



# National Center for Biotechnology Information

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## OMIM Home Page -- Online Mendelian Inheritance in Man

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# OMIM reaches 10,000 Entries!!



The total number of entries in OMIM exceeded 10,000 on December 1, 1998.  
(The entries in the first print edition of 1968 numbered 1487.)

Welcome to OMIM(TM), Online Mendelian Inheritance in Man. This database is a catalog of human genes and genetic disorders authored and edited by Dr. Victor A. McKusick and his colleagues at Johns Hopkins and elsewhere, and developed for the World Wide Web by [NCBI](#), the National Center for Biotechnology Information. The database contains textual information, pictures, and reference information. It also contains copious links to NCBI's [Entrez](#) database of MEDLINE articles and sequence information.

**NEW** The OMIM Morbid Map, a catalog of cytogenetic map locations organized by disease, is now available.

### Browsing OMIM

- [Search the OMIM Database](#)
- [Search the OMIM Gene Map](#)
- [Search the OMIM Morbid Map](#)
- [The OMIM numbering system](#)
- [How to create WWW links to OMIM](#)
- [View the OMIM Update Log](#)
- [OMIM Statistics](#)
- [Citing OMIM in the literature](#)
- [The OMIM Gene List](#)

### OMIM Allied Resources


- [Entrez: the NCBI MEDLINE and GenBank retrieval system](#)
- [Human Gene Nomenclature Home Page](#)

- [The Davis Human/Mouse Homology Map](#)
- [Online Mendelian Inheritance in Animals \(OMIA\)](#)
- [The Alliance of Genetic Support Groups](#)
- [The Cardiff Human Gene Mutation Database \(HGMD\)](#)
- [The Jackson Laboratory: Courses, mouse resources, mouse databases](#)
- [RetNet: Genes causing Retinal Diseases](#)
- [MitoMap: the Emory University mitochondrial genome database](#)
- [HUM-MOLGEN: Courses, resources, databases, etc.](#)
  - There is also an [American mirror site](#).
- [Locus-specific mutation databases](#)

## Available for FTP

- The [complete text of OMIM, compressed](#) (about 23 megabytes)
- The [OMIM Gene Map](#) (about 625 kilobytes)
- The [OMIM Gene Map key](#) (about 6 kilobytes)
- The [OMIM Morbid Map](#) (about 115 kilobytes)

Note: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. The images associated with some OMIM entries are links to an image archive at the [Neonatology on the Web](#) site which is solely responsible for the online publishing of these images. Non-medical users, and professional users viewing these images outside their areas of expertise are strongly urged to seek the assistance of an expert in the interpretation of OMIM text and images.

A note on symbols: pressing the  symbol will find the MEDLINE articles whose text most closely matches the text of the preceding paragraph, using the Entrez MEDLINE neighboring function.

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For questions about the OMIM database in general,  
Comments and questions? Send mail to the [NCBI Help Desk](#).

Credits : Brandon Brylawski

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## Online Mendelian Inheritance in Man

[Search the OMIM Gene Map](#)

[Search the OMIM Morbid Map](#)

### Search OMIM Articles

Enter one or more search keywords:

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To Search all fields, leave the following boxes unchecked. To narrow your search to certain specific fields, check the boxes next to those fields' names. See [Query Help](#) for other ways to search.

Title:	OMIM Number:	Allelic Variants:
Text:	References:	Clinical Synopsis:
Gene Map Disorder:		Contributors:

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See only records which have been changed in the past

Display at most

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# Select Entries from OMIM -- Online Mendelian Inheritance in Man

477 entries found, searching for "limb"

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- [271665](#) SPONDYLOMETAEPIPHYSEAL DYSPLASIA, SHORT LIMB-HAND TYPE
- [#253600](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2A; LGMD2A
- [#253601](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2B; LGMD2B
- [\\*276820](#) ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY
- [211990](#) CAMPTOMELIC SYNDROME, LONG-LIMB TYPE
- [\\*600900](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2E; LGMD2E
- [\\*159000](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1A; LGMD1A
- [\\*253700](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2C; LGMD2C
- [\\*100300](#) ABSENCE DEFECT OF LIMBS, SCALP, AND SKULL
- [\\*600119](#) SARCOGLYCAN, ALPHA; SGCA
- [\\*136535](#) FORMIN; FMN
- [#601287](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2F; LGMD2F
- [\\*601253](#) CAVEOLIN 3; CAV3
- [\\*229850](#) FRYNS SYNDROME; FRNS
- [\\*254110](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2H
- [#156810](#) MICROGASTRIA-LIMB REDUCTION DEFECTS ASSOCIATION; MLRD
- [\\*159001](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B; LGMD1B
- [#148820](#) KLEIN-WAARDENBURG SYNDROME
- [\\*603511](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1D; LGMD1D
- [103300](#) AGLOSSIA-ADACTYLIA
- [146160](#) HYPOMELIA WITH MULLERIAN DUCT ANOMALIES
- [154400](#) ACROFACIAL DYSOSTOSIS 1, NAGER TYPE; AFD1
- [600506](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 4; LGMD4
- [\\*603543](#) LIMB-MAMMARY SYNDROME
- [601163](#) DIAPHRAGMATIC DEFECTS, LIMB DEFICIENCIES, AND OSSIFICATION DEFECTS OF SKULL
- [159400](#) MYASTHENIA, FAMILIAL LIMB-GIRDLE
- [\\*601954](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2G; LGMD2G
- [271510](#) SPONASTRIME DYSPLASIA
- [223610](#) DWARFISM, SHORT-LIMB, WITH ABSENT FIBULAS AND VERY SHORT DIGITS
- [\\*261540](#) PETERS ANOMALY WITH SHORT-LIMB DWARFISM
- [#601173](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL RECESSIVE
- [183300](#) SPLENOGONADAL FUSION WITH LIMB DEFECTS AND MICROGNATHIA
- [246560](#) LIMB DEFICIENCIES, DISTAL, WITH MICROGNATHIA
- [246570](#) LIMB DEFICIENCY-HEART MALFORMATION SYNDROME
- [266910](#) RENAL DYSPLASIA-LIMB DEFECTS SYNDROME

[601356](#) LETHAL SHORT-LIMB SKELETAL DYSPLASIA, AL GAZALI TYPE  
\*[600794](#) SPINAL MUSCULAR ATROPHY, DISTAL, WITH UPPER LIMB PREDOMINANCE  
[249710](#) MESOMELIC LIMB SHORTENING AND BOWING  
[268305](#) ROBIN SEQUENCE WITH CLEFT MANDIBLE AND LIMB ANOMALIES  
[246555](#) LIMB DEFECTS, DISTAL TRANSVERSE, WITH MENTAL RETARDATION AND SPASTICITY  
[273740](#) THORACOMELIC DYSPLASIA  
#[226670](#) EPIDERMOLYSIS BULLOSA SIMPLEX AND LIMB-GIRDLE MUSCULAR DYSTROPHY  
[165660](#) OSLAM SYNDROME  
\*[206920](#) ANOPHTHALMOS WITH LIMB ANOMALIES  
[223340](#) DK--PHOCOMELIA SYNDROME  
[215850](#) CLEFT-LIMB-HEART MALFORMATION SYNDROME  
[601016](#) MIDLINE MALFORMATIONS, MULTIPLE, WITH LIMB ABNORMALITIES AND HYPOPITUITARISM  
[602484](#) PELVIC HYPOPLASIA WITH LOWER-LIMB ARTHROGRYPOSIS  
\*[194350](#) WT LIMB-BLOOD SYNDROME  
[191440](#) ULNAR HYPOPLASIA  
[273680](#) THANATOPHORIC DYSPLASIA, GLASGOW VARIANT  
\*[603009](#) DYSFERLIN; DYSF  
#[142900](#) HOLT-ORAM SYNDROME; HOS  
\*[114240](#) CALPAIN, LARGE POLYPEPTIDE L3; CAPN3  
\*[601411](#) SARCOGLYCAN, DELTA; SGCD  
[122470](#) CORNELIA DE LANGE SYNDROME 1; CDL1  
\*[183600](#) SPLIT-HAND/FOOT MALFORMATION, TYPE 1; SHFM1  
#[181450](#) ULNAR-MAMMARY SYNDROME; UMS  
\*[602575](#) LIM HOMEO BOX TRANSCRIPTION FACTOR 1, BETA; LMX1B  
\*[107269](#) CD44 ANTIGEN; CD44  
\*[310200](#) MUSCULAR DYSTROPHY, PSEUDOHYPERTROPHIC PROGRESSIVE, DUCHENNE AND BECKER TYPES  
#[158810](#) MYOPATHY, BENIGN CONGENITAL, WITH CONTRACTURES  
#[254130](#) MIYOSHI MYOPATHY; MM  
\*[601621](#) T-BOX 3; TBX3  
#[181350](#) EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT; EDMD2  
#[100800](#) ACHONDROPLASIA; ACH  
\*[604140](#) DEATH INDUCER-OBLITERATOR 1  
\*[601620](#) T-BOX 5; TBX5  
\*[193500](#) WAARDENBURG SYNDROME, TYPE I; WS1  
\*[142989](#) HOMEO BOX D13; HOXD13  
\*[600483](#) FIBROBLAST GROWTH FACTOR 8; FGF8  
[217100](#) CONSTRICTING BANDS, CONGENITAL  
\*[142984](#) HOMEO BOX D10; HOXD10  
[308050](#) ICHTHYOSIFORM ERYTHRODERMA, UNILATERAL, WITH IPSILATERAL MALFORMATIONS, ESPECIALLY ABSENCE DEFORMITY OF LIMBS

[223200](#) DISORGANIZATION, MOUSE, HOMOLOG OF  
[\\*131290](#) ENGRAILED 1; EN1  
[\\*600725](#) SONIC HEDGEHOG, DROSOPHILA, HOMOLOG OF; SHH  
[\\*157900](#) MOEBIUS SYNDROME; MBS  
[\\*602115](#) FIBROBLAST GROWTH FACTOR 10; FGF10  
[\\*263750](#) POSTAXIAL ACROFACIAL DYSOSTOSIS; POADS  
[\\*176943](#) FIBROBLAST GROWTH FACTOR RECEPTOR 2; FGFR2  
[\\*274000](#) THROMBOCYTOPENIA--ABSENT RADIUS SYNDROME  
[\\*600535](#) MESENCHYME HOMEO BOX 2; MEOX2  
[\\*142983](#) MSH, DROSOPHILA, HOMEO BOX, HOMOLOG OF, 1; MSX1  
[\\*268300](#) ROBERTS SYNDROME; RBS  
[#200700](#) CHONDRODYSPLASIA, GREBE TYPE  
[\\*600664](#) CONSERVED HELIX-LOOP-HELIX UBIQUITOUS KINASE; CHUK  
[\\*200500](#) ACHEIROPODY  
[\\*601146](#) GROWTH/DIFFERENTIATION FACTOR 5; GDF5  
[141400](#) HEMIFACIAL MICROSOMIA WITH RADIAL DEFECTS  
[\\*142987](#) HOMEO BOX D1; HOXD1  
[603446](#) OROACRAL SYNDROME, VERLOES-KOULISCHER TYPE  
[\\*601570](#) WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 7A; WNT7A  
[\\*600334](#) TIBIAL MUSCULAR DYSTROPHY, TARDIVE  
[#101200](#) APERT SYNDROME  
[258860](#) ORAL-FACIAL-DIGITAL SYNDROME, TYPE IV  
[\\*602077](#) ECTRODACTYLY, ECTODERMAL DYSPLASIA, AND CLEFT LIP/PALATE SYNDROME 2; EEC2  
[\\*232500](#) GLYCOGEN STORAGE DISEASE IV  
[\\*232800](#) GLYCOGEN STORAGE DISEASE VII  
[#249700](#) MESOMELIC DWARFISM OF THE HYPOPLASTIC ULNA, FIBULA AND MANDIBLE TYPE  
[\\*602570](#) JAGGED 2; JAG2  
[\\*134920](#) FIBROBLAST GROWTH FACTOR 2; FGF2  
[\\*216550](#) COHEN SYNDROME; COH1  
[200900](#) ACHONDROPLASIA, SO-CALLED, AND SWISS-TYPE AGAMMAGLOBULINEMIA  
[173800](#) POLAND SYNDROME  
[#601462](#) MYASTHENIC SYNDROME, SLOW-CHANNEL CONGENITAL; SCCMS  
[108145](#) ARTHROGRYPOSIS WITH OCULOMOTOR LIMITATION AND ELECTRORETINAL ABNORMALITIES  
[\\*114290](#) CAMPOMELIC DYSPLASIA  
[#269000](#) SC PHOCOMELIA SYNDROME  
[#164300](#) OCULOPHARYNGEAL MUSCULAR DYSTROPHY; OPMD  
[\\*128100](#) DYSTONIA 1, TORSION; DYT1  
[\\*601622](#) TWIST, DROSOPHILA, HOMOLOG OF; TWIST  
[#601678](#) BARTTER SYNDROME, ANTENATAL HYPERCALCIURIC FORM  
[\\*602991](#) NOGGIN, MOUSE, HOMOLOG OF; NOG  
[#105830](#) ANGELMAN SYNDROME; AS

- [\\*100690](#) CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA POLYPEPTIDE 1; CHRNA1
- [#603596](#) POLYDACTYLY
- [#161200](#) NAIL-PATELLA SYNDROME; NPS
- [\\*129900](#) ECTRODACTYLY, ECTODERMAL DYSPLASIA, AND CLEFT LIP/PALATE SYNDROME 1; EEC1
- [\\*601285](#) DELETED IN SPLIT-HAND/SPLIT-FOOT 1 REGION
- [\\*163400](#) NIEVERGELT SYNDROME
- [\\*601239](#) DYSTROBREVIN, ALPHA; DTNA
- [603671](#) ACROMELIC FRONTAL NASAL DYSOSTOSIS
- [#146510](#) PALLISTER-HALL SYNDROME; PHS
- [\\*142988](#) HOMEO BOX D12; HOXD12
- [\\*145410](#) HYPERTELOMERISM WITH ESOPHAGEAL ABNORMALITY AND HYPOSPADIAS
- [\\*164980](#) FIBROBLAST GROWTH FACTOR 4; FGF4
- [\\*165240](#) GLI-KRUPPEL FAMILY MEMBER 3; GLI3
- [#128230](#) DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION
- [\\*142986](#) HOMEO BOX D11; HOXD11
- [\\*603732](#) CRYPTOCHROME 2; CRY2
- [\\*600095](#) SPLIT-HAND/FOOT MALFORMATION, TYPE 3; SHFM3
- [#530000](#) KEARNS-SAYRE SYNDROME; KSS
- [\\*603803](#) DACHSHUND, DROSOPHILA, HOMOLOG OF; DACH
- [\\*312865](#) SHORT STATURE; SS
- [\\*310300](#) EMERY-DREIFUSS MUSCULAR DYSTROPHY
- [\\*134797](#) FIBRILLIN 1; FBN1
- [\\*601933](#) CRYPTOCHROME 1; CRY1
- [\\*134934](#) FIBROBLAST GROWTH FACTOR RECEPTOR 3; FGFR3
- [\\*254210](#) MYASTHENIA GRAVIS, FAMILIAL INFANTILE; FIMG
- [174400](#) POLYDACTYLY, PREAXIAL I
- [\\*301000](#) WISKOTT-ALDRICH SYNDROME; WAS
- [274205](#) THUMB, HYPOPLASTIC, WITH CHOROID COLOBOMA, POORLY DEVELOPED ANTIHELIX, AND DEAFNESS
- [\\*121050](#) CONTRACTURAL ARACHNODACTYLY, CONGENITAL; CCA
- [207770](#) APROSENCEPHALY SYNDROME
- [260150](#) PALANT CLEFT PALATE SYNDROME
- [\\*112261](#) BONE MORPHOGENETIC PROTEIN 2; BMP2
- [\\*191900](#) MUCKLE-WELLS SYNDROME
- [184705](#) STEINFELD SYNDROME
- [\\*268800](#) SANDHOFF DISEASE
- [192350](#) VATER ASSOCIATION
- [\\*227650](#) FANCONI ANEMIA, COMPLEMENTATION GROUP A; FANCA
- [181250](#) SCALP DEFECTS AND POSTAXIAL POLYDACTYLY
- [\\*185900](#) SYNDACTYLY, TYPE I
- [\\*229300](#) FRIEDREICH ATAXIA 1; FRDA
- [\\*265000](#) PTERYGIUM SYNDROME

[119580](#) BLEPHAROCHEILODONTIC SYNDROME

\*[604103](#) MYOTILIN

[261575](#) PHAVER SYNDROME

#[241200](#) HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA

\*[120220](#) COLLAGEN, TYPE VI, ALPHA-1; COL6A1

\*[126800](#) DUANE RETRACTION SYNDROME 1; DURS1

\*[176946](#) EPHRIN RECEPTOR EphA2; EPHA2

\*[164860](#) MET PROTOONCOGENE; MET

[179280](#) RADIAL-RENAL SYNDROME

\*[162660](#) NEUROTROPHIN 3; NTF3

\*[156225](#) LAMININ, ALPHA-2; LAMA2

[174100](#) POLYDACTYLY, IMPERFORATE ANUS, AND VERTEBRAL ANOMALIES

[151050](#) LENZ-MAJEWSKI HYPEROSTOTIC DWARFISM

#[187600](#) THANATOPHORIC DYSPLASIA; TD

\*[182970](#) SPINAL MUSCULAR ATROPHY, FACIOSCAPULOHUMERAL TYPE; FSHSMA

\*[149730](#) LACRIMO-AURICULODENTODIGITAL SYNDROME; LADD

[149000](#) KLIPPEL-TRENAUNAY-WEBER SYNDROME

[160550](#) MYOPATHY, MITOCHONDRIAL, WITH CATARACT

\*[158900](#) FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A

\*[180800](#) ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA

[184260](#) SPONDYLOMETAPHYSEAL DYSPLASIA WITH DENTINOGENESIS IMPERFECTA

\*[192315](#) VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY

[155950](#) MELORHEOSTOSIS

[193050](#) VIBRATORY ANGIOEDEMA

\*[193090](#) VITAMIN B12-BINDING PROTEIN

[147421](#) INCLUSION BODY MYOSITIS

[187510](#) TETRAMELIC MONODACTYLY

[121070](#) CONTRACTURES OF FINGERS AND JAW

[134780](#) FEMORAL-FACIAL SYNDROME; FFS

#[146000](#) HYPOCHONDROPLASIA; HCH

\*[142981](#) HOMEO BOX D4; HOXD4

\*[142958](#) HOMEO BOX 1I; HOX1I

\*[142409](#) HEPATOCYTE GROWTH FACTOR; HGF

[201170](#) ACROFACIAL DYSOSTOSIS SYNDROME OF RODRIGUEZ

[201180](#) ACROFRONTOFACIONASAL DYSOSTOSIS SYNDROME

[141300](#) HEMIFACIAL ATROPHY, PROGRESSIVE; HFA

\*[167416](#) PAIRED BOX GENE 9; PAX9

\*[169400](#) PELGER-HUET ANOMALY

\*[603959](#) PARACELLIN 1

[302900](#) CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY AND FRIEDREICH ATAXIA, COMBINED

\*[300108](#) DIAPHANOUS, DROSOPHILA, HOMOLOG OF, 2; DIAPH2

[300004](#) CORPUS CALLOSUM, AGENESIS OF, WITH SEIZURES AND MICRENCEPHALY

- [\\*113100](#) BRACHYDACTYLY, TYPE C; BDC
- [\\*208100](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA, NEUROGENIC TYPE; AMCN
- [\\*208150](#) PENA-SHOKEIR SYNDROME, TYPE I
- [\\*601653](#) EYES ABSENT 1; EYA1
- [#115470](#) CAT EYE SYNDROME; CES
- [\\*209900](#) BARDET-BIEDL SYNDROME, TYPE 2; BBS2
- [\\*263650](#) POPLITEAL PTERYGIUM SYNDROME, LETHAL TYPE
- [#259420](#) OSTEOGENESIS IMPERFECTA, PROGRESSIVELY DEFORMING, WITH NORMAL SCLERAE
- [212135](#) CARDIOSKELETAL SYNDROME, KUWAITI TYPE
- [\\*212780](#) CENANI SYNDACTYLISM
- [214800](#) CHOANAL ATRESIA, POSTERIOR; PCA
- [259050](#) OSSIFIED EAR CARTILAGES WITH MENTAL DEFICIENCY, MUSCLE WASTING, AND BONY CHANGES
- [215140](#) CHONDRODYSTROPHY, HYDROPIK AND PRENATALLY LETHAL TYPE
- [258315](#) OMODYSPLASIA, GENERALIZED FORM
- [#550900](#) MYONEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME; MNGIE
- [309950](#) MUSCULAR DYSTROPHY, HEMIZYGOUS LETHAL TYPE
- [255140](#) MYOPATHY WITH GIANT ABNORMAL MITOCHONDRIA
- [603936](#) GROWTH/DIFFERENTIATION FACTOR 11
- [\\*603680](#) SPINOCEREBELLAR ATAXIA 8; SCA8
- [218600](#) CRANIOSYNOSTOSIS WITH RADIAL DEFECTS
- [219250](#) CUTIS MARMORATA TELANGIECTATICA CONGENITA; CMTC
- [\\*100500](#) ACANTHOCYTOSIS WITH NEUROLOGIC DISORDER
- [603280](#) PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA WITH HYPOGONADISM
- [\\*100710](#) CHOLINERGIC RECEPTOR, NICOTINIC, BETA POLYPEPTIDE 1; CHRNB1
- [#277610](#) WEISSENBACHER-ZWEYMULLER SYNDROME; WZS
- [120400](#) COLOBOMA OF MACULA WITH TYPE B BRACHYDACTYLY
- [\\*602629](#) DYSTONIA 6, TORSION; DYT6
- [276950](#) VACTERL ASSOCIATION WITH HYDROCEPHALUS
- [\\*602504](#) SHORT STATURE HOMEO BOX 2; SHOX2
- [602418](#) WEYERS ULNAR RAY/OLIGODACTYLY SYNDROME
- [\\*225500](#) ELLIS-VAN CREVELD SYNDROME; EVC
- [\\*602023](#) CHLORIDE CHANNEL, KIDNEY, B; CLCNKB
- [601779](#) PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA WITH SEVERE CARDIOMYOPATHY, AUTOSOMAL RECESSIVE
- [\\*601655](#) EYES ABSENT 3; EYA3
- [\\*601472](#) CHARCOT-MARIE-TOOTH DISEASE, NEURONAL TYPE, D
- [271150](#) SPINAL MUSCULAR ATROPHY, PROXIMAL, ADULT TYPE
- [254200](#) MYASTHENIA GRAVIS; MG
- [601170](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH SEVERE CENTRAL NERVOUS SYSTEM ATROPHY AND ABSENCE OF LARGE MYELINATED FIBERS
- [108130](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE II
- [\\*232300](#) GLYCOGEN STORAGE DISEASE II

[#118200](#) CHARCOT-MARIE-TOOTH DISEASE, TYPE 1B; CMT1B  
[\\*600890](#) HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA  
 THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT; HADHA  
[254100](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH RAPID PROGRESSION  
[\\*600785](#) VITAMIN D-DEPENDENT RICKETS, RECEPTOR-POSITIVE TYPE  
[\\*116860](#) CEREBRAL CAVERNOUS MALFORMATIONS 1; CCM1  
[\\*600175](#) SPINAL MUSCULAR ATROPHY, CONGENITAL NONPROGRESSIVE, OF LOWER  
 LIMBS  
[108720](#) ATELOSTEOGENESIS, TYPE I  
[\\*236700](#) MCKUSICK-KAUFMAN SYNDROME; MKKS  
[600251](#) OCULOMAXILLOFACIAL DYSPLASIA WITH OBLIQUE FACIAL CLEFTS  
[241800](#) HYPOTHALAMIC HAMARTOMAS  
[\\*245200](#) KRABBE DISEASE  
[\\*119100](#) CLEFT HAND AND ABSENT TIBIA  
[\\*600341](#) TYRO3 PROTEIN TYROSINE KINASE; TYRO3  
[\\*600359](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 1;  
 KCNJ1  
[\\*601205](#) SINE OCULIS HOMEO BOX, DROSOPHILA, HOMOLOG OF, 1; SIX1  
[\\*590050](#) TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1; MTTL1  
[249420](#) MELNICK-NEEDLES SYNDROME  
[268650](#) RUDIGER SYNDROME  
[#550500](#) MYOGLOBINURIA, RECURRENT  
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[\\*250250](#) CARTILAGE-HAIR HYPOPLASIA; CHH  
[#107480](#) TOWNES-BROCKS SYNDROME; TBS  
[251455](#) MICROMELIC DYSPLASIA, CONGENITAL, WITH DISLOCATION OF RADIUS  
[#601419](#) DESMIN-RELATED MYOPATHY  
[#253400](#) SPINAL MUSCULAR ATROPHY III; SMA3  
[\\*108721](#) ATELOSTEOGENESIS, TYPE III  
[\\*601654](#) EYES ABSENT 2; EYA2  
[\\*257600](#) OCULAR MYOPATHY WITH CURARE SENSITIVITY  
[\\*602067](#) CARDIOMYOPATHY, DILATED, 1F; CMD1F  
[\\*602183](#) BAGPIPE HOMEO BOX, DROSOPHILA, HOMOLOG OF, 1; BAPX1  
[\\*300100](#) ADRENOLEUKODYSTROPHY; ALD  
[304120](#) CRANIOORODIGITAL SYNDROME  
[\\*305450](#) FG SYNDROME; FGS1  
[257300](#) NONDISJUNCTION  
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- [\\*601580](#) CAPPING PROTEIN, MUSCLE Z-LINE, ALPHA-1; CAPZA1
- [\\*601572](#) CAPPING PROTEIN, MUSCLE Z-LINE, BETA; CAPZB
- [\\*603967](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE IV, ALPHA SUBUNIT; SCN4A
- [\\*160900](#) DYSTROPHIA MYOTONICA; DMPK
- [\\*310400](#) MYOTUBULAR MYOPATHY 1; MTM1
- [\\*232400](#) GLYCOGEN STORAGE DISEASE III
- [#530000](#) KEARNS-SAYRE SYNDROME; KSS
- [\\*261670](#) PHOSPHOGLYCERATE MUTASE, DEFICIENCY OF M SUBUNIT OF
- [\\*232300](#) GLYCOGEN STORAGE DISEASE II
- [#255110](#) MYOPATHY WITH DEFICIENCY OF CARNITINE PALMITOYLTRANSFERASE II
- [\\*600119](#) SARCOGLYCAN, ALPHA; SGCA
- [#601003](#) BRODY MYOPATHY
- [\\*102770](#) ADENOSINE MONOPHOSPHATE DEAMINASE 1; AMPD1
- [#145600](#) MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 1; MHS1
- [\\*310300](#) EMERY-DREIFUSS MUSCULAR DYSTROPHY
- [#220110](#) COMPLEX IV, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF
- [\\*590050](#) TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 1; MTTL1
- [\\*156225](#) LAMININ, ALPHA-2; LAMA2
- [\\*255125](#) MYOPATHY WITH DEFICIENCY OF SUCCINATE DEHYDROGENASE AND ACONITASE
- [\\*108730](#) ATPase, Ca(2+)-TRANSPORTING, FAST-TWITCH 1; ATP2A1
- [\\*172490](#) PHOSPHORYLASE KINASE, BETA SUBUNIT; PHKB
- [251900](#) MITOCHONDRIAL MYOPATHY
- [\\*125660](#) DESMIN; DES
- [#212140](#) CARNITINE DEFICIENCY, SYSTEMIC PRIMARY; CDSP
- [\\*201470](#) ACYL-CoA DEHYDROGENASE, SHORT-CHAIN; ACADS
- [#255700](#) MYOTONIA, GENERALIZED
- [#255120](#) HYPOGLYCEMIA, HYPOKETOTIC, WITH DEFICIENCY OF CARNITINE

## PALMITOYLTRANSFERASE I

- [\\*158900](#) FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1A; FSHMD1A
- [\\*306000](#) GLYCOGEN STORAGE DISEASE VIII
- [\\*128240](#) UTROPHIN; UTRN
- [\\*600818](#) TRANSGELIN; TAGLN
- [\\*601788](#) GROWTH/DIFFERENTIATION FACTOR 8; GDF8
- [\\*590060](#) TRANSFER RNA, MITOCHONDRIAL, LYSINE; MTTK
- [\\*147670](#) INSULIN RECEPTOR; INSR
- [255310](#) MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION
- [\\*253800](#) FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY; FCMD
- [\\*253700](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2C; LGMD2C
- [\\*248800](#) MARINESCO-SJOGREN SYNDROME; MSS
- [\\*163731](#) NITRIC OXIDE SYNTHASE 1; NOS1
- [#161800](#) NEMALINE MYOPATHY 1, AUTOSOMAL DOMINANT; NEM1
- [\\*236670](#) WALKER-WARBURG SYNDROME
- [\\*600536](#) INTEGRIN, ALPHA-7; ITGA7
- [\\*212160](#) CARNITINE DEFICIENCY, MYOPATHIC
- [#254130](#) MIYOSHI MYOPATHY; MM
- [\\*103850](#) ALDOLASE A, FRUCTOSE-BISPHOSPHATE; ALDOA
- [\\*114207](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-1 SUBUNIT; CACNB1
- [\\*238600](#) HYPERLIPOPROTEINEMIA, TYPE I
- [\\*590055](#) TRANSFER RNA, MITOCHONDRIAL, LEUCINE, 2; MTTL2
- [\\*600900](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2E; LGMD2E
- [245340](#) LACTATE TRANSPORTER DEFECT, MYOPATHY DUE TO
- [#160800](#) MYOTONIA CONGENITA, DOMINANT
- [\\*161650](#) NEBULIN; NEB
- [\\*302060](#) BARTH SYNDROME; BTHS
- [\\*191039](#) TROPONIN C, FAST; TNNC2
- [\\*603009](#) DYSFERLIN; DYSF
- [\\*128239](#) DYSTROPHIN-ASSOCIATED GLYCOPROTEIN 1; DAG1
- [\\*268200](#) RHABDOMYOLYSIS, ACUTE RECURRENT
- [#164300](#) OCULOPHARYNGEAL MUSCULAR DYSTROPHY; OPMD
- [\\*160780](#) MYOSIN, LIGHT POLYPEPTIDE 1, ALKALI, SKELETAL, FAST; MYL1
- [\\*601411](#) SARCOGLYCAN, DELTA; SGCD
- [\\*160565](#) MYOPATHY, TUBULAR AGGREGATE
- [\\*600660](#) MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE A; MEF2A
- [\\*147470](#) INSULIN-LIKE GROWTH FACTOR II; IGF2
- [\\*159980](#) MYOGENIC FACTOR 4; MYOG
- [#256030](#) NEMALINE MYOPATHY 2, AUTOSOMAL RECESSIVE; NEM2
- [232330](#) GLYCOGEN STORAGE DISEASE IIb
- [\\*601253](#) CAVEOLIN 3; CAV3
- [\\*600737](#) INCLUSION BODY MYOPATHY 2, AUTOSOMAL RECESSIVE; IBM2
- [\\*160790](#) MYOSIN, LIGHT CHAIN 3, ALKALI, VENTRICULAR, SKELETAL, SLOW; MYL3

- [\\*138190](#) SOLUTE CARRIER FAMILY 2, MEMBER 4; SLC2A4
- [\\*232700](#) GLYCOGEN STORAGE DISEASE VI
- [#253600](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2A; LGMD2A
- [\\*160150](#) MYOPATHY, CENTRONUCLEAR
- [#550900](#) MYONEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME; MNGIE
- [\\*208100](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA, NEUROGENIC TYPE; AMCN
- [\\*191030](#) TROPOMYOSIN 3; TPM3
- [#252010](#) COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF
- [#313200](#) SPINAL AND BULBAR MUSCULAR ATROPHY, X-LINKED 1; SMAX1
- [\\*159990](#) MYOGENIC FACTOR 5; MYF5
- [\\*258870](#) ORNITHINE AMINOTRANSFERASE DEFICIENCY
- [#226670](#) EPIDERMOLYSIS BULLOSA SIMPLEX AND LIMB-GIRDLE MUSCULAR DYSTROPHY
- [\\*254210](#) MYASTHENIA GRAVIS, FAMILIAL INFANTILE; FIMG
- [#118220](#) CHARCOT-MARIE-TOOTH DISEASE, TYPE 1A; CMT1A
- [#160120](#) MYOKYMIA WITH PERIODIC ATAXIA
- [\\*602044](#) UNCOUPLING PROTEIN 3; UCP3
- [\\*602127](#) SMOOTHELIN; SMTN
- [\\*600662](#) MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE C; MEF2C
- [\\*601162](#) SPASTIC PARAPLEGIA 9, AUTOSOMAL DOMINANT; SPG9
- [254195](#) MYASTHENIA, CONGENITAL, PREDOMINANTLY FACIAL AND MASTICATORY
- [\\*516050](#) CYTOCHROME c OXIDASE III; MTCO3
- [147421](#) INCLUSION BODY MYOSITIS
- [#181350](#) EMERY-DREIFUSS MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT; EDMD2
- [\\*172471](#) PHOSPHORYLASE KINASE, TESTIS/LIVER, GAMMA-2; PHKG2
- [#601419](#) DESMIN-RELATED MYOPATHY
- [\\*601442](#) COFILIN 1; CFL1
- [\\*601227](#) PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA, TYPE 3; PEO3
- [\\*602419](#) EARLY GROWTH RESPONSE 3; EGR3
- [255100](#) MYOPATHY WITH ABNORMAL LIPID METABOLISM
- [\\*138550](#) GLYCOGEN PHOSPHORYLASE, BRAIN TYPE; PYGB
- [\\*603942](#) GLYCOGENIN; GYG
- [\\*603628](#) MUSCULIN; MSC
- [\\*212138](#) CARNITINE-ACYLCARNITINE TRANSLOCASE; CACT
- [\\*600917](#) PROTEIN PHOSPHATASE 1, REGULATORY INHIBITOR SUBUNIT 3; PPP1R3
- [\\*255320](#) MYOPATHY, CONGENITAL MULTICORE, WITH EXTERNAL OPHTHALMOPLEGIA
- [157650](#) MITOCHONDRIAL MYOPATHY, LIPID TYPE
- [\\*600334](#) TIBIAL MUSCULAR DYSTROPHY, TARDIVE
- [\\*600109](#) PROXIMAL MYOTONIC MYOPATHY; PROMM
- [\\*179503](#) RAS-RELATED ASSOCIATED WITH DIABETES; RRAD
- [\\*600535](#) MESENCHYME HOMEBOX 2; MEOX2
- [\\*172405](#) PHOSPHOLAMBAN; PLN

- [\\*602873](#) NEBULIN-RELATED ANCHORING PROTEIN; NRAP
- [\\*150000](#) LACTATE DEHYDROGENASE-A; LDHA
- [#540000](#) MITOCHONDRIAL MYOPATHY, ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKELIKE EPISODES; MELAS
- [\\*303631](#) COLLAGEN, TYPE IV, ALPHA-6; COL4A6
- [\\*158590](#) MUSCULAR ATROPHY, ADULT SPINAL
- [255200](#) MYOPATHY, CENTRONUCLEAR
- [\\*601625](#) ADP-RIBOSYLTRANSFERASE 1; ART1
- [\\*114209](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, GAMMA SUBUNIT 1; CACNG1
- [#253300](#) SPINAL MUSCULAR ATROPHY I; SMA1
- [\\*191045](#) TROPONIN T2, CARDIAC; TNNT2
- [\\*600922](#) MYOSIN LIGHT CHAIN KINASE; MYLK
- [601170](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH SEVERE CENTRAL NERVOUS SYSTEM ATROPHY AND ABSENCE OF LARGE MYELINATED FIBERS
- [\\*160781](#) MYOSIN, LIGHT CHAIN 2, REGULATORY, CARDIAC, SLOW; MYL2
- [\\*180902](#) RYANODINE RECEPTOR 2; RYR2
- [#302045](#) CARDIOMYOPATHY, DILATED, X-LINKED; XLCM
- [\\*602415](#) DYSTROBREVIN, BETA; DTNB
- [#545000](#) MYOCLONIC EPILEPSY ASSOCIATED WITH RAGGED-RED FIBERS; MERRF
- [#168300](#) PARAMYOTONIA CONGENITA OF VON EULENBURG; PMC
- [\\*602790](#) FOUR-AND-A-HALF LIM DOMAINS 3; FHL3
- [#192600](#) CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 1; CMH1
- [\\*246900](#) LIPOAMIDE DEHYDROGENASE DEFICIENCY, LACTIC ACIDOSIS DUE TO
- [\\*602771](#) RIGID SPINE MUSCULAR DYSTROPHY 1; RSMD1
- [\\*516040](#) COMPLEX IV, CYTOCHROME c OXIDASE SUBUNIT II; MTCO2
- [\\*255800](#) SCHWARTZ-JAMPEL SYNDROME, TYPE 1; SJS1
- [252011](#) COMPLEX II, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF
- [171850](#) PHOSPHOFRUCTOKINASE, RED CELL
- [\\*305900](#) GLUCOSE-6-PHOSPHATE DEHYDROGENASE; G6PD
- [\\*600824](#) CYSTEINE- AND GLYCINE-RICH PROTEIN 3; CSRP3
- [#601287](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2F; LGMD2F
- [\\*516003](#) COMPLEX I, SUBUNIT ND4; MTND4
- [#603174](#) HOMOCYSTEINEMIA
- [\\*155310](#) MEGADUODENUM AND/OR MEGACYSTIS
- [#158810](#) MYOPATHY, BENIGN CONGENITAL, WITH CONTRACTURES
- [\\*156610](#) MICHELIN TIRE BABY SYNDROME
- [\\*115200](#) CARDIOMYOPATHY, DILATED, 1A; CMD1A
- [\\*154275](#) MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 2; MHS2
- [\\*103320](#) AGRIN; AGRN
- [#308940](#) LEIOMYOMATOSIS, ESOPHAGEAL AND VULVAL, WITH NEPHROPATHY
- [#153480](#) MACROCEPHALY, MULTIPLE LIPOMAS, AND HEMANGIOMATA
- [\\*300163](#) FOUR-AND-A-HALF LIM DOMAINS 1; FHL1
- [139210](#) GROWTH-MENTAL DEFICIENCY SYNDROME OF MYHRE

[254950](#) MYOPATHY, GRANULOVACUOLAR LOBULAR, WITH ELECTRICAL MYOTONIA  
[#253601](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2B; LGMD2B  
[\\*102630](#) ACTIN, BETA; ACTB  
[\\*600467](#) MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 4; MHS4  
[\\*600650](#) CARNITINE PALMITOYLTRANSFERASE II; CPT2  
[\\*114204](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-2/DELTA SUBUNIT;  
CACNA2D1  
[\\*275630](#) TRIGLYCERIDE STORAGE DISEASE WITH IMPAIRED LONG-CHAIN FATTY ACID  
OXIDATION  
[\\*309600](#) MENTAL RETARDATION, X-LINKED, WITH HYPOTONIA  
[\\*231680](#) GLUTARICACIDURIA IIA  
[\\*272800](#) TAY-SACHS DISEASE; TSD  
[\\*602203](#) SARCOLIPIN; SLN  
[\\*102573](#) ACTININ, ALPHA-2; ACTN2  
[261740](#) PHOSPHORYLASE KINASE DEFICIENCY OF HEART  
[\\*268800](#) SANDHOFF DISEASE  
[#170400](#) PERIODIC PARALYSIS I  
[#601887](#) MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 5; MHS5  
[\\*600692](#) TROPONIN T3, FAST SKELETAL; TNNT3  
[157550](#) MINICORE MYOPATHY, DOMINANT  
[188580](#) THYROTOXIC PERIODIC PARALYSIS  
[\\*120220](#) COLLAGEN, TYPE VI, ALPHA-1; COL6A1  
[\\*590035](#) TRANSFER RNA, MITOCHONDRIAL, GLYCINE; MTTG  
[\\*171860](#) PHOSPHOFRUCTOKINASE, LIVER TYPE; PFKL  
[#551500](#) NEUROPATHY, ATAXIA, AND RETINITIS PIGMENTOSA  
[\\*208150](#) PENA-SHOKEIR SYNDROME, TYPE I  
[\\*172250](#) PHOSPHOGLYCERATE MUTASE 1; PGAM1  
[\\*182980](#) SPINAL MUSCULAR ATROPHY, PROXIMAL, ADULT TYPE  
[161400](#) NARCOLEPSY  
[123320](#) CREATINE PHOSPHOKINASE, ELEVATED SERUM  
[\\*601226](#) PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA, TYPE 2; PEO2  
[\\*159970](#) MYOGENIC DIFFERENTIATION ANTIGEN 1; MYOD1  
[\\*191041](#) TROPONIN T1, SKELETAL, SLOW; TNNT1  
[#550000](#) MITOCHONDRIAL DNA BREAKAGE SYNDROME, SECONDARY TO NUCLEAR  
MUTATION  
[160990](#) MYOTONIC MYOPATHY WITH CYLINDRICAL SPIRALS  
[\\*601239](#) DYSTROBREVIN, ALPHA; DTNA  
[#535000](#) LEBER OPTIC ATROPHY  
[\\*229300](#) FRIEDREICH ATAXIA 1; FRDA  
[309660](#) MENTAL RETARDATION, SCAPULOPERONEAL MUSCULAR DYSTROPHY, AND  
LETHAL CARDIOMYOPATHY  
[\\*232500](#) GLYCOGEN STORAGE DISEASE IV  
[\\*158600](#) MUSCULAR ATROPHY, JUVENILE SPINAL  
[\\*156600](#) MICROCORIA, CONGENITAL

- [\\*160000](#) MYOGLOBIN; MB
- [\\*160010](#) MYOGLOBINURIA, AUTOSOMAL DOMINANT
- [160570](#) MYOPATHY WITH STORAGE OF GLYCOPROTEINS AND GLYCOSAMINOGLYCANS
- [\\*160100](#) MYOKYMIA
- [\\*123870](#) CYTOCHROME c OXIDASE, SUBUNIT VIII; COX8
- [305550](#) FINGERPRINT BODY MYOPATHY
- [151800](#) LIPOMATOSIS, FAMILIAL BENIGN CERVICAL
- [602541](#) MUSCULAR DYSTROPHY, CONGENITAL, MEGAONIAL TYPE
- [\\*212350](#) CATARACT AND CARDIOMYOPATHY
- [#510000](#) CARDIOMYOPATHY, IDIOPATHIC DILATED, MITOCHONDRIAL
- [\\*160750](#) MYOSITIS
- [\\*233690](#) GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL CYTOCHROME-b-NEGATIVE FORM
- [\\*314555](#) VOLTAGE-DEPENDENT ANION CHANNEL 1; VDAC1
- [160550](#) MYOPATHY, MITOCHONDRIAL, WITH CATARACT
- [\\*601609](#) L-3-@HYDROXYACYL-CoA DEHYDROGENASE, SHORT CHAIN; HADHSC
- [\\*147940](#) ISLET AMYLOID POLYPEPTIDE; IAPP
- [251880](#) MITOCHONDRIAL DNA DEPLETION SYNDROME
- [\\*123590](#) CRYSTALLIN, ALPHA-B; CRYAB
- [#105400](#) AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1
- [\\*160793](#) MYOSIN-BINDING PROTEIN C, FAST TYPE; MYBPC2
- [\\*147450](#) SUPEROXIDE DISMUTASE 1; SOD1
- [\\*601455](#) NEUROPATHY, HEREDITARY MOTOR AND SENSORY, LOM TYPE
- [\\*301830](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, X-LINKED
- [#550500](#) MYOGLOBINURIA, RECURRENT
- [\\*159001](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B; LGMD1B
- [#253400](#) SPINAL MUSCULAR ATROPHY III; SMA3
- [\\*107400](#) PROTEASE INHIBITOR 1; PI
- [\\*221350](#) DEAFNESS, CONGENITAL, WITH VITILIGO AND ACHALASIA
- [#253550](#) SPINAL MUSCULAR ATROPHY II; SMA II
- [#557000](#) PEARSON MARROW-PANCREAS SYNDROME
- [\\*162100](#) NEURITIS WITH BRACHIAL PREDILECTION; NAPB
- [\\*204300](#) CEROID LIPOFUSCINOSIS, NEURONAL 4; CLN4
- [\\*601714](#) TEA DOMAIN FAMILY MEMBER 4; TEAD4
- [#560000](#) RENAL TUBULOPATHY, DIABETES MELLITUS, AND CEREBELLAR ATAXIA DUE TO DUPLICATION OF MITOCHONDRIAL DNA
- [\\*603951](#) POTASSIUM CHANNEL, CALCIUM-ACTIVATED, LARGE CONDUCTANCE, SUBFAMILY M, BETA MEMBER 1; KCNMB1
- [\\*142409](#) HEPATOCYTE GROWTH FACTOR; HGF
- [\\*248500](#) MANNOSIDOSIS, ALPHA B, LYSOSOMAL
- [\\*311800](#) PHOSPHOGLYCERATE KINASE 1; PGK1
- [\\*601592](#) RECEPTOR-ASSOCIATED PROTEIN OF THE SYNAPSE, 43-KD; RAPSN
- [\\*108740](#) ATPase, Ca(2+)-TRANSPORTING, SLOW-TWITCH; ATP2A2

- [\\*604063](#) INTEGRIN, ALPHA-8; ITGA8
- [\\*130160](#) ELASTIN; ELN
- [\\*601792](#) PROTEIN PHOSPHATASE 1, REGULATORY INHIBITOR SUBUNIT 2; PPP1R2
- [#601850](#) RETINITIS PIGMENTOSA-DEAFNESS SYNDROME 1, AUTOSOMAL DOMINANT
- [\\*250100](#) METACHROMATIC LEUKODYSTROPHY
- [\\*102575](#) ACTININ, ALPHA-1; ACTN1
- [255140](#) MYOPATHY WITH GIANT ABNORMAL MITOCHONDRIA
- [\\*164860](#) MET PROTOONCOGENE; MET
- [\\*164920](#) V-KIT HARDY-ZUCKERMAN 4 FELINE SARCOMA VIRAL ONCOGENE  
HOMOLOG; KIT
- [251945](#) MITOCHONDRIAL MYOPATHY WITH A DEFECT IN MITOCHONDRIAL-PROTEIN  
TRANSPORT
- [\\*277320](#) VISCERAL MYOPATHY, FAMILIAL, WITH EXTERNAL OPHTHALMOPLEGIA
- [\\*602067](#) CARDIOMYOPATHY, DILATED, 1F; CMD1F
- [\\*603283](#) TRIADIN
- [#263800](#) POTASSIUM AND MAGNESIUM DEPLETION
- [\\*590085](#) TRANSFER RNA, MITOCHONDRIAL, SERINE, 2; MTT52
- [600091](#) SPINAL MUSCULAR ATROPHY, PROXIMAL, CHILDHOOD-ONSET, AUTOSOMAL  
DOMINANT
- [#268210](#) RHABDOMYOSARCOMA 1; RMS1
- [\\*600963](#) SINE OCULIS HOMEO BOX, DROSOPHILA, HOMOLOG OF, 5; SIX5
- [\\*310440](#) MYOPATHY, X-LINKED, WITH EXCESSIVE AUTOPHAGY; MEAX
- [\\*219800](#) CYSTINOSIS, NEPHROPATHIC; CTNS
- [\\*600890](#) HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA  
THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT; HADHA
- [185070](#) STORMORKEN SYNDROME
- [\\*600937](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 11;  
KCNJ11
- [\\*600308](#) AQUAPORIN 4; AQP4
- [\\*602959](#) EUKARYOTIC TRANSLATION ELONGATION FACTOR 1, ALPHA-2; EEF1A2
- [\\*182920](#) SPHEROID BODY MYOPATHY
- [\\*172400](#) GLUCOSE-6-PHOSPHATE ISOMERASE; GPI
- [#600649](#) HYPOGLYCEMIA, HYPOKETOTIC, WITH DEFICIENCY OF CARNITINE  
PALMITOYLTRANSFERASE II
- [\\*180245](#) RETINOID X RECEPTOR, ALPHA; RXRA
- [\\*181430](#) SCAPULOPERONEAL MYOPATHY; SPM
- [601152](#) PERIPHERAL NEUROPATHY AND OPTIC ATROPHY
- [\\*162660](#) NEUROTROPHIN 3; NTF3
- [162380](#) NEUROPATHY, HEREDITARY SENSORIMOTOR, WITH UPPER MOTOR NEURON,  
VISUAL PATHWAY AND AUTONOMIC DISTURBANCE
- [\\*602943](#) RAR-RELATED ORPHAN RECEPTOR C; RORC
- [\\*601273](#) CLATHRIN, HEAVY POLYPEPTIDE-LIKE 1; CLTCL1
- [#118300](#) CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS
- [\\*602740](#) PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, BETA-1; PRKAB1

[\\*602741](#) PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, BETA-2; PRKAB2  
[159400](#) MYASTHENIA, FAMILIAL LIMB-GIRDLE  
[167320](#) PAGETOID AMYOTROPHIC LATERAL SCLEROSIS  
[#601462](#) MYASTHENIC SYNDROME, SLOW-CHANNEL CONGENITAL; SCCMS  
[153460](#) MCARDLE SYNDROME  
[\\*147420](#) INCLUSION BODY MYOPATHY  
[\\*601643](#) PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), ALPHA; PPP2R5A  
[\\*600837](#) GLIAL CELL LINE-DERIVED NEUROTROPHIC FACTOR; GDNF  
[\\*601645](#) PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), GAMMA; PPP2R5C  
[\\*601652](#) MYOCILIN; MYOC  
[\\*160500](#) MYOPATHY, DISTAL 1; MPD1  
[\\*143450](#) HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA  
THIOLASE/ENOYL-CoA HYDRATASE, BETA SUBUNIT; HADHB  
[\\*131240](#) ENDOTHELIN 1; EDN1  
[\\*157640](#) PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA, TYPE 1; PEO1  
[\\*114206](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA-1D SUBUNIT;  
CACNA1D  
[\\*136850](#) FUMARATE HYDRATASE; FH  
[\\*601954](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2G; LGMD2G  
[\\*131243](#) ENDOTHELIN RECEPTOR, TYPE A; EDNRA  
[\\*131244](#) ENDOTHELIN RECEPTOR, TYPE B; EDNRB  
[\\*600759](#) ALZHEIMER DISEASE, FAMILIAL, TYPE 4  
[\\*133170](#) ERYTHROPOIETIN; EPO  
[\\*182390](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE II, ALPHA-1 SUBUNIT; SCN2A1  
[\\*131360](#) ENOLASE 2; ENO2  
[600706](#) PROXIMAL MYOPATHY WITH FOCAL DEPLETION OF MITOCHONDRIA  
[224550](#) DYSTONIA WITH RINGBINDEN  
[262900](#) PLEOCONIAL MYOPATHY WITH SALT CRAVING  
[271120](#) SPINAL MUSCULAR ATROPHY, DISTAL  
[271150](#) SPINAL MUSCULAR ATROPHY, PROXIMAL, ADULT TYPE  
[\\*176914](#) PROTEIN PHOSPHATASE 1, CATALYTIC SUBUNIT, GAMMA ISOFORM; PPP1CC  
[258470](#) OPHTHALMOPLEGIC NEUROMUSCULAR DISORDER WITH ABNORMAL  
MITOCHONDRIA  
[\\*103050](#) ADENYLOSUCCINATE LYASE; ADSL  
[\\*300021](#) SPINAL MUSCULAR ATROPHY, X-LINKED LETHAL INFANTILE  
[\\*256550](#) NEURAMINIDASE DEFICIENCY  
[\\*114105](#) PROTEIN PHOSPHATASE 3, CATALYTIC SUBUNIT, ALPHA ISOFORM; PPP3CA  
[\\*107254](#) ANTIGEN MIC12 IDENTIFIED BY MONOCLONAL ANTIBODY 30.2A8; MIC12  
[227255](#) FACIAL DYSMORPHISM WITH MULTIPLE MALFORMATIONS  
[109130](#) AXIAL OSTEOMALACIA  
[\\*603033](#) COLLAGENIC TAIL OF ENDPLATE ACETYLCHOLINESTERASE; COLQ  
[\\*192240](#) VASCULAR ENDOTHELIAL GROWTH FACTOR; VEGF  
[255995](#) NATIVE AMERICAN MYOPATHY  
[\\*600528](#) CARNITINE PALMITOYLTRANSFERASE I, LIVER; CPT1A

- [\\*300075](#) RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 3; RPS6KA3
- [\\*102574](#) ACTININ, ALPHA-3; ACTN3
- [\\*310490](#) NEUROPATHY, MOTOR-SENSORY, TYPE II, WITH DEAFNESS AND MENTAL RETARDATION
- [254960](#) MYOPATHY DUE TO MALATE-ASPARTATE SHUTTLE DEFECT
- [\\*603114](#) S100 CALCIUM-BINDING PROTEIN A11; S100A11
- [\\*107970](#) ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 1; ARVD1
- [\\*254780](#) EPILEPSY, PROGRESSIVE MYOCLONIC 2; EPM2A
- [\\*194070](#) WILMS TUMOR 1; WT1
- [#246200](#) LEPRECHAUNISM
- [\\*230400](#) GALACTOSEMIA
- [251950](#) MITOCHONDRIAL MYOPATHY WITH LACTIC ACIDOSIS
- [254190](#) MYASTHENIA, CONGENITAL, REFRACTORY TO ACETYLCHOLINESTERASE INHIBITORS
- [600416](#) MUSCULAR DYSTROPHY, SCAPULOHUMERAL
- [\\*182250](#) SINGLETON-MERTEN SYNDROME
- [#520000](#) DIABETES-DEAFNESS SYNDROME, MATERNALLY TRANSMITTED
- [\\*254110](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2H
- [#205000](#) AMYOTONIA CONGENITA
- [\\*603963](#) INTEGRIN, ALPHA-9; ITGA9
- [\\*516060](#) ATP SYNTHASE 6; MTATP6
- [253590](#) MUSCULAR DYSTROPHY, ADULT-ONSET, WITH LEUKOENCEPHALOPATHY
- [\\*590025](#) TRANSFER RNA, MITOCHONDRIAL, GLUTAMIC ACID; MTTE
- [253320](#) MULTICORE MYOPATHY WITH MENTAL RETARDATION, SHORT STATURE, AND HYPOGONADOTROPIC HYPOGONADISM
- [\\*109150](#) MACHADO-JOSEPH DISEASE; MJD
- [\\*590075](#) TRANSFER RNA, MITOCHONDRIAL, PROLINE; MTPP
- [\\*600175](#) SPINAL MUSCULAR ATROPHY, CONGENITAL NONPROGRESSIVE, OF LOWER LIMBS
- [\\*113503](#) BRADYKININ RECEPTOR B2; BDKRB2
- [\\*590080](#) TRANSFER RNA, MITOCHONDRIAL, SERINE, 1; MTTTS1
- [603280](#) PROGRESSIVE EXTERNAL OPHTHALMOPLÉGIA WITH HYPOGONADISM
- [\\*600362](#) FLIGHTLESS I, DROSOPHILA, HOMOLOG OF; FLII
- [\\*600354](#) SURVIVAL OF MOTOR NEURON 1, TELOMERIC; SMN1
- [\\*171840](#) PHOSPHOFRUCTOKINASE, PLATELET TYPE
- [\\*172430](#) ENOLASE 1; ENO1
- [\\*182520](#) SORCIN; SRI
- [184850](#) STIFF MAN SYNDROME, HEREDITARY FORM OF
- [#176270](#) PRADER-WILLI SYNDROME; PWS
- [\\*600489](#) NUCLEAR FACTOR OF ACTIVATED T CELLS, CYTOPLASMIC, 1; NFATC1
- [\\*600882](#) CHARCOT-MARIE-TOOTH DISEASE, NEURONAL TYPE, B; CMT2B
- [\\*600221](#) TEK TYROSINE KINASE, ENDOTHELIAL; TEK
- [\\*600363](#) SPASTIC PARAPLEGIA 6, AUTOSOMAL DOMINANT; SPG6
- [\\*600163](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE V, ALPHA SUBUNIT; SCN5A

[\\*600576](#) GATA-BINDING PROTEIN 4; GATA4  
[\\*600884](#) CARDIOMYOPATHY, DILATED, 1B; CMD1B  
[\\*602999](#) PROTEIN PHOSPHATASE 1, REGULATORY INHIBITOR SUBUNIT 5; PPP1R5  
[600096](#) PUERTO RICAN INFANT HYPOTONIA SYNDROME  
[#118200](#) CHARCOT-MARIE-TOOTH DISEASE, TYPE 1B; CMT1B  
[\\*188840](#) TITIN; TTN  
[\\*118210](#) CHARCOT-MARIE-TOOTH DISEASE, NEURONAL TYPE, A; CMT2A  
[\\*600794](#) SPINAL MUSCULAR ATROPHY, DISTAL, WITH UPPER LIMB PREDOMINANCE  
[\\*600026](#) SYNTROPHIN, BETA-1; SNT2B1  
[\\*590105](#) TRANSFER RNA, MITOCHONDRIAL, VALINE; MTTV  
[\\*601011](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA 1A SUBUNIT; CACNA1A  
[\\*114213](#) CALDESMON 1; CALD1  
[\\*190450](#) TRIOSEPHOSPHATE ISOMERASE 1; TPI1  
[\\*190930](#) TROPOMODULIN; TMOD  
[\\*603271](#) PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 21; PTPN21  
[600506](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 4; LGMD4  
[\\*166700](#) OSTEOPOROSIS  
[168350](#) PARAMYOTONIA WITHOUT COLD PARALYSIS  
[603376](#) LONG-CHAIN FATTY ACIDS, DEFECT IN TRANSPORT OF  
[601164](#) EXTRAPYRAMIDAL DISORDER, PROGRESSIVE, WITH PRIMARY HYPOGONADISM AND ALOPECIA  
[\\*603377](#) SOLUTE CARRIER FAMILY 22, MEMBER 5; SLC22A5  
[#600257](#) BOR-DUANE HYDROCEPHALUS CONTIGUOUS GENE SYNDROME  
[\\*114760](#) CARBONIC ANHYDRASE IV; CA4  
[#601173](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL RECESSIVE  
[\\*600857](#) SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT A, FLAVOPROTEIN; SDHA  
[#179800](#) RENAL TUBULAR ACIDOSIS, DISTAL, AUTOSOMAL DOMINANT  
[\\*191760](#) URIDYL DIPHOSPHATE GLUCOSE PYROPHOSPHORYLASE 2; UGP2  
[\\*183050](#) SPINOCEREBELLAR ATAXIA WITH RIGIDITY AND PERIPHERAL NEUROPATHY  
[\\*590070](#) TRANSFER RNA, MITOCHONDRIAL, PHENYLALANINE; MTTF  
[\\*602399](#) MITOGEN-ACTIVATED PROTEIN KINASE 12; MAPK12  
[192400](#) VEINS, PATTERN OF, ON ANTERIOR THORAX  
[\\*601569](#) UBIQUITIN-CONJUGATING ENZYME E2G; UBE2G  
[\\*245100](#) KETOACIDURIA WITH MENTAL DEFICIENCY AND OTHER FEATURES  
[164310](#) OCULOPHARYNGODISTAL MYOPATHY  
[\\*163729](#) NITRIC OXIDE SYNTHASE 3; NOS3  
[268950](#) SARCOTUBULAR MYOPATHY  
[\\*602281](#) MILK FAT GLOBULE-EGF FACTOR 8; MFGE8  
[\\*162400](#) NEUROPATHY, HEREDITARY SENSORY, TYPE I; HSN1  
[#194050](#) WILLIAMS-BEUREN SYNDROME; WBS  
[\\*245400](#) LACTIC ACIDOSIS, CONGENITAL INFANTILE  
[\\*602230](#) SH3-BINDING DOMAIN GLUTAMIC ACID-RICH PROTEIN; SH3BGR

- [\\*263570](#) POLYGLUCOSAN BODY DISEASE, ADULT FORM; APBD
- [\\*601557](#) ACETYL-CoA CARBOXYLASE-BETA; ACACB
- [\\*602124](#) DYSTONIA 7, TORSION; DYT7
- [275400](#) TRICHOMEGALY WITH MENTAL RETARDATION, DWARFISM, AND PIGMENTARY DEGENERATION OF RETINA
- [\\*602086](#) ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 3; ARVD3
- [#162300](#) MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB; MEN2B
- [\\*162200](#) NEUROFIBROMATOSIS, TYPE I; NF1
- [118230](#) CHARCOT-MARIE-TOOTH DISEASE, GUADALAJARA NEURONAL TYPE
- [#241200](#) HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA
- [#556500](#) PARKINSON DISEASE
- [\\*134600](#) FANCONI RENOTUBULAR SYNDROME
- [\\*601278](#) FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY REGION GENE 1; FRG1
- [#205400](#) HIGH DENSITY LIPOPROTEIN DEFICIENCY, TANGIER TYPE 1; HDLDT1
- [118301](#) CHARCOT-MARIE-TOOTH DISEASE WITH PTOSIS AND PARKINSONISM
- [271220](#) SPINAL MUSCULAR ATROPHY, SCAPULOPERONEAL
- [\\*601523](#) GROWTH FACTOR RECEPTOR-BOUND PROTEIN 10; GRB10
- [255600](#) MYOSCLEROSIS, CONGENITAL, OF LOWENTHAL
- [\\*603667](#) SOLUTE CARRIER FAMILY 25, MEMBER 12; SLC25A12
- [\\*601865](#) PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE 2; PLOD2
- [\\*300076](#) IMMUNONEUROLOGIC DISORDER, X-LINKED
- [\\*307030](#) HYPERGLYCEROLEMIA
- [\\*601282](#) PLECTIN 1; PLEC1
- [\\*237300](#) HYPERAMMONEMIA DUE TO CARBAMOYLPHOSPHATE SYNTHETASE I DEFICIENCY
- [105500](#) AMYOTROPHIC LATERAL SCLEROSIS-PARKINSONISM/DEMENTIA COMPLEX OF GUAM
- [108110](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA; AMC
- [#520100](#) DIARRHEA, CHRONIC, WITH VILLOUS ATROPHY
- [254200](#) MYASTHENIA GRAVIS; MG
- [\\*300104](#) GDP DISSOCIATION INHIBITOR 1; GDI1
- [\\*236250](#) 5,10-METHYLENETETRAHYDROFOLATE REDUCTASE; MTHFR
- [253850](#) MUSCULAR DYSTROPHY, CONGENITAL, EICHSFELD TYPE
- [\\*309000](#) LOWE OCULOCEREBRORENAL SYNDROME; OCRL
- [\\*211960](#) CAMPTODACTYLY WITH MUSCULAR HYPOPLASIA, SKELETAL DYSPLASIA, AND ABNORMAL PALMAR CREASES
- [\\*601693](#) UNCOUPLING PROTEIN 2; UCP2
- [#502000](#) AGING
- [\\*253900](#) MUSCULAR DYSTROPHY, CONGENITAL, PRODUCING ARTHROGRYPOSIS
- [\\*601659](#) ES1, ZEBRAFISH, HOMOLOG OF
- [\\*129010](#) EARLY GROWTH RESPONSE 2; EGR2
- [\\*601646](#) PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), DELTA; PPP2R5D
- [\\*314850](#) KELL BLOOD GROUP PRECURSOR; XK
- [\\*107910](#) CYTOCHROME P450, SUBFAMILY XIX; CYP19

- [\\*159440](#) MYELIN PROTEIN ZERO; MPZ
- [252320](#) MOTOR NEUROPATHY, PERIPHERAL, WITH DYSAUTONOMIA
- [\\*312170](#) PYRUVATE DEHYDROGENASE COMPLEX, E1-ALPHA POLYPEPTIDE 1; PDHA1
- [\\*603415](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE IX, ALPHA SUBUNIT; SCN9A
- [\\*601647](#) PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), EPSILON; PPP2R5E
- [\\*253310](#) LETHAL CONGENITAL CONTRACTURE SYNDROME
- [#252150](#) MOLYBDENUM COFACTOR DEFICIENCY
- [\\*159050](#) MUSCULAR DYSTROPHY, PSEUDOHYPERTROPHIC, WITH INTERNALIZED CAPILLARIES
- [\\*601644](#) PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B (B56), BETA; PPP2R5B
- [\\*159000](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1A; LGMD1A
- [603936](#) GROWTH/DIFFERENTIATION FACTOR 11
- [\\*602489](#) GAP-ASSOCIATED TYROSINE PHOSPHOPROTEIN, 62-KD
- [\\*102910](#) ATP SYNTHASE, H<sup>+</sup> TRANSPORTING, MITOCHONDRIAL F1 COMPLEX, BETA SUBUNIT; ATP5B
- [\\*223360](#) DOPAMINE BETA-HYDROXYLASE, PLASMA; DBH
- [\\*248700](#) MARDEN-WALKER SYNDROME
- [\\*266250](#) RADICULONEUROPATHY, FATAL NEONATAL
- [310450](#) MYOPATHY, QUADRICEPS
- [\\*602714](#) A DISINTEGRIN AND METALLOPROTEINASE DOMAIN 12; ADAM12
- [601321](#) NEUROFIBROMATOSIS-NOONAN SYNDROME; NFNS
- [248770](#) MARFANOID MENTAL RETARDATION SYNDROME, AUTOSOMAL
- [248810](#) MARINESCO-SJOGREN-LIKE SYNDROME; MSLS
- [227800](#) FANCONI RENOTUBULAR SYNDROME II
- [\\*147545](#) INSULIN RECEPTOR SUBSTRATE 1; IRS1
- [\\*126800](#) DUANE RETRACTION SYNDROME 1; DURS1
- [\\*106180](#) DIPEPTIDYL CARBOXYPEPTIDASE 1; DCP1
- [\\*147440](#) INSULIN-LIKE GROWTH FACTOR I; IGF1
- [#248250](#) HYPOMAGNESEMIA, PRIMARY
- [248010](#) MACROEPIPHYSEAL DYSPLASIA WITH OSTEOPOROSIS, WRINKLED SKIN, AND AGED APPEARANCE
- [\\*231550](#) ACHALASIA-ADDISONIANISM-ALACRIMIA SYNDROME; AAA
- [\\*602668](#) DYSTROPHIA MYOTONICA 2; DM2
- [\\*158300](#) MOUTH, INABILITY TO OPEN COMPLETELY, AND SHORT FINGER-FLEXOR TENDONS
- [\\*158070](#) MONOCLONAL ANTIBODY 4F2, ANTIGEN DEFINED BY; MDU1
- [\\*309583](#) MENTAL RETARDATION, X-LINKED, SNYDER-ROBINSON TYPE
- [\\*603508](#) MYOMESIN 1; MYOM1
- [\\*157700](#) MITRAL VALVE PROLAPSE, FAMILIAL; MVP
- [\\*602532](#) SIX-TWELVE LEUKEMIA GENE
- [157300](#) MIGRAINE
- [\\*138571](#) GLYCOGEN SYNTHASE 2; GYS2
- [\\*602633](#) FOUR-AND-A-HALF LIM DOMAINS 2; FHL2
- [\\*601846](#) MUSCULAR DYSTROPHY, AUTOSOMAL DOMINANT, WITH RIMMED

VACUOLES; MDRV

[\\*604103](#) MYOTILIN

[302803](#) CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY, X-LINKED, WITH APLASIA CUTIS CONGENITA

[\\*270550](#) SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE; SACS

[\\*135700](#) FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL; FEOM

[\\*278300](#) XANTHINURIA, TYPE I

[254300](#) MYASTHENIC MYOPATHY

[\\*300124](#) PSEUDOAUTOSOMAL GTP-BINDING PROTEIN-LIKE

[\\*241150](#) HYPOKALEMIA, FAMILIAL

[\\*102560](#) ACTIN, GAMMA-1; ACTG1

[\\*300036](#) SOLUTE CARRIER FAMILY 6, MEMBER 8; SLC6A8

[\\*276700](#) TYROSINEMIA, TYPE I

[\\*602009](#) CYTOCHROME c OXIDASE, SUBUNIT VIa, POLYPEPTIDE 2; COX6A2

[170600](#) PERIODIC PARALYSIS III

[\\*157655](#) NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 1; NDUFS1

[\\*106150](#) ANGIOTENSIN I; AGT

[105565](#) ANAL SPHINCTER MYOPATHY, INTERNAL

[183020](#) SPINAL MUSCULAR ATROPHY, SEGMENTAL

[\\*160795](#) MYOSIN-BINDING PROTEIN H; MYBPH

[218050](#) CRAMPS, FAMILIAL ADOLESCENT

[#158700](#) MUSCULAR ATROPHY, PROGRESSIVE, WITH AMYOTROPHIC LATERAL SCLEROSIS

[#154700](#) MARFAN SYNDROME; MFS

[\\*234200](#) HALLERVORDEN-SPATZ DISEASE

[\\*230000](#) FUCOSIDOSIS

[\\*247100](#) LIPOID PROTEINOSIS OF URBACH AND WIETHE

[170980](#) PERONEAL NERVE, ACCESSORY DEEP

[230710](#) GANGLIOSIDOSIS, GM2, JUVENILE, A(M)B VARIANT

[\\*182970](#) SPINAL MUSCULAR ATROPHY, FACIOSCAPULOHUMERAL TYPE; FSHSMA

[\\*158580](#) MYOPATHY, DISTAL 2

[159100](#) MUSCULAR HYPOPLASIA, CONGENITAL UNIVERSAL, OF KRABBE

[\\*117550](#) CEREBRAL GIGANTISM

[\\*222745](#) 2,4-DIENOYL-CoA REDUCTASE; DECR

[160200](#) MYOPATHY, CONGENITAL, WITH CRYSTALLINE INTRANUCLEAR INCLUSIONS

[\\*249210](#) MEGACYSTIS-MICROCOLON-INTESTINAL HYPOPERISTALSIS SYNDROME

[249800](#) METACHROMATIC LEUKODYSTROPHY AND AMAUROTIC IDIOCY, COMBINED FEATURES OF

[#249900](#) METACHROMATIC LEUKODYSTROPHY DUE TO DEFICIENCY OF CEREBROSIDE SULFATASE ACTIVATOR

[\\*150330](#) LAMIN A/C; LMNA

[\\*148020](#) KERATIN 19; KRT19

[185480](#) SUPRABULBAR PARESIS, CONGENITAL

[\\*189967](#) TEA DOMAIN FAMILY MEMBER 1; TEAD1

- [\\*107680](#) APOLIPOPROTEIN A-I OF HIGH DENSITY LIPOPROTEIN; APOA1
- [\\*182870](#) SPECTRIN, BETA, ERYTHROCYTIC; SPTB
- [\\*158901](#) FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1B; FSHMD1B
- [219080](#) CUSHING DISEASE, ADRENAL
- [252030](#) MITRAL VALVE PROLAPSE AND OPHTHALMOPLEGIA
- [219090](#) CUSHING DISEASE, PITUITARY
- [\\*123280](#) CREATINE KINASE, BRAIN TYPE; CKB
- [\\*253250](#) MULIBREY NANISM; MUL
- [159700](#) MYOCLONUS AND ATAXIA
- [\\*118946](#) CILIARY NEUROTROPHIC FACTOR RECEPTOR; CNTFR
- [#187300](#) TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER; HHT
- [\\*123290](#) CREATINE KINASE, MITOCHONDRIAL; CKMT
- [182700](#) SPASTIC PARAPLEGIA WITH AMYOTROPHY OF HANDS
- [\\*143100](#) HUNTINGTON DISEASE; HD
- [\\*143040](#) HUMAN LEUKOCYTE ANTIGEN MIC4; MIC4
- [\\*182392](#) SODIUM CHANNEL, NEURONAL TYPE VI, ALPHA SUBUNIT; SCN6A
- [108000](#) ARTERIES, ANOMALIES OF
- [236490](#) HYALINOSIS, SYSTEMIC INFANTILE
- [\\*142600](#) HEXOKINASE 1; HK1
- [\\*182389](#) SODIUM CHANNEL, NEURONAL TYPE I, ALPHA SUBUNIT; SCN1A
- [\\*121014](#) GAP JUNCTION PROTEIN, ALPHA-1; GJA1
- [\\*118508](#) CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, BETA POLYPEPTIDE 3; CHRNB3
- [141405](#) HEMIFACIAL SPASM, FAMILIAL
- [254090](#) MUSCULAR DYSTROPHY, SCLEROATONIC
- [167700](#) PALMOMENTAL REFLEX
- [\\*182310](#) ATPase, Na<sup>+</sup>/K<sup>+</sup> TRANSPORTING, ALPHA-1 POLYPEPTIDE; ATP1A1
- [254120](#) MUSCULAR HYPERTONIA, LETHAL
- [\\*140560](#) HEAT-SHOCK 70-KD PROTEIN 2; HSPA2
- [182210](#) SHPRINTZEN OMPHALOCELE SYNDROME
- [130070](#) EHLERS-DANLOS SYNDROME, PROGEROID FORM
- [#125310](#) DEMENTIA, HEREDITARY MULTI-INFARCT TYPE
- [\\*139139](#) HORMONE RECEPTOR; HMR
- [\\*154276](#) MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 3
- [\\*180903](#) RYANODINE RECEPTOR 3; RYR3
- [\\*160782](#) MYOSIN, LIGHT CHAIN 5, REGULATORY; MYL5
- [\\*208500](#) ASPHYXIATING THORACIC DYSTROPHY; ATD
- [#180849](#) RUBINSTEIN SYNDROME
- [\\*138491](#) GLYCINE RECEPTOR, ALPHA-1 SUBUNIT; GLRA1
- [\\*160775](#) MYOSIN, HEAVY CHAIN 9, NONMUSCLE; MYH9
- [208155](#) ARTHROGRYPOSIS MULTIPLEX CONGENITA WITH WHISTLING FACE
- [\\*138230](#) SOLUTE CARRIER FAMILY 2, MEMBER 5; SLC2A5

- [\\*138140](#) SOLUTE CARRIER FAMILY 2, MEMBER 1; SLC2A1
- [\\*180721](#) ROD OUTER SEGMENT PROTEIN 1; ROM1
- [\\*138130](#) GLUTAMATE DEHYDROGENASE 1; GLUD1
- [\\*208200](#) ARTHROGRYPOSIS-LIKE DISORDER
- [\\*241530](#) HYPOPHOSPHATEMIC RICKETS WITH HYPERCALCIURIA, HEREDITARY; HHRH
- [\\*131195](#) ENDOGLIN; ENG
- [\\*160794](#) MYOSIN-BINDING PROTEIN C, SLOW TYPE; MYBPC1
- [\\*166490](#) SECRETED PHOSPHOPROTEIN 1; SPP1
- [\\*137350](#) GELSOLIN; GSN
- [255900](#) MYXEDEMA
- [\\*180663](#) POLYMERASE II, RNA, SUBUNIT C; POLR2C
- [174400](#) POLYDACTYLY, PREAXIAL I
- [\\*191320](#) UBIQUITIN
- [\\*161015](#) NADH-UBIQUINONE OXIDOREDUCTASE FLAVOPROTEIN 1; NDUFV1
- [\\*256520](#) NEU-LAXOVA SYNDROME; NLS
- [179010](#) PYLORIC STENOSIS, INFANTILE
- [176250](#) POSTERIOR COLUMN ATAXIA
- [108700](#) ATAXIA WITH FASCICULATIONS
- [256851](#) NEUROPATHY, GIANT AXONAL, TUNISIAN FORM
- [\\*256855](#) NEUROPATHY, HEREDITARY MOTOR AND SENSORY, WITH EXCESSIVE MYELIN FOLDING COMPLEX, AUTOSOMAL RECESSIVE
- [256870](#) NEUROPATHY, PAINFUL
- [257300](#) NONDISJUNCTION
- [208081](#) ARTHROGRYPOSIS, DISTAL, WITH MENTAL RETARDATION AND CHARACTERISTIC FACIES
- [108145](#) ARTHROGRYPOSIS WITH OCULOMOTOR LIMITATION AND ELECTRORETINAL ABNORMALITIES
- [\\*101000](#) NEUROFIBROMATOSIS, TYPE II; NF2
- [\\*177400](#) BUTYRYLCHOLINESTERASE; BCHE
- [\\*203700](#) ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS
- [\\*201550](#) ADDUCTED THUMBS SYNDROME
- [\\*152200](#) APOLIPOPROTEIN(a); LPA
- [\\*201475](#) ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF
- [\\*192977](#) VERY LOW DENSITY LIPOPROTEIN RECEPTOR; VLDLR
- [\\*176300](#) TRANSTHYRETIN; TTR
- [\\*176640](#) PRION PROTEIN; PRNP
- [\\*259900](#) OXALOSIS I
- [113600](#) BRANCHIAL CLEFT ANOMALIES
- [\\*100740](#) ACETYLCHOLINESTERASE
- [\\*100735](#) ACETYLCHOLINE RECEPTOR-INDUCING ACTIVITY; ARIA
- [134300](#) FACIAL SPASM
- [\\*100500](#) ACANTHOCYTOSIS WITH NEUROLOGIC DISORDER
- [\\*114130](#) CALCITONIN/CALCITONIN-RELATED POLYPEPTIDE, ALPHA; CALCA

[109500](#) BASILAR IMPRESSION, PRIMARY

\*[108600](#) ATAXIA, SPASTIC

#[192430](#) VELOCARDIOFACIAL SYNDROME

\*[265000](#) PTERYGIUM SYNDROME

\*[192500](#) LONG QT SYNDROME 1

\*[176740](#) PROLIFERATING CELL NUCLEAR ANTIGEN; PCNA

#[225400](#) EHLERS-DANLOS SYNDROME, TYPE VI

\*[131400](#) EOSINOPHILIA, FAMILIAL

[100100](#) ABDOMINAL MUSCLES, ABSENCE OF, WITH URINARY TRACT ABNORMALITY AND CRYPTORCHIDISM

\*[603509](#) MYOMESIN 2; MYOM2

\*[603305](#) POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY H, MEMBER 1; KCNH1

\*[600935](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 8; KCNJ8

\*[600936](#) HYALURONAN-MEDIATED MOTILITY RECEPTOR; HMMR

\*[269700](#) BERARDINELLI-SEIP CONGENITAL LIPODYSTROPHY; BSCL

#[603041](#) MYONEUROGASTROINTESTINAL ENCEPHALOMYOPATHY SYNDROME

#[603034](#) ENDPLATE ACETYLCHOLINESTERASE DEFICIENCY; EAD

\*[603011](#) SPINAL MUSCULAR ATROPHY-RELATED GENE H4F5

[271109](#) SPINAL MUSCULAR ATROPHY WITH MENTAL RETARDATION

[271110](#) SPINAL MUSCULAR ATROPHY WITH MICROCEPHALY AND MENTAL SUBNORMALITY

\*[600855](#) DUAL-SPECIFICITY TYROSINE PHOSPHORYLATION-REGULATED KINASE 1A; DYRK1A

\*[600702](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE VIII, ALPHA SUBUNIT; SCN8A

[602107](#) NEUROPATHY, HEREDITARY THERMOSENSITIVE

\*[600261](#) TENASCIN-XA; TNXA

\*[600264](#) ARGININE VASOPRESSIN RECEPTOR 1B; AVPR1B

[603323](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH CEREBELLAR ATROPHY

[271322](#) SPINOCEREBELLAR DEGENERATION WITH SLOW EYE MOVEMENTS; SDSEM

\*[601010](#) TRANSCRIPTION FACTOR 15; TCF15

\*[603306](#) TRANSCRIPTION FACTOR 21; TCF21

[274240](#) THYROCEREBRORETINAL SYNDROME

#[275000](#) GRAVES DISEASE

\*[603326](#) PROTEIN PHOSPHATASE 1, REGULATORY SUBUNIT 6; PPP1R6

\*[603014](#) PROTEIN KINASE, MITOGEN-ACTIVATED, KINASE 7; PRKMK7

#[551000](#) MYOPATHY, MITOCHONDRIAL, LETHAL INFANTILE; LIMM

\*[602694](#) NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 4; NDUFS4

\*[277100](#) VALINEMIA

\*[602460](#) POU DOMAIN, CLASS 4, TRANSCRIPTION FACTOR 3; POU4F3

\*[278250](#) WRINKLY SKIN SYNDROME; WSS

\*[312600](#) RETINITIS PIGMENTOSA 2; RP2

\*[312080](#) PROTEOLIPID PROTEIN, MYELIN; PLP

#[311150](#) OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA

- [\\*601368](#) DISHEVELLED 3; DVL3
- [\\*601313](#) POLYCYSTIC KIDNEY DISEASE 1; PKD1
- [\\*601240](#) GUANIDINOACETATE METHYLTRANSFERASE; GAMT
- [\\*603768](#) MYOSIN PHOSPHATASE TARGET SUBUNIT 2; MYPT2
- [\\*601205](#) SINE OCULIS HOMEBOX, DROSOPHILA, HOMOLOG OF, 1; SIX1
- [309930](#) MUSCULAR DYSTROPHY, CARDIAC TYPE
- [\\*603602](#) PHOSPHOLIPASE A2, GROUP IVC; PLA2G4C
- [\\*300078](#) NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 1; NDUFA1
- [\\*309845](#) MOESIN; MSN
- [\\*603511](#) MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1D; LGMD1D
- [\\*300091](#) FOS-INDUCED GROWTH FACTOR; FIGF
- [\\*603412](#) PROTEIN KINASE, SERINE/THREONINE, RELATED TO THE MYOTONIC DYSTROPHY PROTEIN KINASE
- [\\*300100](#) ADRENOLEUKODYSTROPHY; ALD
- [\\*603434](#) PHOSPHOPROTEIN ENRICHED IN ASTROCYTES, 15-KD; PEA15
- [\\*603170](#) TEA DOMAIN FAMILY MEMBER 3; TEAD3
- [\\*603060](#) KINESIN FAMILY MEMBER 1C; KIF1C
- [\\*309605](#) MENTAL RETARDATION, X-LINKED, SYNDROMIC 4, WITH CONGENITAL CONTRACTURES AND LOW FINGERTIP ARCHES; MRXS4
- [\\*602454](#) PROTEIN-TYROSINE PHOSPHATASE, RECEPTOR-TYPE, LAMBDA, MOUSE, HOMOLOG OF
- [\\*602413](#) SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT C, INTEGRAL MEMBRANE PROTEIN, 15-KD; SDHC
- [\\*300135](#) ATP-BINDING CASSETTE 7; ABC7
- [\\*602204](#) BICAUDAL-D, DROSOPHILA, HOMOLOG OF, 1; BICD1
- [\\*600694](#) INTERLEUKIN 6 SIGNAL TRANSDUCER; IL6ST
- [600691](#) LONG-CHAIN FATTY ACID TRANSPORT PROTEIN
- [\\*300141](#) PROTEIN PHOSPHATASE 6, CATALYTIC SUBUNIT; PPP6C
- [600721](#) D-2-HYDROXYGLUTARICACIDURIA
- [\\*600663](#) MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE D; MEF2D
- [\\*602121](#) DIAPHANOUS, DROSOPHILA, HOMOLOG OF, 1; DIAPH1
- [601701](#) ARTHROGRYPOSIS AND ECTODERMAL DYSPLASIA
- [\\*300198](#) GLYCOGENIN 2; GYG2
- [600462](#) MYOPATHY WITH LACTIC ACIDOSIS AND SIDEROBLASTIC ANEMIA
- [\\*600438](#) TRANSCRIPTION FACTOR A, MITOCHONDRIAL; TFAM
- [\\*601685](#) RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 2; RPS6KA2
- [#301050](#) ALPORT SYNDROME, X-LINKED; ATS
- [\\*601591](#) PROTEIN KINASE, cGMP-DEPENDENT, TYPE II; PRKG2
- [\\*601531](#) LEUKOTRIENE B4 RECEPTOR; LTB4R
- [\\*600217](#) LISSENCEPHALY GENE 2
- [\\*604001](#) YOTIAO
- [\\*601267](#) CHEMOKINE (C-C) RECEPTOR 2; CCR2
- [\\*601254](#) PROTEIN KINASE, MITOGEN-ACTIVATED, KINASE 6; PRKMK6
- [\\*601084](#) CANCER CACHECTIC FACTOR 1

[\\*603030](#) TOLL-LIKE RECEPTOR 4; TLR4  
[\\*602965](#) FATTY ACID-BINDING PROTEIN 7, BRAIN; FABP7  
[\\*602904](#) MITOGEN-ACTIVATED PROTEIN KINASE 6; MAPK6  
[\\*602527](#) PYRUVATE DEHYDROGENASE KINASE, ISOENZYME 4; PDK4  
[\\*600767](#) GDP-DISSOCIATION INHIBITOR 2; GDI2  
[\\*303630](#) COLLAGEN, TYPE IV, ALPHA-5; COL4A5  
[\\*600661](#) MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE B; MEF2B  
[\\*601365](#) DISHEVELLED 1; DVL1  
[600561](#) SPONDYLOEPIPHYSEAL DYSPLASIA WITH ATLANTOAXIAL INSTABILITY  
[\\*306700](#) HEMOPHILIA A  
[\\*600201](#) AGOUTI SIGNALING PROTEIN; ASIP  
[\\*601517](#) ATAXIN 2; ATX2  
[\\*600276](#) NOTCH, DROSOPHILA, HOMOLOG OF, 3; NOTCH3  
[\\*601656](#) GATA-BINDING PROTEIN 6; GATA6  
[\\*312865](#) SHORT STATURE; SS  
[\\*239500](#) HYPERPROLINEMIA, TYPE I  
[305920](#) GLUTAMYL RIBOSE-5-PHOSPHATE STORAGE DISEASE  
[\\*239000](#) HYPEROSTOSIS CORTICALIS DEFORMANS JUVENILIS  
[\\*306900](#) HEMOPHILIA B; HEMB  
[\\*245349](#) PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X  
[\\*243180](#) INTESTINAL PSEUDOObSTRUCTION DUE TO NEURONAL DISEASE  
[236130](#) HOMOCARNOSINOSIS  
[#304150](#) CUTIS LAXA, X-LINKED  
[238800](#) HYPERMETABOLISM DUE TO DEFECT IN MITOCHONDRIA  
[#303600](#) COFFIN-LOWRY SYNDROME; CLS  
[\\*302910](#) CHLORIDE CHANNEL 4; CLCN4  
[302900](#) CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY AND FRIEDREICH ATAXIA, COMBINED  
[\\*246530](#) LEUKOTRIENE C4 SYNTHASE; LTC4S  
[\\*302802](#) CHARCOT-MARIE-TOOTH NEUROPATHY, X-LINKED RECESSIVE, 3; CMTX3  
[\\*302801](#) CHARCOT-MARIE-TOOTH NEUROPATHY, X-LINKED RECESSIVE, 2; CMTX2  
[#302800](#) CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY, X-LINKED; CMTX1  
[248000](#) MACROCEPHALY  
[\\*307800](#) HYPOPHOSPHATEMIA, VITAMIN D-RESISTANT RICKETS  
[\\*250950](#) 3-METHYLGLUTACONICACIDURIA  
[#307810](#) HYPOPHOSPHATEMIA, HEREDITARY, TYPE II  
[222400](#) DIAPHRAGM, UNILATERAL AGENESIS OF  
[311070](#) OPTIC ATROPHY, POLYNEUROPATHY, AND DEAFNESS  
[308050](#) ICHTHYOSIFORM ERYTHRODERMA, UNILATERAL, WITH IPSILATERAL MALFORMATIONS, ESPECIALLY ABSENCE DEFORMITY OF LIMBS  
[\\*301900](#) BORJESON-FORSSMAN-LEHMANN SYNDROME; BFLS  
[300270](#) ADRENOMYODYSTROPHY  
[\\*311850](#) PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE I; PRPS1

[\\*311860](#) PHOSPHORIBOSYLPYROPHOSPHATE SYNTHETASE II; PRPS2  
[\\*253290](#) MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE  
[\\*236800](#) HYDROXYKYNURENINURIA  
[\\*301500](#) ANGIOKERATOMA, DIFFUSE  
[\\*300300](#) BRUTON AGAMMAGLOBULINEMIA TYROSINE KINASE; BTK  
[\\*308840](#) L1 CELL ADHESION MOLECULE; L1CAM  
[312750](#) RETT SYNDROME; RTT  
[\\*309550](#) FRAGILE SITE MENTAL RETARDATION 1; FMR1  
[#312920](#) SPASTIC PARAPLEGIA, X-LINKED, UNCOMPLICATED  
[#214700](#) CHLORIDE DIARRHEA, FAMILIAL; CLD  
[\\*313650](#) TATA BOX-BINDING PROTEIN-ASSOCIATED FACTOR 2A; TAF2A  
[214380](#) CHARCOT-MARIE-TOOTH DISEASE, PROGRESSIVE ATAXIA, AND TREMOR  
[\\*300201](#) CYSTEINYL LEUKOTRIENE RECEPTOR 1  
[\\*300120](#) F18 GENE  
[\\*212130](#) CARDIOMYOPATHY ASSOCIATED WITH MYOPATHY AND SUDDEN DEATH  
[#212065](#) CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE Ia; CDGS1A  
[\\*300182](#) COFACTOR REQUIRED FOR SP1 TRANSCRIPTIONAL ACTIVATION, SUBUNIT 2, 150-KD; CRSP2  
[\\*300170](#) CHROMOSOME X OPEN READING FRAME 5; CXORF5  
[\\*300145](#) X-PROLYL AMINOPEPTIDASE 2; XPNPEP2  
[210745](#) BLEPHAROPHIMOSIS WITH PTOSIS, SYNDACTYLY, AND SHORT STATURE  
[254000](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH INFANTILE CATARACT AND HYPOGONADISM  
[\\*300139](#) IMMUNOGLOBULIN-BINDING PROTEIN 1; IGBP1  
[\\*300138](#) CHLORIDE INTRACELLULAR CHANNEL 2; CLIC2  
[231970](#) GLUTEAL MUSCLES, ABSENCE OF  
[\\*300137](#) IMMUNOGLOBULIN SUPERFAMILY, MEMBER 1; IGSF1  
[254100](#) MUSCULAR DYSTROPHY, CONGENITAL, WITH RAPID PROGRESSION  
[\\*232000](#) PROPIONICACIDEMIA, TYPE I  
[\\*208800](#) PYRUVATE DECARBOXYLASE DEFICIENCY  
[\\*309801](#) MICROPTHALMIA WITH LINEAR SKIN DEFECTS; MLS  
[\\*206700](#) ANIRIDIA, CEREBELLAR ATAXIA, AND MENTAL DEFICIENCY  
[205250](#) AMYOTROPHIC LATERAL SCLEROSIS WITH POLYGLUCOSAN BODIES  
[258450](#) OPHTHALMOPLEGIA, PROGRESSIVE EXTERNAL  
[\\*231675](#) GLUTARICACIDURIA IIC  
[\\*205100](#) AMYOTROPHIC LATERAL SCLEROSIS 2, JUVENILE; ALS2  
[\\*300118](#) GTPase-ACTIVATING PROTEIN, RHO, 6; ARHGAP6  
[255160](#) MYOPATHY WITH LYSIS OF TYPE I MYOFIBRILS  
[\\*201450](#) ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN; ACADM  
[\\*193500](#) WAARDENBURG SYNDROME, TYPE I; WS1  
[\\*300102](#) GS2 GENE; DXS1283E  
[272480](#) SYRINGOMYELIA  
[\\*300107](#) BOMBESIN-LIKE RECEPTOR 3; BRS3

[#254800](#) MYOCLONIC EPILEPSY OF UNVERRICHT AND LUNDBORG  
[255990](#) NATHALIE SYNDROME  
[\\*300094](#) PROTEIN KINASE C, IOTA FORM; PRKCI  
[\\*300090](#) SIGNAL SEQUENCE RECEPTOR, DELTA; SSR4  
[\\*190231](#) TRANSITION PROTEIN 1; TNP1  
[\\*190180](#) TRANSFORMING GROWTH FACTOR, BETA-1; TGFB1  
[\\*600022](#) PROSTAGLANDIN I2 RECEPTOR; PTGIR  
[\\*255300](#) MYOPATHY, CONGENITAL  
[600027](#) SYNTROPHIN-LIKE; SNTL  
[189600](#) TORTICOLLIS  
[310095](#) MUSCULAR DYSTROPHY, PROGRESSIVE PECTORODORSAL  
[\\*600106](#) INOSITOL POLYPHOSPHATE-5-PHOSPHATASE, 40-KD; INPP5A  
[\\*300070](#) FIBROBLAST GROWTH FACTOR 13; FGF13  
[\\*300056](#) HOLOCYTOCHROME C SYNTHASE; HCCS  
[\\*229600](#) FRUCTOSE INTOLERANCE, HEREDITARY  
[#256000](#) LEIGH SYNDROME; LS  
[228550](#) FIBROMATOSIS, CONGENITAL GENERALIZED; CGF  
[\\*600164](#) GTP-BINDING MITOGEN-INDUCED T-CELL PROTEIN; GEM  
[\\*600167](#) HISTAMINE RECEPTOR H1; HRH1  
[\\*227150](#) ETHANOLAMINOSIS  
[\\*600178](#) MICROTUBULE-ASSOCIATED PROTEIN 1A; MAP1A  
[225740](#) ENCEPHALOPATHY, AXONAL, WITH NECROTIZING MYOPATHY, CARDIOMYOPATHY, AND CATARACTS  
[226350](#) EOSINOPHILIC FASCIITIS  
[\\*600197](#) V-MAF AVIAN MUSCULOAPONEUROTIC FIBROSARCOMA ONCOGENE FAMILY, PROTEIN K; MAFK  
[#256730](#) CEROID LIPOFUSCINOSIS, NEURONAL 1, INFANTILE; CLN1  
[\\*300017](#) FILAMIN A, ALPHA; FLNA  
[\\*186357](#) SYNDECAN 3; SDC3  
[\\*600225](#) GTP CYCLOHYDROLASE I; GCH1  
[\\*600234](#) 3-HYDROXY-3-METHYLGLUTARYL-COENZYME A SYNTHASE 2, MITOCHONDRIAL; HMGCS2  
[\\*600235](#) SODIUM CHANNEL, VOLTAGE-GATED, TYPE I, BETA SUBUNIT; SCN1B  
[\\*600256](#) HYDROCEPHALUS, AUTOSOMAL DOMINANT  
[\\*185490](#) SUPEROXIDE DISMUTASE, EXTRACELLULAR; SOD3  
[\\*300019](#) HOST CELL FACTOR C1; HCFC1  
[\\*311790](#) 6-PHOSPHOFRUCTO-2-KINASE; PFKFB1  
[#600274](#) DISINHIBITION-DEMENTIA-PARKINSONISM-AMYOTROPHY COMPLEX; DDPAC  
[\\*220111](#) LEIGH SYNDROME, FRENCH-CANADIAN TYPE  
[\\*600284](#) ELEVEN NINETEEN LYSINE-RICH LEUKEMIA GENE  
[\\*218040](#) COSTELLO SYNDROME  
[#183900](#) SPONDYLOEPIPHYSEAL DYSPLASIA, CONGENITAL TYPE  
[\\*600317](#) TROPOMYOSIN 4; TPM4  
[600325](#) AMINOPTERIN SYNDROME SINE AMINOPTERIN

[182960](#) SPINAL MUSCULAR ATROPHY, DISTAL  
[182815](#) SPASTIC PARAPLEGIA WITH NEUROPATHY AND POIKILODERMA  
[\\*600337](#) BRADYKININ RECEPTOR B1; BDKRB1  
[\\*300011](#) ATPase, Cu(2+)-TRANSPORTING, ALPHA POLYPEPTIDE; ATP7A  
[\\*182601](#) SPASTIC PARAPLEGIA 4, AUTOSOMAL DOMINANT; SPG4  
[312150](#) PTERYGIUM SYNDROME, X-LINKED  
[\\*182340](#) ATPase, Na<sup>+</sup>/K<sup>+</sup> TRANSPORTING, ALPHA-2 POLYPEPTIDE; ATP1A2  
[\\*218000](#) CORPUS CALLOSUM, AGENESIS OF, WITH NEURONOPATHY  
[#182290](#) SMITH-MAGENIS SYNDROME; SMS  
[#182212](#) SHPRINTZEN-GOLDBERG CRANIOSYNOSTOSIS SYNDROME  
[\\*600434](#) FATTY ACID-BINDING PROTEIN 4, ADIPOCYTE; FABP4  
[\\*208905](#) ATAXIA-TELANGIECTASIA, COMPLEMENTATION GROUP D; ATD; ATDC  
[\\*600447](#) ZIPPER PROTEIN KINASE; ZPK  
[212905](#) CEREBELLAR ATAXIA WITH NEURONAL MIGRATION DEFECT  
[\\*181405](#) AMYOTROPHY, NEUROGENIC SCAPULOPERONEAL, NEW ENGLAND TYPE  
[\\*600477](#) SOLUTE CARRIER FAMILY 9, ISOFORM A5; SLC9A5  
[\\*600480](#) TRANSCRIPTION FACTOR 12; TCF12  
[\\*181400](#) AMYOTROPHY, SCAPULOPERONEAL  
[\\*600497](#) PROTEIN KINASE, AMP-ACTIVATED, CATALYTIC, ALPHA-2; PRKAA2  
[\\*600502](#) IMMUNOGLOBULIN MU BINDING PROTEIN 2; IGHMBP2  
[\\*600504](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 4;  
 KCNJ4  
[\\*180800](#) ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA  
[\\*180247](#) RETINOID X RECEPTOR, GAMMA; RXRG  
[\\*180246](#) RETINOID X RECEPTOR, BETA; RXRB  
[\\*180231](#) RETINOIC ACID-BINDING PROTEIN, CELLULAR, TYPE II; CRABP2  
[\\*600542](#) CHONDROSARCOMA, MYXOID EXTRASKELETAL, FUSED TO EWS; CSMF  
[212750](#) CELIAC DISEASE; CD  
[\\*600563](#) PROSTAGLANDIN F RECEPTOR; PTGFR  
[\\*180220](#) RETINOIC ACID RECEPTOR, BETA; RARB  
[211920](#) CAMPTODACTYLY SYNDROME, GUADALAJARA TYPE II  
[\\*600584](#) CARDIAC-SPECIFIC HOMEO BOX; CSX  
[\\*600615](#) LECTIN, GALACTOSIDE-BINDING, SOLUBLE, 7; LGALS7  
[\\*600616](#) LUMICAN; LUM  
[\\*178600](#) PULMONARY HYPERTENSION, PRIMARY; PPH1  
[\\*178300](#) PTOSIS, HEREDITARY CONGENITAL 1; PTOS1  
[\\*516006](#) COMPLEX I, SUBUNIT ND6; MTND6  
[\\*176912](#) PROTEIN KINASE, cAMP-DEPENDENT, REGULATORY, TYPE II, BETA; PRKAR2B  
[\\*516030](#) COMPLEX IV, CYTOCHROME c OXIDASE SUBUNIT I; MTCO1  
[\\*176894](#) PROTEIN KINASE, cGMP-DEPENDENT, REGULATORY, TYPE I; PRKG1  
[210210](#) BETA-METHYLCROTONYLGLYCINURIA II  
[\\*600681](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 2;  
 KCNJ2

[\\*600682](#) SOLUTE CARRIER FAMILY 16, MEMBER 1; SLC16A1  
[\\*210200](#) BETA-METHYLCROTONYLGLYCINURIA I  
[\\*176885](#) PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 1; PTPN1  
[210000](#) BEHR SYNDROME  
[\\*600696](#) ENOYL-CoA HYDRATASE, PEROXISOMAL; ECH1  
[209010](#) ATHEROSCLEROSIS, PREMATURE, WITH DEAFNESS, NEPHROPATHY, DIABETES MELLITUS, PHOTOMYOCLOSIS, AND DEGENERATIVE NEUROLOGIC DISEASE  
[\\*176876](#) PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR-TYPE, 11; PTPN11  
[\\*176847](#) PROTEASOME SUBUNIT, BETA-TYPE, 10; PSMB10  
[208910](#) ATAXIA-TELANGIECTASIA WITH GENERALIZED SKIN PIGMENTATION AND EARLY DEATH  
[\\*600734](#) POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 5; KCNJ5  
[\\*256731](#) CEROID LIPOFUSCINOSIS, NEURONAL 5; CLN5  
[\\*208900](#) ATAXIA-TELANGIECTASIA; AT  
[\\*600752](#) G PROTEIN-COUPLED RECEPTOR 12; GPR12  
[\\*600758](#) PROTEIN-TYROSINE KINASE, CYTOPLASMIC; PTK2  
[#176500](#) FAMILIAL BRITISH DEMENTIA; FBD  
[\\*257600](#) OCULAR MYOPATHY WITH CURARE SENSITIVITY  
[207740](#) APLASIA OF EXTENSOR MUSCLES OF FINGERS, UNILATERAL, WITH GENERALIZED POLYNEUROPATHY  
[\\*176000](#) PORPHYRIA, ACUTE INTERMITTENT  
[#175200](#) PEUTZ-JEGHERS SYNDROME; PJS  
[\\*600797](#) INSULIN RECEPTOR SUBSTRATE 2; IRS2  
[#174900](#) POLYPOSIS, JUVENILE INTESTINAL  
[\\*174763](#) POLYMERASE, DNA, GAMMA; POLG  
[#257950](#) MUSCULAR DYSTROPHY, OCULOPHARYNGEAL, AUTOSOMAL RECESSIVE  
[\\*173360](#) PLASMINOGEN ACTIVATOR INHIBITOR, TYPE 1; PAI1  
[\\*600828](#) ATP SYNTHASE, H<sup>+</sup> TRANSPORTING, MITOCHONDRIAL F1 COMPLEX, O SUBUNIT; ATP5O  
[\\*173335](#) PHOSPHODIESTERASE I/NUCLEOTIDE PYROPHOSPHATASE 1; PDNPI  
[\\*600840](#) SOLUTE CARRIER FAMILY 12, MEMBER 2; SLC12A2  
[260980](#) PERISYLVIAN SYNDROME, CONGENITAL BILATERAL; CBPS  
[\\*600856](#) CYCLIN-DEPENDENT KINASE INHIBITOR 1C; CDKN1C  
[\\*205200](#) AMYOTROPHIC LATERAL SCLEROSIS, JUVENILE, WITH DEMENTIA  
[\\*600875](#) ACYLPHOSPHATASE, ERYTHROCYTE; ACYP1  
[204730](#) AMINOACIDURIA WITH MENTAL DEFICIENCY, DWARFISM, MUSCULAR DYSTROPHY, OSTEOPOROSIS, AND ACIDOSIS  
[\\*600879](#) NUCLEAR RESPIRATORY FACTOR 1; NRF1  
[\\*172280](#) PHOSPHOGLYCOLATE PHOSPHATASE; PGP  
[\\*171891](#) PHOSPHODIESTERASE 1B  
[#204000](#) AMAUROSIS CONGENITA OF LEBER I  
[\\*171835](#) FRUCTOSE-2,6-BISPHOSPHATASE, CARDIAC ISOZYME; PFKFB2  
[192350](#) VATER ASSOCIATION

[\\*590045](#) TRANSFER RNA, MITOCHONDRIAL, ISOLEUCINE; MTTI  
[\\*171833](#) PHOSPHATIDYLINOSITOL 3-KINASE, REGULATORY, 1; PIK3R1  
[\\*171060](#) P-GLYCOPROTEIN 3; PGY3  
[\\*600933](#) G PROTEIN-COUPLED RECEPTOR 11; GPR11  
[265380](#) PULMONARY HYPERTENSION, FAMILIAL PERSISTENT, OF THE NEWBORN  
[\\*258850](#) ORAL-FACIAL-DIGITAL SYNDROME, TYPE III  
[\\*170390](#) PERIODIC PARALYSIS, POTASSIUM-SENSITIVE CARDIODYSRHYTHMIC TYPE  
[\\*600958](#) MYOSIN-BINDING PROTEIN C, CARDIAC; MYBPC3  
[\\*167420](#) PAIRED MESODERM HOMEODOMAIN BOX 1; PMX1  
[\\*600966](#) LETHAL GIANT LARVAE, DROSOPHILA, HOMOLOG OF, 1; LLGL1  
[\\*258501](#) OPTIC ATROPHY 3; OPA3  
[259610](#) OSTEOLYSIS SYNDROME, RECESSIVE  
[\\*191092](#) TUBEROUS SCLEROSIS 2; TSC2  
[\\*193060](#) VIMENTIN; VIM  
[\\*167040](#) OXYSTEROL BINDING PROTEIN; OSBP  
[192700](#) VENULAR INSUFFICIENCY, SYSTEMIC  
[\\*601018](#) SYNTROPHIN, BETA-2; SNT2B2  
[\\*192315](#) VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY  
[\\*601033](#) LAMININ, ALPHA-5; LAMA5  
[\\*601043](#) INSULIN RECEPTOR GENE TRANSCRIPTION FACTOR 1; IRTF1  
[\\*601047](#) CAVEOLIN 1; CAV1  
[\\*600188](#) AMINO-TERMINAL ENHANCER OF SPLIT; AES  
[\\*190315](#) SOLUTE CARRIER FAMILY 25, MEMBER 1; SLC25A1  
[\\*601092](#) FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 9; FKHL9  
[\\*189971](#) E2F TRANSCRIPTION FACTOR 1; E2F1  
[\\*601102](#) EUKARYOTIC TRANSLATION INITIATION FACTOR 4A, ISOFORM 2; EIF4A2  
[\\*601105](#) CATHEPSIN K; CTSK  
[\\*601116](#) GLUTAMATE RECEPTOR, METABOTROPIC, 8; GRM8  
[\\*601119](#) CLPP PROTEASE  
[\\*601120](#) CADHERIN 5; CDH5  
[\\*601125](#) HEXOKINASE 2; HK2  
[165600](#) ORBITAL MARGIN, HYPOPLASIA OF  
[\\*188061](#) THROMBOSPONDIN II; THBS2  
[\\*165340](#) SKI-LIKE; SKIL  
[601168](#) DIHYDROPYRIMIDINASE-LIKE 3; DPYSL3  
[\\*164795](#) ONCOMODULIN; OCM  
[\\*164500](#) SPINOCEREBELLAR ATAXIA 7; SCA7  
[\\*601176](#) GLUTAMATE-CYSTEINE LIGASE, REGULATORY; GLCLR  
[\\*188060](#) THROMBOSPONDIN I; THBS1  
[\\*601214](#) NAXOS DISEASE  
[\\*601215](#) FRAP-RELATED PROTEIN 1  
[164330](#) ODONTOMA-DYSPHAGIA SYNDROME  
[\\*163730](#) NITRIC OXIDE SYNTHASE 2A; NOS2A

- [\\*600111](#) SOLUTE CARRIER FAMILY 1, MEMBER 3; SLC1A3
- [\\*162641](#) NEUROPEPTIDE Y RECEPTOR Y1; NPY1R
- [600118](#) WARBURG MICRO SYNDROME
- [\\*162332](#) NEUROKININ 3 RECEPTOR; NK3R
- [\\*187380](#) HEXABRACHION; HXB
- [\\*601256](#) ANGIOTENSIN RECEPTOR-LIKE 2; AGTRL2
- [\\*186945](#) FK506-BINDING PROTEIN 1A; FKBP1A
- [\\*162280](#) NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE; NEFL
- [\\*186854](#) SOLUTE CARRIER FAMILY 6, MEMBER 6; SLC6A6
- [\\*160776](#) MYOSIN, HEAVY CHAIN 10, NONMUSCLE; MYH10
- [159950](#) MYOCLONUS, HEREDITARY, WITH PROGRESSIVE DISTAL MUSCULAR ATROPHY
- [\\*159900](#) MYOCLONUS, HEREDITARY ESSENTIAL
- [\\*601299](#) BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE IA; BMPR1A
- [\\*601301](#) PEPTIDYLPROLYL ISOMERASE-LIKE 1; PPIL1
- [\\*600184](#) CARNITINE ACETYLTRANSFERASE; CRAT
- [158800](#) MUSCULAR DYSTROPHY, BARNES TYPE
- [\\*601329](#) LIM DOMAIN KINASE 1; LIMK1
- [\\*601361](#) GROWTH/DIFFERENTIATION FACTOR 10; GDF10
- [271310](#) SPINOCEREBELLAR DEGENERATION AND CORNEAL DYSTROPHY
- [259690](#) OSTEOPENIA AND SPARSE HAIR
- [\\*601373](#) CHEMOKINE (C-C) RECEPTOR 5; CCR5
- [158650](#) MUSCULAR ATROPHY, MALIGNANT NEUROGENIC
- [\\*601398](#) VASCULAR ENDOTHELIAL GROWTH FACTOR B; VEGFB
- [\\*601402](#) MYELOID LEUKEMIA FACTOR 1; MLF1
- [\\*158500](#) MUSCULAR ATROPHY, ATAXIA, RETINITIS PIGMENTOSA, AND DIABETES MELLITUS
- [\\*601413](#) DEIODINASE, IODOTHYRONINE, TYPE II; DIO2
- [155150](#) MEDIAN-ULNAR NERVE COMMUNICATIONS
- [601439](#) SULFONYLUREA RECEPTOR 2
- [\\*601441](#) DIACYLGLYCEROL KINASE, ZETA, 104-KD; DGKZ
- [\\*154870](#) GAMMA-CARBOXYGLUTAMIC ACID PROTEIN, MATRIX; MGP
- [\\*154550](#) MANNOSEPHOSPHATE ISOMERASE; MPI
- [\\*601447](#) UBIQUITIN-SPECIFIC PROTEASE 5; USP5
- [\\*154200](#) MALATE DEHYDROGENASE, SOLUBLE; MDH1
- [\\*154030](#) MAJOR HISTOCOMPATIBILITY COMPLEX, CLASS II, Y BOX-BINDING PROTEIN I
- [\\*601465](#) DEOXYGUANOSINE KINASE; DGUOK
- [\\*601471](#) MOEBIUS SYNDROME 2
- [601477](#) RIBBING DISEASE
- [\\*601485](#) SYNTAXIN 1B; STX1B
- [\\*601487](#) PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR, GAMMA; PPARG
- [\\*601506](#) DIFFERENTIATION 6; DIFF6
- [\\*271245](#) INFANTILE-ONSET SPINOCEREBELLAR ATAXIA; IOSCA

- [\\*153455](#) LYSYL OXIDASE; LOX
- [\\*601524](#) GROWTH FACTOR RECEPTOR-BOUND PROTEIN 14; GRB14
- [\\*600304](#) HYPOKALEMIC PERIODIC PARALYSIS, TYPE II; HOKPP2
- [\\*153430](#) LYMPHOCYTE CYTOSOLIC PROTEIN 1; LCP1
- [\\*601558](#) RNA-BINDING PROTEIN GENE WITH MULTIPLE SPLICING
- [\\*601559](#) SCHWARTZ-JAMPEL SYNDROME, TYPE 2
- [153360](#) LYSOSOMAL GLYCOGEN STORAGE DISEASE WITH NORMAL ACID MALTASE ACTIVITY
- [\\*151750](#) LIPASE, HORMONE-SENSITIVE; LIPE
- [151610](#) LEVATOR-MEDIAL RECTUS SYNKINESIS
- [\\*151385](#) CORE-BINDING FACTOR, RUNT DOMAIN, ALPHA SUBUNIT 2; CBFA2
- [\\*601582](#) INOSITOL POLYPHOSPHATE-5-PHOSPHATASE, 145-KD; INPP5D
- [\\*600346](#) ZINC FINGER PROTEIN 144; ZNF144
- [\\*150800](#) LEIOMYOMA, HEREDITARY MULTIPLE, OF SKIN
- [\\*601599](#) SARCOSPAN; SSPN
- [\\*150325](#) LAMININ, BETA-2; LAMB2
- [601618](#) SOX18, MOUSE, HOMOLOG OF
- [\\*601620](#) T-BOX 5; TBX5
- [\\*147460](#) SUPEROXIDE DISMUTASE 2, MITOCHONDRIAL; SOD2
- [\\*601636](#) HYALURONAN SYNTHASE 2; HAS2
- [\\*601641](#) ACYL-COENZYME A OXIDASE 2, BRANCHED-CHAIN; ACOX2
- [146500](#) HYPOTENSION, ORTHOSTATIC
- [#146200](#) HYPOPARATHYROIDISM, FAMILIAL ISOLATED; FIH
- [#145900](#) HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS
- [\\*145800](#) HYPERTROPHIA MUSCULORUM VERA
- [#144700](#) RENAL CELL CARCINOMA 1; RCC1
- [\\*143850](#) ORTHOSTATIC HYPOTENSIVE DISORDER, STREETEN TYPE
- [\\*182600](#) SPASTIC PARAPLEGIA 3, AUTOSOMAL DOMINANT; SPG3A
- [\\*142983](#) MSH, DROSOPHILA, HOMEO BOX, HOMOLOG OF, 1; MSX1
- [\\*601677](#) NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 5; NDUFA5
- [\\*601684](#) RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 1; RPS6KA1
- [\\*600370](#) PHOSPHATE CARRIER, MITOCHONDRIAL; PHC
- [\\*142704](#) HISTIDINE DECARBOXYLASE; HDC
- [\\*601697](#) PROTEASE INHIBITOR 8, OVALBUMIN TYPE; PI8
- [\\*600581](#) INHIBITOR OF DNA BINDING 4; ID4
- [601708](#) SUPERIOR TRANSVERSE SCAPULAR LIGAMENT, CALCIFICATION OF, FAMILIAL
- [\\*601712](#) APOPTOSIS INHIBITOR 1; API1
- [#142680](#) PERIODIC FEVER, FAMILIAL, AUTOSOMAL DOMINANT
- [601719](#) T-BOX 4
- [\\*601728](#) PHOSPHATASE AND TENSIN HOMOLOG; PTEN
- [\\*601745](#) POTASSIUM CHANNEL, SUBFAMILY K, MEMBER 1; KCNK1
- [\\*601757](#) PEROXISOME BIOGENESIS FACTOR 7; PEX7
- [\\*601766](#) FRIZZLED, DROSOPHILA, HOMOLOG OF, 9; FZD9

- [\\*601770](#) NEUROPEPTIDE Y RECEPTOR Y6; NPY6R
- [601779](#) PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH SEVERE CARDIOMYOPATHY, AUTOSOMAL RECESSIVE
- [\\*601786](#) PHOSPHOMANNOMUTASE 1; PMM1
- [\\*139320](#) GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-STIMULATING ACTIVITY POLYPEPTIDE 1; GNAS1
- [\\*138492](#) GLYCINE RECEPTOR, BETA SUBUNIT; GLRB
- [\\*601807](#) MATRIX METALLOPROTEINASE 19; MMP19
- [\\*601818](#) NONMETASTATIC CELLS 4, PROTEIN EXPRESSED IN; NME4
- [\\*601826](#) DIACYLGLYCEROL KINASE, DELTA, 130-KD; DGKD
- [\\*138350](#) GLUTATHIONE S-TRANSFERASE, MU-1; GSTM1
- [\\*138292](#) GLUTAMINE:FRUCTOSE-6-PHOSPHATE AMIDOTRANSFERASE 1; GFPT1
- [\\*601860](#) 17-@BETA-HYDROXYSTEROID DEHYDROGENASE IV; HSD17B4
- [\\*138000](#) VENOUS MALFORMATIONS WITH GLOMUS CELLS
- [\\*137800](#) GLIOMA OF BRAIN
- [\\*136950](#) PAIRED BASIC AMINO ACID CLEAVING ENZYME; PACE
- [\\*601901](#) SOLUTE CARRIER FAMILY 8, MEMBER 2; SLC8A2
- [\\*601906](#) WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10B; WNT10B
- [\\*601951](#) CDC-LIKE KINASE 1; CLK1
- [\\*136535](#) FORMIN; FMN
- [\\*601958](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-3 SUBUNIT; CACNB3
- [\\*601970](#) VASOACTIVE INTESTINAL PEPTIDE RECEPTOR 2; VIPR2
- [601979](#) HYPERZINCEMIA WITH FUNCTIONAL ZINC DEPLETION
- [\\*136500](#) FOCAL FACIAL DERMAL DYSPLASIA
- [\\*601988](#) LIM DOMAIN KINASE 2; LIMK2
- [\\*601989](#) S100 CALCIUM-BINDING PROTEIN A13; S100A13
- [\\*136435](#) FOLLICLE-STIMULATING HORMONE RECEPTOR; FSHR
- [\\*602012](#) CD39-LIKE 1
- [\\*602020](#) V-MAF AVIAN MUSCULOAPONEUROTIC FIBROSARCOMA ONCOGENE FAMILY, PROTEIN G; MAFG
- [\\*602038](#) DUAL-SPECIFICITY PHOSPHATASE 8; DUSP8
- [\\*135600](#) FIBRONECTIN 1; FN1
- [\\*602054](#) T-BOX 1; TBX1
- [\\*602059](#) IMMUNOGLOBULIN SUPERFAMILY CONTAINING LEUCINE-RICH REPEAT; ISLR
- [\\*135500](#) FIBROMATOSIS, GINGIVAL, WITH ABNORMAL FINGERS, FINGERNAILS, NOSE AND EARS, AND SPLENOMEGALY
- [135400](#) FIBROMATOSIS, GINGIVAL, WITH HYPERTRICHOSIS
- [\\*602087](#) ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 4; ARVD4
- [\\*602100](#) PBX/KNOTTED 1 HOMEODOMAIN BOX 1; PKNOX1
- [271200](#) SPINAL MUSCULAR ATROPHY, RYUKYUAN TYPE
- [\\*600656](#) EUKARYOTIC TRANSLATION ELONGATION FACTOR 1, BETA-3; EEF1B3
- [\\*134797](#) FIBRILLIN 1; FBN1
- [\\*602125](#) CYTOCHROME c OXIDASE, SUBUNIT X; COX10
- [\\*134795](#) FIBRILLARIN; FBL

- [\\*602130](#) MITOGEN-ACTIVATED PROTEIN KINASE-ACTIVATED PROTEIN KINASE 3; MAPKAPK3
- [\\*602141](#) NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 8; NDUFS8
- [\\*602149](#) PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 1; PITX1
- [\\*602150](#) NEURAL CREST TRANSCRIPTION FACTOR SLUG; SLUG
- [\\*602179](#) HEAT-SHOCK 27-KD PROTEIN 2; HSPB2
- [\\*602183](#) BAGPIPE HOMEO BOX, DROSOPHILA, HOMOLOG OF, 1; BAPX1
- [\\*602187](#) ZINC FINGER PROTEIN 195; ZNF195
- [\\*602199](#) MEDIUM CHAIN 3-KETOACYL-COENZYME A THIOLASE DEFICIENCY
- [\\*134570](#) FACTOR XIII, A1 SUBUNIT; F13A1
- [\\*259730](#) OSTEOPETROSIS WITH RENAL TUBULAR ACIDOSIS
- [\\*602223](#) EUKARYOTIC TRANSLATION INITIATION FACTOR 4E-BINDING PROTEIN 1; EIF4EBP1
- [\\*133700](#) EXOSTOSES, MULTIPLE, TYPE I; EXT1
- [\\*602241](#) MITOCHONDRIAL INTERMEDIATE PEPTIDASE; MIPEP
- [\\*602262](#) MATRIX METALLOPROTEINASE 16; MMP16
- [\\*602272](#) TRANSCRIPTION FACTOR 4; TCF4
- [\\*602279](#) POLYADENYLATE-BINDING PROTEIN 2; PABP2
- [\\*133090](#) ERYTHROCYTE SURFACE PROTEIN BAND 7.2; EPB72
- [\\*602292](#) ENOYL-CoA HYDRATASE, SHORT-CHAIN, 1, MITOCHONDRIAL; ECHS1
- [\\*602303](#) p300/CBP-ASSOCIATED FACTOR; PCAF
- [\\*602306](#) RAS-ASSOCIATED PROTEIN RAB2L; RAB2L
- [\\*602315](#) PROTEIN KINASE, MITOGEN-ACTIVATED, KINASE 3; PRKMK3
- [\\*602326](#) RIBOSOMAL PROTEIN L23A; RPL23A
- [\\*602359](#) PHOSPHOLEMMAN; PLM
- [\\*602360](#) L-ARGININE:GLYCINE AMIDINOTRANSFERASE; GATM
- [602361](#) GRACILE BONE DYSPLASIA
- [\\*132800](#) MULTIPLE SELF-HEALING SQUAMOUS EPITHELIOMA; MSSE
- [\\*602374](#) CALPONIN 3; CNN3
- [\\*602375](#) RIBOSOMAL PROTEIN, MITOCHONDRIAL, L12; RPML12
- [\\*602378](#) DYNAMIN 2; DNM2
- [\\*131300](#) ENGELMANN DISEASE
- [\\*602403](#) BLEOMYCIN HYDROLASE; BLMH
- [\\*176590](#) PROFILIN 2; PFN2
- [#130650](#) BECKWITH-WIEDEMANN SYNDROME; BWS
- [\\*130500](#) ERYTHROCYTE MEMBRANE PROTEIN BAND 4.1; EPB41
- [\\*602425](#) MAP/ERK KINASE KINASE 4; MEKK4
- [\\*602426](#) NUCLEAR VALOSIN-CONTAINING PROTEIN-LIKE; NVL
- [602427](#) T-BOX 6; TBX6
- [\\*602438](#) HEAT-SHOCK TRANSCRIPTION FACTOR 4; HSF4
- [\\*602440](#) AMYOTROPHY, MONOMELIC
- [\\*602443](#) X-PROLYL AMINOPEPTIDASE-LIKE; XPNPEPL
- [\\*600750](#) PENTRAXIN II, NEURONAL; NPTX2

- \*[602458](#) SORTILIN; SORT1
- \*[600765](#) MKN28 KINASE
- \*[602461](#) PROTEIN-TYROSINE PHOSPHATASE, NONRECEPTOR TYPE, SUBSTRATE 1; PTPNS1
- \*[602487](#) PERCHLORIC ACID-SOLUBLE PROTEIN, 14.5-KD
- #[128230](#) DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION
- \*[602505](#) PAXILLIN; PXN
- \*[602520](#) PROTEIN KINASE, MITOGEN-ACTIVATED, KINASE 5; PRKMK5
- \*[602521](#) MITOGEN-ACTIVATED PROTEIN KINASE 7; MAPK7
- \*[602525](#) PYRUVATE DEHYDROGENASE KINASE, ISOENZYME 2; PDK2
- \*[602526](#) PYRUVATE DEHYDROGENASE KINASE, ISOENZYME 3; PDK3
- \*[600821](#) ARGININE VASOPRESSIN RECEPTOR 1A; AVPR1A
- #[125853](#) DIABETES MELLITUS, TYPE II
- \*[125597](#) DERMATOPONTIN; DPT
- \*[125305](#) ERYTHROCYTE MEMBRANE PROTEIN BAND 4.9; EPB49
- \*[602569](#) SYNUCLEIN, BETA; SNCB
- \*[602575](#) LIM HOMEODOMAIN BOX TRANSCRIPTION FACTOR 1, BETA; LMX1B
- \*[124060](#) CYTOCHROME P450, SUBFAMILY I, POLYPEPTIDE 2; CYP1A2
- \*[602619](#) C-TERMINAL-BINDING PROTEIN 2; CTBP2
- \*[602623](#) FK506-BINDING PROTEIN 5; FKBP5
- \*[123866](#) CYTOCHROME c OXIDASE, SUBUNIT Vb; COX5B
- \*[602651](#) NARDILYSIN; NRD1
- \*[123740](#) CRYSTALLIN, MU; CRYM
- \*[602676](#) PHOSPHODIESTERASE 6D, cGMP-SPECIFIC, ROD, DELTA; PDE6D
- \*[602679](#) PHOSPHATE CYTIDYLYLTRANSFERASE 2, ETHANOLAMINE; PCYT2
- \*[173321](#) PROTEASE INHIBITOR 6; PI6
- [123540](#) CRYOFIBRINOGENEMIA, FAMILIAL PRIMARY
- \*[123295](#) CREATINE KINASE, SARCOMERIC MITOCHONDRIAL; CKMTS
- \*[602733](#) ALDEHYDE DEHYDROGENASE 9; ALDH9
- \*[122561](#) CORTICOTROPIN-RELEASING HORMONE RECEPTOR 1; CRHR1
- [122470](#) CORNELIA DE LANGE SYNDROME 1; CDL1
- \*[602758](#) PHOSPHATIDYLINOSITOL 4-KINASE, CATALYTIC, BETA; PIK4CB
- \*[120325](#) COLLAGEN, TYPE XV, ALPHA-1; COL15A1
- \*[602774](#) RAD51, S. CEREVISIAE, HOMOLOG OF, C; RAD51C
- \*[602783](#) PARAPLEGIN
- \*[118945](#) CILIARY NEUROTROPHIC FACTOR; CNTF
- \*[118504](#) CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 4; CHRNA4
- \*[602862](#) UDP-N-ACETYLGLUCOSAMINE PYROPHOSPHORYLASE 1; UAP1
- \*[118503](#) CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 3; CHRNA3
- [600878](#) RECEPTOR TYROSINE KINASE NSK2
- \*[602909](#) CLAUDIN 4; CLDN4
- \*[602910](#) CLAUDIN 3; CLDN3

- [602911](#) CALCIUM CHANNEL, VOLTAGE-DEPENDENT NEURONAL, GAMMA SUBUNIT  
\*[602937](#) CBP/p300-INTERACTING TRANSACTIVATOR, WITH GLU/ASP-RICH C-TERMINAL DOMAIN, 2; CITED2  
\*[116930](#) CELL ADHESION MOLECULE, NEURAL, 1; NCAM1  
\*[602958](#) SERUM/GLUCOCORTICOID-REGULATED KINASE; SGK  
\*[115210](#) CARDIOMYOPATHY, FAMILIAL RESTRICTIVE  
\*[171885](#) PHOSPHODIESTERASE 7A, cAMP-SPECIFIC, HIGH-AFFINITY; PDE7A  
\*[602989](#) CDC-LIKE KINASE 2; CLK2  
\*[602990](#) CDC-LIKE KINASE 3; CLK3  
\*[602995](#) UBIQUITIN-CONJUGATING ENZYME E2 VARIANT 1; UBE2V1  
#[115196](#) CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, 3; CMH3  
\*[603004](#) GLIOBLASTOMA AMPLIFIED SEQUENCE; GBAS  
\*[114290](#) CAMPOMELIC DYSPLASIA  
[270805](#) SPASTIC PARAPLEGIA WITH MYOCLONIC EPILEPSY  
\*[600915](#) NESTIN; NES  
\*[603024](#) CHROMOSOME 1 OPEN READING FRAME 4; C1ORF4  
\*[114230](#) CALPAIN, LARGE POLYPEPTIDE L2; CAPN2  
\*[603028](#) TOLL-LIKE RECEPTOR 2; TLR2  
\*[600916](#) INOSITOL POLYPHOSPHATE-4-PHOSPHATASE, TYPE I, 107-KD; INPP4A  
\*[114020](#) CADHERIN 2; CDH2  
\*[264700](#) PSEUDOVITAMIN D DEFICIENCY RICKETS  
#[270400](#) SMITH-LEMLI-OPITZ SYNDROME; SLOS  
[265050](#) PTOSIS OF EYELIDS WITH DIASTASIS RECTI AND HIP DYSPLASIA  
\*[603061](#) ENDOSULFINE, ALPHA; ENSA  
\*[603099](#) LYSOPHOSPHATIDIC ACID ACYLTRANSFERASE-ALPHA  
\*[603102](#) CARBOXYPEPTIDASE D; CPD  
\*[113900](#) BUNDLE BRANCH BLOCK  
\*[603124](#) UBIQUITIN-CONJUGATING ENZYME E2G 2; UBE2G2  
\*[603136](#) CULLIN 3; CUL3  
\*[603142](#) PROTEIN-TYROSINE PHOSPHATASE, RECEPTOR-TYPE, F  
POLYPEPTIDE-INTERACTING PROTEIN-BINDING PROTEIN 2; PPFIBP2  
\*[603145](#) PROTEIN-TYROSINE PHOSPHATASE, RECEPTOR-TYPE, F  
POLYPEPTIDE-INTERACTING PROTEIN ALPHA 4; PPFIA4  
\*[603152](#) ATP SYNTHASE, H<sup>+</sup> TRANSPORTING, MITOCHONDRIAL F0 COMPLEX, SUBUNIT F6; ATP5J  
\*[603168](#) UNC51-LIKE KINASE 1; ULK1  
\*[600976](#) FAT TUMOR SUPPRESSOR (DROSOPHILA) HOMOLOG; FAT  
\*[603171](#) NEURAL PRECURSOR CELL EXPRESSED, DEVELOPMENTALLY  
DOWNREGULATED 8; NEDD8  
\*[113730](#) UNCOUPLING PROTEIN 1; UCP1  
\*[603183](#) RETICULON 2; RTN2  
\*[603235](#) SELENOPROTEIN W, 1; SEPW1  
\*[603264](#) RHOMBOID-LIKE; RHBDL  
\*[603268](#) N-DEACETYLASE/N-SULFOTRANSFERASE 2; NDST2

[110050](#) BLEPHARONASOFACIAL MALFORMATION SYNDROME  
\*[603276](#) REGULATOR OF G PROTEIN SIGNALING 5; RGS5  
[109820](#) BLADDER DIVERTICULUM  
\*[109690](#) BETA-2-ADRENERGIC RECEPTOR; ADRB2  
\*[603298](#) PALMITOYL-PROTEIN THIOESTERASE 2; PPT2  
\*[167055](#) OXYTOCIN RECEPTOR; OXTR  
\*[600997](#) EPHRIN RECEPTOR EphB2; EPHB2  
[603314](#) VERY LONG-CHAIN ACYL-CoA SYNTHETASE-RELATED PROTEIN  
[268700](#) SACCHAROPINURIA  
\*[601017](#) SYNTROPHIN, ALPHA-1; SNT1  
\*[603359](#) NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 8; NDUFA8  
\*[603360](#) PEROXISOME BIOGENESIS FACTOR 16; PEX16  
\*[603369](#) CYCLIN-DEPENDENT KINASE INHIBITOR 2C; CDKN2C  
\*[109580](#) B-CELL TRANSLOCATION GENE 1; BTG1  
\*[109565](#) B-CELL LYMPHOMA 6; BCL6  
[603389](#) OSEBOLD SKELETAL DYSPLASIA/OSTEOLYSIS SYNDROME  
\*[603410](#) FRIZZLED, DROSOPHILA, HOMOLOG OF, 7; FZD7  
\*[601030](#) RIBONUCLEASE A FAMILY, 4; RNASE4  
\*[107741](#) APOLIPOPROTEIN E; APOE  
\*[603427](#) PERIOD, DROSOPHILA, HOMOLOG OF, 3; PER3  
\*[603428](#) ZINC FINGER PROTEIN 207; ZNF207  
\*[601055](#) PLEXIN 1; PLXN1  
\*[603437](#) COLD-SHOCK DOMAIN PROTEIN A; CSDA  
\*[107650](#) APNEA, OBSTRUCTIVE SLEEP  
\*[603493](#) POTASSIUM CHANNEL, SUBFAMILY K, MEMBER 5; KCNK5  
\*[603502](#) INTERFERON-RELATED DEVELOPMENTAL REGULATOR 1; IFRD1  
\*[603504](#) CELL DIVISION CYCLE 14, S. CEREVISIAE, HOMOLOG A; CDC14A  
\*[106165](#) ANGIOTENSIN RECEPTOR 1; AGTR1  
[268500](#) ROWLEY-ROSENBERG SYNDROME  
\*[601097](#) PERIPHERAL MYELIN PROTEIN 22; PMP22  
\*[603519](#) SURVIVAL OF MOTOR NEURON-RELATED PROTEIN  
\*[603539](#) CATHEPSIN F; CTSF  
\*[603593](#) SOLUTE CARRIER FAMILY 7, MEMBER 7; SLC7A7  
\*[603599](#) CASP8- AND FADD-LIKE APOPTOSIS REGULATOR; CFLAR  
[165490](#) OPTIC ATROPHY, DEAFNESS, OPHTHALMOPLEGIA, AND MYOPATHY  
\*[603607](#) RIBOSOMAL PROTEIN S6 KINASE, 90-KD, 5; RPS6KA5  
\*[603621](#) FORKHEAD ACTIVIN SIGNAL TRANSDUCER 1, XENOPUS, HOMOLOG OF  
[106160](#) ANGIOTENSIN II BINDING PROTEIN  
\*[603644](#) CYTOCHROME OXIDASE-DEFICIENT 1, S. CEREVISIAE, HOMOLOG OF; SCOD1  
\*[603645](#) PET112, S. CEREVISIAE, HOMOLOG-LIKE; PET112L  
\*[603646](#) CYTOCHROME c OXIDASE, SUBUNIT 15; COX15  
\*[603665](#) STANNIOCALCIN 2; STC2  
\*[103880](#) ALDEHYDE REDUCTASE 1; ALDR1

- [\\*603679](#) UBIQUITIN-CONJUGATING ENZYME E2N; UBE2N
- [\\*603684](#) LIPASE, ENDOTHELIAL; LIPG
- [\\*103000](#) ADENYLATE KINASE 1; AK1
- [\\*603691](#) GALANIN RECEPTOR 2; GALR2
- [\\*603758](#) GLUTATHIONE S-TRANSFERASE, ZETA-1; GSTZ1
- [\\*603765](#) SYNTAXIN 10; STX10
- [\\*603766](#) MITOCHONDRIAL TRANSLATIONAL INITIATION FACTOR 2; MTIF2
- [\\*601232](#) PHOSPHATIDYLINOSITOL 3-KINASE, CATALYTIC, GAMMA; PIK3CG
- [\\*603770](#) PROTEIN PHOSPHATASE 1B, MAGNESIUM-DEPENDENT, BETA ISOFORM; PPM1B
- [\\*603773](#) CYTOCHROME c OXIDASE, SUBUNIT Va; COX5A
- [\\*603774](#) CYTOCHROME c OXIDASE, SUBUNIT VIIc; COX7C
- [\\*603788](#) POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY G, MEMBER 1; KCNG1
- [\\*603789](#) TRANSCRIPTION ELONGATION FACTOR A-LIKE 1; TCEAL1
- [\\*603796](#) POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 2; KCNE2
- [\\*603797](#) CARBOHYDRATE SULFOTRANSFERASE 1; CHST1
- [\\*603799](#) CARBOHYDRATE SULFOTRANSFERASE 3; CHST3
- [\\*603811](#) BREAKPOINT CLUSTER REGION PROTEIN 1
- [\\*603819](#) STEROID RECEPTOR RNA ACTIVATOR
- [#603830](#) LONG QT SYNDROME 3; LQT3
- [\\*603833](#) NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 4; NDUFA4
- [\\*603850](#) DYNAMIN-RELATED PROTEIN 1
- [\\*603863](#) RING FINGER PROTEIN 7; RNF7
- [\\*603876](#) A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 4; ADAMTS4
- [\\*102776](#) ADENOSINE A2 RECEPTOR; ADORA2A
- [\\*603941](#) SOLUTE CARRIER FAMILY 19, MEMBER 2; SLC19A2
- [\\*102775](#) ADENOSINE A1 RECEPTOR; ADORA1
- [\\*102772](#) ADENOSINE MONOPHOSPHATE DEAMINASE 3; AMPD3
- [\\*102771](#) ADENOSINE MONOPHOSPHATE DEAMINASE 2; AMPD2
- [\\*102720](#) DIPEPTIDYLPEPTIDASE IV; DPP4
- [\\*160994](#) N-ACETYLGLUCOSAMINE RECEPTOR, THYROID
- [\\*604002](#) RHO-ASSOCIATED COILED-COIL-CONTAINING PROTEIN KINASE 2; ROCK2
- [\\*604007](#) XYLAN 1,4-BETA-XYLOSIDASE 1; XBX1
- [\\*604046](#) OXIDATIVE STRESS-RESPONSIVE 1; OSR1
- [\\*604048](#) ORGANIC-CATION TRANSPORTER-LIKE 4; ORCTL4
- [\\*604049](#) XYLULOSE KINASE, H. INFLUENZAE, HOMOLOG OF; XYLB
- [\\*604051](#) ENDONUCLEASE G-LIKE 1; ENDOGL1
- [\\*102642](#) STEROL O-ACYLTRANSFERASE; SOAT
- [\\*604084](#) ZINC FINGER PROTEIN 151; ZNF151
- [\\*604097](#) UROCORTIN 2
- [\\*102576](#) ACTIVIN A RECEPTOR, TYPE I; ACVR1
- [\\*604104](#) GLIA MATURATION FACTOR, GAMMA; GMFG

[604121](#) CEREBELLAR ATAXIA, DEAFNESS, AND NARCOLEPSY

\*[604126](#) SUPERVILLIN; SVIL

\*[604150](#) POLYMERASE II, RNA, SUBUNIT J; POLR2J

\*[604151](#) UBIQUITIN-CONJUGATING ENZYME E2E 3; UBE2E3