN1	Carboxypeptidase N deficiency,212070
EXOSC5	-
EXOSC9	Cerebellar atrophy with spinal motor neuropathy
PERP	-
EXOSC8	Pontocerebellar hypoplasia,type 1C,616081
CD59	Hemolytic anemia,CD59-mediated,with or without immune-mediated polyneuropathy,612300
EXOSC3	Pontocerebellar hypoplasia,type 1B,614678
EXOSC2	Short stature,hearing loss,ritinitis pigmentosa and distinctive facies
GPC6	Omodysplasia 1,258315
CCT5	Neuropathy,hereditary sensory,with spastic paraplegia,256840
CD55	Complement hyperactivation,angiopathic thrombosis,and protein-losing enteropathy
LNPK	Neurodevelopmental disorder with epilepsy and hypoplasia of the corpus callosum
CAVIN1	Lipodystrophy,congenital generalized,type 4 613327
GCH1	Hyperphenylalaninemia,BH4-deficient,B,233910
ERCC6L2	Bone marrow failure syndrome 2,615715
NUBPL	Mitochondrial complex I deficiency,252010
CRYBB1	Cataract 17,multiple types,611544
CRIPT	Short stature with microcephaly and distinctive facies,615789
CRYBB3	Cataract 22,autosomal recessive,609741
AKR1C2	46XY sex reversal 8,614279 Obesity,hyperphagia,and developmental delay
ALDH4A1	Hyperprolinemia,type II,239510
DPY19L2	Spermatogenic failure 9,613958
DPAGT1	Congenital disorder of glycosylation,type Ij,608093 Myasthenic syndrome,congenital,13,with tubular aggregates,614750

SCARB2	Epilepsy,progressive myoclonic 4,with or without renal failure,254900
PLVAP	Diarrhea 10,protein-losing enteropathy type
DCLRE1C	Omenn syndrome,603554Severe combined immunodeficiency,Athabascan type,602450
CD81	Immunodeficiency,common variable,6,613496
MPI	Congenital disorder of glycosylation,type Ib,602579
TRAC	Immunodeficiency 7,TCR-alpha/beta deficient,615387
IHH	Acrocapitofemoral dysplasia,607778 Brachydactyly,type A1,112500
MPL	Thrombocytopenia,congenital amegakaryocytic,604498
CPOX	Coproporphyria,121300 Harderoporphyria,121300
OGDHL	Yoon-Bellen neurodevelopmental syndrome
GIMAP5	-
CFAP300	-
MPO	Myeloperoxidase deficiency,254600 {Alzheimer disease,susceptibility to},104300 {Lung cancer,protection against,in smokers}
FRRS1L	Epileptic encephalopathy,early infantile,37,616981
MPZ	Dejerine-Sottas disease
DUOXA2	Thyroid dyshormonogenesis 5,274900
NPPA	Atrial standstill 2,615745
SCN9A	Insensitivity to pain,congenital,243000
CAPN3	Muscular dystrophy,limb-girdle,type 2A,253600
SNIP1	Psychomotor retardation,epilepsy,and craniofacial dysmorphism,614501
BANF1	Nestor-Guillermo progeria syndrome,614008
CAPN1	Spastic paraplegia 76,autosomal recessive,616907
GGT1	Glutathioninuria 231950
SBF1	Charcot-Marie-Tooth disease,type 4B3,615284
SBF2	Charcot-Marie-Tooth disease,type 4B2,604563
SLC30A9	?Birk-Landau-Perez syndrome
HFM1	Premature ovarian failure 9,615724
CYBA	Chronic granulomatous disease,autosomal,due to deficiency of CYBA,233690
MAT1A	Methionine adenosyltransferase deficiencyautosomal recessive,Hypermethioninemiapersistentautosomal dominantdue to methionine adenosyltransferase I/III deficiency
RPGRIP1L	COACH syndrome,216360 Joubert syndrome 7,611560 Meckel syndrome 5,611561
ATAD3A	Harel-Yoon syndrome
KLHL40	Nemaline myopathy 8,autosomal recessive,615348
KLHL41	Nemaline myopathy 9,615731
CD8A	CD8 deficiency,familial,608957
DOK7	Myasthenic syndrome,congenital,10,254300
GPD1	Hypertriglyceridemia,transient infantile,614480
GEMIN4	Neurodevelopmental disorder with microcephalycataractsand renal abnormalities
SBDS	Shwachman-Diamond syndrome

GEMIN5	-
MASP1	3MC syndrome 1,257920
RBM8A	Thrombocytopenia-absent radius syndrome,274000
NPR2	Acromesomelic dysplasia,Maroteaux type,602875
CHAT	Myasthenic syndrome,congenital,6,presynaptic,254210
NPR3	-
SLC1A1	Dicarboxylic aminoaciduria,222730 {?Schizophrenia susceptibility 18},615232
SLC1A4	Spastic tetraplegia,thin corpus callosum,and progressive microcephaly,616657
NDRG1	Charcot-Marie-Tooth disease,type 4D,601455
RASGRP1	Immunodeficiency 64
ACACA	Acetyl-CoA carboxylase deficiency
TOR1AIP1	?Muscular dystrophylimb-girdletype 2Y
SPTA1	Elliptocytosis-2,130600 Pyropoikilocytosis,266140 Spherocytosis,type 3,270970
FUT8	Congenital disorder of glycosylation with defective fucosylation,618005
CYP11A1	Adrenal insufficiency,congenital,with 46XY sex reversal,partial or complete,613743
DEAF1	?Dyskinesia,seizures,and intellectual developmental disorder
NPHP1	Joubert syndrome 4,609583 Nephronophthisis 1,juvenile,256100 Senior-Loken syndrome-1,266900
NPHP3	Meckel syndrome 7,267010 Nephronophthisis 3,604387 Renal-hepatic-pancreatic dysplasia 1,208540
NPHP4	Nephronophthisis 4,606966 Senior-Loken syndrome 4,606996
TAC3	Hypogonadotropic hypogonadism 10 with or without anosmia,614839
CYC1	Mitochondrial complex III deficiency,nuclear type 6,615453
NDUFV2	Mitochondrial complex I deficiency,252010
PRODH	Hyperprolinemia,type I,239500 {Schizophrenia,susceptibility to,4},600850
VARS2	Combined oxidative phosphorylation deficiency 20,615917
WNT1	Osteogenesis imperfecta,type XV,615220 {Osteoporosis,early-onset,susceptibility to,autosomal dominant},615221
NDUFV1	Mitochondrial complex I deficiency,252010
WNT3	Tetra-amelia syndrome 273395
VARS1	Neurodevelopmental disorder with microcephalyseizuresand cortical atrophy
WNT4	Mullerian aplasia and hyperandrogenism,158330 SERKAL syndrome,611812
VDR	Rickets,vitamin D-resistant,type IIA,277440 ?Osteoporosis,involutional,166710
SLC52A2	Brown-Vialetto-Van Laere syndrome 2,614707
SLC52A3	Brown-Vialetto-Van Laere syndrome 1,211530 Fazio-Londe disease,211500
DOLK	Congenital disorder of glycosylation,type Im,610768
ISG15	Immunodeficiency 38,616126

PHOX2A	Fibrosis of extraocular muscles,congenital,2,602078
QDPR	Hyperphenylalaninemia,BH4-deficient,C,261630
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency,230450 {Myocardial infarction,susceptibility to},608446
BMP1	Osteogenesis imperfecta,type XIII,614856
PROC	Thrombophilia due to protein C deficiency,autosomal recessive,612304
CPS1	Carbamoylphosphate synthetase I deficiency,237300 {Pulmonary hypertension,neonatal,susceptibility to},615371 {Venoocclusive disease after bone marrow transplantation}
CYP11B2	Hypoaldosteronism,congenital,due to CMO I deficiency,203400
CYP11B1	Adrenal hyperplasia,congenital,due to 11-beta-hydroxylase deficiency,202010
STT3A	Congenital disorder of glycosylation,type Iw
STT3B	Congenital disorder of glycosylation,type Ix
LAMC3	Cortical malformations,occipital,614115
DNAAF11	Ciliary dyskinesia,primary,19,614935
ATP2A1	Brody myopathy,601003
LAMC2	Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
MTR	Homocystinuria-megaloblastic anemia,cblG complementation type,250940 {Neural tube defects,folate-sensitive,susceptibility to},601634
SOBP	Mental retardation,anterior maxillary protrusion,and strabismus,613671
GPHN	Molybdenum cofactor deficiency C,615501
ABHD16A	Spastic paraplegia 86, autosomal recessive
CPT2	CPT deficiency,hepatic,type II,600649 CPT II deficiency,lethal neonatal,608836 Myopathy due to CPT II deficiency,255110 {Encephalopathy,acute,infection-induced,4,susceptibility to},614212
NUP62	Striatonigral degeneration,infantile,271930
UQCRFS1	Mitochondrial complex III deficiency, nuclear type 10
SCARF2	Van den Ende-Gupta syndrome,600920
BCKDHA	Maple syrup urine disease,type Ia,248600
DDX59	Orofaciodigital syndrome V,174300
POU1F1	Pituitary hormone deficiency,combined,1,613038
GLRX5	Anemia,sideroblastic,3,pyridoxine-refractory,616860 Spasticity,childhood-onset,with hyperglycinemia,616859
MMP2	Multicentric osteolysis,nodulosis,and arthropathy,259600
BCKDHB	Maple syrup urine disease,type Ib,248600
ARHGEF18	Retinitis pigmentosa 78
DARS2	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation,611105
PLA2G4A	-
PEX2	Peroxisome biogenesis disorder 5A (Zellweger),614866 Peroxisome biogenesis disorder 5B,614867

PEX1	Heimler syndrome 1,234580 Peroxisome biogenesis disorder 1A (Zellweger),214100 Peroxisome biogenesis disorder 1B (NALD/IRD),601539
MMP9	Metaphyseal anadysplasia 2,613073
DARS1	Hypomyelination with brainstem and spinal cord involvement and leg spasticity,615281
TRDN	Ventricular tachycardia,catecholaminergic polymorphic,5,with or without muscle weakness,615441
PEX7	Peroxisome biogenesis disorder 9B,614879 Rhizomelic chondrodysplasia punctata,type 1,215100
LMBRD1	Methylmalonic aciduria and homocystinuria,cblF type,277380
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency,256000
NDUFS7	Leigh syndrome,256000
NDUFS6	Mitochondrial complex I deficiency,252010
PEX3	Peroxisome biogenesis disorder 10A (Zellweger),614882
CDHR1	Cone-rod dystrophy 15,613660 Retinitis pigmentosa 65,613660
PEX6	Heimler syndrome 2,616617 Peroxisome biogenesis disorder 4A (Zellweger),614862 Peroxisome biogenesis disorder 4B,614863
NDUFS4	Leigh syndrome,256000 Mitochondrial complex I deficiency,252010
PEX5	Peroxisome biogenesis disorder 2A (Zellweger),214110 Peroxisome biogenesis disorder 2B,202370 Rhizomelic chondrodysplasia punctata,type 5,616716
TBC1D7	Macrocephaly/megalencephaly syndrome,autosomal recessive,248000
IRF7	?Immunodeficiency 39
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency,256000 Mitochondrial complex I deficiency,252010
IRF8	Immunodeficiency 32B,monocyte and dendritic cell deficiency,autosomal recessive,614894
NDUFS2	Mitochondrial complex I deficiency,252010
NDUFS1	Mitochondrial complex I deficiency,252010
NUP54	-
SCN4A	Myasthenic syndrome,congenital,16,614198
ZP1	Oocyte maturation defect 1,615774
CFAP53	Heterotaxyvisceral6autosomal recessive
BLVRA	Hyperbiliverdinemia,614156
IRF9	Immunodeficiency 65,susceptibility to viral infections
LAMA5	-
MVK	Hyper-IgD syndrome,260920 Mevalonic aciduria,610377 Porokeratosis 3,multiple types,175900
LAMA2	Muscular dystrophy,congenital merosin-deficient,607855 Muscular dystrophy,congenital,due to partial LAMA2 deficiency,607855
LAMA1	Poretti-Boltshauser syndrome,615960

GOSR2	Epilepsy,progressive myoclonic 6,614018
PNKP	Ataxia-oculomotor apraxia 4,616267 Microcephaly,seizures,and developmental delay,613402
LAMA3	Epidermolysis bullosa,generalized atrophic benign,226650 Epidermolysis bullosa,junctional,Herlitz type,226700 Laryngoonychocutaneous syndrome,245660
PARN	Dyskeratosis congenita,autosomal recessive 6,616353 Pulmonary fibrosis and/or bone marrow failure,telomere-related,4,616371
HMGCR	-
CLN8	Ceroid lipofuscinosis,neuronal,8,600143 Ceroid lipofuscinosis,neuronal,8,Northern epilepsy variant,610003
SAMHD1	Aicardi-Goutieres syndrome 5,612952 ?Chilblain lupus 2,614415
AURKC	Spermatogenic failure 5,243060
CLCN2	Leukoencephalopathy with ataxia,615651
CLCN1	Myotonia congenita,recessive,255700
CLN6	Ceroid lipofuscinosis,neuronal,6,601780 Ceroid lipofuscinosis,neuronal,Kufs type,adult onset,204300
CLN5	Ceroid lipofuscinosis,neuronal,5,256731
ATOH7	Persistent hyperplastic primary vitreous,autosomal recessive,221900
CLN3	Ceroid lipofuscinosis,neuronal,3,204200
HMGCL	HMG-CoA lyase deficiency,246450
CLMP	Congenital short bowel syndrome,615237
MARVELD2	Deafness,autosomal recessive 49,610153
SH3TC2	Charcot-Marie-Tooth disease,type 4C,601596 Mononeuropathy of the median nerve,mild,613353
TAF1C	developmental delay severe,cerebral atrophia,lack of speech,lack of ambulation,epilepsy,PMID: 32779182
EPS8L2	Deafness autosomal recessive 106
VHL	Erythrocytosis,familial,2 263400
MBTPS1	?Spondyloepiphyseal dysplasia,Kondo-Fu type
GALNT3	Tumoral calcinosis,hyperphosphatemic,familial,211900
LAMB3	Amelogenesis imperfecta,type IA,104530 Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
EGF	Hypomagnesemia 4,renal,611718
GALNT2	Congenital disorder of glycosylation, type IIt
LAMB2	Nephrotic syndrome,type 5,with or without ocular abnormalities,614199 Pierson syndrome,609049
TAF13	Mental retardation,autosomal recessive 60
LAMB1	Lissencephaly 5,615191
SOD1	Amyotrophic lateral sclerosis 1,105400
CLCN7	Osteopetrosis,autosomal recessive 4,611490
RPIA	Ribose 5-phosphate isomerase deficienc 608611
CYP24A1	Hypercalcemia,infantile,143880
NPHS1	Nephrotic syndrome,type 1,256300

GLB1	GM1-gangliosidosis,type I,230500 GM1-gangliosidosis,type II,230600 GM1-gangliosidosis,type III,230650 Mucopolysaccharidosis type IVB (Morquio),253010
NPHS2	Nephrotic syndrome,type 2,600995
TAF6	Alazami-Yuan syndrome,617126
CCDC174	Hypotonia,infantile,with psychomotor retardation,616816
TAF2	Mental retardation,autosomal recessive 40,615599
NUP37	?Microcephaly 24,primary,autosomal recessive
CLPB	3-methylglutaconic aciduria,type VII,with cataracts,neurologic involvement and neutropenia,616271
GLDC	Glycine encephalopathy,605899
COL12A1	Ullrich congenital muscular dystrophy 2,616470
SERPINE1	Plasminogen activator inhibitor-1 deficiency,613329 {Transcription of plasminogen activator inhibitor,modulator of}
RAX	Microphthalmia,isolated 3,611038
ABAT	GABA-transaminase deficiency,613163
MICOS13	Combined oxidative phosphorylation deficiency 37
ABCA12	Ichthyosis,autosomal recessive 4B (harlequin),242500 Ichthyosis,congenital,autosomal recessive 4A,601277
ENO3	Glycogen storage disease XIII 612932
GLDN	Lethal congenital contracture syndrome 11
GCSH	Glycine encephalopathy,605899
GLE1	Lethal congenital contracture syndrome 1,253310
GNAT2	Achromatopsia-4,613856
CLP1	Pontocerebellar hypoplasia,type 10,615803
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome,607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940
CREB3L1	Osteogenesis imperfectatype XVI
FDXR	Auditory neuropathy and optic atrophy,617717
CYP1B1	Glaucoma 3A,primary open angle,congenital,juvenile,or adult onset,231300 Peters anomaly,604229
HOXA2	Microtia,hearing impairment,and cleft palate (AR) 612290
PLCE1	Nephrotic syndrome,type 3,610725
HOXA1	Athabaskan brainstem dysgenesis syndrome,601536 Bosley-Salih-Alorainy syndrome,601536
RTTN	Microcephaly,short stature,and polymicrogyria with seizures,614833
MMUT	Methylmalonic aciduria,mut(0) type,251000
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type),256810
BVES	?Muscular dystrophylimb-girdletype 2X
EARS2	Combined oxidative phosphorylation deficiency 12,614924
GPR179	Night blindness,congenital stationary (complete),1E,autosomal recessive,614565
CUBN	Megaloblastic anemia-1,Finnish type,261100
MECR	Dystonia,childhood-onset,with optic atrophy and basal ganglia abnormalities

NKX2-6	Conotruncal heart malformations,217095 Persistent truncus arteriosus,217095
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome,608800
SERPINF1	Osteogenesis imperfecta,type VI,613982
SERPINF2	Alpha-2-plasmin inhibitor deficiency,262850
IFT80	Short-rib thoracic dysplasia 2 with or without polydactyly,611263
HOGA1	Hyperoxaluria,primary,type III,613616
UNC13D	Hemophagocytic lymphohistiocytosis,familial,3,608898
SULT2B1	Ichthyosiscongenitalautosomal recessive 14
BIN1	Myopathy,centronuclear,autosomal recessive,255200
IFT81	Short-rib thoracic dysplasia 19 with or without polydactyly
CLPP	Perrault syndrome 3,614129
FDX2	Mitochondrial myopathy,episodic,with optic atrophy and reversible leukoencephalopathy
SERPING1	Angioedema,hereditary,types I and II,106100 Complement component 4,partial deficiency of,120790
YME1L1	Optic atrophy 11
NHLRC1	Epilepsy,progressive myoclonic 2B (Lafora),254780
HOXB1	Facial paresis,hereditary congenital,3,614744
NHLRC2	FINCA syndrome
LINS1	Mental retardation,autosomal recessive 27,614340
RD3	Leber congenital amaurosis 12,610612
MEFV	Familial Mediterranean fever,AD,134610 Familial Mediterranean fever,AR,249100
PRDX3	-
CHST11	?Osteochondrodysplasia,brachydactyly,and overlapping malformed digits
PRDX1	MethylmalonicaciduriaandhomocystinuriacblC typedigenic
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency,610090
SERPINH1	Osteogenesis imperfectatype X
SLC19A3	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2),607483
CHST14	Ehlers-Danlos syndrome,musculocontractural type 1,601776
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome,249270
GRXCR1	Deafness,autosomal recessive 25,613285
WNT10B	Split-hand/foot malformation 6,225300
DNAH11	Ciliary dyskinesia,primary,7,with or without situs inversus,611884
ST14	Ichthyosis,congenital,autosomal recessive 11,602400
WNT10A	Odontoonychodermal dysplasia,257980 Schopf-Schulz-Passarge syndrome,224750 Tooth agenesis,selective,4,150400
ZNF142	Neurodevelopmental disorder with impaired speech and hyperkinetic movements
ACE	Renal tubular dysgenesis,267430
CHKB	Muscular dystrophy,congenital,megaconial type,602541

FZD6	Nail disorder,nonsyndromic congenital,10,(claw-shaped nails),614157
RDX	Deafness,autosomal recessive 24,611022
MTHFR	Homocystinuria due to MTHFR deficiency,236250 {Neural tube defects,susceptibility to},601634 {Schizophrenia,susceptibility to},181500 {Thromboembolism,susceptibility to},188050 {Vascular disease,susceptibility to}
TRIP4	Spinal muscular atrophy with congenital bone fractures 1,616866 ?Muscular dystrophy,congenital,Davignon-Chauveau type,617066
MTHFS	Neurodevelopmental disorder with microcephaly,epilepsy,and hypomyelination
CDC6	Meier-Gorlin syndrome 5,613805
SLC6A5	Hyperekplexia 3,614618
TMEM199	Congenital disorder of glycosylation,type IIp,616829
FA2H	Spastic paraplegia 35,autosomal recessive,612319
FIBP	Thauvin-Robinet-Faivre syndrome
GIPC3	Deafness,autosomal recessive 15,601869
SLC6A9	Glycine encephalopathy with normal serum glycine
OGDH	Alpha-ketoglutarate dehydrogenase deficiency 203740
CDK10	Al Kaissi syndrome ,617694
REN	Renal tubular dysgenesis 267430
ODAPH	Amelogenesis imperfecta,type IIA4,614832
ADA	Adenosine deaminase deficiency,partial,102700 Severe combined immunodeficiency due to ADA deficiency,102700
ITK	Lymphoproliferative syndrome 1,613011
TRMU	Liver failure,transient infantile,613070 {Deafness,mitochondrial,modifier of},580000
SERPINA1	Emphysema due to AAT deficiency,613490 Emphysema-cirrhosis,due to AAT deficiency,613490 Hemorrhagic diathesis due to \'antithrombin\' Pittsburgh,613490 {Pulmonary disease,chronic obstructive,susceptibility to},606963
ADK	Hypermethioninemia due to adenosine kinase deficiency
FMN2	Mental retardation,autosomal recessive 47,616193
SERPINA6	Corticosteroid-binding globulin deficiency,611489
SLC6A3	Parkinsonism-dystonia,infantile,613135 {Nicotine dependence,protection against},188890
LFNG	?Spondylocostal dysostosis 3autosomal recessive
DSTYK	Spastic paraplegia 23
IMPA1	Mental retardation,autosomal recessive 59
TRIM2	Charcot-Marie-Tooth disease,type 2R,615490
RSPO2	?Humerofemoral hypoplasia with radiotibial ray deficiency,Tetraamelia syndrome 2
RSPO1	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal,610644
RSPO4	Anonychia congenita,206800
HAVCR2	T-cell lymphoma,subcutaneous panniculitis-like

IFNGR1	Immunodeficiency 27A,mycobacteriosis,AR,209950
SLC2A10	Arterial tortuosity syndrome,208050
IFNGR2	Immunodeficiency 28,mycobacteriosis,614889
TET2	-
PAX3	Waardenburg syndrome,type 3
TACR3	Hypogonadotropic hypogonadism 11 with or without anosmia,614840
BAAT	Hypercholanemia,familial,607748
SERPINB7	Palmoplantar keratoderma,Nagashima type,615598
PRSS12	Mental retardation,autosomal recessive 1,249500
SERPINB8	Peeling skin syndrome 5
PAX1	Otofaciocervical syndrome 2
MRAP	Glucocorticoid deficiency 2,607398
RNF168	RIDDLE syndrome,611943
PLCB4	Auriculocondylar syndrome 2
RIPPLY2	?Spondylocostal dysostosis 6
PAX7	Myopathy,congenital,progressive,with scoliosis
IVD	Isovaleric acidemia,243500
TET3	-
PLCB1	Epileptic encephalopathy,early infantile,12,613722
RGR	Retinitis pigmentosa 44,613769
OCA2	Albinism,brown oculocutaneous,203200 Albinism,oculocutaneous,type II,203200 [Skin/hair/eye pigmentation 1,blond/brown hair],227220 [Skin/hair/eye pigmentation 1,blue/nonblue eyes],227220
ASAH1	Farber lipogranulomatosis,228000 Spinal muscular atrophy with progressive myoclonic epilepsy,159950
CTDP1	Congenital cataracts,facial dysmorphism,and neuropathy,604168
SERPINC1	Thrombophilia due to antithrombin III deficiency,613118
TMEM165	Congenital disorder of glycosylation,type IIk,614727
CHRND	Multiple pterygium syndrome,lethal type,253290
LMAN1	Combined factor V and VIII deficiency,227300
CHRNG	Escobar syndrome,265000 Multiple pterygium syndrome,lethal type,253290
CHRNE	Myasthenic syndrome,congenital,4A,slow-channel,605809 Myasthenic syndrome,congenital,4B,fast-channel,616324 Myasthenic syndrome,congenital,4C,associated with acetylcholine receptor deficiency,608931
GCNT2	Cataract 13 with adult i phenotype,116700
RHO	Retinitis pigmentosa 4,autosomal dominant or recessive ,613731
SC5D	Lathosterolosis,607330
AGA	Aspartylglucosaminuria,208400
WWOX	Epileptic encephalopathy,early infantile,28
TIMMDC1	Mitochondrial complex I deficiency,nuclear type 31
BRF1	Cerebellofaciodental syndrome,616202
F10	Factor X deficiency,227600
AGK	Cataract 38,autosomal recessive,614691 Sengers syndrome,212350

AGL	Glycogen storage disease IIIa,232400 Glycogen storage disease IIIb,232400
F12	Factor XII deficiency 234000
NDE1	Lissencephaly 4 (with microcephaly),614019 ?Microhydranencephaly,605013
CARD9	Candidiasis,familial,2,autosomal recessive,212050
F11	Factor XI deficiency,autosomal recessive,612416
TRMT1	Mental retardation,autosomal recessive 68
FMO3	Trimethylaminuria,602079
ADAT3	Mental retardation,autosomal recessive 36,615286
AGT	Renal tubular dysgenesis,267430
GTF2H5	Trichothiodystrophy 3,photosensitive,616395
LRPPRC	Leigh syndrome,French-Canadian type,220111
AGBL5	Retinitis pigmentosa 75,617023
FIG4	Charcot-Marie-Tooth disease,type 4J,611228 Yunis-Varon syndrome,216340 ?Polymicrogyria,bilateral temporooccipital,612691
NMNAT1	Leber congenital amaurosis 9,608553
RFWD3	?Fanconi anemiacomplementation group W
TRMT5	Combined oxidative phosphorylation deficiency 26,616539
ALMS1	Alstrom syndrome,203800
HYLS1	Hydrolethalus syndrome,236680
MSRB3	Deafness,autosomal recessive 74,613718
LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein,610798
PLCD1	Nail disorder,nonsyndromic congenital,3,(leukonychia),151600
CHRM3	?Prune belly syndrome
DGKE	Nephrotic syndrome,type 7,615008 {Hemolytic uremic syndrome,atypical,susceptibility to,7},615008
ATP8A2	Cerebellar ataxia,mental retardation,and dysequilibrium syndrome 4
CSF3R	-
EPO	?Diamond-Blackfan anemia-like
TUSC3	Mental retardation,autosomal recessive 7,611093
ITGA2B	Glanzmann thrombasthenia,273800 Thrombocytopenia,neonatal alloimmune,BAK antigen related
AHR	?Retinitis pigmentosa 85
ABHD12	Polyneuropathy,hearing loss,ataxia,retinitis pigmentosa,and cataract,612674
CDH3	Ectodermal dysplasia,ectrodactyly,and macular dystrophy,225280 Hypotrichosis,congenital,with juvenile macular dystrophy,601553
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B),252920 ?Charcot-Marie-Tooth disease,axonal,type 2V,616491
IYD	Thyroid dyshormonogenesis 4,274800
CHP1	?Spastic ataxia 9,autosomal recessive
NBN	Aplastic anemia,609135 Leukemia,acute lymphoblastic,613065 Nijmegen breakage syndrome,251260

GYG1	Polyglucosan body myopathy 2,616199 ?Glycogen storage disease XV,613507
TECTA	Deafness,autosomal recessive 21,603629
LHCGR	Leydig cell hypoplasia with hypergonadotropic hypogonadism,238320
NKX6-2	Spastic ataxia 8autosomal recessivewith hypomyelinating leukodystrophy
GPT2	Mental retardation,autsomal recessive 49
TAP2	Bare lymphocyte syndrome,type I,due to TAP2 deficiency,604571 Wegener-like granulomatosis
TAP1	Bare lymphocyte syndrome,type I,604571
MITF	COMMAD syndrome
TRAIP	Seckel syndrome 9,616777
BSCL2	Encephalopathy,progressive,with or without lipodystrophy,615924 Lipodystrophy,congenital generalized,type 2,269700
CLIP1	intellectual disability
STIM1	Immunodeficiency 10 612783
MADD	Neurodevelopmental disorder with dysmorphic facies, impaired speech and hypotonia,DEEAH syndrome
NHP2	Dyskeratosis congenita,autosomal recessive 2,613987
ADAM9	Cone-rod dystrophy 9,612775
TNIK	Mental retardation,autosomal recessive 54,617028
CSF1R	Brain abnormalities,neurodegeneration,and dysosteosclerosis
CUL7	3-M syndrome 1,273750
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness,609057 [
FECH	Protoporphyria,erythropoietic,autosomal recessive,177000
AK1	Hemolytic anemia due to adenylate kinase deficiency,612631
AK2	Reticular dysgenesis,267500
ILDR1	Deafness,autosomal recessive 42,609646
ITPR1	Gillespie syndrome
ABHD5	Chanarin-Dorfman syndrome,275630
CYP4F22	Ichthyosis,congenital,autosomal recessive 5,604777
GTF2E2	Trichothiodystrophy 6,nonphotosensitive,616943
KLC2	Spastic paraplegia,optic atrophy,and neuropathy,609541
PSAP	Combined SAP deficiency,611721 Gaucher disease,atypical,610539 Krabbe disease,atypical,611722 Metachromatic leukodystrophy due to SAP-b deficiency,249900
RNF170	-
IGFBP7	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis,614224
IMPG2	Retinitis pigmentosa 56,613581
MPDU1	Congenital disorder of glycosylation,type If,609180
STRA6	Microphthalmia,isolated,with coloboma 8,601186 Microphthalmia,syndromic 9,601186
TOR1A	Arthrogryposis multiplex congenita 5

ATP8B1	Cholestasis,benign recurrent intrahepatic,243300 Cholestasis,progressive familial intrahepatic 1,211600
WFS1	Wolfram syndrome,222300
ESRRB	Deafness,autosomal recessive 35,608565
LSM1	PMID: 36100156
ASNS	Asparagine synthetase deficiency,615574
DHCR24	Desmosterolosis,602398
NEB	Nemaline myopathy 2,autosomal recessive,256030
TECRL	Ventricular tachycardiacatecholaminergic polymorphic3
CDK6	?Microcephaly 12primaryautosomal recessive
CDK5	?Lissencephaly 7 with cerebellar hypoplasia
ALB	Analbuminemia,616000 [Dysalbuminemic hyperthyroxinemia],615999
ESAM	-
DHCR7	Smith-Lemli-Opitz syndrome,270400
LZTR1	Noonan syndrome 2
TUBA8	Polymicrogyria with optic nerve hypoplasia,613180
WRAP53	Dyskeratosis congenita,autosomal recessive 3,613988
TREM2	Nasu-Hakola disease,221770
PTPN23	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity
SGCD	Muscular dystrophy,limb-girdle,type 2F,601287
GM2A	GM2-gangliosidosis,AB variant,272750
OPA1	Behr syndrome
SGCA	Muscular dystrophy,limb-girdle,type 2D,608099
SGCB	Muscular dystrophy,limb-girdle,type 2E,604286
OPA3	3-methylglutaconic aciduria,type III
CNGA1	Retinitis pigmentosa 49,613756
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1,242860
HYDIN	Ciliary dyskinesia,primary,5,608647
ZNF526	-
CNGA3	Achromatopsia-2,216900
SOST	Sclerosteosis 1,269500 Van Buchem disease,239100
SLC37A4	Glycogen storage disease Ib,232220 Glycogen storage disease Ic,232240
SGCG	Muscular dystrophy,limb-girdle,type 2C,253700
TAPT1	Osteochondrodysplasia,complex lethal,Symoens-Barnes-Gistelinck type,616897
IQCB1	Senior-Loken syndrome 5,609254
AMN	Megaloblastic anemia-1,Norwegian type,261100
GPX4	Spondylometaphyseal dysplasia,Sedaghatian type,250220
VWF	von Willibrand disease,type 3
COL25A1	Fibrosis of extraocular muscles,congenital,5,616219
ELAC2	Combined oxidative phosphorylation deficiency 17,615440 {Prostate cancer,hereditary,2,susceptibility to},614731
AMT	Glycine encephalopathy,605899
SORD	Sorbitol dehydrogenase deficiency with peripheral neuropathy
PLPBP	Epilepsyearly-onsetvitamin B6-dependent

NGF	Neuropathy,hereditary sensory and autonomic,type V,608654
COASY	Neurodegeneration with brain iron accumulation 6,615643
PATL2	Oocyte maturation defect 4,614661
METTL23	Mental retardation,autosomal recessive 44,615942
ASPM	Microcephaly 5,primary,autosomal recessive,608716
RP1	Retinitis pigmentosa 1,180100
SARS1	?Neurodevelopmental disorder with microcephalyataxiaand seizures
EVC	Ellis-van Creveld syndrome,225500
SARS2	Hyperuricemia,pulmonary hypertension,renal failure,and alkalosis,613845
ASPH	Traboulsi syndrome,601552
CNGB3	Achromatopsia-3,262300 Macular degeneration,juvenile,248200
ZNF513	Retinitis pigmentosa 58,613617
ASPA	Canavan disease,271900
CNGB1	Retinitis pigmentosa 45,613767
SLC24A1	Night blindness,congenital stationary (complete),1D,autosomal recessive,613830
SLC24A4	Amelogenesis imperfecta,type IIA5,615887 [Skin/hair/eye pigmentation 6,blond/brown hair],210750 [Skin/hair/eye pigmentation 6,blue/green eyes],210750
SMARCD2	Specific granule deficiency 2
WBP2	Deafnessautosomal recessive 107
TYRP1	Albinism,oculocutaneous,type III,203290 [Skin/hair/eye pigmentation,variation in,11 (Melanesian blond hair)],612271
MESD	-
MRM2	?Mitochondrial DNA depletion syndrome 17
DCAF17	Woodhouse-Sakati syndrome,241080
RBBP8	Jawad syndrome,251255 Pancreatic carcinoma,somatic Seckel syndrome 2,606744
TNNI3	Cardiomyopathy,dilated,2A,611880
FNIP1	-
SEMA4A	Cone-rod dystrophy 10,Retinitis pigmentosa 35
KCNJ10	Enlarged vestibular aqueduct,digenic,600791 SESAME syndrome,612780
IRX5	Hamamy syndrome,611174
KCNJ11	Hyperinsulinemic hypoglycemia,familial,2,601820
EFL1	Shwachman-Diamond syndrome 2
KCNJ13	Leber congenital amaurosis 16,614186 Snowflake vitreoretinal degeneration,193230
DDHD2	Spastic paraplegia 54,autosomal recessive,615033
KCNJ16	-
DDHD1	Spastic paraplegia 28,autosomal recessive,609340
PTPN14	Choanal atresia and lymphedema,613611
ASS1	Citrullinemia,215700
TRIT1	Combined oxidative phosphorylation deficiency 35

RPGRIP1	Cone-rod dystrophy 13,608194 Leber congenital amaurosis 6,613826
NFASC	Neurodevelopmental disorder with central and peripheral motor dysfunction
NIN	Seckel syndrome 7,614851
RAD50	Nijmegen breakage syndrome-like disorder,613078
GLRB	Hyperekplexia 2,autosomal recessive,614619
SPAG1	Ciliary dyskinesia,primary,28,615505
FOLR1	Neurodegeneration due to cerebral folate transport deficiency,613068
MMACHC	Methylmalonic aciduria and homocystinuria,cblC type,277400
PFKM	Glycogen storage disease VII,232800
SLC24A5	Albinism,oculocutaneous,type VI,113750 [Skin/hair/eye pigmentation 4,fair/dark skin],113750
MYORG	Basal ganglia calcification, idiopathic, 7, autosomal recessive
SLC46A1	Folate malabsorption,hereditary,229050
SPARC	Osteogenesis imperfecta,type XVII,616507
ARL6IP1	?Spastic paraplegia 61autosomal recessive
HPSE2	Urofacial syndrome 1,236730
TP53RK	Galloway-Mowat syndrome 4
GORAB	Geroderma osteodysplasticum,231070
PJVK	Deafness,autosomal recessive 59,610220
ANTXR2	Hyaline fibromatosis syndrome,228600
NADSYN1	Vertebral, cardiac, renal, and limb defects syndrome 3
ANTXR1	GAPO syndrome,230740
GOLGA2	-
LRMDA	Albinism,oculocutaneous,type VII,615179
EDNRB	ABCD syndrome,600501 Waardenburg syndrome,type 4A,277580 {Hirschsprung disease,susceptibility to,2},600155
RUSC2	Mental retardation,autosomal recessive 61
B2M	Immunodeficiency 43,241600 ?Amyloidosis,familial visceral,105200
GLUL	Glutamine deficiency,congenital,610015
EYS	Retinitis pigmentosa 25,602772
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency,210210
SLC6A19	Hartnup disorder,234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC6A17	Mental retardation,autosomal recessive 48,616269
SP110	Hepatic venoocclusive disease with immunodeficiency,235550 {Mycobacterium tuberculosis,susceptibility to},607948
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency,210200
SLC11A2	Anemia,hypochromic microcytic,with iron overload 1,206100
CYP27A1	Cerebrotendinous xanthomatosis,213700
AFG3L2	Ataxia,spastic,5,autosomal recessive,614487
BOLA3	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia,614299
TFAM	?Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)

WDPCP	Bardet-Biedl syndrome 15 615992,Congenital heart defects,hamartomas of tongue,and polysyndactyly 217085
ALDH7A1	Epilepsy,pyridoxine-dependent,266100
TELO2	You-Hoover-Fong syndrome,616954
INTU	?Short-rib throacic dysplasia 20 with polydactyly,?Orofaciodigital syndrome XVII
LMAN2L	Mental retardation,autosomal recessive,52
HEPACAM	Megalencephalic leukoencephalopathy with subcortical cysts 2A,613925
TMTC3	Lissencephaly 8
KLK4	Amelogenesis imperfecta,type IIA1,204700
CYP27B1	Vitamin D-dependent rickets,type I,264700
DGUOK	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type),251880
OTOA	Deafness,autosomal recessive 22,607039
SGPL1	Nephrotic syndrometype 14
DPH1	Developmental delay with short stature,dysmorphic features and sparse hair,616901
INPP5E	Joubert syndrome 1,213300 Mental retardation,truncal obesity,retinal dystrophy,and micropenis,610156
OTOG	Deafness,autosomal recessive 18B,614945
INPP5K	Muscular dystrophy,congenital,with cataracts and intellectual disability
OTOF	Auditory neuropathy,autosomal recessive,1,601071 Deafness,autosomal recessive 9,601071
ASL	Argininosuccinic aciduria,207900
FADD	Infections,recurrent,with encephalopathy,hepatic dysfunction,and cardiovasuclar malformations,613759
CDT1	Meier-Gorlin syndrome 4,613804
NUS1	?Congenital disorder of glycosylationtype 1aa,Mental retardationautosomal dominant 55with seizures
CDSN	Peeling skin syndrome 1,270300
CBLIF	Intrinsic factor deficiency,261000
INSR	Leprechaunism,246200 Rabson-Mendenhall syndrome,262190
TREX1	Aicardi-Goutieres syndrome 1,dominant and recessive,225750 Chilblain lupus,610448 Vasculopathy,retinal,with cerebral leukodystrophy,192315 {Systemic lupus erythematosus,susceptibility to},152700
PDE2A	-
SLC33A1	Congenital cataracts,hearing loss,and neurodegeneration,614482 Spastic paraplegia 42,autosomal dominant,612539
GDF1	Right atrial isomerism (Ivemark),208530
ACADSB	2-methylbutyrylglycinuria,610006
GDF5	Acromesomelic dysplasia,Hunter-Thompson type,201250
CIDEC	?Lipodystrophyfamilial partialtype 5
SPART	Troyer syndrome,275900
CCDC88A	PEHO syndrome,260565

OCLN	Band-like calcification with simplified gyration and polymicrogyria,251290
CDAN1	Dyserythropoietic anemia,congenital,type Ia,224120
STAR	Lipoid adrenal hyperplasia,201710
CCDC88C	Hydrocephalus,nonsyndromic,autosomal recessive,236600
SPRTN	Ruijs-Aalfs syndrome,616200
ATM	Ataxia-telangiectasia,208900
ALDH18A1	Cutis laxa,autosomal dominant 3,616603 Cutis laxa,autosomal recessive,type IIIA,219150 Spastic paraplegia 9A,autosomal dominant,601162 Spastic paraplegia 9B,autosomal recessive,616586
CC2D1A	Mental retardation,autosomal recessive 3,608443
ATR	Seckel syndrome 1,210600
INVS	Nephronophthisis 2,infantile,602088
RYR1	Central core disease,117000 King-Denborough syndrome,145600Minicore myopathy with external ophthalmoplegia,255320Neuromuscular disease,congenital,with uniform type 1 fiber,117000{Malignant hyperthermia susceptibility 1},145600
FASTKD2	Mitochondrial complex IV deficiency 220110
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1,224050
GYS2	Glycogen storage disease 0,liver,240600
GYS1	Glycogen storage disease 0,muscle,611556
HINT1	Neuromyotonia and axonal neuropathy,autosomal recessive,137200
UCHL1	Spastic paraplegia 79autosomal recessive
AUH	3-methylglutaconic aciduria,type I,250950
QARS1	Microcephaly,progressive,seizures,and cerebral and cerebellar atrophy,615760
NNT	Glucocorticoid deficiency 4,614736
DLAT	Pyruvate dehydrogenase E2 deficiency,245348
CC2D2A	COACH syndrome,216360 Joubert syndrome 9,612285 Meckel syndrome 6,612284
RSPRY1	Spondylopeimetaphyseal dsyplasia,Faden-Alkuraya type,616723
MANBA	Mannosidosis,beta,248510
PRMT7	Short stature,brachydactyly,intellectual developmental disability,and seizures
AMPD1	Myopathy due to myoadenylate deaminase deficiency,615511
AMPD2	Pontocerebellar hypoplasia,type 9,615809 ?Spastic paraplegia 63,615686
AP3B1	Hermansky-Pudlak syndrome 2,608233
AP3B2	Epileptic encephalopathy,early infantile,48
IL17RC	Candidiasis,familial,9,616445
IL17RA	Immunodeficiency 51
RAB33B	Smith-McCort dysplasia 2,615222
BBS10	Bardet-Biedl syndrome 10,615987

RAD51C	Fanconi anemia,complementation group O,613390 {Breast-ovarian cancer,familial,susceptibility to,3},613399
BBS12	Bardet-Biedl syndrome 12,615989
TNNT1	Nemaline myopathy 5,Amish type,605355
NME8	Ciliary dyskinesia,primary,6,610852
FAM20C	Raine syndrome,259775
RDH5	Fundus albipunctatus,136880
ALDOB	Fructose intolerance,229600
FAM20A	Amelogenesis imperfecta,type IG (enamel-renal syndrome),204690
MAPT	Supranuclear palsy,progressive atypical,260540
ALDOA	Glycogen storage disease XII,611881
KPTN	Mental retardation,autosomal recessive 41,615637
UMPS	Orotic aciduria,258900
DNASE1L3	Systemic lupus erythematosus 16,614420
RAPSN	Fetal akinesia deformation sequence,208150 Myasthenic syndrome,congenital,11,associated with acetylcholine receptor deficiency,616326
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency,606408
ALOX12B	Ichthyosis,congenital,autosomal recessive 2,242100
TRAK1	Epileptic encephalopathy,early infantile,68 618201
ATP5F1A	?Combined oxidative phosphorylation deficiency 22
KIAA0753	?Orofaciodigital syndrome XV
CTNNA2	Cortical dysplasia,complex,with other brain malformations 9
DCPS	Al-Raqad syndrome,616459
MPZL2	Deafness,autosomal recessive 111
CA12	Hyperchlorhidrosis,isolated,143860
ARNT2	?Webb-Dattani syndrome
AP3D1	Hermansky-Pudlak syndrome 10
STAC3	Native American myopathy,255995
KRT10	Epidermolytic hyperkeratosis,113800 Ichthyosis with confetti,609165 Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602
C19orf12	Neurodegeneration with brain iron accumulation 4,614298 ?Spastic paraplegia 43,autosomal recessive,615043
FAH	Tyrosinemia,type I,276700
ATP5F1D	Mitochondrial complex V (ATP synthase) deficiency 618120
ATP5F1E	Mitochondrial complex V (ATP synthase) deficiency,nuclear type 3 614053
KRT18	Cirrhosis,cryptogenic,215600 {Cirrhosis,noncryptogenic,susceptibility to},215600
KRT14	Epidermolysis bullosa simplex,recessive 1
RAB3GAP2	Martsolf syndrome,212720 Warburg micro syndrome 2,614225
TRIP11	Achondrogenesis,type IA,200600
B9D1	Joubert syndrome 27
B9D2	Meckel syndrome 10,614175
TRIP13	Mosaic variegated aneuploidy syndrome 3
RAB3GAP1	Warburg micro syndrome 1,600118
NEUROG3	Diarrhea 4,malabsorptive,congenital,610370

TJP2	Cholestasis,progressive familial intrahepatic 4,615878
WASHC4	Mental retardation,autosomal recessive 43
WASHC5	Ritscher-Schinzel syndrome 1
CIB2	Deafness,autosomal recessive 48,609439 Usher syndrome,type IJ,614869
SDR9C7	Ichthyosiscongenitalautosomal recessive 13
ESPN	Deafness,autosomal recessive 36,609006 Deafness,neurosensory,without vestibular involvement,autosomal dominant
PSPH	Phosphoserine phosphatase deficiency,614023
ABCG8	Sitosterolemia,210250 {Gallbladder disease 4},611465
GHSR	Growth hormone deficiency,isolated partial,615925
ABCG5	Sitosterolemia,210250
CFAP418	Cone-rod dystrophy 16,614500 Retinitis pigmentosa 64,614500
PAM16	Spondylometaphyseal dysplasia,Megarbane-Dagher-Melike type,613320
TRNT1	Retinitis pigmentosa and erythrocytic microcytosis,616959 Sideroblastic anemia with B-cell immunodeficiency,periodic fevers,and developmental delay,616084
CFAP410	Spondylometaphyseal dysplasiaaxial,Retinal dystrophy with macular staphyloma
PDIA6	polycystic kidney disease, infancy-onset diabetes, and microcephaly
PRSS56	Microphthalmia,isolated 6,613517
KCTD7	Epilepsy,progressive myoclonic 3,with or without intracellular inclusions,611726
ALDH3A2	Sjogren-Larsson syndrome,270200
DPM1	Congenital disorder of glycosylation,type Ie,608799
SGO1	Chronic atrial and intestinal dysrhythmia,616201
DPM2	Congenital disorder of glycosylation,type Iu,615042
SELENON	Muscular dystrophy,rigid spine,1,602771 Myopathy,congenital,with fiber-type disproportion,255310
DPM3	Congenital disorder of glycosylation,type Io,612937
PREPL	Myasthenic syndromecongenital22
TMEM138	Joubert syndrome 16,614465
COL4A4	Alport syndrome,autosomal recessive,203780
COL4A3	Alport syndrome,autosomal recessive,203780
EOGT	Adams-Oliver syndrome 4,615297
ELMO2	Vascular malformation,primary intraosseous
AIPL1	Cone-rod dystrophy,604393 Leber congenital amaurosis 4,604393 Retinitis pigmentosa,juvenile,604393
ISCU	Myopathy with lactic acidosis,hereditary,255125
ATP6V1A	Cutis laxaautosomal recessivetype IID
H6PD	Cortisone reductase deficiency 1,604931
USP53	-
HPCA	Dystonia 2,torsion,autosomal recessive,224500
LTBP4	Cutis laxa,autosomal recessive,type IC,613177
CSF2RB	Surfactant metabolism dysfunction,pulmonary,5,614370
LTBP2	Weill-Marchesani syndrome 3,recessive,614819

LTBP3	Dental anomalies and short stature,601216
STK4	T-cell immunodeficiency,recurrent infections,autoimmunity,and cardiac malformations,614868
CPLX1	Epileptic encephalopathyearly infantile63
LTBP1	Cutis laxa, autosomal recessive, type IIE
NALCN	Congenital contractures of the limbs and face,hypotonia,and developmental delay,616266 Hypotonia,infantile,with psychomotor retardation and characteristic facies 1,615419
DNAJC21	Bone marrow failure syndrome 3,617052
MYH11	-
RIPK1	Immunodeficiency 57
ZC3H14	Mental retardation,autosomal recessive 56
PDZD7	Deafness,autosomal recessive 57 ,618003,Usher syndrome,type IIC,GPR98/PDZD7 digenic,605472 {Retinal disease in Usher syndrome type IIA,modifier of},276901
DNAJC19	3-methylglutaconic aciduria,type V,610198
HSPA9	Even-plus syndrome,616854
STIL	Microcephaly 7,primary,autosomal recessive,612703
TGFB1	Inflammatory bowel disease,immunodeficiency,and encephalopathy
EPHX1	Diphenylhydantoin toxicity Hypercholanemia,familial,607748 ?Fetal hydantoin syndrome {Preeclampsia,susceptibility to},189800
TBCD	Encephalopathy,progressive,early-onset,with brain atrophy and thin corpus callosum
DNAJC12	Hyperphenylalaninemia,mild,non-BH4-deficient
IFT122	Cranioectodermal dysplasia 1,218330
TDRD7	Cataract 36,613887
ACSF3	Combined malonic and methylmalonic aciduria,614265
ESR1	Estrogen resistance,615363
SLC2A9	Hypouricemia,renal,2,612076 {Uric acid concentration,serum,QTL 2},612076
TBCK	Hypotonia,infantile,with psychomotor retardation and characteristic facies 3,616900
DNMBP	Cataract 48
RFXAP	Bare lymphocyte syndrome,type II,complementation group D,209920
TBCE	Hypoparathyroidism-retardation-dysmorphism syndrome,241410 Kenny-Caffey syndrome,type 1,244460
MAN2B1	Mannosidosis,alpha-,types I and II,248500
ABCD4	Methylmalonic aciduria and homocystinuria,cblJ type,614857
DOCK6	Adams-Oliver syndrome 2,614219
ACADVL	VLCAD deficiency,201475
B4GALT1	Congenital disorder of glycosylation,type IId,607091
DOCK3	Neurodevelopmental disorder with impaired intellectual development,hypotonia,and ataxia
TSEN15	Pontocerebellar hypoplasia,type 2F,617026
DOCK8	Hyper-IgE recurrent infection syndrome,autosomal recessive,243700
DOCK7	Epileptic encephalopathy,early infantile,23,615859

SLC2A1	GLUT1 deficiency syndrome 1,infantile onset,severe,606777
SLC2A2	Fanconi-Bickel syndrome,227810 {Diabetes mellitus,noninsulin-dependent},125853
LMOD3	Nemaline myopathy 10,616165
DHTKD1	2-aminoadipic 2-oxoadipic aciduria,204750
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A),252900
GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,14,615350
CA5A	Hyperammonemia due to carbonic anhydrase VA deficiency,615751
CYP4V2	Bietti crystalline corneoretinal dystrophy,210370
KYNU	Vertebralcardiacrenaland limb defects syndrome 2,?Hydroxykynureninuria
GMPPA	Alacrima,achalasia,and mental retardation syndrome,615510
TMEM107	?Joubert syndrome 29,Meckel syndrome 13,Orofaciodigital syndrome XVI
SPEG	Centronuclear myopathy 5,615959
GUSB	Mucopolysaccharidosis VII,253220
POLE	IMAGE-I syndrome ,FILS syndrome
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency,250620
POLH	Xeroderma pigmentosum,variant type,278750
TSHB	Hypothryoidism,congenital,nongoitrous 4,275100
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type),203700
SCYL1	Spinocerebellar ataxia,autosomal recessive 21,616719
FGB	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004 Hypofibrinogenemia,congenital,202400
FGA	Afibrinogenemia,congenital,202400
RUBCN	Spinocerebellar ataxia,autosomal recessive 15
TSFM	Combined oxidative phosphorylation deficiency 3,610505
JAGN1	Neutropenia,severe congenital,6,autosomal recessive,616022
ABCC2	Dubin-Johnson syndrome,237500
ABCC8	Hyperinsulinemic hypoglycemia,familial,1,256450
MARS2	Spastic ataxia 3,autosomal recessive,611390 ?Combined oxidative phosphorylation deficiency 25,616430
ABCC6	Arterial calcification,generalized,of infancy,2,614473 Pseudoxanthoma elasticum,264800
FGG	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004 Hypodysfibrinogenemia,616004 Hypofibrinogenemia,congenital,202400
MARS1	Interstitial lung and liver disease,615486
TWNK	Perrault syndrome 5
USP45	?Leber congenital amaurosis 19
CTPS1	Immunodeficiency 24,615897
HPDL	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities -
SAG	Oguchi disease-1,258100Retinitis pigmentosa 47,613758

DOCK2	Immunodeficiency 40,616433
B4GALT7	Ehlers-Danlos syndrome with short stature and limb anomalies,130070
CILK1	Endocrine-cerebroosteodysplasia,612651
FAN1	Interstitial nephritiskaryomegalic
MCPH1	Microcephaly 1,primary,autosomal recessive,251200
TSEN34	Pontocerebellar hypoplasia type 2C,612390
HPGD	Cranioosteoarthropathy,259100 Digital clubbing,isolated congenital,119900 Hypertrophic osteoarthropathy,primary,autosomal recessive 1,259100
NXN	Robinow syndrome,autosomal recessive 2
CAMK2A	?Mental retardation,autosomal recessive 63
POMP	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma,601952
LCAT	Fish-eye disease,136120 Norum disease,245900
SPG7	Spastic paraplegia 7,autosomal recessive,607259
LTC4S	Leukotriene C4 synthase deficiency 614037
KRT85	Ectodermal dysplasia 4,hair/nail type,602032
USP18	Pseudo-TORCH syndrome 2
DLL3	Spondylocostal dysostosis 1,autosomal recessive,277300
HSPD1	Leukodystrophy,hypomyelinating,4,612233 Spastic paraplegia 13,autosomal dominant,605280
ACAN	Spondyloepimetaphyseal dysplasia,aggrecan type
ERBB3	Lethal congenital contractural syndrome 2,607598
PDE6D	?Joubert syndrome 22
BPNT2	Chondrodysplasia with joint dislocations,GPAPP type,614078
PDE6C	Cone dystrophy 4,613093
PDE6B	Retinitis pigmentosa-40,613801
LRIG2	Urofacial syndrome 2,615112
PDE6A	Retinitis pigmentosa 43,613810
S1PR2	Deafness,autosomal recessive 68,610419
SASH1	?Cancer,alopecia,pigment dyscrasia,onychodystrophy,and keratoderma
MUTYH	Adenomas,multiple colorectal,608456 Colorectal adenomatous polyposis,autosomal recessive,with pilomatricomas,132600 Gastric cancer,somatic,613659
REPS1	?Neurodegeneration with brain iron accumulation 7
STN1	Cerebroretinal microangiopathy with calcifications and cysts 2
PDE6H	Achromatopsia 6,610024 Retinal cone dystrophy 3,610024
B3GAT3	Multiple joint dislocationsshort staturecraniofacial dysmorphismwith or without congenital heart defects
LGI4	Arthrogryposis multiplex congenitaneurogenicwith myelin defect
PDE6G	Retinitis pigmentosa 57,613582
PCDHGC4	-
PCARE	Retinitis pigmentosa 54,613428
DTNBP1	Hermansky-Pudlak syndrome 7,614076
LCA5	Leber congenital amaurosis 5,604537
TSPEAR	Deafness,autosomal recessive 98,614861

GDAP1	Charcot-Marie-Tooth disease,axonal,type 2K,607831 Charcot-Marie-Tooth disease,axonal,with vocal cord paresis,607706 Charcot-Marie-Tooth disease,recessive intermediate,A,608340Charcot-Marie-Tooth disease,type 4A,214400
GDAP2	Spinocerebellar ataxia,autosomal recessive 27
TSHR	Hyperthyroidism,familial gestational,603373 Hyperthyroidism,nonautoimmune,609152 Hypothyroidism,congenital,nongoitrous,1 275200 Thyroid adenoma,hyperfunctioning,somatic Thyroid carcinoma with thyrotoxicosis
POMC	Obesity,adrenal insufficiency,and red hair due to POMC deficiency,609734 {Obesity,early-onset,susceptibility to},601665
XPNPEP3	Nephronophthisis-like nephropathy 1,613159
POMK	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies),type A,12,615249 ?Muscular dystrophy-dystroglycanopathy (limb-girdle),type C,12,616094

Spastic paraplegia 83, autosomal recessive