Unidad de Genética
Clínica Vistahermosa

PANEL AMPLIADO VISTAHERMOSA

Gen	Enfermedad
AAAS	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME
ABCA12	ICHTHYOSIS CONGENITA, HARLEQUIN FETUS TYPE
ABCA3	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 3
ABCB11	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 2
ABCB4	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC
ABCC8	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, 1
ABCD1	ADRENOLEUKODYSTROPHY
ACAD9	DEFICIENCY OF ACYL-CoA DEHYDROGENASE FAMILY MEMBER 9
ACADL	ACYL-CoA DEHYDROGENASE, LONG-CHAIN, DEFICIENCY OF
ACADM	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF
ACADVL	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF
ACAT1	ALPHA-METHYLACETOACETIC ACIDURIA
ACOX1	PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY
ACSL4	MENTAL RETARDATION, X-LINKED 68
ADA	SEVERE COMBINED IMMUNODEFICIENCY, AUT REC, T CELL-NEGATIVE,
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL
ADAMTSL2	GELEOPHYSIC DYSPLASIA
ADCK3	COENZYME Q10 DEFICIENCY
AFF2	MENTAL RETARDATION X-LINKED ASSOCIATED WITH FRAGILE SITE
AGL	GLYCOGEN STORAGE DISEASE III
AGPS	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3
AGTR2	MENTAL RETARDATION X-LINKED 88
AHI1	JOUBERT SYNDROME 3
AIRE	AUTOIMMUNE POLYENDOCRINE SYNDROME TYPE I
ALDH3A2	SJOGREN-LARSSON SYNDROME
ALDH5A1	SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY
ALDH7A1	EPILEPSY, PYRIDOXINE-DEPENDENT; EPD
ALDOB	FRUCTOSE INTOLERANCE, HEREDITARY
ALG1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ik
ALG12	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ig
ALG2	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ii
ALG3	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Id; CDG1D
ALG6	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ic
ALG8	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ih
ALG9	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Il
ALMS1	ALSTROM SYNDROME
ALPL	HYPOPHOSPHATASIA, CHILDHOOD
ALS2	JUVENILE AMYOTROPHIC LATERAL SCLEROSIS 2; PRIMARY LATERAL SCLEROSIS, JUVENILE
AMACR	BILE ACID SYNTHESIS DEFECT, CONGENITAL, 4
AMT	GLYCINE ENCEPHALOPATHY
ANTXR2	FIBROMATOSIS, JUVENILE HYALINE; HYALINOSIS, INFANTILE SYSTEMIC
AP1S2	MENTAL RETARDATION, X-LINKED 59

AP3B1	HERMANSKY-PUDLAK SYNDROME 2
APTX	ATAXIA, EARLY-ONSET, WITH oculomotor apraxia AND HYPOALBUMINEMIA; COENZYME Q10 DEFICIENCY
AR	INFERTILE MALE SYNDROME
ARHGEF6	MENTAL RETARDATION, X-LINKED 46
ARHGEF9	HYPEREKPLEXIA AND EPILEPSY
ARSA	METACHROMATIC LEUKODYSTROPHY
ARSB	MUCOPOLYSACCHARIDOSIS TYPE VI MAROTEAUX-LAMY
ARSE	CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE
ARX	LISSENCEPHALY, X-LINKED, 2; CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENITALIA; EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 1
ASL	ARGININOSUCCINIC ACIDURIA
ASPA	CANAVAN DISEASE
ASS1	CITRULLINEMIA, CLASSIC
ATM	ATAXIA-TELANGIECTASIA
ATP6V0A2	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE II
ATP7A	MENKES DISEASE
ATP7B	WILSON DISEASE
ATP8B1	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1
ATR	SECKEL SYNDROME 1
ATRX	α-THALASSEMIA/MENTAL RETARDATION SYNDROME,NONDELETION TYPE, X-LINKED
AUH	3-METHYLGLUTACONIC ACIDURIA, TYPE I
B4GALT1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IId
BCKDHA	MAPLE SYRUP URINE DISEASE Type Ia
BCKDHB	BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE
BCOR	MICROPHTHALMIA, SYNDROMIC
BCS1L	GRACILE SYNDROME; MITOCHONDRIAL COMPLEX III DEFICIENCY
BLM	BLOOM SYNDROME
BRWD3	MENTAL RETARDATION, X-LINKED 93
BTD	BIOTINIDASE DEFICIENCY
BTK	AGAMMAGLOBULINEMIA, X-LINKED XLA
C10orf2	INFANTILE-ONSET SPINOCEREBELLAR ATAXIA; MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM
CA2	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 3
CASK	MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA
CASP10	Autoimmune lymphoproliferative syndrome
CBS	HOMOCYSTINURIA
CD19	IMMUNODEFICIENCY, COMMON VARIABLE, 3
CD247	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-ZETA
CD3D	SCID, AUT REC, T CELL-NEGATIVE, B CELL+, NK CELL+
CD3E	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-EPSILON
CD3G	IMMUNODEFICIENCY DUE TO DEFECT IN CD3-GAMMA
CD40LG	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1
CDH23	USHER SYNDROME, TYPE ID
CDKL5	Early infantile epileptic encephalopathy
CEP290	JOUBERT SYNDROME 5
CFP	PROPERDIN DEFICIENCY, X-LINKED
CFTR	CYSTIC FIBROSIS

CHRNA1	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE
CHRND	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE
CHRNG	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE; MULTIPLE PTERYGIUM SYNDROME, ESCOBAR VARIANT
CLCN5	DENT DISEASE 1
CLCN7	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 4
CLDN1	ICHTHYOSIS, LEUKOCYTE VACUOLES, ALOPECIA, AND SCLEROSING CHOLANGITIS
CLDN19	HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT
CLN3	NEURONAL CEROID LIPOFUSCINOSIS 3
CLN5	NEURONAL CEROID LIPOFUSCINOSIS 5
CLN6	CEROID LIPOFUSCINOSIS, NEURONAL, 6
CLN8	CEROID LIPOFUSCINOSIS, NEURONAL, 8; CEROID LIPOFUSCINOSIS, NEURONAL, 8, NORTHERN EPILEPSY VARIANT
CLRN1	Usher syndrome type 3A
COG1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIg
COG7	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE IIe
COG8	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIh
COL17A1	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE
COL1A1	Osteogenesis imperfecta, type I
COL1A2	Ehlers-Danlos syndrome, cardiac valvular form
COL4A3	ALPORT SYNDROME, AUTOSOMAL RECESSIVE
COL4A4	ALPORT SYNDROME, AUTOSOMAL RECESSIVE
COL4A5	ALPORT SYNDROME, X-LINKED
COL6A1	Bethlem myopathy 1
COL6A2	Bethlem myopathy 1
COL6A3	Bethlem myopathy 1
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE
COQ2	COENZYME Q10 DEFICIENCY
COQ9	COENZYME Q10 DEFICIENCY
COX10	Complex IV deficiency
COX15	Complex IV deficiency
COX6B1	Complex IV deficiency
CPS1	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY, HYPERAMMONEMIA DUE TO
CPT1A	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY
CPT2	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, INFANTILE; CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LATE-ONSET; CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL
CRLF1	CRISPONI SYNDROME
CRTAP	OSTEOGENESIS IMPERFECTA, TYPE IIB
CSTB	MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG
CTNS	CYSTINOSIS, NEPHROPATHIC; CYSTINOSIS, ADULT NONNEPHROPATHIC; CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT NEPHROPATHIC TYPE
CTSD	CEROID LIPOFUSCINOSIS, NEURONAL, 10
CTSK	PYCNODYSOSTOSIS
CUL4B	MENTAL RETARDATION X-LINKED WITH BRACHYDACTYLY AND MACROGLOSSIA
CYP11A1	LIPOID CONGENITAL ADRENAL HYPERPLASIA
CYP11B1	CONGENITAL ADRENAL HYPERPLASIA
CYP17A1	CONGENITAL ADRENAL HYPERPLASIA
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE DEFICIENCY
CYP27A1	CEREBROTENDINOUS XANTHOMATOSIS

CYP27B1	VITAMIN D-DEPENDENT osteopenia, TYPE I
DBT	MSUD type 2
DCLRE1C	OMENN SYNDROME
DCX	LISSENCEPHALY, X-LINKED, 1
DDB2	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP E
DDC	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY
DGUOK	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM
DHCR24	DESMOSTEROLOSIS
DHCR7	SMITH-LEMLI-OPITZ SYNDROME
DKC1	HOYERAAL-HREIDARSSON SYNDROME
DLD	DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY
DLG3	MENTAL RETARDATION X-LINKED 90
DLL3	Autosomal recessive spondylocostal dysostosis
DMD	MUSCULAR DYSTROPHY, DUCHENNE TYPE
DMP1	HYPOPHOSPHATEMIC osteopenia, AUTOSOMAL RECESSIVE
DNAJC19	3-METHYLGLUTACONIC ACIDURIA, TYPE V
DNMT3B	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME
DOCK8	HYPER-IgE RECURRENT INFECTION SYNDROME, AUTOSOMAL RECESSIVE
DOK7	Congenital myasthenic syndrome
DOLK	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Im
DPAGT1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ij
DPM1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ie
DPYD	DIHYDROPYRIMIDINE DEHYDROGENASE
DSP	EPIDERMOLYSIS BULLOSA, LETHAL ACANTHOLYTIC; SKIN FRAGILITY-WOOLLY HAIR SYNDROME
DYNC2H1	ASPHYXIATING THORACIC DYSTROPHY 3
EDA	ECTODERMAL DYSPLASIA, HYPOHIDROTIC, X-LINKED
EDN3	WAARDENBURG-SHAH SYNDROME
EDNRB	WAARDENBURG-SHAH SYNDROME; ABCD SYNDROME
EFEMP2	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I
EFNB1	CRANIOFRONTONASAL SYNDROME
EGR2	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS; NEUROPATHY, CONGENITAL HYPOMYELINATING: CHARCOT-MARIE-TOOTH DISEASE, TYPE 4E
EIF2AK3	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS
ENPP1	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY
EPM2A	MYOCLONIC EPILEPSY OF LAFORA
ERBB3	LETHAL CONGENITAL CONTRACTURE SYNDROME 2
ERCC2	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP D
ERCC3	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE; XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP B
ERCC4	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F
ERCC5	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G
ERCC6	CEREBROOCULOFACIOSKELETAL SYNDROME 1; COCKAYNE SYNDROME TYPE B; DE SANCTIS-CACCHIONE SYNDROME
ERCC8	COCKAYNE SYNDROME, TYPE A
ESCO2	ROBERTS SYNDROME
ETFA	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY
ETFB	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY
ETFDH	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY

ETHE1	ENCEPHALOPATHY, ETHYLMALONIC
EVC	ELLIS-VAN CREVELD SYNDROME; EVC
EVC2	ELLIS-VAN CREVELD SYNDROME
F8	HEMOPHILIA A; HEMA
F9	HEMOPHILIA B; HEMB
FAH	TYROSINEMIA, TYPE I
FAM126A	LEUKODYSTROPHY, HYPOMYELINATING, 5
FAM20C	RAINE SYNDROME
FANCC	Fanconi anemia type C
FAS	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE I
FASLG	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE I,
FASTKD2	Complex IV deficiency
FBLN5	CUTIS LAXA, AUTOSOMAL RECESSIVE, TYPE I
FERMT3	LEUKOCYTE ADHESION DEFICIENCY TYPE III
FGA	Afibrinogenemia, congenital
FGD1	FACIOGENITAL DYSPLASIA
FGD4	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H
FH	FUMARASE DEFICIENCY
FKRP	MUSCULAR DYSTROPHY, CONGENITAL, 1C; MUSCLE-EYE-BRAIN DISEASE
FKTN	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY
FOLR1	NEURODEGENERATION DUE TO CEREBRAL FOLATE TRANSPORT DEFICIENCY
FOXG1	Rett syndrome, congenital variant
FOXN1	T-CELL IMMUNODEFICIENCY, CONGENITAL ALOPECIA, AND NAIL DYSTROPHY
FOXP3	IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY, X-LINKED
FRAS1	FRASER SYNDROME
FREM2	FRASER SYNDROME
FTSJ1	MENTAL RETARDATION, X-LINKED 9
FUCA1	FUCOSIDOSIS
G6PC	Glycogen storage disease Ia
G6PC3	GLYCOGEN STORAGE DISEASE I VON GIERKE DISEASE
G6PD	GLUCOSE-6-PHOSPHATE DEHYDROGENASE
GAA	GLYCOGEN STORAGE DISEASE II (pompe)
GALC	KRABBE DISEASE
GALK1	GALACTOKINASE DEFICIENCY
GALT	GALACTOSEMIA
GAMT	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY
GBA	GAUCHER DISEASE I; GAUCHER DISEASE II; GAUCHER DISEASE III; GAUCHER DISEASE IIIC; GAUCHER DISEASE, PERINATAL LETHAL
GBE1	GLYCOGEN STORAGE DISEASE IV
GCDH	GLUTARIC ACIDEMIA I
GCSH	GLYCINE ENCEPHALOPATHY
GDAP1	CHARCOT-MARIE-TOOTH DISEASE TYPE 4A
GDI1	MENTAL RETARDATION, X-LINKED 41, 48
GFM1	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1
GJB2	DEAFNESS, CONGENITAL, WITH KERATOPACHYDERMIA AND CONSTRICTIONS OF FINGERS AND TOES
GJC2	LEUKODYSTROPHY, HYPOMYELINATING, 2

GLA	FABRY DISEASE
GLB1	GM1-GANGLIOSIDOSIS TYPE II; GM1-GANGLIOSIDOSIS, TYPE I
GLDC	GLYCINE ENCEPHALOPATHY
GLE1	LETHAL CONGENITAL CONTRACTURE SYNDROME 1
GNPTAB	MUCOLIPIDOSIS II ALPHA/BETA; MUCOLIPIDOSIS III ALPHA/BETA
GNRHR	HYPOGONADOTROPIC HYPOGONADISM
GPC3	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 1
GPR98	USHER SYNDROME, TYPE IIC
GRIK2	MENTAL RETARDATION AUTOSOMAL RECESSIVE 6
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY
GTF2H5	TRICHOTHIODYSTROPHY, PHOTOSENSITIVE
GUSB	MUCOPOLYSACCHARIDOSIS TYPE VII SLY SYNDROME
HADH	3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY
HADHA	HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE,; TRIFUNCTIONAL PROTEIN DEFICIENCY
HADHB	TRIFUNCTIONAL PROTEIN DEFICIENCY
HAMP	HEMOCHROMATOSIS, JUVENILE, TYPE 2B
HAX1	neutropenia, SEVERE CONGENITAL, AUTOSOMAL RECESSIVE 3
HBA1	ALPHA THALASSEMIA
HBB	THALASSEMIA MAJOR; SICKLE CELL ANEMIA
HESX1	PITUITARY DWARFISM III
HEXA	TAY-SACHS DISEASE
HEXB	SANDHOFF DISEASE
HFE2	HEMOCHROMATOSIS, JUVENILE, TYPE 2A
HGSNAT	MUCOPOLYSACCHARIDOSIS TYPE IIIC (Sanfilippo type c)
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HIBCH	BETA-HYDROXYISOBUTYRYL CoA DEACYLASE, DEFICIENCY OF
HLCS	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY
HMGCL	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY
HPD	Tyrosinemia, type III
HPRT1	LESCH-NYHAN SYNDROME
HSD11B2	CORTISOL 11-BETA-KETOREDUCTASE DEFICIENCY
HSD17B10	MENTAL RETARDATION, X-LINKED, SYNDROMIC 10
HSD17B3	17-BETA HYDROXYSTEROID DEHYDROGENASE III DEFICIENCY
HSD17B4	D-BIFUNCTIONAL PROTEIN DEFICIENCY
HSD3B2	3-BETA-HYDROXYSTEROID DEHYDROGENASE DEFICIENCY TYPE II
HSPG2	DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE
HUWE1	MENTAL RETARDATION X-LINKED SYNDROMIC TURNER TYPE
ICOS	ANTIBODY DEFICIENCY DUE TO ICOS DEFECT
IDS	MUCOPOLYSACCHARIDOSIS TYPE II
IDUA	HURLER SYNDROME
IFNGR1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
IFNGR2	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
IFT80	ASPHYXIATING THORACIC DYSTROPHY 2
IGHMBP2	SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 1
IKBKAP	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III

IKBKG	ATYPICAL MYCOBACTERIOSIS, FAMILIAL; ECTODERMAL DYSPLASIA, ANHIDROTIC, W IMMUNODEFICIENCY, OSTEOPETROSIS & LYMPHEDEMA; ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY
IL12B	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
IL12RB1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
IL1RAPL1	MENTAL RETARDATION, X-LINKED 21
IL1RN	Deficiency of Interleukin-1-receptor antagonist
IL2RG	COMBINED IMMUNODEFICIENCY, X-LINKED; SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED
INSR	DONOHUE SYNDROME
INVS	NEPHRONOPHTHISIS 2
IQCB1	SENIOR-LOKEN SYNDROME 5
ITGA6	EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA
ITGB4	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE; EPIDERMOLYSIS BULLOSA JUNCTIONALIS WITH PYLORIC ATRESIA
IVD	ISOVALERIC ACIDEMIA
JAK3	SEVERE COMBINED IMMUNODEFICIENCY, AUT REC, T CELL?, B CELL+, NK CELL?
KCNJ1	BARTTER SYNDROME, ANTENATAL, TYPE 2
KDM5C	MENTAL RETARDATION, X-LINKED, SYNDROMIC
L1CAM	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS; CORPUS CALLOSUM, PARTIAL AGENESIS OF, X-LINKED; MASA SYNDROME
LAMA2	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, 1A
LAMA3	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE; LARYNGOONYCHOCUTANEOUS SYNDROME; EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE
LAMB2	PIERSON SYNDROME
LAMB3	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE; EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE
LAMC2	EPIDERMOLYSIS BULLOSA, JUNCTIONAL, NON-HERLITZ TYPE; EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE
LARGE	MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1D
LBR	HYDROPS-ECTOPIC CALCIFICATION-MOTH-EATEN SKELETAL DYSPLASIA
LEPRE1	OSTEOGENESIS IMPERFECTA, TYPE VIII
LHCGR	PRECOCIOUS PUBERTY, MALE-LIMITED
LHX3	PITUITARY DWARFISM III; PITUITARY HORMONE DEFICIENCY, COMBINED, 3; CPHD3
LIFR	STUVE-WIEDEMANN SYNDROME
LIG4	SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION
LMNA	Mandibuloacral dysplasia; Hutchinson-Gilford progeria
LRP2	DONNAI-BARROW SYNDROME
LRPPRC	LEIGH SYNDROME, FRENCH-CANADIAN TYPE
LYST	CHEDIAK HIGASHI SYNDROME
MAN2B1	MANNOSIDOSIS, ALPHA B, LYSOSOMAL
MBTPS2	ICHTHYOSIS FOLLICULARIS, ATRICHIA, AND PHOTOPHOBIA SYNDROME
MCOLN1	MUCOLIPIDOSIS IV
MECP2	RETT SYNDROME
MED12	LUJAN-FRYNS SYNDROME
MEFV	FAMILIAL MEDITERRANEAN FEVER
MFSD8	CEROID LIPOFUSCINOSIS, NEURONAL, 7
MGAT2	CONGENITAL DISORDER OF GLYCOSYLATIO, TYPE IIa
MID1	OPITZ GBBB SYNDROME, X-LINKED
MKS1	MECKEL SYNDROME TYPE 1
MLC1	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS
MMAA	METHYLMALONIC ACIDURIA, cblA TYPE

MMAB	METHYLMALONIC ACIDURIA, cblB TYPE
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE
MOCS1	MOLYBDENUM COFACTOR DEFICIENCY
MOCS2	MOLYBDENUM COFACTOR DEFICIENCY
MOGS	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIb
MPDU1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE If
MPI	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ib
MPL	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL
MPV17	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM; NAVAJO NEUROHEPATOPATHY
MPZ	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS; NEUROPATHY, CONGENITAL HYPOMYELINATING: CHARCOT-MARIE-TOOTH DISEASE, TYPE 4E
MRPS16	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 2
MRPS22	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 5
MTM1	MYOTUBULAR MYOPATHY 1
MUT	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY
MVK	MEVALONIC ACIDURIA
MYD88	MYD88 DEFICIENCY
MYO5A	GRISCELLI SYNDROME, TYPE 1; ELEJALDE DISEASE
MYO7A	USHER SYNDROME, TYPE I
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY
NBN	NIJMEGEN BREAKAGE SYNDROME
NDP	NORRIE DISEASE
NDUFA1	Complex I Deficiency
NDUFA7	Complex I Deficiency
NDUFAF2	Complex I Deficiency
NDUFAF4	Complex I Deficiency
NDUFS3	Complex I Deficiency
NDUFS4	Complex I Deficiency
NDUFS5	Complex I Deficiency
NDUFS6	Complex I Deficiency
NDUFS7	Complex I Deficiency
NDUFS8	Complex I Deficiency
NDUFV1	Complex I Deficiency
NEB	NEMALINE MYOPATHY 2
NEU1	NEURAMINIDASE DEFICIENCY
NEUROG3	DIARRHEA 4, MALABSORPTIVE, CONGENITAL
NHEJ1	SCID W MICROCEPHALY, GROWTH RETARDATION, & SENS TO IONIZING RADIATION
NHLRC1	MYOCLONIC EPILEPSY OF LAFORA
NHS	NANCE-HORAN SYNDROME; NHS
NLGN4X	X-linked Asperger syndrome-2
NPC1	NIEMANN-PICK DISEASE, TYPE C1
NPC2	NIEMANN-PICK DISEASE, TYPE C2
NPHP1	JOUBERT SYNDROME 4; NEPHRONOPHTHISIS 1; SENIOR-LOKEN SYNDROME 1
NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA
NPHP4	NEPHRONOPHTHISIS 4

NPHS1	NEPHROSIS 1, CONGENITAL, FINNISH TYPE
NPHS2	nephrotic syndrome, STEROID-RESISTANT, AUTOSOMAL RECESSIVE
NR0B1	CONGENITAL ADRENAL HYPOPLASIA
NR5A1	GONADAL DYSGENESIS WITH ADRENAL FAILURE
NSD1	Sotos syndrome 1
NSUN2	Autosomal mental retardation
NTRK1	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS
NUP62	STRIATONIGRAL DEGENERATION, INFANTILE
NXF5	X-linked mental retardation
OCRL	LOWE OCULOCEREBRORENAL SYNDROME
OFD1	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2
OPA3	3-@METHYLGLUTACONIC ACIDURIA, TYPE III
OPHN1	MENTAL RETARDATION, XLR, W CEREBELLAR HYPOPLASIA & DISTINCTIVE FACIAL APPEARANCE
ORAI1	IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 1
OSTM1	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 5
OTC	ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO
OXCT1	Succinyl CoA:3-oxoacid CoA transferase deficiency
PAH	PHENYLKETONURIA
PAK3	MENTAL RETARDATION, X-LINKED 30
PANK2	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 1 (Hallervorden-Spatz)
PC	PYRUVATE CARBOXYLASE DEFICIENCY
PCCA	PROPIONIC ACIDEMIA
PCCB	PROPIONIC ACIDEMIA
PCDH19	EPILEPSY, FEMALE-RESTRICTED, WITH MENTAL RETARDATION
PDHA1	LEIGH SYNDROME, X-LINKED
PDHX	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY
PDP1	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY
PDSS1	COENZYME Q10 DEFICIENCY
PDSS2	COENZYME Q10 DEFICIENCY
PEX1	ZELLWEGER SYNDROME; ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM
PEX10	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX10
PEX12	ZELLWEGER SYNDROME
PEX13	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX13
PEX26	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX26
PEX5	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM | PEX5
PEX7	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA TYPE 1
PKHD1	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE
PKLR	PYRUVATE KINASE DEFICIENCY OF RED CELLS
PLA2G6	INFANTILE NEUROAXONAL DYSTROPHY
PLCE1	nephrotic syndrome, TYPE 3
PLDN	Hermansky Pudlak Syndrome, 9
PLEC	EPIDERMOLYSIS BULLOSA SIMPLEX WITH MUSCULAR DYSTROPHY
PLEKHG5	SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 4
PLG	PLASMINOGEN DEFICIENCY TYPE I
PLOD1	NEVO SYNDROME

PLP1	PELIZAEUS-MERZBACHER DISEASE; SPASTIC PARAPLEGIA 2, X-LINKED
PMM2	CONGENITAL DISORDER OF GLYCOSYLATION TYPE Ia
PMP22	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS
PNPO	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY
POLG	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS
POMGNT1	MUSCLE-EYE-BRAIN DISEASE
POMT1	WALKER-WARBURG SYNDROME
POMT2	WALKER-WARBURG SYNDROME
POR	ANTLEY-BIXLER SYNDROME; DISORDERED STEROIDOGENESIS
POU1F1	PITUITARY DWARFISM III
PPT1	NEURONAL CEROID LIPOFUSCINOSIS 1
PQBP1	RENPENNING SYNDROME 1
PRF1	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 2
PROP1	PITUITARY DWARFISM III
PRPS1	ARTS SYNDROME
PRSS12	MENTAL RETARDATION, AUTOSOMAL RECESSIVE 1
PRX	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS
PSAP	COMBINED SAPOSIN DEFICIENCY; METACHROMATIC LEUKODYSTROPHY DUE TO SAPOSIN B DEFICIENCY
PTEN	VATER association with macrocephaly and ventriculomegaly
PTH1R	CHONDRODYSPLASIA, BLOMSTRAND TYPE
PYGM	McArdle disease
RAB23	CARPENTER SYNDROME
RAB27A	GRISCELLI SYNDROME, TYPE 2
RAB39B	MENTAL RETARDATION X-LINKED 72
RAB3GAP1	WARBURG MICRO SYNDROME
RAB3GAP2	MARTSOLF SYNDROME
RAG1	OMENN SYNDROME; SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE,
RAG2	OMENN SYNDROME; SEVERE COMBINED IMMUNODEFICIENCY, AUTOSOMAL RECESSIVE, T CELL-NEGATIVE,
RAPSN	FETAL AKINESIA DEFORMATION SEQUENCE
RELN	LISSENCEPHALY 2
RFT1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE In
RMRP	CARTILAGE-HAIR HYPOPLASIA; ANAUXETIC DYSPLASIA
RNASEH2A	AICARDI-GOUTIERES SYNDROME 4
RNASEH2B	AICARDI-GOUTIERES SYNDROME 2
RNASEH2C	AICARDI-GOUTIERES SYNDROME 3
RPGRIP1L	MECKEL SYNDROME, TYPE 5
RPL10	X-linked mental retardation
RPS6KA3	COFFIN-LOWRY SYNDROME; MENTAL RETARDATION X-LINKED 19 INCLUDED
RRM2B	mtDNA depletion, encephalomyopathic form
SACS	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE
SAMHD1	AICARDI-GOUTIERES SYNDROME 5
SBDS	SHWACHMAN-DIAMOND SYNDROME
SC5DL	LATHOSTEROLOSIS
SCNN1A	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE
SCNN1B	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE

SCNN1G	PSEUDOHYPOALDOSTERONISM, TYPE I, AUTOSOMAL RECESSIVE
SCO1	Complex IV deficiency
SCO2	Complex IV deficiency; CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE
SEPN1	RIGID SPINE MUSCULAR DYSTROPHY 1
SFTPB	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, 1; RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS
SFTPC	RESPIRATORY DISTRESS SYNDROME IN PREMATURE INFANTS
SGSH	MUCOPOLYSACCHARIDOSIS TYPE IIIA (Sanfilippo type A)
SH2D1A	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 1
SHROOM4	STOCCO DOS SANTOS X-LINKED MENTAL RETARDATION SYNDROME
SIL1	Marinesco-Sjogren Syndrome
SLC12A1	BARTTER SYNDROME, ANTENATAL, TYPE 1
SLC12A6	AGENESIS OF THE CORPUS CALLOSUM WITH PERIPHERAL NEUROPATHY
SLC16A2	ALLAN-HERNDON-DUDLEY SYNDROME
SLC17A5	INFANTILE SIALIC ACID STORAGE DISORDER; SIALURIA, FINNISH TYPE
SLC22A5	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY
SLC25A15	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY
SLC25A22	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 3
SLC26A2	ACHONDROGENESIS, TYPE IB; ATELOSTEOGENESIS, TYPE II; DIASTROPHIC DYSPLASIA
SLC35A1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Iif
SLC35C1	CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIc
SLC35D1	SCHNECKENBECKEN DYSPLASIA
SLC37A4	GLYCOGEN STORAGE DISEASE Ib; GLYCOGEN STORAGE DISEASE Ic
SLC4A11	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS
SLC6A8	CREATINE DEFICIENCY SYNDROME, X-LINKED
SLC9A6	MENTAL RETARDATION, X-LINKED ANGELMAN, SYNDROMIC, CHRISTIANSON
SMN1	SPINAL MUSCULAR ATROPHY TYPE I; SPINAL MUSCULAR ATROPHY TYPE II; SPINAL MUSCULAR ATROPHY TYPE III
SMPD1	NIEMANN-PICK DISEASE, TYPE A; NIEMANN-PICK DISEASE, TYPE B
SMS	MENTAL RETARDATION, X-LINKED, SNYDER-ROBINSON TYPE
SNAP29	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA
SOX3	MENTAL RETARDATION, X-LINKED, WITH PANHYPOPITUITARISM
SP110	HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY
SRD5A2	PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS; PPSH
SRD5A3	Autosomal mental retardation CDG 1Q
ST3GAL3	Autosomal mental retardation
ST3GAL5	AMISH INFANTILE EPILEPSY SYNDROME
STAR	LIPOID CONGENITAL ADRENAL HYPERPLASIA
STAT1	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
STIM1	IMMUNE DYSFUNCTION WITH T-CELL INACTIVATION DUE TO CALCIUM ENTRY DEFECT 2
STRA6	MICROPHTHALMIA, SYNDROMIC 9 (Matthew-Wood syndrome)
STX11	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 4
STXBP2	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 5
SUCLA2	mtDNA depletion, encephalomyopathic form
SUCLG1	LACTIC ACIDOSIS, FATAL INFANTILE (mtDNA depletion)
SUOX	SULFOCYSTEINURIA

SURF1	LEIGH SYNDROME
SYP	MENTAL RETARDATION X-LINKED SYP-RELATED
TAT	Tyrosinemia, type II
TAZ	BARTH SYNDROME; CARDIOMYOPATHY, DILATED, 3A
TBCE	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME
TCF4	Corneal dystrophy, Fuchs endothelial, 3
TCIRG1	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 1
TGM1	ICHTHYOSIS, LAMELLAR, 1
TH	SEGAWA SYNDROME, AUTOSOMAL RECESSIVE
TIMM8A	OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA
TK2	MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC FORM
TLR3	HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 2
TMEM67	JOUBERT SYNDROME 6
TNFRSF11B	PAGET DISEASE, JUVENILE
TPP1	NEURONAL CEROID LIPOFUSCINOSIS 2
TRAPPC9	MENTAL RETARDATION AUTOSOMAL RECESSIVE 13
TREX1	AICARDI-GOUTIERES SYNDROME 1
TRIM37	MULIBREY NANISM
TSEN54	PONTOCEREBELLAR HYPOPLASIA TYPE 2A; PONTOCEREBELLAR HYPOPLASIA TYPE 4
TSFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3
TSHB	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, 4
TSPYL1	SUDDEN INFANT DEATH WITH DYSGENESIS OF THE TESTES SYNDROME
TTPA	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF
TUBA1A	LISSENCEPHALY 3
TUFM	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 4
TUSC3	MENTAL RETARDATION AUTOSOMAL RECESSIVE 7
TYK2	ATYPICAL MYCOBACTERIOSIS, FAMILIAL
TYMP	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME
UBA1	SPINAL MUSCULAR ATROPHY, X-LINKED 2
UBE2A	MENTAL RETARDATION X-LINKED SYNDROMIC UBE2A-RELATED
UB3A	ANGELMAN SYNDROME
UBR1	JOHANSON-BLIZZARD SYNDROME
UNC13D	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 3
UNC93B1	HERPES SIMPLEX ENCEPHALITIS, SUSCEPTIBILITY TO, 1
UPF3B	MENTAL RETARDATION, X-LINKED, SYNDROMIC 14
UQCRB	MITOCHONDRIAL COMPLEX III DEFICIENCY
UQCRQ	MITOCHONDRIAL COMPLEX III DEFICIENCY
UROS	PORPHYRIA, CONGENITAL ERYTHROPOIETIC
USH1C	USHER SYNDROME, TYPE IC
USH1G	USHER SYNDROME, TYPE IG
USH2A	USHER SYNDROME, TYPE IIA
VDR	VITAMIN D-DEPENDENT osteopenia, TYPE II
VIPAR	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 2
VLDLR	CEREBELLAR HYPOPLASIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL
VPS13B	COHEN SYNDROME

VPS33B	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS
WAS	WISKOTT-ALDRICH SYNDROME
WNT10A	ODONTOONYCHODERMAL DYSPLASIA
WNT3	TETRA-AMELIA, AUTOSOMAL RECESSIVE
WNT7A	SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1; FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING AND POLY-, SYN-, AND; ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY
XIAP	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2
XPA	DE SANCTIS-CACCHIONE SYNDROME; XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A
XPC	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP C
ZDHHC9	MENTAL RETARDATION X-LINKED SYNDROMIC ZDHHC9-RELATED
ZEB2	Mowat-Wilson syndrome
ZIC3	HETEROTAXY, VISCERAL, 1, X-LINKED
ZMPSTE24	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY; TIGHT SKIN CONTRACTURE SYNDROME, LETHAL
ZNF41	MENTAL RETARDATION X-LINKED 89
ZNF469	BRITTLE CORNEA SYNDROME (Ehlers-Danlos syndrome type VIB)
ZNF674	MENTAL RETARDATION X-LINKED 92
ZNF711	MENTAL RETARDATION X-LINKED ZNF711-RELATED