Gene	Disease
AAAS	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME; AAAS [AR]
AARS2	LEUKOENCEPHALOPATHY. PROGRESSIVE. WITH OVARIAN FAILURE; LKENP [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 8; COXPD8 [AR]
AASS	HYPERLYSINEMIA. TYPE I [AR]
ABAT	GABA-TRANSAMINASE DEFICIENCY [AR]
ABCA12	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 4B; ARCI4B [AR] | ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 4A; ARCI4A [AR]
ABCA3	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 3; SMDP3 [AR]
ABCA4	MACULAR DEGENERATION. AGE-RELATED. 2; ARMD2 [AD] | STARGARDT DISEASE 1; STGD1 [AR] | RETINITIS PIGMENTOSA 19; RP19 [AR]
ABCB11	CHOLESTASIS. BENIGN RECURRENT INTRAHEPATIC. 2; BRIC2 [AR] | CHOLESTASIS. PROGRESSIVE FAMILIAL INTRAHEPATIC. 2; PFIC2 [AR]
ABCB4	GALLBLADDER DISEASE 1; GBD1 [AD] | CHOLESTASIS. INTRAHEPATIC. OF PREGNANCY 3; ICP3 [AD] | CHOLESTASIS. PROGRESSIVE FAMILIAL INTRAHEPATIC. 3; PFIC3 [AR]
ABCB7	ANEMIA. SIDEROBLASTIC. AND SPINOCEREBELLAR ATAXIA; ASAT [XLR]
ABCC2	DUBIN-JOHNSON SYNDROME; DJS [AR]
ABCC6	ARTERIAL CALCIFICATION. GENERALIZED. OF INFANCY. 2; GACI2 [AR] | PSEUDOXANTHOMA ELASTICUM. FORME FRUSTE [AD] | PSEUDOXANTHOMA ELASTICUM; PXE [AR]
ABCC8	DIABETES MELLITUS. PERMANENT NEONATAL; PNDM [AD] | HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 1; HHF1 [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | HYPOGLYCEMIA. LEUCINE-INDUCED; LIH [AD]
ABCD1	ADRENOLEUKODYSTROPHY; ALD [XLR]
ABCG5	Sitosterolemia 2 618666 (3)
ABCG8	SITOSTEROLEMIA 1; STSL1 [AR]
ABHD12	POLYNEUROPATHY. HEARING LOSS. ATAXIA. RETINITIS PIGMENTOSA. AND CATARACT; PHARC [AR]
ABHD5	CHANARIN-DORFMAN SYNDROME; CDS [AR]
ACAD8	ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY; IBDD [AR]
ACAD9	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 20; MC1DN20 [AR]
ACADM	ACYL-CoA DEHYDROGENASE. MEDIUM-CHAIN. DEFICIENCY OF; ACADMD [AR]
ACADS	ACYL-CoA DEHYDROGENASE. SHORT-CHAIN. DEFICIENCY OF; ACADSD [AR]
ACADSB	2-METHYLBUTYRYL-CoA DEHYDROGENASE DEFICIENCY [AR]
ACADVL	ACYL-CoA DEHYDROGENASE. VERY LONG-CHAIN. DEFICIENCY OF; ACADVLD [AR]
ACAN	SPONDYLOEPIMETAPHYSEAL DYSPLASIA. AGGRECAN TYPE; SEMDAG [AR] | SPONDYLOEPIPHYSEAL DYSPLASIA. KIMBERLEY TYPE; SEDK [AD] | SHORT STATURE AND ADVANCED BONE AGE. WITH OR WITHOUT EARLY-ONSET OSTEOARTHRITIS AND/OR OSTEOCHONDRITIS DISSECANS; SSOAOD [AD]
ACAT1	ALPHA-METHYLACETOACETIC ACIDURIA [AR]
ACE	RENAL TUBULAR DYSGENESIS; RTD [AR]
ACO2	INFANTILE CEREBELLAR-RETINAL DEGENERATION; ICRD [AR] | OPTIC ATROPHY 9; OPA9 [AR]
ACOX1	PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY [AR]
ACP5	SPONDYLOENCHONDRODYSPLASIA WITH IMMUNE DYSREGULATION; SPENCDI [AR]
ACTA1	NEMALINE MYOPATHY 3; NEM3 [AD] | MYOPATHY. SCAPULOHUMEROPERONEAL; SHPM [AD] | MYOPATHY. CONGENITAL. WITH FIBER-TYPE DISPROPORTION; CFTD [AD]
ACY1	AMINOACYLASE 1 DEFICIENCY; ACY1D [AR]
ADA	SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-NEGATIVE. NK CELL-NEGATIVE. DUE TO ADENOSINE DEAMINASE DEFICIENCY [AR]
ADAM9	CONE-ROD DYSTROPHY 9; CORD9 [AR]
ADAMTS10	WEILL-MARCHESANI SYNDROME 1; WMS1 [AR]
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA. HEREDITARY; TTP [AR]
ADAMTS17	WEILL-MARCHESANI SYNDROME 4; WMS4 [AR]
ADAMTS18	MICROCORNEA. MYOPIC CHORIORETINAL ATROPHY. AND TELECANTHUS; MMCAT [AR]

ADAMTS2	EHLERS-DANLOS SYNDROME. DERMATOSPARAXIS TYPE; EDSDERMS [AR]
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1; GPHYSD1 [AR]
ADAMTSL4	ECTOPIA LENTIS 2. ISOLATED. AUTOSOMAL RECESSIVE; ECTOL2 [AR] | ECTOPIA LENTIS ET PUPILLAE [AR]
ADAT3	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 36; MRT36 [AR]
ADK	HYPERMETHIONINEMIA DUE TO ADENOSINE KINASE DEFICIENCY [AR]
ADSL	ADENYLOSUCCINASE DEFICIENCY; ADSLD [AR]
AFF2	MENTAL RETARDATION. X-LINKED. ASSOCIATED WITH FRAGILE SITE FRAXE [XLR]
AFG3L2	SPINOCEREBELLAR ATAXIA 28; SCA28 [AD] | SPASTIC ATAXIA 5. AUTOSOMAL RECESSIVE; SPAX5 [AR]
AGA	ASPARTYLGLUCOSAMINURIA; AGU [AR]
AGK	CATARACT 38; CTRCT38 [AR] | SENGERS SYNDROME [AR]
AGL	GLYCOGEN STORAGE DISEASE III; GSD3 [AR]
AGPAT2	LIPODYSTROPHY. CONGENITAL GENERALIZED. TYPE 1; CGL1 [AR]
AGPS	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA. TYPE 3; RCDP3 [AR]
AGRN	MYASTHENIC SYNDROME. CONGENITAL. 8; CMS8 [AR]
AGT	RENAL TUBULAR DYSGENESIS; RTD [AR] | HYPERTENSION. ESSENTIAL [Multifactorial]
AGTR1	RENAL TUBULAR DYSGENESIS; RTD [AR] | HYPERTENSION. ESSENTIAL [Multifactorial]
AGXT	HYPEROXALURIA. PRIMARY. TYPE I; HP1 [AR]
AHCY	HYPERMETHIONINEMIA WITH S-ADENOSYLHOMOCYSTEINE HYDROLASE DEFICIENCY [AR]
AHI1	JOUBERT SYNDROME 3; JBTS3 [AR]
AICDA	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 2; HIGM2 [AR]
AIFM1	CHARCOT-MARIE-TOOTH DISEASE. X-LINKED RECESSIVE. 4. WITH OR WITHOUT CEREBELLAR ATAXIA; CMTX4 [XLR] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. X-LINKED. WITH HYPOMYELINATING LEUKODYSTROPHY; SEMDHL [XLR] | DEAFNESS. X-LINKED 5. WITH PERIPHERAL NEUROPATHY; DFNX5 [XLR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 6; COXPD6 [XLR]
AIMP1	LEUKODYSTROPHY. HYPOMYELINATING. 3; HLD3 [AR]
AIPL1	LEBER CONGENITAL AMAUROSIS 4; LCA4 [AR]
AIRE	AUTOIMMUNE POLYENDOCRINE SYNDROME. TYPE I. WITH OR WITHOUT REVERSIBLE METAPHYSEAL DYSPLASIA; APS1 [AR]
AK1	ADENYLATE KINASE DEFICIENCY. HEMOLYTIC ANEMIA DUE TO [AR]
AK2	RETICULAR DYSGENESIS [AR]
AKR1C2	46.XY SEX REVERSAL 8; SRXY8 [AR]
AKR1D1	BILE ACID SYNTHESIS DEFECT. CONGENITAL. 2; CBAS2 [AR]
ALAD	PORPHYRIA. ACUTE HEPATIC [AR]
ALAS2	PROTOPORPHYRIA. ERYTHROPOIETIC. X-LINKED; XLEPP [X-linked] | ANEMIA. SIDEROBLASTIC. 1; SIDBA1 [XLR]
ALDH18A1	SPASTIC PARAPLEGIA 9A. AUTOSOMAL DOMINANT; SPG9A [AD] | CUTIS LAXA. AUTOSOMAL DOMINANT 3; ADCL3 [AD] | CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IIIA; ARCL3A [AR] | SPASTIC PARAPLEGIA 9B. AUTOSOMAL RECESSIVE; SPG9B [AR]
ALDH1A3	MICROPHTHALMIA. ISOLATED 8; MCOP8 [AR]
ALDH3A2	SJOGREN-LARSSON SYNDROME; SLS [AR]
ALDH4A1	HYPERPROLINEMIA. TYPE II; HYRPRO2 [AR]
ALDH5A1	SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY; SSADHD [AR]
ALDH6A1	METHYLMALONATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY; MMSDHD [AR]
ALDH7A1	EPILEPSY. PYRIDOXINE-DEPENDENT; EPD [AR]
ALDOA	GLYCOGEN STORAGE DISEASE XII; GSD12 [AR]
ALDOB	FRUCTOSE INTOLERANCE. HEREDITARY [AR]
ALG1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ik; CDG1K [AR]
ALG11	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ip; CDG1P [AR]

ALG3	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Id; CDG1D [AR]
ALG6	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ic; CDG1C [AR]
ALG9	GILLESSEN-KAESBACH-NISHIMURA SYNDROME; GIKANIS [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Il; CDG1L [AR]
ALMS1	ALSTROM SYNDROME; ALMS [AR]
ALOX12B	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 2; ARCI2 [AR]
ALOXE3	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 3; ARCI3 [AR]
ALPL	HYPOPHOSPHATASIA. CHILDHOOD [AR] | HYPOPHOSPHATASIA. INFANTILE [AR] | HYPOPHOSPHATASIA. ADULT [AD]
ALS2	SPASTIC PARALYSIS. INFANTILE-ONSET ASCENDING; IAHSP [AR] | AMYOTROPHIC LATERAL SCLEROSIS 2. JUVENILE; ALS2 [AR] | PRIMARY LATERAL SCLEROSIS. JUVENILE; PLSJ [AR]
ALX3	FRONTONASAL DYSPLASIA 1; FND1 [AR]
AMACR	ALPHA-METHYLACYL-CoA RACEMASE DEFICIENCY; AMACRD [AR] | BILE ACID SYNTHESIS DEFECT. CONGENITAL. 4; CBAS4 [AR]
AMH	PERSISTENT MULLERIAN DUCT SYNDROME. TYPES I AND II; PMDS [Autosomal recessive with male sex limitatio]
AMHR2	PERSISTENT MULLERIAN DUCT SYNDROME. TYPES I AND II; PMDS [Autosomal recessive with male sex limitatio]
AMN	MEGALOBLASTIC ANEMIA 1 [AR]
AMPD1	MYOPATHY DUE TO MYOADENYLATE DEAMINASE DEFICIENCY; MMDD [AR]
AMT	GLYCINE ENCEPHALOPATHY; GCE [AR]
ANGPTL3	HYPOBETALIPOPROTEINEMIA. FAMILIAL. 2; FHBL2 [AR]
ANK1	SPHEROCYTOSIS. TYPE 1; SPH1 [AD]
ANO10	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 10; SCAR10 [AR]
ANO5	GNATHODIAPHYSEAL DYSPLASIA; GDD [AD] | MIYOSHI MUSCULAR DYSTROPHY 3; MMD3 [AR] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 12; LGMDR12 [AR]
ANO6	SCOTT SYNDROME; SCTS [AR]
ANTXR2	HYALINE FIBROMATOSIS SYNDROME; HFS [AR]
AP1S2	PETTIGREW SYNDROME; PGS [XLR]
AP3B1	HERMANSKY-PUDLAK SYNDROME 2; HPS2 [AR]
AP4B1	SPASTIC PARAPLEGIA 47. AUTOSOMAL RECESSIVE; SPG47 [AR]
AP4E1	STUTTERING. FAMILIAL PERSISTENT. 1; STUT1 [AD] | SPASTIC PARAPLEGIA 51. AUTOSOMAL RECESSIVE; SPG51 [AR]
AP4M1	SPASTIC PARAPLEGIA 50. AUTOSOMAL RECESSIVE; SPG50 [AR]
AP4S1	SPASTIC PARAPLEGIA 52. AUTOSOMAL RECESSIVE; SPG52 [AR]
AP5Z1	SPASTIC PARAPLEGIA 48. AUTOSOMAL RECESSIVE; SPG48 [AR]
APOA2	HYPERCHOLESTEROLEMIA. FAMILIAL. 1; FHCL1 [AD]
APOC2	APOLIPOPROTEIN C-II DEFICIENCY [AR]
APOE	ALZHEIMER DISEASE 2; AD2 [Autosomal dominant allele (19q) with additional multifactorial component in late-onset case] | ALZHEIMER DISEASE 3; AD [AD] | SEA-BLUE HISTIOCYTE DISEASE [AR] | MACULAR DEGENERATION. AGE-RELATED. 1; ARMD1 [AD]
APP	CEREBRAL AMYLOID ANGIOPATHY. APP-RELATED [AD] | ALZHEIMER DISEASE; AD [AD]
APRT	ADENINE PHOSPHORIBOSYLTRANSFERASE DEFICIENCY; APRTD [AR]
APTX	ATAXIA. EARLY-ONSET. WITH OCULOMOTOR APRAXIA AND HYPOALBUMINEMIA; EAOH [AR]
AQP2	DIABETES INSIPIDUS. NEPHROGENIC. AUTOSOMAL [AD]
AR	PROSTATE CANCER [Somatic mutation]
ARFGEF2	PERIVENTRICULAR HETEROTOPIA WITH MICROCEPHALY. AUTOSOMAL RECESSIVE; ARPHM [AR]
ARG1	ARGININEMIA [AR]
ARHGEF9	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 8; EIEE8 [XLR]
ARL2BP	RETINITIS PIGMENTOSA 82 WITH OR WITHOUT SITUS INVERSUS; RP82 [AR]
ARL6	BARDET-BIEDL SYNDROME 1; BBS1 [AR] | BARDET-BIEDL SYNDROME 3; BBS3 [AR]
ARMC4	CILIARY DYSKINESIA. PRIMARY. 23; CILD23 [AR]

ARSA	METACHROMATIC LEUKODYSTROPHY; MLD [AR]
ARSB	MUCOPOLYSACCHARIDOSIS. TYPE VI; MPS6 [AR]
ARX	PARTINGTON X-LINKED MENTAL RETARDATION SYNDROME; PRTS [XLR] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 1; EIEE1 [XLR] | MENTAL RETARDATION. X-LINKED. WITH OR WITHOUT SEIZURES. ARX-RELATED; MRXARX [XLR] | CORPUS CALLOSUM. AGENESIS OF. WITH ABNORMAL GENITALIA [X-linked] | LISSENCEPHALY. X-LINKED. 2; LISX2 [X-linked]
ASAH1	SPINAL MUSCULAR ATROPHY WITH PROGRESSIVE MYOCLONIC EPILEPSY; SMAPME [AR] | FARBER LIPOGRANULOMATOSIS; FRBRL [AR]
ASL	ARGININOSUCCINIC ACIDURIA [AR]
ASPA	CANAVAN DISEASE [AR]
ASPM	MICROCEPHALY 5. PRIMARY. AUTOSOMAL RECESSIVE; MCPH5 [AR]
ASS1	CITRULLINEMIA. CLASSIC [AR]
ATCAY	CEREBELLAR ATAXIA. CAYMAN TYPE; ATCAY [AR]
ATIC	AICAR TRANSFORMYLASE/IMP CYCLOHYDROLASE DEFICIENCY [AR]
ATM	BREAST CANCER [AD] | ATAXIA-TELANGIECTASIA; AT [AR]
ATP13A2	SPASTIC PARAPLEGIA 78. AUTOSOMAL RECESSIVE; SPG78 [AR] | KUFOR-RAKEB SYNDROME; KRS [AR]
ATP2A1	BRODY MYOPATHY [AR]
ATP6AP2	PARKINSONISM WITH SPASTICITY. X-LINKED; XPDS [XLR] | MENTAL RETARDATION. X-LINKED. SYNDROMIC. HEDERA TYPE; MRXSH [XLR]
ATP6V0A2	WRINKLY SKIN SYNDROME; WSS [AR] | CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IIA; ARCL2A [AR]
ATP6V0A4	Renal tubular acidosis distal autosomal recessive 602722 (3)
ATP6V1B1	RENAL TUBULAR ACIDOSIS. DISTAL. WITH PROGRESSIVE NERVE DEAFNESS [AR]
ATP7A	MENKES DISEASE; MNK [XLR] | OCCIPITAL HORN SYNDROME; OHS [XLR] | SPINAL MUSCULAR ATROPHY. DISTAL. X-LINKED 3; SMAX3 [XLR]
ATP7B	WILSON DISEASE; WND [AR]
ATP8B1	CHOLESTASIS. INTRAHEPATIC. OF PREGNANCY. 1; ICP1 [AD] | CHOLESTASIS. BENIGN RECURRENT INTRAHEPATIC. 1; BRIC1 [AR] | CHOLESTASIS. PROGRESSIVE FAMILIAL INTRAHEPATIC. 1; PFIC1 [AR]
ATR	SECKEL SYNDROME 1; SCKL1 [AR] | CUTANEOUS TELANGIECTASIA AND CANCER SYNDROME. FAMILIAL; FCTCS [AD]
ATRX	ALPHA-THALASSEMIA/MENTAL RETARDATION SYNDROME. X-LINKED; ATRX [XLD] | MENTAL RETARDATION-HYPOTONIC FACIES SYNDROME. X-LINKED. 1; MRXHF1 [XLR]
AUH	3-METHYLGLUTACONIC ACIDURIA. TYPE I; MGCA1 [AR]
AURKC	SPERMATOGENIC FAILURE 5; SPGF5 [AR]
AVP	DIABETES INSIPIDUS. NEUROHYPOPHYSEAL [AD]
AVPR2	DIABETES INSIPIDUS. NEPHROGENIC. X-LINKED [XLR] | NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS; NSIAD [XLR]
B2M	IMMUNODEFICIENCY 43; IMD43 [AR] | AMYLOIDOSIS. FAMILIAL VISCERAL [AD]
B3GAT3	MULTIPLE JOINT DISLOCATIONS. SHORT STATURE. AND CRANIOFACIAL DYSMORPHISM WITH OR WITHOUT CONGENITAL HEART DEFECTS; JDSCD [AR]
B4GALNT1	SPASTIC PARAPLEGIA 26. AUTOSOMAL RECESSIVE; SPG26 [AR]
B4GALT1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IId; CDG2D [AR]
B4GALT7	EHLERS-DANLOS SYNDROME. SPONDYLODYSPLASTIC TYPE. 1; EDSSPD1 [AR]
B9D2	MECKEL SYNDROME. TYPE 10; MKS10 [AR]
BAAT	HYPERCHOLANEMIA. FAMILIAL; FHCA [AR]
BANF1	NESTOR-GUILLERMO PROGERIA SYNDROME; NGPS [AR]
BBS1	BARDET-BIEDL SYNDROME 1; BBS1 [AR]
BBS10	BARDET-BIEDL SYNDROME 10; BBS10 [AR]
BBS12	BARDET-BIEDL SYNDROME 12; BBS12 [AR]
BBS2	BARDET-BIEDL SYNDROME 2; BBS2 [AR] | RETINITIS PIGMENTOSA 74; RP74 [AR]
BBS4	BARDET-BIEDL SYNDROME 4; BBS4 [AR]
BBS5	BARDET-BIEDL SYNDROME 5; BBS5 [AR]

BBS7	BARDET-BIEDL SYNDROME 7; BBS7 [AR]
BBS9	BARDET-BIEDL SYNDROME 9; BBS9 [AR]
BCHE	BUTYRYLCHOLINESTERASE; BCHE [AR]
BCKDHA	MAPLE SYRUP URINE DISEASE; MSUD [AR]
BCKDHB	MAPLE SYRUP URINE DISEASE; MSUD [AR]
BCS1L	GRACILE SYNDROME [AR] | LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 1; MC3DN1 [AR] | BJORNSTAD SYNDROME; BJS [AR]
BEST1	MACULAR DYSTROPHY. VITELLIFORM. 2; VMD2 [AD] | VITREORETINOCHOROIDOPATHY; VRCP [AD]
BFSP1	CATARACT 33. MULTIPLE TYPES; CTRCT33 [AR]
BIN1	MYOPATHY. CENTRONUCLEAR. 2; CNM2 [AR]
BLM	BLOOM SYNDROME; BLM [AR]
BLVRA	HYPERBILIVERDINEMIA; HBLVD [AD]
BMP1	OSTEOGENESIS IMPERFECTA. TYPE XIII; OI13 [AR]
BMP2	SHORT STATURE. FACIAL DYSMORPHISM. AND SKELETAL ANOMALIES WITH OR WITHOUT CARDIAC ANOMALIES; SSFSC [AD] | BRACHYDACTYLY. TYPE A2; BDA2 [AD] | HEMOCHROMATOSIS. TYPE 1; HFE1 [AR]
BMPER	DIAPHANOSPONDYLODYSOSTOSIS [AR]
BMPR1B	BRACHYDACTYLY. TYPE A1. D; BDA1D [AD] | BRACHYDACTYLY. TYPE A2; BDA2 [AD] | ACROMESOMELIC DYSPLASIA. DEMIRHAN TYPE; AMDD [AR]
BMPR2	PULMONARY HYPERTENSION. PRIMARY. 1; PPH1 [AD] | PULMONARY VENOOCCLUSIVE DISEASE 1. AUTOSOMAL DOMINANT; PVOD1 [AD]
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2 WITH HYPERGLYCINEMIA; MMDS2 [AR]
BPGM	ERYTHROCYTOSIS. FAMILIAL. 8; ECYT8 [AR]
BRAT1	RIGIDITY AND MULTIFOCAL SEIZURE SYNDROME. LETHAL NEONATAL; RMFSL [AR] | NEURODEVELOPMENTAL DISORDER WITH CEREBELLAR ATROPHY AND WITH OR WITHOUT SEIZURES; NEDCAS [AR]
BRCA2	WILMS TUMOR 1; WT1 [AD] | FANCONI ANEMIA. COMPLEMENTATION GROUP D1; FANCD1 [AR] | MEDULLOBLASTOMA; MDB [Somatic mutation] | GLIOMA SUSCEPTIBILITY 3; GLM3 [AR] | BREAST-OVARIAN CANCER. FAMILIAL. SUSCEPTIBILITY TO. 2; BROVCA2 [AD] | BREAST CANCER [AD] | PROSTATE CANCER [Somatic mutation]
BRWD3	MENTAL RETARDATION. X-LINKED 93; MRX93 [XLR]
BSCL2	SPASTIC PARAPLEGIA 17. AUTOSOMAL DOMINANT; SPG17 [AD] | ENCEPHALOPATHY. PROGRESSIVE. WITH OR WITHOUT LIPODYSTROPHY; PELD [AR] | LIPODYSTROPHY. CONGENITAL GENERALIZED. TYPE 2; CGL2 [AR] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VA; HMN5A [AD]
BSND	BARTTER SYNDROME. TYPE 4A. NEONATAL. WITH SENSORINEURAL DEAFNESS; BARTS4A [AR]
BTD	BIOTINIDASE DEFICIENCY [AR]
BTK	ISOLATED GROWTH HORMONE DEFICIENCY. TYPE III. WITH AGAMMAGLOBULINEMIA; IGHD3 [XLR] | AGAMMAGLOBULINEMIA. X-LINKED; XLA [XLR]
BUB1B	PREMATURE CHROMATID SEPARATION TRAIT; PCS [AD] | COLORECTAL CANCER; CRC [Somatic mutation] | MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 1; MVA1 [AR]
C12orf57	TEMTAMY SYNDROME; TEMTYS [AR]
C12orf65	SPASTIC PARAPLEGIA 55. AUTOSOMAL RECESSIVE; SPG55 [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 7; COXPD7 [AR]
C19orf12	SPASTIC PARAPLEGIA 43. AUTOSOMAL RECESSIVE; SPG43 [AR] | NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 4; NBIA4 [AR]
C1QA	C1q DEFICIENCY; C1QD [AR]
C1QB	C1q DEFICIENCY; C1QD [AR]
C1QC	C1q DEFICIENCY; C1QD [AR]
C2	C2 deficiency 217000 (3) | {Macular degeneration age-related 14 reduced risk of} 615489 (3)
C3	C3 deficiency 613779 (3) | {Hemolytic uremic syndrome atypical susceptibility to 5} 612925 (3) | {Macular degeneration age-related 9} 611378 (3)
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY; C4AD [AR]
C4B	C4B deficiency 614379 (3)
C6	C6 deficiency 612446 (3) | Combined C6/C7 deficiency (3)

C7	C7 deficiency 610102 (3)
C8A	COMPLEMENT COMPONENT 8 DEFICIENCY. TYPE I; C8D1 [AR]
C8B	COMPLEMENT COMPONENT 8 DEFICIENCY. TYPE II; C8D2 [AR]
C8orf37	BARDET-BIEDL SYNDROME 21; BBS21 [AR] | CONE-ROD DYSTROPHY 16; CORD16 [AR]
CA12	HYPERCHLORHIDROSIS. ISOLATED; HYCHL [AR]
CA2	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 3; OPTB3 [AR]
CA8	CEREBELLAR ATAXIA. MENTAL RETARDATION. AND DYSEQUILIBRIUM SYNDROME 3; CAMRQ3 [AR]
CABP2	DEAFNESS. AUTOSOMAL RECESSIVE 93; DFNB93 [AR]
CABP4	CONE-ROD SYNAPTIC DISORDER. CONGENITAL NONPROGRESSIVE; CRSD [AR]
CACNA2D4	RETINAL CONE DYSTROPHY 4; RCD4 [AR]
CANT1	EPIPHYSEAL DYSPLASIA. MULTIPLE. 7; EDM7 [AR] | DESBUQUOIS DYSPLASIA 1; DBQD1 [AR]
CAPN3	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 1; LGMDR1 [AR] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL DOMINANT 4; LGMDD4 [AD]
CARD9	CANDIDIASIS. FAMILIAL. 2; CANDF2 [AR]
CASP8	CASPASE 8 DEFICIENCY [AR] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | LUNG CANCER [Somatic mutation] | BREAST CANCER [AD]
CASQ2	VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 2; CPVT2 [AR]
CATSPER1	SPERMATOGENIC FAILURE 7; SPGF7 [AR]
CAV1	PULMONARY HYPERTENSION. PRIMARY. 3; PPH3 [AD] | LIPODYSTROPHY. CONGENITAL GENERALIZED. TYPE 3; CGL3 [AR] | LIPODYSTROPHY. FAMILIAL PARTIAL. TYPE 7; FPLD7 [AD]
CAV3	RIPPLING MUSCLE DISEASE 2; RMD2 [AD] | MYOPATHY. DISTAL. TATEYAMA TYPE; MPDT [AD] | LONG QT SYNDROME 9; LQT9 [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 1; CMH1 [AD] | CREATINE PHOSPHOKINASE. ELEVATED SERUM [AD]
CBS	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY [AR]
CC2D1A	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 3; MRT3 [AR]
CC2D2A	COACH SYNDROME [AR] | JOUBERT SYNDROME 9; JBTS9 [AR] | MECKEL SYNDROME. TYPE 6; MKS6 [AR]
CCBE1	HENNEKAM LYMPHANGIECTASIA-LYMPHEDEMA SYNDROME 1; HKLLS1 [AR]
CCDC39	Ciliary dyskinesia primary 14 613807 (3)
CCDC8	THREE M SYNDROME 3; 3M3 [AR]
CCDC88C	SPINOCEREBELLAR ATAXIA 40; SCA40 [AD] | HYDROCEPHALUS. CONGENITAL. 1; HYC1 [AR]
CCT5	NEUROPATHY. HEREDITARY SENSORY. WITH SPASTIC PARAPLEGIA. AUTOSOMAL RECESSIVE [AR]
CD19	IMMUNODEFICIENCY. COMMON VARIABLE. 3; CVID3 [AR]
CD27	LYMPHOPROLIFERATIVE SYNDROME 2; LPFS2 [AR]
CD320	Methylmalonic aciduria transient due to transcobalamin receptor defect 613646 (3)
CD36	PLATELET GLYCOPROTEIN IV DEFICIENCY [AR]
CD3D	IMMUNODEFICIENCY 19; IMD19 [AR] | SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-POSITIVE [AR]
CD3E	IMMUNODEFICIENCY 18; IMD18 [AR] | SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-POSITIVE [AR]
CD3G	IMMUNODEFICIENCY 17; IMD17 [AR]
CD40	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 3; HIGM3 [AR]
CD40LG	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 1; HIGM1 [XLR]
CD79A	AGAMMAGLOBULINEMIA 3. AUTOSOMAL RECESSIVE; AGM3 [AR]
CD79B	AGAMMAGLOBULINEMIA 6. AUTOSOMAL RECESSIVE; AGM6 [AR]
CD81	IMMUNODEFICIENCY. COMMON VARIABLE. 6; CVID6 [AR]
CD8A	CD8 DEFICIENCY. FAMILIAL [AR]
CD96	C SYNDROME [AD]
CDAN1	ANEMIA. CONGENITAL DYSERYTHROPOIETIC. TYPE Ia; CDAN1A [AR]

CDH23	PITUITARY ADENOMA 5. MULTIPLE TYPES; PITA5 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 12; DFNB12 [AR] | USHER SYNDROME. TYPE ID; USH1D [AR]
CDH3	ECTODERMAL DYSPLASIA. ECTRODACTYLY. AND MACULAR DYSTROPHY SYNDROME; EEMS [AR] | HYPOTRICHOSIS. CONGENITAL. WITH JUVENILE MACULAR DYSTROPHY; HJMD [AR]
CDHR1	CONE-ROD DYSTROPHY 15; CORD15 [AR]
CDK5RAP2	MICROCEPHALY 3. PRIMARY. AUTOSOMAL RECESSIVE; MCPH3 [AR]
CDKL5	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 2; EIEE2 [XLD]
CDSN	HYPOTRICHOSIS 2; HYPT2 [AD] | PEELING SKIN SYNDROME 1; PSS1 [AR]
CDT1	MEIER-GORLIN SYNDROME 4; MGORS4 [AR]
CEBPE	SPECIFIC GRANULE DEFICIENCY 1; SGD1 [AR]
CENPJ	SECKEL SYNDROME 4; SCKL4 [AR] | MICROCEPHALY 6. PRIMARY. AUTOSOMAL RECESSIVE; MCPH6 [AR]
CEP135	MICROCEPHALY 8. PRIMARY. AUTOSOMAL RECESSIVE; MCPH8 [AR]
CEP152	SECKEL SYNDROME 5; SCKL5 [AR] | MICROCEPHALY 9. PRIMARY. AUTOSOMAL RECESSIVE; MCPH9 [AR]
CEP290	SENIOR-LOKEN SYNDROME 6; SLSN6 [AR] | BARDET-BIEDL SYNDROME 14; BBS14 [AR] | JOUBERT SYNDROME 5; JBTS5 [AR] | MECKEL SYNDROME. TYPE 4; MKS4 [AR]
CEP41	JOUBERT SYNDROME 15; JBTS15 [AR]
CEP57	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 2; MVA2 [AR]
CERKL	Retinitis pigmentosa 26 608380 (3)
CERS3	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 9; ARCI9 [AR]
CES1	Drug metabolism altered CES1-related 618057 (3)
CFC1	HETEROTAXY. VISCERAL. 2. AUTOSOMAL; HTX2 [AD]
CFD	COMPLEMENT FACTOR D DEFICIENCY; CFDD [AR]
CFH	BASAL LAMINAR DRUSEN [AD] | HEMOLYTIC UREMIC SYNDROME. ATYPICAL. SUSCEPTIBILITY TO. 1; AHUS1 [AR] | COMPLEMENT FACTOR H DEFICIENCY; CFHD [AD]
CFP	PROPERDIN DEFICIENCY. X-LINKED; CFPD [XLR]
CFTR	VAS DEFERENS. CONGENITAL BILATERAL APLASIA OF; CBAVD [AR] | BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 1; BESC1 [AD] | PANCREATITIS. HEREDITARY; PCTT [AD] | CYSTIC FIBROSIS; CF [AR]
CHAT	MYASTHENIC SYNDROME. CONGENITAL. 6. PRESYNAPTIC; CMS6 [AR]
CHKB	MUSCULAR DYSTROPHY. CONGENITAL. MEGACONIAL TYPE; MDCMC [AR]
CHM	CHOROIDEREMIA; CHM [XLD] | CHOROIDEREMIA. DEAFNESS. AND MENTAL RETARDATION [XLR]
CHMP1A	PONTOCEREBELLAR HYPOPLASIA. TYPE 8; PCH8 [AR]
CHRNA1	MULTIPLE PTERYGIUM SYNDROME. LETHAL TYPE; LMPS [AR] | MYASTHENIC SYNDROME. CONGENITAL. 1B. FAST-CHANNEL; CMS1B [AD] | MYASTHENIC SYNDROME. CONGENITAL. 1A. SLOW-CHANNEL; CMS1A [AD]
CHRNB1	MYASTHENIC SYNDROME. CONGENITAL. 2C. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS2C [AR] | MYASTHENIC SYNDROME. CONGENITAL. 2A. SLOW-CHANNEL; CMS2A [AD]
CHRND	MULTIPLE PTERYGIUM SYNDROME. LETHAL TYPE; LMPS [AR] | MYASTHENIC SYNDROME. CONGENITAL. 3B. FAST-CHANNEL; CMS3B [AR] | MYASTHENIC SYNDROME. CONGENITAL. 3C. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS3C [AR] | MYASTHENIC SYNDROME. CONGENITAL. 3A. SLOW-CHANNEL; CMS3A [AD]
CHRNE	MYASTHENIC SYNDROME. CONGENITAL. 4C. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS4C [AR] | MYASTHENIC SYNDROME. CONGENITAL. 4B. FAST-CHANNEL; CMS4B [AR] | MYASTHENIC SYNDROME. CONGENITAL. 4A. SLOW-CHANNEL; CMS4A [AD]
CHRNG	MULTIPLE PTERYGIUM SYNDROME. LETHAL TYPE; LMPS [AR] | MULTIPLE PTERYGIUM SYNDROME. ESCOBAR VARIANT; EVMPS [AR]
CHST14	EHLERS-DANLOS SYNDROME. MUSCULOCONTRACTURAL TYPE. 1; EDSMC1 [AR]
CHST3	SPONDYLOEPIPHYSEAL DYSPLASIA WITH CONGENITAL JOINT DISLOCATIONS; SEDCJD [AR]
CHST6	MACULAR DYSTROPHY. CORNEAL; MCD [AR]
CHSY1	TEMTAMY PREAXIAL BRACHYDACTYLY SYNDROME; TPBS [AR]
CHUK	Cocoon syndrome 613630 (3)
CIB2	DEAFNESS. AUTOSOMAL RECESSIVE 48; DFNB48 [AR] | USHER SYNDROME. TYPE IJ; USH1J [AR]
CISD2	WOLFRAM SYNDROME 2; WFS2 [AR]

CLCF1	CRISPONI/COLD-INDUCED SWEATING SYNDROME 2; CISS2 [AR]
CLCN1	MYOTONIA CONGENITA. AUTOSOMAL RECESSIVE [AR] | MYOTONIA CONGENITA. AUTOSOMAL DOMINANT [AD]
CLCN5	DENT DISEASE 1 [XLR] | NEPHROLITHIASIS. X-LINKED RECESSIVE. WITH RENAL FAILURE; XRN [XLR] | PROTEINURIA. LOW MOLECULAR WEIGHT. WITH HYPERCALCIURIA AND NEPHROCALCINOSIS [XLR] | HYPOPHOSPHATEMIC RICKETS. X-LINKED RECESSIVE [XLR]
CLCN7	HYPOPIGMENTATION. ORGANOMEGALY. AND DELAYED MYELINATION AND DEVELOPMENT; HOD [AD] | OSTEOPETROSIS. AUTOSOMAL RECESSIVE 4; OPTB4 [AR] | OSTEOPETROSIS. AUTOSOMAL DOMINANT 2; OPTA2 [AD]
CLCNKB	BARTTER SYNDROME. TYPE 3; BARTS3 [AR] | BARTTER SYNDROME. TYPE 4B. NEONATAL. WITH SENSORINEURAL DEAFNESS; BARTS4B [Digenic recessive]
CLDN1	ICHTHYOSIS. LEUKOCYTE VACUOLES. ALOPECIA. AND SCLEROSING CHOLANGITIS; ILVASC [AR]
CLDN14	DEAFNESS. AUTOSOMAL RECESSIVE 29; DFNB29 [AR]
CLDN16	HYPOMAGNESEMIA 3. RENAL; HOMG3 [AR]
CLDN19	HYPOMAGNESEMIA 5. RENAL. WITH OR WITHOUT OCULAR INVOLVEMENT; HOMG5 [AR]
CLEC7A	CANDIDIASIS. FAMILIAL. 4; CANDF4 [AR]
CLN3	CEROID LIPOFUSCINOSIS. NEURONAL. 3; CLN3 [AR]
CLN5	CEROID LIPOFUSCINOSIS. NEURONAL. 5; CLN5 [AR]
CLN6	CEROID LIPOFUSCINOSIS. NEURONAL. 4A. AUTOSOMAL RECESSIVE; CLN4A [AR] | CEROID LIPOFUSCINOSIS. NEURONAL. 6; CLN6 [AR]
CLN8	CEROID LIPOFUSCINOSIS. NEURONAL. 8; CLN8 [AR] | CEROID LIPOFUSCINOSIS. NEURONAL. 8. NORTHERN EPILEPSY VARIANT [AR]
CLPP	PERRAULT SYNDROME 3; PRLTS3 [AR]
CLRN1	USHER SYNDROME. TYPE IIIA; USH3A [AR]
CNGA1	Retinitis pigmentosa 49 613756 (3)
CNGA3	ACHROMATOPSIA 2; ACHM2 [AR]
CNGB1	RETINITIS PIGMENTOSA 45; RP45 [AR]
CNGB3	STARGARDT DISEASE 1; STGD1 [AR] | ACHROMATOPSIA 3; ACHM3 [AR]
CNNM4	JALILI SYNDROME [AR]
CNTNAP2	PITT-HOPKINS-LIKE SYNDROME 1; PTHSL1 [AR]
COG1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIg; CDG2G [AR]
COG4	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIj; CDG2J [AR] | SAUL-WILSON SYNDROME; SWILS [AD]
COG5	Congenital disorder of glycosylation type IIi 613612 (3)
COG7	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIe; CDG2E [AR]
COL11A1	FIBROCHONDROGENESIS 1; FBCG1 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 37; DFNA37 [AD] | STICKLER SYNDROME. TYPE II; STL2 [AD] | MARSHALL SYNDROME; MRSHS [AD]
COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA. AUTOSOMAL RECESSIVE; OSMEDB [AR] | DEAFNESS. AUTOSOMAL DOMINANT 13; DFNA13 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 53; DFNB53 [AR] | FIBROCHONDROGENESIS 2; FBCG2 [AD] | OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA. AUTOSOMAL DOMINANT; OSMEDA [AD]
COL17A1	EPITHELIAL RECURRENT EROSION DYSTROPHY; ERED [AD] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR]
COL18A1	KNOBLOCH SYNDROME 1; KNO1 [AR]
COL1A2	OSTEOGENESIS IMPERFECTA. TYPE II; OI2 [AD] | EHLERS-DANLOS SYNDROME. CARDIAC VALVULAR TYPE; EDSCV [AR] | OSTEOPOROSIS [AD] | EHLERS-DANLOS SYNDROME. ARTHROCHALASIA TYPE. 2; EDSARTH2 [AD] | OSTEOGENESIS IMPERFECTA. TYPE IV; OI4 [AD] | OSTEOGENESIS IMPERFECTA. TYPE III; OI3 [AD]
COL2A1	CZECH DYSPLASIA [AD] | SPONDYLOPERIPHERAL DYSPLASIA [AD] | OSTEOARTHRITIS WITH MILD CHONDRODYSPLASIA; OSCDP [AD] | SPONDYLOEPIPHYSEAL DYSPLASIA. STANESCU TYPE; SEDSTN [AD] | PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA. TORRANCE TYPE; PLSDT [AD] | SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC [AD] | STICKLER SYNDROME. TYPE I. NONSYNDROMIC OCULAR [AD] | LEGG-CALVE-PERTHES DISEASE; LCPD [Autosomal dominant vs. multifactoria] | ACHONDROGENESIS. TYPE II; ACG2 [AD] | AVASCULAR NECROSIS OF FEMORAL HEAD. PRIMARY. 1; ANFH1 [AD] | KNIEST DYSPLASIA [AD] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. STRUDWICK TYPE; SEMDSTWK [AD] | STICKLER SYNDROME. TYPE I; STL1 [AD] | EPIPHYSEAL DYSPLASIA. MULTIPLE. WITH MYOPIA AND CONDUCTIVE DEAFNESS; EDMMD [AD]
COL3A1	POLYMICROGYRIA WITH OR WITHOUT VASCULAR-TYPE EHLERS-DANLOS SYNDROME; PMGEDSV [AR] | EHLERS-DANLOS SYNDROME. VASCULAR TYPE; EDSVASC [AD]
COL4A3	ALPORT SYNDROME 3. AUTOSOMAL DOMINANT; ATS3 [AD] | ALPORT SYNDROME 2. AUTOSOMAL RECESSIVE; ATS2 [AR] | HEMATURIA. BENIGN FAMILIAL; BFH [AD]
COL4A4	ALPORT SYNDROME 2. AUTOSOMAL RECESSIVE; ATS2 [AR] | HEMATURIA. BENIGN FAMILIAL; BFH [AD]

COL4A5	ALPORT SYNDROME 1. X-LINKED; ATS1 [XLD] | LEIOMYOMATOSIS. DIFFUSE. WITH ALPORT SYNDROME; DL-ATS [X-linked]
COL6A1	ULLRICH CONGENITAL MUSCULAR DYSTROPHY 1; UCMD1 [AR] | BETHLEM MYOPATHY 1; BTHLM1 [AD]
COL6A2	ULLRICH CONGENITAL MUSCULAR DYSTROPHY 1; UCMD1 [AR] | BETHLEM MYOPATHY 1; BTHLM1 [AD] | MYOSCLEROSIS. AUTOSOMAL RECESSIVE [AR]
COL6A3	ULLRICH CONGENITAL MUSCULAR DYSTROPHY 1; UCMD1 [AR] | DYSTONIA 27; DYT27 [AR] | BETHLEM MYOPATHY 1; BTHLM1 [AD]
COL7A1	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 8; NDNC8 [AD] | EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN AND DEFORMITY OF NAILS [AD] | EPIDERMOLYSIS BULLOSA DYSTROPHICA. AUTOSOMAL DOMINANT; DDEB [AD] | EPIDERMOLYSIS BULLOSA PRURIGINOSA [AD] | EPIDERMOLYSIS BULLOSA DYSTROPHICA. PRETIBIAL [AD] | TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN; TBDN [AD] | EPIDERMOLYSIS BULLOSA DYSTROPHICA. AUTOSOMAL RECESSIVE; RDEB [AR]
COL9A1	EPIPHYSEAL DYSPLASIA. MULTIPLE. 6; EDM6 [AD]
COL9A2	STICKLER SYNDROME. TYPE V; STL5 [AR] | EPIPHYSEAL DYSPLASIA. MULTIPLE. 2; EDM2 [AD]
COLEC11	3MC SYNDROME 2; 3MC2 [AR]
COLQ	MYASTHENIC SYNDROME. CONGENITAL. 5; CMS5 [AR]
COQ2	COENZYME Q10 DEFICIENCY. PRIMARY. 1; COQ10D1 [AR] | MULTIPLE SYSTEM ATROPHY 1. SUSCEPTIBILITY TO; MSA1 [AD]
COQ6	COENZYME Q10 DEFICIENCY. PRIMARY. 6; COQ10D6 [AR]
COQ9	COENZYME Q10 DEFICIENCY. PRIMARY. 5; COQ10D5 [AR]
COX15	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR] | CARDIOENCEPHALOMYOPATHY. FATAL INFANTILE. DUE TO CYTOCHROME c OXIDASE DEFICIENCY 2; CEMCOX2 [AR]
COX4I2	EXOCRINE PANCREATIC INSUFFICIENCY. DYSERYTHROPOIETIC ANEMIA. AND CALVARIAL HYPEROSTOSIS [AR]
COX6B1	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
CP	Cerebellar ataxia 604290 (3) | Hemosiderosis systemic due to aceruloplasminemia 604290 (3) | [Hypoceruloplasminemia hereditary] 604290 (3)
CPA6	FEBRILE SEIZURES. FAMILIAL. 11; FEB11 [AR] | EPILEPSY. FAMILIAL TEMPORAL LOBE. 5; ETL5 [AD]
CPN1	CARBOXYPEPTIDASE N DEFICIENCY [AR]
CPS1	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY. HYPERAMMONEMIA DUE TO [AR]
CPT1A	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY [AR]
CPT2	ENCEPHALOPATHY. ACUTE. INFECTION-INDUCED. SUSCEPTIBILITY TO. 4; IIAE4 [AD] | CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY. LETHAL NEONATAL [AR] | CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY. INFANTILE [AR] | CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY. MYOPATHIC. STRESS-INDUCED [AR]
CRADD	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 34. WITH VARIANT LISSENCEPHALY; MRT34 [AR]
CRB1	LEBER CONGENITAL AMAUROSIS 8; LCA8 [AR] | RETINITIS PIGMENTOSA 12; RP12 [AR] | PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY; PPCRA [AD]
CRBN	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 2; MRT2 [AR]
CRLF1	CRISPONI/COLD-INDUCED SWEATING SYNDROME 1; CISS1 [AR]
CRTAP	OSTEOGENESIS IMPERFECTA. TYPE VII; OI7 [AR]
CRX	CONE-ROD DYSTROPHY 2; CORD2 [Autosomal dominant (19q13.1-q13.2)]
CRYAA	CATARACT 9. MULTIPLE TYPES; CTRCT9 [AD]
CRYAB	MYOPATHY. MYOFIBRILLAR. FATAL INFANTILE HYPERTONIC. ALPHA-B CRYSTALLIN-RELATED [AR] | CATARACT 16. MULTIPLE TYPES; CTRCT16 [AD] | MYOPATHY. MYOFIBRILLAR. 2; MFM2 [AD] | CARDIOMYOPATHY. DILATED. 1II; CMD1II [AD]
CRYBB1	CATARACT 17. MULTIPLE TYPES; CTRCT17 [AD]
CRYBB3	CATARACT 22. MULTIPLE TYPES; CTRCT22 [AD]
CSF2RB	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 5; SMDP5 [AR]
CSTA	PEELING SKIN SYNDROME 4; PSS4 [AR]
CSTB	MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG [AR]
CTC1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS 1; CRMCC1 [AR]
CTDP1	CONGENITAL CATARACTS. FACIAL DYSMORPHISM. AND NEUROPATHY; CCFDN [AR]
CTH	CYSTATHIONINURIA [AR]
CTNS	CYSTINOSIS. NEPHROPATHIC; CTNS [AR] | CYSTINOSIS. ADULT NONNEPHROPATHIC [AR] | CYSTINOSIS. LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE [AR]

CTSA	GALACTOSIALIDOSIS; GSL [AR]
CTSC	PERIODONTITIS. AGGRESSIVE. 1 [AR] | PAPILLON-LEFEVRE SYNDROME; PALS [AR] | HAIM-MUNK SYNDROME; HMS [AR]
CTSD	CEROID LIPOFUSCINOSIS. NEURONAL. 10; CLN10 [AR]
CTSF	CEROID LIPOFUSCINOSIS. NEURONAL. 13; CLN13 [AR]
CTSK	PYCNODYSOSTOSIS [AR]
CUBN	MEGALOBLASTIC ANEMIA 1 [AR]
CUL4B	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CABEZAS TYPE; MRXSC [XLR]
CUL7	THREE M SYNDROME 1; 3M1 [AR]
CYB5A	METHEMOGLOBINEMIA AND AMBIGUOUS GENITALIA; METAG [AR]
CYB5R3	METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE [AR]
CYBA	GRANULOMATOUS DISEASE. CHRONIC. AUTOSOMAL RECESSIVE. CYTOCHROME b-NEGATIVE [AR]
CYBB	GRANULOMATOUS DISEASE. CHRONIC. X-LINKED; CDGX [XLR] | IMMUNODEFICIENCY 34; IMD34 [XLR]
CYP11A1	Adrenal insufficiency congenital with 46XY sex reversal partial or complete 613743 (3)
CYP11B1	HYPERALDOSTERONISM. FAMILIAL. TYPE I; HALD1 [AD] | ADRENAL HYPERPLASIA. CONGENITAL. DUE TO STEROID 11-BETA-HYDROXYLASE DEFICIENCY [AR]
CYP11B2	HYPERALDOSTERONISM. FAMILIAL. TYPE I; HALD1 [AD] | CORTICOSTERONE METHYLOXIDASE TYPE II DEFICIENCY [AR] | CORTICOSTERONE METHYLOXIDASE TYPE I DEFICIENCY [AR]
CYP17A1	ADRENAL HYPERPLASIA. CONGENITAL. DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY [AR]
CYP19A1	AROMATASE EXCESS SYNDROME; AEXS [Male-limited autosomal dominant vs. autosomal recessive or X-linke]
CYP1B1	GLAUCOMA 3. PRIMARY CONGENITAL. A; GLC3A [AR]
CYP21A2	ADRENAL HYPERPLASIA. CONGENITAL. DUE TO 21-HYDROXYLASE DEFICIENCY [AR]
CYP24A1	HYPERCALCEMIA. INFANTILE. 1; HCINF1 [AR]
CYP27A1	CEREBROTENDINOUS XANTHOMATOSIS; CTX [AR]
CYP27B1	VITAMIN D HYDROXYLATION-DEFICIENT RICKETS. TYPE 1A; VDDR1A [AR]
CYP2A6	TOBACCO ADDICTION. SUSCEPTIBILITY TO [Genetic factors seem to contribute only in light smokers and heavy smoker] | COUMARIN RESISTANCE [AD] | LUNG CANCER [Somatic mutation]
CYP2C19	DRUG METABOLISM. POOR. CYP2C19-RELATED [AR]
CYP2R1	VITAMIN D HYDROXYLATION-DEFICIENT RICKETS. TYPE 1B; VDDR1B [AR]
CYP2U1	SPASTIC PARAPLEGIA 56. AUTOSOMAL RECESSIVE; SPG56 [AR]
CYP4F22	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 5; ARCI5 [AR]
CYP4V2	BIETTI CRYSTALLINE CORNEORETINAL DYSTROPHY; BCD [AR]
CYP7B1	BILE ACID SYNTHESIS DEFECT. CONGENITAL. 3; CBAS3 [AR] | SPASTIC PARAPLEGIA 5A. AUTOSOMAL RECESSIVE; SPG5A [AR]
D2HGDH	D-2-HYDROXYGLUTARIC ACIDURIA 1; D2HGA1 [AR]
DAG1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 9; MDDGA9 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 9; MDDGC9 [AR]
DARS2	LEUKOENCEPHALOPATHY WITH BRAINSTEM AND SPINAL CORD INVOLVEMENT AND LACTATE ELEVATION; LBSL [AR]
DBH	ORTHOSTATIC HYPOTENSION 1; ORTHYP1 [AR]
DBT	MAPLE SYRUP URINE DISEASE; MSUD [AR]
DCAF17	WOODHOUSE-SAKATI SYNDROME; WDSKS [AR]
DCHS1	MITRAL VALVE PROLAPSE 2; MVP2 [AD] | VAN MALDERGEM SYNDROME 1; VMLDS1 [AR]
DCLRE1C	SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION [AR] | OMENN SYNDROME [AR]
DCX	LISSENCEPHALY. X-LINKED. 1; LISX1 [X-linked]
DDB2	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP E [AR]
DDC	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY [AR]
DDHD1	SPASTIC PARAPLEGIA 28. AUTOSOMAL RECESSIVE; SPG28 [AR]

DDHD2	SPASTIC PARAPLEGIA 54. AUTOSOMAL RECESSIVE; SPG54 [AR]
DDR2	SPONDYLOMETAEPIPHYSEAL DYSPLASIA. SHORT LIMB-HAND TYPE [AR] | WARBURG-CINOTTI SYNDROME; WRCN [AD]
DDX11	WARSAW BREAKAGE SYNDROME; WABS [AR]
DGKE	NEPHROTIC SYNDROME. TYPE 7; NPHS7 [AR]
DGUOK	MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (HEPATOCEREBRAL TYPE); MTDPS3 [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL RECESSIVE 4; PEOB4 [AR] | PORTAL HYPERTENSION. NONCIRRHOTIC; NCPH [AR]
DHCR24	DESMOSTEROLOSIS [AR]
DHCR7	SMITH-LEMLI-OPITZ SYNDROME; SLOS [AR]
DHDDS	DEVELOPMENTAL DELAY AND SEIZURES WITH OR WITHOUT MOVEMENT ABNORMALITIES; DEDSM [AD] | RETINITIS PIGMENTOSA 59; RP59 [AR]
DHFR	MEGALOBLASTIC ANEMIA DUE TO DIHYDROFOLATE REDUCTASE DEFICIENCY [AR]
DHH	46.XY SEX REVERSAL 7; SRXY7 [AR]
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS; POADS [AR]
DIS3L2	PERLMAN SYNDROME; PRLMNS [AR]
DKC1	DYSKERATOSIS CONGENITA. X-LINKED; DKCX [XLR]
DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY; PDHDD [AR]
DLD	MAPLE SYRUP URINE DISEASE; MSUD [AR] | DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY; DLDD [AR]
DLL3	SPONDYLOCOSTAL DYSOSTOSIS 1. AUTOSOMAL RECESSIVE; SCDO1 [AR]
DMD	CARDIOMYOPATHY. DILATED. 3B; CMD3B [X-linked] | MUSCULAR DYSTROPHY. DUCHENNE TYPE; DMD [XLR] | MUSCULAR DYSTROPHY. BECKER TYPE; BMD [XLR]
DMGDH	DIMETHYLGLYCINE DEHYDROGENASE DEFICIENCY; DMGDHD [AR]
DMP1	HYPOPHOSPHATEMIC RICKETS. AUTOSOMAL RECESSIVE. 1; ARHR1 [AR]
DNAAF1	CILIARY DYSKINESIA. PRIMARY. 13; CILD13 [AR]
DNAH11	CILIARY DYSKINESIA. PRIMARY. 7; CILD7 [AR]
DNAH5	Ciliary dyskinesia primary 3 with or without situs inversus 608644 (3)
DNAI1	CILIARY DYSKINESIA. PRIMARY. 1; CILD1 [AR]
DNAJB2	SPINAL MUSCULAR ATROPHY. DISTAL. AUTOSOMAL RECESSIVE. 5; DSMA5 [AR]
DNAJC19	3-METHYLGLUTACONIC ACIDURIA. TYPE V; MGCA5 [AR]
DNAL1	CILIARY DYSKINESIA. PRIMARY. 16; CILD16 [AR]
DNASE1L3	SYSTEMIC LUPUS ERYTHEMATOSUS 16; SLEB16 [AR]
DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1; ICF1 [AR]
DOCK6	ADAMS-OLIVER SYNDROME 2; AOS2 [AR]
DOCK8	HYPER-IgE RECURRENT INFECTION SYNDROME 2. AUTOSOMAL RECESSIVE; HIES2 [AR]
DOK7	MYASTHENIC SYNDROME. CONGENITAL. 10; CMS10 [AR] | FETAL AKINESIA DEFORMATION SEQUENCE 1; FADS1 [AR]
DOLK	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Im; CDG1M [AR]
DPAGT1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ij; CDG1J [AR] | MYASTHENIC SYNDROME. CONGENITAL. 13; CMS13 [AR]
DPM1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ie; CDG1E [AR]
DPY19L2	SPERMATOGENIC FAILURE 9; SPGF9 [AR]
DPYD	DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY [AR]
DPYS	DIHYDROPYRIMIDINASE DEFICIENCY; DPYSD [AR]
DSG2	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 10; ARVD10 [AD]
DSG4	HYPOTRICHOSIS 6; HYPT6 [AR]
DSP	EPIDERMOLYSIS BULLOSA. LETHAL ACANTHOLYTIC; EBLA [AR] | CARDIOMYOPATHY. DILATED. WITH WOOLLY HAIR AND KERATODERMA; DCWHK [AR] | KERATOSIS PALMOPLANTARIS STRIATA II; PPKS2 [AD] | ARRHYTHMOGENIC RIGHT VENTRICULAR

	DYSPLASIA. FAMILIAL. 8; ARVD8 [AD] | CARDIOMYOPATHY. DILATED. WITH WOOLLY HAIR. KERATODERMA. AND TOOTH AGENESIS; DCWHKTA [AD] | SKIN FRAGILITY-WOOLLY HAIR SYNDROME; SFWHS [AR]
DST	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE VI; HSAN6 [AR] | EPIDERMOLYSIS BULLOSA SIMPLEX. AUTOSOMAL RECESSIVE 2; EBSB2 [AR]
DTNBP1	HERMANSKY-PUDLAK SYNDROME 7; HPS7 [AR]
DUOX2	THYROID DYSHORMONOGENESIS 6; TDH6 [AR]
DUOXA2	THYROID DYSHORMONOGENESIS 5; TDH5 [AR]
DYM	SMITH-MCCORT DYSPLASIA 1; SMC1 [AR] | DYGGVE-MELCHIOR-CLAUSEN DISEASE; DMC [AR]
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY; SRTD3 [AR]
DYSF	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 2; LGMDR2 [AR] | MYOPATHY. DISTAL. WITH ANTERIOR TIBIAL ONSET; DMAT [AR] | MIYOSHI MUSCULAR DYSTROPHY 1; MMD1 [AR]
ECEL1	ARTHROGRYPOSIS. DISTAL. TYPE 5D; DA5D [AR]
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE [AR]
EDA	TOOTH AGENESIS. SELECTIVE. X-LINKED. 1; STHAGX1 [XLD] | ECTODERMAL DYSPLASIA 1. HYPOHIDROTIC. X-LINKED; XHED [XLR]
EDAR	ECTODERMAL DYSPLASIA 10A. HYPOHIDROTIC/HAIR/NAIL TYPE. AUTOSOMAL DOMINANT; ECTD10A [AD] | ECTODERMAL DYSPLASIA 10B. HYPOHIDROTIC/HAIR/TOOTH TYPE. AUTOSOMAL RECESSIVE; ECTD10B [AR]
EDARADD	ECTODERMAL DYSPLASIA 11A. HYPOHIDROTIC/HAIR/TOOTH TYPE. AUTOSOMAL DOMINANT; ECTD11A [AD] | ECTODERMAL DYSPLASIA 10A. HYPOHIDROTIC/HAIR/NAIL TYPE. AUTOSOMAL DOMINANT; ECTD10A [AD] | ECTODERMAL DYSPLASIA 10B. HYPOHIDROTIC/HAIR/TOOTH TYPE. AUTOSOMAL RECESSIVE; ECTD10B [AR] | ECTODERMAL DYSPLASIA 11B. HYPOHIDROTIC/HAIR/TOOTH TYPE. AUTOSOMAL RECESSIVE; ECTD11B [AR]
EDN3	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD] | WAARDENBURG SYNDROME. TYPE 4B; WS4B [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 4; HSCR4 [AD]
EDNRB	ABCD SYNDROME; ABCDS [AR] | WAARDENBURG SYNDROME. TYPE 4A; WS4A [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 2; HSCR2 [AD]
EFEMP2	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IB; ARCL1B [AR]
EGF	Hypomagnesemia 4 renal 611718 (3)
EGFR	INFLAMMATORY SKIN AND BOWEL DISEASE. NEONATAL. 2; NISBD2 [AR] | LUNG CANCER [Somatic mutation]
EGR2	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS [AD] | NEUROPATHY. CONGENITAL HYPOMYELINATING. 1. AUTOSOMAL RECESSIVE; CHN1 [AR] | CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1D; CMT1D [AD]
EIF2AK3	EPIPHYSEAL DYSPLASIA. MULTIPLE. WITH EARLY-ONSET DIABETES MELLITUS [AR]
EIF2B1	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM [AR]
EIF2B2	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM [AR]
EIF2B3	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM [AR]
EIF2B4	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM [AR]
EIF2B5	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM [AR]
ELANE	CYCLIC NEUTROPENIA [AD] | NEUTROPENIA. SEVERE CONGENITAL. 1. AUTOSOMAL DOMINANT; SCN1 [AD]
ELOVL4	SPINOCEREBELLAR ATAXIA 34; SCA34 [AD] | ICHTHYOSIS. SPASTIC QUADRIPLEGIA. AND MENTAL RETARDATION; ISQMR [AR] | STARGARDT DISEASE 3; STGD3 [AD]
EMD	EMERY-DREIFUSS MUSCULAR DYSTROPHY 1. X-LINKED; EDMD1 [XLR]
EMG1	BOWEN-CONRADI SYNDROME; BWCNS [AR]
ENAM	AMELOGENESIS IMPERFECTA. TYPE IB; AI1B [AD] | AMELOGENESIS IMPERFECTA. TYPE IC; AI1C [AR]
ENPP1	ARTERIAL CALCIFICATION. GENERALIZED. OF INFANCY. 1; GACI1 [AR] | HYPOPHOSPHATEMIC RICKETS. AUTOSOMAL RECESSIVE. 2; ARHR2 [AR] | OBESITY [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | COLE DISEASE; COLED [AD]
EOGT	ADAMS-OLIVER SYNDROME 4; AOS4 [AR]
EPB42	Spherocytosis type 5 612690 (3)
EPCAM	DIARRHEA 5. WITH TUFTING ENTEROPATHY. CONGENITAL; DIAR5 [AR]
EPG5	VICI SYNDROME; VICIS [AR]
EPM2A	MYOCLONIC EPILEPSY OF LAFORA [AR]
ERBB3	ERYTHROLEUKEMIA. FAMILIAL. SUSCEPTIBILITY TO; FERLK [AD] | LETHAL CONGENITAL CONTRACTURE SYNDROME 2; LCCS2 [AR]
ERCC2	TRICHOTHIODYSTROPHY 1. PHOTOSENSITIVE; TTD1 [AR] | XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP D; XPD [AR] | CEREBROOCULOFACIOSKELETAL SYNDROME 2; COFS2 [AR]

ERCC3	TRICHOTHIODYSTROPHY 2. PHOTOSENSITIVE; TTD2 [AR] | XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP B; XPB [AR]
ERCC4	XFE PROGEROID SYNDROME; XFEPS [AR] | XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP F; XPF [AR] | FANCONI ANEMIA. COMPLEMENTATION GROUP Q; FANCQ [AR]
ERCC5	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP G; XPG [AR] | CEREBROOCULOFACIOSKELETAL SYNDROME 3; COFS3 [AR]
ERCC6	CEREBROOCULOFACIOSKELETAL SYNDROME 1; COFS1 [AR] | COCKAYNE SYNDROME B; CSB [AR] | UV-SENSITIVE SYNDROME 1; UVSS1 [AR] | DE SANCTIS-CACCHIONE SYNDROME [AR] | PREMATURE OVARIAN FAILURE 11; POF11 [AD] | LUNG CANCER [Somatic mutation]
ERCC8	COCKAYNE SYNDROME A; CSA [AR] | UV-SENSITIVE SYNDROME 2; UVSS2 [AR]
ERLIN2	SPASTIC PARAPLEGIA 18. AUTOSOMAL RECESSIVE; SPG18 [AR]
ESCO2	ROBERTS SYNDROME; RBS [AR] | SC PHOCOMELIA SYNDROME [AR]
ESPN	USHER SYNDROME. TYPE 1M; USH1M [AR] | DEAFNESS. AUTOSOMAL RECESSIVE 36. WITH OR WITHOUT VESTIBULAR INVOLVEMENT; DFNB36 [AR]
ESRRB	DEAFNESS. AUTOSOMAL RECESSIVE 35; DFNB35 [AR]
ETFA	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD [AR]
ETFB	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD [AR]
ETFDH	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY; MADD [AR]
ETHE1	ENCEPHALOPATHY. ETHYLMALONIC; EE [AR]
EVC	ELLIS-VAN CREVELD SYNDROME; EVC [AR] | WEYERS ACROFACIAL DYSOSTOSIS; WAD [AD]
EVC2	ELLIS-VAN CREVELD SYNDROME; EVC [AR] | WEYERS ACROFACIAL DYSOSTOSIS; WAD [AD]
EXOSC3	PONTOCEREBELLAR HYPOPLASIA. TYPE 1B; PCH1B [AR]
EXPH5	EPIDERMOLYSIS BULLOSA. NONSPECIFIC. AUTOSOMAL RECESSIVE; EBNS [AR]
EXT1	CHONDROSARCOMA [AR] | EXOSTOSES. MULTIPLE. TYPE I; EXT1 [AD] | TRICHORHINOPHALANGEAL SYNDROME. TYPE II; TRPS2 [AD]
EYS	RETINITIS PIGMENTOSA 25; RP25 [AR]
F10	FACTOR X DEFICIENCY [AR]
F11	Factor XI deficiency autosomal dominant 612416 (3) | Factor XI deficiency autosomal recessive 612416 (3)
F12	ANGIOEDEMA. HEREDITARY. TYPE III; HAE3 [AD] | FACTOR XII DEFICIENCY [AR]
F13B	FACTOR XIII. B SUBUNIT. DEFICIENCY OF [AR]
F2	STROKE. ISCHEMIC [Multifactorial predispositio] | THROMBOPHILIA DUE TO THROMBIN DEFECT; THPH1 [AD]
F5	BUDD-CHIARI SYNDROME; BDCHS [AR] | STROKE. ISCHEMIC [Multifactorial predispositio]
F7	Factor VII deficiency 227500 (3) | {Myocardial infarction decreased susceptibility to} 608446 (3)
F8	Hemophilia A 306700 (3)
F9	COUMARIN RESISTANCE [AD]
FA2H	SPASTIC PARAPLEGIA 35. AUTOSOMAL RECESSIVE; SPG35 [AR]
FADD	INFECTIONS. RECURRENT. WITH ENCEPHALOPATHY. HEPATIC DYSFUNCTION. AND CARDIOVASCULAR MALFORMATIONS [AR]
FAH	TYROSINEMIA. TYPE I; TYRSN1 [AR]
FAM126A	LEUKODYSTROPHY. HYPOMYELINATING. 5; HLD5 [AR]
FAM161A	Retinitis pigmentosa 28 606068 (3)
FAM20A	AMELOGENESIS IMPERFECTA. TYPE IG; AI1G [AR]
FAM20C	RAINE SYNDROME; RNS [AR]
FANCA	FANCONI ANEMIA. COMPLEMENTATION GROUP A; FANCA [AR]
FANCB	FANCONI ANEMIA. COMPLEMENTATION GROUP B; FANCB [XLR]
FANCC	FANCONI ANEMIA. COMPLEMENTATION GROUP C; FANCC [Autosomal recessive with at least two loci and multiple allele]
FANCD2	FANCONI ANEMIA. COMPLEMENTATION GROUP D2; FANCD2 [AR]
FANCE	FANCONI ANEMIA. COMPLEMENTATION GROUP E; FANCE [AR]
FANCG	Fanconi anemia complementation group G 614082 (3)
FANCI	FANCONI ANEMIA. COMPLEMENTATION GROUP I; FANCI [AR]

FANCL	FANCONI ANEMIA. COMPLEMENTATION GROUP L; FANCL [AR]
FANCM	PREMATURE OVARIAN FAILURE 15; POF15 [AR] | SPERMATOGENIC FAILURE 28; SPGF28 [AR]
FAS	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS [AD]
FAT4	VAN MALDERGEM SYNDROME 2; VMLDS2 [AR] | HENNEKAM LYMPHANGIECTASIA-LYMPHEDEMA SYNDROME 2; HKLLS2 [AR]
FBLN5	CUTIS LAXA. AUTOSOMAL DOMINANT 2; ADCL2 [AD] | CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IA; ARCL1A [AR] | NEUROPATHY. HEREDITARY. WITH OR WITHOUT AGE-RELATED MACULAR DEGENERATION; HNARMD [AD] | CUTIS LAXA. AUTOSOMAL DOMINANT 1; ADCL1 [AD]
FBN1	ACROMICRIC DYSPLASIA; ACMICD [AD] | MARFAN SYNDROME; MFS [AD] | GELEOPHYSIC DYSPLASIA 2; GPHYSD2 [AD] | MARFANOID-PROGEROID-LIPODYSTROPHY SYNDROME; MFLS [AD] | WEILL-MARCHESANI SYNDROME 2; WMS2 [AD] | MASS SYNDROME [AD] | STIFF SKIN SYNDROME; SSKS [AD] | ECTOPIA LENTIS 1. ISOLATED. AUTOSOMAL DOMINANT; ECTOL1 [AD]
FBP1	FRUCTOSE-1.6-BISPHOSPHATASE DEFICIENCY; FBP1D [AR]
FBXO7	PARKINSON DISEASE 15. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK15 [AR]
FCN3	FICOLIN 3 DEFICIENCY [AR]
FECH	PROTOPORPHYRIA. ERYTHROPOIETIC. 1; EPP1 [AR]
FERMT1	KINDLER SYNDROME; KNDLRS [AR]
FERMT3	LEUKOCYTE ADHESION DEFICIENCY. TYPE III; LAD3 [AR]
FGA	AMYLOIDOSIS. FAMILIAL VISCERAL [AD] | AFIBRINOGENEMIA. CONGENITAL [AR]
FGB	AFIBRINOGENEMIA. CONGENITAL [AR]
FGD1	AARSKOG-SCOTT SYNDROME; AAS [XLR]
FGD4	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4H; CMT4H [AR]
FGF16	METACARPAL 4-5 FUSION; MF4 [XLR]
FGF23	HYPOPHOSPHATEMIC RICKETS. AUTOSOMAL DOMINANT; ADHR [AD] | TUMORAL CALCINOSIS. HYPERPHOSPHATEMIC. FAMILIAL. 1; HFTC1 [AR]
FGF3	DEAFNESS. CONGENITAL. WITH INNER EAR AGENESIS. MICROTIA. AND MICRODONTIA [AR]
FGFR2	SAETHRE-CHOTZEN SYNDROME; SCS [AD] | LACRIMOAURICULODENTODIGITAL SYNDROME; LADD [AD] | APERT SYNDROME [AD] | ANTLEY-BIXLER SYNDROME WITHOUT GENITAL ANOMALIES OR DISORDERED STEROIDOGENESIS; ABS2 [AD] | JACKSON-WEISS SYNDROME; JWS [AD] | BENT BONE DYSPLASIA SYNDROME; BBDS [AD] | CROUZON SYNDROME [AD] | PFEIFFER SYNDROME [AD] | BEARE-STEVENSON CUTIS GYRATA SYNDROME; BSTVS [AD]
FGG	AFIBRINOGENEMIA. CONGENITAL [AR]
FH	Fumarase deficiency 606812 (3) | Leiomyomatosis and renal cell cancer 150800 (3)
FIG4	YUNIS-VARON SYNDROME; YVS [AR] | CHARCOT-MARIE-TOOTH DISEASE. TYPE 4J; CMT4J [AR] | AMYOTROPHIC LATERAL SCLEROSIS 11; ALS11 [AD] | POLYMICROGYRIA. BILATERAL TEMPOROOCCIPITAL; BTOP [AR]
FKBP10	BRUCK SYNDROME 1; BRKS1 [AR] | OSTEOGENESIS IMPERFECTA. TYPE XI; OI11 [AR]
FKBP14	EHLERS-DANLOS SYNDROME. KYPHOSCOLIOTIC TYPE. 2; EDSKSCL2 [AR]
FKRP	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 5; MDDGA5 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 5; MDDGC5 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH OR WITHOUT MENTAL RETARDATION). TYPE B. 5; MDDGB5 [AR]
FKTN	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITHOUT MENTAL RETARDATION). TYPE B. 4; MDDGB4 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 4; MDDGA4 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 4; MDDGC4 [AR] | CARDIOMYOPATHY. DILATED. 1X; CMD1X [AR]
FLNA	TERMINAL OSSEOUS DYSPLASIA; TOD [XLD] | INTESTINAL PSEUDOOBSTRUCTION. NEURONAL. CHRONIC IDIOPATHIC. X-LINKED [XLR] | FRONTOMETAPHYSEAL DYSPLASIA 1; FMD1 [XLR] | FG SYNDROME 2; FGS2 [X-linked] | CARDIAC VALVULAR DYSPLASIA. X-LINKED; CVD1 [X-linked] | OTOPALATODIGITAL SYNDROME. TYPE I; OPD1 [XLD] | OTOPALATODIGITAL SYNDROME. TYPE II; OPD2 [XLD] | MELNICK-NEEDLES SYNDROME; MNS [XLD] | PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1 [XLD]
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME; SCT [AR] | BOOMERANG DYSPLASIA; BOOMD [AD] | LARSEN SYNDROME; LRS [AD] | ATELOSTEOGENESIS. TYPE III; AO3 [AD] | ATELOSTEOGENESIS. TYPE I; AO1 [AD]
FLT4	HEMANGIOMA. CAPILLARY INFANTILE [AD] | LYMPHATIC MALFORMATION 1; LMPHM1 [AD] | CONGENITAL HEART DEFECTS. MULTIPLE TYPES. 7; CHTD7 [AD]
FLVCR1	POSTERIOR COLUMN ATAXIA WITH RETINITIS PIGMENTOSA; AXPC1 [AR]
FLVCR2	PROLIFERATIVE VASCULOPATHY AND HYDRANENCEPHALY-HYDROCEPHALY SYNDROME; PVHH [AR]
FMO3	TRIMETHYLAMINURIA; TMAU [AR]

FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY [AR]
FOXE1	THYROID CANCER. NONMEDULLARY. 4; NMTC4 [AD] | HYPOTHYROIDISM. THYROIDAL OR ATHYROIDAL. WITH SPIKY HAIR AND CLEFT PALATE [AR]
FOXE3	AORTIC ANEURYSM. FAMILIAL THORACIC 11. SUSCEPTIBILITY TO; AAT11 [AD] | ANTERIOR SEGMENT DYSGENESIS 2; ASGD2 [AR]
FOXF1	ALVEOLAR CAPILLARY DYSPLASIA WITH MISALIGNMENT OF PULMONARY VEINS; ACDMPV [AD]
FOXI1	DEAFNESS. AUTOSOMAL RECESSIVE 4. WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4 [AR]
FOXL2	PREMATURE OVARIAN FAILURE 3; POF3 [AD] | BLEPHAROPHIMOSIS. PTOSIS. AND EPICANTHUS INVERSUS; BPES [AD]
FOXN1	T-CELL IMMUNODEFICIENCY. CONGENITAL ALOPECIA. AND NAIL DYSTROPHY; TIDAND [AR]
FOXO1	RHABDOMYOSARCOMA 2; RMS2 [Somatic mutation]
FOXP3	IMMUNODYSREGULATION. POLYENDOCRINOPATHY. AND ENTEROPATHY. X-LINKED; IPEX [XLR]
FOXRED1	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 19; MC1DN19 [AR]
FRAS1	FRASER SYNDROME 1; FRASRS1 [AR]
FREM1	TRIGONOCEPHALY 2; TRIGNO2 [AD] | MANITOBA OCULOTRICHOANAL SYNDROME; MOTA [AR]
FREM2	FRASER SYNDROME 2; FRASRS2 [AR] | CRYPTOPHTHALMOS. UNILATERAL OR BILATERAL. ISOLATED; CRYPTOP [AR] | FRASER SYNDROME 1; FRASRS1 [AR]
FRMD7	NYSTAGMUS 1. CONGENITAL. X-LINKED; NYS1 [X-linked]
FSHB	HYPOGONADOTROPIC HYPOGONADISM 24 WITHOUT ANOSMIA; HH24 [AR]
FSHR	TWINNING. DIZYGOTIC [Autosomal recessive vs. multifactoria] | OVARIAN DYSGENESIS 1; ODG1 [AR] | OVARIAN HYPERSTIMULATION SYNDROME; OHSS [AD]
FTCD	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY [AR]
FTO	GROWTH RETARDATION. DEVELOPMENTAL DELAY. AND FACIAL DYSMORPHISM; GDFD [AR] | BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 14; BMIQ14 [AR]
FUCA1	FUCOSIDOSIS [AR]
FUS	TREMOR. HEREDITARY ESSENTIAL. 4; ETM4 [AD]
FUT6	Fucosyltransferase 6 deficiency 613852 (3)
FXN	FRIEDREICH ATAXIA; FRDA [AR]
FYCO1	CATARACT 18; CTRCT18 [AR]
FZD6	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 1; NDNC1 [AR]
G6PC	GLYCOGEN STORAGE DISEASE Ia; GSD1A [AR]
G6PC3	NEUTROPENIA. SEVERE CONGENITAL. 4. AUTOSOMAL RECESSIVE; SCN4 [AR]
G6PD	ANEMIA. NONSPHEROCYTIC HEMOLYTIC. DUE TO G6PD DEFICIENCY [XLD]
GAA	GLYCOGEN STORAGE DISEASE II; GSD2 [AR]
GALC	KRABBE DISEASE [AR]
GALE	GALACTOSE EPIMERASE DEFICIENCY [AR]
GALK1	GALACTOKINASE DEFICIENCY [AR]
GALNS	MUCOPOLYSACCHARIDOSIS. TYPE IVA; MPS4A [AR]
GALNT3	TUMORAL CALCINOSIS. HYPERPHOSPHATEMIC. FAMILIAL. 1; HFTC1 [AR]
GALT	GALACTOSEMIA [AR]
GAMT	CEREBRAL CREATINE DEFICIENCY SYNDROME 2; CCDS2 [AR]
GAN	GIANT AXONAL NEUROPATHY 1. AUTOSOMAL RECESSIVE; GAN1 [AR]
GATA1	THROMBOCYTOPENIA. X-LINKED. WITH OR WITHOUT DYSERYTHROPOIETIC ANEMIA; XLTDA [XLR] | DOWN SYNDROME [Isolated cases] | ANEMIA. X-LINKED. WITH OR WITHOUT NEUTROPENIA AND/OR PLATELET ABNORMALITIES; XLANP [XLR] | THROMBOCYTOPENIA WITH BETA-THALASSEMIA. X-LINKED; XLTT [XLR]
GBA	PARKINSON DISEASE. LATE-ONSET; PD [AD] | GAUCHER DISEASE. TYPE II [AR] | DEMENTIA. LEWY BODY; DLB [AD] | GAUCHER DISEASE. TYPE IIIC [AR] | GAUCHER DISEASE. TYPE III [AR] | GAUCHER DISEASE. PERINATAL LETHAL [AR] | GAUCHER DISEASE. TYPE I [AR]
GBA2	SPASTIC PARAPLEGIA 46. AUTOSOMAL RECESSIVE; SPG46 [AR]
GBE1	GLYCOGEN STORAGE DISEASE IV; GSD4 [AR] | POLYGLUCOSAN BODY NEUROPATHY. ADULT FORM; APBN [AR]

GCDH	GLUTARIC ACIDEMIA I; GA1 [AR]
GCH1	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. B; HPABH4B [AR] | DYSTONIA. DOPA-RESPONSIVE; DRD [AD]
GCLC	GAMMA-GLUTAMYLCYSTEINE SYNTHETASE DEFICIENCY. HEMOLYTIC ANEMIA DUE TO [AR]
GCM2	HYPOPARATHYROIDISM. FAMILIAL ISOLATED; FIH [AD] | HYPERPARATHYROIDISM 4; HRPT4 [AD]
GDAP1	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4A; CMT4A [AR] | CHARCOT-MARIE-TOOTH DISEASE. RECESSIVE INTERMEDIATE A; CMTRIA [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. WITH VOCAL CORD PARESIS. AUTOSOMAL RECESSIVE [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2K; CMT2K [AR]
GDF5	BRACHYDACTYLY. TYPE A1. C; BDA1C [AR] | CHONDRODYSPLASIA. GREBE TYPE [AR] | SYMPHALANGISM. PROXIMAL. 1B; SYM1B [AD] | BRACHYDACTYLY. TYPE A2; BDA2 [AD] | MULTIPLE SYNOSTOSES SYNDROME 2; SYNS2 [AD] | DU PAN SYNDROME; DUPANS [AR] | BRACHYDACTYLY. TYPE C; BDC [AD] | MULTIPLE SYNOSTOSES SYNDROME 1; SYNS1 [AD] | ACROMESOMELIC DYSPLASIA. HUNTER-THOMPSON TYPE; AMDH [AR]
GFER	Myopathy mitochondrial progressive with congenital cataract hearing loss and developmental delay 613076 (3)
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1; COXPD1 [AR]
GFPT1	MYASTHENIC SYNDROME. CONGENITAL. 12; CMS12 [AR]
GGCX	VITAMIN K-DEPENDENT CLOTTING FACTORS. COMBINED DEFICIENCY OF. 1; VKCFD1 [AR]
GH1	KOWARSKI SYNDROME [AR] | ISOLATED GROWTH HORMONE DEFICIENCY. TYPE IA; IGHD1A [AR] | ISOLATED GROWTH HORMONE DEFICIENCY. TYPE II; IGHD2 [AD]
GHR	GROWTH HORMONE INSENSITIVITY. PARTIAL; GHIP [AD] | LARON SYNDROME [AR] | HYPERCHOLESTEROLEMIA. FAMILIAL. 1; FHCL1 [AD]
GHRHR	Growth hormone deficiency isolated type IV 618157 (3)
GHSR	GROWTH HORMONE DEFICIENCY. ISOLATED PARTIAL; GHDP [AD]
GIPC3	DEAFNESS. AUTOSOMAL RECESSIVE 15; DFNB15 [AR]
GJA1	OCULODENTODIGITAL DYSPLASIA. AUTOSOMAL RECESSIVE [AR] | OCULODENTODIGITAL DYSPLASIA; ODDD [AD] | SYNDACTYLY. TYPE III [AD] | HYPOPLASTIC LEFT HEART SYNDROME 1; HLHS1 [Autosomal recessive vs. multifactoria] | CRANIOMETAPHYSEAL DYSPLASIA. AUTOSOMAL RECESSIVE; CMDR [AR] | ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 3; EKVP3 [AD] | PALMOPLANTAR KERATODERMA AND CONGENITAL ALOPECIA 1; PPKCA1 [AD] | ATRIOVENTRICULAR SEPTAL DEFECT 3; AVSD3 [Autosomal dominant with variable expression and incomplete penetranc]
GJB2	BART-PUMPHREY SYNDROME; BAPS [AD] | ICHTHYOSIS. HYSTRIX-LIKE. WITH DEAFNESS [AD] | DEAFNESS. AUTOSOMAL DOMINANT 3A; DFNA3A [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 1A; DFNB1A [AR] | KERATITIS-ICHTHYOSIS-DEAFNESS SYNDROME. AUTOSOMAL DOMINANT; KIDAD [AD] | VOHWINKEL SYNDROME; VOWNKL [AD] | KERATODERMA. PALMOPLANTAR. WITH DEAFNESS [AD]
GJB3	ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 1; EKVP1 [AD] | DEAFNESS. AUTOSOMAL DOMINANT 2B; DFNA2B [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 1A; DFNB1A [AR]
GJB4	ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 2; EKVP2 [AD]
GJB6	DEAFNESS. AUTOSOMAL RECESSIVE 1A; DFNB1A [AR] | CLOUSTON SYNDROME [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 1B; DFNB1B [AR] | DEAFNESS. AUTOSOMAL DOMINANT 3B; DFNA3B [AD]
GJC2	LEUKODYSTROPHY. HYPOMYELINATING. 2; HLD2 [AR] | LYMPHATIC MALFORMATION 3; LMPHM3 [AD] | SPASTIC PARAPLEGIA 44. AUTOSOMAL RECESSIVE; SPG44 [AR]
GLA	FABRY DISEASE [X-linked]
GLB1	GM1-GANGLIOSIDOSIS. TYPE II [AR] | GM1-GANGLIOSIDOSIS. TYPE I [AR] | MUCOPOLYSACCHARIDOSIS. TYPE IVB; MPS4B [AR] | GM1-GANGLIOSIDOSIS. TYPE III [AR]
GLDC	GLYCINE ENCEPHALOPATHY; GCE [AR]
GLE1	LETHAL CONGENITAL CONTRACTURE SYNDROME 1; LCCS1 [AR] | CONGENITAL ARTHROGRYPOSIS WITH ANTERIOR HORN CELL DISEASE; CAAHD [AR]
GLI3	PALLISTER-HALL SYNDROME; PHS [AD] | HYPOTHALAMIC HAMARTOMAS [AR] | POLYDACTYLY. PREAXIAL IV; PPD4 [AD] | GREIG CEPHALOPOLYSYNDACTYLY SYNDROME; GCPS [AD] | POLYDACTYLY. POSTAXIAL. TYPE A1; PAPA1 [AD]
GLIS2	Nephronophthisis 7 611498 (3)
GLIS3	DIABETES MELLITUS. NEONATAL. WITH CONGENITAL HYPOTHYROIDISM; NDH [AR]
GLRA1	HYPEREKPLEXIA 1; HKPX1 [AD]
GLRB	HYPEREKPLEXIA 2; HKPX2 [AR]
GLRX5	ANEMIA. SIDEROBLASTIC. 3. PYRIDOXINE-REFRACTORY; SIDBA3 [AR] | SPASTICITY. CHILDHOOD-ONSET. WITH HYPERGLYCINEMIA; SPAHGC [AR] | ANEMIA. SIDEROBLASTIC. 2. PYRIDOXINE-REFRACTORY; SIDBA2 [AR]
GLUL	GLUTAMINE DEFICIENCY. CONGENITAL [AR]
GLYCTK	D-GLYCERIC ACIDURIA [AR]
GM2A	GM2-GANGLIOSIDOSIS. AB VARIANT [AR]

GNAT2	Achromatopsia 4 613856 (3)
GNE	SIALURIA [AD] | NONAKA MYOPATHY; NM [AR]
GNMT	GLYCINE N-METHYLTRANSFERASE DEFICIENCY [AR]
GNPAT	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA. TYPE 2; RCDP2 [AR]
GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA [AR] | MUCOLIPIDOSIS III ALPHA/BETA [AR]
GNPTG	MUCOLIPIDOSIS III GAMMA [AR]
GNRHR	HYPOGONADOTROPIC HYPOGONADISM 7 WITH OR WITHOUT ANOSMIA; HH7 [AR]
GNS	MUCOPOLYSACCHARIDOSIS. TYPE IIID; MPS3D [AR]
GORAB	GERODERMA OSTEODYSPLASTICUM; GO [AR]
GOSR2	EPILEPSY. PROGRESSIVE MYOCLONIC. 6; EPM6 [AR]
GP1BA	BERNARD-SOULIER SYNDROME; BSS [AR] | NONARTERITIC ANTERIOR ISCHEMIC OPTIC NEUROPATHY. SUSCEPTIBILITY TO [AR] | PSEUDO-VON WILLEBRAND DISEASE; VWDP [AD] | BERNARD-SOULIER SYNDROME. TYPE A2. AUTOSOMAL DOMINANT; BSSA2 [AD]
GP1BB	BERNARD-SOULIER SYNDROME; BSS [AR]
GP6	BLEEDING DISORDER. PLATELET-TYPE. 11; BDPLT11 [AR]
GP9	BERNARD-SOULIER SYNDROME; BSS [AR]
GPC6	OMODYSPLASIA 1; OMOD1 [AR]
GPD1	HYPERTRIGLYCERIDEMIA. TRANSIENT INFANTILE; HTGTI [AR]
GPHN	MOLYBDENUM COFACTOR DEFICIENCY. COMPLEMENTATION GROUP C; MOCODC [AR]
GPI	HEMOLYTIC ANEMIA. NONSPHEROCYTIC. DUE TO GLUCOSE PHOSPHATE ISOMERASE DEFICIENCY [AR]
GPR143	ALBINISM. OCULAR. TYPE I; OA1 [X-linked] | NYSTAGMUS 6. CONGENITAL. X-LINKED; NYS6 [XLR]
GPR179	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1E; CSNB1E [AR]
GPSM2	CHUDLEY-MCCULLOUGH SYNDROME; CMCS [AR]
GRHPR	HYPEROXALURIA. PRIMARY. TYPE II; HP2 [AR]
GRIK2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 6; MRT6 [AR]
GRM1	SPINOCEREBELLAR ATAXIA 44; SCA44 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 13; SCAR13 [AR]
GRM6	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1B; CSNB1B [AR]
GRN	CEROID LIPOFUSCINOSIS. NEURONAL. 11; CLN11 [AR] | FRONTOTEMPORAL LOBAR DEGENERATION WITH TDP43 INCLUSIONS. GRN-RELATED [AD]
GRXCR1	DEAFNESS. AUTOSOMAL RECESSIVE 25; DFNB25 [AR]
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY; GSSD [AR] | GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES. HEMOLYTIC ANEMIA DUE TO; GSSDE [AR]
GTF2H5	Trichothiodystrophy 3 photosensitive 616395 (3)
GUCY2D	CHOROIDAL DYSTROPHY. CENTRAL AREOLAR. 1; CACD1 [AD] | LEBER CONGENITAL AMAUROSIS 1; LCA1 [AR] | CONE-ROD DYSTROPHY 6; CORD6 [AD] | NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE1I; CSNB1I [AR]
GUSB	MUCOPOLYSACCHARIDOSIS. TYPE VII; MPS7 [AR]
GYG1	GLYCOGEN STORAGE DISEASE XV; GSD15 [AR] | POLYGLUCOSAN BODY MYOPATHY 2; PGBM2 [AR]
GYS1	GLYCOGEN STORAGE DISEASE 0. MUSCLE; GSD0B [AR]
GYS2	GLYCOGEN STORAGE DISEASE 0. LIVER; GSD0A [AR]
HADH	HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 4; HHF4 [AR] | 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY [AR]
HADHA	MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY; MTPD [AR]
HADHB	MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY; MTPD [AR]
HAMP	HEMOCHROMATOSIS. TYPE 2B; HFE2B [AR]
HAX1	NEUTROPENIA. SEVERE CONGENITAL. 3. AUTOSOMAL RECESSIVE; SCN3 [AR]
HBA1	HEINZ BODY ANEMIAS [AD]
HBB	HEINZ BODY ANEMIAS [AD] | SICKLE CELL ANEMIA [AR] | FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 [AD]

HEPACAM	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 2B. REMITTING. WITH OR WITHOUT MENTAL RETARDATION; MLC2B [AD] | MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 2A; MLC2A [AR]
HERC2	SKIN/HAIR/EYE PIGMENTATION. VARIATION IN. 1; SHEP1 [AR] | MENTAL RETARDATION. AUTOSOMAL RECESSIVE 38; MRT38 [AR]
HES7	SPONDYLOCOSTAL DYSOSTOSIS 4. AUTOSOMAL RECESSIVE; SCDO4 [AR]
HESX1	SEPTOOPTIC DYSPLASIA [AD]
HEXA	TAY-SACHS DISEASE; TSD [AR]
HEXB	SANDHOFF DISEASE [Autosomal recessive with multiple alleles and compound]
HFE	VARIEGATE PORPHYRIA; VP [AD] | PORPHYRIA CUTANEA TARDA [AD] | ALZHEIMER DISEASE; AD [AD] | HEMOCHROMATOSIS. TYPE 1; HFE1 [AR]
HGD	ALKAPTONURIA; AKU [AR]
HGF	DEAFNESS. AUTOSOMAL RECESSIVE 39; DFNB39 [AR]
HGSNAT	RETINITIS PIGMENTOSA 73; RP73 [AR] | MUCOPOLYSACCHARIDOSIS. TYPE IIIC; MPS3C [AR]
HIBCH	3-HYDROXYISOBUTYRYL-CoA HYDROLASE DEFICIENCY; HIBCHD [AR]
HINT1	NEUROMYOTONIA AND AXONAL NEUROPATHY. AUTOSOMAL RECESSIVE; NMAN [AR]
HK1	HEMOLYTIC ANEMIA. NONSPHEROCYTIC. DUE TO HEXOKINASE DEFICIENCY [AR] | NEURODEVELOPMENTAL DISORDER WITH VISUAL DEFECTS AND BRAIN ANOMALIES; NEDVIBA [AD] | NEUROPATHY. HEREDITARY MOTOR AND SENSORY. RUSSE TYPE; HMSNR [AR] | RETINITIS PIGMENTOSA 79; RP79 [AD]
HLCS	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY [AR]
HMBS	PORPHYRIA. ACUTE INTERMITTENT; AIP [AD]
HMGCL	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY; HMGCLD [AR]
HMOX1	Heme oxygenase-1 deficiency 614034 (3) | {Pulmonary disease chronic obstructive susceptibility to} 606963 (3)
HMX1	OCULOAURICULAR SYNDROME; OCACS [AR]
HOXA1	Athabaskan brainstem dysgenesis syndrome 601536 (3) | Bosley-Salih-Alorainy syndrome 601536 (3)
HOXA2	MICROTIA. HEARING IMPAIRMENT. AND CLEFT PALATE [AD]
HPD	TYROSINEMIA. TYPE III; TYRSN3 [AR] | HAWKINSINURIA [AD]
HPGD	HYPERTROPHIC OSTEOARTHROPATHY. PRIMARY. AUTOSOMAL RECESSIVE. 1; PHOAR1 [AR] | DIGITAL CLUBBING. ISOLATED CONGENITAL [AR]
HPRT1	LESCH-NYHAN SYNDROME; LNS [XLR] | KELLEY-SEEGMILLER SYNDROME [XLR]
HPS1	HERMANSKY-PUDLAK SYNDROME 1; HPS1 [AR]
HPS3	HERMANSKY-PUDLAK SYNDROME 3; HPS3 [AR]
HPS6	HERMANSKY-PUDLAK SYNDROME 6; HPS6 [AR]
HPSE2	UROFACIAL SYNDROME 1; UFS1 [AR]
HR	Alopecia universalis 203655 (3) | Atrichia with papular lesions 209500 (3) | Hypotrichosis 4 146550 (3)
HSD11B2	APPARENT MINERALOCORTICOID EXCESS; AME [AR]
HSD17B10	HSD10 MITOCHONDRIAL DISEASE; HSD10MD [XLD]
HSD17B3	17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY [AR]
HSD17B4	D-BIFUNCTIONAL PROTEIN DEFICIENCY [AR] | PERRAULT SYNDROME 1; PRLTS1 [AR]
HSD3B7	BILE ACID SYNTHESIS DEFECT. CONGENITAL. 1; CBAS1 [AR]
HSPB1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2F; CMT2F [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE IIB; HMN2B [AD]
HSPD1	SPASTIC PARAPLEGIA 13. AUTOSOMAL DOMINANT; SPG13 [AD] | LEUKODYSTROPHY. HYPOMYELINATING. 4; HLD4 [AR]
HSPG2	SCHWARTZ-JAMPEL SYNDROME. TYPE 1; SJS1 [AR] | DYSSEGMENTAL DYSPLASIA. SILVERMAN-HANDMAKER TYPE; DDSH [AR]
HTRA1	CEREBRAL ARTERIOPATHY. AUTOSOMAL RECESSIVE. WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY; CARASIL [AR] | CEREBRAL ARTERIOPATHY. AUTOSOMAL DOMINANT. WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY. TYPE 2; CADASIL2 [AD]
HYDIN	CILIARY DYSKINESIA. PRIMARY. 5; CILD5 [AR]
HYLS1	HYDROLETHALUS SYNDROME 1; HLS1 [AR]

ICOS	IMMUNODEFICIENCY. COMMON VARIABLE. 1; CVID1 [AR]
IDH3B	Retinitis pigmentosa 46 612572 (3)
IDS	MUCOPOLYSACCHARIDOSIS. TYPE II; MPS2 [XLR]
IDUA	SCHEIE SYNDROME [AR] | HURLER SYNDROME [AR] | HURLER-SCHEIE SYNDROME [AR]
IER3IP1	MICROCEPHALY. EPILEPSY. AND DIABETES SYNDROME; MEDS [AR]
IFNGR1	IMMUNODEFICIENCY 27A; IMD27A [AR] | HELICOBACTER PYLORI INFECTION. SUSCEPTIBILITY TO [Not determine] | IMMUNODEFICIENCY 27B; IMD27B [AD]
IFT122	CRANIOECTODERMAL DYSPLASIA 1; CED1 [AR]
IFT43	CRANIOECTODERMAL DYSPLASIA 3; CED3 [AR] | RETINITIS PIGMENTOSA 81; RP81 [AR] | SHORT-RIB THORACIC DYSPLASIA 18 WITH POLYDACTYLY; SRTD18 [AR]
IFT80	SHORT-RIB THORACIC DYSPLASIA 2 WITH OR WITHOUT POLYDACTYLY; SRTD2 [AR]
IGBP1	CORPUS CALLOSUM. AGENESIS OF. WITH MENTAL RETARDATION. OCULAR COLOBOMA. AND MICROGNATHIA [XLR]
IGF1	INSULIN-LIKE GROWTH FACTOR I DEFICIENCY [AR]
IGF1R	INSULIN-LIKE GROWTH FACTOR I. RESISTANCE TO; IGF1RES [AD]
IGFBP7	RETINAL ARTERIAL MACROANEURYSM WITH SUPRAVALVULAR PULMONIC STENOSIS; RAMSVPS [AR]
IGHG2	IgG2 deficiency selective (3)
IGHM	AGAMMAGLOBULINEMIA 1. AUTOSOMAL RECESSIVE; AGM1 [AR]
IGHMBP2	SPINAL MUSCULAR ATROPHY. DISTAL. AUTOSOMAL RECESSIVE. 1; DSMA1 [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2S; CMT2S [AR]
IGLL1	AGAMMAGLOBULINEMIA 2. AUTOSOMAL RECESSIVE; AGM2 [AR]
IHH	BRACHYDACTYLY. TYPE A1; BDA1 [AD] | ACROCAPITOFEMORAL DYSPLASIA; ACFD [AR]
IKBKG	INCONTINENTIA PIGMENTI; IP [XLD] | IMMUNODEFICIENCY 33; IMD33 [XLR]
IL10RA	INFLAMMATORY BOWEL DISEASE 28. AUTOSOMAL RECESSIVE; IBD28 [AR]
IL11RA	CRANIOSYNOSTOSIS AND DENTAL ANOMALIES; CRSDA [AR]
IL12B	IMMUNODEFICIENCY 29; IMD29 [AR]
IL1RAPL1	MENTAL RETARDATION. X-LINKED 21; MRX21 [XLR]
IL1RN	OSTEOMYELITIS. STERILE MULTIFOCAL. WITH PERIOSTITIS AND PUSTULOSIS; OMPP [AR] | GASTRIC CANCER. HEREDITARY DIFFUSE; HDGC [AD]
IL2RG	SEVERE COMBINED IMMUNODEFICIENCY. X-LINKED; SCIDX1 [XLR] | COMBINED IMMUNODEFICIENCY. X-LINKED; CIDX [XLR]
IL36RN	PSORIASIS 14. PUSTULAR; PSORS14 [AR]
IL7R	SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-POSITIVE [AR]
ILDR1	DEAFNESS. AUTOSOMAL RECESSIVE 42; DFNB42 [AR]
IMPAD1	CHONDRODYSPLASIA WITH JOINT DISLOCATIONS, GPAPP TYPE [AR]
IMPDH1	RETINITIS PIGMENTOSA 10; RP10 [AD] | LEBER CONGENITAL AMAUROSIS 11; LCA11 [AD]
IMPG2	MACULAR DYSTROPHY. VITELLIFORM. 5; VMD5 [AD] | RETINITIS PIGMENTOSA 56; RP56 [AR]
ING1	SQUAMOUS CELL CARCINOMA. HEAD AND NECK; HNSCC [AR]
INPP5E	MENTAL RETARDATION. TRUNCAL OBESITY. RETINAL DYSTROPHY. AND MICROPENIS SYNDROME; MORMS [AR] | JOUBERT SYNDROME 1; JBTS1 [AR]
INSR	DONOHUE SYNDROME [AR] | PINEAL HYPERPLASIA. INSULIN-RESISTANT DIABETES MELLITUS. AND SOMATIC ABNORMALITIES [AR] | HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 5; HHF5 [AD]
INVS	NEPHRONOPHTHISIS 2; NPHP2 [AR]
IQCB1	SENIOR-LOKEN SYNDROME 5; SLSN5 [AR]
IRAK4	IRAK4 deficiency 607676 (3) | Invasive pneumococcal disease recurrent isolated 1 610799 (3)
IRF1	LUNG CANCER [Somatic mutation]
IRF8	IMMUNODEFICIENCY 32A; IMD32A [AD] | IMMUNODEFICIENCY 32B; IMD32B [AR]
IRX5	HAMAMY SYNDROME; HMMS [AR]
ISCU	MYOPATHY WITH LACTIC ACIDOSIS. HEREDITARY; HML [AR]

ITCH	AUTOIMMUNE DISEASE. MULTISYSTEM. WITH FACIAL DYSMORPHISM; ADMFD [AR]
ITGA2B	BLEEDING DISORDER. PLATELET-TYPE. 16; BDPLT16 [AD] | GLANZMANN THROMBASTHENIA; GT [AR]
ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA [AR]
ITGA7	MUSCULAR DYSTROPHY. CONGENITAL. DUE TO INTEGRIN ALPHA-7 DEFICIENCY [AR]
ITGB2	LEUKOCYTE ADHESION DEFICIENCY. TYPE I; LAD [AR]
ITGB3	INTEGRIN. BETA-3; ITGB3 [AR] | BLEEDING DISORDER. PLATELET-TYPE. 16; BDPLT16 [AD] | GLANZMANN THROMBASTHENIA; GT [AR]
ITGB4	EPIDERMOLYSIS BULLOSA SIMPLEX. LOCALIZED [AD] | EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA [AR] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR]
ITK	LYMPHOPROLIFERATIVE SYNDROME 1; LPFS1 [AR]
IVD	ISOVALERIC ACIDEMIA; IVA [AR]
IYD	THYROID DYSHORMONOGENESIS 4; TDH4 [AR]
JAK2	LEUKEMIA. ACUTE MYELOID; AML [AD] | ERYTHROCYTOSIS. FAMILIAL. 1; ECYT1 [AD] | POLYCYTHEMIA VERA; PV [Somatic mutation] | MYELOFIBROSIS [Somatic mutation] | THROMBOCYTHEMIA 3; THCYT3 [AD] | BUDD-CHIARI SYNDROME; BDCHS [AR]
JAK3	SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-NEGATIVE [AR]
JAM3	HEMORRHAGIC DESTRUCTION OF THE BRAIN. SUBEPENDYMAL CALCIFICATION. AND CATARACTS; HDBSCC [AR]
JUP	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 12; ARVD12 [AD] | NAXOS DISEASE; NXD [AR]
KAT6B	OHDO SYNDROME. SBBYS VARIANT; SBBYSS [AD] | GENITOPATELLAR SYNDROME; GTPTS [AD]
KCNJ1	ATRIAL FIBRILLATION. FAMILIAL. 9; ATFB9 [AD] | BARTTER SYNDROME. TYPE 2. ANTENATAL; BARTS2 [AR]
KCNJ10	DEAFNESS. AUTOSOMAL RECESSIVE 4. WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4 [AR] | SEIZURES. SENSORINEURAL DEAFNESS. ATAXIA. MENTAL RETARDATION. AND ELECTROLYTE IMBALANCE; SESAMES [AR]
KCNJ11	DIABETES MELLITUS. PERMANENT NEONATAL; PNDM [AD] | HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 2; HHF2 [AR] | MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 13; MODY13 [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | DIABETES MELLITUS. TRANSIENT NEONATAL. 3 [AD]
KCNJ13	VITREORETINAL DEGENERATION. SNOWFLAKE TYPE; SVD [AD] | LEBER CONGENITAL AMAUROSIS 16; LCA16 [AR]
KCNQ1	SHORT QT SYNDROME 2; SQT2 [AD] | ATRIAL FIBRILLATION. FAMILIAL. 3; ATFB3 [AD] | LONG QT SYNDROME 1; LQT1 [AD] | JERVELL AND LANGE-NIELSEN SYNDROME 1; JLNS1 [AR]
KCNV2	RETINAL CONE DYSTROPHY 3B; RCD3B [AR]
KCTD7	EPILEPSY. PROGRESSIVE MYOCLONIC. 3. WITH OR WITHOUT INTRACELLULAR INCLUSIONS; EPM3 [AR]
KDM5C	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CLAES-JENSEN TYPE; MRXSCJ [XLR]
KERA	CORNEA PLANA 2. AUTOSOMAL RECESSIVE; CNA2 [AR]
KHDC3L	HYDATIDIFORM MOLE. RECURRENT. 2; HYDM2 [AR]
KIF1A	NESCAV SYNDROME; NESCAVS [AD] | NEUROPATHY. HEREDITARY SENSORY. TYPE IIC; HSN2C [AR] | SPASTIC PARAPLEGIA 30. AUTOSOMAL RECESSIVE; SPG30 [AR]
KIF7	AL-GAZALI-BAKALINOVA SYNDROME; AGBK [AR] | ACROCALLOSAL SYNDROME; ACLS [AR] | HYDROLETHALUS SYNDROME 2; HLS2 [AR]
KISS1R	HYPOGONADOTROPIC HYPOGONADISM 8 WITH OR WITHOUT ANOSMIA; HH8 [AR] | PRECOCIOUS PUBERTY. CENTRAL. 1; CPPB1 [AD]
KLHL3	PSEUDOHYPOALDOSTERONISM. TYPE IID; PHA2D [AD]
KLHL40	NEMALINE MYOPATHY 8; NEM8 [AR]
KLK4	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA1; AI2A1 [AR]
KLKB1	PREKALLIKREIN DEFICIENCY [AR]
KRAS	LEUKEMIA. ACUTE MYELOID; AML [AD] | BLADDER CANCER [Somatic mutation] | SCHIMMELPENNING-FEUERSTEIN-MIMS SYNDROME; SFM [Somatic mosaicism] | JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML [AD] | PANCREATIC CANCER [AD] | OCULOECTODERMAL SYNDROME; OES [Somatic mutation] | CARDIOFACIOCUTANEOUS SYNDROME 2; CFC2 [AD] | ARTERIOVENOUS MALFORMATIONS OF THE BRAIN [Somatic mutation] | RAS-ASSOCIATED AUTOIMMUNE LEUKOPROLIFERATIVE DISORDER; RALD [AD] | NOONAN SYNDROME 3; NS3 [AD] | GASTRIC CANCER. HEREDITARY DIFFUSE; HDGC [AD] | BREAST CANCER [AD] | LUNG CANCER [Somatic mutation]
KRT14	NAEGELI-FRANCESCHETTI-JADASSOHN SYNDROME; NFJS [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. LOCALIZED [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. GENERALIZED [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. DOWLING-MEARA TYPE; EBSDM [AD] | DERMATOPATHIA PIGMENTOSA RETICULARIS; DPR [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. AUTOSOMAL RECESSIVE 1; EBSB1 [AR]
KRT18	CIRRHOSIS. FAMILIAL [Autosomal recessive cases, probably heterogeneou]
KRT5	DOWLING-DEGOS DISEASE 1; DDD1 [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. LOCALIZED [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. GENERALIZED [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX WITH MOTTLED PIGMENTATION; EBSMP [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. DOWLING-MEARA TYPE; EBSDM [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX. AUTOSOMAL RECESSIVE 1; EBSB1 [AR]

KRT8	CIRRHOSIS. FAMILIAL [Autosomal recessive cases, probably heterogeneou]
L1CAM	MASA SYNDROME [XLR] | HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; HSAS [XLR] | CORPUS CALLOSUM. PARTIAL AGENESIS OF. X-LINKED [XLR]
L2HGDH	L-2-HYDROXYGLUTARIC ACIDURIA; L2HGA [AR]
LAMA2	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 23; LGMDR23 [AR] | MUSCULAR DYSTROPHY. CONGENITAL MEROSIN-DEFICIENT. 1A; MDC1A [AR]
LAMA3	LARYNGOONYCHOCUTANEOUS SYNDROME; LOCS [AR] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. HERLITZ TYPE [AR] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR]
LAMB2	PIERSON SYNDROME [AR]
LAMB3	EPIDERMOLYSIS BULLOSA. JUNCTIONAL. HERLITZ TYPE [AR] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR] | AMELOGENESIS IMPERFECTA. TYPE IA; AI1A [AD]
LAMC2	EPIDERMOLYSIS BULLOSA. JUNCTIONAL. HERLITZ TYPE [AR] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR]
LAMC3	CORTICAL MALFORMATIONS. OCCIPITAL; OCCM [AR]
LAMTOR2	IMMUNODEFICIENCY DUE TO DEFECT IN MAPBP-INTERACTING PROTEIN [AR]
LARP7	ALAZAMI SYNDROME; ALAZS [AR]
LBR	REYNOLDS SYNDROME [AD] | GREENBERG DYSPLASIA; GRBGD [AR] | PELGER-HUET ANOMALY; PHA [AD]
LCA5	LEBER CONGENITAL AMAUROSIS 5; LCA5 [AR]
LCAT	FISH-EYE DISEASE; FED [AR] | LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY [AR]
LCT	LACTASE DEFICIENCY. CONGENITAL [AR]
LDHA	GLYCOGEN STORAGE DISEASE XI; GSD11 [AR]
LDLR	HYPERCHOLESTEROLEMIA. FAMILIAL. 1; FHCL1 [AD]
LDLRAP1	HYPERCHOLESTEROLEMIA. FAMILIAL. 4; FHCL4 [AR]
LEPR	LEPTIN RECEPTOR DEFICIENCY; LEPRD [AR]
LFNG	SPONDYLOCOSTAL DYSOSTOSIS 3. AUTOSOMAL RECESSIVE; SCDO3 [AR]
LHCGR	LEYDIG CELL HYPOPLASIA. TYPE I [AR] | PRECOCIOUS PUBERTY. MALE-LIMITED [Sex-limited autosomal dominant]
LHFPL5	DEAFNESS. AUTOSOMAL RECESSIVE 67; DFNB67 [AR]
LHX3	PITUITARY HORMONE DEFICIENCY. COMBINED. 3; CPHD3 [AR]
LIAS	HYPERGLYCINEMIA. LACTIC ACIDOSIS. AND SEIZURES; HGCLAS [AR]
LIFR	STUVE-WIEDEMANN SYNDROME; STWS [AR]
LIG4	LIG4 SYNDROME [AR] | MYELOMA. MULTIPLE [Somatic mutation]
LIM2	CATARACT 19. MULTIPLE TYPES; CTRCT19 [AR]
LIPA	LYSOSOMAL ACID LIPASE DEFICIENCY [AR]
LIPH	HYPOTRICHOSIS 7; HYPT7 [AR]
LIPN	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 8; ARCI8 [AR]
LMAN1	FACTOR V AND FACTOR VIII. COMBINED DEFICIENCY OF. 1; F5F8D1 [AR]
LMBR1	ACHEIROPODY; ACHP [AR] | LAURIN-SANDROW SYNDROME; LSS [AD] | POLYDACTYLY. PREAXIAL II; PPD2 [AD] | SYNDACTYLY. TYPE IV; SDTY4 [AD] | TIBIA. HYPOPLASIA OR APLASIA OF. WITH POLYDACTYLY; THYP [AD]
LMBRD1	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA. cblF TYPE; MAHCF [AR]
LMF1	LIPASE DEFICIENCY. COMBINED [AR]
LMNA	HUTCHINSON-GILFORD PROGERIA SYNDROME; HGPS [AD] | MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2B1; CMT2B1 [AR] | EMERY-DREIFUSS MUSCULAR DYSTROPHY 2. AUTOSOMAL DOMINANT; EDMD2 [AD] | EMERY-DREIFUSS MUSCULAR DYSTROPHY 3. AUTOSOMAL RECESSIVE; EDMD3 [AR] | MUSCULAR DYSTROPHY. CONGENITAL. LMNA-RELATED [AD] | RESTRICTIVE DERMOPATHY. LETHAL [AR] | CARDIOMYOPATHY. DILATED. 1A; CMD1A [AD] | HEART-HAND SYNDROME. SLOVENIAN TYPE [AD] | LIPODYSTROPHY. FAMILIAL PARTIAL. TYPE 2; FPLD2 [AD] | CARDIOMYOPATHY. DILATED. WITH HYPERGONADOTROPIC HYPOGONADISM [AD]
LOXHD1	DEAFNESS. AUTOSOMAL RECESSIVE 77; DFNB77 [AR]
LPAR6	HYPOTRICHOSIS 8; HYPT8 [AR]
LPIN1	MYOGLOBINURIA. ACUTE RECURRENT. AUTOSOMAL RECESSIVE [AR]

LPIN2	Majeed syndrome 609628 (3)
LPL	HYPERLIPOPROTEINEMIA. TYPE I [AR] | HYPERLIPIDEMIA. FAMILIAL COMBINED. 3; FCHL3 [AD]
LRAT	LEBER CONGENITAL AMAUROSIS 14; LCA14 [AR]
LRBA	IMMUNODEFICIENCY. COMMON VARIABLE. 8. WITH AUTOIMMUNITY; CVID8 [AR]
LRIT3	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1F; CSNB1F [AR]
LRP2	DONNAI-BARROW SYNDROME [AR]
LRP4	SCLEROSTEOSIS 2; SOST2 [AD] | MYASTHENIC SYNDROME. CONGENITAL. 17; CMS17 [AR] | CENANI-LENZ SYNDACTYLY SYNDROME; CLSS [AR]
LRP5	EXUDATIVE VITREORETINOPATHY 4; EVR4 [AD] | OSTEOPOROSIS [AD] | BONE MINERAL DENSITY QUANTITATIVE TRAIT LOCUS 1; BMND1 [AD] | POLYCYSTIC LIVER DISEASE 4 WITH OR WITHOUT KIDNEY CYSTS; PCLD4 [AD] | OSTEOPETROSIS. AUTOSOMAL DOMINANT 1; OPTA1 [AD] | VAN BUCHEM DISEASE. TYPE 2 [AD] | OSTEOPOROSIS-PSEUDOGLIOMA SYNDROME; OPPG [AR] | ENDOSTEAL HYPEROSTOSIS. AUTOSOMAL DOMINANT [AD]
LRPAP1	MYOPIA 23. AUTOSOMAL RECESSIVE; MYP23 [AR]
LRPPRC	LEIGH SYNDROME. FRENCH CANADIAN TYPE; LSFC [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
LRSAM1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2P; CMT2P [AD]
LRTOMT	DEAFNESS. AUTOSOMAL RECESSIVE 63; DFNB63 [AR]
LTBP2	MICROSPHEROPHAKIA AND/OR MEGALOCORNEA. WITH ECTOPIA LENTIS AND WITH OR WITHOUT SECONDARY GLAUCOMA; MSPKA [AR] | WEILL-MARCHESANI SYNDROME 3; WMS3 [AR]
LTBP3	GELEOPHYSIC DYSPLASIA 3; GPHYSD3 [AD] | DENTAL ANOMALIES AND SHORT STATURE; DASS [AR]
LTBP4	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IC; ARCL1C [AR]
LYST	CHEDIAK-HIGASHI SYNDROME; CHS [AR]
MAK	RETINITIS PIGMENTOSA 62; RP62 [AR]
MAMLD1	HYPOSPADIAS 2. X-LINKED; HYSP2 [XLR]
MAN1B1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 15; MRT15 [AR]
MAN2B1	MANNOSIDOSIS. ALPHA B. LYSOSOMAL; MANSA [AR]
MANBA	MANNOSIDOSIS. BETA A. LYSOSOMAL; MANSB [AR]
MAP3K8	LUNG CANCER [Somatic mutation]
MAPT	FRONTOTEMPORAL DEMENTIA; FTD [AD] | PARKINSON DISEASE. LATE-ONSET; PD [AD] | PARKINSON-DEMENTIA SYNDROME [Autosomal recessive vs. dominant with low penetrance and expressio] | SUPRANUCLEAR PALSY. PROGRESSIVE. 1; PSNP1 [AD] | PICK DISEASE OF BRAIN [AD]
MARVELD2	DEAFNESS. AUTOSOMAL RECESSIVE 49; DFNB49 [AR]
MASP1	3MC SYNDROME 1; 3MC1 [AR]
MAT1A	METHIONINE ADENOSYLTRANSFERASE I/III DEFICIENCY [AD]
MATN3	OSTEOARTHRITIS SUSCEPTIBILITY 2; OS2 [Sex-influenced autosomal dominan] | OSTEOARTHRITIS SUSCEPTIBILITY 3; OS3 [AD] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. MATRILIN-3 RELATED [AR] | EPIPHYSEAL DYSPLASIA. MULTIPLE. 5; EDM5 [AD]
MBTPS2	OSTEOGENESIS IMPERFECTA. TYPE XIX; OI19 [XLR] | KERATOSIS FOLLICULARIS SPINULOSA DECALVANS. X-LINKED; KFSDX [XLR] | PALMOPLANTAR KERATODERMA. MUTILATING. WITH PERIORIFICIAL KERATOTIC PLAQUES. X-LINKED [XLR] | IFAP SYNDROME WITH OR WITHOUT BRESHECK SYNDROME [XLR]
MC2R	GLUCOCORTICOID DEFICIENCY 1; GCCD1 [AR]
MC4R	BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 20; BMIQ20 [AD] | OBESITY [AD]
MCCC1	3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY; MCC1D [AR]
MCCC2	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY; MCC2D [AR]
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY [AR]
MCM4	IMMUNODEFICIENCY 54; IMD54 [AR]
MCM6	LACTOSE INTOLERANCE. ADULT TYPE [AD]
MCOLN1	MUCOLIPIDOSIS IV; ML4 [AR]
MCPH1	MICROCEPHALY 1. PRIMARY. AUTOSOMAL RECESSIVE; MCPH1 [AR]

MECP2	ENCEPHALOPATHY. NEONATAL SEVERE. DUE TO MECP2 MUTATIONS [XLR] | AUTISM. SUSCEPTIBILITY TO. X-LINKED 3; AUTSX3 [X-linked] | MENTAL RETARDATION. X-LINKED. SYNDROMIC 13; MRXS13 [XLR] | LUBS X-LINKED MENTAL RETARDATION SYNDROME; MRXSL [XLR] | RETT SYNDROME; RTT [XLD]
MED12	OPITZ-KAVEGGIA SYNDROME; OKS [XLR] | INTELLECTUAL DEVELOPMENTAL DISORDER. X-LINKED. SYNDROMIC. LUJAN-FRYNS TYPE; MRXSLF [XLR] | OHDO SYNDROME. X-LINKED; OHDOX [XLR]
MED23	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 18; MRT18 [AR]
MED25	BASEL-VANAGAITE-SMIRIN-YOSEF SYNDROME; BVSYS [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2B2; CMT2B2 [AR]
MEFV	FAMILIAL MEDITERRANEAN FEVER. AUTOSOMAL DOMINANT [AD] | FAMILIAL MEDITERRANEAN FEVER; FMF [AR]
MEGF10	MYOPATHY. AREFLEXIA. RESPIRATORY DISTRESS. AND DYSPHAGIA. EARLY-ONSET; EMARDD [AR]
MEGF8	CARPENTER SYNDROME 2; CRPT2 [AR]
MERTK	RETINITIS PIGMENTOSA 38; RP38 [AR]
MESP2	SPONDYLOCOSTAL DYSOSTOSIS 2. AUTOSOMAL RECESSIVE; SCDO2 [AR]
MFN2	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. AUTOSOMAL DOMINANT. TYPE 2A2A; CMT2A2A [AD] | NEUROPATHY. HEREDITARY MOTOR AND SENSORY. TYPE VIA. WITH OPTIC ATROPHY; HMSN6A [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. AUTOSOMAL RECESSIVE. TYPE 2A2B; CMT2A2B [AR]
MFRP	MICROPHTHALMIA. ISOLATED 5; MCOP5 [AR]
MFSD8	CEROID LIPOFUSCINOSIS. NEURONAL. 7; CLN7 [AR] | MACULAR DYSTROPHY WITH CENTRAL CONE INVOLVEMENT; CCMD [AR]
MGAT2	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIa; CDG2A [AR]
MGP	KEUTEL SYNDROME; KTLS [AR]
MID1	OPITZ GBBB SYNDROME. TYPE I; GBBB1 [XLR]
MKKS	BARDET-BIEDL SYNDROME 6; BBS6 [AR] | MCKUSICK-KAUFMAN SYNDROME; MKKS [AR]
MKS1	JOUBERT SYNDROME 28; JBTS28 [AR] | BARDET-BIEDL SYNDROME 13; BBS13 [AR] | MECKEL SYNDROME. TYPE 1; MKS1 [AR]
MLC1	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 1; MLC1 [AR]
MLH1	MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR] | MUIR-TORRE SYNDROME; MRTES [AD]
MLPH	GRISCELLI SYNDROME. TYPE 3; GS3 [AR]
MLYCD	MALONYL-CoA DECARBOXYLASE DEFICIENCY [AR]
MMAA	METHYLMALONIC ACIDURIA. cblA TYPE [AR]
MMAB	METHYLMALONIC ACIDURIA. cblB TYPE [AR]
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA. cblC TYPE; MAHCC [AR]
MMADHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA. cblD TYPE; MAHCD [AR]
MMP1	EPIDERMOLYSIS BULLOSA DYSTROPHICA. AUTOSOMAL RECESSIVE; RDEB [AR]
MMP13	METAPHYSEAL DYSPLASIA. SPAHR TYPE; MDST [AR] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. MISSOURI TYPE [AD]
MMP2	MULTICENTRIC OSTEOLYSIS. NODULOSIS. AND ARTHROPATHY; MONA [AR]
MMP20	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA2; AI2A2 [AR]
MMP9	Metaphyseal anadysplasia 2 613073 (3)
MOCOS	XANTHINURIA. TYPE II; XAN2 [AR]
MOCS1	MOLYBDENUM COFACTOR DEFICIENCY. COMPLEMENTATION GROUP A; MOCODA [AR]
MOCS2	MOLYBDENUM COFACTOR DEFICIENCY. COMPLEMENTATION GROUP B; MOCODB [AR]
MOGS	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIb; CDG2B [AR]
MPDZ	HYDROCEPHALUS. CONGENITAL. 2. WITH OR WITHOUT BRAIN OR EYE ANOMALIES; HYC2 [AR]
MPI	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ib; CDG1B [AR]
MPL	THROMBOCYTHEMIA 2; THCYT2 [AD] | MYELOFIBROSIS [Somatic mutation] | AMEGAKARYOCYTIC THROMBOCYTOPENIA. CONGENITAL; CAMT [AR]
MPLKIP	TRICHOTHIODYSTROPHY 4. NONPHOTOSENSITIVE; TTD4 [AR]
MPV17	MITOCHONDRIAL DNA DEPLETION SYNDROME 6 (HEPATOCEREBRAL TYPE); MTDPS6 [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2EE; CMT2EE [AR]

MPZ	CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1B; CMT1B [AD] | ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2I; CMT2I [AD] | HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2J; CMT2J [AD] | NEUROPATHY. CONGENITAL HYPOMYELINATING. 2; CHN2 [AD] | CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE D; CMTDID [AD]
MRAP	GLUCOCORTICOID DEFICIENCY 2; GCCD2 [AR]
MRPS16	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2; COXPD2 [AR]
MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5; COXPD5 [AR] | OVARIAN DYSGENESIS 7; ODG7 [AR]
MS4A1	IMMUNODEFICIENCY. COMMON VARIABLE. 5; CVID5 [AR]
MSH6	COLORECTAL CANCER. HEREDITARY NONPOLYPOSIS. TYPE 5; HNPCC5 [AD] | ENDOMETRIAL CANCER [Somatic mutation] | MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR]
MSRB3	DEAFNESS. AUTOSOMAL RECESSIVE 74; DFNB74 [AR]
MSTN	Muscle hypertrophy 614160 (3)
MTHFR	NEURAL TUBE DEFECTS. FOLATE-SENSITIVE; NTDFS [AR] | THROMBOPHILIA DUE TO THROMBIN DEFECT; THPH1 [AD] | HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5.10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY [AR] | SCHIZOPHRENIA; SCZD [AD]
MTM1	MYOPATHY. CENTRONUCLEAR. X-LINKED; CNMX [XLR]
MTMR2	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4B1; CMT4B1 [AR]
MTR	NEURAL TUBE DEFECTS. FOLATE-SENSITIVE; NTDFS [AR] | HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA. cblG COMPLEMENTATION TYPE; HMAG [AR]
MTRR	HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA. cblE COMPLEMENTATION TYPE; HMAE [AR] | NEURAL TUBE DEFECTS. FOLATE-SENSITIVE; NTDFS [AR]
MTTP	ABDOMINAL OBESITY-METABOLIC SYNDROME 1; AOMS1 [AD] | ABETALIPOPROTEINEMIA; ABL [AR]
MUSK	MYASTHENIC SYNDROME. CONGENITAL. 9. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS9 [AR] | FETAL AKINESIA DEFORMATION SEQUENCE 1; FADS1 [AR]
MUTYH	FAMILIAL ADENOMATOUS POLYPOSIS 2; FAP2 [AR]
MVK	HYPER-IgD SYNDROME; HIDS [AR] | MEVALONIC ACIDURIA; MEVA [AR] | POROKERATOSIS 3. MULTIPLE TYPES; POROK3 [AD]
MYBPC1	ARTHROGRYPOSIS. DISTAL. TYPE 1B; DA1B [AD] | LETHAL CONGENITAL CONTRACTURE SYNDROME 4; LCCS4 [AR] | MYOPATHY. CONGENITAL. WITH TREMOR; MYOTREM [AD]
MYBPC3	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 4; CMH4 [Autosomal dominant (incomplete penetrance)] | LEFT VENTRICULAR NONCOMPACTION 10; LVNC10 [AD]
MYD88	MACROGLOBULINEMIA. WALDENSTROM. SUSCEPTIBILITY TO. 1; WM1 [AD]
MYL3	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 8; CMH8 [AD]
MYO15A	DEAFNESS. AUTOSOMAL RECESSIVE 3; DFNB3 [AR]
MYO1E	FOCAL SEGMENTAL GLOMERULOSCLEROSIS 6; FSGS6 [AR]
MYO3A	DEAFNESS. AUTOSOMAL RECESSIVE 30; DFNB30 [AR]
MYO5A	ELEJALDE DISEASE [AR] | GRISCELLI SYNDROME. TYPE 1; GS1 [AR] | GRISCELLI SYNDROME. TYPE 3; GS3 [AR]
MYO5B	DIARRHEA 2. WITH MICROVILLUS ATROPHY; DIAR2 [AR]
MYO6	DEAFNESS. AUTOSOMAL RECESSIVE 37; DFNB37 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 22; DFNA22 [AD]
MYO7A	DEAFNESS. AUTOSOMAL DOMINANT 11; DFNA11 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 2; DFNB2 [AR] | USHER SYNDROME. TYPE I; USH1 [AR]
MYOC	GLAUCOMA 1. OPEN ANGLE. A; GLC1A [AD]
NAA10	MICROPHTHALMIA. SYNDROMIC 1; MCOPS1 [X-linked] | OGDEN SYNDROME; OGDNS [XLR]
NAGA	KANZAKI DISEASE [AR] | SCHINDLER DISEASE. TYPE I [AR]
NAGLU	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2V; CMT2V [AD] | MUCOPOLYSACCHARIDOSIS. TYPE IIIB; MPS3B [AR]
NAGS	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY; NAGSD [AR]
NALCN	HYPOTONIA. INFANTILE. WITH PSYCHOMOTOR RETARDATION AND CHARACTERISTIC FACIES 1; IHPRF1 [AR] | CONGENITAL CONTRACTURES OF THE LIMBS AND FACE. HYPOTONIA. AND DEVELOPMENTAL DELAY; CLIFAHDD [AD]
NBEAL2	GRAY PLATELET SYNDROME; GPS [AR]
NBN	NIJMEGEN BREAKAGE SYNDROME; NBS [AR]
NCF1	GRANULOMATOUS DISEASE. CHRONIC. AUTOSOMAL RECESSIVE. CYTOCHROME b-POSITIVE. TYPE I; CDG1 [AR]

NCF2	GRANULOMATOUS DISEASE. CHRONIC. AUTOSOMAL RECESSIVE. CYTOCHROME b-POSITIVE. TYPE II; CDG2 [AR]
NDE1	MICROHYDRANENCEPHALY; MHAC [AR] | LISSENCEPHALY 4; LIS4 [AR]
NDP	NORRIE DISEASE; ND [XLR] | EXUDATIVE VITREORETINOPATHY 2. X-LINKED; EVR2 [XLR]
NDRG1	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4D; CMT4D [AR]
NDUFA1	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 12; MC1DN12 [XLR]
NDUFA10	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 22; MC1DN22 [AR]
NDUFA11	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 14; MC1DN14 [AR]
NDUFA9	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 26; MC1DN26 [AR]
NDUFAF1	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 11; MC1DN11 [AR]
NDUFAF2	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 10; MC1DN10 [AR]
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 18; MC1DN18 [AR]
NDUFAF4	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 15; MC1DN15 [AR]
NDUFAF5	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 16; MC1DN16 [AR]
NDUFAF6	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 17; MC1DN17 [AR]
NDUFB3	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 25; MC1DN25 [AR]
NDUFS1	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 5; MC1DN5 [AR]
NDUFS2	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 6; MC1DN6 [AR]
NDUFS3	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 8; MC1DN8 [AR]
NDUFS4	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 1; MC1DN1 [AR]
NDUFS6	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 9; MC1DN9 [AR]
NDUFS7	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 3; MC1DN3 [AR] | LEIGH SYNDROME; LS [AR]
NDUFS8	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 2; MC1DN2 [AR]
NDUFV1	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 4; MC1DN4 [AR]
NDUFV2	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 7; MC1DN7 [AR]
NEB	NEMALINE MYOPATHY 2; NEM2 [AR]
NEFL	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2E; CMT2E [AD] | CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE G; CMTDIG [AD] | CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1F; CMT1F [AD]
NEK1	AMYOTROPHIC LATERAL SCLEROSIS. SUSCEPTIBILITY TO. 24; ALS24 [AD] | SHORT-RIB THORACIC DYSPLASIA 6 WITH OR WITHOUT POLYDACTYLY; SRTD6 [AR] | SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY; SRTD3 [AR]
NEK8	RENAL-HEPATIC-PANCREATIC DYSPLASIA 2; RHPD2 [AR]
NEU1	NEURAMINIDASE DEFICIENCY [AR]
NEUROG3	DIARRHEA 4. MALABSORPTIVE. CONGENITAL; DIAR4 [AR]
NFU1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 1; MMDS1 [AR]
NGF	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE V; HSAN5 [AR]
NHEJ1	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation 611291 (3)
NHLRC1	MYOCLONIC EPILEPSY OF LAFORA [AR]
NHP2	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 2; DKCB2 [AR]
NIPAL4	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 6; ARCI6 [AR]
NKX2-6	Conotruncal heart malformations 217095 (3) | Persistent truncus arteriosus 217095 (3)
NLRP7	HYDATIDIFORM MOLE. RECURRENT. 1; HYDM1 [AR]
NME8	CILIARY DYSKINESIA. PRIMARY. 6; CILD6 [AR]
NOP10	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 1; DKCB1 [AR]
NPC1	NIEMANN-PICK DISEASE. TYPE C2; NPC2 [AR] | NIEMANN-PICK DISEASE. TYPE C1; NPC1 [AR]
NPC2	NIEMANN-PICK DISEASE. TYPE C2; NPC2 [AR]

NPHP1	NEPHRONOPHTHISIS 1; NPHP1 [AR] | SENIOR-LOKEN SYNDROME 1; SLSN1 [AR] | JOUBERT SYNDROME 4; JBTS4 [AR]
NPHP3	MECKEL SYNDROME. TYPE 7; MKS7 [AR] | NEPHRONOPHTHISIS 3; NPHP3 [AR] | RENAL-HEPATIC-PANCREATIC DYSPLASIA 1; RHPD1 [AR]
NPHP4	NEPHRONOPHTHISIS 4; NPHP4 [AR] | SENIOR-LOKEN SYNDROME 4; SLSN4 [AR]
NPHS1	NEPHROTIC SYNDROME. TYPE 1; NPHS1 [AR]
NPHS2	NEPHROTIC SYNDROME. TYPE 2; NPHS2 [AR]
NPR2	ACROMESOMELIC DYSPLASIA. MAROTEAUX TYPE; AMDM [AR] | SHORT STATURE WITH NONSPECIFIC SKELETAL ABNORMALITIES; SNSK [AD] | EPIPHYSEAL CHONDRODYSPLASIA. MIURA TYPE; ECDM [AD]
NR0B1	ADRENAL HYPOPLASIA. CONGENITAL; AHC [XLR] | 46.XY SEX REVERSAL 2; SRXY2 [X-linked]
NR2E3	ENHANCED S-CONE SYNDROME; ESCS [AR] | RETINITIS PIGMENTOSA 37; RP37 [AD]
NR5A1	46.XX SEX REVERSAL 4; SRXX4 [AD] | PREMATURE OVARIAN FAILURE 7; POF7 [AD] | SPERMATOGENIC FAILURE 8; SPGF8 [AD] | 46.XY SEX REVERSAL 3; SRXY3 [AD]
NRL	RETINITIS PIGMENTOSA 27; RP27 [AD]
NRXN1	PITT-HOPKINS-LIKE SYNDROME 2; PTHSL2 [AR]
NSUN2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 5; MRT5 [AR]
NT5C3A	URIDINE 5-PRIME MONOPHOSPHATE HYDROLASE DEFICIENCY. HEMOLYTIC ANEMIA DUE TO [AR]
NT5E	CALCIFICATION OF JOINTS AND ARTERIES; CALJA [AR]
NTRK1	INSENSITIVITY TO PAIN. CONGENITAL. WITH ANHIDROSIS; CIPA [AR]
NUBPL	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 21; MC1DN21 [AR]
NUP62	STRIATONIGRAL DEGENERATION. INFANTILE; SNDI [AR]
OAT	GYRATE ATROPHY OF CHOROID AND RETINA; GACR [AR]
OCA2	SKIN/HAIR/EYE PIGMENTATION. VARIATION IN. 1; SHEP1 [AR] | ALBINISM. OCULOCUTANEOUS. TYPE II; OCA2 [AR]
OCLN	PSEUDO-TORCH SYNDROME 1; PTORCH1 [AR]
OCRL	LOWE OCULOCEREBRORENAL SYNDROME; OCRL [XLR] | DENT DISEASE 2 [XLR]
OFD1	SIMPSON-GOLABI-BEHMEL SYNDROME. TYPE 2; SGBS2 [XLR] | JOUBERT SYNDROME 10; JBTS10 [XLR] | RETINITIS PIGMENTOSA 23; RP23 [XLR] | OROFACIODIGITAL SYNDROME I; OFD1 [XLD]
OPA3	OPTIC ATROPHY 3. AUTOSOMAL DOMINANT; OPA3 [AD] | 3-METHYLGLUTACONIC ACIDURIA. TYPE III; MGCA3 [AR]
OPHN1	MENTAL RETARDATION. X-LINKED. WITH CEREBELLAR HYPOPLASIA AND DISTINCTIVE FACIAL APPEARANCE [XLR]
OPLAH	5-OXOPROLINASE DEFICIENCY; OPLAHD [AR]
OPN1LW	COLORBLINDNESS. PARTIAL. PROTAN SERIES; CBP [X-linked] | BLUE CONE MONOCHROMACY; BCM [XLR]
OPN1MW	COLORBLINDNESS. PARTIAL. DEUTAN SERIES; CBD [X-linked] | BLUE CONE MONOCHROMACY; BCM [XLR]
OPTN	GLAUCOMA. PRIMARY OPEN ANGLE; POAG [AD]
ORAI1	MYOPATHY. TUBULAR AGGREGATE. 2; TAM2 [AD] | IMMUNODEFICIENCY 9; IMD9 [AR]
ORC1	MEIER-GORLIN SYNDROME 1; MGORS1 [AR]
ORC4	MEIER-GORLIN SYNDROME 2; MGORS2 [AR]
ORC6	MEIER-GORLIN SYNDROME 3; MGORS3 [AR]
OSTM1	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 5; OPTB5 [AR]
OTC	ORNITHINE TRANSCARBAMYLASE DEFICIENCY. HYPERAMMONEMIA DUE TO [XLR]
OTOA	DEAFNESS. AUTOSOMAL RECESSIVE 22; DFNB22 [AR]
OTOF	DEAFNESS. AUTOSOMAL RECESSIVE 9; DFNB9 [AR]
OTOG	DEAFNESS. AUTOSOMAL RECESSIVE 18B; DFNB18B [AR]
OTOGL	DEAFNESS. AUTOSOMAL RECESSIVE 84B; DFNB84B [AR]
P2RY12	BLEEDING DISORDER. PLATELET-TYPE. 8; BDPLT8 [AR]
PAH	PHENYLKETONURIA; PKU [AR]
PAK3	MENTAL RETARDATION. X-LINKED 30; MRX30 [XLR]
PALB2	BREAST CANCER [AD]

PANK2	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1; NBIA1 [AR] | HYPOPREBETALIPOPROTEINEMIA. ACANTHOCYTOSIS. RETINITIS PIGMENTOSA. AND PALLIDAL DEGENERATION [AR]
PAPSS2	BRACHYOLMIA TYPE 4 WITH MILD EPIPHYSEAL AND METAPHYSEAL CHANGES; BCYM4 [AR]
PARK7	PARKINSON DISEASE 7. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK7 [AR]
PAX3	RHABDOMYOSARCOMA 2; RMS2 [Somatic mutation] | WAARDENBURG SYNDROME. TYPE 1; WS1 [AD] | WAARDENBURG SYNDROME. TYPE 3; WS3 [AD] | CRANIOFACIAL-DEAFNESS-HAND SYNDROME; CDHS [AD]
PAX6	COLOBOMA OF OPTIC NERVE [AD] | KERATITIS. HEREDITARY [AD] | COLOBOMA. OCULAR. AUTOSOMAL DOMINANT [AD] | OPTIC NERVE HYPOPLASIA. BILATERAL [AD] | FOVEAL HYPOPLASIA 1; FVH1 [AD] | ANIRIDIA 1; AN1 [AD]
PAX7	MYOPATHY. CONGENITAL. PROGRESSIVE. WITH SCOLIOSIS; MYOSCO [AR] | RHABDOMYOSARCOMA 2; RMS2 [Somatic mutation]
PAX8	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 2; CHNG2 [AD]
PC	Pyruvate carboxylase deficiency 266150 (3)
PCBD1	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. D; HPABH4D [AR]
PCCA	PROPIONIC ACIDEMIA [AR]
PCCB	PROPIONIC ACIDEMIA [AR]
PCDH15	DEAFNESS. AUTOSOMAL RECESSIVE 23; DFNB23 [AR] | USHER SYNDROME. TYPE ID; USH1D [AR] | USHER SYNDROME. TYPE IF; USH1F [AR]
PCNT	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM. TYPE II; MOPD2 [AR]
PDE6A	Retinitis pigmentosa 43 613810 (3)
PDE6B	NIGHT BLINDNESS. CONGENITAL STATIONARY. AUTOSOMAL DOMINANT 2; CSNBAD2 [AD] | RETINITIS PIGMENTOSA 40; RP40 [AR]
PDE6C	CONE DYSTROPHY 4; COD4 [AR]
PDE6G	RETINITIS PIGMENTOSA 57; RP57 [AR]
PDE6H	RETINAL CONE DYSTROPHY 3A; RCD3A [AD]
PDHA1	PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY; PDHAD [XLD]
PDHX	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY; PDHXD [AR]
PDP1	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY; PDHPD [AR]
PDSS1	COENZYME Q10 DEFICIENCY. PRIMARY. 2; COQ10D2 [AR]
PDSS2	COENZYME Q10 DEFICIENCY. PRIMARY. 3; COQ10D3 [AR]
PDX1	PANCREATIC AGENESIS 1; PAGEN1 [AR] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
PDZD7	USHER SYNDROME. TYPE IIC; USH2C [AR] | USHER SYNDROME. TYPE IIA; USH2A [AR] | DEAFNESS. AUTOSOMAL RECESSIVE 57; DFNB57 [AR]
PEPD	PROLIDASE DEFICIENCY [AR]
PEX1	HEIMLER SYNDROME 1; HMLR1 [AR] | PEROXISOME BIOGENESIS DISORDER 1B; PBD1B [AR] | PEROXISOME BIOGENESIS DISORDER 1A (ZELLWEGER); PBD1A [AR]
PEX10	PEROXISOME BIOGENESIS DISORDER 6B; PBD6B [AR] | PEROXISOME BIOGENESIS DISORDER 6A (ZELLWEGER); PBD6A [AR]
PEX13	PEROXISOME BIOGENESIS DISORDER 11A (ZELLWEGER); PBD11A [AR] | PEROXISOME BIOGENESIS DISORDER 11B; PBD11B [AR]
PEX14	PEROXISOME BIOGENESIS DISORDER 13A (ZELLWEGER); PBD13A [AR]
PEX19	PEROXISOME BIOGENESIS DISORDER 12A (ZELLWEGER); PBD12A [AR]
PEX2	PEROXISOME BIOGENESIS DISORDER 5B; PBD5B [AR] | PEROXISOME BIOGENESIS DISORDER 3B; PBD3B [AR] | PEROXISOME BIOGENESIS DISORDER 5A (ZELLWEGER); PBD5A [AR]
PEX26	PEROXISOME BIOGENESIS DISORDER 7B; PBD7B [AR] | PEROXISOME BIOGENESIS DISORDER 7A (ZELLWEGER); PBD7A [AR]
PEX3	PEROXISOME BIOGENESIS DISORDER 10A (ZELLWEGER); PBD10A [AR] | PEROXISOME BIOGENESIS DISORDER 10B; PBD10B [AR]
PEX5	PEROXISOME BIOGENESIS DISORDER 2A (ZELLWEGER); PBD2A [Autosomal recessive, several form] | PEROXISOME BIOGENESIS DISORDER 2B; PBD2B [AR] | RHIZOMELIC CHONDRODYSPLASIA PUNCTATA. TYPE 5; RCDP5 [AR]
PEX6	PEROXISOME BIOGENESIS DISORDER 4A (ZELLWEGER); PBD4A [AR] | PEROXISOME BIOGENESIS DISORDER 4B; PBD4B [AR] | HEIMLER SYNDROME 2; HMLR2 [AR]
PEX7	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA. TYPE 1; RCDP1 [AR] | PEROXISOME BIOGENESIS DISORDER 9B; PBD9B [AR] | REFSUM DISEASE. CLASSIC [AR]
PFKM	GLYCOGEN STORAGE DISEASE VII; GSD7 [AR]
PGAM2	GLYCOGEN STORAGE DISEASE X; GSD10 [AR]

PGAP2	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 3; HPMRS3 [AR]
PGK1	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY [XLR]
PGM1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE It; CDG1T [AR]
PHF6	BORJESON-FORSSMAN-LEHMANN SYNDROME; BFLS [XLR]
PHGDH	PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY; PHGDHD [AR] | NEU-LAXOVA SYNDROME 1; NLS1 [AR]
PHKA1	GLYCOGEN STORAGE DISEASE. TYPE IXd; GSD9D [XLR]
PHKA2	GLYCOGEN STORAGE DISEASE IXa1; GSD9A1 [XLR]
PHKB	GLYCOGEN STORAGE DISEASE IXb; GSD9B [AR]
PHKG2	GLYCOGEN STORAGE DISEASE IXc; GSD9C [AR]
PHOX2A	FIBROSIS OF EXTRAOCULAR MUSCLES. CONGENITAL. 2; CFEOM2 [AR]
PHYH	REFSUM DISEASE. CLASSIC [AR]
PIGL	COLOBOMA. CONGENITAL HEART DISEASE. ICHTHYOSIFORM DERMATOSIS. MENTAL RETARDATION. AND EAR ANOMALIES SYNDROME; CHIME [AR]
PIGM	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 1; GPIBD1 [AR]
PIGN	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 1; MCAHS1 [AR]
PIGV	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 1; HPMRS1 [AR]
PIK3R1	SHORT SYNDROME [AD] | IMMUNODEFICIENCY 36; IMD36 [AD] | AGAMMAGLOBULINEMIA 7. AUTOSOMAL RECESSIVE; AGM7 [AR]
PINK1	PARKINSON DISEASE 6. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK6 [AR]
PIP5K1C	LETHAL CONGENITAL CONTRACTURE SYNDROME 3; LCCS3 [AR]
PKHD1	POLYCYSTIC KIDNEY DISEASE 4 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD4 [AR]
PKLR	ADENOSINE TRIPHOSPHATE. ELEVATED. OF ERYTHROCYTES [AD] | PYRUVATE KINASE DEFICIENCY OF RED CELLS [AR]
PLA2G6	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 2A; NBIA2A [AR] | NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 2B; NBIA2B [AR] | PARKINSON DISEASE 14. AUTOSOMAL RECESSIVE; PARK14 [AR]
PLA2G7	IgE RESPONSIVENESS. ATOPIC; IGER [AD] | ASTHMA. SUSCEPTIBILITY TO [Autosomal dominant vs. multifactoria] | PLATELET-ACTIVATING FACTOR ACETYLHYDROLASE DEFICIENCY; PAFAD [AR]
PLCB1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 12; EIEE12 [AR]
PLCD1	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 3; NDNC3 [AD]
PLCE1	NEPHROTIC SYNDROME. TYPE 3; NPHS3 [AR]
PLEC	EPIDERMOLYSIS BULLOSA SIMPLEX WITH NAIL DYSTROPHY; EBSND [AR] | EPIDERMOLYSIS BULLOSA SIMPLEX WITH PYLORIC ATRESIA; EBSPA [AR] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 17; LGMDR17 [AR] | EPIDERMOLYSIS BULLOSA SIMPLEX. OGNA TYPE; EBSOG [AD] | EPIDERMOLYSIS BULLOSA SIMPLEX WITH MUSCULAR DYSTROPHY; EBSMD [AR]
PLEKHG5	SPINAL MUSCULAR ATROPHY. DISTAL. AUTOSOMAL RECESSIVE. 4; DSMA4 [AR] | CHARCOT-MARIE-TOOTH DISEASE. RECESSIVE INTERMEDIATE C; CMTRIC [AR]
PLEKHM1	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 6; OPTB6 [AR] | OSTEOPETROSIS. AUTOSOMAL DOMINANT 3; OPTA3 [AD]
PLG	PLASMINOGEN DEFICIENCY. TYPE I [AR]
PLOD1	EHLERS-DANLOS SYNDROME. KYPHOSCOLIOTIC TYPE. 1; EDSKSCL1 [AR]
PLOD2	BRUCK SYNDROME 2; BRKS2 [AR]
PLOD3	BONE FRAGILITY WITH CONTRACTURES. ARTERIAL RUPTURE. AND DEAFNESS [AR]
PLP1	PELIZAEUS-MERZBACHER DISEASE; PMD [XLR] | SPASTIC PARAPLEGIA 2. X-LINKED; SPG2 [XLR]
PMM2	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ia; CDG1A [AR]
PMP22	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS [AD] | GUILLAIN-BARRE SYNDROME. FAMILIAL; GBS [AD] | NEUROPATHY. HEREDITARY. WITH LIABILITY TO PRESSURE PALSIES; HNPP [AD] | CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1A; CMT1A [AD] | ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA [AD] | HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS [AD]
PMS2	MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR]
PNKP	MICROCEPHALY. SEIZURES. AND DEVELOPMENTAL DELAY; MCSZ [AR] | ATAXIA-OCULOMOTOR APRAXIA 4; AOA4 [AR]
PNP	PURINE NUCLEOSIDE PHOSPHORYLASE DEFICIENCY [AR]
PNPLA1	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 10; ARCI10 [AR]

PNPLA6	SPASTIC PARAPLEGIA 39. AUTOSOMAL RECESSIVE; SPG39 [AR] | OLIVER-MCFARLANE SYNDROME; OMCS [AR] | BOUCHER-NEUHAUSER SYNDROME; BNHS [AR] | LAURENCE-MOON SYNDROME; LNMS [AR]
PNPO	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY; PNPOD [AR]
PNPT1	DEAFNESS. AUTOSOMAL RECESSIVE 70; DFNB70 [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 13; COXPD13 [AR]
POC1A	SHORT STATURE. ONYCHODYSPLASIA. FACIAL DYSMORPHISM. AND HYPOTRICHOSIS; SOFT [AR]
POLG	MITOCHONDRIAL DNA DEPLETION SYNDROME 4B (MNGIE TYPE); MTDPS4B [AR] | SENSORY ATAXIC NEUROPATHY. DYSARTHRIA. AND OPHTHALMOPARESIS; SANDO [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 1; PEOA1 [AD] | MITOCHONDRIAL DNA DEPLETION SYNDROME 4A (ALPERS TYPE); MTDPS4A [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL RECESSIVE 1; PEOB1 [AR]
POLH	XERODERMA PIGMENTOSUM. VARIANT TYPE; XPV [AR]
POLR1C	LEUKODYSTROPHY. HYPOMYELINATING. 11; HLD11 [AR] | TREACHER COLLINS SYNDROME 3; TCS3 [AR]
POLR3A	WIEDEMANN-RAUTENSTRAUCH SYNDROME; WDRTS [AR] | LEUKODYSTROPHY. HYPOMYELINATING. 7. WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM; HLD7 [AR]
POLR3B	LEUKODYSTROPHY. HYPOMYELINATING. 8. WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM; HLD8 [AR]
POMC	OBESITY [AD] | OBESITY. EARLY-ONSET. WITH ADRENAL INSUFFICIENCY AND RED HAIR; OBAIRH [AR]
POMGNT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION). TYPE B. 3; MDDGB3 [AR] | RETINITIS PIGMENTOSA 76; RP76 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 3; MDDGA3 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 3; MDDGC3 [AR]
POMP	KERATOSIS LINEARIS WITH ICHTHYOSIS CONGENITA AND SCLEROSING KERATODERMA; KLICK [AR] | PROTEASOME-ASSOCIATED AUTOINFLAMMATORY SYNDROME 2; PRAAS2 [AD]
POMT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION). TYPE B. 1; MDDGB1 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 1; MDDGA1 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 1; MDDGC1 [AR]
POMT2	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION). TYPE B. 2; MDDGB2 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 2; MDDGC2 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 2; MDDGA2 [AR]
POR	ANTLEY-BIXLER SYNDROME WITH GENITAL ANOMALIES AND DISORDERED STEROIDOGENESIS; ABS1 [AR]
POU1F1	PITUITARY HORMONE DEFICIENCY. COMBINED. 1; CPHD1 [AD]
POU3F4	DEAFNESS. X-LINKED 2; DFNX2 [XLR] | CHOROIDEREMIA. DEAFNESS. AND MENTAL RETARDATION [XLR]
PPIB	OSTEOGENESIS IMPERFECTA. TYPE IX; OI9 [AR]
PPP2R1B	LUNG CANCER [Somatic mutation]
PPT1	CEROID LIPOFUSCINOSIS. NEURONAL. 1; CLN1 [AR]
PQBP1	RENPENNING SYNDROME 1; RENS1 [XLR]
PRCD	Retinitis pigmentosa 36 610599 (3)
PRDM5	BRITTLE CORNEA SYNDROME 2; BCS2 [AR]
PREPL	MYASTHENIC SYNDROME. CONGENITAL. 22; CMS22 [AR]
PRF1	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 2; FHL2 [AR]
PRG4	CAMPTODACTYLY-ARTHROPATHY-COXA VARA-PERICARDITIS SYNDROME; CACP [AR]
PRICKLE1	EPILEPSY. PROGRESSIVE MYOCLONIC. 1B; EPM1B [AR]
PRKAG2	GLYCOGEN STORAGE DISEASE OF HEART. LETHAL CONGENITAL [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 6; CMH6 [AD] | WOLFF-PARKINSON-WHITE SYNDROME [AD]
PRKAR1A	CARNEY COMPLEX. TYPE 1; CNC1 [AD] | MYXOMA. INTRACARDIAC [AD] | ACRODYSOSTOSIS 1 WITH OR WITHOUT HORMONE RESISTANCE; ACRDYS1 [AD] | PIGMENTED NODULAR ADRENOCORTICAL DISEASE. PRIMARY. 1; PPNAD1 [AD]
PRKCG	SPINOCEREBELLAR ATAXIA 14; SCA14 [AD]
PRKRA	DYSTONIA 16; DYT16 [AR]
PROC	THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY. AUTOSOMAL DOMINANT; THPH3 [AD] | THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY. AUTOSOMAL RECESSIVE; THPH4 [AR]
PROKR2	HYPOGONADOTROPIC HYPOGONADISM 3 WITH OR WITHOUT ANOSMIA; HH3 [AD]
PROM1	RETINITIS PIGMENTOSA 41; RP41 [AR] | MACULAR DYSTROPHY. RETINAL. 2; MCDR2 [AD]
PROP1	PITUITARY HORMONE DEFICIENCY. COMBINED. 2; CPHD2 [AR]

PROS1	THROMBOPHILIA DUE TO PROTEIN S DEFICIENCY. AUTOSOMAL DOMINANT; THPH5 [AD] | THROMBOPHILIA DUE TO PROTEIN S DEFICIENCY. AUTOSOMAL RECESSIVE; THPH6 [AR]
PRPH2	CHOROIDAL DYSTROPHY. CENTRAL AREOLAR 2; CACD2 [AD] | RETINITIS PIGMENTOSA 7; RP7 [AD] | MACULAR DYSTROPHY. PATTERNED. 1; MDPT1 [AD] | MACULAR DYSTROPHY. VITELLIFORM. 3; VMD3 [AD] | FUNDUS ALBIPUNCTATUS [AD]
PRPS1	PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE SUPERACTIVITY [XLR] | CHARCOT-MARIE-TOOTH DISEASE. X-LINKED RECESSIVE. 5; CMTX5 [XLR] | DEAFNESS. X-LINKED 1; DFNX1 [X-linked] | ARTS SYNDROME; ARTS [XLR]
PRRX1	AGNATHIA-OTOCEPHALY COMPLEX; AGOTC [AR]
PRSS1	PANCREATITIS. HEREDITARY; PCTT [AD]
PRSS12	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 1; MRT1 [AR]
PRSS56	MICROPHTHALMIA. ISOLATED 6; MCOP6 [AR]
PRX	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS [AD] | CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 4F; CMT4F [AR]
PSAP	COMBINED SAPOSIN DEFICIENCY [AR] | KRABBE DISEASE. ATYPICAL. DUE TO SAPOSIN A DEFICIENCY [AR] | METACHROMATIC LEUKODYSTROPHY DUE TO SAPOSIN B DEFICIENCY [AR]
PSAT1	NEU-LAXOVA SYNDROME 2; NLS2 [AR] | PHOSPHOSERINE AMINOTRANSFERASE DEFICIENCY; PSATD [AR]
PSMB8	PROTEASOME-ASSOCIATED AUTOINFLAMMATORY SYNDROME 1; PRAAS1 [AR]
PSMC3IP	OVARIAN DYSGENESIS 3; ODG3 [AR]
PSPH	PHOSPHOSERINE PHOSPHATASE DEFICIENCY; PSPHD [AR]
PTF1A	PANCREATIC AGENESIS 2; PAGEN2 [AR] | PANCREATIC AND CEREBELLAR AGENESIS; PACA [AR]
PTH	HYPOPARATHYROIDISM. FAMILIAL ISOLATED; FIH [AD]
PTH1R	CHONDRODYSPLASIA. BLOMSTRAND TYPE; BOCD [AR] | EIKEN SYNDROME [AR] | FAILURE OF TOOTH ERUPTION. PRIMARY; PFE [AD] | METAPHYSEAL CHONDRODYSPLASIA. JANSEN TYPE; MCDJ [AD]
PTPN14	CHOANAL ATRESIA AND LYMPHEDEMA; CATLPH [AR]
PTPRC	MULTIPLE SCLEROSIS. SUSCEPTIBILITY TO; MS [Multifactorial] | SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-POSITIVE [AR]
PTPRO	NEPHROTIC SYNDROME. TYPE 6; NPHS6 [AR]
PTPRQ	DEAFNESS. AUTOSOMAL DOMINANT 73; DFNA73 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 84A; DFNB84A [AR]
PTS	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. A; HPABH4A [AR]
PUS1	MYOPATHY. LACTIC ACIDOSIS. AND SIDEROBLASTIC ANEMIA 1; MLASA1 [AR]
PYCR1	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IIB; ARCL2B [AR]
PYGL	GLYCOGEN STORAGE DISEASE VI; GSD6 [AR]
PYGM	GLYCOGEN STORAGE DISEASE V; GSD5 [AR]
QDPR	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. C; HPABH4C [AR]
RAB23	CARPENTER SYNDROME 1; CRPT1 [AR]
RAB27A	GRISCELLI SYNDROME. TYPE 2; GS2 [AR]
RAB28	CONE-ROD DYSTROPHY 18; CORD18 [AR]
RAB39B	MENTAL RETARDATION. X-LINKED 72; MRX72 [XLR] | WAISMAN SYNDROME; WSMN [XLR]
RAB3GAP1	WARBURG MICRO SYNDROME 1; WARBM1 [AR]
RAB3GAP2	MARTSOLF SYNDROME [AR] | WARBURG MICRO SYNDROME 2; WARBM2 [AR]
RAD51C	FANCONI ANEMIA. COMPLEMENTATION GROUP O; FANCO [AR]
RAG1	COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS WITH GRANULOMAS; CCHIDG [AR] | SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-NEGATIVE. NK CELL-POSITIVE [AR] | OMENN SYNDROME [AR]
RAG2	COMBINED CELLULAR AND HUMORAL IMMUNE DEFECTS WITH GRANULOMAS; CCHIDG [AR] | SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-NEGATIVE. NK CELL-POSITIVE [AR] | OMENN SYNDROME [AR]
RAPSN	MYASTHENIC SYNDROME. CONGENITAL. 11. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS11 [AR] | FETAL AKINESIA DEFORMATION SEQUENCE 1; FADS1 [AR]
RARB	MICROPHTHALMIA. SYNDROMIC 12; MCOPS12 [AD]
RAX	MICROPHTHALMIA. ISOLATED 3; MCOP3 [AR]
RBBP8	SECKEL SYNDROME 2; SCKL2 [AR] | JAWAD SYNDROME; JWDS [AR]
RBM8A	THROMBOCYTOPENIA-ABSENT RADIUS SYNDROME; TAR [AR]

RD3	LEBER CONGENITAL AMAUROSIS 12; LCA12 [AR]
RDH12	LEBER CONGENITAL AMAUROSIS 13; LCA13 [Autosomal dominant (in one RP53 family)]
RDH5	FUNDUS ALBIPUNCTATUS [AD]
RDX	DEAFNESS. AUTOSOMAL RECESSIVE 24; DFNB24 [AR]
RECQL4	ROTHMUND-THOMSON SYNDROME. TYPE 2; RTS2 [AR] | RAPADILINO SYNDROME [AR] | BALLER-GEROLD SYNDROME; BGS [AR]
RELN	LISSENCEPHALY 2; LIS2 [AR] | EPILEPSY. FAMILIAL TEMPORAL LOBE. 7; ETL7 [AD]
REN	HYPERURICEMIC NEPHROPATHY. FAMILIAL JUVENILE. 2; HNFJ2 [AD] | RENAL TUBULAR DYSGENESIS; RTD [AR]
RFX5	BARE LYMPHOCYTE SYNDROME. TYPE II [AR]
RFX6	MITCHELL-RILEY SYNDROME; MTCHRS [AR]
RFXANK	BARE LYMPHOCYTE SYNDROME. TYPE II [AR]
RFXAP	BARE LYMPHOCYTE SYNDROME. TYPE II [AR]
RGR	Retinitis pigmentosa 44 613769 (3)
RGS9	Bradyopsia 608415 (3)
RHAG	OVERHYDRATED HEREDITARY STOMATOCYTOSIS; OHST [AD] | RH-NULL. REGULATOR TYPE; RHNR [AD]
RHO	RETINITIS PIGMENTOSA 4; RP4 [AD] | FUNDUS ALBIPUNCTATUS [AD]
RIN2	MACS SYNDROME [AR]
RIPK4	CHAND SYNDROME; CHANDS [AR] | BARTSOCAS-PAPAS SYNDROME; BPS [AR]
RLBP1	BOTHNIA RETINAL DYSTROPHY [AR] | FUNDUS ALBIPUNCTATUS [AD]
RMRP	ANAUXETIC DYSPLASIA 1; ANXD1 [AR] | CARTILAGE-HAIR HYPOPLASIA; CHH [AR] | METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS; MDWH [AR]
RNASEH2B	AICARDI-GOUTIERES SYNDROME 2; AGS2 [AR]
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3; AGS3 [AR]
RNASET2	LEUKOENCEPHALOPATHY. CYSTIC. WITHOUT MEGALENCEPHALY [AR]
RNU4ATAC	ROIFMAN SYNDROME; RFMN [AR] | MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM. TYPE I; MOPD1 [AR]
ROBO3	GAZE PALSY. FAMILIAL HORIZONTAL. WITH PROGRESSIVE SCOLIOSIS 1; HGPPS1 [AR]
ROR2	BRACHYDACTYLY. TYPE B1; BDB1 [AD] | ROBINOW SYNDROME. AUTOSOMAL RECESSIVE 1; RRS1 [AR]
RP1	RETINITIS PIGMENTOSA 1; RP1 [AD]
RP2	RETINITIS PIGMENTOSA 2; RP2 [X-linked]
RPE65	LEBER CONGENITAL AMAUROSIS 2; LCA2 [AR] | RETINITIS PIGMENTOSA 20; RP20 [AR] | RETINITIS PIGMENTOSA 87 WITH CHOROIDAL INVOLVEMENT; RP87 [AD]
RPGR	MACULAR DEGENERATION. X-LINKED ATROPHIC [XLR] | CONE-ROD DYSTROPHY. X-LINKED. 1; CORDX1 [X-linke]
RPGRIP1	LEBER CONGENITAL AMAUROSIS 6; LCA6 [AR]
RPGRIP1L	JOUBERT SYNDROME 7; JBTS7 [AR] | COACH SYNDROME [AR] | MECKEL SYNDROME. TYPE 5; MKS5 [AR]
RPIA	RIBOSE 5-PHOSPHATE ISOMERASE DEFICIENCY; RPIAD [AR]
RRM2B	MITOCHONDRIAL DNA DEPLETION SYNDROME 8A (ENCEPHALOMYOPATHIC TYPE WITH RENAL TUBULOPATHY); MTDPS8A [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 5; PEOA5 [AD]
RS1	RETINOSCHISIS 1. X-LINKED. JUVENILE; RS1 [XLR]
RSPH9	Ciliary dyskinesia primary 12 612650 (3)
RSPO4	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 4; NDNC4 [AR]
RTEL1	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 5; DKCB5 [AD] | PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE. TELOMERE-RELATED. 3; PFBMFT3 [AD]
RTTN	MICROCEPHALY. SHORT STATURE. AND POLYMICROGYRIA WITH OR WITHOUT SEIZURES; MSSP [AR]
RYR1	CENTRAL CORE DISEASE OF MUSCLE; CCD [AD] | MALIGNANT HYPERTHERMIA. SUSCEPTIBILITY TO. 1; MHS1 [AD] | MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGIA [AR]
SACS	SPASTIC ATAXIA. CHARLEVOIX-SAGUENAY TYPE; SACS [AR]
SAG	OGUCHI DISEASE 1 [AR]

SAMD9	MIRAGE SYNDROME; MIRAGE [AD] | TUMORAL CALCINOSIS. NORMOPHOSPHATEMIC. FAMILIAL; NFTC [AR]
SAR1B	CHYLOMICRON RETENTION DISEASE; CMRD [AR]
SARS2	HYPERURICEMIA. PULMONARY HYPERTENSION. RENAL FAILURE. AND ALKALOSIS SYNDROME; HUPRAS [AR]
SBDS	SHWACHMAN-DIAMOND SYNDROME 1; SDS1 [AR]
SBF2	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4B2; CMT4B2 [AR]
SC5D	LATHOSTEROLOSIS [AR]
SCARB2	EPILEPSY. PROGRESSIVE MYOCLONIC. 4. WITH OR WITHOUT RENAL FAILURE; EPM4 [AR]
SCARF2	VAN DEN ENDE-GUPTA SYNDROME; VDEGS [AR]
SCN5A	PROGRESSIVE FAMILIAL HEART BLOCK. TYPE IA; PFHB1A [AD] | BRUGADA SYNDROME 1; BRGDA1 [AD] | CARDIOMYOPATHY. DILATED. 1E; CMD1E [AD] | SICK SINUS SYNDROME 1; SSS1 [AR] | LONG QT SYNDROME 3; LQT3 [AD] | SUDDEN INFANT DEATH SYNDROME [Autosomal recessive form vs. multifactoria] | ATRIAL FIBRILLATION. FAMILIAL. 10; ATFB10 [AD]
SCN9A	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 6; EIEE6 [AD] | PAROXYSMAL EXTREME PAIN DISORDER [AD] | GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS. TYPE 7; GEFSP7 [AD] | ERYTHERMALGIA. PRIMARY [AD] | INDIFFERENCE TO PAIN. CONGENITAL. AUTOSOMAL RECESSIVE; CIP [AR]
SCNN1A	PSEUDOHYPOALDOSTERONISM. TYPE I. AUTOSOMAL RECESSIVE; PHA1B [AR] | LIDDLE SYNDROME 3; LIDLS3 [AD] | BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 2; BESC2 [AD]
SCNN1B	PSEUDOHYPOALDOSTERONISM. TYPE I. AUTOSOMAL RECESSIVE; PHA1B [AR] | LIDDLE SYNDROME 1; LIDLS1 [AD] | BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 1; BESC1 [AD]
SCNN1G	PSEUDOHYPOALDOSTERONISM. TYPE I. AUTOSOMAL RECESSIVE; PHA1B [AR] | LIDDLE SYNDROME 2; LIDLS2 [AD] | BRONCHIECTASIS WITH OR WITHOUT ELEVATED SWEAT CHLORIDE 3; BESC3 [AD]
SCO2	MYOPIA 6; MYP6 [AD] | CARDIOENCEPHALOMYOPATHY. FATAL INFANTILE. DUE TO CYTOCHROME c OXIDASE DEFICIENCY 1; CEMCOX1 [AR]
SDCCAG8	BARDET-BIEDL SYNDROME 16; BBS16 [AR]
SDHA	LEIGH SYNDROME; LS [AR] | PARAGANGLIOMAS 5; PGL5 [AD] | MITOCHONDRIAL COMPLEX II DEFICIENCY [AR]
SDHAF1	MITOCHONDRIAL COMPLEX II DEFICIENCY [AR]
SEC23A	CRANIOLENTICULOSUTURAL DYSPLASIA; CLSD [AR]
SEC23B	ANEMIA. CONGENITAL DYSERYTHROPOIETIC. TYPE II; CDAN2 [AR] | COWDEN SYNDROME 7; CWS7 [AD]
SEMA4A	CONE-ROD DYSTROPHY 10; CORD10 [AR] | RETINITIS PIGMENTOSA 35; RP35 [AD]
SEPSECS	PONTOCEREBELLAR HYPOPLASIA. TYPE 2D; PCH2D [AR]
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD [AR]
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY [AD]
SERPINC1	ANTITHROMBIN III DEFICIENCY; AT3D [AD]
SERPINE1	PLASMINOGEN ACTIVATOR INHIBITOR-1 DEFICIENCY [AR]
SERPINF1	OSTEOGENESIS IMPERFECTA. TYPE VI; OI6 [AR]
SERPINF2	ALPHA-2-PLASMIN INHIBITOR DEFICIENCY [AR]
SERPING1	ANGIOEDEMA. HEREDITARY. TYPE I; HAE1 [AD] | COMPLEMENT COMPONENT 4. PARTIAL DEFICIENCY OF [AD]
SERPINH1	OSTEOGENESIS IMPERFECTA. TYPE X; OI10 [AR]
SETX	AMYOTROPHIC LATERAL SCLEROSIS 4. JUVENILE; ALS4 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE. WITH AXONAL NEUROPATHY 2; SCAN2 [AR]
SFTPB	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 1; SMDP1 [AR]
SFTPC	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 2; SMDP2 [AD]
SGCA	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 3; LGMDR3 [AR]
SGCB	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 4; LGMDR4 [AR]
SGCD	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 6; LGMDR6 [AR]
SGCG	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 5; LGMDR5 [AR]
SGSH	MUCOPOLYSACCHARIDOSIS. TYPE IIIA; MPS3A [AR]
SH2D1A	LYMPHOPROLIFERATIVE SYNDROME. X-LINKED. 1; XLP1 [XLR]
SH3PXD2B	FRANK-TER HAAR SYNDROME; FTHS [AR]

SH3TC2	MONONEUROPATHY OF THE MEDIAN NERVE. MILD; MNMN [AD] | CHARCOT-MARIE-TOOTH DISEASE. TYPE 4C; CMT4C [AR]
SHOX	LERI-WEILL DYSCHONDROSTEOSIS; LWD [AD] | LANGER MESOMELIC DYSPLASIA; LMD [AR]
SI	Sucrase-isomaltase deficiency congenital 222900 (3)
SIL1	MARINESCO-SJOGREN SYNDROME; MSS [AR]
SIX6	OPTIC DISC ANOMALIES WITH RETINAL AND/OR MACULAR DYSTROPHY; ODRMD [AR]
SLC10A2	BILE ACID MALABSORPTION. PRIMARY; PBAM [AR]
SLC11A2	ANEMIA. HYPOCHROMIC MICROCYTIC. WITH IRON OVERLOAD 1; AHMIO1 [AR]
SLC12A1	BARTTER SYNDROME. TYPE 1. ANTENATAL; BARTS1 [AR]
SLC12A3	GITELMAN SYNDROME; GTLMNS [AR]
SLC12A6	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY; ACCPN [AR]
SLC16A2	ALLAN-HERNDON-DUDLEY SYNDROME; AHDS [X-linked]
SLC17A5	SALLA DISEASE; SD [AR] | INFANTILE SIALIC ACID STORAGE DISEASE; ISSD [AR]
SLC19A2	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME; TRMA [AR]
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 (BIOTIN- OR THIAMINE-RESPONSIVE TYPE); THMD2 [AR]
SLC22A12	HYPOURICEMIA. RENAL. 1; RHUC1 [AR]
SLC22A18	BREAST CANCER [AD] | LUNG CANCER [Somatic mutation] | RHABDOMYOSARCOMA. EMBRYONAL. 1; RMSE1 [AR]
SLC22A5	CARNITINE DEFICIENCY. SYSTEMIC PRIMARY; CDSP [AR]
SLC24A1	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1D; CSNB1D [AR]
SLC25A12	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 39; EIEE39 [AR]
SLC25A13	CITRULLINEMIA. TYPE II. ADULT-ONSET; CTLN2 [AR] | CITRULLINEMIA. TYPE II. NEONATAL-ONSET [AR]
SLC25A15	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME; HHHS [AR]
SLC25A19	MICROCEPHALY. AMISH TYPE; MCPHA [AR] | THIAMINE METABOLISM DYSFUNCTION SYNDROME 4 (BILATERAL STRIATAL DEGENERATION AND PROGRESSIVE POLYNEUROPATHY TYPE); THMD4 [AR]
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY; CACTD [AR]
SLC25A22	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 3; EIEE3 [AR]
SLC25A38	ANEMIA. SIDEROBLASTIC. 2. PYRIDOXINE-REFRACTORY; SIDBA2 [AR]
SLC25A4	MITOCHONDRIAL DNA DEPLETION SYNDROME 12A (CARDIOMYOPATHIC TYPE). AUTOSOMAL DOMINANT; MTDPS12A [AD] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 2; PEOA2 [AD] | MITOCHONDRIAL DNA DEPLETION SYNDROME 12B (CARDIOMYOPATHIC TYPE). AUTOSOMAL RECESSIVE; MTDPS12B [AR]
SLC26A2	EPIPHYSEAL DYSPLASIA. MULTIPLE. 4; EDM4 [AR] | ACHONDROGENESIS. TYPE IB; ACG1B [AR] | DIASTROPHIC DYSPLASIA; DTD [AR] | ATELOSTEOGENESIS. TYPE II; AO2 [AR]
SLC26A3	DIARRHEA 1. SECRETORY CHLORIDE. CONGENITAL; DIAR1 [AR]
SLC26A4	DEAFNESS. AUTOSOMAL RECESSIVE 4. WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4 [AR] | PENDRED SYNDROME; PDS [AR]
SLC27A4	Ichthyosis prematurity syndrome 608649 (3)
SLC29A3	HISTIOCYTOSIS-LYMPHADENOPATHY PLUS SYNDROME [AR]
SLC2A1	GLUT1 DEFICIENCY SYNDROME 1; GLUT1DS1 [AD] | DYSTONIA 9; DYT9 [AD] | EPILEPSY. IDIOPATHIC GENERALIZED. SUSCEPTIBILITY TO. 12; EIG12 [AD] | GLUT1 DEFICIENCY SYNDROME 2; GLUT1DS2 [AD] | STOMATIN-DEFICIENT CRYOHYDROCYTOSIS WITH NEUROLOGIC DEFECTS; SDCHCN [AD]
SLC2A10	ARTERIAL TORTUOSITY SYNDROME; ATORS [AR]
SLC2A2	FANCONI-BICKEL SYNDROME; FBS [AR] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
SLC33A1	CONGENITAL CATARACTS. HEARING LOSS. AND NEURODEGENERATION; CCHLND [AR] | SPASTIC PARAPLEGIA 42. AUTOSOMAL DOMINANT; SPG42 [AD]
SLC34A1	HYPERCALCEMIA. INFANTILE. 2; HCINF2 [AR] | FANCONI RENOTUBULAR SYNDROME 2; FRTS2 [AR] | NEPHROLITHIASIS/OSTEOPOROSIS. HYPOPHOSPHATEMIC. 1; NPHLOP1 [AD]
SLC34A2	PULMONARY ALVEOLAR MICROLITHIASIS; PULAM [AR]
SLC34A3	HYPOPHOSPHATEMIC RICKETS WITH HYPERCALCIURIA. HEREDITARY; HHRH [AR]
SLC35A1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIf; CDG2F [AR]

SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIc; CDG2C [AR]
SLC35D1	SCHNECKENBECKEN DYSPLASIA; SHNKND [AR]
SLC37A4	GLYCOGEN STORAGE DISEASE Ic; GSD1C [AR] | GLYCOGEN STORAGE DISEASE Ib; GSD1B [AR]
SLC39A13	EHLERS-DANLOS SYNDROME. SPONDYLODYSPLASTIC TYPE. 3; EDSSPD3 [AR]
SLC39A4	ACRODERMATITIS ENTEROPATHICA. ZINC-DEFICIENCY TYPE; AEZ [AR]
SLC3A1	CYSTINURIA [AD]
SLC45A2	SKIN/HAIR/EYE PIGMENTATION. VARIATION IN. 5; SHEP5 [AR] | ALBINISM. OCULOCUTANEOUS. TYPE IV; OCA4 [AR]
SLC46A1	FOLATE MALABSORPTION. HEREDITARY [AR]
SLC4A1	SPHEROCYTOSIS. TYPE 4; SPH4 [AD] | RENAL TUBULAR ACIDOSIS. DISTAL. WITH HEMOLYTIC ANEMIA [AR] | RENAL TUBULAR ACIDOSIS. DISTAL. AUTOSOMAL DOMINANT [AD] | OVALOCYTOSIS. SOUTHEAST ASIAN; SAO [AD] | CRYOHYDROCYTOSIS; CHC [AD]
SLC4A11	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS; CDPD [AR] | CORNEAL ENDOTHELIAL DYSTROPHY; CHED [AR]
SLC4A4	RENAL TUBULAR ACIDOSIS. PROXIMAL. WITH OCULAR ABNORMALITIES AND MENTAL RETARDATION [AR]
SLC52A2	BROWN-VIALETTO-VAN LAERE SYNDROME 2; BVVLS2 [AR]
SLC52A3	BROWN-VIALETTO-VAN LAERE SYNDROME 1; BVVLS1 [AR] | FAZIO-LONDE DISEASE [AR]
SLC5A1	GLUCOSE/GALACTOSE MALABSORPTION; GGM [AR]
SLC5A2	RENAL GLUCOSURIA; GLYS [AD]
SLC5A5	THYROID DYSHORMONOGENESIS 1; TDH1 [AR]
SLC6A19	HARTNUP DISORDER; HND [AR] | IMINOGLYCINURIA [AR] | HYPERGLYCINURIA [AD]
SLC6A20	IMINOGLYCINURIA [AR] | HYPERGLYCINURIA [AD]
SLC6A3	TOBACCO ADDICTION. SUSCEPTIBILITY TO [Genetic factors seem to contribute only in light smokers and heavy smoker] | PARKINSONISM-DYSTONIA. INFANTILE. 1; PKDYS1 [AR]
SLC6A5	HYPEREKPLEXIA 3; HKPX3 [AD]
SLC6A8	CEREBRAL CREATINE DEFICIENCY SYNDROME 1; CCDS1 [XLR]
SLC7A7	LYSINURIC PROTEIN INTOLERANCE; LPI [AR]
SLC7A9	CYSTINURIA [AD]
SLC9A6	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CHRISTIANSON TYPE; MRXSCH [XLD]
SLCO1B1	HYPERBILIRUBINEMIA. ROTOR TYPE; HBLRR [Digenic recessive]
SLCO1B3	HYPERBILIRUBINEMIA. ROTOR TYPE; HBLRR [Digenic recessive]
SLCO2A1	HYPERTROPHIC OSTEOARTHROPATHY. PRIMARY. AUTOSOMAL RECESSIVE. 2; PHOAR2 [AR]
SLURP1	MAL DE MELEDA; MDM [AR]
SLX4	FANCONI ANEMIA. COMPLEMENTATION GROUP P; FANCP [AR]
SMARCAL1	SCHIMKE IMMUNOOSSEOUS DYSPLASIA; SIOD [AR]
SMN1	SPINAL MUSCULAR ATROPHY. TYPE III; SMA3 [AR] | SPINAL MUSCULAR ATROPHY. TYPE II; SMA2 [AR] | SPINAL MUSCULAR ATROPHY. TYPE I; SMA1 [AR] | SPINAL MUSCULAR ATROPHY. TYPE IV; SMA4 [AR]
SMOC1	MICROPHTHALMIA WITH LIMB ANOMALIES; MLA [AR]
SMOC2	DENTIN DYSPLASIA. TYPE I; DTDP1 [AR]
SMPD1	NIEMANN-PICK DISEASE. TYPE B [AR] | NIEMANN-PICK DISEASE. TYPE A [AR]
SMS	MENTAL RETARDATION. X-LINKED. SYNDROMIC. SNYDER-ROBINSON TYPE; MRXSSR [XLR]
SNAI2	PIEBALD TRAIT; PBT [AD] | WAARDENBURG SYNDROME. TYPE 2D; WS2D [AR]
SNAP29	CEREBRAL DYSGENESIS. NEUROPATHY. ICHTHYOSIS. AND PALMOPLANTAR KERATODERMA SYNDROME [AR]
SNIP1	PSYCHOMOTOR RETARDATION. EPILEPSY. AND CRANIOFACIAL DYSMORPHISM; PMRED [AR]
SNX10	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 8; OPTB8 [AR]
SOBP	MENTAL RETARDATION. ANTERIOR MAXILLARY PROTRUSION. AND STRABISMUS; MRAMS [AR]
SOD1	SPASTIC TETRAPLEGIA AND AXIAL HYPOTONIA. PROGRESSIVE; STAHP [AR] | AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1 [AD]

SOST	SCLEROSTEOSIS 1; SOST1 [AR] | CRANIODIAPHYSEAL DYSPLASIA. AUTOSOMAL DOMINANT; CDD [AD] | VAN BUCHEM DISEASE; VBCH [AR]
SOX10	WAARDENBURG SYNDROME. TYPE 2E; WS2E [AD] | WAARDENBURG SYNDROME. TYPE 4C; WS4C [AD] | PERIPHERAL DEMYELINATING NEUROPATHY. CENTRAL DYSMYELINATION. WAARDENBURG SYNDROME. AND HIRSCHSPRUNG DISEASE; PCWH [AD]
SOX18	HYPOTRICHOSIS-LYMPHEDEMA-TELANGIECTASIA-RENAL DEFECT SYNDROME; HLTRS [AD] | HYPOTRICHOSIS-LYMPHEDEMA-TELANGIECTASIA SYNDROME; HLTS [AR]
SOX3	46.XX SEX REVERSAL 3; SRXX3 [XLD] | PANHYPOPITUITARISM. X-LINKED; PHPX [X-linked]
SP110	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY; VODI [AR]
SP7	OSTEOGENESIS IMPERFECTA. TYPE XII; OI12 [AR]
SPATA7	Leber congenital amaurosis 3 604232 (3) | Retinitis pigmentosa juvenile autosomal recessive 604232 (3)
SPG11	AMYOTROPHIC LATERAL SCLEROSIS 5. JUVENILE; ALS5 [AR] | SPASTIC PARAPLEGIA 11. AUTOSOMAL RECESSIVE; SPG11 [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2X; CMT2X [AR]
SPG21	MAST SYNDROME [AR]
SPG7	SPASTIC PARAPLEGIA 7. AUTOSOMAL RECESSIVE; SPG7 [AR]
SPINK1	PANCREATITIS. HEREDITARY; PCTT [AD] | TROPICAL CALCIFIC PANCREATITIS [AD]
SPINK5	NETHERTON SYNDROME; NETH [AR]
SPINT2	DIARRHEA 3. SECRETORY SODIUM. CONGENITAL. WITH OR WITHOUT OTHER CONGENITAL ANOMALIES; DIAR3 [AR]
SPTA1	PYROPOIKILOCYTOSIS. HEREDITARY; HPP [AR] | ELLIPTOCYTOSIS 2; EL2 [AD] | SPHEROCYTOSIS. TYPE 3; SPH3 [AR]
SPTBN2	SPINOCEREBELLAR ATAXIA 5; SCA5 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 14; SCAR14 [AR]
SRD5A2	PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS; PPSH [AR]
SRD5A3	KAHRIZI SYNDROME; KHRZ [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Iq; CDG1Q [AR]
ST14	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 11; ARCI11 [AR]
ST3GAL3	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 15; EIEE15 [AR] | MENTAL RETARDATION. AUTOSOMAL RECESSIVE 12; MRT12 [AR]
ST3GAL5	SALT AND PEPPER DEVELOPMENTAL REGRESSION SYNDROME; SPDRS [AR]
STAC3	MYOPATHY. CONGENITAL. BAILEY-BLOCH; MYPBB [AR]
STAR	LIPOID CONGENITAL ADRENAL HYPERPLASIA; LCAH [AR]
STAT1	IMMUNODEFICIENCY 31C; IMD31C [AD] | IMMUNODEFICIENCY 31B; IMD31B [AR] | IMMUNODEFICIENCY 31A; IMD31A [AD]
STAT5B	Growth hormone insensitivity with immunodeficiency 245590 (3) | Leukemia acute promyelocytic somatic 102578 (3)
STIL	MICROCEPHALY 7. PRIMARY. AUTOSOMAL RECESSIVE; MCPH7 [AR]
STIM1	IMMUNODEFICIENCY 10; IMD10 [AR] | MYOPATHY. TUBULAR AGGREGATE. 1; TAM1 [AD] | STORMORKEN SYNDROME; STRMK [AD]
STK4	T-cell immunodeficiency recurrent infections autoimmunity and cardiac malformations 614868 (3)
STRA6	MICROPHTHALMIA. SYNDROMIC 9; MCOPS9 [AR]
STRADA	POLYHYDRAMNIOS. MEGALENCEPHALY. AND SYMPTOMATIC EPILEPSY; PMSE [AR]
STRC	DEAFNESS. AUTOSOMAL RECESSIVE 16; DFNB16 [AR] | DEAFNESS-INFERTILITY SYNDROME; DIS [AR]
STS	ICHTHYOSIS. X-LINKED; XLI [XLR]
STX11	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 4; FHL4 [AR]
STXBP1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 4; EIEE4 [AD]
STXBP2	Hemophagocytic lymphohistiocytosis familial 5 613101 (3)
SUCLA2	MITOCHONDRIAL DNA DEPLETION SYNDROME 5 (ENCEPHALOMYOPATHIC WITH OR WITHOUT METHYLMALONIC ACIDURIA); MTDPS5 [AR]
SUCLG1	MITOCHONDRIAL DNA DEPLETION SYNDROME 9 (ENCEPHALOMYOPATHIC TYPE WITH METHYLMALONIC ACIDURIA); MTDPS9 [AR]
SUMF1	MULTIPLE SULFATASE DEFICIENCY; MSD [AR]
SUOX	SULFITE OXIDASE DEFICIENCY. ISOLATED; ISOD [AR]
SURF1	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR] | CHARCOT-MARIE-TOOTH DISEASE. TYPE 4K; CMT4K [AR]
SYN1	EPILEPSY. X-LINKED. WITH VARIABLE LEARNING DISABILITIES AND BEHAVIOR DISORDERS [XLR]
SYNE1	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 8; SCAR8 [AR] | EMERY-DREIFUSS MUSCULAR DYSTROPHY 4. AUTOSOMAL DOMINANT; EDMD4 [AD] | ARTHROGRYPOSIS MULTIPLEX CONGENITA. MYOGENIC TYPE; AMCM [AR]

SYNE4	DEAFNESS. AUTOSOMAL RECESSIVE 76; DFNB76 [AR]
SYNJ1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 53; EIEE53 [AR] | PARKINSON DISEASE 20. EARLY-ONSET; PARK20 [AR]
SZT2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 18; EIEE18 [AR]
TACR3	HYPOGONADOTROPIC HYPOGONADISM 11 WITH OR WITHOUT ANOSMIA; HH11 [AR]
TACSTD2	CORNEAL DYSTROPHY. GELATINOUS DROP-LIKE; GDLD [AR]
TAF1	MENTAL RETARDATION. X-LINKED. SYNDROMIC 33; MRXS33 [XLR] | DYSTONIA 3. TORSION. X-LINKED; DYT3 [XLR]
TAP1	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAP2	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAPBP	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAT	TYROSINEMIA. TYPE II; TYRSN2 [AR]
TAZ	BARTH SYNDROME; BTHS [XLR]
TBC1D24	DEAFNESS. ONYCHODYSTROPHY. OSTEODYSTROPHY. MENTAL RETARDATION. AND SEIZURES SYNDROME; DOORS [AR] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 16; EIEE16 [AR] | EPILEPSY. ROLANDIC. WITH PAROXYSMAL EXERCISE-INDUCED DYSTONIA AND WRITER'S CRAMP; EPRPDC [AR] | MYOCLONIC EPILEPSY. FAMILIAL INFANTILE; FIME [AR] | DEAFNESS. AUTOSOMAL RECESSIVE 86; DFNB86 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 65; DFNA65 [AD]
TBCE	ENCEPHALOPATHY. PROGRESSIVE. WITH AMYOTROPHY AND OPTIC ATROPHY; PEAMO [AR] | KENNY-CAFFEY SYNDROME. TYPE 1; KCS1 [AR] | HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME; HRDS [AR]
TBX15	COUSIN SYNDROME [AR]
TBX19	ACTH DEFICIENCY. ISOLATED; IAD [AR]
TBX21	ASTHMA. NASAL POLYPS. AND ASPIRIN INTOLERANCE [AR]
TBXAS1	GHOSAL HEMATODIAPHYSEAL DYSPLASIA; GHDD [AR]
TCAP	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 25; CMH25 [AD] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 7; LGMDR7 [AR]
TCIRG1	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 1; OPTB1 [AR]
TCN2	TRANSCOBALAMIN II DEFICIENCY [AR]
TCTN1	JOUBERT SYNDROME 13; JBTS13 [AR]
TCTN3	JOUBERT SYNDROME 18; JBTS18 [AR] | OROFACIODIGITAL SYNDROME IV; OFD4 [AR]
TDRD7	CATARACT 36; CTRCT36 [AR]
TECPR2	SPASTIC PARAPLEGIA 49. AUTOSOMAL RECESSIVE; SPG49 [AR]
TECR	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 14; MRT14 [AR]
TECTA	DEAFNESS. AUTOSOMAL DOMINANT 12; DFNA12 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 21; DFNB21 [AR]
TF	Atransferrinemia 209300 (3)
TFR2	HEMOCHROMATOSIS. TYPE 3; HFE3 [AR]
TG	Thyroid dyshormonogenesis 3 274700 (3) | {Autoimmune thyroid disease susceptibility to 3} 608175 (3)
TGFB1	INFLAMMATORY BOWEL DISEASE. IMMUNODEFICIENCY. AND ENCEPHALOPATHY; IBDIMDE [AR] | CYSTIC FIBROSIS; CF [AR] | CAMURATI-ENGELMANN DISEASE; CAEND [AD]
TGFBI	CORNEAL DYSTROPHY. THIEL-BEHNKE TYPE; CDTB [AD] | CORNEAL DYSTROPHY. LATTICE TYPE IIIA; CDL3A [AD] | CORNEAL DYSTROPHY. GROENOUW TYPE I; CDGG1 [Autosomal dominant (5q22-q33.3)] | CORNEAL DYSTROPHY. AVELLINO TYPE; CDA [AD] | CORNEAL DYSTROPHY. EPITHELIAL BASEMENT MEMBRANE; EBMD [AD] | CORNEAL DYSTROPHY. LATTICE TYPE I; CDL1 [AD]
TGM1	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 1; ARCI1 [AR]
TGM5	PEELING SKIN SYNDROME 2; PSS2 [AR]
TH	Segawa syndrome recessive 605407 (3)
THRB	THYROID HORMONE RESISTANCE. SELECTIVE PITUITARY; PRTH [AD] | THYROID HORMONE RESISTANCE. GENERALIZED. AUTOSOMAL RECESSIVE; GRTH [AR] | THYROID HORMONE RESISTANCE. GENERALIZED. AUTOSOMAL DOMINANT; GRTH [AD]
TIMM8A	MOHR-TRANEBJAERG SYNDROME; MTS [XLR]
TIMP3	SORSBY FUNDUS DYSTROPHY; SFD [AD]
TJP2	HYPERCHOLANEMIA. FAMILIAL; FHCA [AR] | DEAFNESS. AUTOSOMAL DOMINANT 51; DFNA51 [AD] | CHOLESTASIS. PROGRESSIVE FAMILIAL INTRAHEPATIC. 4; PFIC4 [AR]

TK2	MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (MYOPATHIC TYPE); MTDPS2 [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL RECESSIVE 3; PEOB3 [AR]
TMC1	DEAFNESS. AUTOSOMAL RECESSIVE 7; DFNB7 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 36; DFNA36 [AD]
TMC6	EPIDERMODYSPLASIA VERRUCIFORMIS. SUSCEPTIBILITY TO. 1; EV1 [AR]
TMC8	EPIDERMODYSPLASIA VERRUCIFORMIS. SUSCEPTIBILITY TO. 1; EV1 [AR]
TMCO1	CRANIOFACIAL DYSMORPHISM. SKELETAL ANOMALIES. AND MENTAL RETARDATION SYNDROME; CFSMR [AR]
TMEM126A	OPTIC ATROPHY 7 WITH OR WITHOUT AUDITORY NEUROPATHY; OPA7 [AR]
TMEM138	JOUBERT SYNDROME 16; JBTS16 [AR]
TMEM216	JOUBERT SYNDROME 2; JBTS2 [AR] | MECKEL SYNDROME. TYPE 2; MKS2 [AR]
TMEM237	JOUBERT SYNDROME 14; JBTS14 [AR]
TMEM38B	Osteogenesis imperfecta type XIV 615066 (3)
TMEM67	COACH SYNDROME [AR] | BARDET-BIEDL SYNDROME 14; BBS14 [AR] | RHYNS SYNDROME; RHYNS [AR] | NEPHRONOPHTHISIS 11; NPHP11 [AR] | MECKEL SYNDROME. TYPE 3; MKS3 [AR] | JOUBERT SYNDROME 6; JBTS6 [AR]
TMEM70	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY. NUCLEAR TYPE 1; MC5DN1 [AR] | MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY. NUCLEAR TYPE 2; MC5DN2 [AR]
TMIE	DEAFNESS. AUTOSOMAL RECESSIVE 6; DFNB6 [AR]
TMPRSS15	ENTEROKINASE DEFICIENCY [AR]
TMPRSS3	DEAFNESS. AUTOSOMAL RECESSIVE 8; DFNB8 [AR]
TMPRSS6	IRON-REFRACTORY IRON DEFICIENCY ANEMIA; IRIDA [AR]
TNFRSF10B	SQUAMOUS CELL CARCINOMA. HEAD AND NECK; HNSCC [AR]
TNFRSF11A	FAMILIAL EXPANSILE OSTEOLYSIS; FEO [AD] | OSTEOPETROSIS. AUTOSOMAL RECESSIVE 7; OPTB7 [AR] | PAGET DISEASE OF BONE 2. EARLY-ONSET; PDB2 [AD]
TNFRSF11B	PAGET DISEASE OF BONE 5. JUVENILE-ONSET; PDB5 [AR]
TNFRSF13B	IMMUNODEFICIENCY. COMMON VARIABLE. 2; CVID2 [AR]
TNFRSF13C	IMMUNODEFICIENCY. COMMON VARIABLE. 4; CVID4 [AR]
TNFSF11	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 2; OPTB2 [AR]
TNNI3	CARDIOMYOPATHY. FAMILIAL RESTRICTIVE. 1; RCM1 [AD] | CARDIOMYOPATHY. DILATED. 2A; CMD2A [AR] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 7; CMH7 [AD]
TNNT1	NEMALINE MYOPATHY 5; NEM5 [AR]
TNXB	EHLERS-DANLOS SYNDROME. HYPERMOBILITY TYPE; EDSHMB [AD] | VESICOURETERAL REFLUX 8; VUR8 [AD] | EHLERS-DANLOS SYNDROME. CLASSIC-LIKE; EDSCLL [AR]
TPI1	TRIOSEPHOSPHATE ISOMERASE DEFICIENCY; TPID [AR]
TPK1	THIAMINE METABOLISM DYSFUNCTION SYNDROME 5 (EPISODIC ENCEPHALOPATHY TYPE); THMD5 [AR]
TPM3	NEMALINE MYOPATHY 1; NEM1 [AD] | MYOPATHY. CONGENITAL. WITH FIBER-TYPE DISPROPORTION; CFTD [AD]
TPO	THYROID DYSHORMONOGENESIS 2A; TDH2A [AR]
TPP1	CEROID LIPOFUSCINOSIS. NEURONAL. 2; CLN2 [AR] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 7; SCAR7 [AR]
TPRN	DEAFNESS. AUTOSOMAL RECESSIVE 79; DFNB79 [AR]
TRAPPC11	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 18; LGMDR18 [AR]
TRAPPC2	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA. X-LINKED; SEDT [XLR]
TRAPPC9	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 13; MRT13 [AR]
TREM2	POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY 1; PLOSL1 [AR]
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS; SLE [AD] | VASCULOPATHY. RETINAL. WITH CEREBRAL LEUKODYSTROPHY; RVCL [AD] | AICARDI-GOUTIERES SYNDROME 1; AGS1 [AR] | CHILBLAIN LUPUS 1; CHBL1 [AD]
TRIM32	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 8; LGMDR8 [AR] | BARDET-BIEDL SYNDROME 11; BBS11 [AR]
TRIM37	MULIBREY NANISM [AR]
TRIOBP	DEAFNESS. AUTOSOMAL RECESSIVE 28; DFNB28 [AR]
TRIP11	ACHONDROGENESIS. TYPE IA; ACG1A [AR] | ODONTOCHONDRODYSPLASIA; ODCD [AR]

TRMU	LIVER FAILURE. INFANTILE. TRANSIENT; LFIT [AR] | DEAFNESS. AMINOGLYCOSIDE-INDUCED [Mitochondrial]
TRPM1	Night blindness congenital stationary (complete) 1C autosomal recessive 613216 (3)
TRPM6	HYPOMAGNESEMIA 1. INTESTINAL; HOMG1 [AR]
TSEN54	PONTOCEREBELLAR HYPOPLASIA. TYPE 5; PCH5 [AR] | PONTOCEREBELLAR HYPOPLASIA. TYPE 2A; PCH2A [AR] | PONTOCEREBELLAR HYPOPLASIA. TYPE 4; PCH4 [AR]
TSFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3; COXPD3 [AR]
TSHB	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 4; CHNG4 [AR]
TSHR	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 1; CHNG1 [AR] | HYPERTHYROIDISM. NONAUTOIMMUNE [AD]
TSPEAR	ECTODERMAL DYSPLASIA 14. HAIR/TOOTH TYPE WITH OR WITHOUT HYPOHIDROSIS; ECTD14 [AR] | DEAFNESS. AUTOSOMAL RECESSIVE 98; DFNB98 [AR]
TSPYL1	SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME; SIDDT [AR]
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 2; MC3DN2 [AR]
TTC21B	NEPHRONOPHTHISIS 12; NPHP12 [AD] | SHORT-RIB THORACIC DYSPLASIA 4 WITH OR WITHOUT POLYDACTYLY; SRTD4 [AR]
TTC7A	GASTROINTESTINAL DEFECTS AND IMMUNODEFICIENCY SYNDROME; GIDID [AR]
TTC8	BARDET-BIEDL SYNDROME 8; BBS8 [AR] | RETINITIS PIGMENTOSA 51; RP51 [AR]
TTI2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 39; MRT39 [AR]
TTN	MYOPATHY. MYOFIBRILLAR. 9. WITH EARLY RESPIRATORY FAILURE; MFM9 [AD] | SALIH MYOPATHY; SALMY [AR] | TIBIAL MUSCULAR DYSTROPHY. TARDIVE; TMD [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 9; CMH9 [AD] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 10; LGMDR10 [AR]
TTPA	VITAMIN E. FAMILIAL ISOLATED DEFICIENCY OF; VED [AR]
TUBA8	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 8; CDCBM8 [AR]
TUFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4; COXPD4 [AR]
TULP1	LEBER CONGENITAL AMAUROSIS 15; LCA15 [AR] | RETINITIS PIGMENTOSA 14; RP14 [AR]
TUSC3	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 7; MRT7 [AR]
TWIST2	BARBER-SAY SYNDROME; BBRSAY [AD] | FOCAL FACIAL DERMAL DYSPLASIA 3. SETLEIS TYPE; FFDD3 [AR] | ABLEPHARON-MACROSTOMIA SYNDROME; AMS [AD]
TYK2	IMMUNODEFICIENCY 35; IMD35 [AR]
TYMP	MITOCHONDRIAL DNA DEPLETION SYNDROME 1 (MNGIE TYPE); MTDPS1 [AR]
TYR	ALBINISM. OCULOCUTANEOUS. TYPE IA; OCA1A [AR] | ALBINISM. OCULOCUTANEOUS. TYPE IB; OCA1B [AR] | SKIN/HAIR/EYE PIGMENTATION. VARIATION IN. 3; SHEP3 [AD]
TYROBP	POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY 1; PLOSL1 [AR]
TYRP1	ALBINISM. OCULOCUTANEOUS. TYPE III; OCA3 [AR]
UBA1	SPINAL MUSCULAR ATROPHY. X-LINKED 2; SMAX2 [XLR]
UBE2A	MENTAL RETARDATION. X-LINKED. SYNDROMIC. NASCIMENTO TYPE; MRXSN [XLR]
UBR1	JOHANSON-BLIZZARD SYNDROME; JBS [AR]
UGT1A1	BILIRUBIN. SERUM LEVEL OF. QUANTITATIVE TRAIT LOCUS 1; BILIQTL1 [Major gene effect in 11.5% of persons analyze] | GILBERT SYNDROME [AR] | CRIGLER-NAJJAR SYNDROME. TYPE I [AR] | HYPERBILIRUBINEMIA. TRANSIENT FAMILIAL NEONATAL; HBLRTFN [AR] | CRIGLER-NAJJAR SYNDROME. TYPE II [AR]
UMPS	OROTIC ACIDURIA [AR]
UNC13D	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 3; FHL3 [AR]
UNG	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 5; HIGM5 [AR]
UPB1	BETA-UREIDOPROPIONASE DEFICIENCY; UPB1D [AR]
UPF3B	MENTAL RETARDATION. X-LINKED. SYNDROMIC 14; MRXS14 [XLR]
UQCRB	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 3; MC3DN3 [AR]
UQCRQ	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 4; MC3DN4 [AR]
UROS	PORPHYRIA. CONGENITAL ERYTHROPOIETIC; CEP [AR]
USB1	POIKILODERMA WITH NEUTROPENIA; PN [AR]

USH1C	DEAFNESS. AUTOSOMAL RECESSIVE 18A; DFNB18A [AR] | USHER SYNDROME. TYPE IC; USH1C [AR]
USH1G	USHER SYNDROME. TYPE IG; USH1G [AR]
USH2A	USHER SYNDROME. TYPE IIA; USH2A [AR]
VDR	VITAMIN D-DEPENDENT RICKETS. TYPE 2A; VDDR2A [AR]
VHL	VON HIPPEL-LINDAU SYNDROME; VHLS [AD] | ERYTHROCYTOSIS. FAMILIAL. 2. AUTOSOMAL RECESSIVE; ECYT2 [AR] | PHEOCHROMOCYTOMA [AD]
VIPAS39	ARTHROGRYPOSIS. RENAL DYSFUNCTION. AND CHOLESTASIS 2; ARCS2 [AR]
VLDLR	CEREBELLAR ATAXIA. MENTAL RETARDATION. AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 [AR]
VPS13A	CHOREOACANTHOCYTOSIS; CHAC [AR]
VPS13B	COHEN SYNDROME; COH1 [AR]
VPS33B	ARTHROGRYPOSIS. RENAL DYSFUNCTION. AND CHOLESTASIS 1; ARCS1 [AR]
VPS37A	SPASTIC PARAPLEGIA 53. AUTOSOMAL RECESSIVE; SPG53 [AR]
VPS45	NEUTROPENIA. SEVERE CONGENITAL. 5. AUTOSOMAL RECESSIVE; SCN5 [AR]
VRK1	PONTOCEREBELLAR HYPOPLASIA. TYPE 1A; PCH1A [AR]
VSX2	Microphthalmia with coloboma 3 610092 (3) | Microphthalmia isolated 2 610093 (3)
VWF	VON WILLEBRAND DISEASE. TYPE 2; VWD2 [AD] | VON WILLEBRAND DISEASE. TYPE 3; VWD3 [AR] | VON WILLEBRAND DISEASE. TYPE 1; VWD1 [AD]
WAS	NEUTROPENIA. SEVERE CONGENITAL. X-LINKED; SCNX [XLR] | THROMBOCYTOPENIA 1; THC1 [XLR] | WISKOTT-ALDRICH SYNDROME; WAS [XLR]
WDR19	NEPHRONOPHTHISIS 13; NPHP13 [AR] | SENIOR-LOKEN SYNDROME 8; SLSN8 [AR] | SHORT-RIB THORACIC DYSPLASIA 5 WITH OR WITHOUT POLYDACTYLY; SRTD5 [AR] | CRANIOECTODERMAL DYSPLASIA 4; CED4 [AR]
WDR35	CRANIOECTODERMAL DYSPLASIA 2; CED2 [AR] | SHORT-RIB THORACIC DYSPLASIA 7 WITH OR WITHOUT POLYDACTYLY; SRTD7 [AR]
WDR62	MICROCEPHALY 2. PRIMARY. AUTOSOMAL RECESSIVE. WITH OR WITHOUT CORTICAL MALFORMATIONS; MCPH2 [AR]
WDR72	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA3; AI2A3 [AR]
WDR81	CEREBELLAR ATAXIA. MENTAL RETARDATION. AND DYSEQUILIBRIUM SYNDROME 2; CAMRQ2 [AR] | HYDROCEPHALUS. CONGENITAL. 3. WITH BRAIN ANOMALIES; HYC3 [AR]
WFS1	CATARACT 41; CTRCT41 [AD] | WOLFRAM SYNDROME 1; WFS1 [AR] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | WOLFRAM-LIKE SYNDROME. AUTOSOMAL DOMINANT; WFSL [AD] | DEAFNESS. AUTOSOMAL DOMINANT 6; DFNA6 [AD]
WNK1	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE IIA; HSAN2A [AR] | PSEUDOHYPOALDOSTERONISM. TYPE IIC; PHA2C [AD]
WNT1	OSTEOGENESIS IMPERFECTA. TYPE XV; OI15 [AR]
WNT10A	ODONTOONYCHODERMAL DYSPLASIA; OODD [AR] | SCHOPF-SCHULZ-PASSARGE SYNDROME; SSPS [AR] | TOOTH AGENESIS. SELECTIVE. 4; STHAG4 [AD]
WNT10B	SPLIT-HAND/FOOT MALFORMATION 6; SHFM6 [AR] | TOOTH AGENESIS. SELECTIVE. 8; STHAG8 [AD]
WNT4	46.XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS. ADRENALS. AND LUNGS; SERKAL [AR] | MULLERIAN APLASIA AND HYPERANDROGENISM [AD]
WNT7A	FIBULAR APLASIA OR HYPOPLASIA. FEMORAL BOWING AND POLY-. SYN-. AND OLIGODACTYLY [AR] | ULNA AND FIBULA. ABSENCE OF. WITH SEVERE LIMB DEFICIENCY [AR]
WRAP53	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 3; DKCB3 [AR]
WRN	WERNER SYNDROME; WRN [AR]
WT1	MESOTHELIOMA. MALIGNANT; MESOM [Somatic mutation] | WILMS TUMOR 1; WT1 [AD] | DENYS-DRASH SYNDROME; DDS [AD] | NEPHROTIC SYNDROME. TYPE 4; NPHS4 [AD] | WILMS TUMOR. ANIRIDIA. GENITOURINARY ANOMALIES. AND MENTAL RETARDATION SYNDROME; WAGR [AD] | FRASIER SYNDROME [AD]
XDH	XANTHINURIA. TYPE I; XAN1 [AR]
XPA	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP A; XPA [AR]
XPC	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP C; XPC [AR]
XPNPEP3	NEPHRONOPHTHISIS-LIKE NEPHROPATHY 1; NPHPL1 [AR]
YARS2	MYOPATHY. LACTIC ACIDOSIS. AND SIDEROBLASTIC ANEMIA 2; MLASA2 [AR]
ZAP70	AUTOIMMUNE DISEASE. MULTISYSTEM. INFANTILE-ONSET. 2; ADMIO2 [AR] | IMMUNODEFICIENCY 48; IMD48 [AR]
ZBTB16	SKELETAL DEFECTS. GENITAL HYPOPLASIA. AND MENTAL RETARDATION [AR]

ZBTB24	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 2; ICF2 [AR]
ZFYVE26	SPASTIC PARAPLEGIA 15. AUTOSOMAL RECESSIVE; SPG15 [AR]
ZIC3	VACTERL ASSOCIATION. X-LINKED. WITH OR WITHOUT HYDROCEPHALUS; VACTERLX [XLR] | HETEROTAXY. VISCERAL. 1. X-LINKED; HTX1 [XLR]
ZMPSTE24	RESTRICTIVE DERMOPATHY. LETHAL [AR] | MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY; MADB [AR]
ZNF335	MICROCEPHALY 10. PRIMARY. AUTOSOMAL RECESSIVE; MCPH10 [AR]
ZNF469	BRITTLE CORNEA SYNDROME 1; BCS1 [AR]