Gene	Disease
A2M	ALZHEIMER DISEASE; AD [AD]
A4GALT	NOR polyagglutination syndrome 111400 (3) | [Blood group P1Pk system P(2) phenotype] 111400 (3) | [Blood group P1Pk system p phenotype] 111400 (3)
AAAS	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME; AAAS [AR]
AAGAB	PALMOPLANTAR KERATODERMA. PUNCTATE TYPE IA; PPKP1A [AD]
AARS1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2N; CMT2N [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 29; EIEE29 [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]
ABCA1	TANGIER DISEASE; TGD [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]
ABCB6	MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 7; MCOPCB7 [AD] | DYSCHROMATOSIS UNIVERSALIS HEREDITARIA 3; DUH3 [AD] | PSEUDOHYPERKALEMIA. FAMILIAL. 2. DUE TO RED CELL LEAK; PSHK2 [AD]
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]
ABCC9	CANTU SYNDROME [AD] | ATRIAL FIBRILLATION. FAMILIAL. 12; ATFB12 [AD] | CARDIOMYOPATHY. DILATED. 1O; CMD1O [AD]
ABCD1	ADRENOLEUKODYSTROPHY; ALD [XLR]
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA. cblJ TYPE; MAHCJ [AR]
ABCG2	URIC ACID CONCENTRATION. SERUM. QUANTITATIVE TRAIT LOCUS 1; UAQTL1 [AD]
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]
ABL1	LEUKEMIA. CHRONIC MYELOID; CML [Somatic mutation] | CONGENITAL HEART DEFECTS AND SKELETAL MALFORMATIONS SYNDROME; CHDSKM [AD]
ACACA	ACETYL-CoA CARBOXYLASE DEFICIENCY; ACACAD [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]
ACOX2	BILE ACID SYNTHESIS DEFECT. CONGENITAL. 6; CBAS6 [AR]
ACP4	AMELOGENESIS IMPERFECTA. TYPE IJ; AI1J [AR]
ACP5	SPONDYLOENCHONDRODYSPLASIA WITH IMMUNE DYSREGULATION; SPENCDI [AR]
ACSF3	Combined malonic and methylmalonic aciduria 614265 (3)
ACSL4	MENTAL RETARDATION. X-LINKED 63; MRX63 [XLD]
ACSL6	Myelodysplastic syndrome (3) | Myelogenous leukemia acute (3)
ACSM3	{?Hypertension essential} (1)
ACTA1	NEMALINE MYOPATHY 3; NEM3 [AD] | MYOPATHY. SCAPULOHUMEROPERONEAL; SHPM [AD] | MYOPATHY. CONGENITAL. WITH FIBER-TYPE DISPROPORTION; CFTD [AD]
ACTA2	AORTIC ANEURYSM. FAMILIAL THORACIC 6; AAT6 [AD] | MULTISYSTEMIC SMOOTH MUSCLE DYSFUNCTION SYNDROME; MSMDS [AD]
ACTB	DYSTONIA. JUVENILE-ONSET; DJO [AD] | BARAITSER-WINTER SYNDROME 1; BRWS1 [AD]
ACTC1	ATRIAL SEPTAL DEFECT 5; ASD5 [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 11; CMH11 [AD] | CARDIOMYOPATHY. DILATED. 1R; CMD1R [AD]
ACTG1	BARAITSER-WINTER SYNDROME 2; BRWS2 [AD] | DEAFNESS. AUTOSOMAL DOMINANT 20; DFNA20 [AD]
ACTG2	VISCERAL MYOPATHY; VSCM [AD]
ACTL6B	INTELLECTUAL DEVELOPMENTAL DISORDER WITH SEVERE SPEECH AND AMBULATION DEFECTS; IDDSSAD [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 76; EIEE76 [AR]
ACTN1	BLEEDING DISORDER. PLATELET-TYPE. 15; BDPLT15 [AD]
ACTN2	MYOPATHY. CONGENITAL. WITH STRUCTURED CORES AND Z-LINE ABNORMALITIES; MYOCOZ [AD] | CARDIOMYOPATHY. DILATED. 1AA. WITH OR WITHOUT LEFT VENTRICULAR NONCOMPACTION; CMD1AA [AD] | MYOPATHY. DISTAL. 6. ADULT-ONSET. AUTOSOMAL DOMINANT; MPD6 [AD]
ACTN4	FOCAL SEGMENTAL GLOMERULOSCLEROSIS 1; FSGS1 [AD]
ACVR1	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA; FOP [AD]
ACVR1B	Pancreatic cancer somatic (3)
ACVR2B	Heterotaxy visceral 4 autosomal 613751 (3)
ACVRL1	TELANGIECTASIA. HEREDITARY HEMORRHAGIC. TYPE 2; HHT2 [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]
ADA2	VASCULITIS. AUTOINFLAMMATION. IMMUNODEFICIENCY. AND HEMATOLOGIC DEFECTS SYNDROME; VAIHS [AR] | SNEDDON SYNDROME; SNDNS [AR]
ADAM10	RETICULATE ACROPIGMENTATION OF KITAMURA; RAK [AD]
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]
ADAMTS3	HENNEKAM LYMPHANGIECTASIA-LYMPHEDEMA SYNDROME 3; HKLLS3 [AR]
ADAMTSL2	GELEOPHYSIC DYSPLASIA 1; GPHYSD1 [AR]
ADAMTSL4	ECTOPIA LENTIS 2. ISOLATED. AUTOSOMAL RECESSIVE; ECTOL2 [AR] | ECTOPIA LENTIS ET PUPILLAE [AR]
ADAR	DYSCHROMATOSIS SYMMETRICA HEREDITARIA; DSH [AD] | AICARDI-GOUTIERES SYNDROME 6; AGS6 [AR]
ADAT3	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 36; MRT36 [AR]
ADCY5	DYSKINESIA. FAMILIAL. WITH FACIAL MYOKYMIA; FDFM [AD]
ADD3	CEREBRAL PALSY. SPASTIC QUADRIPLEGIC. 3; CPSQ3 [AR]

ADGRE2	VIBRATORY URTICARIA; VBU [AD]
ADGRG1	POLYMICROGYRIA. BILATERAL FRONTOPARIETAL; BFPP [AR]
ADGRG2	VAS DEFERENS. CONGENITAL BILATERAL APLASIA OF. X-LINKED; CBAVDX [X-linked]
ADGRG6	LETHAL CONGENITAL CONTRACTURE SYNDROME 9; LCCS9 [AR]
ADGRV1	USHER SYNDROME. TYPE IIC; USH2C [AR] | FEBRILE SEIZURES. FAMILIAL. 4; FEB4 [AD]
ADIPOQ	Adiponectin deficiency 612556 (3)
ADK	HYPERMETHIONINEMIA DUE TO ADENOSINE KINASE DEFICIENCY [AR]
ADNP	HELSMOORTEL-VAN DER AA SYNDROME; HVDAS [AD]
ADPRS	NEURODEGENERATION. CHILDHOOD-ONSET. STRESS-INDUCED. WITH VARIABLE ATAXIA AND SEIZURES; CONDSIAS [AR]
ADRB2	ASTHMA. SUSCEPTIBILITY TO [Autosomal dominant vs. multifactoria] | OBESITY [AD]
ADSL	ADENYLOSUCCINASE DEFICIENCY; ADSLD [AR]
ADSS1	MYOPATHY. DISTAL. 5; MPD5 [AR]
AEBP1	EHLERS-DANLOS SYNDROME. CLASSIC-LIKE. 2; EDSCLL2 [AR]
AFF2	MENTAL RETARDATION. X-LINKED. ASSOCIATED WITH FRAGILE SITE FRAXE [XLR]
AFF4	CHOPS SYNDROME; CHOPS [AD]
AFG3L2	SPINOCEREBELLAR ATAXIA 28; SCA28 [AD] | SPASTIC ATAXIA 5. AUTOSOMAL RECESSIVE; SPAX5 [AR]
AFP	ALPHA-FETOPROTEIN. HEREDITARY PERSISTENCE OF; HPAFP [AD] | ALPHA-FETOPROTEIN DEFICIENCY; AFPD [AR]
AGA	ASPARTYLGLUCOSAMINURIA; AGU [AR]
AGBL1	CORNEAL DYSTROPHY. FUCHS ENDOTHELIAL. 8; FECD8 [AD]
AGBL5	RETINITIS PIGMENTOSA 75; RP75 [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]
AGTPBP1	NEURODEGENERATION. CHILDHOOD-ONSET. WITH CEREBELLAR ATROPHY; CONDCA [AR]
AGTR1	RENAL TUBULAR DYSGENESIS; RTD [AR] | HYPERTENSION. ESSENTIAL [Multifactorial]
AGXT	HYPEROXALURIA. PRIMARY. TYPE I; HP1 [AR]
AHCY	HYPERMETHIONINEMIA WITH S-ADENOSYLHOMOCYSTEINE HYDROLASE DEFICIENCY [AR]
AHDC1	XIA-GIBBS SYNDROME; XIGIS [AD]
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]
AIMP2	LEUKODYSTROPHY. HYPOMYELINATING. 17; HLD17 [AR]
AIP	PITUITARY ADENOMA 1. MULTIPLE TYPES; PITA1 [AD]
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]
AKT1	OVARIAN CANCER [Somatic mutation] | BREAST CANCER [AD] | COLORECTAL CANCER; CRC [Somatic mutation] | PROTEUS SYNDROME [Somatic mutation]
AKT2	HYPOINSULINEMIC HYPOGLYCEMIA WITH HEMIHYPERTROPHY; HIHGHH [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
AKT3	MEGALENCEPHALY-POLYMICROGYRIA-POLYDACTYLY-HYDROCEPHALUS SYNDROME 2; MPPH2 [AD]
ALAD	PORPHYRIA. ACUTE HEPATIC [AR]
ALAS2	PROTOPORPHYRIA. ERYTHROPOIETIC. X-LINKED; XLEPP [X-linked] | ANEMIA. SIDEROBLASTIC. 1; SIDBA1 [XLR]
ALB	Analbuminemia 616000 (3) | [Dysalbuminemic hyperthyroxinemia] 615999 (3)
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]
ALDH2	ALDEHYDE DEHYDROGENASE 2 FAMILY; ALDH2 [AD] | ALCOHOL SENSITIVITY. ACUTE [AD]
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]
ALG12	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ig; CDG1G [AR]
ALG13	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 36; EIEE36 [XLD]
ALG2	MYASTHENIC SYNDROME. CONGENITAL. 14; CMS14 [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ii; CDG1I [AR]
ALG3	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Id; CDG1D [AR]
ALG6	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ic; CDG1C [AR]
ALG8	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ih; CDG1H [AR] | POLYCYSTIC LIVER DISEASE 3 WITH OR WITHOUT KIDNEY CYSTS; PCLD3 [AD]
ALG9	GILLESSEN-KAESBACH-NISHIMURA SYNDROME; GIKANIS [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Il; CDG1L [AR]
ALKBH8	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 71; MRT71 [AR]
ALMS1	ALSTROM SYNDROME; ALMS [AR]
ALOX12B	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 2; ARCI2 [AR]
ALOXE3	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 3; ARCI3 [AR]
ALPK3	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC 27; CMH27 [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]
ALX4	PARIETAL FORAMINA 2; PFM2 [AD] | CRANIOSYNOSTOSIS 5. SUSCEPTIBILITY TO; CRS5 [AD] | FRONTONASAL DYSPLASIA 2; FND2 [AR]
AMACR	ALPHA-METHYLACYL-CoA RACEMASE DEFICIENCY; AMACRD [AR] | BILE ACID SYNTHESIS DEFECT. CONGENITAL. 4; CBAS4 [AR]
AMBN	AMELOGENESIS IMPERFECTA. TYPE IF; AI1F [AR]
AMELX	AMELOGENESIS IMPERFECTA. TYPE IE; AI1E [XLD]
AMER1	OSTEOPATHIA STRIATA WITH CRANIAL SCLEROSIS; OSCS [XLD]
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]

AMMECR1	MIDFACE HYPOPLASIA. HEARING IMPAIRMENT. ELLIPTOCYTOSIS. AND NEPHROCALCINOSIS; MFHIEN [XLR]
AMN	MEGALOBLASTIC ANEMIA 1 [AR]
AMPD1	MYOPATHY DUE TO MYOADENYLATE DEAMINASE DEFICIENCY; MMDD [AR]
AMPD2	SPASTIC PARAPLEGIA 63. AUTOSOMAL RECESSIVE; SPG63 [AR] | PONTOCEREBELLAR HYPOPLASIA. TYPE 9; PCH9 [AR]
AMT	GLYCINE ENCEPHALOPATHY; GCE [AR]
ANAPC1	ROTHMUND-THOMSON SYNDROME. TYPE 1; RTS1 [AR]
ANG	Amyotrophic lateral sclerosis 9 611895 (3)
ANGPTL3	HYPOBETALIPOPROTEINEMIA. FAMILIAL. 2; FHBL2 [AR]
ANGPTL4	PLASMA TRIGLYCERIDE LEVEL QUANTITATIVE TRAIT LOCUS; TGQTL [AD]
ANK1	SPHEROCYTOSIS. TYPE 1; SPH1 [AD]
ANK2	CARDIAC ARRHYTHMIA. ANKYRIN-B-RELATED [AD]
ANKH	CRANIOMETAPHYSEAL DYSPLASIA. AUTOSOMAL DOMINANT; CMDD [AD] | CHONDROCALCINOSIS 2; CCAL2 [AD]
ANKLE2	MICROCEPHALY 16. PRIMARY. AUTOSOMAL RECESSIVE; MCPH16 [AR]
ANKRD11	KBG SYNDROME; KBGS [AD]
ANKRD26	THROMBOCYTOPENIA 2; THC2 [AD]
ANKS6	NEPHRONOPHTHISIS 16; NPHP16 [AR]
ANLN	FOCAL SEGMENTAL GLOMERULOSCLEROSIS 8; FSGS8 [AD]
ANO10	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 10; SCAR10 [AR]
ANO3	DYSTONIA 24; DYT24 [AD]
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]
ANOS1	HYPOGONADOTROPIC HYPOGONADISM 1 WITH OR WITHOUT ANOSMIA; HH1 [XLR]
ANTXR2	HYALINE FIBROMATOSIS SYNDROME; HFS [AR]
ANXA11	AMYOTROPHIC LATERAL SCLEROSIS 23; ALS23 [AD]
AP1S1	MENTAL RETARDATION. ENTEROPATHY. DEAFNESS. PERIPHERAL NEUROPATHY. ICHTHYOSIS. AND KERATODERMA; MEDNIK [AR]
AP1S2	PETTIGREW SYNDROME; PGS [XLR]
AP2M1	INTELLECTUAL DEVELOPMENTAL DISORDER 60 WITH SEIZURES; MRD60 [AD]
AP2S1	HYPOCALCIURIC HYPERCALCEMIA. FAMILIAL. TYPE III; HHC3 [AD]
AP3B1	HERMANSKY-PUDLAK SYNDROME 2; HPS2 [AR]
AP3B2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 48; EIEE48 [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]
APC	DESMOID DISEASE. HEREDITARY; DESMD [AD] | FAMILIAL ADENOMATOUS POLYPOSIS 1; FAP1 [AD] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation]
APCDD1	HYPOTRICHOSIS 1; HYPT1 [AD]
APCS	{?Amyloidosis secondary susceptibility to} (1)
APOA1	AMYLOIDOSIS. FAMILIAL VISCERAL [AD]
APOA2	HYPERCHOLESTEROLEMIA. FAMILIAL. 1; FHCL1 [AD]
APOA5	HYPERLIPOPROTEINEMIA. TYPE V [AD] | HYPERTRIGLYCERIDEMIA. FAMILIAL [AD]
APOB	HYPERCHOLESTEROLEMIA. FAMILIAL. 2; FCHL2 [AD] | HYPOBETALIPOPROTEINEMIA. FAMILIAL. 1; FHBL1 [AR]

APOC2	APOLIPOPROTEIN C-II DEFICIENCY [AR]
APOC3	Apolipoprotein C-III deficiency 614028 (3)
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]
AQP5	PALMOPLANTAR KERATODERMA. BOTHNIAN TYPE; PPKB [AD]
AR	PROSTATE CANCER [Somatic mutation]
ARCN1	SHORT STATURE. RHIZOMELIC. WITH MICROCEPHALY. MICROGNATHIA. AND DEVELOPMENTAL DELAY; SRMMD [AD]
ARF1	PERIVENTRICULAR NODULAR HETEROTOPIA 8; PVNH8 [AD]
ARFGEF2	PERIVENTRICULAR HETEROTOPIA WITH MICROCEPHALY. AUTOSOMAL RECESSIVE; ARPHM [AR]
ARG1	ARGININEMIA [AR]
ARHGAP26	JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML [AD]
ARHGAP31	ADAMS-OLIVER SYNDROME 1; AOS1 [AD]
ARHGDIA	NEPHROTIC SYNDROME. TYPE 8; NPHS8 [AR]
ARHGEF18	RETINITIS PIGMENTOSA 78; RP78 [AR]
ARHGEF9	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 8; EIEE8 [XLR]
ARID1A	COFFIN-SIRIS SYNDROME 2; CSS2 [AD]
ARID1B	COFFIN-SIRIS SYNDROME 1; CSS1 [AD]
ARID2	COFFIN-SIRIS SYNDROME 6; CSS6 [AD]
ARL13B	JOUBERT SYNDROME 8; JBTS8 [AR]
ARL2BP	RETINITIS PIGMENTOSA 82 WITH OR WITHOUT SITUS INVERSUS; RP82 [AR]
ARL3	RETINITIS PIGMENTOSA 83; RP83 [AD] | JOUBERT SYNDROME 35; JBTS35 [AR]
ARL6	BARDET-BIEDL SYNDROME 1; BBS1 [AR] | BARDET-BIEDL SYNDROME 3; BBS3 [AR]
ARMC2	SPERMATOGENIC FAILURE 38; SPGF38 [AR]
ARMC4	CILIARY DYSKINESIA. PRIMARY. 23; CILD23 [AR]
ARMC5	ACTH-INDEPENDENT MACRONODULAR ADRENAL HYPERPLASIA 2; AIMAH2 [AD]
ARMC9	JOUBERT SYNDROME 30; JBTS30 [AR]
ARPC1B	PLATELET ABNORMALITIES WITH EOSINOPHILIA AND IMMUNE-MEDIATED INFLAMMATORY DISEASE; PLTEID [AR]
ARR3	MYOPIA 26. X-LINKED. FEMALE-LIMITED; MYP26 [X-linked (female limited)]
ARSA	METACHROMATIC LEUKODYSTROPHY; MLD [AR]
ARSB	MUCOPOLYSACCHARIDOSIS. TYPE VI; MPS6 [AR]
ARSG	USHER SYNDROME. TYPE IV; USH4 [AR]
ARSL	CHONDRODYSPLASIA PUNCTATA 1. X-LINKED RECESSIVE; CDPX1 [XLR]
ARV1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 38; EIEE38 [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]
ASB10	Glaucoma 1 open angle F 603383 (3)
ASCC1	SPINAL MUSCULAR ATROPHY WITH CONGENITAL BONE FRACTURES 2; SMABF2 [AR]
ASCL1	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD]

ASH1L	MENTAL RETARDATION. AUTOSOMAL DOMINANT 52; MRD52 [AD]
ASL	ARGININOSUCCINIC ACIDURIA [AR]
ASNS	ASPARAGINE SYNTHETASE DEFICIENCY; ASNSD [AR]
ASPA	CANAVAN DISEASE [AR]
ASPH	FACIAL DYSMORPHISM. LENS DISLOCATION. ANTERIOR SEGMENT ABNORMALITIES. AND SPONTANEOUS FILTERING BLEBS; FDLAB [AR]
ASPM	MICROCEPHALY 5. PRIMARY. AUTOSOMAL RECESSIVE; MCPH5 [AR]
ASPSCR1	Alveolar soft-part sarcoma 606243 (3)
ASS1	CITRULLINEMIA. CLASSIC [AR]
ASXL1	BOHRING-OPITZ SYNDROME; BOPS [AD]
ASXL2	SHASHI-PENA SYNDROME; SHAPNS [AD]
ASXL3	BAINBRIDGE-ROPERS SYNDROME; BRPS [AD]
ATAD1	HYPEREKPLEXIA 4; HKPX4 [AR]
ATAD3A	HAREL-YOON SYNDROME; HAYOS [AD]
ATCAY	CEREBELLAR ATAXIA. CAYMAN TYPE; ATCAY [AR]
ATF6	ACHROMATOPSIA 7; ACHM7 [AR]
ATIC	AICAR TRANSFORMYLASE/IMP CYCLOHYDROLASE DEFICIENCY [AR]
ATL1	SPASTIC PARAPLEGIA 3. AUTOSOMAL DOMINANT; SPG3A [AD] | NEUROPATHY. HEREDITARY SENSORY. TYPE ID; HSN1D [AD]
ATL3	NEUROPATHY. HEREDITARY SENSORY. TYPE IF; HSN1F [AD]
ATM	BREAST CANCER [AD] | ATAXIA-TELANGIECTASIA; AT [AR]
ATN1	CONGENITAL HYPOTONIA. EPILEPSY. DEVELOPMENTAL DELAY. AND DIGITAL ANOMALIES; CHEDDA [AD] | DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY; DRPLA [AD]
ATOH7	PERSISTENT HYPERPLASTIC PRIMARY VITREOUS. AUTOSOMAL RECESSIVE; PHPVAR [AR]
ATP13A2	SPASTIC PARAPLEGIA 78. AUTOSOMAL RECESSIVE; SPG78 [AR] | KUFOR-RAKEB SYNDROME; KRS [AR]
ATP1A1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2DD; CMT2DD [AD] | HYPOMAGNESEMIA. SEIZURES. AND MENTAL RETARDATION 2; HOMGSMR2 [AD]
ATP1A2	MIGRAINE. FAMILIAL HEMIPLEGIC. 2; FHM2 [AD] | ALTERNATING HEMIPLEGIA OF CHILDHOOD 1; AHC1 [AD]
ATP1A3	ALTERNATING HEMIPLEGIA OF CHILDHOOD 2; AHC2 [AD] | DYSTONIA 12; DYT12 [AD] | CEREBELLAR ATAXIA. AREFLEXIA. PES CAVUS. OPTIC ATROPHY. AND SENSORINEURAL HEARING LOSS; CAPOS [AD]
ATP2A1	BRODY MYOPATHY [AR]
ATP2A2	DARIER-WHITE DISEASE; DAR [AD] | ACROKERATOSIS VERRUCIFORMIS; AKV [AD]
ATP2C1	BENIGN CHRONIC PEMPHIGUS; BCPM [AD]
ATP5F1D	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY. NUCLEAR TYPE 5; MC5DN5 [AR]
ATP6AP1	IMMUNODEFICIENCY 47; IMD47 [XLR]
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)
ATP6V1A	EPILEPTIC ENCEPHALOPATHY. INFANTILE OR EARLY CHILDHOOD. 3; IECEE3 [AD] | CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IID; ARCL2D [AR]
ATP6V1B1	RENAL TUBULAR ACIDOSIS. DISTAL. WITH PROGRESSIVE NERVE DEAFNESS [AR]
ATP6V1B2	ZIMMERMANN-LABAND SYNDROME 2; ZLS2 [AD] | DEAFNESS. CONGENITAL. WITH ONYCHODYSTROPHY. AUTOSOMAL DOMINANT; DDOD [AD]
ATP6V1E1	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IIC; ARCL2C [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]
ATXN1	SPINOCEREBELLAR ATAXIA 1; SCA1 [AD]
ATXN10	SPINOCEREBELLAR ATAXIA 10; SCA10 [AD]
ATXN2	SPINOCEREBELLAR ATAXIA 2; SCA2 [AD] | PARKINSON DISEASE. LATE-ONSET; PD [AD]
ATXN3	MACHADO-JOSEPH DISEASE; MJD [AD]
ATXN7	SPINOCEREBELLAR ATAXIA 7; SCA7 [AD]
ATXN8	SPINOCEREBELLAR ATAXIA 8; SCA8 [AD]
ATXN8OS	PARKINSON DISEASE. LATE-ONSET; PD [AD] | SPINOCEREBELLAR ATAXIA 8; SCA8 [AD]
AUH	3-METHYLGLUTACONIC ACIDURIA. TYPE I; MGCA1 [AR]
AURKC	SPERMATOGENIC FAILURE 5; SPGF5 [AR]
AUTS2	MENTAL RETARDATION. AUTOSOMAL DOMINANT 26; MRD26 [AD]
AVIL	NEPHROTIC SYNDROME. TYPE 21; NPHS21 [AR]
AVP	DIABETES INSIPIDUS. NEUROHYPOPHYSEAL [AD]
AVPR2	DIABETES INSIPIDUS. NEPHROGENIC. X-LINKED [XLR] | NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS; NSIAD [XLR]
AXIN1	HEPATOCELLULAR CARCINOMA [Somatic mutation]
AXIN2	OLIGODONTIA-COLORECTAL CANCER SYNDROME; ODCRCS [AD] | COLORECTAL CANCER; CRC [Somatic mutation]
B2M	IMMUNODEFICIENCY 43; IMD43 [AR] | AMYLOIDOSIS. FAMILIAL VISCERAL [AD]
B3GALNT2	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 11; MDDGA11 [AR]
B3GALT6	EHLERS-DANLOS SYNDROME. SPONDYLODYSPLASTIC TYPE. 2; EDSSPD2 [AR] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY. TYPE 1. WITH OR WITHOUT FRACTURES; SEMDJL1 [AR]
B3GAT3	MULTIPLE JOINT DISLOCATIONS. SHORT STATURE. AND CRANIOFACIAL DYSMORPHISM WITH OR WITHOUT CONGENITAL HEART DEFECTS; JDSCD [AR]
B3GLCT	PETERS-PLUS SYNDROME; PTRPLS [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]
B4GAT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 13; MDDGA13 [AR]
B9D1	JOUBERT SYNDROME 27; JBTS27 [AR] | MECKEL SYNDROME. TYPE 9; MKS9 [AR]
B9D2	MECKEL SYNDROME. TYPE 10; MKS10 [AR]
BAAT	HYPERCHOLANEMIA. FAMILIAL; FHCA [AR]
BACH2	IMMUNODEFICIENCY 60; IMD60 [AD]
BAG3	CARDIOMYOPATHY. DILATED. 1HH; CMD1HH [AD] | MYOPATHY. MYOFIBRILLAR. 6; MFM6 [AD]
BANF1	NESTOR-GUILLERMO PROGERIA SYNDROME; NGPS [AR]
BAP1	TUMOR PREDISPOSITION SYNDROME; TPDS [AD]
BAX	COLORECTAL CANCER; CRC [Somatic mutation]
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]

BCAP31	DEAFNESS. DYSTONIA. AND CEREBRAL HYPOMYELINATION; DDCH [XLR]
BCHE	BUTYRYLCHOLINESTERASE; BCHE [AR]
BCKDHA	MAPLE SYRUP URINE DISEASE; MSUD [AR]
BCKDHB	MAPLE SYRUP URINE DISEASE; MSUD [AR]
BCKDK	Branched-chain ketoacid dehydrogenase kinase deficiency 614923 (3)
BCL10	IMMUNODEFICIENCY 37; IMD37 [AR] | MESOTHELIOMA. MALIGNANT; MESOM [Somatic mutation] | TESTICULAR GERM CELL TUMOR; TGCT [Somatic mutation]
BCL11A	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 5; HBFQTL5 [AD] | INTELLECTUAL DEVELOPMENTAL DISORDER WITH PERSISTENCE OF FETAL HEMOGLOBIN [AD]
BCL11B	IMMUNODEFICIENCY 49; IMD49 [AD] | INTELLECTUAL DEVELOPMENTAL DISORDER WITH SPEECH DELAY. DYSMORPHIC FACIES. AND T-CELL ABNORMALITIES; IDDSFTA [AD]
BCL2	Leukemia/lymphoma B-cell 2 (3)
BCL3	Leukemia/lymphoma B-cell 3 (2)
BCL6	Lymphoma B-cell (2)
BCL7A	B-cell non-Hodgkin lymphoma high-grade (3)
BCOR	MICROPHTHALMIA. SYNDROMIC 2; MCOPS2 [XLD]
BCORL1	SHUKLA-VERNON SYNDROME; SHUVER [XLR]
BCR	LEUKEMIA. CHRONIC MYELOID; CML [Somatic mutation]
BCS1L	GRACILE SYNDROME [AR] | LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 1; MC3DN1 [AR] | BJORNSTAD SYNDROME; BJS [AR]
BEAN1	SPINOCEREBELLAR ATAXIA 31; SCA31 [AD]
BEST1	MACULAR DYSTROPHY. VITELLIFORM. 2; VMD2 [AD] | VITREORETINOCHOROIDOPATHY; VRCP [AD]
BFSP1	CATARACT 33. MULTIPLE TYPES; CTRCT33 [AR]
BFSP2	CATARACT 12. MULTIPLE TYPES; CTRCT12 [AD]
BGN	MEESTER-LOEYS SYNDROME; MRLS [X-linked] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. X-LINKED; SEMDX [XLR]
BHLHA9	SYNDACTYLY. MESOAXIAL SYNOSTOTIC. WITH PHALANGEAL REDUCTION; MSSD [AR] | CAMPTOSYNPOLYDACTYLY. COMPLEX; CCSPD [AR]
BICD2	SPINAL MUSCULAR ATROPHY. LOWER EXTREMITY-PREDOMINANT. 2A. CHILDHOOD ONSET. AUTOSOMAL DOMINANT; SMALED2A [AD] | SPINAL MUSCULAR ATROPHY. LOWER EXTREMITY-PREDOMINANT. 2B. PRENATAL ONSET. AUTOSOMAL DOMINANT; SMALED2B [AD]
BIN1	MYOPATHY. CENTRONUCLEAR. 2; CNM2 [AR]
BLK	MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 11; MODY11 [AD]
BLM	BLOOM SYNDROME; BLM [AR]
BLOC1S3	HERMANSKY-PUDLAK SYNDROME 8; HPS8 [AR]
BLVRA	HYPERBILIVERDINEMIA; HBLVD [AD]
BMP1	OSTEOGENESIS IMPERFECTA. TYPE XIII; OI13 [AR]
BMP15	OVARIAN DYSGENESIS 2; ODG2 [X-linked]
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]
BMP4	OROFACIAL CLEFT 11; OFC11 [Familial predispositio] | MICROPHTHALMIA. SYNDROMIC 6; MCOPS6 [AD]
BMPER	DIAPHANOSPONDYLODYSOSTOSIS [AR]
BMPR1A	JUVENILE POLYPOSIS SYNDROME; JPS [AD]
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]
BNC2	LOWER URINARY TRACT OBSTRUCTION. CONGENITAL; LUTO [AD]
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2 WITH HYPERGLYCINEMIA; MMDS2 [AR]
BPGM	ERYTHROCYTOSIS. FAMILIAL. 8; ECYT8 [AR]
BPTF	NEURODEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND DISTAL LIMB ANOMALIES; NEDDFL [AD]

BRAF	NOONAN SYNDROME 7; NS7 [AD] | CARDIOFACIOCUTANEOUS SYNDROME 1; CFC1 [AD] | LUNG CANCER [Somatic mutation] | LEOPARD SYNDROME 3; LPRD3 [AD]
BRAT1	RIGIDITY AND MULTIFOCAL SEIZURE SYNDROME. LETHAL NEONATAL; RMFSL [AR] | NEURODEVELOPMENTAL DISORDER WITH CEREBELLAR ATROPHY AND WITH OR WITHOUT SEIZURES; NEDCAS [AR]
BRCA1	FANCONI ANEMIA. COMPLEMENTATION GROUP S; FANCS [AR] | BREAST CANCER [AD] | BREAST-OVARIAN CANCER. FAMILIAL. SUSCEPTIBILITY TO. 1; BROVCA1 [AD]
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]
BRF1	CEREBELLOFACIODENTAL SYNDROME; CFDS [AR]
BRIP1	BREAST CANCER [AD]
BRPF1	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND PTOSIS; IDDDFP [AD]
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]
BUB1	COLORECTAL CANCER; CRC [Somatic mutation]
BUB1B	PREMATURE CHROMATID SEPARATION TRAIT; PCS [AD] | COLORECTAL CANCER; CRC [Somatic mutation] | MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 1; MVA1 [AR]
BVES	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 25; LGMDR25 [AR]
C11orf80	Hydatidiform mole recurrent 4 618432 (3)
C12orf4	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 66; MRT66 [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]
C1GALT1C1	Tn polyagglutination syndrome somatic 300622 (3)
C1QA	C1q DEFICIENCY; C1QD [AR]
C1QB	C1q DEFICIENCY; C1QD [AR]
C1QBP	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 33; COXPD33 [AR]
C1QC	C1q DEFICIENCY; C1QD [AR]
C1QTNF5	LATE-ONSET RETINAL DEGENERATION; LORD [AD]
C1R	EHLERS-DANLOS SYNDROME. PERIODONTAL TYPE. 1; EDSPD1 [AD]
C1S	EHLERS-DANLOS SYNDROME. PERIODONTAL TYPE. 2; EDSPD2 [AD]
C2	C2 deficiency 217000 (3) | {Macular degeneration age-related 14 reduced risk of} 615489 (3)
C2CD3	OROFACIODIGITAL SYNDROME XIV; OFD14 [AR]
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)
C5	C5 deficiency 609536 (3) | [Eculizumab poor response to] 615749 (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]
C9	C9 deficiency 613825 (3) | {Macular degeneration age-related 15 susceptibility to} 615591 (3)
C9orf72	FRONTOTEMPORAL DEMENTIA AND/OR AMYOTROPHIC LATERAL SCLEROSIS 1; FTDALS1 [AD]
CA12	HYPERCHLORHIDROSIS. ISOLATED; HYCHL [AR]
CA2	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 3; OPTB3 [AR]
CA4	RETINITIS PIGMENTOSA 17; RP17 [AD]
CA5A	CARBONIC ANHYDRASE VA DEFICIENCY. HYPERAMMONEMIA DUE TO; CA5AD [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]
CACNA1A	MIGRAINE. FAMILIAL HEMIPLEGIC. 1; FHM1 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 42; EIEE42 [AD] | SPINOCEREBELLAR ATAXIA 6; SCA6 [AD] | EPISODIC ATAXIA. TYPE 2; EA2 [AD]
CACNA1B	DYSTONIA 23; DYT23 [AD] | NEURODEVELOPMENTAL DISORDER WITH SEIZURES AND NONEPILEPTIC HYPERKINETIC MOVEMENTS; NEDNEH [AR]
CACNA1C	TIMOTHY SYNDROME; TS [AD]
CACNA1D	SINOATRIAL NODE DYSFUNCTION AND DEAFNESS; SANDD [AR] | PRIMARY ALDOSTERONISM. SEIZURES. AND NEUROLOGIC ABNORMALITIES; PASNA [AD]
CACNA1E	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 69; EIEE69 [AD]
CACNA1F	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 2A; CSNB2A [X-linked form] | ALAND ISLAND EYE DISEASE; AIED [X-linked] | CONE-ROD DYSTROPHY. X-LINKED. 3; CORDX3 [XLR]
CACNA1G	SPINOCEREBELLAR ATAXIA 42; SCA42 [AD] | SPINOCEREBELLAR ATAXIA 42. EARLY-ONSET. SEVERE. WITH NEURODEVELOPMENTAL DEFICITS; SCA42ND [AD]
CACNA1H	HYPERALDOSTERONISM. FAMILIAL. TYPE IV; HALD4 [AD]
CACNA1S	THYROTOXIC PERIODIC PARALYSIS. SUSCEPTIBILITY TO. 1; TTPP1 [AD] | HYPOKALEMIC PERIODIC PARALYSIS. TYPE 1; HOKPP1 [AD] | MALIGNANT HYPERTHERMIA. SUSCEPTIBILITY TO. 5; MHS5 [AD]
CACNA2D2	CEREBELLAR ATROPHY WITH SEIZURES AND VARIABLE DEVELOPMENTAL DELAY; CASVDD [AR]
CACNA2D4	RETINAL CONE DYSTROPHY 4; RCD4 [AR]
CACNB2	Brugada syndrome 4 611876 (3)
CACNB4	EPISODIC ATAXIA. TYPE 5; EA5 [AD] | EPILEPSY. IDIOPATHIC GENERALIZED. SUSCEPTIBILITY TO. 9; EIG9 [AD]
CAD	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 50; EIEE50 [AR]
CALM1	VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 4; CPVT4 [AD] | LONG QT SYNDROME 14; LQT14 [AD]
CALM2	LONG QT SYNDROME 15; LQT15 [AD]
CALR	THROMBOCYTHEMIA 1; THCYT1 [AD] | MYELOFIBROSIS [Somatic mutation]
CAMK2A	MENTAL RETARDATION. AUTOSOMAL DOMINANT 53; MRD53 [AD] | MENTAL RETARDATION. AUTOSOMAL RECESSIVE 63; MRT63 [AR]
CAMK2B	MENTAL RETARDATION. AUTOSOMAL DOMINANT 54; MRD54 [AD]
CAMK2G	INTELLECTUAL DEVELOPMENTAL DISORDER 59; MRD59 [AD]
CAMTA1	CEREBELLAR ATAXIA. NONPROGRESSIVE. WITH MENTAL RETARDATION; CANPMR [AD]
CANT1	EPIPHYSEAL DYSPLASIA. MULTIPLE. 7; EDM7 [AR] | DESBUQUOIS DYSPLASIA 1; DBQD1 [AR]
CAPN1	SPASTIC PARAPLEGIA 76. AUTOSOMAL RECESSIVE; SPG76 [AR]
CAPN3	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 1; LGMDR1 [AR] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL DOMINANT 4; LGMDD4 [AD]
CAPN5	VITREORETINOPATHY. NEOVASCULAR INFLAMMATORY; VRNI [AD]
CARD11	B-CELL EXPANSION WITH NFKB AND T-CELL ANERGY; BENTA [AD] | IMMUNODEFICIENCY 11B WITH ATOPIC DERMATITIS; IMD11B [AD] | IMMUNODEFICIENCY 11; IMD11A [AR]
CARD14	PITYRIASIS RUBRA PILARIS; PRP [AD] | PSORIASIS 2; PSORS2 [AD]
CARD9	CANDIDIASIS. FAMILIAL. 2; CANDF2 [AR]
CARMIL2	IMMUNODEFICIENCY 58; IMD58 [AR]
CARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 27; COXPD27 [AR]

CARTPT	OBESITY [AD]
CASK	MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA; MICPCH [XLD]
CASP10	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME. TYPE IIA; ALPS2A [AD]
CASP14	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 12; ARCI12 [AR]
CASP8	CASPASE 8 DEFICIENCY [AR] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | LUNG CANCER [Somatic mutation] | BREAST CANCER [AD]
CASQ1	MYOPATHY. VACUOLAR. WITH CASQ1 AGGREGATES; VMCQA [AD]
CASQ2	VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 2; CPVT2 [AR]
CASR	HYPOCALCIURIC HYPERCALCEMIA. FAMILIAL. TYPE I; HHC1 [AD] | HYPOCALCEMIA. AUTOSOMAL DOMINANT 1; HYPOC1 [AD] | HYPERPARATHYROIDISM. NEONATAL SEVERE; NSHPT [AR]
CAST	PEELING SKIN WITH LEUKONYCHIA. ACRAL PUNCTATE KERATOSES. CHEILITIS. AND KNUCKLE PADS; PLACK [AR]
CAT	Acatalasemia 614097 (3)
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]
CAVIN1	LIPODYSTROPHY. CONGENITAL GENERALIZED. TYPE 4; CGL4 [AR]
CBFB	LEUKEMIA. ACUTE MYELOID; AML [AD]
CBL	JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML [AD] | NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MYELOMONOCYTIC LEUKEMIA; NSLL [AD]
CBLIF	INTRINSIC FACTOR DEFICIENCY; IFD [AR]
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]
CCDC103	CILIARY DYSKINESIA. PRIMARY. 17; CILD17 [AR]
CCDC114	CILIARY DYSKINESIA. PRIMARY. 20; CILD20 [AR]
CCDC115	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIo; CDG2O [AR]
CCDC151	CILIARY DYSKINESIA. PRIMARY. 30; CILD30 [AR]
CCDC174	HYPOTONIA. INFANTILE. WITH PSYCHOMOTOR RETARDATION; IHPMR [AR]
CCDC22	RITSCHER-SCHINZEL SYNDROME 2; RTSC2 [XLR]
CCDC39	Ciliary dyskinesia primary 14 613807 (3)
CCDC40	Ciliary dyskinesia primary 15 613808 (3)
CCDC47	TRICHOHEPATONEURODEVELOPMENTAL SYNDROME; THNS [AR]
CCDC65	CILIARY DYSKINESIA. PRIMARY. 27; CILD27 [AR]
CCDC8	THREE M SYNDROME 3; 3M3 [AR]
CCDC88C	SPINOCEREBELLAR ATAXIA 40; SCA40 [AD] | HYDROCEPHALUS. CONGENITAL. 1; HYC1 [AR]
CCM2	CEREBRAL CAVERNOUS MALFORMATIONS 2; CCM2 [AD]
CCN6	ARTHROPATHY. PROGRESSIVE PSEUDORHEUMATOID. OF CHILDHOOD; PPAC [AR]
CCND2	MEGALENCEPHALY-POLYMICROGYRIA-POLYDACTYLY-HYDROCEPHALUS SYNDROME 3; MPPH3 [AD]
CCNO	CILIARY DYSKINESIA. PRIMARY. 29; CILD29 [AR]
CCNQ	TOE SYNDACTYLY. TELECANTHUS. AND ANOGENITAL AND RENAL MALFORMATIONS; STAR [XLD]
CCT5	NEUROPATHY. HEREDITARY SENSORY. WITH SPASTIC PARAPLEGIA. AUTOSOMAL RECESSIVE [AR]
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness 609057 (3) | [Blood group Raph] 179620 (3)
CD19	IMMUNODEFICIENCY. COMMON VARIABLE. 3; CVID3 [AR]

CD207	[?Birbeck granule deficiency] 613393 (3)
CD27	LYMPHOPROLIFERATIVE SYNDROME 2; LPFS2 [AR]
CD2AP	Glomerulosclerosis focal segmental 3 607832 (3)
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]
CD4	OKT4 epitope deficiency 613949 (3)
CD40	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 3; HIGM3 [AR]
CD40LG	IMMUNODEFICIENCY WITH HYPER-IgM. TYPE 1; HIGM1 [XLR]
CD55	COMPLEMENT HYPERACTIVATION. ANGIOPATHIC THROMBOSIS. AND PROTEIN-LOSING ENTEROPATHY; CHAPLE [AR] | BLOOD GROUP. CROMER SYSTEM; CROM [AR]
CD59	HEMOLYTIC ANEMIA. CD59-MEDIATED. WITH OR WITHOUT IMMUNE-MEDIATED POLYNEUROPATHY; HACD59 [AR]
CD70	LYMPHOPROLIFERATIVE SYNDROME 3; LPFS3 [AR]
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]
CDC14A	DEAFNESS. AUTOSOMAL RECESSIVE 32. WITH OR WITHOUT IMMOTILE SPERM; DFNB32 [AR]
CDC42	TAKENOUCHI-KOSAKI SYNDROME; TKS [AD]
CDC45	MEIER-GORLIN SYNDROME 7; MGORS7 [AR]
CDC73	HYPERPARATHYROIDISM 2 WITH JAW TUMORS; HRPT2 [AD] | HYPERPARATHYROIDISM 1; HRPT1 [AD]
CDCA7	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 3; ICF3 [AR]
CDH1	OVARIAN CANCER [Somatic mutation] | GASTRIC CANCER. HEREDITARY DIFFUSE; HDGC [AD] | PROSTATE CANCER [Somatic mutation] | BLEPHAROCHEILODONTIC SYNDROME 1; BCDS1 [AD] | BREAST CANCER [AD] | ENDOMETRIAL CANCER [Somatic mutation]
CDH11	ELSAHY-WATERS SYNDROME; ESWS [AR]
CDH15	Mental retardation autosomal dominant 3 612580 (3)
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]
CDK10	AL KAISSI SYNDROME; ALKAS [AR]
CDK13	CONGENITAL HEART DEFECTS. DYSMORPHIC FACIAL FEATURES. AND INTELLECTUAL DEVELOPMENTAL DISORDER; CHDFIDD [AD]
CDK5RAP2	MICROCEPHALY 3. PRIMARY. AUTOSOMAL RECESSIVE; MCPH3 [AR]
CDKL5	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 2; EIEE2 [XLD]
CDKN1B	MULTIPLE ENDOCRINE NEOPLASIA. TYPE IV; MEN4 [AD]
CDKN1C	INTRAUTERINE GROWTH RETARDATION. METAPHYSEAL DYSPLASIA. ADRENAL HYPOPLASIA CONGENITA. AND GENITAL ANOMALIES; IMAGE [AD] | BECKWITH-WIEDEMANN SYNDROME; BWS [AD]
CDKN2A	MELANOMA-PANCREATIC CANCER SYNDROME [AD] | MELANOMA. CUTANEOUS MALIGNANT. SUSCEPTIBILITY TO. 2; CMM2 [AD] | MELANOMA-ASTROCYTOMA SYNDROME [AD]
CDON	HOLOPROSENCEPHALY 11; HPE11 [AD]
CDSN	HYPOTRICHOSIS 2; HYPT2 [AD] | PEELING SKIN SYNDROME 1; PSS1 [AR]

CDT1	MEIER-GORLIN SYNDROME 4; MGORS4 [AR]
CEACAM16	DEAFNESS. AUTOSOMAL RECESSIVE 113; DFNB113 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 4B; DFNA4B [AD]
CEBPA	LEUKEMIA. ACUTE MYELOID; AML [AD]
CEBPE	SPECIFIC GRANULE DEFICIENCY 1; SGD1 [AR]
CEL	MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 8. WITH EXOCRINE DYSFUNCTION; MODY8 [AD]
CELA2A	ABDOMINAL OBESITY-METABOLIC SYNDROME 4; AOMS4 [AD]
CENPF	STROMME SYNDROME; STROMS [AR]
CENPJ	SECKEL SYNDROME 4; SCKL4 [AR] | MICROCEPHALY 6. PRIMARY. AUTOSOMAL RECESSIVE; MCPH6 [AR]
CEP104	JOUBERT SYNDROME 25; JBTS25 [AR]
CEP120	JOUBERT SYNDROME 31; JBTS31 [AR] | SHORT-RIB THORACIC DYSPLASIA 13 WITH OR WITHOUT POLYDACTYLY; SRTD13 [AR]
CEP135	MICROCEPHALY 8. PRIMARY. AUTOSOMAL RECESSIVE; MCPH8 [AR]
CEP152	SECKEL SYNDROME 5; SCKL5 [AR] | MICROCEPHALY 9. PRIMARY. AUTOSOMAL RECESSIVE; MCPH9 [AR]
CEP164	NEPHRONOPHTHISIS 15; NPHP15 [AR]
CEP19	MORBID OBESITY AND SPERMATOGENIC FAILURE; MOSPGF [AR]
CEP250	CONE-ROD DYSTROPHY AND HEARING LOSS 2; CRDHL2 [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]
CEP55	MULTINUCLEATED NEURONS. ANHYDRAMNIOS. RENAL DYSPLASIA. CEREBELLAR HYPOPLASIA. AND HYDRANENCEPHALY; MARCH [AR]
CEP57	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 2; MVA2 [AR]
CEP78	CONE-ROD DYSTROPHY AND HEARING LOSS 1; CRDHL1 [AR]
CEP83	NEPHRONOPHTHISIS 18; NPHP18 [AR]
CERKL	Retinitis pigmentosa 26 608380 (3)
CERS3	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 9; ARCI9 [AR]
CERT1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 34; MRD34 [AD]
CES1	Drug metabolism altered CES1-related 618057 (3)
CETP	HYPERALPHALIPOPROTEINEMIA 1; HALP1 [AD]
CFAP298	CILIARY DYSKINESIA. PRIMARY. 26; CILD26 [AR]
CFAP300	CILIARY DYSKINESIA. PRIMARY. 38; CILD38 [AR]
CFAP410	SPONDYLOMETAPHYSEAL DYSPLASIA. AXIAL; SMDAX [AR] | RETINAL DYSTROPHY WITH OR WITHOUT MACULAR STAPHYLOMA; RDMS [AR]
CFAP43	HYDROCEPHALUS. NORMAL-PRESSURE. 1; HYDNP1 [AD] | SPERMATOGENIC FAILURE 19; SPGF19 [AR]
CFAP53	HETEROTAXY. VISCERAL. 6. AUTOSOMAL; HTX6 [AR]
CFAP69	SPERMATOGENIC FAILURE 24; SPGF24 [AR]
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]
CFHR5	CFHR5 DEFICIENCY [AD]
CFI	HEMOLYTIC UREMIC SYNDROME. ATYPICAL. SUSCEPTIBILITY TO. 3; AHUS3 [AD] | COMPLEMENT FACTOR I DEFICIENCY; CFID [AR] | MACULAR DEGENERATION. AGE-RELATED. 13; ARMD13 [AD]
CFL2	NEMALINE MYOPATHY 7; NEM7 [AR]
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]
CHAMP1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 40; MRD40 [AD]

CHAT	MYASTHENIC SYNDROME. CONGENITAL. 6. PRESYNAPTIC; CMS6 [AR]
CHCHD10	FRONTOTEMPORAL DEMENTIA AND/OR AMYOTROPHIC LATERAL SCLEROSIS 2; FTDALS2 [AD] | SPINAL MUSCULAR ATROPHY. JOKELA TYPE; SMAJ [AD] | MYOPATHY. ISOLATED MITOCHONDRIAL. AUTOSOMAL DOMINANT; IMMD [AD]
CHCHD2	PARKINSON DISEASE 22. AUTOSOMAL DOMINANT; PARK22 [AD]
CHD1	PILAROWSKI-BJORNSSON SYNDROME; PILBOS [AD]
CHD2	EPILEPTIC ENCEPHALOPATHY. CHILDHOOD-ONSET; EEOC [AD]
CHD3	SNIJDERS BLOK-CAMPEAU SYNDROME; SNIBCPS [AD]
CHD4	SIFRIM-HITZ-WEISS SYNDROME; SIHIWES [AD]
CHD7	HYPOGONADOTROPIC HYPOGONADISM 5 WITH OR WITHOUT ANOSMIA; HH5 [AD] | CHARGE SYNDROME [AD]
CHEK2	BREAST CANCER [AD] | OSTEOGENIC SARCOMA [Somatic mutation] | PROSTATE CANCER [Somatic mutation]
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]
CHMP2B	FRONTOTEMPORAL DEMENTIA. CHROMOSOME 3-LINKED; FTD3 [AD] | AMYOTROPHIC LATERAL SCLEROSIS 17; ALS17 [AD]
CHMP4B	CATARACT 31. MULTIPLE TYPES; CTRCT31 [AD]
CHN1	DUANE RETRACTION SYNDROME 2; DURS2 [AD]
CHRDL1	MEGALOCORNEA; MGC1 [XLR]
CHRNA1	MULTIPLE PTERYGIUM SYNDROME. LETHAL TYPE; LMPS [AR] | MYASTHENIC SYNDROME. CONGENITAL. 1B. FAST-CHANNEL; CMS1B [AD] | MYASTHENIC SYNDROME. CONGENITAL. 1A. SLOW-CHANNEL; CMS1A [AD]
CHRNA2	EPILEPSY. NOCTURNAL FRONTAL LOBE. 4; ENFL4 [AD]
CHRNA4	EPILEPSY. NOCTURNAL FRONTAL LOBE. 1; ENFL1 [AD] | TOBACCO ADDICTION. SUSCEPTIBILITY TO [Genetic factors seem to contribute only in light smokers and heavy smoker]
CHRNB1	MYASTHENIC SYNDROME. CONGENITAL. 2C. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS2C [AR] | MYASTHENIC SYNDROME. CONGENITAL. 2A. SLOW-CHANNEL; CMS2A [AD]
CHRNB2	Epilepsy nocturnal frontal lobe 3 605375 (3)
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)
CIB1	EPIDERMODYSPLASIA VERRUCIFORMIS. SUSCEPTIBILITY TO. 3; EV3 [AR]
CIB2	DEAFNESS. AUTOSOMAL RECESSIVE 48; DFNB48 [AR] | USHER SYNDROME. TYPE IJ; USH1J [AR]
CIC	MENTAL RETARDATION. AUTOSOMAL DOMINANT 45; MRD45 [AD]
CIITA	RHEUMATOID ARTHRITIS; RA [Unproved inheritanc] | BARE LYMPHOCYTE SYNDROME. TYPE II [AR]
CILK1	EPILEPSY. JUVENILE MYOCLONIC. SUSCEPTIBILITY TO. 10; EJM10 [AD] | ENDOCRINE-CEREBROOSTEODYSPLASIA; ECO [AR]
CISD2	WOLFRAM SYNDROME 2; WFS2 [AR]
CIT	MICROCEPHALY 17. PRIMARY. AUTOSOMAL RECESSIVE; MCPH17 [AR]
CITED2	VENTRICULAR SEPTAL DEFECT 2; VSD2 [AD] | ATRIAL SEPTAL DEFECT 8; ASD8 [AD]
CKAP2L	FILIPPI SYNDROME; FLPIS [AR]
CLCF1	CRISPONI/COLD-INDUCED SWEATING SYNDROME 2; CISS2 [AR]

CLCN1	MYOTONIA CONGENITA. AUTOSOMAL RECESSIVE [AR] | MYOTONIA CONGENITA. AUTOSOMAL DOMINANT [AD]
CLCN2	HYPERALDOSTERONISM. FAMILIAL. TYPE II; HALD2 [AD] | LEUKOENCEPHALOPATHY WITH ATAXIA; LKPAT [AR] | EPILEPSY. IDIOPATHIC GENERALIZED. SUSCEPTIBILITY TO. 11; EIG11 [AD] | EPILEPSY. JUVENILE ABSENCE. SUSCEPTIBILITY TO. 1; EJA1 [AD]
CLCN4	RAYNAUD-CLAES SYNDROME; MRXSRC [XLD]
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]
CLCNKA	BARTTER SYNDROME. TYPE 4B. NEONATAL. WITH SENSORINEURAL DEAFNESS; BARTS4B [Digenic recessive]
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]
CLDN10	HELIX SYNDROME; HELIX [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]
CLEC4M	SARS infection protection against (2)
CLEC7A	CANDIDIASIS. FAMILIAL. 4; CANDF4 [AR]
CLMP	CONGENITAL SHORT BOWEL SYNDROME; CSBS [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]
CLP1	PONTOCEREBELLAR HYPOPLASIA. TYPE 10; PCH10 [AR]
CLPB	3-METHYLGLUTACONIC ACIDURIA. TYPE VII; MGCA7 [AR]
CLPP	PERRAULT SYNDROME 3; PRLTS3 [AR]
CLRN1	USHER SYNDROME. TYPE IIIA; USH3A [AR]
CLTC	MENTAL RETARDATION. AUTOSOMAL DOMINANT 56; MRD56 [AD]
CNBP	MYOTONIC DYSTROPHY 2; DM2 [AD]
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]
CNKSR2	MENTAL RETARDATION. X-LINKED. SYNDROMIC. HOUGE TYPE; MRXSHG [X-linked]
CNNM2	HYPOMAGNESEMIA. SEIZURES. AND MENTAL RETARDATION 1; HOMGSMR1 [AD] | HYPOMAGNESEMIA 6. RENAL; HOMG6 [AD]
CNNM4	JALILI SYNDROME [AR]
CNOT1	HOLOPROSENCEPHALY 12 WITH OR WITHOUT PANCREATIC AGENESIS; HPE12 [AD]
CNOT2	INTELLECTUAL DEVELOPMENTAL DISORDER WITH NASAL SPEECH. DYSMORPHIC FACIES. AND VARIABLE SKELETAL ANOMALIES; IDNADFS [AD]
CNPY3	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 60; EIEE60 [AR]
CNTNAP1	LETHAL CONGENITAL CONTRACTURE SYNDROME 7; LCCS7 [AR] | NEUROPATHY. CONGENITAL HYPOMYELINATING. 3; CHN3 [AR]
CNTNAP2	PITT-HOPKINS-LIKE SYNDROME 1; PTHSL1 [AR]
COA6	CARDIOENCEPHALOMYOPATHY. FATAL INFANTILE. DUE TO CYTOCHROME c OXIDASE DEFICIENCY 4; CEMCOX4 [AR]
COA7	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE. WITH AXONAL NEUROPATHY 3; SCAN3 [AR]
COA8	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
COASY	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 6; NBIA6 [AR] | PONTOCEREBELLAR HYPOPLASIA. TYPE 12; PCH12 [AR]

COCH	DEAFNESS. AUTOSOMAL RECESSIVE 110; DFNB110 [AR] | DEAFNESS. AUTOSOMAL DOMINANT 9; DFNA9 [AD]
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)
COG6	SHAHEEN SYNDROME; SHNS [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE III; CDG2L [AR]
COG7	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIe; CDG2E [AR]
COG8	Congenital disorder of glycosylation type IIh 611182 (3)
COL10A1	METAPHYSEAL CHONDRODYSPLASIA. SCHMID TYPE; MCDS [AD]
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]
COL12A1	BETHLEM MYOPATHY 2; BTHLM2 [AD]
COL13A1	MYASTHENIC SYNDROME. CONGENITAL. 19; CMS19 [AR]
COL17A1	EPITHELIAL RECURRENT EROSION DYSTROPHY; ERED [AD] | EPIDERMOLYSIS BULLOSA. JUNCTIONAL. NON-HERLITZ TYPE [AR]
COL18A1	KNOBLOCH SYNDROME 1; KNO1 [AR]
COL1A1	OSTEOGENESIS IMPERFECTA. TYPE II; OI2 [AD] | OSTEOGENESIS IMPERFECTA. TYPE I; OI1 [AD] | OSTEOPOROSIS [AD] | CAFFEY DISEASE; CAFYD [AD] | OSTEOGENESIS IMPERFECTA. TYPE IV; OI4 [AD] | OSTEOGENESIS IMPERFECTA. TYPE III; OI3 [AD] | EHLERS-DANLOS SYNDROME. ARTHROCHALASIA TYPE. 1; EDSARTH1 [AD]
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]
COL25A1	FIBROSIS OF EXTRAOCULAR MUSCLES. CONGENITAL. 5; CFEOM5 [AR]
COL27A1	STEEL SYNDROME; STLS [AR]
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]
COL4A1	MICROANGIOPATHY AND LEUKOENCEPHALOPATHY. PONTINE. AUTOSOMAL DOMINANT; PADMAL [AD] | BRAIN SMALL VESSEL DISEASE 1 WITH OR WITHOUT OCULAR ANOMALIES; BSVD1 [AD] | RETINAL ARTERIES. TORTUOSITY OF; RATOR [AD] | ANGIOPATHY. HEREDITARY. WITH NEPHROPATHY. ANEURYSMS. AND MUSCLE CRAMPS; HANAC [AD]
COL4A2	BRAIN SMALL VESSEL DISEASE 2; BSVD2 [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]
COL5A1	EHLERS-DANLOS SYNDROME. CLASSIC TYPE. 1; EDSCL1 [AD]
COL5A2	EHLERS-DANLOS SYNDROME. CLASSIC TYPE. 1; EDSCL1 [AD] | EHLERS-DANLOS SYNDROME. CLASSIC TYPE. 2; EDSCL2 [AD]
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]
COL8A2	CORNEAL DYSTROPHY. FUCHS ENDOTHELIAL. 1; FECD1 [AD] | CORNEAL DYSTROPHY. POSTERIOR POLYMORPHOUS. 2; PPCD2 [AD]

COL9A1	EPIPHYSEAL DYSPLASIA. MULTIPLE. 6; EDM6 [AD]
COL9A2	STICKLER SYNDROME. TYPE V; STL5 [AR] | EPIPHYSEAL DYSPLASIA. MULTIPLE. 2; EDM2 [AD]
COL9A3	EPIPHYSEAL DYSPLASIA. MULTIPLE. 3; EDM3 [AD]
COLEC10	3MC SYNDROME 3; 3MC3 [AR]
COLEC11	3MC SYNDROME 2; 3MC2 [AR]
COLGALT1	BRAIN SMALL VESSEL DISEASE 3; BSVD3 [AR]
COLQ	MYASTHENIC SYNDROME. CONGENITAL. 5; CMS5 [AR]
COMP	PSEUDOACHONDROPLASIA; PSACH [AD] | EPIPHYSEAL DYSPLASIA. MULTIPLE. 1; EDM1 [AD]
COMT	PANIC DISORDER 1; PAND1 [AD] | SCHIZOPHRENIA; SCZD [AD]
COQ2	COENZYME Q10 DEFICIENCY. PRIMARY. 1; COQ10D1 [AR] | MULTIPLE SYSTEM ATROPHY 1. SUSCEPTIBILITY TO; MSA1 [AD]
COQ4	COENZYME Q10 DEFICIENCY. PRIMARY. 7; COQ10D7 [AR]
COQ6	COENZYME Q10 DEFICIENCY. PRIMARY. 6; COQ10D6 [AR]
COQ8A	COENZYME Q10 DEFICIENCY. PRIMARY. 4; COQ10D4 [AR]
COQ8B	NEPHROTIC SYNDROME. TYPE 9; NPHS9 [AR]
COQ9	COENZYME Q10 DEFICIENCY. PRIMARY. 5; COQ10D5 [AR]
CORIN	Preeclampsia/eclampsia 5 614595 (3)
CORO1A	IMMUNODEFICIENCY 8; IMD8 [AR]
COX10	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
COX15	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR] | CARDIOENCEPHALOMYOPATHY. FATAL INFANTILE. DUE TO CYTOCHROME c OXIDASE DEFICIENCY 2; CEMCOX2 [AR]
COX20	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
COX4I2	EXOCRINE PANCREATIC INSUFFICIENCY. DYSERYTHROPOIETIC ANEMIA. AND CALVARIAL HYPEROSTOSIS [AR]
COX6A1	CHARCOT-MARIE-TOOTH DISEASE. RECESSIVE INTERMEDIATE D; CMTRID [AR]
COX6A2	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
COX6B1	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
COX7B	LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 2; LSDMCA2 [XLD]
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]
CPAMD8	ANTERIOR SEGMENT DYSGENESIS 8; ASGD8 [AR]
CPLANE1	JOUBERT SYNDROME 17; JBTS17 [AR] | OROFACIODIGITAL SYNDROME VI; OFD6 [AR]
CPLX1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 63; EIEE63 [AR]
CPN1	CARBOXYPEPTIDASE N DEFICIENCY [AR]
CPOX	COPROPORPHYRIA. HEREDITARY; HCP [AD]
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]
CR2	IMMUNODEFICIENCY. COMMON VARIABLE. 7; CVID7 [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]
CRB2	FOCAL SEGMENTAL GLOMERULOSCLEROSIS 9; FSGS9 [AR] | VENTRICULOMEGALY WITH CYSTIC KIDNEY DISEASE; VMCKD [AR]
CRBN	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 2; MRT2 [AR]
CREB1	Histiocytoma angiomatoid fibrous somatic 612160 (3)

CREB3L1	OSTEOGENESIS IMPERFECTA. TYPE XVI; OI16 [AR]
CREBBP	RUBINSTEIN-TAYBI SYNDROME 1; RSTS1 [AD]
CRELD1	ATRIOVENTRICULAR SEPTAL DEFECT. SUSCEPTIBILITY TO. 2; AVSD2 [AD]
CRIPT	SHORT STATURE WITH MICROCEPHALY AND DISTINCTIVE FACIES; SSMCF [AR]
CRLF1	CRISPONI/COLD-INDUCED SWEATING SYNDROME 1; CISS1 [AR]
CRPPA	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 7; MDDGA7 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 7; MDDGC7 [AR]
CRTAP	OSTEOGENESIS IMPERFECTA. TYPE VII; OI7 [AR]
CRTC1	Mucoepidermoid salivary gland carcinoma (3)
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]
CRYBA1	CATARACT 10. MULTIPLE TYPES; CTRCT10 [AD]
CRYBA4	Cataract 23 610425 (3)
CRYBB1	CATARACT 17. MULTIPLE TYPES; CTRCT17 [AD]
CRYBB2	CATARACT 3. MULTIPLE TYPES; CTRCT3 [AD]
CRYBB3	CATARACT 22. MULTIPLE TYPES; CTRCT22 [AD]
CRYGB	CATARACT 39. MULTIPLE TYPES; CTRCT39 [AD]
CRYGC	CATARACT 2. MULTIPLE TYPES; CTRCT2 [AD]
CRYGD	CATARACT 4. MULTIPLE TYPES; CTRCT4 [AD]
CRYGS	CATARACT 20. MULTIPLE TYPES; CTRCT20 [AD]
CRYM	DEAFNESS. AUTOSOMAL DOMINANT 40; DFNA40 [AD]
CSF1R	LEUKOENCEPHALOPATHY. HEREDITARY DIFFUSE. WITH SPHEROIDS; HDLS [AD] | BRAIN ABNORMALITIES. NEURODEGENERATION. AND DYSOSTEOSCLEROSIS; BANDDOS [AR]
CSF2RA	Surfactant metabolism dysfunction pulmonary 4 300770 (3)
CSF2RB	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 5; SMDP5 [AR]
CSF3R	NEUTROPHILIA. HEREDITARY [AD] | NEUTROPENIA. SEVERE CONGENITAL. 7. AUTOSOMAL RECESSIVE; SCN7 [AR]
CSNK1D	ADVANCED SLEEP PHASE SYNDROME. FAMILIAL. 2; FASPS2 [AD]
CSNK2A1	OKUR-CHUNG NEURODEVELOPMENTAL SYNDROME; OCNDS [AD]
CSPP1	JOUBERT SYNDROME 21; JBTS21 [AR]
CSRP3	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 12; CMH12 [AD]
CST3	CEREBRAL AMYLOID ANGIOPATHY. CST3-RELATED [AD]
CSTA	PEELING SKIN SYNDROME 4; PSS4 [AR]
CSTB	MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG [AR]
CTBP1	HYPOTONIA. ATAXIA. DEVELOPMENTAL DELAY. AND TOOTH ENAMEL DEFECT SYNDROME; HADDTS [AD]
CTC1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS 1; CRMCC1 [AR]
CTCF	MENTAL RETARDATION. AUTOSOMAL DOMINANT 21; MRD21 [AD]
CTDP1	CONGENITAL CATARACTS. FACIAL DYSMORPHISM. AND NEUROPATHY; CCFDN [AR]
CTH	CYSTATHIONINURIA [AR]
CTHRC1	Barrett esophagus/esophageal adenocarcinoma 614266 (3)
CTLA4	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME. TYPE V; ALPS5 [AD] | HASHIMOTO THYROIDITIS [AD] | SYSTEMIC LUPUS ERYTHEMATOSUS; SLE [AD]
CTNNA1	MACULAR DYSTROPHY. PATTERNED. 2; MDPT2 [AD]
CTNNA2	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 9; CDCBM9 [AR]

CTNNA3	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 13; ARVD13 [AD]
CTNNB1	OVARIAN CANCER [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation] | NEURODEVELOPMENTAL DISORDER WITH SPASTIC DIPLEGIA AND VISUAL DEFECTS; NEDSDV [AD] | MEDULLOBLASTOMA; MDB [Somatic mutation] | PILOMATRIXOMA [Somatic mutation] | EXUDATIVE VITREORETINOPATHY 7; EVR7 [AD] | HEPATOCELLULAR CARCINOMA [Somatic mutation]
CTNND1	BLEPHAROCHEILODONTIC SYNDROME 2; BCDS2 [AD]
CTNS	CYSTINOSIS. NEPHROPATHIC; CTNS [AR] | CYSTINOSIS. ADULT NONNEPHROPATHIC [AR] | CYSTINOSIS. LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE [AR]
CTPS1	IMMUNODEFICIENCY 24; IMD24 [AR]
CTSA	GALACTOSIALIDOSIS; GSL [AR]
CTSB	KERATOLYTIC WINTER ERYTHEMA; KWE [AD]
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]
CTU2	MICROCEPHALY. FACIAL DYSMORPHISM. RENAL AGENESIS. AND AMBIGUOUS GENITALIA SYNDROME; MFRG [AR]
CUBN	MEGALOBLASTIC ANEMIA 1 [AR]
CUL3	PSEUDOHYPOALDOSTERONISM. TYPE IIE; PHA2E [AD]
CUL4B	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CABEZAS TYPE; MRXSC [XLR]
CUL7	THREE M SYNDROME 1; 3M1 [AR]
CUX1	GLOBAL DEVELOPMENTAL DELAY WITH OR WITHOUT IMPAIRED INTELLECTUAL DEVELOPMENT; GDDI [AD]
CUX2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 67; EIEE67 [AD]
CWC27	RETINITIS PIGMENTOSA WITH OR WITHOUT SKELETAL ANOMALIES; RPSKA [AR]
CWF19L1	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 17; SCAR17 [AR]
CXCR4	WHIM SYNDROME; WHIMS [AD]
CYB561	ORTHOSTATIC HYPOTENSION 2; ORTHYP2 [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]
CYC1	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 6; MC3DN6 [AR]
CYCS	THROMBOCYTOPENIA 4; THC4 [AD]
CYFIP2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 65; EIEE65 [AD]
CYLD	BROOKE-SPIEGLER SYNDROME; BRSS [AD] | CYLINDROMATOSIS. FAMILIAL [AD] | TRICHOEPITHELIOMA. MULTIPLE FAMILIAL. 1 [AD]
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]
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies 614416 (3)
CYP26C1	FOCAL FACIAL DERMAL DYSPLASIA 4; FFDD4 [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]
CYP2B6	Efavirenz poor metabolism of 614546 (3) | {Efavirenz central nervous system toxicity susceptibility to} 614546 (3)
CYP2C19	DRUG METABOLISM. POOR. CYP2C19-RELATED [AR]
CYP2C9	COUMARIN RESISTANCE [AD]
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]
DAB1	SPINOCEREBELLAR ATAXIA 37; SCA37 [AD]
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]
DARS1	HYPOMYELINATION WITH BRAINSTEM AND SPINAL CORD INVOLVEMENT AND LEG SPASTICITY; HBSL [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]
DCC	GAZE PALSY. FAMILIAL HORIZONTAL. WITH PROGRESSIVE SCOLIOSIS 2. WITH IMPAIRED INTELLECTUAL DEVELOPMENT; HGPPS2 [AR] | ESOPHAGEAL CANCER [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation] | MIRROR MOVEMENTS 1; MRMV1 [AD]
DCDC2	DEAFNESS. AUTOSOMAL RECESSIVE 66; DFNB66 [AR] | NEPHRONOPHTHISIS 19; NPHP19 [AR] | SCLEROSING CHOLANGITIS. NEONATAL; NSC [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]
DCN	CORNEAL DYSTROPHY. CONGENITAL STROMAL; CSCD [AD]
DCPS	AL-RAQAD SYNDROME; ARS [AR]
DCTN1	NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VIIB; HMN7B [AD] | PERRY SYNDROME [AD] | AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1 [AD]
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]
DDRGK1	SPONDYLOEPIMETAPHYSEAL DYSPLASIA. SHOHAT TYPE; SEMDSH [AR]
DDX11	WARSAW BREAKAGE SYNDROME; WABS [AR]
DDX3X	MENTAL RETARDATION. X-LINKED 102; MRX102 [XLD]
DDX58	SINGLETON-MERTEN SYNDROME 2; SGMRT2 [AD]
DDX59	OROFACIODIGITAL SYNDROME V; OFD5 [AR]
DDX6	INTELLECTUAL DEVELOPMENTAL DISORDER WITH IMPAIRED LANGUAGE AND DYSMORPHIC FACIES; IDDILF [AD]
DEAF1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 24; MRD24 [AD] | DYSKINESIA. SEIZURES. AND INTELLECTUAL DEVELOPMENTAL DISORDER; DYSEIDD [AR]
DEGS1	LEUKODYSTROPHY. HYPOMYELINATING. 18; HLD18 [AR]
DEK	Leukemia acute nonlymphocytic (2)
DENND5A	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 49; EIEE49 [AR]

DEPDC5	EPILEPSY. FAMILIAL FOCAL. WITH VARIABLE FOCI 1; FFEVF1 [AD]
DES	CARDIOMYOPATHY. DILATED. 1I; CMD1I [AD] | SCAPULOPERONEAL SYNDROME. NEUROGENIC. KAESER TYPE; SCPNK [AD] | MYOPATHY. MYOFIBRILLAR. 1; MFM1 [AD]
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]
DHPS	NEURODEVELOPMENTAL DISORDER WITH SEIZURES AND SPEECH AND WALKING IMPAIRMENT; NEDSSWI [AR]
DHTKD1	2-AMINOADIPIC 2-OXOADIPIC ACIDURIA; AMOXAD [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2Q; CMT2Q [AD]
DHX30	NEURODEVELOPMENTAL DISORDER WITH SEVERE MOTOR IMPAIRMENT AND ABSENT LANGUAGE; NEDMIAL [AD]
DHX38	RETINITIS PIGMENTOSA 84; RP84 [AR]
DIABLO	DEAFNESS. AUTOSOMAL DOMINANT 64; DFNA64 [AD]
DIAPH1	SEIZURES. CORTICAL BLINDNESS. AND MICROCEPHALY SYNDROME; SCBMS [AR] | DEAFNESS. AUTOSOMAL DOMINANT 1; DFNA1 [AD]
DIAPH3	AUDITORY NEUROPATHY. AUTOSOMAL DOMINANT. 1; AUNA1 [AD]
DICER1	GLOBAL DEVELOPMENTAL DELAY. LUNG CYSTS. OVERGROWTH. AND WILMS TUMOR; GLOW [Somatic mosaicism] | GOITER. MULTINODULAR 1. WITH OR WITHOUT SERTOLI-LEYDIG CELL TUMORS; MNG1 [AD] | PLEUROPULMONARY BLASTOMA; PPB [AD]
DIP2B	MENTAL RETARDATION. FRA12A TYPE [AD]
DIS3L2	PERLMAN SYNDROME; PRLMNS [AR]
DISC2	SCHIZOPHRENIA; SCZD [AD]
DKC1	DYSKERATOSIS CONGENITA. X-LINKED; DKCX [XLR]
DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY; PDHDD [AR]
DLC1	COLORECTAL CANCER; CRC [Somatic mutation]
DLD	MAPLE SYRUP URINE DISEASE; MSUD [AR] | DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY; DLDD [AR]
DLG3	MENTAL RETARDATION. X-LINKED 90; MRX90 [XLR]
DLL3	SPONDYLOCOSTAL DYSOSTOSIS 1. AUTOSOMAL RECESSIVE; SCDO1 [AR]
DLL4	ADAMS-OLIVER SYNDROME 6; AOS6 [AD]
DLST	PARAGANGLIOMAS 7; PGL7 [AD]
DLX3	AMELOGENESIS IMPERFECTA. TYPE IV; AI4 [AD] | TRICHODENTOOSSEOUS SYNDROME; TDO [AD]
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]
DMPK	MYOTONIC DYSTROPHY 1; DM1 [AD]
DMXL2	POLYENDOCRINE-POLYNEUROPATHY SYNDROME; PEPNS [AR] | DEAFNESS. AUTOSOMAL DOMINANT 71; DFNA71 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 81; EIEE81 [AR]
DNA2	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 6; PEOA6 [AD] | SECKEL SYNDROME 8; SCKL8 [AR]
DNAAF1	CILIARY DYSKINESIA. PRIMARY. 13; CILD13 [AR]
DNAAF2	Ciliary dyskinesia primary 10 612518 (3)
DNAAF3	CILIARY DYSKINESIA. PRIMARY. 2; CILD2 [AR]
DNAAF4	CILIARY DYSKINESIA. PRIMARY. 25; CILD25 [AR] | DYSLEXIA. SUSCEPTIBILITY TO. 1; DYX1 [AD]

DNAAF5	CILIARY DYSKINESIA. PRIMARY. 18; CILD18 [AR]
DNAH1	SPERMATOGENIC FAILURE 18; SPGF18 [AR] | CILIARY DYSKINESIA. PRIMARY. 37; CILD37 [AR]
DNAH11	CILIARY DYSKINESIA. PRIMARY. 7; CILD7 [AR]
DNAH17	SPERMATOGENIC FAILURE 39; SPGF39 [AR]
DNAH5	Ciliary dyskinesia primary 3 with or without situs inversus 608644 (3)
DNAH9	CILIARY DYSKINESIA. PRIMARY. 40; CILD40 [AR]
DNAI1	CILIARY DYSKINESIA. PRIMARY. 1; CILD1 [AR]
DNAI2	Ciliary dyskinesia primary 9 with or without situs inversus 612444 (3)
DNAJB11	POLYCYSTIC KIDNEY DISEASE 6 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD6 [AD]
DNAJB13	CILIARY DYSKINESIA. PRIMARY. 34; CILD34 [AR]
DNAJB2	SPINAL MUSCULAR ATROPHY. DISTAL. AUTOSOMAL RECESSIVE. 5; DSMA5 [AR]
DNAJB6	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL DOMINANT 1; LGMDD1 [AD]
DNAJC12	HYPERPHENYLALANINEMIA. MILD. NON-BH4-DEFICIENT; HPANBH4 [AR]
DNAJC19	3-METHYLGLUTACONIC ACIDURIA. TYPE V; MGCA5 [AR]
DNAJC21	BONE MARROW FAILURE SYNDROME 3; BMFS3 [AR]
DNAJC5	CEROID LIPOFUSCINOSIS. NEURONAL. 4B. AUTOSOMAL DOMINANT; CLN4B [AD]
DNAJC6	PARKINSON DISEASE 19A. JUVENILE-ONSET; PARK19A [AR]
DNAL1	CILIARY DYSKINESIA. PRIMARY. 16; CILD16 [AR]
DNASE1L3	SYSTEMIC LUPUS ERYTHEMATOSUS 16; SLEB16 [AR]
DNM1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 31; EIEE31 [AD]
DNM1L	ENCEPHALOPATHY DUE TO DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION 1; EMPF1 [AD] | OPTIC ATROPHY 5; OPA5 [AD]
DNM2	LETHAL CONGENITAL CONTRACTURE SYNDROME 5; LCCS5 [AR] | CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE B; CMTDIB [AD] | MYOPATHY. CENTRONUCLEAR. 1; CNM1 [AD]
DNMBP	CATARACT 48; CTRCT48 [AR]
DNMT1	CEREBELLAR ATAXIA. DEAFNESS. AND NARCOLEPSY. AUTOSOMAL DOMINANT; ADCADN [AD] | NEUROPATHY. HEREDITARY SENSORY. TYPE IE; HSN1E [AD]
DNMT3A	LEUKEMIA. ACUTE MYELOID; AML [AD] | TATTON-BROWN-RAHMAN SYNDROME; TBRS [AD] | HEYN-SPROUL-JACKSON SYNDROME; HESJAS [AD]
DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1; ICF1 [AR]
DOCK2	IMMUNODEFICIENCY 40; IMD40 [AR]
DOCK3	NEURODEVELOPMENTAL DISORDER WITH IMPAIRED INTELLECTUAL DEVELOPMENT. HYPOTONIA. AND ATAXIA; NEDIDHA [AR]
DOCK6	ADAMS-OLIVER SYNDROME 2; AOS2 [AR]
DOCK7	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 23; EIEE23 [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]
DONSON	MICROCEPHALY-MICROMELIA SYNDROME; MIMIS [AR] | MICROCEPHALY. SHORT STATURE. AND LIMB ABNORMALITIES; MISSLA [AR]
DPAGT1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ij; CDG1J [AR] | MYASTHENIC SYNDROME. CONGENITAL. 13; CMS13 [AR]
DPF2	COFFIN-SIRIS SYNDROME 7; CSS7 [AD]
DPH1	DEVELOPMENTAL DELAY WITH SHORT STATURE. DYSMORPHIC FACIAL FEATURES. AND SPARSE HAIR; DEDSSH [AR]
DPM1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ie; CDG1E [AR]
DPM2	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Iu; CDG1U [AR]
DPM3	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 15; MDDGC15 [AR]
DPP6	VENTRICULAR FIBRILLATION. PAROXYSMAL FAMILIAL. 2; VF2 [AD]
DPY19L2	SPERMATOGENIC FAILURE 9; SPGF9 [AR]

DPYD	DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY [AR]
DPYS	DIHYDROPYRIMIDINASE DEFICIENCY; DPYSD [AR]
DRAM2	CONE-ROD DYSTROPHY 21; CORD21 [AR]
DRC1	CILIARY DYSKINESIA. PRIMARY. 21; CILD21 [AR]
DSC2	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 11; ARVD11 [AD]
DSE	EHLERS-DANLOS SYNDROME. MUSCULOCONTRACTURAL TYPE. 2; EDSMC2 [AR]
DSG1	PALMOPLANTAR KERATODERMA I. STRIATE. FOCAL. OR DIFFUSE; PPKS1 [AD] | ERYTHRODERMA. CONGENITAL. WITH PALMOPLANTAR KERATODERMA. HYPOTRICHOSIS. AND HYPER-IgE; EPKHE [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]
DSPP	DENTINOGENESIS IMPERFECTA 1; DGI1 [AD] | DEAFNESS. AUTOSOMAL DOMINANT 39. WITH DENTINOGENESIS IMPERFECTA 1 [AD] | DENTINOGENESIS IMPERFECTA. SHIELDS TYPE III [AD] | DENTIN DYSPLASIA. TYPE II; DTDP2 [AD]
DST	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE VI; HSAN6 [AR] | EPIDERMOLYSIS BULLOSA SIMPLEX. AUTOSOMAL RECESSIVE 2; EBSB2 [AR]
DSTYK	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT 1; CAKUT1 [AD] | SPASTIC PARAPLEGIA 23. AUTOSOMAL RECESSIVE; SPG23 [AR]
DTNA	LEFT VENTRICULAR NONCOMPACTION 1; LVNC1 [AD]
DTNBP1	HERMANSKY-PUDLAK SYNDROME 7; HPS7 [AR]
DUOX2	THYROID DYSHORMONOGENESIS 6; TDH6 [AR]
DUOXA2	THYROID DYSHORMONOGENESIS 5; TDH5 [AR]
DUSP6	HYPOGONADOTROPIC HYPOGONADISM 19 WITH OR WITHOUT ANOSMIA; HH19 [Autosomal dominant (see MISCELLANEOUS below)]
DVL1	ROBINOW SYNDROME. AUTOSOMAL DOMINANT 2; DRS2 [AD]
DVL3	ROBINOW SYNDROME. AUTOSOMAL DOMINANT 3; DRS3 [AD]
DYM	SMITH-MCCORT DYSPLASIA 1; SMC1 [AR] | DYGGVE-MELCHIOR-CLAUSEN DISEASE; DMC [AR]
DYNC1H1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 13; MRD13 [AD] | SPINAL MUSCULAR ATROPHY. LOWER EXTREMITY-PREDOMINANT. 1. AUTOSOMAL DOMINANT; SMALED1 [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2O; CMT2O [AD]
DYNC1I2	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY AND STRUCTURAL BRAIN ANOMALIES; NEDMIBA [AR]
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY; SRTD3 [AR]
DYNC2LI1	SHORT-RIB THORACIC DYSPLASIA 15 WITH POLYDACTYLY; SRTD15 [AR]
DYRK1A	MENTAL RETARDATION. AUTOSOMAL DOMINANT 7; MRD7 [AD]
DYRK1B	ABDOMINAL OBESITY-METABOLIC SYNDROME 3; AOMS3 [AD]
DYSF	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 2; LGMDR2 [AR] | MYOPATHY. DISTAL. WITH ANTERIOR TIBIAL ONSET; DMAT [AR] | MIYOSHI MUSCULAR DYSTROPHY 1; MMD1 [AR]
DZIP1L	POLYCYSTIC KIDNEY DISEASE 5; PKD5 [AR]
EARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 12; COXPD12 [AR]
EBF3	HYPOTONIA. ATAXIA. AND DELAYED DEVELOPMENT SYNDROME; HADDS [AD]
EBP	CHONDRODYSPLASIA PUNCTATA 2. X-LINKED DOMINANT; CDPX2 [XLD] | MEND SYNDROME; MEND [XLR] | CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS [XLD]
ECEL1	ARTHROGRYPOSIS. DISTAL. TYPE 5D; DA5D [AR]
ECHS1	MITOCHONDRIAL SHORT-CHAIN ENOYL-CoA HYDRATASE 1 DEFICIENCY; ECHS1D [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]
EDN1	AURICULOCONDYLAR SYNDROME 3; ARCND3 [AR] | QUESTION MARK EARS. ISOLATED; QME [AD]
EDN3	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD] | WAARDENBURG SYNDROME. TYPE 4B; WS4B [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 4; HSCR4 [AD]
EDNRA	MANDIBULOFACIAL DYSOSTOSIS WITH ALOPECIA; MFDA [AD] | MIGRAINE WITH OR WITHOUT AURA. SUSCEPTIBILITY TO. 1 [AD]
EDNRB	ABCD SYNDROME; ABCDS [AR] | WAARDENBURG SYNDROME. TYPE 4A; WS4A [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 2; HSCR2 [AD]
EED	COHEN-GIBSON SYNDROME; COGIS [AD]
EEF1A2	MENTAL RETARDATION. AUTOSOMAL DOMINANT 38; MRD38 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 33; EIEE33 [AD]
EEF1AKNMT	DEAFNESS. AUTOSOMAL RECESSIVE 26. MODIFIER OF; DFNB26M [AD]
EFEMP1	DOYNE HONEYCOMB RETINAL DYSTROPHY; DHRD [AD]
EFEMP2	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IB; ARCL1B [AR]
EFL1	SHWACHMAN-DIAMOND SYNDROME 2; SDS2 [AR]
EFNB1	CRANIOFRONTONASAL SYNDROME; CFNS [XLD]
EFTUD2	MANDIBULOFACIAL DYSOSTOSIS. GUION-ALMEIDA TYPE; MFDGA [AD]
EGF	Hypomagnesemia 4 renal 611718 (3)
EGFR	INFLAMMATORY SKIN AND BOWEL DISEASE. NEONATAL. 2; NISBD2 [AR] | LUNG CANCER [Somatic mutation]
EGLN1	ERYTHROCYTOSIS. FAMILIAL. 3; ECYT3 [AD] | HEMOGLOBIN. HIGH ALTITUDE ADAPTATION; HALAH [AD]
EGR2	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS [AD] | NEUROPATHY. CONGENITAL HYPOMYELINATING. 1. AUTOSOMAL RECESSIVE; CHN1 [AR] | CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1D; CMT1D [AD]
EHMT1	KLEEFSTRA SYNDROME 1; KLEFS1 [AD]
EIF2AK3	EPIPHYSEAL DYSPLASIA. MULTIPLE. WITH EARLY-ONSET DIABETES MELLITUS [AR]
EIF2AK4	PULMONARY VENOOCCLUSIVE DISEASE 2. AUTOSOMAL RECESSIVE; PVOD2 [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]
EIF2S3	MEHMO SYNDROME; MEHMO [XLR]
EIF3F	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 67; MRT67 [AR]
EIF4A3	ROBIN SEQUENCE WITH CLEFT MANDIBLE AND LIMB ANOMALIES [AR]
ELAC2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 17; COXPD17 [AR]
ELANE	CYCLIC NEUTROPENIA [AD] | NEUTROPENIA. SEVERE CONGENITAL. 1. AUTOSOMAL DOMINANT; SCN1 [AD]
ELMO2	VASCULAR MALFORMATION. PRIMARY INTRAOSSEOUS [AR]
ELN	SUPRAVALVULAR AORTIC STENOSIS; SVAS [AD] | CUTIS LAXA. AUTOSOMAL DOMINANT 1; ADCL1 [AD]
ELOVL1	ICHTHYOTIC KERATODERMA. SPASTICITY. HYPOMYELINATION. AND DYSMORPHIC FACIAL FEATURES; IKSHD [AD]
ELOVL4	SPINOCEREBELLAR ATAXIA 34; SCA34 [AD] | ICHTHYOSIS. SPASTIC QUADRIPLEGIA. AND MENTAL RETARDATION; ISQMR [AR] | STARGARDT DISEASE 3; STGD3 [AD]
ELOVL5	SPINOCEREBELLAR ATAXIA 38; SCA38 [AD]
ELP1	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE III; HSAN3 [AR]
ELP2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 58; MRT58 [AR]
EMC1	CEREBELLAR ATROPHY. VISUAL IMPAIRMENT. AND PSYCHOMOTOR RETARDATION; CAVIPMR [AR]
EMD	EMERY-DREIFUSS MUSCULAR DYSTROPHY 1. X-LINKED; EDMD1 [XLR]
EMG1	BOWEN-CONRADI SYNDROME; BWCNS [AR]
EML1	BAND HETEROTOPIA; BH [AR]

EMP2	NEPHROTIC SYNDROME. TYPE 10; NPHS10 [AR]
EMX2	Schizencephaly 269160 (3)
ENAM	AMELOGENESIS IMPERFECTA. TYPE IB; AI1B [AD] | AMELOGENESIS IMPERFECTA. TYPE IC; AI1C [AR]
ENG	TELANGIECTASIA. HEREDITARY HEMORRHAGIC. TYPE 1; HHT1 [AD]
ENO1	Enolase deficiency (1)
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]
ENTPD1	SPASTIC PARAPLEGIA 64. AUTOSOMAL RECESSIVE; SPG64 [AR]
EOGT	ADAMS-OLIVER SYNDROME 4; AOS4 [AR]
EP300	RUBINSTEIN-TAYBI SYNDROME 2; RSTS2 [AD] | COLORECTAL CANCER; CRC [Somatic mutation]
EPAS1	Erythrocytosis familial 4 611783 (3)
EPB41	ELLIPTOCYTOSIS 1; EL1 [AD]
EPB42	Spherocytosis type 5 612690 (3)
EPCAM	DIARRHEA 5. WITH TUFTING ENTEROPATHY. CONGENITAL; DIAR5 [AR]
EPG5	VICI SYNDROME; VICIS [AR]
EPHA2	CATARACT 6. MULTIPLE TYPES; CTRCT6 [AD]
EPHB4	LYMPHATIC MALFORMATION 7; LMPHM7 [AD] | CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION 2; CMAVM2 [AD]
EPM2A	MYOCLONIC EPILEPSY OF LAFORA [AR]
EPO	DIAMOND-BLACKFAN ANEMIA-LIKE; DBAL [AR] | ERYTHROCYTOSIS. FAMILIAL. 5; ECYT5 [AD]
EPRS1	LEUKODYSTROPHY. HYPOMYELINATING. 15; HLD15 [AR]
EPS8L2	DEAFNESS. AUTOSOMAL RECESSIVE 106; DFNB106 [AR]
ERAL1	PERRAULT SYNDROME 6; PRLTS6 [AR]
ERBB2	GLIOMA SUSCEPTIBILITY 1; GLM1 [AD] | LUNG CANCER [Somatic mutation]
ERBB3	ERYTHROLEUKEMIA. FAMILIAL. SUSCEPTIBILITY TO; FERLK [AD] | LETHAL CONGENITAL CONTRACTURE SYNDROME 2; LCCS2 [AR]
ERBB4	AMYOTROPHIC LATERAL SCLEROSIS 19; ALS19 [AD]
ERCC1	CEREBROOCULOFACIOSKELETAL SYNDROME 4; COFS4 [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]
ERCC6L2	BONE MARROW FAILURE SYNDROME 2; BMFS2 [AR]
ERCC8	COCKAYNE SYNDROME A; CSA [AR] | UV-SENSITIVE SYNDROME 2; UVSS2 [AR]
ERF	CHITAYAT SYNDROME; CHYTS [AD] | CRANIOSYNOSTOSIS 4; CRS4 [AD]
ERLIN1	SPASTIC PARAPLEGIA 62. AUTOSOMAL RECESSIVE; SPG62 [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]
ESR1	MIGRAINE WITH OR WITHOUT AURA. SUSCEPTIBILITY TO. 1 [AD] | BREAST CANCER [AD] | ESTROGEN RESISTANCE; ESTRR [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]
ETV6	THROMBOCYTOPENIA 5; THC5 [AD] | LEUKEMIA. ACUTE MYELOID; AML [AD]
EVC	ELLIS-VAN CREVELD SYNDROME; EVC [AR] | WEYERS ACROFACIAL DYSOSTOSIS; WAD [AD]
EVC2	ELLIS-VAN CREVELD SYNDROME; EVC [AR] | WEYERS ACROFACIAL DYSOSTOSIS; WAD [AD]
EWSR1	Ewing sarcoma 612219 (3) | Neuroepithelioma 612219 (3)
EXOC6B	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY. TYPE 3; SEMDJL3 [AR]
EXOSC2	SHORT STATURE. HEARING LOSS. RETINITIS PIGMENTOSA. AND DISTINCTIVE FACIES; SHRF [AR]
EXOSC3	PONTOCEREBELLAR HYPOPLASIA. TYPE 1B; PCH1B [AR]
EXOSC8	PONTOCEREBELLAR HYPOPLASIA. TYPE 1C; PCH1C [AR]
EXOSC9	PONTOCEREBELLAR HYPOPLASIA. TYPE 1D; PCH1D [AR]
EXPH5	EPIDERMOLYSIS BULLOSA. NONSPECIFIC. AUTOSOMAL RECESSIVE; EBNS [AR]
EXT1	CHONDROSARCOMA [AR] | EXOSTOSES. MULTIPLE. TYPE I; EXT1 [AD] | TRICHORHINOPHALANGEAL SYNDROME. TYPE II; TRPS2 [AD]
EXT2	SEIZURES. SCOLIOSIS. AND MACROCEPHALY/MICROCEPHALY SYNDROME; SSMS [AR] | EXOSTOSES. MULTIPLE. TYPE II; EXT2 [AD]
EXTL3	IMMUNOSKELETAL DYSPLASIA WITH NEURODEVELOPMENTAL ABNORMALITIES; ISDNA [AR]
EYA1	OTOFACIOCERVICAL SYNDROME 1; OTFCS [AD] | BRANCHIOOTIC SYNDROME 1; BOS1 [AD] | BRANCHIOOTORENAL SYNDROME 1; BOR1 [AD]
EYA4	DEAFNESS. AUTOSOMAL DOMINANT 10; DFNA10 [AD] | CARDIOMYOPATHY. DILATED. 1J; CMD1J [AD]
EYS	RETINITIS PIGMENTOSA 25; RP25 [AR]
EZH2	WEAVER SYNDROME; WVS [AD]
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]
F13A1	FACTOR XIII. A SUBUNIT. DEFICIENCY OF [AR] | THROMBOPHILIA DUE TO THROMBIN DEFECT; THPH1 [AD]
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]
FAM111A	GRACILE BONE DYSPLASIA; GCLEB [AD] | KENNY-CAFFEY SYNDROME. TYPE 2; KCS2 [AD]
FAM111B	POIKILODERMA. HEREDITARY FIBROSING. WITH TENDON CONTRACTURES. MYOPATHY. AND PULMONARY FIBROSIS; POIKTMP [AD]
FAM126A	LEUKODYSTROPHY. HYPOMYELINATING. 5; HLD5 [AR]
FAM161A	Retinitis pigmentosa 28 606068 (3)
FAM20A	AMELOGENESIS IMPERFECTA. TYPE IG; AI1G [AR]
FAM20C	RAINE SYNDROME; RNS [AR]
FAM83H	AMELOGENESIS IMPERFECTA. TYPE IIIA; AI3A [AD]
FAN1	INTERSTITIAL NEPHRITIS. KARYOMEGALIC; KMIN [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]
FANCF	Fanconi anemia complementation group F 603467 (3)
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]
FAR1	PEROXISOMAL FATTY ACYL-CoA REDUCTASE 1 DISORDER; PFCRD [AR]
FARS2	SPASTIC PARAPLEGIA 77. AUTOSOMAL RECESSIVE; SPG77 [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 14; COXPD14 [AR]
FARSB	RAJAB INTERSTITIAL LUNG DISEASE WITH BRAIN CALCIFICATIONS; RILDBC [AR]
FAS	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS [AD]
FASLG	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME; ALPS [AD] | LUNG CANCER [Somatic mutation]
FAT2	SPINOCEREBELLAR ATAXIA 45; SCA45 [AD]
FAT4	VAN MALDERGEM SYNDROME 2; VMLDS2 [AR] | HENNEKAM LYMPHANGIECTASIA-LYMPHEDEMA SYNDROME 2; HKLLS2 [AR]
FBLN1	SYNPOLYDACTYLY 2; SPD2 [AD]
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]
FBN2	CONTRACTURAL ARACHNODACTYLY. CONGENITAL; CCA [AD] | MACULAR DEGENERATION. EARLY-ONSET; EOMD [AD]
FBP1	FRUCTOSE-1.6-BISPHOSPHATASE DEFICIENCY; FBP1D [AR]
FBXL3	INTELLECTUAL DEVELOPMENTAL DISORDER WITH SHORT STATURE. FACIAL ANOMALIES. AND SPEECH DEFECTS; IDDSFAS [AR]
FBXL4	MITOCHONDRIAL DNA DEPLETION SYNDROME 13 (ENCEPHALOMYOPATHIC TYPE); MTDPS13 [AR]
FBXO11	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND BEHAVIORAL ABNORMALITIES; IDDFBA [AD]
FBXO38	NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE IID; HMN2D [AD]
FBXO7	PARKINSON DISEASE 15. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK15 [AR]
FCGR3A	IMMUNODEFICIENCY 20; IMD20 [AR]
FCGR3B	Neutropenia alloimmune neonatal (3)
FCN3	FICOLIN 3 DEFICIENCY [AR]
FCSK	CONGENITAL DISORDER OF GLYCOSYLATION WITH DEFECTIVE FUCOSYLATION 2; CDGF2 [AR]
FDFT1	SQUALENE SYNTHASE DEFICIENCY; SQSD [AR]
FDPS	POROKERATOSIS 9. MULTIPLE TYPES; POROK9 [AD]
FDX2	MITOCHONDRIAL MYOPATHY. EPISODIC. WITH OR WITHOUT OPTIC ATROPHY AND REVERSIBLE LEUKOENCEPHALOPATHY; MEOAL [AR]
FDXR	AUDITORY NEUROPATHY AND OPTIC ATROPHY; ANOA [AR]
FECH	PROTOPORPHYRIA. ERYTHROPOIETIC. 1; EPP1 [AR]
FERMT1	KINDLER SYNDROME; KNDLRS [AR]
FERMT3	LEUKOCYTE ADHESION DEFICIENCY. TYPE III; LAD3 [AR]
FEZF1	HYPOGONADOTROPIC HYPOGONADISM 22 WITH OR WITHOUT ANOSMIA; HH22 [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]
FGF10	APLASIA OF LACRIMAL AND SALIVARY GLANDS; ALSG [AD] | LACRIMOAURICULODENTODIGITAL SYNDROME; LADD [AD]
FGF12	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 47; EIEE47 [AD]
FGF14	SPINOCEREBELLAR ATAXIA 27; SCA27 [AD]
FGF16	METACARPAL 4-5 FUSION; MF4 [XLR]
FGF17	HYPOGONADOTROPIC HYPOGONADISM 20 WITH OR WITHOUT ANOSMIA; HH20 [Autosomal dominant (see MISCELLANEOUS below)]
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]
FGF5	TRICHOMEGALY; TCMGLY [AR]
FGF8	HYPOGONADOTROPIC HYPOGONADISM 6 WITH OR WITHOUT ANOSMIA; HH6 [AD]
FGF9	MULTIPLE SYNOSTOSES SYNDROME 3; SYNS3 [AD]
FGFR1	ENCEPHALOCRANIOCUTANEOUS LIPOMATOSIS; ECCL [Somatic mosaicism] | HARTSFIELD SYNDROME; HRTFDS [AD] | OSTEOGLOPHONIC DYSPLASIA; OGD [AD] | JACKSON-WEISS SYNDROME; JWS [AD] | TRIGONOCEPHALY 1; TRIGNO1 [AD] | PFEIFFER SYNDROME [AD] | HYPOGONADOTROPIC HYPOGONADISM 2 WITH OR WITHOUT ANOSMIA; HH2 [AD]
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]
FGFR3	CAMPTODACTYLY. TALL STATURE. AND HEARING LOSS SYNDROME; CATSHLS [AD] | THANATOPHORIC DYSPLASIA. TYPE II; TD2 [AD] | TESTICULAR GERM CELL TUMOR; TGCT [Somatic mutation] | BLADDER CANCER [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation] | MUENKE SYNDROME; MNKES [AD] | LACRIMOAURICULODENTODIGITAL SYNDROME; LADD [AD] | CROUZON SYNDROME WITH ACANTHOSIS NIGRICANS; CAN [AD] | NEVUS. EPIDERMAL [Somatic mutation] | ACHONDROPLASIA. SEVERE. WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN [AD] | HYPOCHONDROPLASIA; HCH [AD] | ACHONDROPLASIA; ACH [AD] | THANATOPHORIC DYSPLASIA. TYPE I; TD1 [AD]
FGG	AFIBRINOGENEMIA. CONGENITAL [AR]
FH	Fumarase deficiency 606812 (3) | Leiomyomatosis and renal cell cancer 150800 (3)
FHL1	SCAPULOPERONEAL MYOPATHY. X-LINKED DOMINANT; SPM [XLD] | URUGUAY FACIOCARDIOMUSCULOSKELETAL SYNDROME; FCMSU [XLR] | HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 1; FHL1 [AR] | REDUCING BODY MYOPATHY. X-LINKED 1B. WITH LATE CHILDHOOD OR ADULT ONSET; RBMX1B [X-linked] | MYOPATHY. X-LINKED. WITH POSTURAL MUSCLE ATROPHY; XMPMA [XLR] | REDUCING BODY MYOPATHY. X-LINKED 1A. SEVERE. WITH INFANTILE OR EARLY CHILDHOOD ONSET; RBMX1A [XLD]
FIBP	THAUVIN-ROBINET-FAIVRE SYNDROME; TROFAS [AR]
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]
FIGLA	PREMATURE OVARIAN FAILURE 6; POF6 [AD]
FITM2	SIDDIQI SYNDROME; SIDDIS [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]
FLAD1	LIPID STORAGE MYOPATHY DUE TO FLAVIN ADENINE DINUCLEOTIDE SYNTHETASE DEFICIENCY; LSMFLAD [AR]
FLCN	PNEUMOTHORAX. PRIMARY SPONTANEOUS [AD] | BIRT-HOGG-DUBE SYNDROME; BHD [AD] | COLORECTAL CANCER; CRC [Somatic mutation]
FLG	ICHTHYOSIS VULGARIS [AD]
FLG2	PEELING SKIN SYNDROME 6; PSS6 [AR]
FLI1	BLEEDING DISORDER. PLATELET-TYPE. 21; BDPLT21 [AD] | THROMBOCYTOPENIA. PARIS-TROUSSEAU TYPE; TCPT [Isolated cases]
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]
FLNC	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 26; CMH26 [AD] | MYOPATHY. DISTAL. 4; MPD4 [AD] | MYOPATHY. MYOFIBRILLAR. 5; MFM5 [AD]
FLRT3	HYPOGONADOTROPIC HYPOGONADISM 21 WITH OR WITHOUT ANOSMIA; HH21 [Autosomal dominant (see MISCELLANEOUS below)]
FLT3	LEUKEMIA. ACUTE MYELOID; AML [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]
FMN2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 47; MRT47 [AR]
FMO3	TRIMETHYLAMINURIA; TMAU [AR]
FMR1	PREMATURE OVARIAN FAILURE 1; POF1 [X-linked] | FRAGILE X TREMOR/ATAXIA SYNDROME; FXTAS [XLD] | FRAGILE X SYNDROME; FXS [XLD]
FN1	SPONDYLOMETAPHYSEAL DYSPLASIA. CORNER FRACTURE TYPE; SMDCF [AD] | GLOMERULOPATHY WITH FIBRONECTIN DEPOSITS 2; GFND2 [AD]
FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY [AR]
FOXC1	AXENFELD-RIEGER SYNDROME. TYPE 3; RIEG3 [AD] | ANTERIOR SEGMENT DYSGENESIS 3; ASGD3 [AD]
FOXC2	LYMPHEDEMA-DISTICHIASIS SYNDROME; LPHDST [AD]
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]
FOXG1	RETT SYNDROME. CONGENITAL VARIANT [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]
FOXP1	MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND WITH OR WITHOUT AUTISTIC FEATURES [AD]
FOXP2	SPEECH-LANGUAGE DISORDER 1; SPCH1 [AD]
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]
FRMPD4	MENTAL RETARDATION. X-LINKED 104; MRX104 [X-linked]
FRRS1L	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 37; EIEE37 [AR]
FSCN2	Retinitis pigmentosa 30 607921 (3)
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]
FSIP2	SPERMATOGENIC FAILURE 34; SPGF34 [AR]
FTCD	GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY [AR]
FTL	HYPERFERRITINEMIA WITH OR WITHOUT CATARACT; HRFTC [AD] | L-FERRITIN DEFICIENCY; LFTD [AD] | NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 3; NBIA3 [AD]

FTO	GROWTH RETARDATION. DEVELOPMENTAL DELAY. AND FACIAL DYSMORPHISM; GDFD [AR] | BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 14; BMIQ14 [AR]
FTSJ1	MENTAL RETARDATION. X-LINKED 9; MRX9 [XLR]
FUCA1	FUCOSIDOSIS [AR]
FUS	TREMOR. HEREDITARY ESSENTIAL. 4; ETM4 [AD]
FUT6	Fucosyltransferase 6 deficiency 613852 (3)
FUT8	CONGENITAL DISORDER OF GLYCOSYLATION WITH DEFECTIVE FUCOSYLATION 1; CDGF1 [AR]
FXN	FRIEDREICH ATAXIA; FRDA [AR]
FXYD2	HYPOMAGNESEMIA 2. RENAL; HOMG2 [AD]
FYB1	THROMBOCYTOPENIA 3; THC3 [AR]
FYCO1	CATARACT 18; CTRCT18 [AR]
FZD2	OMODYSPLASIA 2; OMOD2 [AD]
FZD4	EXUDATIVE VITREORETINOPATHY 1; EVR1 [AD]
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]
GABBR2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 59; EIEE59 [AD] | TOBACCO ADDICTION. SUSCEPTIBILITY TO [Genetic factors seem to contribute only in light smokers and heavy smoker] | NEURODEVELOPMENTAL DISORDER WITH POOR LANGUAGE AND LOSS OF HAND SKILLS; NDPLHS [AD]
GABRA1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 19; EIEE19 [AD]
GABRA2	ALCOHOL DEPENDENCE [Probably multifactorial, genetically influence] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 78; EIEE78 [AD]
GABRA5	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 79; EIEE79 [AD]
GABRB1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 45; EIEE45 [AD]
GABRB2	EPILEPTIC ENCEPHALOPATHY. INFANTILE OR EARLY CHILDHOOD. 2; IECEE2 [AD]
GABRB3	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 43; EIEE43 [AD]
GABRG2	FEBRILE SEIZURES. FAMILIAL. 8; FEB8 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 74; EIEE74 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 6; EIEE6 [AD]
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]
GANAB	POLYCYSTIC KIDNEY DISEASE 3 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD3 [AD]
GARS1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2D; CMT2D [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VA; HMN5A [AD]
GAS8	CILIARY DYSKINESIA. PRIMARY. 33; CILD33 [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]
GATA2	LYMPHEDEMA. PRIMARY. WITH MYELODYSPLASIA [AD] | LEUKEMIA. ACUTE MYELOID; AML [AD] | IMMUNODEFICIENCY 21; IMD21 [AD]
GATA3	HYPOPARATHYROIDISM. SENSORINEURAL DEAFNESS. AND RENAL DISEASE; HDR [AD]

GATA4	ATRIOVENTRICULAR SEPTAL DEFECT 4; AVSD4 [AD] | VENTRICULAR SEPTAL DEFECT 1; VSD1 [AD] | TESTICULAR ANOMALIES WITH OR WITHOUT CONGENITAL HEART DISEASE; TACHD [AD] | ATRIAL SEPTAL DEFECT 2; ASD2 [AD] | TETRALOGY OF FALLOT; TOF [AD]
GATA5	CONGENITAL HEART DEFECTS. MULTIPLE TYPES. 5; CHTD5 [AD]
GATA6	ATRIAL SEPTAL DEFECT 9; ASD9 [AD] | HEART DEFECTS. CONGENITAL. AND OTHER CONGENITAL ANOMALIES; HDCA [AD] | ATRIOVENTRICULAR SEPTAL DEFECT 5; AVSD5 [AD] | TETRALOGY OF FALLOT; TOF [AD]
GATAD2B	MENTAL RETARDATION. AUTOSOMAL DOMINANT 18; MRD18 [AD]
GATM	CEREBRAL CREATINE DEFICIENCY SYNDROME 3; CCDS3 [AR]
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]
GCK	MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 2; MODY2 [AD] | DIABETES MELLITUS. PERMANENT NEONATAL; PNDM [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 3; HHF3 [AD]
GCLC	GAMMA-GLUTAMYLCYSTEINE SYNTHETASE DEFICIENCY. HEMOLYTIC ANEMIA DUE TO [AR]
GCM2	HYPOPARATHYROIDISM. FAMILIAL ISOLATED; FIH [AD] | HYPERPARATHYROIDISM 4; HRPT4 [AD]
GCNT2	CATARACT 13 WITH ADULT i PHENOTYPE; CTRCT13 [AR] | BLOOD GROUP. I SYSTEM; Ii [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]
GDAP2	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 27; SCAR27 [AR]
GDF1	RIGHT ATRIAL ISOMERISM; RAI [AR] | CONGENITAL HEART DEFECTS. MULTIPLE TYPES. 6; CHTD6 [AD]
GDF2	TELANGIECTASIA. HEREDITARY HEMORRHAGIC. TYPE 5; HHT5 [AD]
GDF3	MICROPHTHALMIA. ISOLATED 7; MCOP7 [AD] | MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 6; MCOPCB6 [AD]
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]
GDF6	LEBER CONGENITAL AMAUROSIS 17; LCA17 [AR] | MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 6; MCOPCB6 [AD] | MULTIPLE SYNOSTOSES SYNDROME 4; SYNS4 [AD] | KLIPPEL-FEIL SYNDROME 1. AUTOSOMAL DOMINANT; KFS1 [AD]
GDI1	MENTAL RETARDATION. X-LINKED 41; MRX41 [XLD]
GDNF	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 3; HSCR3 [AD] | PHEOCHROMOCYTOMA [AD]
GEMIN4	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. CATARACTS. AND RENAL ABNORMALITIES; NEDMCR [AR]
GFAP	ALEXANDER DISEASE; ALXDRD [AD]
GFER	Myopathy mitochondrial progressive with congenital cataract hearing loss and developmental delay 613076 (3)
GFI1B	BLEEDING DISORDER. PLATELET-TYPE. 17; BDPLT17 [AD]
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1; COXPD1 [AR]
GFM2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 39; COXPD39 [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]
GHRH	?Isolated growth hormone deficiency due to defect in GHRF (1) | Gigantism due to GHRF hypersecretion (1)
GHRHR	Growth hormone deficiency isolated type IV 618157 (3)
GHSR	GROWTH HORMONE DEFICIENCY. ISOLATED PARTIAL; GHDP [AD]
GINS1	IMMUNODEFICIENCY 55; IMD55 [AR]

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]
GJA3	CATARACT 14. MULTIPLE TYPES; CTRCT14 [AD]
GJA5	ATRIAL STANDSTILL 1; ATRST1 [AD] | ATRIAL FIBRILLATION. FAMILIAL. 11; ATFB11 [AD]
GJA8	CATARACT 1. MULTIPLE TYPES; CTRCT1 [AD]
GJB1	CHARCOT-MARIE-TOOTH DISEASE. X-LINKED DOMINANT. 1; CMTX1 [XLD]
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]
GK	Glycerol kinase deficiency 307030 (3)
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]
GLDN	LETHAL CONGENITAL CONTRACTURE SYNDROME 11; LCCS11 [AR]
GLE1	LETHAL CONGENITAL CONTRACTURE SYNDROME 1; LCCS1 [AR] | CONGENITAL ARTHROGRYPOSIS WITH ANTERIOR HORN CELL DISEASE; CAAHD [AR]
GLI1	POLYDACTYLY. PREAXIAL I; PPD1 [AR] | POLYDACTYLY. POSTAXIAL. TYPE A8; PAPA8 [AR]
GLI2	CULLER-JONES SYNDROME; CJS [AD] | HOLOPROSENCEPHALY 9; HPE9 [AD]
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]
GLMN	GLOMUVENOUS MALFORMATIONS; GVM [AD]
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]
GLS	GLOBAL DEVELOPMENTAL DELAY. PROGRESSIVE ATAXIA. AND ELEVATED GLUTAMINE; GDPAG [AR] | INFANTILE CATARACT. SKIN ABNORMALITIES. GLUTAMATE EXCESS. AND IMPAIRED INTELLECTUAL DEVELOPMENT; CASGID [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 71; EIEE71 [AR]
GLUD1	HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 6; HHF6 [AD]
GLUL	GLUTAMINE DEFICIENCY. CONGENITAL [AR]
GLYCTK	D-GLYCERIC ACIDURIA [AR]
GM2A	GM2-GANGLIOSIDOSIS. AB VARIANT [AR]
GMNN	MEIER-GORLIN SYNDROME 6; MGORS6 [AD]
GMPPA	ALACRIMA. ACHALASIA. AND MENTAL RETARDATION SYNDROME; AAMR [AR]
GMPPB	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 14; MDDGA14 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 14; MDDGC14 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION). TYPE B. 14; MDDGB14 [AR]
GNA11	HYPOCALCIURIC HYPERCALCEMIA. FAMILIAL. TYPE II; HHC2 [Autosomal dominant with locus heterogeneity (19p13.3] | HYPOCALCEMIA. AUTOSOMAL DOMINANT 2; HYPOC2 [AD]

GNAI2	VENTRICULAR TACHYCARDIA. FAMILIAL [AD]
GNAI3	AURICULOCONDYLAR SYNDROME 1; ARCND1 [AD]
GNAL	DYSTONIA 25; DYT25 [AD]
GNAO1	NEURODEVELOPMENTAL DISORDER WITH INVOLUNTARY MOVEMENTS; NEDIM [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 17; EIEE17 [AD]
GNAQ	STURGE-WEBER SYNDROME; SWS [Somatic mosaicism] | CAPILLARY MALFORMATIONS. CONGENITAL; CMC [AD]
GNAS	PSEUDOHYPOPARATHYROIDISM. TYPE IA; PHP1A [AD] | OSSEOUS HETEROPLASIA. PROGRESSIVE; POH [AD] | ACTH-INDEPENDENT MACRONODULAR ADRENAL HYPERPLASIA; AIMAH1 [Somatic mutation] | PSEUDOHYPOPARATHYROIDISM. TYPE IC; PHP1C [AD] | PSEUDOPSEUDOHYPOPARATHYROIDISM; PPHP [AD] | MCCUNE-ALBRIGHT SYNDROME; MAS [Somatic mosaicism] | PSEUDOHYPOPARATHYROIDISM. TYPE IB; PHP1B [AD]
GNAS-AS1	PSEUDOHYPOPARATHYROIDISM. TYPE IB; PHP1B [AD]
GNAT1	NIGHT BLINDNESS. CONGENITAL STATIONARY. AUTOSOMAL DOMINANT 3; CSNBAD3 [AD] | NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1G; CSNB1G [AR]
GNAT2	Achromatopsia 4 613856 (3)
GNB1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 42; MRD42 [AD]
GNB3	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1H; CSNB1H [AR] | HYPERTENSION. ESSENTIAL [Multifactorial]
GNB4	CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE F; CMTDIF [AD]
GNB5	INTELLECTUAL DEVELOPMENTAL DISORDER WITH CARDIAC ARRHYTHMIA; IDDCA [AR] | LANGUAGE DELAY AND ATTENTION DEFICIT-HYPERACTIVITY DISORDER/COGNITIVE IMPAIRMENT WITH OR WITHOUT CARDIAC ARRHYTHMIA; LADCI [AR]
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]
GOT1	Aspartate aminotransferase serum level of QTL1 614419 (3)
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]
GPAA1	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 15; GPIBD15 [AR]
GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME. TYPE 1; SGBS1 [XLR] | WILMS TUMOR 1; WT1 [AD]
GPC4	KEIPERT SYNDROME; KPTS [XLR]
GPC6	OMODYSPLASIA 1; OMOD1 [AR]
GPD1	HYPERTRIGLYCERIDEMIA. TRANSIENT INFANTILE; HTGTI [AR]
GPD1L	Brugada syndrome 2 611777 (3)
GPHN	MOLYBDENUM COFACTOR DEFICIENCY. COMPLEMENTATION GROUP C; MOCODC [AR]
GPI	HEMOLYTIC ANEMIA. NONSPHEROCYTIC. DUE TO GLUCOSE PHOSPHATE ISOMERASE DEFICIENCY [AR]
GPIHBP1	HYPERLIPOPROTEINEMIA. TYPE ID [AR]
GPNMB	AMYLOIDOSIS. PRIMARY LOCALIZED CUTANEOUS. 3; PLCA3 [AR]
GPR101	PITUITARY ADENOMA 2. GROWTH HORMONE-SECRETING; PITA2 [X-linked]
GPR143	ALBINISM. OCULAR. TYPE I; OA1 [X-linked] | NYSTAGMUS 6. CONGENITAL. X-LINKED; NYS6 [XLR]
GPR179	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1E; CSNB1E [AR]

GPR68	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA6; AI2A6 [AR]
GPSM2	CHUDLEY-MCCULLOUGH SYNDROME; CMCS [AR]
GPT2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 49; MRT49 [AR]
GPX1	GLUTATHIONE PEROXIDASE DEFICIENCY; GPXD [AR]
GPX4	SPONDYLOMETAPHYSEAL DYSPLASIA. SEDAGHATIAN TYPE; SMDS [AR]
GRAP	DEAFNESS. AUTOSOMAL RECESSIVE 114; DFNB114 [AR]
GREB1L	RENAL HYPODYSPLASIA/APLASIA 3; RHDA3 [AD]
GREM2	TOOTH AGENESIS. SELECTIVE. 9; STHAG9 [AD]
GRHL2	CORNEAL DYSTROPHY. POSTERIOR POLYMORPHOUS. 4; PPCD4 [AD] | ECTODERMAL DYSPLASIA/SHORT STATURE SYNDROME; ECTDS [AR] | DEAFNESS. AUTOSOMAL DOMINANT 28; DFNA28 [AD]
GRHL3	VAN DER WOUDE SYNDROME 2; VWS2 [AD]
GRHPR	HYPEROXALURIA. PRIMARY. TYPE II; HP2 [AR]
GRIA3	MENTAL RETARDATION. X-LINKED. SYNDROMIC. WU TYPE; MRXSW [XLR]
GRIA4	NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT SEIZURES AND GAIT ABNORMALITIES; NEDSGA [AD]
GRID2	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 18; SCAR18 [AR]
GRIK2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 6; MRT6 [AR]
GRIN1	NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT HYPERKINETIC MOVEMENTS AND SEIZURES. AUTOSOMAL RECESSIVE; NDHMSR [AR] | NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT HYPERKINETIC MOVEMENTS AND SEIZURES. AUTOSOMAL DOMINANT; NDHMSD [AD]
GRIN2A	EPILEPSY. FOCAL. WITH SPEECH DISORDER AND WITH OR WITHOUT MENTAL RETARDATION; FESD [AD]
GRIN2B	MENTAL RETARDATION. AUTOSOMAL DOMINANT 6. WITH OR WITHOUT SEIZURES; MRD6 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 27; EIEE27 [AD]
GRIN2D	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 46; EIEE46 [AD]
GRIP1	FRASER SYNDROME 3; FRASRS3 [AR]
GRK1	Oguchi disease-2 613411 (3)
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]
GSC	SHORT STATURE. AUDITORY CANAL ATRESIA. MANDIBULAR HYPOPLASIA. AND SKELETAL ABNORMALITIES; SAMS [AR]
GSDME	DEAFNESS. AUTOSOMAL DOMINANT 5; DFNA5 [AD]
GSN	AMYLOIDOSIS. FINNISH TYPE [AD]
GSR	HEMOLYTIC ANEMIA DUE TO GLUTATHIONE REDUCTASE DEFICIENCY [AR]
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY; GSSD [AR] | GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES. HEMOLYTIC ANEMIA DUE TO; GSSDE [AR]
GSX2	DIENCEPHALIC-MESENCEPHALIC JUNCTION DYSPLASIA SYNDROME 2; DMJDS2 [AR]
GTF2E2	TRICHOTHIODYSTROPHY 6. NONPHOTOSENSITIVE; TTD6 [AR]
GTF2H5	Trichothiodystrophy 3 photosensitive 616395 (3)
GTPBP2	JABERI-ELAHI SYNDROME; JABELS [AR]
GTPBP3	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 23; COXPD23 [AR]
GUCA1A	CONE DYSTROPHY 3; COD3 [AD]
GUCA1B	Retinitis pigmentosa 48 613827 (3)
GUCY1A1	MOYAMOYA DISEASE 6 WITH OR WITHOUT ACHALASIA; MYMY6 [AR]
GUCY2C	MECONIUM ILEUS [AR] | DIARRHEA 6; DIAR6 [AD]
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]

GULOP	Scurvy (3)
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]
GZF1	JOINT LAXITY. SHORT STATURE. AND MYOPIA; JLSM [AR]
H1-4	RAHMAN SYNDROME; RMNS [AD]
H19	BECKWITH-WIEDEMANN SYNDROME; BWS [AD] | SILVER-RUSSELL SYNDROME; SRS [Autosomal dominant (loss of paternal allele)] | WILMS TUMOR 2; WT2 [AD]
H6PD	CORTISONE REDUCTASE DEFICIENCY 1; CORTRD1 [AR]
HAAO	VERTEBRAL. CARDIAC. RENAL. AND LIMB DEFECTS SYNDROME 1; VCRL1 [AR]
HABP2	THYROID CANCER. NONMEDULLARY. 5; NMTC5 [AD] | THROMBOPHILIA DUE TO THROMBIN DEFECT; THPH1 [AD]
HACE1	SPASTIC PARAPLEGIA AND PSYCHOMOTOR RETARDATION WITH OR WITHOUT SEIZURES; SPPRS [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]
HARS1	USHER SYNDROME. TYPE IIIB; USH3B [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2W; CMT2W [AD]
HAVCR2	T-CELL LYMPHOMA. SUBCUTANEOUS PANNICULITIS-LIKE; SPTCL [AR]
HAX1	NEUTROPENIA. SEVERE CONGENITAL. 3. AUTOSOMAL RECESSIVE; SCN3 [AR]
HBA1	HEINZ BODY ANEMIAS [AD]
HBA2	HEINZ BODY ANEMIAS [AD]
HBB	HEINZ BODY ANEMIAS [AD] | SICKLE CELL ANEMIA [AR] | FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 [AD]
HBD	Thalassemia due to Hb Lepore (3) | Thalassemia delta- (3)
HBG1	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 [AD]
HBG2	CYANOSIS. TRANSIENT NEONATAL; TNCY [AD] | FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 [AD]
HCCS	LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 1; LSDMCA1 [XLD]
HCFC1	METHYLMALONIC ACIDEMIA AND HOMOCYSTEINEMIA. cblX TYPE [XLR]
HCN1	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS. TYPE 10; GEFSP10 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 24; EIEE24 [AD]
HCN4	SICK SINUS SYNDROME 2; SSS2 [AD]
HDAC8	CORNELIA DE LANGE SYNDROME 5; CDLS5 [XLD]
HECW2	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA. SEIZURES. AND ABSENT LANGUAGE; NDHSAL [AD]
HELLS	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 4; ICF4 [AR]
HEPACAM	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 2B. REMITTING. WITH OR WITHOUT MENTAL RETARDATION; MLC2B [AD] | MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS 2A; MLC2A [AR]
HERC1	MACROCEPHALY. DYSMORPHIC FACIES. AND PSYCHOMOTOR RETARDATION; MDFPMR [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]
HFM1	PREMATURE OVARIAN FAILURE 9; POF9 [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]
HIKESHI	LEUKODYSTROPHY. HYPOMYELINATING. 13; HLD13 [AR]
HINT1	NEUROMYOTONIA AND AXONAL NEUROPATHY. AUTOSOMAL RECESSIVE; NMAN [AR]
HIVEP2	MENTAL RETARDATION. AUTOSOMAL DOMINANT 43; MRD43 [AD]
HJV	HEMOCHROMATOSIS. TYPE 2A; HFE2A [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]
HMGA2	LEIOMYOMA. UTERINE; UL [Somatic mutation]
HMGCL	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY; HMGCLD [AR]
HMGCS2	3-HYDROXY-3-METHYLGLUTARYL-CoA SYNTHASE-2 DEFICIENCY; HMGCS2D [AR]
HMOX1	Heme oxygenase-1 deficiency 614034 (3) | {Pulmonary disease chronic obstructive susceptibility to} 606963 (3)
HMX1	OCULOAURICULAR SYNDROME; OCACS [AR]
HNF1A	DIABETES MELLITUS. TYPE I [Autosomal recessive susceptibility] | MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 3; MODY3 [Autosomal dominant (12q22-qter)] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | HEPATIC ADENOMAS. FAMILIAL [AD]
HNF1B	RENAL CYSTS AND DIABETES SYNDROME; RCAD [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
HNF4A	MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 1; MODY1 [Autosomal dominant (20q12-q13.1)] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | FANCONI RENOTUBULAR SYNDROME 4 WITH MATURITY-ONSET DIABETES OF THE YOUNG; FRTS4 [AD]
HNMT	ASTHMA. SUSCEPTIBILITY TO [Autosomal dominant vs. multifactoria] | MENTAL RETARDATION. AUTOSOMAL RECESSIVE 51; MRT51 [AR]
HNRNPA1	AMYOTROPHIC LATERAL SCLEROSIS 20; ALS20 [AD] | INCLUSION BODY MYOPATHY WITH EARLY-ONSET PAGET DISEASE WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA 3; IBMPFD3 [AD]
HNRNPDL	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL DOMINANT 3; LGMDD3 [AD]
HNRNPH2	MENTAL RETARDATION. X-LINKED. SYNDROMIC. BAIN TYPE; MRXSB [XLD]
HNRNPK	AU-KLINE SYNDROME; AUKS [AD]
HNRNPU	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 54; EIEE54 [AD]
HOGA1	HYPEROXALURIA. PRIMARY. TYPE III; HP3 [AR]
HOXA1	Athabaskan brainstem dysgenesis syndrome 601536 (3) | Bosley-Salih-Alorainy syndrome 601536 (3)
HOXA11	RADIOULNAR SYNOSTOSIS WITH AMEGAKARYOCYTIC THROMBOCYTOPENIA 1; RUSAT1 [AD]
HOXA13	HAND-FOOT-GENITAL SYNDROME; HFG [AD] | PREAXIAL DEFICIENCY. POSTAXIAL POLYDACTYLY. AND HYPOSPADIAS [AD]
HOXA2	MICROTIA. HEARING IMPAIRMENT. AND CLEFT PALATE [AD]
HOXB1	FACIAL PARESIS. HEREDITARY CONGENITAL. 3; HCFP3 [AR]
HOXC13	ECTODERMAL DYSPLASIA 9. HAIR/NAIL TYPE; ECTD9 [AR]
HOXD10	VERTICAL TALUS. CONGENITAL; CVT [AD]
HOXD13	BRACHYDACTYLY. TYPE E1; BDE1 [AD] | VATER/VACTERL ASSOCIATION [Isolated cases] | BRACHYDACTYLY. TYPE D; BDD [AD] | SYNPOLYDACTYLY 1; SPD1 [AD] | SYNDACTYLY. TYPE V; SDTY5 [AD]
HPCA	DYSTONIA 2. TORSION. AUTOSOMAL RECESSIVE; DYT2 [AR]
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]
HPS4	HERMANSKY-PUDLAK SYNDROME 4; HPS4 [AR]
HPS5	HERMANSKY-PUDLAK SYNDROME 5; HPS5 [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)
HRAS	BLADDER CANCER [Somatic mutation] | MELANOCYTIC NEVUS SYNDROME. CONGENITAL; CMNS [Somatic mutation] | THYROID CANCER. NONMEDULLARY. 2; NMTC2 [AD] | SCHIMMELPENNING-FEUERSTEIN-MIMS SYNDROME; SFM [Somatic mosaicism] | NEVUS. EPIDERMAL [Somatic mutation] | COSTELLO SYNDROME; CSTLO [AD]
HRG	THROMBOPHILIA DUE TO HISTIDINE-RICH GLYCOPROTEIN DEFICIENCY; THPH11 [AD]
HSD11B1	CORTISONE REDUCTASE DEFICIENCY 2; CORTRD2 [AD]
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]
HSD3B2	ADRENAL HYPERPLASIA. CONGENITAL. DUE TO 3-BETA-HYDROXYSTEROID DEHYDROGENASE 2 DEFICIENCY [AR]
HSD3B7	BILE ACID SYNTHESIS DEFECT. CONGENITAL. 1; CBAS1 [AR]
HSF4	CATARACT 5. MULTIPLE TYPES; CTRCT5 [AD]
HSPA9	ANEMIA. SIDEROBLASTIC. 4; SIDBA4 [Autosomal dominant (another allele in trans may be required)] | EVEN-PLUS SYNDROME; EVPLS [AR]
HSPB1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2F; CMT2F [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE IIB; HMN2B [AD]
HSPB8	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2L; CMT2L [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE IIA; HMN2A [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]
HTR1A	PERIODIC FEVER. MENSTRUAL CYCLE-DEPENDENT [AD]
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]
HTRA2	3-METHYLGLUTACONIC ACIDURIA. TYPE VIII; MGCA8 [AR]
HTT	HUNTINGTON DISEASE; HD [AD] | LOPES-MACIEL-RODAN SYNDROME; LOMARS [AR]
HUWE1	MENTAL RETARDATION. X-LINKED. SYNDROMIC. TURNER TYPE; MRXST [X-linked]
HYDIN	CILIARY DYSKINESIA. PRIMARY. 5; CILD5 [AR]
HYLS1	HYDROLETHALUS SYNDROME 1; HLS1 [AR]
IARS1	GROWTH RETARDATION. IMPAIRED INTELLECTUAL DEVELOPMENT. HYPOTONIA. AND HEPATOPATHY; GRIDHH [AR]
IBA57	SPASTIC PARAPLEGIA 74. AUTOSOMAL RECESSIVE; SPG74 [AR] | MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 3; MMDS3 [AR]
ICOS	IMMUNODEFICIENCY. COMMON VARIABLE. 1; CVID1 [AR]
IDH2	D-2-hydroxyglutaric aciduria 2 613657 (3)
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]
IFIH1	AICARDI-GOUTIERES SYNDROME 7; AGS7 [AD] | SINGLETON-MERTEN SYNDROME 1; SGMRT1 [AD]
IFITM5	OSTEOGENESIS IMPERFECTA. TYPE V; OI5 [AD]

IFNA1	Interferon alpha deficiency (1)
IFNGR1	IMMUNODEFICIENCY 27A; IMD27A [AR] | HELICOBACTER PYLORI INFECTION. SUSCEPTIBILITY TO [Not determine] | IMMUNODEFICIENCY 27B; IMD27B [AD]
IFNGR2	IMMUNODEFICIENCY 28; IMD28 [AR]
IFT122	CRANIOECTODERMAL DYSPLASIA 1; CED1 [AR]
IFT140	SHORT-RIB THORACIC DYSPLASIA 9 WITH OR WITHOUT POLYDACTYLY; SRTD9 [AR] | RETINITIS PIGMENTOSA 80; RP80 [AR]
IFT172	RETINITIS PIGMENTOSA 71; RP71 [AR] | SHORT-RIB THORACIC DYSPLASIA 10 WITH OR WITHOUT POLYDACTYLY; SRTD10 [AR]
IFT43	CRANIOECTODERMAL DYSPLASIA 3; CED3 [AR] | RETINITIS PIGMENTOSA 81; RP81 [AR] | SHORT-RIB THORACIC DYSPLASIA 18 WITH POLYDACTYLY; SRTD18 [AR]
IFT52	SHORT-RIB THORACIC DYSPLASIA 16 WITH OR WITHOUT POLYDACTYLY; SRTD16 [AR]
IFT80	SHORT-RIB THORACIC DYSPLASIA 2 WITH OR WITHOUT POLYDACTYLY; SRTD2 [AR]
IFT81	SHORT-RIB THORACIC DYSPLASIA 19 WITH OR WITHOUT POLYDACTYLY; SRTD19 [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]
IGF2R	HEPATOCELLULAR CARCINOMA [Somatic mutation]
IGFALS	Acid-labile subunit deficiency of 615961 (3)
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]
IGKC	IMMUNOGLOBULIN KAPPA LIGHT CHAIN DEFICIENCY; IGKCD [AR]
IGLL1	AGAMMAGLOBULINEMIA 2. AUTOSOMAL RECESSIVE; AGM2 [AR]
IGSF1	HYPOTHYROIDISM. CENTRAL. WITH TESTICULAR ENLARGEMENT; CHTE [XLR]
IHH	BRACHYDACTYLY. TYPE A1; BDA1 [AD] | ACROCAPITOFEMORAL DYSPLASIA; ACFD [AR]
IKBKB	IMMUNODEFICIENCY 15B; IMD15B [AR] | IMMUNODEFICIENCY 15A; IMD15A [AD]
IKBKG	INCONTINENTIA PIGMENTI; IP [XLD] | IMMUNODEFICIENCY 33; IMD33 [XLR]
IKZF1	IMMUNODEFICIENCY. COMMON VARIABLE. 13; CVID13 [AD]
IL10RA	INFLAMMATORY BOWEL DISEASE 28. AUTOSOMAL RECESSIVE; IBD28 [AR]
IL10RB	INFLAMMATORY BOWEL DISEASE 25. AUTOSOMAL RECESSIVE; IBD25 [AR]
IL11RA	CRANIOSYNOSTOSIS AND DENTAL ANOMALIES; CRSDA [AR]
IL12B	IMMUNODEFICIENCY 29; IMD29 [AR]
IL12RB1	IMMUNODEFICIENCY 30; IMD30 [AR]
IL17RA	IMMUNODEFICIENCY 51; IMD51 [AR]
IL17RC	CANDIDIASIS. FAMILIAL. 9; CANDF9 [AR]
IL17RD	HYPOGONADOTROPIC HYPOGONADISM 18 WITH OR WITHOUT ANOSMIA; HH18 [Autosomal dominant (see MISCELLANEOUS below)]
IL18BP	HEPATITIS. FULMINANT VIRAL. SUSCEPTIBILITY TO; FVH [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]
IL21R	IgE RESPONSIVENESS. ATOPIC; IGER [AD] | IMMUNODEFICIENCY 56; IMD56 [AR]
IL2RA	DIABETES MELLITUS. INSULIN-DEPENDENT. 10; IDDM10 [Autosomal locus and allelic heterogeneit] | IMMUNODEFICIENCY 41 WITH LYMPHOPROLIFERATION AND AUTOIMMUNITY; IMD41 [AR]
IL2RB	IMMUNODEFICIENCY 63 WITH LYMPHOPROLIFERATION AND AUTOIMMUNITY; IMD63 [AR]
IL2RG	SEVERE COMBINED IMMUNODEFICIENCY. X-LINKED; SCIDX1 [XLR] | COMBINED IMMUNODEFICIENCY. X-LINKED; CIDX [XLR]

IL36RN	PSORIASIS 14. PUSTULAR; PSORS14 [AR]
IL6ST	HYPER-IgE RECURRENT INFECTION SYNDROME 4. AUTOSOMAL RECESSIVE; HIES4 [AR]
IL7	EPIDERMODYSPLASIA VERRUCIFORMIS. SUSCEPTIBILITY TO. 5; EV5 [AR]
IL7R	SEVERE COMBINED IMMUNODEFICIENCY. AUTOSOMAL RECESSIVE. T CELL-NEGATIVE. B CELL-POSITIVE. NK CELL-POSITIVE [AR]
ILDR1	DEAFNESS. AUTOSOMAL RECESSIVE 42; DFNB42 [AR]
IMPA1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 59; MRT59 [AR]
IMPDH1	RETINITIS PIGMENTOSA 10; RP10 [AD] | LEBER CONGENITAL AMAUROSIS 11; LCA11 [AD]
IMPG1	MACULAR DYSTROPHY. VITELLIFORM. 4; VMD4 [AD]
IMPG2	MACULAR DYSTROPHY. VITELLIFORM. 5; VMD5 [AD] | RETINITIS PIGMENTOSA 56; RP56 [AR]
INF2	CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE E; CMTDIE [AD]
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]
INPP5K	MUSCULAR DYSTROPHY. CONGENITAL. WITH CATARACTS AND INTELLECTUAL DISABILITY; MDCCAID [AR]
INPPL1	OPSISMODYSPLASIA; OPSMD [AR]
INS	HYPERPROINSULINEMIA [AD] | MATURITY-ONSET DIABETES OF THE YOUNG. TYPE 10; MODY10 [AD] | DIABETES MELLITUS. INSULIN-DEPENDENT. 2 [AD] | DIABETES MELLITUS. PERMANENT NEONATAL; PNDM [AD]
INSL3	CRYPTORCHIDISM. UNILATERAL OR BILATERAL [AD]
INSR	DONOHUE SYNDROME [AR] | PINEAL HYPERPLASIA. INSULIN-RESISTANT DIABETES MELLITUS. AND SOMATIC ABNORMALITIES [AR] | HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 5; HHF5 [AD]
INTS1	NEURODEVELOPMENTAL DISORDER WITH CATARACTS. POOR GROWTH. AND DYSMORPHIC FACIES; NDCAGF [AR]
INVS	NEPHRONOPHTHISIS 2; NPHP2 [AR]
IQCB1	SENIOR-LOKEN SYNDROME 5; SLSN5 [AR]
IQSEC2	MENTAL RETARDATION. X-LINKED 1; MRX1 [XLD]
IRAK4	IRAK4 deficiency 607676 (3) | Invasive pneumococcal disease recurrent isolated 1 610799 (3)
IREB2	NEURODEGENERATION. EARLY-ONSET. WITH CHOREOATHETOID MOVEMENTS AND MICROCYTIC ANEMIA; NDCAMA [AR]
IRF1	LUNG CANCER [Somatic mutation]
IRF2BPL	NEURODEVELOPMENTAL DISORDER WITH REGRESSION. ABNORMAL MOVEMENTS. LOSS OF SPEECH. AND SEIZURES; NEDAMSS [AD]
IRF6	OROFACIAL CLEFT 6. SUSCEPTIBILITY TO; OFC6 [AD] | POPLITEAL PTERYGIUM SYNDROME; PPS [AD] | VAN DER WOUDE SYNDROME 1; VWS1 [AD]
IRF8	IMMUNODEFICIENCY 32A; IMD32A [AD] | IMMUNODEFICIENCY 32B; IMD32B [AR]
IRF9	IMMUNODEFICIENCY 65. SUSCEPTIBILITY TO VIRAL INFECTIONS; IMD65 [AR]
IRS4	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 9; CHNG9 [XLR]
IRX5	HAMAMY SYNDROME; HMMS [AR]
ISCA1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 5; MMDS5 [AR]
ISCA2	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 4; MMDS4 [AR]
ISCU	MYOPATHY WITH LACTIC ACIDOSIS. HEREDITARY; HML [AR]
ISG15	IMMUNODEFICIENCY 38 WITH BASAL GANGLIA CALCIFICATION; IMD38 [AR]
ITCH	AUTOIMMUNE DISEASE. MULTISYSTEM. WITH FACIAL DYSMORPHISM; ADMFD [AR]
ITGA2B	BLEEDING DISORDER. PLATELET-TYPE. 16; BDPLT16 [AD] | GLANZMANN THROMBASTHENIA; GT [AR]
ITGA3	INTERSTITIAL LUNG DISEASE. NEPHROTIC SYNDROME. AND EPIDERMOLYSIS BULLOSA. CONGENITAL; ILNEB [AR]
ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA [AR]
ITGA7	MUSCULAR DYSTROPHY. CONGENITAL. DUE TO INTEGRIN ALPHA-7 DEFICIENCY [AR]
ITGA8	RENAL HYPODYSPLASIA/APLASIA 1; RHDA1 [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]
ITGB6	AMELOGENESIS IMPERFECTA. TYPE IH; AI1H [AR]
ITK	LYMPHOPROLIFERATIVE SYNDROME 1; LPFS1 [AR]
ITM2B	CEREBRAL AMYLOID ANGIOPATHY. ITM2B-RELATED. 2 [AD] | CEREBRAL AMYLOID ANGIOPATHY. ITM2B-RELATED. 1 [AD] | RETINAL DYSTROPHY WITH INNER RETINAL DYSFUNCTION AND GANGLION CELL ABNORMALITIES; RDGCA [AD]
ITPA	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 35; EIEE35 [AR]
ITPR1	SPINOCEREBELLAR ATAXIA 15; SCA15 [AD] | SPINOCEREBELLAR ATAXIA 29; SCA29 [AD] | GILLESPIE SYNDROME; GLSP [AD]
IVD	ISOVALERIC ACIDEMIA; IVA [AR]
IYD	THYROID DYSHORMONOGENESIS 4; TDH4 [AR]
JAG1	ALAGILLE SYNDROME 1; ALGS1 [AD] | TETRALOGY OF FALLOT; TOF [AD]
JAGN1	NEUTROPENIA. SEVERE CONGENITAL. 6. AUTOSOMAL RECESSIVE; SCN6 [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]
JPH2	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 17; CMH17 [AD]
JPH3	HUNTINGTON DISEASE-LIKE 2; HDL2 [AD]
JUP	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 12; ARVD12 [AD] | NAXOS DISEASE; NXD [AR]
KANK1	Cerebral palsy spastic quadriplegic 2 612900 (3)
KANK2	PALMOPLANTAR KERATODERMA AND WOOLLY HAIR; PPKWH [AR] | NEPHROTIC SYNDROME. TYPE 16; NPHS16 [AR]
KANSL1	KOOLEN-DE VRIES SYNDROME; KDVS [AD]
KARS1	DEAFNESS. AUTOSOMAL RECESSIVE 89; DFNB89 [AR] | CHARCOT-MARIE-TOOTH DISEASE. RECESSIVE INTERMEDIATE B; CMTRIB [AR]
KAT6A	ARBOLEDA-THAM SYNDROME; ARTHS [AD]
KAT6B	OHDO SYNDROME. SBBYS VARIANT; SBBYSS [AD] | GENITOPATELLAR SYNDROME; GTPTS [AD]
KATNB1	LISSENCEPHALY 6 WITH MICROCEPHALY; LIS6 [AR]
KBTBD13	NEMALINE MYOPATHY 6; NEM6 [AD]
KCNA1	EPISODIC ATAXIA. TYPE 1; EA1 [AD]
KCNA2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 32; EIEE32 [AD]
KCNA4	MICROCEPHALY. CATARACTS. IMPAIRED INTELLECTUAL DEVELOPMENT. AND DYSTONIA WITH ABNORMAL STRIATUM; MCIDDS [AR]
KCNA5	ATRIAL FIBRILLATION. FAMILIAL. 7; ATFB7 [AD]
KCNB1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 26; EIEE26 [AD]
KCNC1	EPILEPSY. PROGRESSIVE MYOCLONIC 7; EPM7 [AD]
KCNC3	SPINOCEREBELLAR ATAXIA 13; SCA13 [AD]
KCND3	BRUGADA SYNDROME 9; BRGDA9 [AD] | SPINOCEREBELLAR ATAXIA 19; SCA19 [AD]
KCNE1	JERVELL AND LANGE-NIELSEN SYNDROME 2; JLNS2 [AR] | LONG QT SYNDROME 5; LQT5 [AD]
KCNE2	LONG QT SYNDROME 6; LQT6 [AD]
KCNH1	TEMPLE-BARAITSER SYNDROME; TMBTS [AD] | ZIMMERMANN-LABAND SYNDROME 1; ZLS1 [AD]
KCNH2	LONG QT SYNDROME 2; LQT2 [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]
KCNJ2	ATRIAL FIBRILLATION. FAMILIAL. 9; ATFB9 [AD] | ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS [AD]
KCNJ5	HYPERALDOSTERONISM. FAMILIAL. TYPE III; HALD3 [AD] | LONG QT SYNDROME 13; LQT13 [AD]
KCNJ6	KEPPEN-LUBINSKY SYNDROME; KPLBS [AD]
KCNK3	PULMONARY HYPERTENSION. PRIMARY. 4; PPH4 [AD]
KCNK4	FACIAL DYSMORPHISM. HYPERTRICHOSIS. EPILEPSY. INTELLECTUAL/DEVELOPMENTAL DELAY. AND GINGIVAL OVERGROWTH SYNDROME; FHEIG [AD]
KCNK9	Birk-Barel mental retardation dysmorphism syndrome 612292 (3)
KCNMA1	LIANG-WANG SYNDROME; LIWAS [AD] | EPILEPSY. IDIOPATHIC GENERALIZED. SUSCEPTIBILITY TO. 16; EIG16 [AD] | PAROXYSMAL NONKINESIGENIC DYSKINESIA. 3. WITH OR WITHOUT GENERALIZED EPILEPSY; PNKD3 [AD] | CEREBELLAR ATROPHY. DEVELOPMENTAL DELAY. AND SEIZURES; CADEDS [AR]
KCNN3	ZIMMERMANN-LABAND SYNDROME 3; ZLS3 [AD]
KCNN4	DEHYDRATED HEREDITARY STOMATOCYTOSIS 2; DHS2 [AD]
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]
KCNQ1OT1	BECKWITH-WIEDEMANN SYNDROME; BWS [AD]
KCNQ2	SEIZURES. BENIGN FAMILIAL NEONATAL. 1; BFNS1 [AD] | FEBRILE SEIZURES. FAMILIAL. 1; FEB1 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 7; EIEE7 [AD]
KCNQ3	SEIZURES. BENIGN FAMILIAL NEONATAL. 2; BFNS2 [AD]
KCNQ4	DEAFNESS. AUTOSOMAL DOMINANT 2A; DFNA2A [AD]
KCNQ5	MENTAL RETARDATION. AUTOSOMAL DOMINANT 46; MRD46 [AD]
KCNT1	EPILEPSY. NOCTURNAL FRONTAL LOBE. 5; ENFL5 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 14; EIEE14 [AD]
KCNV2	RETINAL CONE DYSTROPHY 3B; RCD3B [AR]
KCTD1	SCALP-EAR-NIPPLE SYNDROME; SENS [AD]
KCTD17	DYSTONIA 26. MYOCLONIC; DYT26 [AD]
KCTD7	EPILEPSY. PROGRESSIVE MYOCLONIC. 3. WITH OR WITHOUT INTRACELLULAR INCLUSIONS; EPM3 [AR]
KDM1A	CLEFT PALATE. PSYCHOMOTOR RETARDATION. AND DISTINCTIVE FACIAL FEATURES; CPRF [AD]
KDM5B	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 65; MRT65 [AR]
KDM5C	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CLAES-JENSEN TYPE; MRXSCJ [XLR]
KDM6A	KABUKI SYNDROME 2; KABUK2 [XLD]
KDM6B	NEURODEVELOPMENTAL DISORDER WITH COARSE FACIES AND MILD DISTAL SKELETAL ABNORMALITIES; NEDCFSA [AD]
KDR	HEMANGIOMA. CAPILLARY INFANTILE [AD]
KDSR	ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 4; EKVP4 [AR]
KERA	CORNEA PLANA 2. AUTOSOMAL RECESSIVE; CNA2 [AR]
KHDC3L	HYDATIDIFORM MOLE. RECURRENT. 2; HYDM2 [AR]
KIAA0586	SHORT-RIB THORACIC DYSPLASIA 14 WITH POLYDACTYLY; SRTD14 [AR] | JOUBERT SYNDROME 23; JBTS23 [AR]
KIAA0825	POLYDACTYLY. POSTAXIAL. TYPE A10; PAPA10 [AR]
KIAA1109	ALKURAYA-KUCINSKAS SYNDROME; ALKKUCS [AR]
KIAA1549	RETINITIS PIGMENTOSA 86; RP86 [AR]
KIDINS220	SPASTIC PARAPLEGIA. INTELLECTUAL DISABILITY. NYSTAGMUS. AND OBESITY; SINO [AD]
KIF11	MICROCEPHALY WITH OR WITHOUT CHORIORETINOPATHY. LYMPHEDEMA. OR MENTAL RETARDATION; MCLMR [AD]
KIF14	MICROCEPHALY 20. PRIMARY. AUTOSOMAL RECESSIVE; MCPH20 [AR] | MECKEL SYNDROME 12; MKS12 [AR]
KIF1A	NESCAV SYNDROME; NESCAVS [AD] | NEUROPATHY. HEREDITARY SENSORY. TYPE IIC; HSN2C [AR] | SPASTIC PARAPLEGIA 30. AUTOSOMAL RECESSIVE; SPG30 [AR]
KIF1B	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2A1; CMT2A1 [AD] | PHEOCHROMOCYTOMA [AD] | NEUROBLASTOMA. SUSCEPTIBILITY TO. 1; NBLST1 [AD]
KIF1C	SPASTIC ATAXIA 2. AUTOSOMAL RECESSIVE; SPAX2 [AR]

KIF21A	FIBROSIS OF EXTRAOCULAR MUSCLES. CONGENITAL. 1; CFEOM1 [AD]
KIF22	SPONDYLOEPIMETAPHYSEAL DYSPLASIA WITH JOINT LAXITY. TYPE 2; SEMDJL2 [AD]
KIF2A	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 3; CDCBM3 [AD]
KIF5A	SPASTIC PARAPLEGIA 10. AUTOSOMAL DOMINANT; SPG10 [AD] | MYOCLONUS. INTRACTABLE. NEONATAL; NEIMY [AD] | AMYOTROPHIC LATERAL SCLEROSIS. SUSCEPTIBILITY TO. 25; ALS25 [AD]
KIF5C	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 2; CDCBM2 [AD]
KIF7	AL-GAZALI-BAKALINOVA SYNDROME; AGBK [AR] | ACROCALLOSAL SYNDROME; ACLS [AR] | HYDROLETHALUS SYNDROME 2; HLS2 [AR]
KIFBP	GOLDBERG-SHPRINTZEN SYNDROME; GOSHS [AR]
KISS1R	HYPOGONADOTROPIC HYPOGONADISM 8 WITH OR WITHOUT ANOSMIA; HH8 [AR] | PRECOCIOUS PUBERTY. CENTRAL. 1; CPPB1 [AD]
KIT	GASTROINTESTINAL STROMAL TUMOR; GIST [AD] | PIEBALD TRAIT; PBT [AD] | MASTOCYTOSIS. CUTANEOUS; MASTC [AD] | TESTICULAR GERM CELL TUMOR; TGCT [Somatic mutation] | LEUKEMIA. ACUTE MYELOID; AML [AD]
KITLG	HYPERPIGMENTATION WITH OR WITHOUT HYPOPIGMENTATION. FAMILIAL PROGRESSIVE; FPHH [AD] | DEAFNESS. AUTOSOMAL DOMINANT 69; DFNA69 [AD]
KIZ	RETINITIS PIGMENTOSA 69; RP69 [AR]
KLC2	SPASTIC PARAPLEGIA. OPTIC ATROPHY. AND NEUROPATHY; SPOAN [AR]
KLF1	ANEMIA. CONGENITAL DYSERYTHROPOIETIC. TYPE IV; CDAN4 [AD]
KLF11	Maturity-onset diabetes of the young type VII 610508 (3)
KLF6	PROSTATE CANCER [Somatic mutation]
KLHL10	SPERMATOGENIC FAILURE 11; SPGF11 [AD]
KLHL15	MENTAL RETARDATION. X-LINKED 103; MRX103 [XLR]
KLHL24	EPIDERMOLYSIS BULLOSA SIMPLEX. GENERALIZED. WITH SCARRING AND HAIR LOSS; EBSSH [AD]
KLHL3	PSEUDOHYPOALDOSTERONISM. TYPE IID; PHA2D [AD]
KLHL40	NEMALINE MYOPATHY 8; NEM8 [AR]
KLHL41	NEMALINE MYOPATHY 9; NEM9 [AR]
KLHL7	RETINITIS PIGMENTOSA 42; RP42 [AD] | PERCHING SYNDROME; PERCHING [AR]
KLK4	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA1; AI2A1 [AR]
KLKB1	PREKALLIKREIN DEFICIENCY [AR]
KLLN	Cowden syndrome 4 615107 (3)
KMT2A	WIEDEMANN-STEINER SYNDROME; WDSTS [AD]
KMT2B	DYSTONIA 28. CHILDHOOD-ONSET; DYT28 [AD]
KMT2C	KLEEFSTRA SYNDROME 2; KLEFS2 [AD]
KMT2D	KABUKI SYNDROME 1; KABUK1 [AD]
KMT2E	O'DONNELL-LURIA-RODAN SYNDROME; ODLURO [AD]
KMT5B	MENTAL RETARDATION. AUTOSOMAL DOMINANT 51; MRD51 [AD]
KNL1	MICROCEPHALY 4. PRIMARY. AUTOSOMAL RECESSIVE; MCPH4 [AR]
KPTN	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 41; MRT41 [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]
KREMEN1	ECTODERMAL DYSPLASIA 13. HAIR/TOOTH TYPE; ECTD13 [AR]
KRIT1	CEREBRAL CAVERNOUS MALFORMATIONS; CCM [AD]
KRT1	PALMOPLANTAR KERATODERMA. NONEPIDERMOLYTIC; NEPPK [AD] | ICHTHYOSIS. CYCLIC. WITH EPIDERMOLYTIC HYPERKERATOSIS [AD] | ICHTHYOSIS HYSTRIX. CURTH-MACKLIN TYPE; IHCM [AD] | PALMOPLANTAR KERATODERMA. EPIDERMOLYTIC; EPPK [AD] | EPIDERMOLYTIC HYPERKERATOSIS; EHK [AD]
KRT10	EPIDERMOLYTIC HYPERKERATOSIS; EHK [AD] | ERYTHRODERMA. ICHTHYOSIFORM. CONGENITAL RETICULAR; CRIE [AD] | ICHTHYOSIS. CYCLIC. WITH EPIDERMOLYTIC HYPERKERATOSIS [AD]

KRT12	CORNEAL DYSTROPHY. MEESMANN. 1; MECD1 [AD]
KRT13	WHITE SPONGE NEVUS 2; WSN2 [AD]
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]
KRT16	PACHYONYCHIA CONGENITA 1; PC1 [AD] | PALMOPLANTAR KERATODERMA. NONEPIDERMOLYTIC. FOCAL 1; FNEPPK1 [AD]
KRT17	STEATOCYSTOMA MULTIPLEX [AD] | PACHYONYCHIA CONGENITA 2; PC2 [AD]
KRT18	CIRRHOSIS. FAMILIAL [Autosomal recessive cases, probably heterogeneou]
KRT2	ICHTHYOSIS BULLOSA OF SIEMENS; IBS [AD]
KRT25	WOOLLY HAIR. AUTOSOMAL RECESSIVE 3; ARWH3 [AR]
KRT3	Meesmann corneal dystrophy 2 618767 (3)
KRT4	WHITE SPONGE NEVUS 1; WSN1 [AD]
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]
KRT6A	PACHYONYCHIA CONGENITA 1; PC1 [AD]
KRT6B	PACHYONYCHIA CONGENITA 2; PC2 [AD]
KRT6C	PALMOPLANTAR KERATODERMA. NONEPIDERMOLYTIC. FOCAL OR DIFFUSE; PPKNEFD [AD]
KRT74	WOOLLY HAIR. AUTOSOMAL DOMINANT; ADWH [AD] | ECTODERMAL DYSPLASIA 7. HAIR/NAIL TYPE; ECTD7 [AR] | HYPOTRICHOSIS 3; HYPT3 [AD]
KRT8	CIRRHOSIS. FAMILIAL [Autosomal recessive cases, probably heterogeneou]
KRT81	MONILETHRIX; MNLIX [AD]
KRT83	ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 5; EKVP5 [AR] | MONILETHRIX; MNLIX [AD]
KRT85	ECTODERMAL DYSPLASIA 4. HAIR/NAIL TYPE; ECTD4 [AR]
KRT86	MONILETHRIX; MNLIX [AD]
KRT9	PALMOPLANTAR KERATODERMA. EPIDERMOLYTIC; EPPK [AD]
KY	Myopathy myofibrillar 7 617114 (3)
KYNU	VERTEBRAL. CARDIAC. RENAL. AND LIMB DEFECTS SYNDROME 2; VCRL2 [AR] | HYDROXYKYNURENINURIA [AR]
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]
LAGE3	GALLOWAY-MOWAT SYNDROME 2. X-LINKED; GAMOS2 [XLR]
LAMA1	PORETTI-BOLTSHAUSER SYNDROME; PTBHS [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]
LAMA4	CARDIOMYOPATHY. DILATED. 1JJ; CMD1JJ [AD]
LAMB1	LISSENCEPHALY 5; LIS5 [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]
LAMP2	DANON DISEASE [XLD]
LAMTOR2	IMMUNODEFICIENCY DUE TO DEFECT IN MAPBP-INTERACTING PROTEIN [AR]
LARGE1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION). TYPE B. 6; MDDGB6 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 6; MDDGA6 [AR]
LARP7	ALAZAMI SYNDROME; ALAZS [AR]

LARS2	PERRAULT SYNDROME 4; PRLTS4 [AR] | HYDROPS. LACTIC ACIDOSIS. AND SIDEROBLASTIC ANEMIA; HLASA [AR]
LAS1L	WILSON-TURNER X-LINKED MENTAL RETARDATION SYNDROME; WTS [XLR]
LAT	IMMUNODEFICIENCY 52; IMD52 [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]
LDB3	CARDIOMYOPATHY. DILATED. 1C. WITH OR WITHOUT LEFT VENTRICULAR NONCOMPACTION; CMD1C [AD] | MYOPATHY. MYOFIBRILLAR. 4; MFM4 [AD]
LDHA	GLYCOGEN STORAGE DISEASE XI; GSD11 [AR]
LDHD	D-LACTIC ACIDURIA; DLACD [AR]
LDLR	HYPERCHOLESTEROLEMIA. FAMILIAL. 1; FHCL1 [AD]
LDLRAP1	HYPERCHOLESTEROLEMIA. FAMILIAL. 4; FHCL4 [AR]
LEF1	Sebaceous tumors somatic (3)
LEMD2	CATARACT 46. JUVENILE-ONSET. WITH OR WITHOUT ARRHYTHMIC CARDIOMYOPATHY; CTRCT46 [AR]
LEMD3	BUSCHKE-OLLENDORFF SYNDROME; BOS [AD]
LEP	LEPTIN DEFICIENCY OR DYSFUNCTION; LEPD [AR]
LEPR	LEPTIN RECEPTOR DEFICIENCY; LEPRD [AR]
LFNG	SPONDYLOCOSTAL DYSOSTOSIS 3. AUTOSOMAL RECESSIVE; SCDO3 [AR]
LGI1	EPILEPSY. FAMILIAL TEMPORAL LOBE. 1; ETL1 [AD]
LGI4	ARTHROGRYPOSIS MULTIPLEX CONGENITA. NEUROGENIC. WITH MYELIN DEFECT; AMCNMY [AR]
LHB	HYPOGONADOTROPIC HYPOGONADISM 23 WITHOUT ANOSMIA; HH23 [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]
LHX4	PITUITARY HORMONE DEFICIENCY. COMBINED. 4; CPHD4 [AD]
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]
LINGO1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 64; MRT64 [AR]
LINS1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 27; MRT27 [AR]
LIPA	LYSOSOMAL ACID LIPASE DEFICIENCY [AR]
LIPC	HEPATIC LIPASE DEFICIENCY [AR] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
LIPE	LIPODYSTROPHY. FAMILIAL PARTIAL. TYPE 6; FPLD6 [AR]
LIPH	HYPOTRICHOSIS 7; HYPT7 [AR]
LIPN	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 8; ARCI8 [AR]
LIPT1	LIPOYLTRANSFERASE 1 DEFICIENCY; LIPT1D [AR]
LIPT2	ENCEPHALOPATHY. NEONATAL SEVERE. WITH LACTIC ACIDOSIS AND BRAIN ABNORMALITIES; NELABA [AR]
LITAF	CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1C; CMT1C [AD]
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]
LMNB1	LEUKODYSTROPHY. DEMYELINATING. ADULT-ONSET. AUTOSOMAL DOMINANT; ADLD [AD]
LMO1	Leukemia T-cell acute lymphoblastic (2)
LMO2	Leukemia acute T-cell (2)
LMOD3	NEMALINE MYOPATHY 10; NEM10 [AR]
LMX1B	NAIL-PATELLA SYNDROME; NPS [AD]
LNPK	NEURODEVELOPMENTAL DISORDER WITH EPILEPSY AND HYPOPLASIA OF THE CORPUS CALLOSUM; NEDEHCC [AR]
LONP1	CODAS SYNDROME [AR]
LORICRIN	VOHWINKEL SYNDROME. VARIANT FORM [AD]
LOX	AORTIC ANEURYSM. FAMILIAL THORACIC 10; AAT10 [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]
LPP	LEUKEMIA. ACUTE MYELOID; AML [AD]
LRAT	LEBER CONGENITAL AMAUROSIS 14; LCA14 [AR]
LRBA	IMMUNODEFICIENCY. COMMON VARIABLE. 8. WITH AUTOIMMUNITY; CVID8 [AR]
LRIG2	UROFACIAL SYNDROME 2; UFS2 [AR]
LRIT3	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1F; CSNB1F [AR]
LRMDA	ALBINISM. OCULOCUTANEOUS. TYPE VII; OCA7 [AR]
LRP12	OCULOPHARYNGODISTAL MYOPATHY 1; OPDM1 [AD]
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]
LRP6	TOOTH AGENESIS. SELECTIVE. 7; STHAG7 [AD] | CORONARY ARTERY DISEASE. AUTOSOMAL DOMINANT 2; ADCAD2 [AD]
LRPAP1	MYOPIA 23. AUTOSOMAL RECESSIVE; MYP23 [AR]
LRPPRC	LEIGH SYNDROME. FRENCH CANADIAN TYPE; LSFC [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
LRRC56	CILIARY DYSKINESIA. PRIMARY. 39; CILD39 [AR]
LRRC6	CILIARY DYSKINESIA. PRIMARY. 19; CILD19 [AR]
LRSAM1	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2P; CMT2P [AD]
LRTOMT	DEAFNESS. AUTOSOMAL RECESSIVE 63; DFNB63 [AR]
LSS	HYPOTRICHOSIS 14; HYPT14 [AR] | CATARACT 44; CTRCT44 [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]

LTC4S	LEUKOTRIENE C4 SYNTHASE DEFICIENCY [AR]
LYL1	Leukemia T-cell acute lymphoblastoid (2)
LYRM7	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 8; MC3DN8 [AR]
LYST	CHEDIAK-HIGASHI SYNDROME; CHS [AR]
LYZ	AMYLOIDOSIS. FAMILIAL VISCERAL [AD]
LZTFL1	BARDET-BIEDL SYNDROME 17; BBS17 [AR]
LZTR1	NOONAN SYNDROME 2; NS2 [AR] | NOONAN SYNDROME 10; NS10 [AD] | SCHWANNOMATOSIS 2; SWNTS2 [AD]
LZTS1	ESOPHAGEAL CANCER [Somatic mutation]
MAB21L1	CEREBELLAR. OCULAR. CRANIOFACIAL. AND GENITAL SYNDROME; COFG [AR]
MAB21L2	MICROPHTHALMIA/COLOBOMA AND SKELETAL DYSPLASIA SYNDROME; MCSKS [AD]
MACF1	LISSENCEPHALY 9 WITH COMPLEX BRAINSTEM MALFORMATION; LIS9 [AD]
MAD1L1	PROSTATE CANCER [Somatic mutation]
MAF	AYME-GRIPP SYNDROME; AYGRP [AD] | CATARACT 21. MULTIPLE TYPES; CTRCT21 [AD]
MAFA	INSULINOMATOSIS AND DIABETES MELLITUS; INSDM [AD]
MAFB	MULTICENTRIC CARPOTARSAL OSTEOLYSIS SYNDROME; MCTO [AD] | DUANE RETRACTION SYNDROME 3 WITH OR WITHOUT DEAFNESS; DURS3 [AD]
MAG	SPASTIC PARAPLEGIA 75. AUTOSOMAL RECESSIVE; SPG75 [AR]
MAGED2	BARTTER SYNDROME. TYPE 5. ANTENATAL. TRANSIENT; BARTS5 [XLR]
MAGEL2	SCHAAF-YANG SYNDROME; SHFYNG [AD]
MAGI2	NEPHROTIC SYNDROME. TYPE 15; NPHS15 [AR]
MAGT1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Icc; CDG1CC [XLR] | IMMUNODEFICIENCY. X-LINKED. WITH MAGNESIUM DEFECT. EPSTEIN-BARR VIRUS INFECTION. AND NEOPLASIA; XMEN [XLR]
MAK	RETINITIS PIGMENTOSA 62; RP62 [AR]
MALT1	IMMUNODEFICIENCY 12; IMD12 [AR]
MAML2	Mucoepidermoid salivary gland carcinoma (3)
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]
MAOA	BRUNNER SYNDROME; BRNRS [XLR]
MAP2K1	CARDIOFACIOCUTANEOUS SYNDROME 3; CFC3 [AD]
MAP2K2	CARDIOFACIOCUTANEOUS SYNDROME 4; CFC4 [AD]
MAP3K1	46.XY SEX REVERSAL 6; SRXY6 [AD]
MAP3K20	MYOPATHY. CENTRONUCLEAR. 6. WITH FIBER-TYPE DISPROPORTION; CNM6 [AR] | SPLIT-FOOT MALFORMATION WITH MESOAXIAL POLYDACTYLY; SFMMP [AR]
MAP3K7	FRONTOMETAPHYSEAL DYSPLASIA 2; FMD2 [AD] | CARDIOSPONDYLOCARPOFACIAL SYNDROME; CSCF [AD]
MAP3K8	LUNG CANCER [Somatic mutation]
MAPK8IP3	NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT VARIABLE BRAIN ABNORMALITIES; NEDBA [AD]
MAPKBP1	NEPHRONOPHTHISIS 20; NPHP20 [AR]
MAPRE2	SKIN CREASES. CONGENITAL SYMMETRIC CIRCUMFERENTIAL. 2; CSCSC2 [AD]
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]
MARCHF6	EPILEPSY. FAMILIAL ADULT MYOCLONIC. 3; FAME3 [AD]
MARS1	INTERSTITIAL LUNG AND LIVER DISEASE; ILLD [AR] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2U; CMT2U [AD]
MARS2	SPASTIC ATAXIA 3. AUTOSOMAL RECESSIVE; SPAX3 [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 25; COXPD25 [AR]

MARVELD2	DEAFNESS. AUTOSOMAL RECESSIVE 49; DFNB49 [AR]
MASP1	3MC SYNDROME 1; 3MC1 [AR]
MASP2	MASP2 DEFICIENCY [AR]
MAST1	MEGA-CORPUS-CALLOSUM SYNDROME WITH CEREBELLAR HYPOPLASIA AND CORTICAL MALFORMATIONS; MCCCHCM [AD]
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]
MATR3	AMYOTROPHIC LATERAL SCLEROSIS 21; ALS21 [AD]
MBD5	MENTAL RETARDATION. AUTOSOMAL DOMINANT 1; MRD1 [AD]
MBOAT7	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 57; MRT57 [AR]
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]
MCC	COLORECTAL CANCER; CRC [Somatic mutation]
MCCC1	3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY; MCC1D [AR]
MCCC2	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY; MCC2D [AR]
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY [AR]
MCFD2	Factor V and factor VIII combined deficiency of 613625 (3)
MCM3AP	PERIPHERAL NEUROPATHY. AUTOSOMAL RECESSIVE. WITH OR WITHOUT IMPAIRED INTELLECTUAL DEVELOPMENT; PNRIID [AR]
MCM4	IMMUNODEFICIENCY 54; IMD54 [AR]
MCM6	LACTOSE INTOLERANCE. ADULT TYPE [AD]
MCM9	OVARIAN DYSGENESIS 4; ODG4 [AR]
MCOLN1	MUCOLIPIDOSIS IV; ML4 [AR]
MCPH1	MICROCEPHALY 1. PRIMARY. AUTOSOMAL RECESSIVE; MCPH1 [AR]
MDH2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 51; EIEE51 [AR]
MECOM	RADIOULNAR SYNOSTOSIS WITH AMEGAKARYOCYTIC THROMBOCYTOPENIA 2; RUSAT2 [AD]
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]
MECR	DYSTONIA. CHILDHOOD-ONSET. WITH OPTIC ATROPHY AND BASAL GANGLIA ABNORMALITIES; DYTOABG [AR]
MED12	OPITZ-KAVEGGIA SYNDROME; OKS [XLR] | INTELLECTUAL DEVELOPMENTAL DISORDER. X-LINKED. SYNDROMIC. LUJAN-FRYNS TYPE; MRXSLF [XLR] | OHDO SYNDROME. X-LINKED; OHDOX [XLR]
MED13L	TRANSPOSITION OF THE GREAT ARTERIES. DEXTRO-LOOPED 1; DTGA1 [AD] | MENTAL RETARDATION AND DISTINCTIVE FACIAL FEATURES WITH OR WITHOUT CARDIAC DEFECTS; MRFACD [AD]
MED17	MICROCEPHALY. POSTNATAL PROGRESSIVE. WITH SEIZURES AND BRAIN ATROPHY [AR]
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]
MEF2C	MENTAL RETARDATION. AUTOSOMAL DOMINANT 20; MRD20 [AD]
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]
MEI1	HYDATIDIFORM MOLE. RECURRENT. 3; HYDM3 [AR]
MEIS2	CLEFT PALATE. CARDIAC DEFECTS. AND MENTAL RETARDATION; CPCMR [AD]
MEN1	MULTIPLE ENDOCRINE NEOPLASIA. TYPE I; MEN1 [AD]
MEOX1	KLIPPEL-FEIL SYNDROME 2. AUTOSOMAL RECESSIVE; KFS2 [AR]

MERTK	RETINITIS PIGMENTOSA 38; RP38 [AR]
MESD	OSTEOGENESIS IMPERFECTA. TYPE XX; OI20 [AR]
MESP2	SPONDYLOCOSTAL DYSOSTOSIS 2. AUTOSOMAL RECESSIVE; SCDO2 [AR]
MET	OSTEOFIBROUS DYSPLASIA. SUSCEPTIBILITY TO; OSFD [AD] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | DEAFNESS. AUTOSOMAL RECESSIVE 97; DFNB97 [AR]
METTL23	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 44; MRT44 [AR]
METTL5	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 72; MRT72 [AR]
MFAP5	AORTIC ANEURYSM. FAMILIAL THORACIC 9; AAT9 [AD]
MFF	ENCEPHALOPATHY DUE TO DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION 2; EMPF2 [AR]
MFHAS1	Malignant fibrous histiocytoma (2)
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]
MFSD2A	MICROCEPHALY 15. PRIMARY. AUTOSOMAL RECESSIVE; MCPH15 [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]
MGME1	MITOCHONDRIAL DNA DEPLETION SYNDROME 11; MTDPS11 [AR]
MGP	KEUTEL SYNDROME; KTLS [AR]
MIB1	LEFT VENTRICULAR NONCOMPACTION 7; LVNC7 [AD]
MICOS13	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 37; COXPD37 [AR]
MICU1	MYOPATHY WITH EXTRAPYRAMIDAL SIGNS; MPXPS [AR]
MID1	OPITZ GBBB SYNDROME. TYPE I; GBBB1 [XLR]
MIP	CATARACT 15. MULTIPLE TYPES; CTRCT15 [AD]
MIPEP	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 31; COXPD31 [AR]
MIR140	SPONDYLOEPIPHYSEAL DYSPLASIA. NISHIMURA TYPE; SEDN [AD]
MIR17HG	FEINGOLD SYNDROME 2; FGLDS2 [AD]
MIR184	EDICT SYNDROME; EDICT [AD]
MIR96	DEAFNESS. AUTOSOMAL DOMINANT 50; DFNA50 [AD]
MITF	COLOBOMA. OSTEOPETROSIS. MICROPHTHALMIA. MACROCEPHALY. ALBINISM. AND DEAFNESS; COMMAD [AR] | WAARDENBURG SYNDROME. TYPE 2A; WS2A [AD] | TIETZ ALBINISM-DEAFNESS SYNDROME; TADS [AD]
MKKS	BARDET-BIEDL SYNDROME 6; BBS6 [AR] | MCKUSICK-KAUFMAN SYNDROME; MKKS [AR]
MKRN3	PRECOCIOUS PUBERTY. CENTRAL. 2; CPPB2 [AD]
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]
MLH3	COLORECTAL CANCER; CRC [Somatic mutation] | ENDOMETRIAL CANCER [Somatic mutation]
MLLT10	LEUKEMIA. ACUTE MYELOID; AML [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]
MME	SPINOCEREBELLAR ATAXIA 43; SCA43 [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2T; CMT2T [AR]

MMP1	EPIDERMOLYSIS BULLOSA DYSTROPHICA. AUTOSOMAL RECESSIVE; RDEB [AR]
MMP13	METAPHYSEAL DYSPLASIA. SPAHR TYPE; MDST [AR] | SPONDYLOEPIMETAPHYSEAL DYSPLASIA. MISSOURI TYPE [AD]
MMP19	CAVITARY OPTIC DISC ANOMALIES; CODA [AD]
MMP2	MULTICENTRIC OSTEOLYSIS. NODULOSIS. AND ARTHROPATHY; MONA [AR]
MMP20	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA2; AI2A2 [AR]
MMP21	HETEROTAXY. VISCERAL. 7. AUTOSOMAL; HTX7 [AR]
MMP9	Metaphyseal anadysplasia 2 613073 (3)
MMUT	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY [AR]
MN1	MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD] | CEBALID SYNDROME; CEBALID [AD]
MNX1	CURRARINO SYNDROME [AD]
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]
MORC2	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2Z; CMT2Z [AD]
MPC1	MITOCHONDRIAL PYRUVATE CARRIER DEFICIENCY; MPYCD [AR]
MPDU1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE If; CDG1F [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]
MPO	ALZHEIMER DISEASE; AD [AD] | MYELOPEROXIDASE DEFICIENCY; MPOD [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]
MPZL2	DEAFNESS. AUTOSOMAL RECESSIVE 111; DFNB111 [AR]
MRAP	GLUCOCORTICOID DEFICIENCY 2; GCCD2 [AR]
MRAP2	BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 18; BMIQ18 [AD]
MRAS	NOONAN SYNDROME 11; NS11 [AD]
MRE11	ATAXIA-TELANGIECTASIA-LIKE DISORDER 1; ATLD1 [AR]
MRPL3	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 9; COXPD9 [AR]
MRPS16	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2; COXPD2 [AR]
MRPS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 36; COXPD36 [AR]
MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5; COXPD5 [AR] | OVARIAN DYSGENESIS 7; ODG7 [AR]
MRPS34	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 32; COXPD32 [AR]
MRTFA	Megakaryoblastic leukemia acute (3)
MS4A1	IMMUNODEFICIENCY. COMMON VARIABLE. 5; CVID5 [AR]
MSH2	LYNCH SYNDROME I [AD] | MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR] | MUIR-TORRE SYNDROME; MRTES [AD]
MSH3	FAMILIAL ADENOMATOUS POLYPOSIS 4; FAP4 [AR] | ENDOMETRIAL CANCER [Somatic mutation]
MSH6	COLORECTAL CANCER. HEREDITARY NONPOLYPOSIS. TYPE 5; HNPCC5 [AD] | ENDOMETRIAL CANCER [Somatic mutation] | MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR]
MSL3	Basilicata-Akhtar syndrome 301032 (3)

MSMO1	MICROCEPHALY. CONGENITAL CATARACT. AND PSORIASIFORM DERMATITIS; MCCPD [AR]
MSN	IMMUNODEFICIENCY 50; IMD50 [XLR]
MSR1	Barrett esophagus/esophageal adenocarcinoma 614266 (3)
MSRB3	DEAFNESS. AUTOSOMAL RECESSIVE 74; DFNB74 [AR]
MSTN	Muscle hypertrophy 614160 (3)
MSTO1	MYOPATHY. MITOCHONDRIAL. AND ATAXIA; MMYAT [AR]
MSX1	WITKOP SYNDROME [AD] | OROFACIAL CLEFT 1; OFC1 [AD] | TOOTH AGENESIS. SELECTIVE. 1; STHAG1 [AD]
MSX2	PARIETAL FORAMINA 1; PFM1 [AD] | CRANIOSYNOSTOSIS 2; CRS2 [AD] | PARIETAL FORAMINA WITH CLEIDOCRANIAL DYSPLASIA; PFMCCD [AD]
MTAP	DIAPHYSEAL MEDULLARY STENOSIS WITH MALIGNANT FIBROUS HISTIOCYTOMA; DMSMFH [AD]
MTFMT	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 15; COXPD15 [AR] | LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 27; MC1DN27 [AR]
MTHFD1	NEURAL TUBE DEFECTS. FOLATE-SENSITIVE; NTDFS [AR] | COMBINED IMMUNODEFICIENCY AND MEGALOBLASTIC ANEMIA WITH OR WITHOUT HYPERHOMOCYSTEINEMIA; CIMAH [AR]
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]
MTHFS	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. EPILEPSY. AND HYPOMYELINATION; NEDMEHM [AR]
MTM1	MYOPATHY. CENTRONUCLEAR. X-LINKED; CNMX [XLR]
MTMR2	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4B1; CMT4B1 [AR]
MTO1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 10; COXPD10 [AR]
MTOR	SMITH-KINGSMORE SYNDROME; SKS [AD] | FOCAL CORTICAL DYSPLASIA. TYPE II; FCORD2 [Somatic mutation]
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]
MUC1	MEDULLARY CYSTIC KIDNEY DISEASE 1; MCKD1 [AD]
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]
MVD	POROKERATOSIS 7. MULTIPLE TYPES; POROK7 [AD]
MVK	HYPER-IgD SYNDROME; HIDS [AR] | MEVALONIC ACIDURIA; MEVA [AR] | POROKERATOSIS 3. MULTIPLE TYPES; POROK3 [AD]
MXI1	PROSTATE CANCER [Somatic mutation]
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]
MYC	BURKITT LYMPHOMA; BL [Isolated cases]
MYCN	FEINGOLD SYNDROME 1; FGLDS1 [AD]
MYD88	MACROGLOBULINEMIA. WALDENSTROM. SUSCEPTIBILITY TO. 1; WM1 [AD]
MYF5	OPHTHALMOPLEGIA. EXTERNAL. WITH RIB AND VERTEBRAL ANOMALIES; EORVA [AR]
MYH11	AORTIC ANEURYSM. FAMILIAL THORACIC 4; AAT4 [AD]
MYH14	PERIPHERAL NEUROPATHY. MYOPATHY. HOARSENESS. AND HEARING LOSS; PNMHH [AD] | DEAFNESS. AUTOSOMAL DOMINANT 4A; DFNA4A [AD]
MYH2	MYOPATHY. PROXIMAL. WITH OPHTHALMOPLEGIA; MYPOP [AD]
MYH3	ARTHROGRYPOSIS. DISTAL. TYPE 2A; DA2A [AD] | CONTRACTURES. PTERYGIA. AND SPONDYLOCARPOTARSAL FUSION SYNDROME 1A; CPSFS1A [AD] | ARTHROGRYPOSIS. DISTAL. TYPE 2B1; DA2B1 [AD] | ARTHROGRYPOSIS. DISTAL. TYPE 2B3; DA2B3 [AD] | CONTRACTURES. PTERYGIA. AND SPONDYLOCARPOTARSAL FUSION SYNDROME 1B; CPSFS1B [AR]
MYH6	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 14; CMH14 [AD]

MYH7	SCAPULOPERONEAL MYOPATHY. MYH7-RELATED; SPMM [AD] | CARDIOMYOPATHY. DILATED. 1S; CMD1S [AD] | MYOPATHY. DISTAL. 1; MPD1 [AD] | MYOPATHY. MYOSIN STORAGE. AUTOSOMAL RECESSIVE; MSMB [AR] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 1; CMH1 [AD] | MYOPATHY. MYOSIN STORAGE. AUTOSOMAL DOMINANT; MSMA [AD]
MYH8	ARTHROGRYPOSIS. DISTAL. TYPE 7; DA7 [AD]
MYH9	DEAFNESS. AUTOSOMAL DOMINANT 17; DFNA17 [AD] | MACROTHROMBOCYTOPENIA AND GRANULOCYTE INCLUSIONS WITH OR WITHOUT NEPHRITIS OR SENSORINEURAL HEARING LOSS; MATINS [AD]
MYL1	MYOPATHY. CONGENITAL. WITH FAST-TWITCH (TYPE II) FIBER ATROPHY; MYOFTA [AR]
MYL2	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 10; CMH10 [AD]
MYL3	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 8; CMH8 [AD]
MYLK	AORTIC ANEURYSM. FAMILIAL THORACIC 7; AAT7 [AD] | MEGACYSTIS-MICROCOLON-INTESTINAL HYPOPERISTALSIS SYNDROME; MMIHS [AR]
MYLK2	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 1; CMH1 [AD]
MYMK	CAREY-FINEMAN-ZITER SYNDROME; CFZS [AR]
MYO15A	DEAFNESS. AUTOSOMAL RECESSIVE 3; DFNB3 [AR]
MYO18B	KLIPPEL-FEIL SYNDROME 4. AUTOSOMAL RECESSIVE. WITH NEMALINE MYOPATHY AND FACIAL DYSMORPHISM; KFS4 [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]
MYO9A	MYASTHENIC SYNDROME. CONGENITAL. 24. PRESYNAPTIC; CMS24 [AR]
MYOC	GLAUCOMA 1. OPEN ANGLE. A; GLC1A [AD]
MYORG	BASAL GANGLIA CALCIFICATION. IDIOPATHIC. 7. AUTOSOMAL RECESSIVE; IBGC7 [AR]
MYOT	MYOPATHY. MYOFIBRILLAR. 3; MFM3 [AD] | MYOPATHY. SPHEROID BODY [AD]
MYOZ2	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 16; CMH16 [AD]
MYPN	CARDIOMYOPATHY. DILATED. 1KK; CMD1KK [AD] | NEMALINE MYOPATHY 11. AUTOSOMAL RECESSIVE; NEM11 [AR]
MYRF	ENCEPHALITIS/ENCEPHALOPATHY. MILD. WITH REVERSIBLE MYELIN VACUOLIZATION; MMERV [AD] | CARDIAC-UROGENITAL SYNDROME; CUGS [AD]
MYSM1	BONE MARROW FAILURE SYNDROME 4; BMFS4 [AR]
MYT1L	MENTAL RETARDATION. AUTOSOMAL DOMINANT 39; MRD39 [AD]
NAA10	MICROPHTHALMIA. SYNDROMIC 1; MCOPS1 [X-linked] | OGDEN SYNDROME; OGDNS [XLR]
NAA15	MENTAL RETARDATION. AUTOSOMAL DOMINANT 50; MRD50 [AD]
NACC1	NEURODEVELOPMENTAL DISORDER WITH EPILEPSY. CATARACTS. FEEDING DIFFICULTIES. AND DELAYED BRAIN MYELINATION; NECFM [AD]
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]
NANOS1	SPERMATOGENIC FAILURE 12; SPGF12 [AD]
NANS	SPONDYLOEPIMETAPHYSEAL DYSPLASIA. GENEVIEVE TYPE; SEMDG [AR]
NARS2	DEAFNESS. AUTOSOMAL RECESSIVE 94; DFNB94 [AR] | COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 24; COXPD24 [AR]
NAXD	ENCEPHALOPATHY. PROGRESSIVE. EARLY-ONSET. WITH BRAIN EDEMA AND/OR LEUKOENCEPHALOPATHY. 2; PEBEL2 [AR]
NAXE	ENCEPHALOPATHY. PROGRESSIVE. EARLY-ONSET. WITH BRAIN EDEMA AND/OR LEUKOENCEPHALOPATHY. 1; PEBEL1 [AR]
NBAS	INFANTILE LIVER FAILURE SYNDROME 2; ILFS2 [AR] | SHORT STATURE. OPTIC NERVE ATROPHY. AND PELGER-HUET ANOMALY; SOPH [AR]

NBEAL2	GRAY PLATELET SYNDROME; GPS [AR]
NBN	NIJMEGEN BREAKAGE SYNDROME; NBS [AR]
NCAPD3	MICROCEPHALY 22. PRIMARY. AUTOSOMAL RECESSIVE; MCPH22 [AR]
NCAPG2	KHAN-KHAN-KATSANIS SYNDROME; 3KS [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]
NCSTN	ACNE INVERSA. FAMILIAL. 1; ACNINV1 [AD]
NDE1	MICROHYDRANENCEPHALY; MHAC [AR] | LISSENCEPHALY 4; LIS4 [AR]
NDN	PRADER-WILLI SYNDROME; PWS [Autosomal dominant (loss of paternal allele)]
NDP	NORRIE DISEASE; ND [XLR] | EXUDATIVE VITREORETINOPATHY 2. X-LINKED; EVR2 [XLR]
NDRG1	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4D; CMT4D [AR]
NDST1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 46; MRT46 [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]
NDUFA6	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 33; MC1DN33 [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]
NDUFB11	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 30; MC1DN30 [X-linked] | LINEAR SKIN DEFECTS WITH MULTIPLE CONGENITAL ANOMALIES 3; LSDMCA3 [XLD]
NDUFB3	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 25; MC1DN25 [AR]
NDUFB8	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 32; MC1DN32 [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]
NECTIN1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME; CLPED1 [AR]
NECTIN4	ECTODERMAL DYSPLASIA-SYNDACTYLY SYNDROME 1; EDSS1 [AR]
NEDD4L	PERIVENTRICULAR NODULAR HETEROTOPIA 7; PVNH7 [AD]
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]

NEK9	NEVUS COMEDONICUS; NC [Somatic mutation] | LETHAL CONGENITAL CONTRACTURE SYNDROME 10; LCCS10 [AR] | ARTHROGRYPOSIS. PERTHES DISEASE. AND UPWARD GAZE PALSY; APUG [AR]
NEU1	NEURAMINIDASE DEFICIENCY [AR]
NEUROD1	DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
NEUROD2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 72; EIEE72 [AD]
NEUROG3	DIARRHEA 4. MALABSORPTIVE. CONGENITAL; DIAR4 [AR]
NEXMIF	MENTAL RETARDATION. X-LINKED 98; MRX98 [XLD]
NEXN	CARDIOMYOPATHY. DILATED. 1CC; CMD1CC [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 20; CMH20 [AD]
NF1	NEUROFIBROMATOSIS. FAMILIAL SPINAL [AD] | JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML [AD] | NEUROFIBROMATOSIS. TYPE I; NF1 [AD] | WATSON SYNDROME; WTSN [AD] | NEUROFIBROMATOSIS-NOONAN SYNDROME; NFNS [AD]
NF2	MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD] | SCHWANNOMATOSIS 1; SWNTS1 [AD] | NEUROFIBROMATOSIS. TYPE II; NF2 [AD]
NFASC	NEURODEVELOPMENTAL DISORDER WITH CENTRAL AND PERIPHERAL MOTOR DYSFUNCTION; NEDCPMD [AR]
NFE2L2	IMMUNODEFICIENCY. DEVELOPMENTAL DELAY. AND HYPOHOMOCYSTEINEMIA; IMDDHH [AD]
NFIA	BRAIN MALFORMATIONS WITH OR WITHOUT URINARY TRACT DEFECTS; BRMUTD [AD]
NFIB	MACROCEPHALY. ACQUIRED. WITH IMPAIRED INTELLECTUAL DEVELOPMENT; MACID [AD]
NFIX	MARSHALL-SMITH SYNDROME; MRSHSS [AD] | SOTOS SYNDROME 2; SOTOS2 [AD]
NFKB1	IMMUNODEFICIENCY. COMMON VARIABLE. 12; CVID12 [AD]
NFKB2	IMMUNODEFICIENCY. COMMON VARIABLE. 10; CVID10 [AD]
NFKBIA	ECTODERMAL DYSPLASIA AND IMMUNODEFICIENCY 2; EDAID2 [AD]
NFU1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 1; MMDS1 [AR]
NGF	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE V; HSAN5 [AR]
NGLY1	CONGENITAL DISORDER OF DEGLYCOSYLATION; CDDG [AR]
NHEJ1	Severe combined immunodeficiency with microcephaly growth retardation and sensitivity to ionizing radiation 611291 (3)
NHLRC1	MYOCLONIC EPILEPSY OF LAFORA [AR]
NHLRC2	FIBROSIS. NEURODEGENERATION. AND CEREBRAL ANGIOMATOSIS; FINCA [AR]
NHP2	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 2; DKCB2 [AR]
NHS	CATARACT 40; CTRCT40 [X-linked] | NANCE-HORAN SYNDROME; NHS [XLD]
NIPA1	SPASTIC PARAPLEGIA 6. AUTOSOMAL DOMINANT; SPG6 [AD]
NIPAL4	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 6; ARCI6 [AR]
NIPBL	CORNELIA DE LANGE SYNDROME 1; CDLS1 [AD]
NKX2-1	THYROID CANCER. NONMEDULLARY. 1; NMTC1 [AD] | CHOREOATHETOSIS AND CONGENITAL HYPOTHYROIDISM WITH OR WITHOUT PULMONARY DYSFUNCTION; CAHTP [AD] | CHOREA. BENIGN HEREDITARY; BHC [AD]
NKX2-5	VENTRICULAR SEPTAL DEFECT 3; VSD3 [AD] | HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 5; CHNG5 [AD] | ATRIAL SEPTAL DEFECT 7 WITH OR WITHOUT ATRIOVENTRICULAR CONDUCTION DEFECTS; ASD7 [AD] | HYPOPLASTIC LEFT HEART SYNDROME 2; HLHS2 [AD] | TETRALOGY OF FALLOT; TOF [AD]
NKX2-6	Conotruncal heart malformations 217095 (3) | Persistent truncus arteriosus 217095 (3)
NKX3-2	SPONDYLO-MEGAEPIPHYSEAL-METAPHYSEAL DYSPLASIA; SMMD [AR]
NKX6-2	SPASTIC ATAXIA 8. AUTOSOMAL RECESSIVE. WITH HYPOMYELINATING LEUKODYSTROPHY; SPAX8 [AR]
NLGN4X	ASPERGER SYNDROME. X-LINKED. SUSCEPTIBILITY TO. 2; ASPGX2 [X-linked] | AUTISM. SUSCEPTIBILITY TO. X-LINKED 2; AUTSX2 [X-linked]
NLRC4	AUTOINFLAMMATION WITH INFANTILE ENTEROCOLITIS; AIFEC [AD] | FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 4; FCAS4 [AD]
NLRP1	AUTOINFLAMMATION WITH ARTHRITIS AND DYSKERATOSIS; AIADK [AD] | RESPIRATORY PAPILLOMATOSIS. JUVENILE RECURRENT. CONGENITAL; JRRP [AR] | PALMOPLANTAR CARCINOMA. MULTIPLE SELF-HEALING; MSPC [AD]
NLRP12	FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 2; FCAS2 [AD]
NLRP3	MUCKLE-WELLS SYNDROME; MWS [AD] | DEAFNESS. AUTOSOMAL DOMINANT 34. WITH OR WITHOUT INFLAMMATION; DFNA34 [AD] | KERATOENDOTHELIITIS FUGAX HEREDITARIA; KEFH [AD] | CINCA SYNDROME; CINCA [AD] | FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 1; FCAS1 [AD]
NLRP7	HYDATIDIFORM MOLE. RECURRENT. 1; HYDM1 [AR]

NME8	CILIARY DYSKINESIA. PRIMARY. 6; CILD6 [AR]
NMNAT1	LEBER CONGENITAL AMAUROSIS 9; LCA9 [AR]
NNMT	Homocysteine plasma level (2)
NNT	GLUCOCORTICOID DEFICIENCY 4 WITH OR WITHOUT MINERALOCORTICOID DEFICIENCY; GCCD4 [AR]
NOBOX	PREMATURE OVARIAN FAILURE 5; POF5 [AD]
NOD2	INFLAMMATORY BOWEL DISEASE (CROHN DISEASE) 1; IBD1 [Multifactorial] | BLAU SYNDROME; BLAUS [AD] | YAO SYNDROME; YAOS [Multifactorial]
NODAL	HETEROTAXY. VISCERAL. 5. AUTOSOMAL; HTX5 [AD]
NOG	MULTIPLE SYNOSTOSES SYNDROME 1; SYNS1 [AD] | STAPES ANKYLOSIS WITH BROAD THUMBS AND TOES [AD] | BRACHYDACTYLY. TYPE B2; BDB2 [AD] | SYMPHALANGISM. PROXIMAL. 1A; SYM1A [AD] | TARSAL-CARPAL COALITION SYNDROME; TCC [AD]
NONO	MENTAL RETARDATION. X-LINKED. SYNDROMIC 34; MRXS34 [X-linked]
NOP10	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 1; DKCB1 [AR]
NOP56	SPINOCEREBELLAR ATAXIA 36; SCA36 [AD]
NOTCH1	ADAMS-OLIVER SYNDROME 5; AOS5 [AD] | AORTIC VALVE DISEASE 1; AOVD1 [AD]
NOTCH2	ALAGILLE SYNDROME 2; ALGS2 [AD] | HAJDU-CHENEY SYNDROME; HJCYS [AD]
NOTCH2NLC	NEURONAL INTRANUCLEAR INCLUSION DISEASE; NIID [AD]
NOTCH3	LATERAL MENINGOCELE SYNDROME; LMNS [AD] | CEREBRAL ARTERIOPATHY. AUTOSOMAL DOMINANT. WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY. TYPE 1; CADASIL1 [AD] | MYOFIBROMATOSIS. INFANTILE. 2; IMF2 [AD]
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]
NPM1	LEUKEMIA. ACUTE MYELOID; AML [AD]
NPPA	ATRIAL STANDSTILL 2; ATRST2 [AR] | ATRIAL FIBRILLATION. FAMILIAL. 6; ATFB6 [AD]
NPR2	ACROMESOMELIC DYSPLASIA. MAROTEAUX TYPE; AMDM [AR] | SHORT STATURE WITH NONSPECIFIC SKELETAL ABNORMALITIES; SNSK [AD] | EPIPHYSEAL CHONDRODYSPLASIA. MIURA TYPE; ECDM [AD]
NPRL2	EPILEPSY. FAMILIAL FOCAL. WITH VARIABLE FOCI 2; FFEVF2 [AD]
NPRL3	EPILEPSY. FAMILIAL FOCAL. WITH VARIABLE FOCI 3; FFEVF3 [AD]
NQO2	BREAST CANCER [AD]
NR0B1	ADRENAL HYPOPLASIA. CONGENITAL; AHC [XLR] | 46.XY SEX REVERSAL 2; SRXY2 [X-linked]
NR0B2	OBESITY [AD]
NR1H4	CHOLESTASIS. PROGRESSIVE FAMILIAL INTRAHEPATIC. 5; PFIC5 [AR]
NR2E3	ENHANCED S-CONE SYNDROME; ESCS [AR] | RETINITIS PIGMENTOSA 37; RP37 [AD]
NR2F1	BOSCH-BOONSTRA-SCHAAF OPTIC ATROPHY SYNDROME; BBSOAS [AD]
NR2F2	CONGENITAL HEART DEFECTS. MULTIPLE TYPES. 4; CHTD4 [AD]
NR3C1	GLUCOCORTICOID RESISTANCE. GENERALIZED; GCCR [AD]
NR3C2	PSEUDOHYPOALDOSTERONISM. TYPE I. AUTOSOMAL DOMINANT; PHA1A [AD]
NR4A3	Chondrosarcoma extraskeletal myxoid 612237 (3)
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]
NRAS	NOONAN SYNDROME 6; NS6 [AD] | COLORECTAL CANCER; CRC [Somatic mutation] | MELANOCYTIC NEVUS SYNDROME. CONGENITAL; CMNS [Somatic mutation] | THYROID CANCER. NONMEDULLARY. 2; NMTC2 [AD] | SCHIMMELPENNING-FEUERSTEIN-MIMS SYNDROME; SFM [Somatic mosaicism] | NEVUS. EPIDERMAL [Somatic mutation] | RAS-ASSOCIATED AUTOIMMUNE LEUKOPROLIFERATIVE DISORDER; RALD [AD] | MELANOSIS. NEUROCUTANEOUS; NCMS [Somatic mutation]

NRG1	{?Schizophrenia susceptibility to} 603013 (1)
NRL	RETINITIS PIGMENTOSA 27; RP27 [AD]
NRXN1	PITT-HOPKINS-LIKE SYNDROME 2; PTHSL2 [AR]
NSD1	SOTOS SYNDROME 1; SOTOS1 [AD] | BECKWITH-WIEDEMANN SYNDROME; BWS [AD]
NSDHL	CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS [XLD] | CK SYNDROME [XLR]
NSMCE2	SECKEL SYNDROME 10; SCKL10 [AR]
NSMCE3	LUNG DISEASE. IMMUNODEFICIENCY. AND CHROMOSOME BREAKAGE SYNDROME; LICS [AR]
NSMF	HYPOGONADOTROPIC HYPOGONADISM 9 WITH OR WITHOUT ANOSMIA; HH9 [AD]
NSUN2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 5; MRT5 [AR]
NT5C2	SPASTIC PARAPLEGIA 45. AUTOSOMAL RECESSIVE; SPG45 [AR]
NT5C3A	URIDINE 5-PRIME MONOPHOSPHATE HYDROLASE DEFICIENCY. HEMOLYTIC ANEMIA DUE TO [AR]
NT5E	CALCIFICATION OF JOINTS AND ARTERIES; CALJA [AR]
NTF4	Glaucoma 1 open angle 1O 613100 (3)
NTHL1	FAMILIAL ADENOMATOUS POLYPOSIS 3; FAP3 [AR]
NTN1	MIRROR MOVEMENTS 4; MRMV4 [AD]
NTRK1	INSENSITIVITY TO PAIN. CONGENITAL. WITH ANHIDROSIS; CIPA [AR]
NTRK2	OBESITY. HYPERPHAGIA. AND DEVELOPMENTAL DELAY; OBHD [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 58; EIEE58 [AD]
NUBPL	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 21; MC1DN21 [AR]
NUMA1	Leukemia acute promyelocytic somatic 612376 (3)
NUP107	GALLOWAY-MOWAT SYNDROME 7; GAMOS7 [AR] | NEPHROTIC SYNDROME. TYPE 11; NPHS11 [AR] | OVARIAN DYSGENESIS 6; ODG6 [AR]
NUP133	NEPHROTIC SYNDROME. TYPE 18; NPHS18 [AR] | GALLOWAY-MOWAT SYNDROME 8; GAMOS8 [AR]
NUP214	LEUKEMIA. ACUTE MYELOID; AML [AD] | ENCEPHALOPATHY. ACUTE. INFECTION-INDUCED. SUSCEPTIBILITY TO. 9; IIAE9 [AR]
NUP62	STRIATONIGRAL DEGENERATION. INFANTILE; SNDI [AR]
NUP85	NEPHROTIC SYNDROME. TYPE 17; NPHS17 [AR]
NUP88	FETAL AKINESIA DEFORMATION SEQUENCE 4; FADS4 [AR]
NUP93	NEPHROTIC SYNDROME. TYPE 12; NPHS12 [AR]
NUS1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 55. WITH SEIZURES; MRD55 [AD] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Iaa; CDG1AA [AR]
NXN	ROBINOW SYNDROME. AUTOSOMAL RECESSIVE 2; RRS2 [AR]
NYX	NIGHT BLINDNESS. CONGENITAL STATIONARY. TYPE 1A; CSNB1A [XLR]
OAT	GYRATE ATROPHY OF CHOROID AND RETINA; GACR [AR]
OBSL1	THREE M SYNDROME 2; 3M2 [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]
ODAPH	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA4; AI2A4 [AR]
OFD1	SIMPSON-GOLABI-BEHMEL SYNDROME. TYPE 2; SGBS2 [XLR] | JOUBERT SYNDROME 10; JBTS10 [XLR] | RETINITIS PIGMENTOSA 23; RP23 [XLR] | OROFACIODIGITAL SYNDROME I; OFD1 [XLD]
OGDH	ALPHA-KETOGLUTARATE DEHYDROGENASE DEFICIENCY [AR]
OGG1	Renal cell carcinoma clear cell somatic 144700 (3)
OGT	MENTAL RETARDATION. X-LINKED 106; MRX106 [XLR]
OPA1	OPTIC ATROPHY WITH OR WITHOUT DEAFNESS. OPHTHALMOPLEGIA. MYOPATHY. ATAXIA. AND NEUROPATHY [AD] | OPTIC ATROPHY 1; OPA1 [AD] | BEHR SYNDROME; BEHRS [AR]
OPA3	OPTIC ATROPHY 3. AUTOSOMAL DOMINANT; OPA3 [AD] | 3-METHYLGLUTACONIC ACIDURIA. TYPE III; MGCA3 [AR]

OPCML	OVARIAN CANCER [Somatic mutation]
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]
OPN1SW	TRITANOPIA [Autosomal dominant (7q31.3-q32)]
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]
OSBPL2	DEAFNESS. AUTOSOMAL DOMINANT 67; DFNA67 [AD]
OSGEP	GALLOWAY-MOWAT SYNDROME 3; GAMOS3 [AR]
OSMR	AMYLOIDOSIS. PRIMARY LOCALIZED CUTANEOUS. 1; PLCA1 [AD]
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]
OTUD6B	INTELLECTUAL DEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES. SEIZURES. AND DISTAL LIMB ANOMALIES; IDDFSDA [AR]
OTULIN	AUTOINFLAMMATION. PANNICULITIS. AND DERMATOSIS SYNDROME; AIPDS [AR]
OTX2	PITUITARY HORMONE DEFICIENCY. COMBINED. 6; CPHD6 [AD] | MICROPHTHALMIA. SYNDROMIC 5; MCOPS5 [AD]
OVOL2	CORNEAL DYSTROPHY. POSTERIOR POLYMORPHOUS. 1; PPCD1 [AD]
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency 245050 (3)
P2RX2	DEAFNESS. AUTOSOMAL DOMINANT 41; DFNA41 [AD]
P2RY12	BLEEDING DISORDER. PLATELET-TYPE. 8; BDPLT8 [AR]
P3H1	OSTEOGENESIS IMPERFECTA. TYPE VIII; OI8 [AR]
P3H2	MYOPIA. HIGH. WITH CATARACT AND VITREORETINAL DEGENERATION; MCVD [AR]
P4HA2	MYOPIA 25. AUTOSOMAL DOMINANT; MYP25 [AD]
P4HB	COLE-CARPENTER SYNDROME 1; CLCRP1 [AD]
P4HTM	HYPOTONIA. HYPOVENTILATION. IMPAIRED INTELLECTUAL DEVELOPMENT. DYSAUTONOMIA. EPILEPSY. AND EYE ABNORMALITIES; HIDEA [AR]
PABPN1	OCULOPHARYNGEAL MUSCULAR DYSTROPHY; OPMD [AD]
PACS1	SCHUURS-HOEIJMAKERS SYNDROME; SHMS [AD]
PACS2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 66; EIEE66 [AD]
PADI3	UNCOMBABLE HAIR SYNDROME 1; UHS1 [AR]
PADI6	PREIMPLANTATION EMBRYONIC LETHALITY 2; PREMBL2 [AR]
PAFAH1B1	LISSENCEPHALY 1; LIS1 [AD]
PAH	PHENYLKETONURIA; PKU [AR]
PAK1	INTELLECTUAL DEVELOPMENTAL DISORDER WITH MACROCEPHALY. SEIZURES. AND SPEECH DELAY; IDDMSSD [AD]
PAK3	MENTAL RETARDATION. X-LINKED 30; MRX30 [XLR]
PALB2	BREAST CANCER [AD]

PAM16	SPONDYLOMETAPHYSEAL DYSPLASIA. MEGARBANE-DAGHER-MELKI TYPE; SMDMDM [AR]
PANK2	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1; NBIA1 [AR] | HYPOPREBETALIPOPROTEINEMIA. ACANTHOCYTOSIS. RETINITIS PIGMENTOSA. AND PALLIDAL DEGENERATION [AR]
PANX1	OOCYTE MATURATION DEFECT 7; OOMD7 [AD]
PAPSS2	BRACHYOLMIA TYPE 4 WITH MILD EPIPHYSEAL AND METAPHYSEAL CHANGES; BCYM4 [AR]
PARK7	PARKINSON DISEASE 7. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK7 [AR]
PARN	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 6; DKCB6 [AR] | PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE. TELOMERE-RELATED. 4; PFBMFT4 [AD]
PARS2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 75; EIEE75 [AR]
PATL2	OOCYTE MATURATION DEFECT 4; OOMD4 [AR]
PAX2	PAPILLORENAL SYNDROME; PAPRS [AD] | FOCAL SEGMENTAL GLOMERULOSCLEROSIS 7; FSGS7 [AD]
PAX3	RHABDOMYOSARCOMA 2; RMS2 [Somatic mutation] | WAARDENBURG SYNDROME. TYPE 1; WS1 [AD] | WAARDENBURG SYNDROME. TYPE 3; WS3 [AD] | CRANIOFACIAL-DEAFNESS-HAND SYNDROME; CDHS [AD]
PAX4	DIABETES MELLITUS. KETOSIS-PRONE; KPD [Autosomal dominant (in 1 patient)] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [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]
PAX9	TOOTH AGENESIS. SELECTIVE. 3; STHAG3 [AD]
PBX1	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT SYNDROME WITH OR WITHOUT HEARING LOSS. ABNORMAL EARS. OR DEVELOPMENTAL DELAY; CAKUTHED [AD]
PC	Pyruvate carboxylase deficiency 266150 (3)
PCARE	Retinitis pigmentosa 54 613428 (3)
PCBD1	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. D; HPABH4D [AR]
PCCA	PROPIONIC ACIDEMIA [AR]
PCCB	PROPIONIC ACIDEMIA [AR]
PCDH12	DIENCEPHALIC-MESENCEPHALIC JUNCTION DYSPLASIA SYNDROME 1; DMJDS1 [AR]
PCDH15	DEAFNESS. AUTOSOMAL RECESSIVE 23; DFNB23 [AR] | USHER SYNDROME. TYPE ID; USH1D [AR] | USHER SYNDROME. TYPE IF; USH1F [AR]
PCDH19	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 9; EIEE9 [X-linked]
PCGF2	TURNPENNY-FRY SYNDROME; TPFS [AD]
PCK2	PHOSPHOENOLPYRUVATE CARBOXYKINASE DEFICIENCY. MITOCHONDRIAL; PCKDM [AR]
PCNT	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM. TYPE II; MOPD2 [AR]
PCSK1	PROPROTEIN CONVERTASE 1/3 DEFICIENCY [AR]
PCSK9	HYPERCHOLESTEROLEMIA. FAMILIAL. 3; FHCL3 [AD]
PCYT1A	SPONDYLOMETAPHYSEAL DYSPLASIA WITH CONE-ROD DYSTROPHY; SMDCRD [AR]
PDCD10	Cerebral cavernous malformations 3 603285 (3)
PDE10A	STRIATAL DEGENERATION. AUTOSOMAL DOMINANT 2; ADSD2 [AD] | DYSKINESIA. LIMB AND OROFACIAL. INFANTILE-ONSET; IOLOD [AR]
PDE11A	PIGMENTED NODULAR ADRENOCORTICAL DISEASE. PRIMARY. 2; PPNAD2 [AD]
PDE3A	HYPERTENSION AND BRACHYDACTYLY SYNDROME; HTNB [AD]
PDE4D	ACRODYSOSTOSIS 2 WITH OR WITHOUT HORMONE RESISTANCE; ACRDYS2 [AD]
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]

PDE8B	STRIATAL DEGENERATION. AUTOSOMAL DOMINANT 1; ADSD1 [AD]
PDGFB	BASAL GANGLIA CALCIFICATION. IDIOPATHIC. 5; IBGC5 [AD] | MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD]
PDGFRA	GASTROINTESTINAL STROMAL TUMOR; GIST [AD] | GIST-PLUS SYNDROME; GISTPS [AD] | HYPEREOSINOPHILIC SYNDROME. IDIOPATHIC; HES [Somatic mutation]
PDGFRB	MYELOPROLIFERATIVE DISORDER. CHRONIC. WITH EOSINOPHILIA [AD] | PREMATURE AGING SYNDROME. PENTTINEN TYPE; PENTT [AD] | MYOFIBROMATOSIS. INFANTILE. 1; IMF1 [AD] | BASAL GANGLIA CALCIFICATION. IDIOPATHIC. 4; IBGC4 [AD] | KOSAKI OVERGROWTH SYNDROME; KOGS [AD]
PDGFRL	HEPATOCELLULAR CARCINOMA [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation]
PDHA1	PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY; PDHAD [XLD]
PDHB	Pyruvate dehydrogenase E1-beta deficiency 614111 (3)
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]
PDXK	NEUROPATHY. HEREDITARY MOTOR AND SENSORY. TYPE VIC. WITH OPTIC ATROPHY; HMSN6C [AR]
PDYN	SPINOCEREBELLAR ATAXIA 23; SCA23 [AD]
PDZD7	USHER SYNDROME. TYPE IIC; USH2C [AR] | USHER SYNDROME. TYPE IIA; USH2A [AR] | DEAFNESS. AUTOSOMAL RECESSIVE 57; DFNB57 [AR]
PEPD	PROLIDASE DEFICIENCY [AR]
PERCC1	DIARRHEA 11. MALABSORPTIVE. CONGENITAL; DIAR11 [AR]
PET100	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV 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]
PEX12	PEROXISOME BIOGENESIS DISORDER 3B; PBD3B [AR] | PEROXISOME BIOGENESIS DISORDER 3A (ZELLWEGER); PBD3A [AR]
PEX13	PEROXISOME BIOGENESIS DISORDER 11A (ZELLWEGER); PBD11A [AR] | PEROXISOME BIOGENESIS DISORDER 11B; PBD11B [AR]
PEX14	PEROXISOME BIOGENESIS DISORDER 13A (ZELLWEGER); PBD13A [AR]
PEX16	PEROXISOME BIOGENESIS DISORDER 8A (ZELLWEGER); PBD8A [AR] | PEROXISOME BIOGENESIS DISORDER 8B; PBD8B [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]
PFKL	Hemolytic anemia due to phosphofructokinase deficiency (1)
PFKM	GLYCOGEN STORAGE DISEASE VII; GSD7 [AR]
PFN1	Amyotrophic lateral sclerosis 18 614808 (3)
PGAM2	GLYCOGEN STORAGE DISEASE X; GSD10 [AR]
PGAP1	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 42; MRT42 [AR]
PGAP2	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 3; HPMRS3 [AR]
PGAP3	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 4; HPMRS4 [AR]
PGK1	PHOSPHOGLYCERATE KINASE 1 DEFICIENCY [XLR]

PGM1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE It; CDG1T [AR]
PGM3	IMMUNODEFICIENCY 23; IMD23 [AR]
PHACTR1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 70; EIEE70 [AD]
PHEX	HYPOPHOSPHATEMIC RICKETS. X-LINKED DOMINANT; XLHR [XLD]
PHF6	BORJESON-FORSSMAN-LEHMANN SYNDROME; BFLS [XLR]
PHF8	SIDERIUS X-LINKED MENTAL RETARDATION SYNDROME; MRXSSD [XLR]
PHGDH	PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY; PHGDHD [AR] | NEU-LAXOVA SYNDROME 1; NLS1 [AR]
PHIP	CHUNG-JANSEN SYNDROME; CHUJANS [AD]
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]
PHOX2B	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD]
PHYH	REFSUM DISEASE. CLASSIC [AR]
PHYKPL	[?Phosphohydroxylysinuria] 615011 (3)
PI4KA	POLYMICROGYRIA. PERISYLVIAN. WITH CEREBELLAR HYPOPLASIA AND ARTHROGRYPOSIS; PMGYCHA [AR]
PIBF1	JOUBERT SYNDROME 33; JBTS33 [AR]
PICALM	LEUKEMIA. ACUTE MYELOID; AML [AD]
PIEZO1	DEHYDRATED HEREDITARY STOMATOCYTOSIS 1 WITH OR WITHOUT PSEUDOHYPERKALEMIA AND/OR PERINATAL EDEMA; DHS1 [AD] | LYMPHATIC MALFORMATION 6; LMPHM6 [AR]
PIEZO2	ARTHROGRYPOSIS. DISTAL. TYPE 5; DA5 [AD] | MARDEN-WALKER SYNDROME; MWKS [AD] | ARTHROGRYPOSIS. DISTAL. WITH IMPAIRED PROPRIOCEPTION AND TOUCH; DAIPT [AR] | ARTHROGRYPOSIS. DISTAL. TYPE 3; DA3 [AD]
PIGA	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 2; MCAHS2 [XLR] | PAROXYSMAL NOCTURNAL HEMOGLOBINURIA 1; PNH1 [Somatic mutation]
PIGB	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 80; EIEE80 [AR]
PIGC	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 16; GPIBD16 [AR]
PIGG	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 53; MRT53 [AR]
PIGH	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 17; GPIBD17 [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]
PIGO	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 2; HPMRS2 [AR]
PIGQ	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 77; EIEE77 [AR]
PIGS	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 18; GPIBD18 [AR]
PIGT	MULTIPLE CONGENITAL ANOMALIES-HYPOTONIA-SEIZURES SYNDROME 3; MCAHS3 [AR] | PAROXYSMAL NOCTURNAL HEMOGLOBINURIA 2; PNH2 [AD]
PIGU	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 21; GPIBD21 [AR]
PIGV	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 1; HPMRS1 [AR]
PIGW	GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT 11; GPIBD11 [AR]
PIGY	HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 6; HPMRS6 [AR]
PIK3C2A	OCULOSKELETODENTAL SYNDROME; OCSKD [AR]
PIK3CA	MACRODACTYLY [Somatic mutation] | OVARIAN CANCER [Somatic mutation] | COLORECTAL CANCER; CRC [Somatic mutation] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | KERATOSIS. SEBORRHEIC [AD] | NEVUS. EPIDERMAL [Somatic mutation] | CAPILLARY MALFORMATION OF THE LOWER LIP. LYMPHATIC MALFORMATION OF FACE AND NECK. ASYMMETRY OF FACE AND LIMBS. AND PARTIAL/GENERALIZED OVERGROWTH [Somatic mutation] | MEGALENCEPHALY-CAPILLARY MALFORMATION-POLYMICROGYRIA

	SYNDROME; MCAP [Somatic mutation] | BREAST CANCER [AD] | LUNG CANCER [Somatic mutation] | CONGENITAL LIPOMATOUS OVERGROWTH. VASCULAR MALFORMATIONS. AND EPIDERMAL NEVI [Somatic mosaicism]
PIK3CD	IMMUNODEFICIENCY 14; IMD14 [AD]
PIK3R1	SHORT SYNDROME [AD] | IMMUNODEFICIENCY 36; IMD36 [AD] | AGAMMAGLOBULINEMIA 7. AUTOSOMAL RECESSIVE; AGM7 [AR]
PIK3R2	MEGALENCEPHALY-POLYMICROGYRIA-POLYDACTYLY-HYDROCEPHALUS SYNDROME 1; MPPH1 [AD]
PIK3R5	ATAXIA-OCULOMOTOR APRAXIA 3; AOA3 [AR]
PIKFYVE	CORNEAL DYSTROPHY. FLECK [AD]
PINK1	PARKINSON DISEASE 6. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK6 [AR]
PIP5K1C	LETHAL CONGENITAL CONTRACTURE SYNDROME 3; LCCS3 [AR]
PITPNM3	CONE-ROD DYSTROPHY 5; CORD5 [AD]
PITX1	LIEBENBERG SYNDROME; LBNBG [AD] | CLUBFOOT. CONGENITAL. WITH OR WITHOUT DEFICIENCY OF LONG BONES AND/OR MIRROR-IMAGE POLYDACTYLY; CCF [AD]
PITX2	RING DERMOID OF CORNEA; RDC [AD] | ANTERIOR SEGMENT DYSGENESIS 4; ASGD4 [AD] | AXENFELD-RIEGER SYNDROME. TYPE 1; RIEG1 [AD]
PITX3	CATARACT 11. MULTIPLE TYPES; CTRCT11 [AD] | ANTERIOR SEGMENT DYSGENESIS 1; ASGD1 [AD]
PJVK	DEAFNESS. AUTOSOMAL RECESSIVE 59; DFNB59 [AR]
PKD1	POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD1 [AD]
PKD1L1	HETEROTAXY. VISCERAL. 8. AUTOSOMAL; HTX8 [AR]
PKD2	POLYCYSTIC KIDNEY DISEASE 2 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE; PKD2 [AD]
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]
PKP1	ECTODERMAL DYSPLASIA/SKIN FRAGILITY SYNDROME; EDSFS [AR]
PKP2	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 9; ARVD9 [AD]
PLA2G2A	COLORECTAL CANCER; CRC [Somatic mutation]
PLA2G4A	GASTROINTESTINAL ULCERATION. RECURRENT. WITH DYSFUNCTIONAL PLATELETS; GURDP [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]
PLAA	NEURODEVELOPMENTAL DISORDER WITH PROGRESSIVE MICROCEPHALY. SPASTICITY. AND BRAIN ANOMALIES; NDMSBA [AR]
PLAG1	SALIVARY GLAND ADENOMA. PLEOMORPHIC [Somatic mutation]
PLAT	Hyperfibrinolysis familial due to increased release of PLAT 612348 (1) | Thrombophilia familial due to decreased release of PLAT 612348 (1)
PLAU	ALZHEIMER DISEASE; AD [AD] | QUEBEC PLATELET DISORDER; QPD [AD]
PLCB1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 12; EIEE12 [AR]
PLCB2	Platelet PLC beta-2 deficiency (1)
PLCB4	AURICULOCONDYLAR SYNDROME 2; ARCND2 [AD]
PLCD1	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 3; NDNC3 [AD]
PLCE1	NEPHROTIC SYNDROME. TYPE 3; NPHS3 [AR]
PLCG2	FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 3; FCAS3 [AD] | AUTOINFLAMMATION. ANTIBODY DEFICIENCY. AND IMMUNE DYSREGULATION; APLAID [AD]
PLD1	CARDIAC VALVULAR DEFECT. DEVELOPMENTAL; CVDD [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]
PLEKHG2	LEUKODYSTROPHY AND ACQUIRED MICROCEPHALY WITH OR WITHOUT DYSTONIA; LDAMD [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]
PLIN1	LIPODYSTROPHY. FAMILIAL PARTIAL. TYPE 4; FPLD4 [AD]
PLK4	MICROCEPHALY AND CHORIORETINOPATHY. AUTOSOMAL RECESSIVE. 2; MCCRP2 [AR]
PLN	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 18; CMH18 [AD]
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]
PLPBP	EPILEPSY. EARLY-ONSET. VITAMIN B6-DEPENDENT; EPVB6D [AR]
PLS3	BONE MINERAL DENSITY QUANTITATIVE TRAIT LOCUS 18; BMND18 [XLD]
PLVAP	DIARRHEA 10. PROTEIN-LOSING ENTEROPATHY TYPE; DIAR10 [AR]
PMFBP1	SPERMATOGENIC FAILURE 31; SPGF31 [AR]
PML	Leukemia acute promyelocytic PML/RARA type (3)
PMM2	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Ia; CDG1A [AR]
PMP2	CHARCOT-MARIE-TOOTH DISEASE. DEMYELINATING. TYPE 1G; CMT1G [AD]
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]
PMPCA	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 2; SCAR2 [AR]
PMPCB	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 6; MMDS6 [AR]
PMS2	MISMATCH REPAIR CANCER SYNDROME; MMRCS [AR]
PMVK	POROKERATOSIS 1. MULTIPLE TYPES; POROK1 [AD]
PNKD	PAROXYSMAL NONKINESIGENIC DYSKINESIA 1; PNKD1 [AD]
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]
PNPLA2	NEUTRAL LIPID STORAGE DISEASE WITH MYOPATHY; NLSDM [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]
POC1B	CONE-ROD DYSTROPHY 20; CORD20 [AR]
POFUT1	DOWLING-DEGOS DISEASE 2; DDD2 [AD]
POGLUT1	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 21; LGMDR21 [AR] | DOWLING-DEGOS DISEASE 4; DDD4 [AD]
POGZ	WHITE-SUTTON SYNDROME; WHSUS [AD]
POLA1	VAN ESCH-O'DRISCOLL SYNDROME; VEODS [XLR] | PIGMENTARY DISORDER. RETICULATE. WITH SYSTEMIC MANIFESTATIONS. X-LINKED; PDR [XLR]
POLD1	MANDIBULAR HYPOPLASIA. DEAFNESS. PROGEROID FEATURES. AND LIPODYSTROPHY SYNDROME; MDPL [AD] | COLORECTAL CANCER. SUSCEPTIBILITY TO. 10; CRCS10 [AD]
POLE	FACIAL DYSMORPHISM. IMMUNODEFICIENCY. LIVEDO. AND SHORT STATURE; FILS [AR] | COLORECTAL CANCER. SUSCEPTIBILITY TO. 12; CRCS12 [AD] | INTRAUTERINE GROWTH RETARDATION. METAPHYSEAL DYSPLASIA. ADRENAL HYPOPLASIA CONGENITA. GENITAL ANOMALIES. AND IMMUNODEFICIENCY; IMAGEI [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]
POLG2	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 4; PEOA4 [AD] | MITOCHONDRIAL DNA DEPLETION SYNDROME 16 (HEPATIC TYPE); MTDPS16 [AR]

POLH	XERODERMA PIGMENTOSUM. VARIANT TYPE; XPV [AR]
POLR1A	ACROFACIAL DYSOSTOSIS. CINCINNATI TYPE; AFDCIN [AD]
POLR1C	LEUKODYSTROPHY. HYPOMYELINATING. 11; HLD11 [AR] | TREACHER COLLINS SYNDROME 3; TCS3 [AR]
POLR1D	TREACHER COLLINS SYNDROME 2; TCS2 [AD]
POLR2A	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA AND VARIABLE INTELLECTUAL AND BEHAVIORAL ABNORMALITIES; NEDHIB [AD]
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]
POMGNT2	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 8; MDDGA8 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 8; MDDGC8 [AR]
POMK	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 12; MDDGA12 [AR] | MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE). TYPE C. 12; MDDGC12 [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]
POP1	ANAUXETIC DYSPLASIA 2; ANXD2 [AR]
POR	ANTLEY-BIXLER SYNDROME WITH GENITAL ANOMALIES AND DISORDERED STEROIDOGENESIS; ABS1 [AR]
PORCN	FOCAL DERMAL HYPOPLASIA; FDH [XLD]
POU1F1	PITUITARY HORMONE DEFICIENCY. COMBINED. 1; CPHD1 [AD]
POU3F3	SNIJDERS BLOK-FISHER SYNDROME; SNIBFIS [AD]
POU3F4	DEAFNESS. X-LINKED 2; DFNX2 [XLR] | CHOROIDEREMIA. DEAFNESS. AND MENTAL RETARDATION [XLR]
POU4F3	DEAFNESS. AUTOSOMAL DOMINANT 15; DFNA15 [AD]
PPA2	SUDDEN CARDIAC FAILURE. ALCOHOL-INDUCED; SCFAI [AR] | SUDDEN CARDIAC FAILURE. INFANTILE; SCFI [AR]
PPARG	OBESITY [AD] | DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD] | LIPODYSTROPHY. FAMILIAL PARTIAL. TYPE 3; FPLD3 [AD]
PPCS	CARDIOMYOPATHY. DILATED. 2C; CMD2C [AR]
PPIB	OSTEOGENESIS IMPERFECTA. TYPE IX; OI9 [AR]
PPIP5K2	DEAFNESS. AUTOSOMAL RECESSIVE 100; DFNB100 [AR]
PPM1D	BREAST CANCER [AD] | JANSEN-DE VRIES SYNDROME; JDVS [AD]
PPOX	VARIEGATE PORPHYRIA; VP [AD]
PPP1CB	NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR 2; NSLH2 [AD]
PPP1R15B	MICROCEPHALY. SHORT STATURE. AND IMPAIRED GLUCOSE METABOLISM 2; MSSGM2 [AR]
PPP1R3A	DIABETES MELLITUS. NONINSULIN-DEPENDENT; NIDDM [AD]
PPP2CA	NEURODEVELOPMENTAL DISORDER AND LANGUAGE DELAY WITH OR WITHOUT STRUCTURAL BRAIN ABNORMALITIES; NEDLBA [AD]
PPP2R1A	MENTAL RETARDATION. AUTOSOMAL DOMINANT 36; MRD36 [AD]
PPP2R1B	LUNG CANCER [Somatic mutation]
PPP2R2B	SPINOCEREBELLAR ATAXIA 12; SCA12 [AD]
PPP2R3C	GONADAL DYSGENESIS. DYSMORPHIC FACIES. RETINAL DYSTROPHY. AND MYOPATHY; GDRM [AR] | SPERMATOGENIC FAILURE 36; SPGF36 [AD]
PPP2R5D	MENTAL RETARDATION. AUTOSOMAL DOMINANT 35; MRD35 [AD]

PPP3CA	EPILEPTIC ENCEPHALOPATHY. INFANTILE OR EARLY CHILDHOOD. 1; IECEE1 [AD] | ARTHROGRYPOSIS. CLEFT PALATE. CRANIOSYNOSTOSIS. AND IMPAIRED INTELLECTUAL DEVELOPMENT; ACCIID [AD]
PPT1	CEROID LIPOFUSCINOSIS. NEURONAL. 1; CLN1 [AR]
PQBP1	RENPENNING SYNDROME 1; RENS1 [XLR]
PRCC	Renal cell carcinoma papillary 605074 (3)
PRCD	Retinitis pigmentosa 36 610599 (3)
PRDM12	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE VIII; HSAN8 [AR]
PRDM16	LEFT VENTRICULAR NONCOMPACTION 8; LVNC8 [AD]
PRDM5	BRITTLE CORNEA SYNDROME 2; BCS2 [AR]
PRDM6	PATENT DUCTUS ARTERIOSUS 3; PDA3 [AD]
PRDX1	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA. cblC TYPE; MAHCC [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]
PRIMPOL	MYOPIA 22. AUTOSOMAL DOMINANT; MYP22 [AD]
PRKACA	PIGMENTED NODULAR ADRENOCORTICAL DISEASE. PRIMARY. 4; PPNAD4 [AD]
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]
PRKCA	Pituitary tumor invasive (3)
PRKCD	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME. TYPE III; ALPS3 [AR]
PRKCG	SPINOCEREBELLAR ATAXIA 14; SCA14 [AD]
PRKCSH	POLYCYSTIC LIVER DISEASE 1 WITH OR WITHOUT KIDNEY CYSTS; PCLD1 [AD]
PRKD1	CONGENITAL HEART DEFECTS AND ECTODERMAL DYSPLASIA; CHDED [AD]
PRKDC	IMMUNODEFICIENCY 26 WITH OR WITHOUT NEUROLOGIC ABNORMALITIES; IMD26 [AR]
PRKG1	AORTIC ANEURYSM. FAMILIAL THORACIC 8; AAT8 [AD]
PRKN	PARKINSON DISEASE 2. AUTOSOMAL RECESSIVE JUVENILE; PARK2 [AR] | OVARIAN CANCER [Somatic mutation] | LUNG CANCER [Somatic mutation]
PRKRA	DYSTONIA 16; DYT16 [AR]
PRLR	MULTIPLE FIBROADENOMAS OF THE BREAST; MFAB [AD] | HYPERPROLACTINEMIA; HPRL [AD]
PRMT7	SHORT STATURE. BRACHYDACTYLY. INTELLECTUAL DEVELOPMENTAL DISABILITY. AND SEIZURES; SBIDDS [AR]
PRNP	SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES [AD] | KURU. SUSCEPTIBILITY TO ['Slow virus' etiology with genetic factor] | HUNTINGTON DISEASE-LIKE 1; HDL1 [AD] | CREUTZFELDT-JAKOB DISEASE; CJD [AD] | FATAL FAMILIAL INSOMNIA; FFI [AD] | GERSTMANN-STRAUSSLER DISEASE; GSD [AD]
PROC	THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY. AUTOSOMAL DOMINANT; THPH3 [AD] | THROMBOPHILIA DUE TO PROTEIN C DEFICIENCY. AUTOSOMAL RECESSIVE; THPH4 [AR]
PRODH	SCHIZOPHRENIA 4; SCZD4 [AD] | HYPERPROLINEMIA. TYPE I; HYRPRO1 [AR]
PROK2	HYPOGONADOTROPIC HYPOGONADISM 4 WITH OR WITHOUT ANOSMIA; HH4 [AD]
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]
PRPF3	RETINITIS PIGMENTOSA 18; RP18 [AD]
PRPF31	RETINITIS PIGMENTOSA 11; RP11 [AD]
PRPF4	RETINITIS PIGMENTOSA 70; RP70 [AD]

PRPF6	RETINITIS PIGMENTOSA 60; RP60 [AD]
PRPF8	RETINITIS PIGMENTOSA 13; RP13 [AD]
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]
PRRT2	EPISODIC KINESIGENIC DYSKINESIA 1; EKD1 [AD] | SEIZURES. BENIGN FAMILIAL INFANTILE. 2; BFIS2 [AD] | CONVULSIONS. FAMILIAL INFANTILE. WITH PAROXYSMAL CHOREOATHETOSIS; ICCA [AD]
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]
PRUNE1	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. HYPOTONIA. AND VARIABLE BRAIN ANOMALIES; NMIHBA [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]
PSEN1	CARDIOMYOPATHY. DILATED. 1U; CMD1U [AD] | FRONTOTEMPORAL DEMENTIA; FTD [AD] | ALZHEIMER DISEASE 3; AD [AD] | ACNE INVERSA. FAMILIAL. 3; ACNINV3 [AD] | PICK DISEASE OF BRAIN [AD]
PSEN2	ALZHEIMER DISEASE 4 [AD] | CARDIOMYOPATHY. DILATED. 1V; CMD1V [AD]
PSENEN	ACNE INVERSA. FAMILIAL. 2. WITH OR WITHOUT DOWLING-DEGOS DISEASE; ACNINV2 [AD]
PSMB8	PROTEASOME-ASSOCIATED AUTOINFLAMMATORY SYNDROME 1; PRAAS1 [AR]
PSMC3IP	OVARIAN DYSGENESIS 3; ODG3 [AR]
PSMD12	STANKIEWICZ-ISIDOR SYNDROME; STISS [AD]
PSPH	PHOSPHOSERINE PHOSPHATASE DEFICIENCY; PSPHD [AR]
PSTPIP1	PYOGENIC STERILE ARTHRITIS. PYODERMA GANGRENOSUM. AND ACNE [AD]
PTCH1	BASAL CELL NEVUS SYNDROME; BCNS [AD] | HOLOPROSENCEPHALY 7; HPE7 [AD]
PTCH2	BASAL CELL NEVUS SYNDROME; BCNS [AD] | MEDULLOBLASTOMA; MDB [Somatic mutation]
PTDSS1	LENZ-MAJEWSKI HYPEROSTOTIC DWARFISM; LMHD [AD]
PTEN	MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD] | PROSTATE CANCER [Somatic mutation] | COWDEN SYNDROME 1; CWS1 [AD] | MACROCEPHALY/AUTISM SYNDROME [AD]
PTF1A	PANCREATIC AGENESIS 2; PAGEN2 [AR] | PANCREATIC AND CEREBELLAR AGENESIS; PACA [AR]
PTGIS	HYPERTENSION. ESSENTIAL [Multifactorial]
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]
PTHLH	BRACHYDACTYLY. TYPE E2; BDE2 [AD]
PTPN11	JUVENILE MYELOMONOCYTIC LEUKEMIA; JMML [AD] | NOONAN SYNDROME 1; NS1 [AD] | METACHONDROMATOSIS; METCDS [AD] | LEOPARD SYNDROME 1; LPRD1 [AD]
PTPN12	COLORECTAL CANCER; CRC [Somatic mutation]
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]
PTPRJ	COLORECTAL CANCER; CRC [Somatic mutation]
PTPRO	NEPHROTIC SYNDROME. TYPE 6; NPHS6 [AR]
PTPRQ	DEAFNESS. AUTOSOMAL DOMINANT 73; DFNA73 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 84A; DFNB84A [AR]
PTRH2	NEUROLOGIC. ENDOCRINE. AND PANCREATIC DISEASE. MULTISYSTEM. INFANTILE-ONSET; IMNEPD [AR]
PTS	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. A; HPABH4A [AR]
PUF60	VERHEIJ SYNDROME; VRJS [AD]

PUM1	SPINOCEREBELLAR ATAXIA 47; SCA47 [AD]
PURA	MENTAL RETARDATION. AUTOSOMAL DOMINANT 31; MRD31 [AD]
PUS1	MYOPATHY. LACTIC ACIDOSIS. AND SIDEROBLASTIC ANEMIA 1; MLASA1 [AR]
PUS3	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 55; MRT55 [AR]
PUS7	INTELLECTUAL DEVELOPMENTAL DISORDER WITH ABNORMAL BEHAVIOR. MICROCEPHALY. AND SHORT STATURE; IDDABS [AR]
PXDN	ANTERIOR SEGMENT DYSGENESIS 7; ASGD7 [AR]
PYCR1	CUTIS LAXA. AUTOSOMAL RECESSIVE. TYPE IIB; ARCL2B [AR]
PYCR2	LEUKODYSTROPHY. HYPOMYELINATING. 10; HLD10 [AR]
PYGL	GLYCOGEN STORAGE DISEASE VI; GSD6 [AR]
PYGM	GLYCOGEN STORAGE DISEASE V; GSD5 [AR]
PYROXD1	MYOPATHY. MYOFIBRILLAR. 8; MFM8 [AR]
QARS1	MICROCEPHALY. PROGRESSIVE. WITH SEIZURES AND CEREBRAL AND CEREBELLAR ATROPHY; MSCCA [AR]
QDPR	HYPERPHENYLALANINEMIA. BH4-DEFICIENT. C; HPABH4C [AR]
QRICH1	VERVERI-BRADY SYNDROME; VERBRAS [AD]
QRICH2	SPERMATOGENIC FAILURE 35; SPGF35 [AR]
RAB11B	NEURODEVELOPMENTAL DISORDER WITH ATAXIC GAIT. ABSENT SPEECH. AND DECREASED CORTICAL WHITE MATTER; NDAGSCW [AD]
RAB18	WARBURG MICRO SYNDROME 3; WARBM3 [AR]
RAB23	CARPENTER SYNDROME 1; CRPT1 [AR]
RAB27A	GRISCELLI SYNDROME. TYPE 2; GS2 [AR]
RAB28	CONE-ROD DYSTROPHY 18; CORD18 [AR]
RAB33B	SMITH-MCCORT DYSPLASIA 2; SMC2 [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]
RAB7A	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2B; CMT2B [AD]
RAC1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 48; MRD48 [AD]
RAC2	Neutrophil immunodeficiency syndrome 608203 (3)
RAC3	NEURODEVELOPMENTAL DISORDER WITH STRUCTURAL BRAIN ANOMALIES AND DYSMORPHIC FACIES; NEDBAF [AD]
RAD21	CORNELIA DE LANGE SYNDROME 4; CDLS4 [AD] | MUNGAN SYNDROME; MGS [AR]
RAD50	Nijmegen breakage syndrome-like disorder 613078 (3)
RAD51	FANCONI ANEMIA. COMPLEMENTATION GROUP R; FANCR [AD] | MIRROR MOVEMENTS 2; MRMV2 [AD] | BREAST CANCER [AD]
RAD51C	FANCONI ANEMIA. COMPLEMENTATION GROUP O; FANCO [AR]
RAD54B	COLORECTAL CANCER; CRC [Somatic mutation]
RAD54L	BREAST CANCER [AD]
RAF1	CARDIOMYOPATHY. DILATED. 1NN; CMD1NN [AD] | NOONAN SYNDROME 5; NS5 [AD]
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]
RAI1	SMITH-MAGENIS SYNDROME; SMS [AD]
RAP1GDS1	Lymphocytic leukemia acute T-cell (3)
RAPSN	MYASTHENIC SYNDROME. CONGENITAL. 11. ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY; CMS11 [AR] | FETAL AKINESIA DEFORMATION SEQUENCE 1; FADS1 [AR]
RARA	Leukemia acute promyelocytic 612376 (1)
RARB	MICROPHTHALMIA. SYNDROMIC 12; MCOPS12 [AD]

RARS1	LEUKODYSTROPHY. HYPOMYELINATING. 9; HLD9 [AR]
RARS2	PONTOCEREBELLAR HYPOPLASIA. TYPE 6; PCH6 [AR]
RASA1	CAPILLARY MALFORMATION-ARTERIOVENOUS MALFORMATION 1; CMAVM1 [AD]
RASGRP1	IMMUNODEFICIENCY 64; IMD64 [AR]
RAX	MICROPHTHALMIA. ISOLATED 3; MCOP3 [AR]
RAX2	CONE-ROD DYSTROPHY 11; CORD11 [AD]
RB1	BLADDER CANCER [Somatic mutation] | RETINOBLASTOMA; RB1 [AD] | OSTEOGENIC SARCOMA [Somatic mutation] | SMALL CELL CANCER OF THE LUNG [AD]
RB1CC1	BREAST CANCER [AD]
RBBP8	SECKEL SYNDROME 2; SCKL2 [AR] | JAWAD SYNDROME; JWDS [AR]
RBCK1	POLYGLUCOSAN BODY MYOPATHY 1 WITH OR WITHOUT IMMUNODEFICIENCY; PGBM1 [AR]
RBM10	TARP SYNDROME; TARPS [XLR]
RBM15	Megakaryoblastic leukemia acute (2)
RBM20	CARDIOMYOPATHY. DILATED. 1DD; CMD1DD [AD]
RBM8A	THROMBOCYTOPENIA-ABSENT RADIUS SYNDROME; TAR [AR]
RBP4	RETINAL DYSTROPHY. IRIS COLOBOMA. AND COMEDOGENIC ACNE SYNDROME; RDCCAS [AR] | MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 10; MCOPCB10 [AD]
RBPJ	ADAMS-OLIVER SYNDROME 3; AOS3 [AD]
RCBTB1	RETINAL DYSTROPHY WITH OR WITHOUT EXTRAOCULAR ANOMALIES; RDEOA [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]
REEP1	SPASTIC PARAPLEGIA 31. AUTOSOMAL DOMINANT; SPG31 [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VB; HMN5B [AD]
REEP6	RETINITIS PIGMENTOSA 77; RP77 [AR]
RELN	LISSENCEPHALY 2; LIS2 [AR] | EPILEPSY. FAMILIAL TEMPORAL LOBE. 7; ETL7 [AD]
RELT	AMELOGENESIS IMPERFECTA. TYPE IIIC; AI3C [AR]
REN	HYPERURICEMIC NEPHROPATHY. FAMILIAL JUVENILE. 2; HNFJ2 [AD] | RENAL TUBULAR DYSGENESIS; RTD [AR]
RERE	NEURODEVELOPMENTAL DISORDER WITH OR WITHOUT ANOMALIES OF THE BRAIN. EYE. OR HEART; NEDBEH [AD]
REST	DEAFNESS. AUTOSOMAL DOMINANT 27; DFNA27 [AD] | FIBROMATOSIS. GINGIVAL. 5; GINGF5 [AD]
RET	CENTRAL HYPOVENTILATION SYNDROME. CONGENITAL; CCHS [AD] | HIRSCHSPRUNG DISEASE. SUSCEPTIBILITY TO. 1; HSCR1 [AD] | MULTIPLE ENDOCRINE NEOPLASIA. TYPE IIA; MEN2A [AD] | MULTIPLE ENDOCRINE NEOPLASIA. TYPE IIB; MEN2B [AD] | PHEOCHROMOCYTOMA [AD] | THYROID CARCINOMA. FAMILIAL MEDULLARY; MTC [AD]
RETREG1	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE IIB; HSAN2B [AR]
RFC1	CEREBELLAR ATAXIA. NEUROPATHY. AND VESTIBULAR AREFLEXIA SYNDROME; CANVAS [AR]
RFT1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE In; CDG1N [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)
RGS9BP	Bradyopsia 608415 (3)
RHAG	OVERHYDRATED HEREDITARY STOMATOCYTOSIS; OHST [AD] | RH-NULL. REGULATOR TYPE; RHNR [AD]

RHBDF2	TYLOSIS WITH ESOPHAGEAL CANCER; TOC [AD]
RHCE	Rh-null disease amorph type 617970 (3) | [Blood group Rhesus] (3)
RHO	RETINITIS PIGMENTOSA 4; RP4 [AD] | FUNDUS ALBIPUNCTATUS [AD]
RHOBTB2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 64; EIEE64 [AD]
RHOH	EPIDERMODYSPLASIA VERRUCIFORMIS. SUSCEPTIBILITY TO. 4; EV4 [AR]
RIMS1	Cone-rod dystrophy 7 603649 (3)
RIN2	MACS SYNDROME [AR]
RINT1	INFANTILE LIVER FAILURE SYNDROME 3; ILFS3 [AR]
RIPK1	IMMUNODEFICIENCY 57; IMD57 [AR]
RIPK4	CHAND SYNDROME; CHANDS [AR] | BARTSOCAS-PAPAS SYNDROME; BPS [AR]
RIT1	NOONAN SYNDROME 8; NS8 [AD]
RLBP1	BOTHNIA RETINAL DYSTROPHY [AR] | FUNDUS ALBIPUNCTATUS [AD]
RLIM	TONNE-KALSCHEUER SYNDROME; TOKAS [X-linked]
RMND1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 11; COXPD11 [AR]
RMRP	ANAUXETIC DYSPLASIA 1; ANXD1 [AR] | CARTILAGE-HAIR HYPOPLASIA; CHH [AR] | METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS; MDWH [AR]
RNASEH1	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL RECESSIVE 2; PEOB2 [AR]
RNASEH2A	AICARDI-GOUTIERES SYNDROME 4; AGS4 [AR]
RNASEH2B	AICARDI-GOUTIERES SYNDROME 2; AGS2 [AR]
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3; AGS3 [AR]
RNASEL	PROSTATE CANCER. HEREDITARY. 1; HPC1 [AD]
RNASET2	LEUKOENCEPHALOPATHY. CYSTIC. WITHOUT MEGALENCEPHALY [AR]
RNF125	TENORIO SYNDROME; TNORS [AD]
RNF13	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 73; EIEE73 [AD]
RNF139	Renal cell carcinoma 144700 (3)
RNF168	RIDDLE SYNDROME; RIDL [AR]
RNF170	ATAXIA. SENSORY. 1. AUTOSOMAL DOMINANT; SNAX1 [AD]
RNF212	Recombination rate QTL 1 612042 (3)
RNF216	GORDON HOLMES SYNDROME; GDHS [AR]
RNF43	SESSILE SERRATED POLYPOSIS CANCER SYNDROME; SSPCS [AD]
RNF6	ESOPHAGEAL CANCER [Somatic mutation]
RNU4ATAC	ROIFMAN SYNDROME; RFMN [AR] | MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM. TYPE I; MOPD1 [AR]
ROBO2	VESICOURETERAL REFLUX 2; VUR2 [AD]
ROBO3	GAZE PALSY. FAMILIAL HORIZONTAL. WITH PROGRESSIVE SCOLIOSIS 1; HGPPS1 [AR]
ROBO4	AORTIC VALVE DISEASE 3; AOVD3 [AD]
ROGDI	KOHLSCHUTTER-TONZ SYNDROME; KTZS [AR]
ROM1	RETINITIS PIGMENTOSA 7; RP7 [AD]
ROR2	BRACHYDACTYLY. TYPE B1; BDB1 [AD] | ROBINOW SYNDROME. AUTOSOMAL RECESSIVE 1; RRS1 [AR]
RORA	INTELLECTUAL DEVELOPMENTAL DISORDER WITH OR WITHOUT EPILEPSY OR CEREBELLAR ATAXIA; IDDECA [AD]
RORC	IMMUNODEFICIENCY 42; IMD42 [AR]
RP1	RETINITIS PIGMENTOSA 1; RP1 [AD]
RP1L1	OCCULT MACULAR DYSTROPHY; OCMD [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]
RPL10	MENTAL RETARDATION. X-LINKED. SYNDROMIC. 35; MRXS35 [XLR]
RPL11	DIAMOND-BLACKFAN ANEMIA 7; DBA7 [AD]
RPL21	HYPOTRICHOSIS 12; HYPT12 [AD]
RPL35A	DIAMOND-BLACKFAN ANEMIA 5; DBA5 [AD]
RPL5	DIAMOND-BLACKFAN ANEMIA 6; DBA6 [AD]
RPS10	DIAMOND-BLACKFAN ANEMIA 9; DBA9 [AD]
RPS14	CHROMOSOME 5q DELETION SYNDROME [Somatic mutation]
RPS17	DIAMOND-BLACKFAN ANEMIA 4; DBA4 [AD]
RPS19	DIAMOND-BLACKFAN ANEMIA 1; DBA1 [AD]
RPS23	BRACHYCEPHALY. TRICHOMEGALY. AND DEVELOPMENTAL DELAY; BTDD [AD]
RPS24	DIAMOND-BLACKFAN ANEMIA 3; DBA3 [AD]
RPS26	DIAMOND-BLACKFAN ANEMIA 10; DBA10 [AD]
RPS28	DIAMOND-BLACKFAN ANEMIA 15 WITH MANDIBULOFACIAL DYSOSTOSIS; DBA15 [AD]
RPS29	DIAMOND-BLACKFAN ANEMIA 13; DBA13 [AD]
RPS6KA3	COFFIN-LOWRY SYNDROME; CLS [XLD] | MENTAL RETARDATION. X-LINKED 19; MRX19 [XLD]
RPS7	DIAMOND-BLACKFAN ANEMIA 8; DBA8 [AD]
RPSA	ASPLENIA. ISOLATED CONGENITAL; ICAS [AD]
RRAS2	NOONAN SYNDROME 12; NS12 [AD]
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]
RSPH1	CILIARY DYSKINESIA. PRIMARY. 24; CILD24 [AR]
RSPH3	CILIARY DYSKINESIA. PRIMARY. 32; CILD32 [AR]
RSPH4A	Ciliary dyskinesia primary 11 612649 (3)
RSPH9	Ciliary dyskinesia primary 12 612650 (3)
RSPO1	PALMOPLANTAR HYPERKERATOSIS WITH SQUAMOUS CELL CARCINOMA OF SKIN AND 46.XX SEX REVERSAL [AR]
RSPO2	TETRAAMELIA SYNDROME 2; TETAMS2 [AR] | HUMEROFEMORAL HYPOPLASIA WITH RADIOTIBIAL RAY DEFICIENCY; HHRRD [AR]
RSPO4	NAIL DISORDER. NONSYNDROMIC CONGENITAL. 4; NDNC4 [AR]
RSPRY1	SPONDYLOEPIMETAPHYSEAL DYSPLASIA. FADEN-ALKURAYA TYPE; SEMDFA [AR]
RSRC1	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 70; MRT70 [AR]
RTEL1	DYSKERATOSIS CONGENITA. AUTOSOMAL RECESSIVE 5; DKCB5 [AD] | PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE. TELOMERE-RELATED. 3; PFBMFT3 [AD]
RTN2	SPASTIC PARAPLEGIA 12. AUTOSOMAL DOMINANT; SPG12 [AD]
RTN4IP1	OPTIC ATROPHY 10 WITH OR WITHOUT ATAXIA. MENTAL RETARDATION. AND SEIZURES; OPA10 [AR]
RTTN	MICROCEPHALY. SHORT STATURE. AND POLYMICROGYRIA WITH OR WITHOUT SEIZURES; MSSP [AR]
RUNX1	LEUKEMIA. ACUTE MYELOID; AML [AD] | PLATELET DISORDER. FAMILIAL. WITH ASSOCIATED MYELOID MALIGNANCY; FPDMM [AD]
RUNX2	CLEIDOCRANIAL DYSPLASIA; CCD [AD] | METAPHYSEAL DYSPLASIA WITH MAXILLARY HYPOPLASIA WITH OR WITHOUT BRACHYDACTYLY; MDMHB [AD]
RUSC2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 61; MRT61 [AR]

RXYLT1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH BRAIN AND EYE ANOMALIES). TYPE A. 10; MDDGA10 [AR]
RYR1	CENTRAL CORE DISEASE OF MUSCLE; CCD [AD] | MALIGNANT HYPERTHERMIA. SUSCEPTIBILITY TO. 1; MHS1 [AD] | MINICORE MYOPATHY WITH EXTERNAL OPHTHALMOPLEGIA [AR]
RYR2	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 2; ARVD2 [AD] | VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 1. WITH OR WITHOUT ATRIAL DYSFUNCTION AND/OR DILATED CARDIOMYOPATHY; CPVT1 [AD]
S1PR2	DEAFNESS. AUTOSOMAL RECESSIVE 68; DFNB68 [AR]
SACS	SPASTIC ATAXIA. CHARLEVOIX-SAGUENAY TYPE; SACS [AR]
SAG	OGUCHI DISEASE 1 [AR]
SALL1	TOWNES-BROCKS SYNDROME 1; TBS1 [AD]
SALL4	IVIC SYNDROME [AD] | DUANE-RADIAL RAY SYNDROME; DRRS [AD]
SAMD12	EPILEPSY. FAMILIAL ADULT MYOCLONIC. 1; FAME1 [AD]
SAMD9	MIRAGE SYNDROME; MIRAGE [AD] | TUMORAL CALCINOSIS. NORMOPHOSPHATEMIC. FAMILIAL; NFTC [AR]
SAMD9L	ATAXIA-PANCYTOPENIA SYNDROME; ATXPC [AD]
SAMHD1	CHILBLAIN LUPUS 2; CHBL2 [AD] | AICARDI-GOUTIERES SYNDROME 5; AGS5 [AR]
SAR1B	CHYLOMICRON RETENTION DISEASE; CMRD [AR]
SARS2	HYPERURICEMIA. PULMONARY HYPERTENSION. RENAL FAILURE. AND ALKALOSIS SYNDROME; HUPRAS [AR]
SASH1	CANCER. ALOPECIA. PIGMENT DYSCRASIA. ONYCHODYSTROPHY. AND KERATODERMA; CAPOK [AR] | DYSCHROMATOSIS UNIVERSALIS HEREDITARIA 1; DUH1 [AD]
SATB2	GLASS SYNDROME; GLASS [AD]
SBDS	SHWACHMAN-DIAMOND SYNDROME 1; SDS1 [AR]
SBF1	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4B3; CMT4B3 [AR]
SBF2	CHARCOT-MARIE-TOOTH DISEASE. TYPE 4B2; CMT4B2 [AR]
SC5D	LATHOSTEROLOSIS [AR]
SCAPER	INTELLECTUAL DEVELOPMENTAL DISORDER AND RETINITIS PIGMENTOSA; IDDRP [AR]
SCARB2	EPILEPSY. PROGRESSIVE MYOCLONIC. 4. WITH OR WITHOUT RENAL FAILURE; EPM4 [AR]
SCARF2	VAN DEN ENDE-GUPTA SYNDROME; VDEGS [AR]
SCN10A	EPISODIC PAIN SYNDROME. FAMILIAL. 2; FEPS2 [AD]
SCN11A	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE VII; HSAN7 [AD] | EPISODIC PAIN SYNDROME. FAMILIAL. 3; FEPS3 [AD]
SCN1A	MIGRAINE. FAMILIAL HEMIPLEGIC. 3; FHM3 [AD] | GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS. TYPE 2; GEFSP2 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 6; EIEE6 [AD]
SCN1B	ATRIAL FIBRILLATION. FAMILIAL. 13; ATFB13 [AD] | GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS. TYPE 1; GEFSP1 [AD] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 52; EIEE52 [AR]
SCN2A	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 11; EIEE11 [AD] | SEIZURES. BENIGN FAMILIAL INFANTILE. 3; BFIS3 [AD]
SCN2B	ATRIAL FIBRILLATION. FAMILIAL. 14; ATFB14 [AD]
SCN3A	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 62; EIEE62 [AD] | EPILEPSY. FAMILIAL FOCAL. WITH VARIABLE FOCI 4; FFEVF4 [AD]
SCN3B	BRUGADA SYNDROME 7; BRGDA7 [AD]
SCN4A	HYPERKALEMIC PERIODIC PARALYSIS; HYPP [AD] | MYOTONIA. POTASSIUM-AGGRAVATED [AD] | HYPOKALEMIC PERIODIC PARALYSIS. TYPE 2; HOKPP2 [AD] | MYASTHENIC SYNDROME. CONGENITAL. 16; CMS16 [AR] | PARAMYOTONIA CONGENITA OF VON EULENBURG; PMC [AD]
SCN4B	LONG QT SYNDROME 10; LQT10 [AD]
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]
SCN8A	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 13; EIEE13 [AD] | COGNITIVE IMPAIRMENT WITH OR WITHOUT CEREBELLAR ATAXIA; CIAT [AD] | MYOCLONUS. FAMILIAL. 2; MYOCL2 [AD] | SEIZURES. BENIGN FAMILIAL INFANTILE. 5; BFIS5 [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]
SCO1	MITOCHONDRIAL COMPLEX IV DEFICIENCY [AR]
SCO2	MYOPIA 6; MYP6 [AD] | CARDIOENCEPHALOMYOPATHY. FATAL INFANTILE. DUE TO CYTOCHROME c OXIDASE DEFICIENCY 1; CEMCOX1 [AR]
SCYL1	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 21; SCAR21 [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]
SDHAF2	PARAGANGLIOMAS 2; PGL2 [AD]
SDHB	GASTROINTESTINAL STROMAL TUMOR; GIST [AD] | PHEOCHROMOCYTOMA [AD] | PARAGANGLIOMAS 4; PGL4 [AD]
SDHC	GASTROINTESTINAL STROMAL TUMOR; GIST [AD] | PARAGANGLIOMAS 3; PGL3 [AD]
SDHD	PHEOCHROMOCYTOMA [AD] | PARAGANGLIOMAS 1; PGL1 [AD] | MITOCHONDRIAL COMPLEX II DEFICIENCY [AR]
SDR9C7	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 13; ARCI13 [AR]
SEC23A	CRANIOLENTICULOSUTURAL DYSPLASIA; CLSD [AR]
SEC23B	ANEMIA. CONGENITAL DYSERYTHROPOIETIC. TYPE II; CDAN2 [AR] | COWDEN SYNDROME 7; CWS7 [AD]
SEC24D	COLE-CARPENTER SYNDROME 2; CLCRP2 [AR]
SEC61A1	HYPERURICEMIC NEPHROPATHY. FAMILIAL JUVENILE. 4; HNFJ4 [AD]
SEC63	POLYCYSTIC LIVER DISEASE 2 WITH OR WITHOUT KIDNEY CYSTS; PCLD2 [AD]
SECISBP2	THYROID HORMONE METABOLISM. ABNORMAL; THMA [AR]
SELENBP1	EXTRAORAL HALITOSIS DUE TO METHANETHIOL OXIDASE DEFICIENCY; EHMTO [AR]
SELENON	RIGID SPINE MUSCULAR DYSTROPHY 1; RSMD1 [AR] | MYOPATHY. CONGENITAL. WITH FIBER-TYPE DISPROPORTION; CFTD [AD]
SEMA4A	CONE-ROD DYSTROPHY 10; CORD10 [AR] | RETINITIS PIGMENTOSA 35; RP35 [AD]
SEPSECS	PONTOCEREBELLAR HYPOPLASIA. TYPE 2D; PCH2D [AR]
SEPTIN12	SPERMATOGENIC FAILURE 10; SPGF10 [AD]
SEPTIN9	AMYOTROPHY. HEREDITARY NEURALGIC; HNA [AD]
SERAC1	3-METHYLGLUTACONIC ACIDURIA WITH DEAFNESS. ENCEPHALOPATHY. AND LEIGH-LIKE SYNDROME; MEGDEL [AR]
SERPINA1	ALPHA-1-ANTITRYPSIN DEFICIENCY; A1ATD [AR]
SERPINA3	Alpha-1-antichymotrypsin deficiency (3) | Cerebrovascular disease occlusive (3)
SERPINA6	CORTICOSTEROID-BINDING GLOBULIN DEFICIENCY [AD]
SERPINB7	PALMOPLANTAR KERATODERMA. NAGASHIMA TYPE; PPKN [AR]
SERPINB8	PEELING SKIN SYNDROME 5; PSS5 [AR]
SERPINC1	ANTITHROMBIN III DEFICIENCY; AT3D [AD]
SERPIND1	HEPARIN COFACTOR II DEFICIENCY [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]
SERPINI1	ENCEPHALOPATHY. FAMILIAL. WITH NEUROSERPIN INCLUSION BODIES; FENIB [AD]
SET	MENTAL RETARDATION. AUTOSOMAL DOMINANT 58; MRD58 [AD]
SETBP1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 29; MRD29 [AD] | SCHINZEL-GIEDION MIDFACE RETRACTION SYNDROME [AD]
SETD2	LUSCAN-LUMISH SYNDROME; LLS [AD]

SETD5	MENTAL RETARDATION. AUTOSOMAL DOMINANT 23; MRD23 [AD]
SETX	AMYOTROPHIC LATERAL SCLEROSIS 4. JUVENILE; ALS4 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE. WITH AXONAL NEUROPATHY 2; SCAN2 [AR]
SF3B1	Myelodysplastic syndrome somatic 614286 (3)
SF3B4	ACROFACIAL DYSOSTOSIS 1. NAGER TYPE; AFD1 [AD]
SFRP4	PYLE DISEASE; PYL [AR]
SFTPA2	PULMONARY FIBROSIS. IDIOPATHIC; IPF [AD]
SFTPB	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 1; SMDP1 [AR]
SFTPC	SURFACTANT METABOLISM DYSFUNCTION. PULMONARY. 2; SMDP2 [AD]
SFXN4	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 18; COXPD18 [AR]
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]
SGCE	DYSTONIA 11. MYOCLONIC; DYT11 [AD]
SGCG	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 5; LGMDR5 [AR]
SGMS2	CALVARIAL DOUGHNUT LESIONS WITH BONE FRAGILITY; CDL [AD]
SGO1	CHRONIC ATRIAL AND INTESTINAL DYSRHYTHMIA; CAID [AR]
SGPL1	NEPHROTIC SYNDROME. TYPE 14; NPHS14 [AR]
SGSH	MUCOPOLYSACCHARIDOSIS. TYPE IIIA; MPS3A [AR]
SH2B3	THROMBOCYTHEMIA 1; THCYT1 [AD] | MYELOFIBROSIS [Somatic mutation] | ERYTHROCYTOSIS. FAMILIAL. 1; ECYT1 [AD]
SH2D1A	LYMPHOPROLIFERATIVE SYNDROME. X-LINKED. 1; XLP1 [XLR]
SH3BP2	CHERUBISM [AD]
SH3GL1	LEUKEMIA. ACUTE MYELOID; AML [AD]
SH3PXD2B	FRANK-TER HAAR SYNDROME; FTHS [AR]
SH3TC2	MONONEUROPATHY OF THE MEDIAN NERVE. MILD; MNMN [AD] | CHARCOT-MARIE-TOOTH DISEASE. TYPE 4C; CMT4C [AR]
SHANK3	SCHIZOPHRENIA 15; SCZD15 [AD] | PHELAN-MCDERMID SYNDROME; PHMDS [AD]
SHH	MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 5; MCOPCB5 [AD] | SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR; SMMCI [AD] | HOLOPROSENCEPHALY 3; HPE3 [AD]
SHOC2	NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR 1; NSLH1 [AD]
SHOX	LERI-WEILL DYSCHONDROSTEOSIS; LWD [AD] | LANGER MESOMELIC DYSPLASIA; LMD [AR]
SHROOM4	STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME; SDSX [X-linked]
SI	Sucrase-isomaltase deficiency congenital 222900 (3)
SIK1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 30; EIEE30 [AD]
SIL1	MARINESCO-SJOGREN SYNDROME; MSS [AR]
SIN3A	WITTEVEEN-KOLK SYNDROME; WITKOS [AD]
SIX1	BRANCHIOOTIC SYNDROME 3; BOS3 [AD] | DEAFNESS. AUTOSOMAL DOMINANT 23; DFNA23 [AD]
SIX3	HOLOPROSENCEPHALY 2; HPE2 [AD]
SIX5	Branchiootorenal syndrome 2 610896 (3)
SIX6	OPTIC DISC ANOMALIES WITH RETINAL AND/OR MACULAR DYSTROPHY; ODRMD [AR]
SKI	SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME; SGS [AD]
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2; THES2 [AR]
SLC10A2	BILE ACID MALABSORPTION. PRIMARY; PBAM [AR]
SLC10A7	SHORT STATURE. AMELOGENESIS IMPERFECTA. AND SKELETAL DYSPLASIA WITH SCOLIOSIS; SSASKS [AR]
SLC11A2	ANEMIA. HYPOCHROMIC MICROCYTIC. WITH IRON OVERLOAD 1; AHMIO1 [AR]

SLC12A1	BARTTER SYNDROME. TYPE 1. ANTENATAL; BARTS1 [AR]
SLC12A3	GITELMAN SYNDROME; GTLMNS [AR]
SLC12A5	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 34; EIEE34 [AR] | EPILEPSY. IDIOPATHIC GENERALIZED. SUSCEPTIBILITY TO. 14; EIG14 [AD]
SLC12A6	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY; ACCPN [AR]
SLC13A3	LEUKOENCEPHALOPATHY. ACUTE REVERSIBLE. WITH INCREASED URINARY ALPHA-KETOGLUTARATE; ARLIAK [AR]
SLC13A5	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 25. WITH AMELOGENESIS IMPERFECTA; EIEE25 [AR]
SLC16A1	HYPERINSULINEMIC HYPOGLYCEMIA. FAMILIAL. 7; HHF7 [AD] | MONOCARBOXYLATE TRANSPORTER 1 DEFICIENCY; MCT1D [AD] | ERYTHROCYTE LACTATE TRANSPORTER DEFECT [AD]
SLC16A12	CATARACT 47; CTRCT47 [AD]
SLC16A2	ALLAN-HERNDON-DUDLEY SYNDROME; AHDS [X-linked]
SLC17A5	SALLA DISEASE; SD [AR] | INFANTILE SIALIC ACID STORAGE DISEASE; ISSD [AR]
SLC17A8	DEAFNESS. AUTOSOMAL DOMINANT 25; DFNA25 [AD]
SLC17A9	POROKERATOSIS 8. DISSEMINATED SUPERFICIAL ACTINIC TYPE; POROK8 [AD]
SLC18A3	MYASTHENIC SYNDROME. CONGENITAL. 21. PRESYNAPTIC; CMS21 [AR]
SLC19A2	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME; TRMA [AR]
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 (BIOTIN- OR THIAMINE-RESPONSIVE TYPE); THMD2 [AR]
SLC1A2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 41; EIEE41 [AD]
SLC1A3	EPISODIC ATAXIA. TYPE 6; EA6 [AD]
SLC1A4	SPASTIC TETRAPLEGIA. THIN CORPUS CALLOSUM. AND PROGRESSIVE MICROCEPHALY; SPATCCM [AR]
SLC20A2	BASAL GANGLIA CALCIFICATION. IDIOPATHIC. 1; IBGC1 [AD]
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]
SLC24A4	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA5; AI2A5 [AR] | SKIN/HAIR/EYE PIGMENTATION. VARIATION IN. 6; SHEP6 [AR]
SLC24A5	ALBINISM. OCULOCUTANEOUS. TYPE VI; OCA6 [AR]
SLC25A1	COMBINED D-2- AND L-2-HYDROXYGLUTARIC ACIDURIA; D2L2AD [AR] | MYASTHENIC SYNDROME. CONGENITAL. 23. PRESYNAPTIC; CMS23 [AR]
SLC25A11	PARAGANGLIOMAS 6; PGL6 [AD]
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]
SLC25A24	FONTAINE PROGEROID SYNDROME; FPS [AD]
SLC25A26	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 28; COXPD28 [AR]
SLC25A3	Mitochondrial phosphate carrier deficiency 610773 (3)
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]
SLC25A42	METABOLIC CRISES. RECURRENT. WITH VARIABLE ENCEPHALOMYOPATHIC FEATURES AND NEUROLOGIC REGRESSION; MECREN [AR]
SLC25A46	NEUROPATHY. HEREDITARY MOTOR AND SENSORY. TYPE VIB. WITH OPTIC ATROPHY; HMSN6B [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]
SLC26A8	SPERMATOGENIC FAILURE 3; SPGF3 [AD]
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]
SLC2A9	HYPOURICEMIA. RENAL. 2; RHUC2 [AD]
SLC30A10	HYPERMANGANESEMIA WITH DYSTONIA 1; HMNDYT1 [AR]
SLC30A2	ZINC DEFICIENCY. TRANSIENT NEONATAL; TNZD [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]
SLC35A2	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIm; CDG2M [XLD]
SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIc; CDG2C [AR]
SLC35D1	SCHNECKENBECKEN DYSPLASIA; SHNKND [AR]
SLC36A2	IMINOGLYCINURIA [AR] | HYPERGLYCINURIA [AD]
SLC37A4	GLYCOGEN STORAGE DISEASE Ic; GSD1C [AR] | GLYCOGEN STORAGE DISEASE Ib; GSD1B [AR]
SLC38A8	FOVEAL HYPOPLASIA 2; FVH2 [AR]
SLC39A13	EHLERS-DANLOS SYNDROME. SPONDYLODYSPLASTIC TYPE. 3; EDSSPD3 [AR]
SLC39A14	HYPERMANGANESEMIA WITH DYSTONIA 2; HMNDYT2 [AR] | HYPEROSTOSIS CRANIALIS INTERNA; HCIN [AD]
SLC39A4	ACRODERMATITIS ENTEROPATHICA. ZINC-DEFICIENCY TYPE; AEZ [AR]
SLC39A5	MYOPIA 24. AUTOSOMAL DOMINANT; MYP24 [AD]
SLC39A8	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIn; CDG2N [AR]
SLC3A1	CYSTINURIA [AD]
SLC40A1	HEMOCHROMATOSIS. TYPE 4; HFE4 [AD]
SLC45A1	INTELLECTUAL DEVELOPMENTAL DISORDER WITH NEUROPSYCHIATRIC FEATURES; IDDNPF [AR]
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]
SLC52A1	RIBOFLAVIN DEFICIENCY; RBFVD [AD]
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]
SLC5A7	MYASTHENIC SYNDROME. CONGENITAL. 20. PRESYNAPTIC; CMS20 [AR] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VIIA; HMN7A [AD]
SLC6A1	MYOCLONIC-ATONIC EPILEPSY; MAE [AD]
SLC6A17	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 48; MRT48 [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]
SLC6A9	GLYCINE ENCEPHALOPATHY WITH NORMAL SERUM GLYCINE [AR]
SLC7A14	RETINITIS PIGMENTOSA 68; RP68 [AR]
SLC7A7	LYSINURIC PROTEIN INTOLERANCE; LPI [AR]
SLC7A9	CYSTINURIA [AD]
SLC9A3	DIARRHEA 8. SECRETORY SODIUM. CONGENITAL; DIAR8 [AR]
SLC9A3R1	NEPHROLITHIASIS/OSTEOPOROSIS. HYPOPHOSPHATEMIC. 2; NPHLOP2 [AD]
SLC9A6	MENTAL RETARDATION. X-LINKED. SYNDROMIC. CHRISTIANSON TYPE; MRXSCH [XLD]
SLC9A7	INTELLECTUAL DEVELOPMENTAL DISORDER. X-LINKED 108; MRX108 [XLR]
SLC9A9	{?Autism susceptibility 16} 613410 (3)
SLCO1B1	HYPERBILIRUBINEMIA. ROTOR TYPE; HBLRR [Digenic recessive]
SLCO1B3	HYPERBILIRUBINEMIA. ROTOR TYPE; HBLRR [Digenic recessive]
SLCO2A1	HYPERTROPHIC OSTEOARTHROPATHY. PRIMARY. AUTOSOMAL RECESSIVE. 2; PHOAR2 [AR]
SLFN14	BLEEDING DISORDER. PLATELET-TYPE. 20; BDPLT20 [AD]
SLITRK1	GILLES DE LA TOURETTE SYNDROME; GTS [AD] | TRICHOTILLOMANIA; TTM [AD]
SLITRK6	DEAFNESS AND MYOPIA; DFNMYP [AR]
SLURP1	MAL DE MELEDA; MDM [AR]
SLX4	FANCONI ANEMIA. COMPLEMENTATION GROUP P; FANCP [AR]
SMAD3	LOEYS-DIETZ SYNDROME 3; LDS3 [AD]
SMAD4	JUVENILE POLYPOSIS SYNDROME; JPS [AD] | PANCREATIC CANCER [AD] | JUVENILE POLYPOSIS/HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME; JPHT [AD] | MYHRE SYNDROME; MYHRS [AD]
SMAD6	AORTIC VALVE DISEASE 2; AOVD2 [AD] | CRANIOSYNOSTOSIS 7; CRS7 [Autosomal dominant (see qualifier below)]
SMAD9	PULMONARY HYPERTENSION. PRIMARY. 2; PPH2 [AD]
SMARCA2	NICOLAIDES-BARAITSER SYNDROME; NCBRS [AD]
SMARCA4	RHABDOID TUMOR PREDISPOSITION SYNDROME 2; RTPS2 [AD] | COFFIN-SIRIS SYNDROME 4; CSS4 [AD]
SMARCAD1	ADERMATOGLYPHIA; ADERM [AD] | HURIEZ SYNDROME; HRZ [AD] | BASAN SYNDROME [AD]
SMARCAL1	SCHIMKE IMMUNOOSSEOUS DYSPLASIA; SIOD [AR]
SMARCB1	COFFIN-SIRIS SYNDROME 3; CSS3 [AD] | RHABDOID TUMOR PREDISPOSITION SYNDROME 1; RTPS1 [AD] | SCHWANNOMATOSIS 1; SWNTS1 [AD]
SMARCC2	COFFIN-SIRIS SYNDROME 8; CSS8 [AD]
SMARCD2	SPECIFIC GRANULE DEFICIENCY 2; SGD2 [AR]
SMARCE1	MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD] | COFFIN-SIRIS SYNDROME 5; CSS5 [AD]
SMC1A	CORNELIA DE LANGE SYNDROME 2; CDLS2 [XLD]
SMC3	CORNELIA DE LANGE SYNDROME 3; CDLS3 [AD]

SMCHD1	BOSMA ARHINIA MICROPHTHALMIA SYNDROME; BAMS [AD]
SMG9	HEART AND BRAIN MALFORMATION SYNDROME; HBMS [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]
SMO	CURRY-JONES SYNDROME; CRJS [Somatic mosaicism]
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]
SMPD4	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. ARTHROGRYPOSIS. AND STRUCTURAL BRAIN ANOMALIES; NEDMABA [AR]
SMPX	DEAFNESS. X-LINKED 4; DFNX4 [XLD]
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]
SNCA	PARKINSON DISEASE 1. AUTOSOMAL DOMINANT; PARK1 [AD] | PARKINSON DISEASE 4. AUTOSOMAL DOMINANT; PARK4 [AD] | DEMENTIA. LEWY BODY; DLB [AD]
SNCB	DEMENTIA. LEWY BODY; DLB [AD]
SNIP1	PSYCHOMOTOR RETARDATION. EPILEPSY. AND CRANIOFACIAL DYSMORPHISM; PMRED [AR]
SNORD118	LEUKOENCEPHALOPATHY. BRAIN CALCIFICATIONS. AND CYSTS; LCC [AR]
SNRNP200	Retinitis pigmentosa 33 610359 (3)
SNRPB	CEREBROCOSTOMANDIBULAR SYNDROME; CCMS [AD]
SNRPE	HYPOTRICHOSIS 11; HYPT11 [AD]
SNRPN	PRADER-WILLI SYNDROME; PWS [Autosomal dominant (loss of paternal allele)]
SNTA1	LONG QT SYNDROME 12; LQT12 [AD]
SNX10	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 8; OPTB8 [AR]
SNX14	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 20; SCAR20 [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]
SOHLH1	SPERMATOGENIC FAILURE 32; SPGF32 [AD] | OVARIAN DYSGENESIS 5; ODG5 [AR]
SON	ZTTK SYNDROME; ZTTKS [AD]
SOS1	NOONAN SYNDROME 4; NS4 [AD] | FIBROMATOSIS. GINGIVAL. 1; GINGF1 [AD]
SOS2	NOONAN SYNDROME 9; NS9 [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]
SOX11	COFFIN-SIRIS SYNDROME 9; CSS9 [AD]
SOX17	VESICOURETERAL REFLUX 3; VUR3 [AD]
SOX18	HYPOTRICHOSIS-LYMPHEDEMA-TELANGIECTASIA-RENAL DEFECT SYNDROME; HLTRS [AD] | HYPOTRICHOSIS-LYMPHEDEMA-TELANGIECTASIA SYNDROME; HLTS [AR]
SOX2	MICROPHTHALMIA. SYNDROMIC 3; MCOPS3 [AD]
SOX3	46.XX SEX REVERSAL 3; SRXX3 [XLD] | PANHYPOPITUITARISM. X-LINKED; PHPX [X-linked]
SOX4	COFFIN-SIRIS SYNDROME 10; CSS10 [AD]
SOX5	LAMB-SHAFFER SYNDROME; LAMSHF [AD]
SOX9	CAMPOMELIC DYSPLASIA [AD] | 46.XY SEX REVERSAL 10; SRXY10 [AD] | 46.XX SEX REVERSAL 2; SRXX2 [AD]
SP110	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY; VODI [AR]
SP7	OSTEOGENESIS IMPERFECTA. TYPE XII; OI12 [AR]

SPAG1	CILIARY DYSKINESIA. PRIMARY. 28; CILD28 [AR]
SPARC	OSTEOGENESIS IMPERFECTA. TYPE XVII; OI17 [AR]
SPART	SPASTIC PARAPLEGIA 20. AUTOSOMAL RECESSIVE; SPG20 [AR]
SPAST	SPASTIC PARAPLEGIA 4. AUTOSOMAL DOMINANT; SPG4 [AD]
SPATA5	EPILEPSY. HEARING LOSS. AND MENTAL RETARDATION SYNDROME; EHLMRS [AR]
SPATA7	Leber congenital amaurosis 3 604232 (3) | Retinitis pigmentosa juvenile autosomal recessive 604232 (3)
SPECC1L	OPITZ GBBB SYNDROME. TYPE II; GBBB2 [AD] | HYPERTELORISM. TEEBI TYPE; TBHS [AD] | FACIAL CLEFTING. OBLIQUE. 1; OBLFC1 [AD]
SPEG	MYOPATHY. CENTRONUCLEAR. 5; CNM5 [AR]
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]
SPRED1	LEGIUS SYNDROME; LGSS [AD]
SPRTN	RUIJS-AALFS SYNDROME; RJALS [AR]
SPRY2	IgA NEPHROPATHY. SUSCEPTIBILITY TO. 3; IGAN3 [AD]
SPRY4	HYPOGONADOTROPIC HYPOGONADISM 17 WITH OR WITHOUT ANOSMIA; HH17 [Autosomal dominant (see MISCELLANEOUS below)]
SPTA1	PYROPOIKILOCYTOSIS. HEREDITARY; HPP [AR] | ELLIPTOCYTOSIS 2; EL2 [AD] | SPHEROCYTOSIS. TYPE 3; SPH3 [AR]
SPTAN1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 5; EIEE5 [AD]
SPTB	SPHEROCYTOSIS. TYPE 2; SPH2 [AD]
SPTBN2	SPINOCEREBELLAR ATAXIA 5; SCA5 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 14; SCAR14 [AR]
SPTBN4	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA. NEUROPATHY. AND DEAFNESS; NEDHND [AR]
SPTLC1	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE IA; HSAN1A [AD]
SPTLC2	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE IC; HSAN1C [AD]
SQSTM1	PAGET DISEASE OF BONE 3; PDB3 [AD] | FRONTOTEMPORAL DEMENTIA AND/OR AMYOTROPHIC LATERAL SCLEROSIS 3; FTDALS3 [AD] | MYOPATHY. DISTAL. WITH RIMMED VACUOLES; DMRV [AD] | NEURODEGENERATION WITH ATAXIA. DYSTONIA. AND GAZE PALSY. CHILDHOOD-ONSET; NADGP [AR]
SRC	THROMBOCYTOPENIA 6; THC6 [AD] | COLORECTAL CANCER; CRC [Somatic mutation]
SRCAP	FLOATING-HARBOR SYNDROME; FLHS [AD]
SRD5A2	PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS; PPSH [AR]
SRD5A3	KAHRIZI SYNDROME; KHRZ [AR] | CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Iq; CDG1Q [AR]
SRP72	BONE MARROW FAILURE SYNDROME 1; BMFS1 [AD]
SRY	46.XY SEX REVERSAL 1; SRXY1 [Y-linked] | 46.XX SEX REVERSAL 1; SRXX1 [XLD]
SS18	Sarcoma synovial (1)
SSR4	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE Iy; CDG1Y [XLR]
SSTR5	Somatostatin analog resistance to (3)
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]
STAG1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 47; MRD47 [AD]
STAG2	MULLEGAMA-KLEIN-MARTINEZ SYNDROME; MKMS [X-linked]

STAG3	PREMATURE OVARIAN FAILURE 8; POF8 [AR]
STAMBP	MICROCEPHALY-CAPILLARY MALFORMATION SYNDROME; MICCAP [AR]
STAR	LIPOID CONGENITAL ADRENAL HYPERPLASIA; LCAH [AR]
STARD7	EPILEPSY. FAMILIAL ADULT MYOCLONIC. 2; FAME2 [AD]
STAT1	IMMUNODEFICIENCY 31C; IMD31C [AD] | IMMUNODEFICIENCY 31B; IMD31B [AR] | IMMUNODEFICIENCY 31A; IMD31A [AD]
STAT2	IMMUNODEFICIENCY 44; IMD44 [AR]
STAT3	HYPER-IgE RECURRENT INFECTION SYNDROME 1. AUTOSOMAL DOMINANT; HIES1 [AD] | AUTOIMMUNE DISEASE. MULTISYSTEM. INFANTILE-ONSET. 1; ADMIO1 [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]
STING1	STING-ASSOCIATED VASCULOPATHY. INFANTILE-ONSET; SAVI [AD]
STK11	PEUTZ-JEGHERS SYNDROME; PJS [AD] | TESTICULAR GERM CELL TUMOR; TGCT [Somatic mutation] | PANCREATIC CANCER [AD]
STK4	T-cell immunodeficiency recurrent infections autoimmunity and cardiac malformations 614868 (3)
STN1	CEREBRORETINAL MICROANGIOPATHY WITH CALCIFICATIONS AND CYSTS 2; CRMCC2 [AR]
STOX1	Preeclampsia/eclampsia 4 609404 (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]
STUB1	SPINOCEREBELLAR ATAXIA 48; SCA48 [AD] | SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 16; SCAR16 [AR]
STX11	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 4; FHL4 [AR]
STX16	PSEUDOHYPOPARATHYROIDISM. TYPE IB; PHP1B [AD]
STX1B	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS. TYPE 9; GEFSP9 [AD]
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]
SUFU	BASAL CELL NEVUS SYNDROME; BCNS [AD] | MENINGIOMA. FAMILIAL. SUSCEPTIBILITY TO [AD] | MEDULLOBLASTOMA; MDB [Somatic mutation] | JOUBERT SYNDROME 32; JBTS32 [AR]
SUGCT	GLUTARIC ACIDURIA III; GA3 [AR]
SULT2B1	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 14; ARCI14 [AR]
SUMF1	MULTIPLE SULFATASE DEFICIENCY; MSD [AR]
SUN5	SPERMATOGENIC FAILURE 16; SPGF16 [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]
SVBP	NEURODEVELOPMENTAL DISORDER WITH ATAXIA. HYPOTONIA. AND MICROCEPHALY; NEDAHM [AR]
SYCP3	SPERMATOGENIC FAILURE 4; SPGF4 [AD]
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]
SYNE2	EMERY-DREIFUSS MUSCULAR DYSTROPHY 5. AUTOSOMAL DOMINANT; EDMD5 [AD]
SYNE4	DEAFNESS. AUTOSOMAL RECESSIVE 76; DFNB76 [AR]
SYNGAP1	MENTAL RETARDATION. AUTOSOMAL DOMINANT 5; MRD5 [AD]

SYNJ1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 53; EIEE53 [AR] | PARKINSON DISEASE 20. EARLY-ONSET; PARK20 [AR]
SYP	MENTAL RETARDATION. X-LINKED 96; MRX96 [XLR]
SYT1	BAKER-GORDON SYNDROME; BAGOS [AD]
SYT2	MYASTHENIC SYNDROME. CONGENITAL. 7. PRESYNAPTIC; CMS7 [AD]
SZT2	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 18; EIEE18 [AR]
TAB2	CONGENITAL HEART DEFECTS. MULTIPLE TYPES. 2; CHTD2 [AD]
TAC3	HYPOGONADOTROPIC HYPOGONADISM 10 WITH OR WITHOUT ANOSMIA; HH10 [AR]
TACO1	LEIGH SYNDROME; LS [AR] | MITOCHONDRIAL COMPLEX IV DEFICIENCY [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]
TAF13	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 60; MRT60 [AR]
TAF15	Chondrosarcoma extraskeletal myxoid 612237 (1)
TAF2	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 40; MRT40 [AR]
TAF6	ALAZAMI-YUAN SYNDROME; ALYUS [AR]
TAL1	Leukemia T-cell acute lymphocytic somatic 613065 (3)
TAL2	Leukemia T-cell acute lymphocytic somatic 613065 (3)
TALDO1	TRANSALDOLASE DEFICIENCY [AR]
TANGO2	METABOLIC CRISES. RECURRENT. WITH RHABDOMYOLYSIS. CARDIAC ARRHYTHMIAS. AND NEURODEGENERATION; MECRCN [AR]
TAP1	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAP2	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAPBP	BARE LYMPHOCYTE SYNDROME. TYPE I [AR]
TAPT1	OSTEOCHONDRODYSPLASIA. COMPLEX LETHAL. SYMOENS-BARNES-GISTELINCK TYPE; OCLSBG [AR]
TARDBP	AMYOTROPHIC LATERAL SCLEROSIS 10 WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA; ALS10 [AD]
TARS1	TRICHOTHIODYSTROPHY 7. NONPHOTOSENSITIVE; TTD7 [AR]
TAS2R16	BETA-GLUCOPYRANOSIDE TASTING; BGLPT [AD]
TAT	TYROSINEMIA. TYPE II; TYRSN2 [AR]
TAZ	BARTH SYNDROME; BTHS [XLR]
TBC1D20	WARBURG MICRO SYNDROME 4; WARBM4 [AR]
TBC1D23	PONTOCEREBELLAR HYPOPLASIA. TYPE 11; PCH11 [AR]
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]
TBC1D7	MACROCEPHALY/MEGALENCEPHALY SYNDROME. AUTOSOMAL RECESSIVE; MGCPH [AR]
TBC1D8B	NEPHROTIC SYNDROME. TYPE 20; NPHS20 [X-linked]
TBCD	ENCEPHALOPATHY. PROGRESSIVE. EARLY-ONSET. WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM; PEBAT [AR]
TBCE	ENCEPHALOPATHY. PROGRESSIVE. WITH AMYOTROPHY AND OPTIC ATROPHY; PEAMO [AR] | KENNY-CAFFEY SYNDROME. TYPE 1; KCS1 [AR] | HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME; HRDS [AR]
TBCK	HYPOTONIA. INFANTILE. WITH PSYCHOMOTOR RETARDATION AND CHARACTERISTIC FACIES 3; IHPRF3 [AR]
TBK1	FRONTOTEMPORAL DEMENTIA AND/OR AMYOTROPHIC LATERAL SCLEROSIS 4; FTDALS4 [AD] | ENCEPHALOPATHY. ACUTE. INFECTION-INDUCED (HERPES-SPECIFIC). SUSCEPTIBILITY TO. 8; IIAE8 [AD]
TBL1X	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 8; CHNG8 [X-linked]
TBL1XR1	PIERPONT SYNDROME; PRPTS [AD] | MENTAL RETARDATION. AUTOSOMAL DOMINANT 41; MRD41 [AD]
TBP	SPINOCEREBELLAR ATAXIA 17; SCA17 [AD] | PARKINSON DISEASE. LATE-ONSET; PD [AD]

TBR1	INTELLECTUAL DEVELOPMENTAL DISORDER WITH AUTISM AND SPEECH DELAY; IDDAS [AD]
TBX1	DIGEORGE SYNDROME; DGS [AD] | TETRALOGY OF FALLOT; TOF [AD] | VELOCARDIOFACIAL SYNDROME [AD]
TBX15	COUSIN SYNDROME [AR]
TBX18	CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT 2; CAKUT2 [AD]
TBX19	ACTH DEFICIENCY. ISOLATED; IAD [AR]
TBX2	VERTEBRAL ANOMALIES AND VARIABLE ENDOCRINE AND T-CELL DYSFUNCTION; VETD [AD]
TBX20	Atrial septal defect 4 611363 (3)
TBX21	ASTHMA. NASAL POLYPS. AND ASPIRIN INTOLERANCE [AR]
TBX22	CLEFT PALATE WITH OR WITHOUT ANKYLOGLOSSIA. X-LINKED; CPX [X-linked] | ABRUZZO-ERICKSON SYNDROME; ABERS [X-linked]
TBX3	ULNAR-MAMMARY SYNDROME; UMS [AD]
TBX4	ISCHIOCOXOPODOPATELLAR SYNDROME WITH OR WITHOUT PULMONARY ARTERIAL HYPERTENSION; ICPPS [AD] | AMELIA. POSTERIOR. WITH PELVIC AND PULMONARY HYPOPLASIA SYNDROME; PAPPAS [AR]
TBX5	HOLT-ORAM SYNDROME; HOS [AD]
TBX6	SPONDYLOCOSTAL DYSOSTOSIS 5; SCDO5 [AR]
TBXAS1	GHOSAL HEMATODIAPHYSEAL DYSPLASIA; GHDD [AR]
TBXT	NEURAL TUBE DEFECTS. SUSCEPTIBILITY TO; NTD [AD] | SACRAL AGENESIS WITH VERTEBRAL ANOMALIES; SAVA [AR]
TCAP	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 25; CMH25 [AD] | MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 7; LGMDR7 [AR]
TCF12	CRANIOSYNOSTOSIS 3; CRS3 [AD]
TCF20	DEVELOPMENTAL DELAY WITH VARIABLE INTELLECTUAL IMPAIRMENT AND BEHAVIORAL ABNORMALITIES; DDVIBA [AD]
TCF3	AGAMMAGLOBULINEMIA 8. AUTOSOMAL DOMINANT; AGM8 [AD]
TCF4	CORNEAL DYSTROPHY. FUCHS ENDOTHELIAL. 3; FECD3 [AD] | PITT-HOPKINS SYNDROME; PTHS [AD]
TCIRG1	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 1; OPTB1 [AR]
TCL1A	Leukemia/lymphoma T-cell (2)
TCL1B	Leukemia/lymphoma T-cell (2)
TCN2	TRANSCOBALAMIN II DEFICIENCY [AR]
TCOF1	TREACHER COLLINS SYNDROME 1; TCS1 [AD]
TCTEX1D2	SHORT-RIB THORACIC DYSPLASIA 17 WITH OR WITHOUT POLYDACTYLY; SRTD17 [AR]
TCTN1	JOUBERT SYNDROME 13; JBTS13 [AR]
TCTN2	JOUBERT SYNDROME 24; JBTS24 [AR] | MECKEL SYNDROME. TYPE 8; MKS8 [AR]
TCTN3	JOUBERT SYNDROME 18; JBTS18 [AR] | OROFACIODIGITAL SYNDROME IV; OFD4 [AR]
TDGF1	Forebrain defects (3)
TDO2	HYPERTRYPTOPHANEMIA; HYPTRP [AR]
TDP2	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 23; SCAR23 [AR]
TDRD7	CATARACT 36; CTRCT36 [AR]
TEAD1	SVEINSSON CHORIORETINAL ATROPHY; SCRA [AD]
TECPR2	SPASTIC PARAPLEGIA 49. AUTOSOMAL RECESSIVE; SPG49 [AR]
TECR	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 14; MRT14 [AR]
TECRL	VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 3; CPVT3 [AR]
TECTA	DEAFNESS. AUTOSOMAL DOMINANT 12; DFNA12 [AD] | DEAFNESS. AUTOSOMAL RECESSIVE 21; DFNB21 [AR]
TEK	VENOUS MALFORMATIONS. MULTIPLE CUTANEOUS AND MUCOSAL; VMCM [AD] | GLAUCOMA 3. PRIMARY CONGENITAL. E; GLC3E [AD]
TELO2	YOU-HOOVER-FONG SYNDROME; YHFS [AR]
TENM3	MICROPHTHALMIA. ISOLATED. WITH COLOBOMA 9; MCOPCB9 [AR]
TENM4	TREMOR. HEREDITARY ESSENTIAL. 5; ETM5 [AD]

TENT5A	OSTEOGENESIS IMPERFECTA. TYPE XVIII; OI18 [AR]
TERC	DYSKERATOSIS CONGENITA. AUTOSOMAL DOMINANT 1; DKCA1 [AD] | PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE. TELOMERE-RELATED. 2; PFBMFT2 [AD]
TET2	Myelodysplastic syndrome somatic 614286 (3)
TEX11	SPERMATOGENIC FAILURE. X-LINKED. 2; SPGFX2 [XLR]
TEX15	SPERMATOGENIC FAILURE 25; SPGF25 [AR]
TF	Atransferrinemia 209300 (3)
TFAP2A	BRANCHIOOCULOFACIAL SYNDROME; BOFS [AD]
TFAP2B	PATENT DUCTUS ARTERIOSUS 2; PDA2 [AD] | CHAR SYNDROME; CHAR [AD]
TFE3	Renal cell carcinoma papillary 1 300854 (3)
TFG	NEUROPATHY. HEREDITARY MOTOR AND SENSORY. OKINAWA TYPE; HMSNO [AD] | SPASTIC PARAPLEGIA 57. AUTOSOMAL RECESSIVE; SPG57 [AR]
TFR2	HEMOCHROMATOSIS. TYPE 3; HFE3 [AR]
TFRC	IMMUNODEFICIENCY 46; IMD46 [AR]
TG	Thyroid dyshormonogenesis 3 274700 (3) | {Autoimmune thyroid disease susceptibility to 3} 608175 (3)
TGDS	CATEL-MANZKE SYNDROME; CATMANS [AR]
TGFB1	INFLAMMATORY BOWEL DISEASE. IMMUNODEFICIENCY. AND ENCEPHALOPATHY; IBDIMDE [AR] | CYSTIC FIBROSIS; CF [AR] | CAMURATI-ENGELMANN DISEASE; CAEND [AD]
TGFB2	LOEYS-DIETZ SYNDROME 4; LDS4 [AD]
TGFB3	LOEYS-DIETZ SYNDROME 5; LDS5 [AD] | ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 1; ARVD1 [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]
TGFBR1	MULTIPLE SELF-HEALING SQUAMOUS EPITHELIOMA. SUSCEPTIBILITY TO; MSSE [AD] | LOEYS-DIETZ SYNDROME 1; LDS1 [AD]
TGFBR2	LOEYS-DIETZ SYNDROME 2; LDS2 [AD] | ESOPHAGEAL CANCER [Somatic mutation]
TGIF1	HOLOPROSENCEPHALY 4; HPE4 [AD]
TGM1	ICHTHYOSIS. CONGENITAL. AUTOSOMAL RECESSIVE 1; ARCI1 [AR]
TGM5	PEELING SKIN SYNDROME 2; PSS2 [AR]
TGM6	SPINOCEREBELLAR ATAXIA 35; SCA35 [AD]
TH	Segawa syndrome recessive 605407 (3)
THAP1	DYSTONIA 6. TORSION; DYT6 [AD]
THBD	HEMOLYTIC UREMIC SYNDROME. ATYPICAL. SUSCEPTIBILITY TO. 6; AHUS6 [AD]
THOC2	MENTAL RETARDATION. X-LINKED 12; MRX12 [XLR]
THOC6	BEAULIEU-BOYCOTT-INNES SYNDROME; BBIS [AR]
THPO	THROMBOCYTHEMIA 1; THCYT1 [AD]
THRA	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 6; CHNG6 [AD]
THRB	THYROID HORMONE RESISTANCE. SELECTIVE PITUITARY; PRTH [AD] | THYROID HORMONE RESISTANCE. GENERALIZED. AUTOSOMAL RECESSIVE; GRTH [AR] | THYROID HORMONE RESISTANCE. GENERALIZED. AUTOSOMAL DOMINANT; GRTH [AD]
TIA1	WELANDER DISTAL MYOPATHY; WDM [AD]
TIMM50	3-METHYLGLUTACONIC ACIDURIA. TYPE IX; MGCA9 [AR]
TIMM8A	MOHR-TRANEBJAERG SYNDROME; MTS [XLR]
TIMMDC1	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 31; MC1DN31 [AR]
TIMP3	SORSBY FUNDUS DYSTROPHY; SFD [AD]
TINF2	DYSKERATOSIS CONGENITA. AUTOSOMAL DOMINANT 3; DKCA3 [AD] | REVESZ SYNDROME [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]

TKT	SHORT STATURE. DEVELOPMENTAL DELAY. AND CONGENITAL HEART DEFECTS; SDDHD [AR]
TLE6	PREIMPLANTATION EMBRYONIC LETHALITY 1; PREMBL1 [AR]
TLK2	MENTAL RETARDATION. AUTOSOMAL DOMINANT 57; MRD57 [AD]
TLL1	ATRIAL SEPTAL DEFECT 6; ASD6 [AD]
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]
TMEM106B	LEUKODYSTROPHY. HYPOMYELINATING. 16; HLD16 [AD]
TMEM107	MECKEL SYNDROME 13; MKS13 [AR] | OROFACIODIGITAL SYNDROME XVI; OFD16 [AR]
TMEM126A	OPTIC ATROPHY 7 WITH OR WITHOUT AUDITORY NEUROPATHY; OPA7 [AR]
TMEM126B	MITOCHONDRIAL COMPLEX I DEFICIENCY. NUCLEAR TYPE 29; MC1DN29 [AR]
TMEM138	JOUBERT SYNDROME 16; JBTS16 [AR]
TMEM165	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIk; CDG2K [AR]
TMEM199	CONGENITAL DISORDER OF GLYCOSYLATION. TYPE IIp; CDG2P [AR]
TMEM216	JOUBERT SYNDROME 2; JBTS2 [AR] | MECKEL SYNDROME. TYPE 2; MKS2 [AR]
TMEM231	JOUBERT SYNDROME 20; JBTS20 [AR] | MECKEL SYNDROME. TYPE 11; MKS11 [AR]
TMEM237	JOUBERT SYNDROME 14; JBTS14 [AR]
TMEM240	SPINOCEREBELLAR ATAXIA 21; SCA21 [AD]
TMEM260	STRUCTURAL HEART DEFECTS AND RENAL ANOMALIES SYNDROME; SHDRA [AR]
TMEM38B	Osteogenesis imperfecta type XIV 615066 (3)
TMEM43	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA. FAMILIAL. 5; ARVD5 [AD] | EMERY-DREIFUSS MUSCULAR DYSTROPHY 7. AUTOSOMAL DOMINANT; EDMD7 [AD]
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]
TMEM94	INTELLECTUAL DEVELOPMENTAL DISORDER WITH CARDIAC DEFECTS AND DYSMORPHIC FACIES; IDDCDF [AR]
TMEM98	NANOPHTHALMOS 4; NNO4 [AD]
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]
TMTC3	LISSENCEPHALY 8; LIS8 [AR]
TNC	DEAFNESS. AUTOSOMAL DOMINANT 56; DFNA56 [AD]
TNFAIP3	AUTOINFLAMMATORY SYNDROME. FAMILIAL. BEHCET-LIKE; AISBL [AD]
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]
TNFRSF1A	PERIODIC FEVER. FAMILIAL. AUTOSOMAL DOMINANT; FPF [AD]
TNFSF11	OSTEOPETROSIS. AUTOSOMAL RECESSIVE 2; OPTB2 [AR]
TNIK	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 54; MRT54 [AR]

TNNC1	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 13; CMH13 [AD]
TNNI2	ARTHROGRYPOSIS. DISTAL. TYPE 2B1; DA2B1 [AD]
TNNI3	CARDIOMYOPATHY. FAMILIAL RESTRICTIVE. 1; RCM1 [AD] | CARDIOMYOPATHY. DILATED. 2A; CMD2A [AR] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 7; CMH7 [AD]
TNNI3K	CARDIAC CONDUCTION DISEASE WITH OR WITHOUT DILATED CARDIOMYOPATHY; CCDD [AD]
TNNT1	NEMALINE MYOPATHY 5; NEM5 [AR]
TNNT2	CARDIOMYOPATHY. FAMILIAL RESTRICTIVE. 3; RCM3 [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 2; CMH2 [AD] | CARDIOMYOPATHY. DILATED. 1D; CMD1D [AD]
TNNT3	ARTHROGRYPOSIS. DISTAL. TYPE 2B2; DA2B2 [AD] | ARTHROGRYPOSIS. DISTAL. TYPE 2B1; DA2B1 [AD]
TNPO3	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL DOMINANT 2; LGMDD2 [AD]
TNXB	EHLERS-DANLOS SYNDROME. HYPERMOBILITY TYPE; EDSHMB [AD] | VESICOURETERAL REFLUX 8; VUR8 [AD] | EHLERS-DANLOS SYNDROME. CLASSIC-LIKE; EDSCLL [AR]
TOE1	PONTOCEREBELLAR HYPOPLASIA. TYPE 7; PCH7 [AR]
TONSL	SPONDYLOEPIMETAPHYSEAL DYSPLASIA. SPONASTRIME TYPE; SEMDSP [AR]
TOP1	DNA topoisomerase I camptothecin-resistant (3)
TOP2A	DNA topoisomerase II resistance to inhibition of by amsacrine (3)
TOP3A	MICROCEPHALY. GROWTH RESTRICTION. AND INCREASED SISTER CHROMATID EXCHANGE 2; MGRISCE2 [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL RECESSIVE 5; PEOB5 [AR]
TOPORS	Retinitis pigmentosa 31 609923 (3)
TOR1A	DYSTONIA 1. TORSION. AUTOSOMAL DOMINANT; DYT1 [AD]
TP53	GLIOMA SUSCEPTIBILITY 1; GLM1 [AD] | LI-FRAUMENI SYNDROME; LFS [AD] | COLORECTAL CANCER; CRC [Somatic mutation] | BREAST CANCER [AD] | ADRENOCORTICAL CARCINOMA. HEREDITARY; ADCC [AD] | BONE MARROW FAILURE SYNDROME 5; BMFS5 [AD] | OSTEOGENIC SARCOMA [Somatic mutation] | PANCREATIC CANCER [AD] | BASAL CELL CARCINOMA. SUSCEPTIBILITY TO. 7; BCC7 [AD] | HEPATOCELLULAR CARCINOMA [Somatic mutation] | PAPILLOMA OF CHOROID PLEXUS; CPP [AD]
TP53RK	GALLOWAY-MOWAT SYNDROME 4; GAMOS4 [AR]
TP63	LIMB-MAMMARY SYNDROME; LMS [AD] | ECTRODACTYLY. ECTODERMAL DYSPLASIA. AND CLEFT LIP/PALATE SYNDROME 3; EEC3 [AD] | ADULT SYNDROME [AD] | ANKYLOBLEPHARON-ECTODERMAL DEFECTS-CLEFT LIP/PALATE; AEC [AD] | RAPP-HODGKIN SYNDROME; RHS [AD] | SPLIT-HAND/FOOT MALFORMATION 4; SHFM4 [AD]
TPI1	TRIOSEPHOSPHATE ISOMERASE DEFICIENCY; TPID [AR]
TPK1	THIAMINE METABOLISM DYSFUNCTION SYNDROME 5 (EPISODIC ENCEPHALOPATHY TYPE); THMD5 [AR]
TPM1	CARDIOMYOPATHY. DILATED. 1Y; CMD1Y [AD] | CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 3; CMH3 [AD]
TPM2	NEMALINE MYOPATHY 4; NEM4 [AD] | ARTHROGRYPOSIS. DISTAL. TYPE 1A; DA1A [AD] | ARTHROGRYPOSIS. DISTAL. TYPE 2B1; DA2B1 [AD]
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]
TPRKB	GALLOWAY-MOWAT SYNDROME 5; GAMOS5 [AR]
TPRN	DEAFNESS. AUTOSOMAL RECESSIVE 79; DFNB79 [AR]
TRAC	T-CELL RECEPTOR-ALPHA/BETA DEFICIENCY [AR]
TRAF3	{?Encephalopathy acute infection-induced (herpes-specific) susceptibility to 5} 614849 (3)
TRAF3IP1	SENIOR-LOKEN SYNDROME 9; SLSN9 [AR]
TRAF7	CARDIAC. FACIAL. AND DIGITAL ANOMALIES WITH DEVELOPMENTAL DELAY; CAFDADD [AD]
TRAIP	SECKEL SYNDROME 9; SCKL9 [AR]
TRAK1	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 68; EIEE68 [AR]
TRAPPC11	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 18; LGMDR18 [AR]
TRAPPC12	ENCEPHALOPATHY. PROGRESSIVE. EARLY-ONSET. WITH BRAIN ATROPHY AND SPASTICITY; PEBAS [AR]
TRAPPC2	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA. X-LINKED; SEDT [XLR]
TRAPPC2L	ENCEPHALOPATHY. PROGRESSIVE. EARLY-ONSET. WITH EPISODIC RHABDOMYOLYSIS; PEERB [AR]

TRAPPC6B	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. EPILEPSY. AND BRAIN ATROPHY; NEDMEBA [AR]
TRAPPC9	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 13; MRT13 [AR]
TRDN	VENTRICULAR TACHYCARDIA. CATECHOLAMINERGIC POLYMORPHIC. 5. WITH OR WITHOUT MUSCLE WEAKNESS; CPVT5 [AR]
TREH	TREHALASE DEFICIENCY [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]
TRH	THYROTROPIN-RELEASING HORMONE DEFICIENCY [AR]
TRHR	HYPOTHYROIDISM. CONGENITAL. NONGOITROUS. 7; CHNG7 [AR]
TRIM2	CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2R; CMT2R [AR]
TRIM32	MUSCULAR DYSTROPHY. LIMB-GIRDLE. AUTOSOMAL RECESSIVE 8; LGMDR8 [AR] | BARDET-BIEDL SYNDROME 11; BBS11 [AR]
TRIM37	MULIBREY NANISM [AR]
TRIM71	HYDROCEPHALUS. CONGENITAL COMMUNICATING. 1; HYDCC1 [AD]
TRIO	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL DOMINANT 44. WITH MICROCEPHALY; MRD44 [AD]
TRIOBP	DEAFNESS. AUTOSOMAL RECESSIVE 28; DFNB28 [AR]
TRIP11	ACHONDROGENESIS. TYPE IA; ACG1A [AR] | ODONTOCHONDRODYSPLASIA; ODCD [AR]
TRIP12	MENTAL RETARDATION. AUTOSOMAL DOMINANT 49; MRD49 [AD]
TRIP13	MOSAIC VARIEGATED ANEUPLOIDY SYNDROME 3; MVA3 [AR]
TRIP4	MUSCULAR DYSTROPHY. CONGENITAL. DAVIGNON-CHAUVEAU TYPE; MDCDC [AR] | SPINAL MUSCULAR ATROPHY WITH CONGENITAL BONE FRACTURES 1; SMABF1 [AR]
TRIT1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 35; COXPD35 [AR]
TRMT1	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 68; MRT68 [AR]
TRMT10A	MICROCEPHALY. SHORT STATURE. AND IMPAIRED GLUCOSE METABOLISM 1; MSSGM1 [AR]
TRMT10C	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 30; COXPD30 [AR]
TRMT5	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 26; COXPD26 [AR]
TRMU	LIVER FAILURE. INFANTILE. TRANSIENT; LFIT [AR] | DEAFNESS. AMINOGLYCOSIDE-INDUCED [Mitochondrial]
TRNT1	SIDEROBLASTIC ANEMIA WITH B-CELL IMMUNODEFICIENCY. PERIODIC FEVERS. AND DEVELOPMENTAL DELAY; SIFD [AR] | RETINITIS PIGMENTOSA AND ERYTHROCYTIC MICROCYTOSIS; RPEM [AR]
TRPC6	FOCAL SEGMENTAL GLOMERULOSCLEROSIS 2; FSGS2 [AD]
TRPM1	Night blindness congenital stationary (complete) 1C autosomal recessive 613216 (3)
TRPM4	PROGRESSIVE FAMILIAL HEART BLOCK. TYPE IB; PFHB1B [AD] | ERYTHROKERATODERMIA VARIABILIS ET PROGRESSIVA 6; EKVP6 [AD]
TRPM6	HYPOMAGNESEMIA 1. INTESTINAL; HOMG1 [AR]
TRPS1	TRICHORHINOPHALANGEAL SYNDROME. TYPE III; TRPS3 [AD] | TRICHORHINOPHALANGEAL SYNDROME. TYPE I; TRPS1 [AD] | TRICHORHINOPHALANGEAL SYNDROME. TYPE II; TRPS2 [AD]
TRPV3	PALMOPLANTAR KERATODERMA. MUTILATING. WITH PERIORIFICIAL KERATOTIC PLAQUES [AD] | PALMOPLANTAR KERATODERMA. NONEPIDERMOLYTIC. FOCAL 2; FNEPPK2 [AD]
TRPV4	AVASCULAR NECROSIS OF FEMORAL HEAD. PRIMARY. 2; ANFH2 [AD] | PARASTREMMATIC DWARFISM [AD] | HEREDITARY MOTOR AND SENSORY NEUROPATHY. TYPE IIC; HMSN2C [AD] | METATROPIC DYSPLASIA [AD] | BRACHYOLMIA TYPE 3; BCYM3 [AD] | DIGITAL ARTHROPATHY-BRACHYDACTYLY. FAMILIAL; FDAB [AD] | SPONDYLOEPIPHYSEAL DYSPLASIA. MAROTEAUX TYPE [AD] | SCAPULOPERONEAL SPINAL MUSCULAR ATROPHY; SPSMA [AD] | NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE VIII; HMN8 [AD] | SPONDYLOMETAPHYSEAL DYSPLASIA. KOZLOWSKI TYPE; SMDK [AD]
TRPV6	HYPERPARATHYROIDISM. TRANSIENT NEONATAL; HRPTTN [AR]
TRRAP	DEAFNESS. AUTOSOMAL DOMINANT 75; DFNA75 [AD] | DEVELOPMENTAL DELAY WITH OR WITHOUT DYSMORPHIC FACIES AND AUTISM; DEDDFA [AD]
TSC1	FOCAL CORTICAL DYSPLASIA. TYPE II; FCORD2 [Somatic mutation] | TUBEROUS SCLEROSIS 1; TSC1 [AD]
TSC2	FOCAL CORTICAL DYSPLASIA. TYPE II; FCORD2 [Somatic mutation] | TUBEROUS SCLEROSIS 2; TSC2 [AD]
TSEN15	PONTOCEREBELLAR HYPOPLASIA. TYPE 2F; PCH2F [AR]
TSEN2	PONTOCEREBELLAR HYPOPLASIA. TYPE 2B; PCH2B [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]
TSHZ1	AURAL ATRESIA. CONGENITAL; CAA [AD]
TSPAN12	EXUDATIVE VITREORETINOPATHY 5; EVR5 [AD]
TSPAN7	MENTAL RETARDATION. X-LINKED 58; MRX58 [XLR]
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]
TTBK2	SPINOCEREBELLAR ATAXIA 11; SCA11 [AD]
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 2; MC3DN2 [AR]
TTC21A	SPERMATOGENIC FAILURE 37; SPGF37 [AR]
TTC21B	NEPHRONOPHTHISIS 12; NPHP12 [AD] | SHORT-RIB THORACIC DYSPLASIA 4 WITH OR WITHOUT POLYDACTYLY; SRTD4 [AR]
TTC25	CILIARY DYSKINESIA. PRIMARY. 35; CILD35 [AR]
TTC37	TRICHOHEPATOENTERIC SYNDROME 1; THES1 [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]
TTLL5	CONE-ROD DYSTROPHY 19; CORD19 [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]
TTR	HYPERTHYROXINEMIA. DYSTRANSTHYRETINEMIC; DTTRH [AD] | CARPAL TUNNEL SYNDROME; CTS1 [AD] | AMYLOIDOSIS. HEREDITARY. TRANSTHYRETIN-RELATED [AD]
TUBA1A	LISSENCEPHALY 3; LIS3 [AD]
TUBA3D	KERATOCONUS 9; KTCN9 [AD]
TUBA4A	AMYOTROPHIC LATERAL SCLEROSIS 22 WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA; ALS22 [AD]
TUBA8	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 8; CDCBM8 [AR]
TUBB	SKIN CREASES. CONGENITAL SYMMETRIC CIRCUMFERENTIAL. 1; CSCSC1 [AD] | CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 6; CDCBM6 [AD]
TUBB1	MACROTHROMBOCYTOPENIA. AUTOSOMAL DOMINANT. TUBB1-RELATED [AD]
TUBB2A	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 5; CDCBM5 [AD]
TUBB2B	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 7; CDCBM7 [AD]
TUBB3	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 1; CDCBM1 [AD] | FIBROSIS OF EXTRAOCULAR MUSCLES. CONGENITAL. 3A. WITH OR WITHOUT EXTRAOCULAR INVOLVEMENT; CFEOM3A [AD]
TUBB4A	DYSTONIA 4. TORSION. AUTOSOMAL DOMINANT; DYT4 [AD] | LEUKODYSTROPHY. HYPOMYELINATING. 6; HLD6 [AD]
TUBB4B	LEBER CONGENITAL AMAUROSIS WITH EARLY-ONSET DEAFNESS; LCAEOD [AD]
TUBB8	OOCYTE MATURATION DEFECT 2; OOMD2 [AD]
TUBG1	CORTICAL DYSPLASIA. COMPLEX. WITH OTHER BRAIN MALFORMATIONS 4; CDCBM4 [AD]
TUBGCP4	MICROCEPHALY AND CHORIORETINOPATHY. AUTOSOMAL RECESSIVE. 3; MCCRP3 [AR]
TUBGCP6	MICROCEPHALY AND CHORIORETINOPATHY. AUTOSOMAL RECESSIVE. 1; MCCRP1 [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]

TWIST1	ROBINOW-SORAUF SYNDROME [AD] | CRANIOSYNOSTOSIS 1; CRS1 [AD] | SAETHRE-CHOTZEN SYNDROME; SCS [AD] | SWEENEY-COX SYNDROME; SWCOS [AD]
TWIST2	BARBER-SAY SYNDROME; BBRSAY [AD] | FOCAL FACIAL DERMAL DYSPLASIA 3. SETLEIS TYPE; FFDD3 [AR] | ABLEPHARON-MACROSTOMIA SYNDROME; AMS [AD]
TWNK	MITOCHONDRIAL DNA DEPLETION SYNDROME 7 (HEPATOCEREBRAL TYPE); MTDPS7 [AR] | PERRAULT SYNDROME 5; PRLTS5 [AR] | PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS. AUTOSOMAL DOMINANT 3; PEOA3 [AD]
TXNL4A	BURN-MCKEOWN SYNDROME; BMKS [AR]
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]
UBA5	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 24; SCAR24 [AR] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 44; EIEE44 [AR]
UBAP1	SPASTIC PARAPLEGIA 80. AUTOSOMAL DOMINANT; SPG80 [AD]
UBE2A	MENTAL RETARDATION. X-LINKED. SYNDROMIC. NASCIMENTO TYPE; MRXSN [XLR]
UBE2T	FANCONI ANEMIA. COMPLEMENTATION GROUP T; FANCT [AR]
UBE3A	ANGELMAN SYNDROME; AS [Autosomal dominant (loss of maternal allele)]
UBE3B	KAUFMAN OCULOCEREBROFACIAL SYNDROME; KOS [AR]
UBIAD1	SCHNYDER CORNEAL DYSTROPHY; SCCD [AD]
UBQLN2	AMYOTROPHIC LATERAL SCLEROSIS 15 WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA; ALS15 [XLD]
UBR1	JOHANSON-BLIZZARD SYNDROME; JBS [AR]
UBTF	NEURODEGENERATION. CHILDHOOD-ONSET. WITH BRAIN ATROPHY; CONDBA [AD]
UFC1	NEURODEVELOPMENTAL DISORDER WITH SPASTICITY AND POOR GROWTH; NEDSG [AR]
UFM1	LEUKODYSTROPHY. HYPOMYELINATING. 14; HLD14 [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]
UMOD	HYPERURICEMIC NEPHROPATHY. FAMILIAL JUVENILE. 1; HNFJ1 [AD]
UMPS	OROTIC ACIDURIA [AR]
UNC13D	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS. FAMILIAL. 3; FHL3 [AR]
UNC80	HYPOTONIA. INFANTILE. WITH PSYCHOMOTOR RETARDATION AND CHARACTERISTIC FACIES 2; IHPRF2 [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]
UQCC2	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 7; MC3DN7 [AR]
UQCRB	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 3; MC3DN3 [AR]
UQCRC2	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 5; MC3DN5 [AR]
UQCRQ	MITOCHONDRIAL COMPLEX III DEFICIENCY. NUCLEAR TYPE 4; MC3DN4 [AR]
UROD	PORPHYRIA CUTANEA TARDA [AD]
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]

USP18	PSEUDO-TORCH SYNDROME 2; PTORCH2 [AR]
USP27X	MENTAL RETARDATION. X-LINKED 105; MRX105 [XLR]
USP8	PITUITARY ADENOMA 4. ACTH-SECRETING; PITA4 [Autosomal recessive vs. dominant MEN]
USP9X	MENTAL RETARDATION. X-LINKED 99; MRX99 [XLR] | MENTAL RETARDATION. X-LINKED 99. SYNDROMIC. FEMALE-RESTRICTED; MRXS99F [XLD]
USP9Y	SPERMATOGENIC FAILURE. Y-LINKED. 2; SPGFY2 [Y-linked (Yq11.23]
UVSSA	UV-SENSITIVE SYNDROME 3; UVSS3 [AR]
VAC14	STRIATONIGRAL DEGENERATION. CHILDHOOD-ONSET; SNDC [AR]
VAMP1	SPASTIC ATAXIA 1. AUTOSOMAL DOMINANT; SPAX1 [AD] | MYASTHENIC SYNDROME. CONGENITAL. 25. PRESYNAPTIC; CMS25 [AR]
VANGL1	SACRAL DEFECT WITH ANTERIOR MENINGOCELE [AD] | NEURAL TUBE DEFECTS. SUSCEPTIBILITY TO; NTD [AD]
VANGL2	NEURAL TUBE DEFECTS. SUSCEPTIBILITY TO; NTD [AD]
VAPB	AMYOTROPHIC LATERAL SCLEROSIS 8; ALS8 [AD] | SPINAL MUSCULAR ATROPHY. LATE-ONSET. FINKEL TYPE; SMAFK [AD]
VARS1	NEURODEVELOPMENTAL DISORDER WITH MICROCEPHALY. SEIZURES. AND CORTICAL ATROPHY; NDMSCA [AR]
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20; COXPD20 [AR]
VCAN	WAGNER VITREORETINOPATHY; WGVRP [AD]
VCL	CARDIOMYOPATHY. FAMILIAL HYPERTROPHIC. 15; CMH15 [AD]
VCP	INCLUSION BODY MYOPATHY WITH EARLY-ONSET PAGET DISEASE WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA 1; IBMPFD1 [AD] | CHARCOT-MARIE-TOOTH DISEASE. AXONAL. TYPE 2Y; CMT2Y [AD]
VDR	VITAMIN D-DEPENDENT RICKETS. TYPE 2A; VDDR2A [AR]
VEGFC	LYMPHATIC MALFORMATION 4; LMPHM4 [AD]
VHL	VON HIPPEL-LINDAU SYNDROME; VHLS [AD] | ERYTHROCYTOSIS. FAMILIAL. 2. AUTOSOMAL RECESSIVE; ECYT2 [AR] | PHEOCHROMOCYTOMA [AD]
VIM	CATARACT 30. MULTIPLE TYPES; CTRCT30 [AD]
VIPAS39	ARTHROGRYPOSIS. RENAL DYSFUNCTION. AND CHOLESTASIS 2; ARCS2 [AR]
VKORC1	COUMARIN RESISTANCE [AD]
VLDLR	CEREBELLAR ATAXIA. MENTAL RETARDATION. AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 [AR]
VMA21	MYOPATHY. X-LINKED. WITH EXCESSIVE AUTOPHAGY; MEAX [XLR]
VPS11	LEUKODYSTROPHY. HYPOMYELINATING. 12; HLD12 [AR]
VPS13A	CHOREOACANTHOCYTOSIS; CHAC [AR]
VPS13B	COHEN SYNDROME; COH1 [AR]
VPS13C	PARKINSON DISEASE 23. AUTOSOMAL RECESSIVE EARLY-ONSET; PARK23 [AR]
VPS13D	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 4; SCAR4 [AR]
VPS33A	MUCOPOLYSACCHARIDOSIS-PLUS SYNDROME; MPSPS [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]
VPS51	PONTOCEREBELLAR HYPOPLASIA. TYPE 13; PCH13 [AR]
VPS53	PONTOCEREBELLAR HYPOPLASIA. TYPE 2E; PCH2E [AR]
VRK1	PONTOCEREBELLAR HYPOPLASIA. TYPE 1A; PCH1A [AR]
VSX1	KERATOCONUS 1; KTCN1 [AD]
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]
WAC	DESANTO-SHINAWI SYNDROME; DESSH [AD]
WARS1	NEURONOPATHY. DISTAL HEREDITARY MOTOR. TYPE IX; HMN9 [AD]

WARS2	NEURODEVELOPMENTAL DISORDER. MITOCHONDRIAL. WITH ABNORMAL MOVEMENTS AND LACTIC ACIDOSIS. WITH OR WITHOUT SEIZURES; NEMMLAS [AR]
WAS	NEUTROPENIA. SEVERE CONGENITAL. X-LINKED; SCNX [XLR] | THROMBOCYTOPENIA 1; THC1 [XLR] | WISKOTT-ALDRICH SYNDROME; WAS [XLR]
WASHC5	SPASTIC PARAPLEGIA 8. AUTOSOMAL DOMINANT; SPG8 [AD] | RITSCHER-SCHINZEL SYNDROME 1; RTSC1 [AR]
WBP2	DEAFNESS. AUTOSOMAL RECESSIVE 107; DFNB107 [AR]
WDR11	HYPOGONADOTROPIC HYPOGONADISM 14 WITH OR WITHOUT ANOSMIA; HH14 [AD]
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]
WDR26	SKRABAN-DEARDORFF SYNDROME; SKDEAS [AD]
WDR34	SHORT-RIB THORACIC DYSPLASIA 11 WITH OR WITHOUT POLYDACTYLY; SRTD11 [AR]
WDR35	CRANIOECTODERMAL DYSPLASIA 2; CED2 [AR] | SHORT-RIB THORACIC DYSPLASIA 7 WITH OR WITHOUT POLYDACTYLY; SRTD7 [AR]
WDR36	Glaucoma 1 open angle G 609887 (3)
WDR37	NEUROOCULOCARDIOGENITOURINARY SYNDROME; NOCGUS [AD]
WDR4	MICROCEPHALY. GROWTH DEFICIENCY. SEIZURES. AND BRAIN MALFORMATIONS; MIGSB [AR] | GALLOWAY-MOWAT SYNDROME 6; GAMOS6 [AR]
WDR45	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 5; NBIA5 [XLD]
WDR45B	NEURODEVELOPMENTAL DISORDER WITH SPASTIC QUADRIPLEGIA AND BRAIN ABNORMALITIES WITH OR WITHOUT SEIZURES; NEDSBAS [AR]
WDR60	SHORT-RIB THORACIC DYSPLASIA 8 WITH OR WITHOUT POLYDACTYLY; SRTD8 [AR]
WDR62	MICROCEPHALY 2. PRIMARY. AUTOSOMAL RECESSIVE. WITH OR WITHOUT CORTICAL MALFORMATIONS; MCPH2 [AR]
WDR72	AMELOGENESIS IMPERFECTA. HYPOMATURATION TYPE. IIA3; AI2A3 [AR]
WDR73	GALLOWAY-MOWAT SYNDROME 1; GAMOS1 [AR]
WDR81	CEREBELLAR ATAXIA. MENTAL RETARDATION. AND DYSEQUILIBRIUM SYNDROME 2; CAMRQ2 [AR] | HYDROCEPHALUS. CONGENITAL. 3. WITH BRAIN ANOMALIES; HYC3 [AR]
WEE2	OOCYTE MATURATION DEFECT 5; OOMD5 [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]
WHRN	DEAFNESS. AUTOSOMAL RECESSIVE 31; DFNB31 [AR] | USHER SYNDROME. TYPE IID; USH2D [AR]
WNK1	NEUROPATHY. HEREDITARY SENSORY AND AUTONOMIC. TYPE IIA; HSAN2A [AR] | PSEUDOHYPOALDOSTERONISM. TYPE IIC; PHA2C [AD]
WNK4	PSEUDOHYPOALDOSTERONISM. TYPE IIB; PHA2B [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]
WNT2B	DIARRHEA 9; DIAR9 [AR]
WNT4	46.XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS. ADRENALS. AND LUNGS; SERKAL [AR] | MULLERIAN APLASIA AND HYPERANDROGENISM [AD]
WNT5A	ROBINOW SYNDROME. AUTOSOMAL DOMINANT 1; DRS1 [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]
WWOX	SPINOCEREBELLAR ATAXIA. AUTOSOMAL RECESSIVE 12; SCAR12 [AR] | ESOPHAGEAL CANCER [Somatic mutation] | EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 28; EIEE28 [AR]
XDH	XANTHINURIA. TYPE I; XAN1 [AR]
XIAP	LYMPHOPROLIFERATIVE SYNDROME. X-LINKED. 2; XLP2 [XLR]
XIST	X-inactivation familial skewed 300087 (3)

XK	McLeod syndrome with or without chronic granulomatous disease 300842 (3)
XPA	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP A; XPA [AR]
XPC	XERODERMA PIGMENTOSUM. COMPLEMENTATION GROUP C; XPC [AR]
XPNPEP3	NEPHRONOPHTHISIS-LIKE NEPHROPATHY 1; NPHPL1 [AR]
XPR1	BASAL GANGLIA CALCIFICATION. IDIOPATHIC. 6; IBGC6 [AD]
XRCC4	SHORT STATURE. MICROCEPHALY. AND ENDOCRINE DYSFUNCTION; SSMED [AR]
XYLT1	PSEUDOXANTHOMA ELASTICUM; PXE [AR] | DESBUQUOIS DYSPLASIA 2; DBQD2 [AR]
XYLT2	PSEUDOXANTHOMA ELASTICUM; PXE [AR] | SPONDYLOOCULAR SYNDROME; SOS [AR]
YAP1	COLOBOMA. OCULAR. WITH OR WITHOUT HEARING IMPAIRMENT. CLEFT LIP/PALATE. AND/OR MENTAL RETARDATION; COB1 [AD]
YARS1	CHARCOT-MARIE-TOOTH DISEASE. DOMINANT INTERMEDIATE C; CMTDIC [AD]
YARS2	MYOPATHY. LACTIC ACIDOSIS. AND SIDEROBLASTIC ANEMIA 2; MLASA2 [AR]
YWHAG	EPILEPTIC ENCEPHALOPATHY. EARLY INFANTILE. 56; EIEE56 [AD]
YY1	GABRIELE-DE VRIES SYNDROME; GADEVS [AD]
YY1AP1	GRANGE SYNDROME; GRNG [AR]
ZAP70	AUTOIMMUNE DISEASE. MULTISYSTEM. INFANTILE-ONSET. 2; ADMIO2 [AR] | IMMUNODEFICIENCY 48; IMD48 [AR]
ZBTB11	INTELLECTUAL DEVELOPMENTAL DISORDER. AUTOSOMAL RECESSIVE 69; MRT69 [AR]
ZBTB16	SKELETAL DEFECTS. GENITAL HYPOPLASIA. AND MENTAL RETARDATION [AR]
ZBTB18	MENTAL RETARDATION. AUTOSOMAL DOMINANT 22; MRD22 [AD]
ZBTB20	PRIMROSE SYNDROME; PRIMS [AD]
ZBTB24	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 2; ICF2 [AR]
ZC3H14	MENTAL RETARDATION. AUTOSOMAL RECESSIVE 56; MRT56 [AR]
ZC4H2	WIEACKER-WOLFF SYNDROME; WRWF [XLR]
ZDHHC9	Mental retardation X-linked syndromic Raymond type 300799 (3)
ZEB1	Corneal dystrophy Fuchs endothelial 6 613270 (3) | Corneal dystrophy posterior polymorphous 3 609141 (3)
ZEB2	MOWAT-WILSON SYNDROME; MOWS [AD]
ZFHX3	PROSTATE CANCER [Somatic mutation]
ZFP57	DIABETES MELLITUS. TRANSIENT NEONATAL. 1; TNDM1 [Autosomal dominant (loss of maternal allele)]
ZFPM2	46.XY SEX REVERSAL 9; SRXY9 [AD] | TETRALOGY OF FALLOT; TOF [AD]
ZFYVE26	SPASTIC PARAPLEGIA 15. AUTOSOMAL RECESSIVE; SPG15 [AR]
ZFYVE27	SPASTIC PARAPLEGIA 33. AUTOSOMAL DOMINANT; SPG33 [AD]
ZIC1	STRUCTURAL BRAIN ANOMALIES WITH IMPAIRED INTELLECTUAL DEVELOPMENT AND CRANIOSYNOSTOSIS; BAIDCS [AD] | CRANIOSYNOSTOSIS 6; CRS6 [AD]
ZIC2	HOLOPROSENCEPHALY 5; HPE5 [AD]
ZIC3	VACTERL ASSOCIATION. X-LINKED. WITH OR WITHOUT HYDROCEPHALUS; VACTERLX [XLR] | HETEROTAXY. VISCERAL. 1. X-LINKED; HTX1 [XLR]
ZMIZ1	NEURODEVELOPMENTAL DISORDER WITH DYSMORPHIC FACIES AND DISTAL SKELETAL ANOMALIES; NEDDFSA [AD]
ZMPSTE24	RESTRICTIVE DERMOPATHY. LETHAL [AR] | MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY; MADB [AR]
ZMYND10	CILIARY DYSKINESIA. PRIMARY. 22; CILD22 [AR]
ZMYND11	MENTAL RETARDATION. AUTOSOMAL DOMINANT 30; MRD30 [AD]
ZNF142	NEURODEVELOPMENTAL DISORDER WITH IMPAIRED SPEECH AND HYPERKINETIC MOVEMENTS; NEDISHM [AR]
ZNF148	GLOBAL DEVELOPMENTAL DELAY. ABSENT OR HYPOPLASTIC CORPUS CALLOSUM. AND DYSMORPHIC FACIES; GDACCF [AD]
ZNF335	MICROCEPHALY 10. PRIMARY. AUTOSOMAL RECESSIVE; MCPH10 [AR]
ZNF341	HYPER-IgE RECURRENT INFECTION SYNDROME 3. AUTOSOMAL RECESSIVE; HIES3 [AR]
ZNF408	EXUDATIVE VITREORETINOPATHY 6; EVR6 [AD] | RETINITIS PIGMENTOSA 72; RP72 [AR]

ZNF423	NEPHRONOPHTHISIS 14; NPHP14 [AD]
ZNF462	WEISS-KRUSZKA SYNDROME; WSKA [AD]
ZNF469	BRITTLE CORNEA SYNDROME 1; BCS1 [AR]
ZNF644	MYOPIA 21. AUTOSOMAL DOMINANT; MYP21 [AD]
ZNF687	PAGET DISEASE OF BONE 6; PDB6 [AD]
ZNF711	MENTAL RETARDATION. X-LINKED 97; MRX97 [X-linked]
ZNF750	Seborrhea-like dermatitis with psoriasiform elements 610227 (3)
ZNHIT3	PEHO SYNDROME; PEHO [AR]
ZP1	OOCYTE MATURATION DEFECT 1; OOMD1 [AR]
ZP2	OOCYTE MATURATION DEFECT 6; OOMD6 [AR]
ZP3	OOCYTE MATURATION DEFECT 3; OOMD3 [AD]
ZSWIM6	ACROMELIC FRONTONASAL DYSOSTOSIS; AFND [AD] | NEURODEVELOPMENTAL DISORDER WITH MOVEMENT ABNORMALITIES. ABNORMAL GAIT. AND AUTISTIC FEATURES; NEDMAGA [AD]