Gene	Pathology
AAAS	Achalasia-Addisonism-syndrome
ABCA12	Ichthyosis, autosomal recessive 4B (harlequin)
	Ichthyosis, congenital, autosomal recessive 4A
ABCA3	Dysfunction of surfactant metabolism, pulmonary, 3
ABCA4	Dystrophy 3
	Fundus flavimaculatus
	Age-related macular degeneration 2
	Retinal dystrophy, severe with early onset
	Retinitis pigmentosa 19
	Stargardt disease 1
ABCB11	Cholestasis, recurrent benign intrahepatic, 2
	Cholestasis, progressive familial intrahepatic 2
ABCB4	Cholestasis, intrahepatic, pregnancy, 3
	Cholestasis, intrahepatic familial progressive 3
	Draw the gallbladder 1
ABCB7	Anemia, sideroblastic, with ataxia
ABCC2	Dubin-Johnson syndrome
ABCC6	Arterial calcification, generalized, of childhood, 2
	Pseudoxanthoma elasticum
	Pseudoxanthoma elasticum, stem form
ABCC8	Diabetes mellitus, noninsulin-dependent
	Diabetes mellitus, permanent neonatal
	Diabetes mellitus, transient neonatal 2
	Familial hyperinsulinemic hypoglycemia 1
	Infancy hypoglycemia, sensitive to leucine
ABCD1	Adrenoleukodystrophy
ABCD4	Methylmalonic aciduria and homocystinuria, CBLJ type
ACAD8	Isobutyryl-CoA dehydrogenase deficiency
ACAD9	Acyl coa dehydrogenase deficiency 9
ACADL	Acyl-coa dehydrogenase, long chain
ACADM	Medium-chain acyl-COA dehydrogenase deficiency
ACADS	Acyl coa dehydrogenase, short chain, deficiency
ACADSB	2-methylbutyrylglycinuria
ACADVL	Deficiency of very long chain acyl-CoA dehydrogenase
ACAT1	Ketoacidosis due to beta-ketoolase deficiency

Gene	Pathology
ACE	Angiotensin I converting enzyme, benign serum increase
	Microvascular complications of diabetes 3
	Myocardial infarction, susceptibility to
	Renal tubular dysgenesis
	SARS, progression of
	Stroke, hemorrhagic
ACOX1	Peroxisomal acyl-coa oxidase deficiency
ACSF3	Combined malonic and methylmalonic aciduria
ACSL4	Mental retardation, linked to X 63
ACTN4	Glomerulosclerosis, focal segmental, 1
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency
ADAMTS13	Thrombotic thrombocytopenic purpura, familial
ADAMTS2	Ehlers-Danlos Syndrome, Type of dermatosparaxis
ADAMTSL2	Gelaeophyseal dysplasia 1
ADCK3	Autosomal recessive ataxia due to ubiquinone deficiency
ADGRG1	Polymicrogia, bilateral frontoparietal
	Polymicrogia, bilateral perisylvian
ADK	Hypermethioninemia due to adenosine kinase deficiency
AFF2	Mentally retarded, frax-linked types, fraxe
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease due to deficiency of glycogen debranching enzymes
AGPS	Rhizomelic chondrodysplasia punctata type 3
AGT	Hypertension, essential, susceptibility to
	Preeclampsia, susceptibility to
	Renal tubular dysgenesis
AGTR1	Hypertension, essential
	Renal tubular dysgenesis
AGTR2	Angiotensin II receptor, type 2
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency
AHI1	Joubert syndrome with ocular defect
AIPL1	Cone-rod dystrophy
	Leber congenital Amaurosis 4
	Retinitis pigmentosa, juvenile
AIRE	Autoimmune polyedocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia

Gene	Pathology
ALAS2	Anemia, sideroblastic, 1
	Protoporphyria, erythropoietic, X-linked
ALDH3A2	Sjogren-Larsson syndrome
ALDH4A1	Hyperprolinemia, type II
ALDH5A1	4-hydroxybutyric aciduria
ALDH7A1	Epilepsy, pyridoxine-dependent
ALDOA	Glycogen storage disease XII
ALDOB	Hereditary fructose intolerance
ALG1	Congenital disorder of the IK glycosylation type
ALG12	Congenital disorder of glycosylation, type IG
ALG2	Congenital glycosylation disorder, type II
ALG3	Congenital disorder of glycosylation, type ID
ALG6	Congenital disorder of the IC glycosylation type
ALG8	Congenital glycosylation disorder, type IH
ALG9	Congenital disorder of glycosylation, type IL
ALMS1	Alström syndrome
ALPL	Infantile onset hypophosphatasia
	Infantile hypophosphatasia
ALS2	Amyotrophic lateral sclerosis 2, juvenile
	Primary, juvenile lateral sclerosis
	Spastic paralysis, ascending infantile onset
AMACR	Alpha-methylacyl-CoA racemase deficiency
	Congenital bile acid synthesis defect Type 4
AMPD1	Myopathy due to myoadenylate deaminase deficiency
AMT	Glycine encephalopathy
ANO5	Gnathodiaphysal dysplasia
	Miyoshi muscular dystrophy 3
	Muscular dystrophy, club limbs, autosomal recessive 12
ANTXR2	Hyaline fibromatosis syndrome
AP1S1	Mednik syndrome
AP1S2	Mental retardation, syndromic linked to X 5
AP3B1	Hermansky-pudlak syndrome 2
APTX	Ataxia - Oculomotor Apraxia Type 1
AQP2	Diabetes insipidus, nephrogenic
AR	Complete androgen insensitivity syndrome
	Kennedy's disease

Gene	Pathology
	Partial androgen insensitivity syndrome
ARG1	Argininemia
ARHGEF6	Mental retardation, X-linked 46
ARHGEF9	Epileptic encephalopathy, early infantile, 8
ARL13B	Joubert syndrome 8
ARL6	Bardet-Biedl syndrome 1, modifier of
	Bardet-Biedl syndrome 3
	Retinitis pigmentosa 55
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis type 6
ARSE	Brachytelephalangic chondrodysplasia punctata
ARSF	Arylsulfatase f
ARX	Early infantile epileptic encephalopathy
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASPM	Microcephaly 5, primary, autosomal recessive
ASS1	Citrullinemia Type I.
ATIC	Aica-ribosiduria due to atic deficiency
ATM	Ataxia-telangiectasia
ATP6AP2	Mental retardation, syndromic, syndromic, ivy type
	Parkinsonism with spasticity, X-linked
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA
	Wrinkled skin syndrome
ATP6V1B1	Renal tubular acidosis with deafness
ATP7A	Menkes disease
	Occipital horn syndrome
	X-linked distal spinal muscular atrophy
ATP7B	Wilson's disease
ATP8B1	Cholestasis, benign intrahepatic
	Cholestasis, intrahepatic, pregnancy, 1
	Cholestasis, progressive familial intrahepatic 1
ATR	Seckel's syndrome
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic
	Alpha-thalassemia/mental retardation syndrome
	Facies mental retardation-hypotonic syndrome, X-linked

Gene	Pathology
AUH	3-methylglutaconic aciduria Type 1
B4GALT1	Congenital disorder of the 2D glycosylation type
B9D2	Joubert syndrome 34
	Meckel syndrome 10
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BBS2	Bardet-Biedl syndrome 2
	Retinitis pigmentosa 74
BCHE	Apnea, postanesthetic, susceptibility to, due to BCHE deficiency
	Butyrylcholinesterase deficiency
BCKDHA	Maple syrup urine disease (BCKDHA gene)
BCKDHB	Maple syrup urine disease (BCKDHB gene)
BCOR	Microphthalmia, syndromic 2
BCS1L	Björnstad syndrome
	Gracilis syndrome
	Isolated coq-cytochrome C deficiency
	Leigh's syndrome
BEST1	Bestrophinopathy, autosomal recessive
	Macular dystrophy, vitelliform, 2
	Microcornea, rod cell dystrophy, cataract and posterior staphyloma
	Retinitis pigmentosa, concentric
	Retinitis pigmentosa-50
	Vitreoretinochoroidopathy
BLM	Bloom's syndrome
BRCA2	Breast cancer, male, susceptibility to
	Breast-ovarian cancer, familial, 2
	Fanconi Anemia, Complementation Group D1
	Glioblastoma 3
	Medulloblastoma
	Pancreatic cancer 2
	Prostate cancer
	Wilms tumor
BRIP1	Breast cancer, early onset, susceptibility to
	Fanconi Anemia, Complementation Group J
BRWD3	Mentally retarded, X-linked 93

Gene	Pathology
BSCL2	Encephalopathy, progressive, with or without lipodystrophy
	Lipodycolphy, generalized congenital, Type 2
	Neuropathy, distal hereditary motor, type VA
	Silver spastic paraplegia syndrome
BSND	Bartter Syndrome, Type 4A
	Sensory deafness with mild renal dysfunction
BTD	Biotinidase deficiency
BTK	Isolated growth hormone deficiency Type III
	X-linked agammaglobulinemia
C10orf2	Infantile onset spinocerebellar ataxia
C3	C3 deficiency
	Hemolytic uremic syndrome, atypical, susceptibility to, 5
	Macular degeneration, age-related, 9
CA2	Osteopetrosis with renal tubular acidosis
CANT1	Desbuquois Desplasia 1
	Epiphyseal dysplasia, multiple, 7
CAPN3	Muscular dystrophy, club limbs, autosomal dominant 4
	Muscular dystrophy, club of limbs, autosomal recessive 1
CASK	FG syndrome 4
	Mental retardation and microcephaly with pontine and cerebellar hypoplasia
	Mental retardation, with or without nystagmus
CASP10	Autoimmune lymphoproliferative syndrome, type II
CASQ2	Ventricular tachycardia, Polymorphic catecholaminergic, 2
CBS	Classical homocystinuria
CC2D2A	Coach Syndrome
	Joubert syndrome 9
	Meckel syndrome 6
CCDC103	Ciliary dyskinesia, primary, 17
CCDC39	Ciliary dyskinesia, primary, 14
CD19	Immunodeficiency, common variable, 3
CD247	Immunodeficiency 25
CD2AP	Glomerulosclerosis, focal segmental, 3
CD320	Methylmalonic aciduria, transient, due to defective tranchobalamin receptor
CD3D	Immunodeficiency 19
CD3E	Immunodeficiency 18, SCID variant
CD3G	Immunodeficiency 17, gamma CD3 deficient

Gene	Pathology
CD40LG	X-linked hyper-IgM syndrome
CDH23	DFNB12 autosomal non-syndromic deafness sensorimic type Dfnb12
CDH3	Ectodermal dysplasia, ectrodactyly and macular dystrophy
	Hypotrichosis, congenital, with juvenile macular dystrophy
CDHR1	Cone-rod dystrophy 15
	Retinitis pigmentosa 65
CDK5RAP2	Microcephaly 3, primary, autosomal recessive
CDKL5	Epileptic encephalopathy, early infantile, 2
CENPJ	Microcephaly 6, primary, autosomal recessive
	Seckel syndrome 4
CEP152	Microcephaly 9, primary, autosomal recessive
	Seckel syndrome 5
CEP290	Joubert syndrome with oculorenal defect 5
	Senior syndrome
CERKL	Retinitis pigmentosa 26
CFH	Basal laminar drusen
	Complement factor H deficiency
	Hemolytic uremic syndrome, atypical, susceptibility to, 1
	Age-related macular degeneration 4
CFP	Corrected dyne deficiency, linked to x
CFTR	Cystic fibrosis; mucoviscidosis
CHM	Choroiremia
CHRNA1	Multiple pterygium syndrome, lethal type
	Myostenic syndrome, congenital fast channel
	Myasthenic syndrome, congenital slow canal
CHRND	Multiple pterygium syndrome, lethal type
	Myostenic syndrome, congenital fast channel
	Myasthenic syndrome, congenital slow canal
CHRNE	Myostenic syndrome, congenital, 4a, slow canal
	Myostenic syndrome, congenital, 4b, fast canal
	Myostenic syndrome, congenital, 4c, associated with acetylcholine receptor deficiency
CHRNG	Escobar syndrome
	Multiple pterygium syndrome, lethal type
	Myasthenia gravis, neonatal transient
CHST6	Macular corneal dystrophy
CIITA	Bare lymphocyte syndrome, type II, complementation group A

Gene	Pathology
	Rheumatoid arthritis, susceptibility to
CLCN1	Myotonia Congenita, dominant
	Congenital, recessive myotonia
	Myotonia Levior, recessive
CLCN5	Disease in bruise
	Hypophosphatemic rachits
	Nephrolithiasis, type I
	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis 308990
CLCN7	Autosomal recessive malignant osteopetrosis 4
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia and sclerosing cholangitis 607626
CLDN14	Deafness, autosomal recessive 29
CLDN19	Familial hypomagnesemia - hypercalciuria - nephrocalcinosis - severe ocular involvement
CLN3	Juvenile neuronal ceroid lipofuscinosis 3
CLN5	Late infantile neuronal ceroid lipofuscinosis 5
CLN6	Adult neuronal ceroid lipofuscinosis 4a
	Late infantile neuronal ceroid lipofuscinosis 6
CLN8	Late infantile neuronal ceroid lipofuscinosis 8
	Progressive epilepsy - Intellectual deficits, Finnish type
CLRN1	Usher Syndrome Type 3A
CNGA1	Retinitis pigmentosa 49
CNGA3	Achromatopsia 2
CNGB1	Retinitis pigmentosa 45
CNGB3	Achromatopsia 3
	Macular degeneration, juvenile
COG1	Congenital glycosylation disorder, type IIG
COG7	Congenital glycosylation disorder, type IIE
COG8	Congenital glycosylation disorder, type IIH
COL11A1	Fibhondrogenesis 1
	Lumbar disc herniation, susceptibility to
	Marshall syndrome
	Stickler syndrome, Type II
COL17A1	Generalized junctional epidermolysis bullosa, non-erlitz type
COL18A1	Knobloch Syndrome, Type 1
COL1A1	Caffey's disease
	Ehlers-Danlos syndrome, type I
	Ehlers-Danlos syndrome, type VIIA

Gene	Pathology
	Osteogenesis imperfecta, type I
	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
COL1A2	Ehlers-Danlos syndrome, heart valve type
COL2A1	Achondrogenesis, type II or hypochondrogenesis
	Avascular necrosis of the femoral head
	Czech dysplasia
	Epiphyseal dysplasia, multiple, with myopia and deafness
	Most cited dysplasia
	Legg-calve-perthes disease
	Osteoarthritis with mild chondrodysplasia
	Platyspondyl skeletal dysplasia, Torrance type
	Congenital sed
	Smed Strudwick Type
	Spondyloepiphyseal dysplasia, Stanescu type
	Spondyloperipheral dysplasia
	Stickler Sydrome, type I, ocular nonsyndromic
	Stickler syndrome, Type I
	Vitreoretinopathy with epiphyseal phalangeal dysplasia
COL4A3	Autosomal Recessive Alport Syndrome (COL4A3 Gene)
COL4A4	Autosomal Recessive Alport Syndrome (COL4A4 Gene)
COL4A5	Alport syndrome
COL6A1	Bethlem myopathy
	Ullrich congenital muscular dystrophy
COL6A2	Bethlem myopathy
	Ullrich congenital muscular dystrophy
COL6A3	Bethlem myopathy
	Ullrich congenital muscular dystrophy
COL7A1	Dystrophic epidermolysis bullosa pruriginosa
	Severe recessive generalized dystrophic epidermolysis bullosa
COL9A1	Epiphyseal dysplasia, multiple, 6
	Stickler syndrome, Type IV
COL9A2	Epiphyseal dysplasia, multiple, 2
	Stickler syndrome, Type V
COQ2	Leigh syndrome with nephrotic syndrome

Gene	Pathology
COQ9	Coenzyme Q10 deficiency, primary, 5
COX10	Leigh syndrome due to mitochondrial Cox4 deficiency
	Mitochondrial complex IV deficiency
COX15	Cardioencephalomyopathy, fatal in childhood, due to cytochrome 2 oxidase C deficiency
	Leigh syndrome due to cytochrome C oxidase deficiency
COX6B1	Mitochondrial complex IV deficiency
CPS1	Carbamoyl phosphate synthetase deficiency
CPT1A	Carnitine palmitoyltransferase 1a deficiency
CPT2	Carnitine palmitoyl Transferase II deficiency, infantile form
	Carnitine palmitoyl transferase II deficiency, neonatal form
CRB1	Leber congenital Amaurosis 8
	Pigmented paravenous chorioretinal atrophy
	Retinitis pigmentosa-12
CRLF1	Cold-induced sweating syndrome
CRTAP	Osteogenesis imperfecta type VII
CRX	Dystrophy-2 retina-2 cone-rod
	Leber congenital Amaurosis 7
CSTB	Unverricht-Lundborg disease
CTH	Cystathioninuria
	Homocysteine, total plasma, elevated
CTNS	Cystinosis
CTSC	Haim-Munk syndrome
	Papillon-Lefevre syndrome
	Periodontitis 1, juvenile
CTSD	Adult neuronal ceroid lipofuscinosis 10
CTSK	Pycnodysostosis
CUL4B	Mental retardation, syndromic, syndromic X (Cabeza type)
CYBA	Chronic, autosomal granulomatous disease due to CYBA deficiency
CYBB	Chronic, X-linked granulomatous disease
	Immunodeficiency 34, mycobacteriosis, X-ligands
CYP11A1	Adrenal insufficiency, congenital, with sexual reversal 46xy, partial or complete
CYP11B1	Adrenal hyperplasia, congenital, due to 11 beta-hydroxylase deficiency
	Aldosteronism, remedy with glucocorticoid
CYP11B2	Aldosterone to Renin ratio raised
	Hypoaldosteronism, congenital, due to cMO I deficiency
	Hypoaldosteronism, congenital, due to CMO II deficiency

Gene	Pathology
	Low-renin hypertension, susceptibility to
CYP17A1	17-alpha-hydroxylase/17,20 deficiency
CYP19A1	Aromatase deficiency
	Aromatase excess syndrome
CYP1B1	Anterior segment dysgenesis 6, multiple subtypes
	Glaucoma 3A, primary open angle, congenital, juvenile or adult
CYP27A1	Cerebrotenndous xanthomatosis
CYP27B1	VITAMIN D-dependent RICKET, Type I
CYP4V2	Bietti Cristalline corneoretinal Dystrophy
CYP7B1	Defect in bile acid synthesis, congenital, 3
	Spastic paraplegia 5a, autosomal recessive
D2HGDH	D-2-hydroxyglutaric aciduria
DBT	Classic maple syrup urine disease
DCLRE1C	Omenn syndrome
	Severe combined immunodeficiency due to DCLRE1c deficiency
DCX	Lissencephaly, X-linked
	Subcortical, X-linked laminal heteropia
DDB2	Xeroderma pigment complementation group E
DDC	Aromatic L-amino acid decarboxylase deficiency
DFNB59	Deafness, autosomal recessive 59
DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK 3 deficiency
DHCR24	Desmosterosis
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Congenital glycosylation disorder, type 1BB
	Developmental delay and seizures with or without movement abnormalities
	Retinitis pigmentosa 59
DKC1	X-linked congenital dyskatosis
	Hoyeraal-Hreidarsson syndrome
DLD	Leigh's syndrome
	Maple syrup urine disease
DLG3	Mentally retarded, linked to X 90
DLL3	Autosomal recessive spondylocostal dysostosis 1
DMD	Becker Muscular Dystrophy
	Duchenne muscular dystrophy
DMP1	Autosomal recessive hypophosphatemic rachits 1
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus

Gene	Pathology
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus
DNAJC19	Dilated cardiomyopathy with ataxia
DNAL1	Ciliary dyskinesia, primary, 16
DNMT3B	Centromeric instability anomalies immunodeficiency syndrome 1
DOCK8	Recurrent hyper-IGE infection syndrome, autosomal recessive
DOK7	Deformation sequence of Fetal Akinesia
	Myasthenia, girle of the limbs, family members
DOLK	Congenital glycosylation disorder, MI type
DPAGT1	Congenital glycosylation disorder type 1J
DPM1	Congenital glycosylation disorder type 1E
DPYD	Dihydropyrimidine dehydrogenase deficiency
DSP	Lethal acantholytic epidermolysis bullosa
DUOX2	Thyroid dyshormonogenesis 6
DUOXA2	Thyroid dyshormonogenesis 5
DYNC2H1	Short kibble thoracic dysplasia 3 with or without polydacty
DYSF	Miyoshi muscular dystrophy 1
	Muscular dystrophy, club limbs, autosomal recessive 2
	Myopathy, distal, with anterior tibial onset
EDA	Muscular dystrophy, club limbs, autosomal recessive 2
	Ectodermal dysplasia 1, hypohidrotic, X-linked
	Tooth agenesis, selective, X-linked 1
EDAR	Ectodermal dysplasia 10a, hypohidrotic/hair/nail type, autosomal dominant
	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive
	Hair morphology 1, hair thickness
EDN3	Waardenburg-Shah 4B syndrome
EDNRB	ABCD syndrome
	Waardenburg-Shah syndrome 4A
EFEMP2	Cutis laxa, autosomal recessive, type Ib
EFNB1	Craniofrontonasal dysplasia
EGR2	Charcot-Marie-tooth disease type 4e
EIF2AK3	Wolcott-Rallison syndrome
EIF2B5	Leukoencephalopathy with escaping white matter
	Ovarioleukodystrophy
ELK1	Member of the ETS Oncogene family
EMD	Muscular dystrophy 1, X-linked.

Gene	Pathology
ENO3	Glycogen storage disease XIII
ENPP1	AUTOSOMAL RICKET Recessive hypophosphatemics 2
EPM2A	Epilepsy, Progressive Myoclonic 2A (Lafora)
ERBB3	Lethal congenital contractual syndrome 2
ERCC2	Xeroderma Pigmentosum/Cockayne Syndrome Complementation complex D.
ERCC3	Xeroderma Pigmentosum/Cockayne Syndrome Complementation Complex Group
ERCC4	Xeroderma Pigmentosum/Cockayne Syndrome Complementation complex F F F F
ERCC5	Xeroderma Pigmentosum Syndrome/Cockayne Syndrome Complex Complementation Group G
ERCC6	Cockayne syndrome type B
	COFS Syndrome 1
ERCC8	Cockayne syndrome Type A
ESCO2	Roberts syndrome
ESPN	Deafness, autosomal recessive 36
	Deafness, sensorineural, without vestibular involvement, autosomal dominant
ESRRB	Deafness, autosomal recessive 35
ETFA	Glutaric Acidemia Type 2 (ETFA Gene)
ETFB	Glutaric acidemia type 2 (ETB gene)
ETFDH	Glutaric acidemia type 2 (ETFDH gene)
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-Van Creveld syndrome
	Weyers acrodental dysostosis
EVC2	Ellis-Van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
EYS	Retinitis pigmentosa 25
F11	Factor XI deficiency, autosomal dominant
	Factor XI Deficiency, autosomal recessive
F2	Dysprothrombinemia
	Hypoprothrombinemia
	Pregnancy loss, recurrent, susceptibility to, 2
	Stroke, ischemic, susceptibility to
	Thrombophilia due to thrombin defect
F5	Budd-Chiari syndrome
	Factor V deficiency
	Pregnancy loss, recurrent, susceptibility to, 1
	Stroke, ischemic, susceptibility to

Gene	Pathology
	Thrombophilia due to resistance to activated protein C
	Thrombophilia, susceptibility to, due to factor v leiden
F8	Hemophilia a
F9	Hemophilia b
FAH	Tyrosinemia Type 1
FAM126A	Hypomyelination - congenital cataract
FAM161A	Retinitis pigmentosa 28
FAM20C	Lethal osteosclerotic bone dysplasia
FANCA	Fanconi anemia, complementation group A
FANCB	Fanconi Anemia, Complementation Group B
FANCC	Fanconi anemia complementation group C.
FANCD2	Fanconi Anemia, Complementation Group D2
FANCE	Fanconi Anemia, Complementation Group E
FANCG	Fanconi Anemia, Complementation Group G
FANCI	Fanconi Anemia, Complementation Group I
FANCL	Fanconi Anemia, Complementation Group L
FANCM	Premature ovarian failure 15
	Spermatogenic insufficiency 28
FAS	Autoimmune lymphoproliferative syndrome, type IA
FASLG	Autoimmune lymphoproliferative syndrome, type IB
FASTKD2	Mitochondrial complex IV deficiency
FBLN5	Cutis Laxa, autosomal dominant 2
	Cutis laxa, autosomal recessive, type IA
	Age-related macular degeneration 3
FERMT3	Deficiency of leukocyte adhesion, type III
FGA	Congenital fibrinogen deficiency (FGA gene)
FGB	Afibrinogenemia, congenital/hypofibrinogenemia, congenital
	Dysfibrinogenemia, congenital
FGD1	Aarskog-Scott syndrome
	Mental retardation, X-linked syndromic 16
FGD4	Charcot-Marie-tooth type 4h disease
FH	Fumaric aciduria
FHL1	Uruguay Faciocardiomusculoskeletal Syndrome
	Emery-Dreifuss muscular dystrophy 6, X-linked
	Myopathy, X-linked, with postural muscle atrophy
	Reduce body myopathy, X-linked 1A, severe, infantile or early childhood onset

Gene	Pathology
	Reduce body myopathy, X-linked 1b, with late childhood or adult onset
	Scapuloperoneal myopathy, X-linked dominant
FIG4	Amyotrophic lateral sclerosis 11
	Charcot-Marie-Tooth Disease, Type 4J
	Polymicrogia, bilateral tempotoocpipital
	Yunis-Varon syndrome
FKRP	Autosomal recessive muscle dystrophy type 2i type 2i
	Congenital muscular dystrophy type 5b
	Muscle-eye-brain disease
FKTN	Autosomal recessive muscular dystrophy Type 2M
	Congenital muscular dystrophy type 4b
	Fukuyama congenital muscular dystrophy
FLNA	Cardiac valvular dysplasia, x-linked
	Congenital short bowel syndrome
	FG syndrome 2
	Frontometaphysal dysplasia 1
	Heterotopia, periventricular, 1
	Intestinal, neuronal pseudoobstruction
	Melnick-Needles syndrome
	Otopalatodigital syndrome, type I
	Otopalatodigital syndrome, type II
	Terminal bone dysplasia
FLVCR1	Ataxia, posterior column, with retinitis pigmentosa
FMR1	Fragile X syndrome
	Fragile x tremor/ataxia syndrome
	Premature ovarian failure 1
FOLR1	Neurodegeneration due to deficiency of cerebral folate transport
FOXG1	Rett syndrome, congenital variant
FOXN1	Severe T-cell immunodeficiency - Congenital alopecia - nail dystrophy
FOXP3	Immunodysregulation, polyedocrinopathy and entheopathy, X-linked
FRAS1	Fraser Syndrome (FRAS1 Gene)
FREM2	Fraser Syndrome (FRAS2 Gene)
FTCD	Glutamate formiminotransferase deficiency
FTSJ1	Mentally retarded, X-linked 9
FUCA1	Fucosidosis
FXN	Friedreich's ataxia

Gene	Pathology
	Friedreich's ataxia with retained reflexes
G6PC	Glycogen storage disease due to glucose-6-phosphatase type 1A deficiency
G6PC3	Dursun syndrome
G6PD	Favism
	Hemolytic anemia due to G6PD deficiency
GAA	Glycogen storage disease due to acid maltase deficiency
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis IVA
GALNT3	Tumor calcinosis, hyperphosphatemic, familial, 1
GALT	Classic galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GAN	Giant axonal neuropathy-1
GBA	Fetal gaucher disease
	Type 2 Gaucher disease
	Type 3 Gaucher disease
	Gaucher disease Type 3C
GBE1	Glycogen storage disease due to deficiency of branched glycogen enzymes, infantile combined hepatic and myopathic form
GCDH	Glutaryl-CoA dehydrogenase deficiency
GCH1	Dystonia, dopa sensitive, with or without hyperphenylalaninemia
	Hyperphenylalaninemia, with BH4 deficiency, b
GCSH	Glycine encephalopathy
GDAP1	Autosomal dominant Charcot-Marie-Marco disease Type 2K
	Autosomal recessive carcotta-marina disease with hoarseness
	Autosomal Intermediate Recessive Charcot-Marie-Tooth-Tooth Type A
	Charcot-Marie-tooth disease type 4a
GDF5	Acromesomelic dysplasia, Hunter-Thompson type
	Brachydactyly, type A1, c
	Brachydactyly, type A2
	Brachydactyly, type C
	Chondrodysplasia, Grebe type
	Du Pan syndrome
	Multiple synostosis syndrome 2
	Osteoarthritis-5

Gene	Pathology
	Symphalangism, proximal, 1b
GDI1	Mental retardation, X-linked 41
GFM1	Hepaenencephalopathy due to combined deficiency of type 1 oxidative phosphorylation
GHRHR	Growth hormone deficiency, isolated, type IV
GJA1	Atrioventricular septal defect 3
	Craniometaphysal dysplasia, autosomal recessive
	Erithrokeratodermia varias et Progressiva 3
	Hypoplastic left heart syndrome 1
	Oculodentodigital dysplasia
	Oculodentodigital dysplasia, autosomal recessive
	Palmoplantar keratoderma with congenital alopecia
	Syndyly, type III
GJB1	Carcotta-maritime neuropathy, X-linked dominant, 1
GJB2	Autosomal Recessive Non-Syndromic Sensory Deafness DFNB1A (GJB2 Gene)
GJB3	Deafness, autosomal dominant 2b
	Deafness, autosomal dominant, with peripheral neuropathy
	Deafness, autosomal recessive
	Deafness, digenic, gjb2/gjb3
	Erithrokeratodermia varias et progressivea 1
GJB6	Deafness, autosomal dominant 3b
	Deafness, autosomal recessive 1b
	Deafness, digenic gjb2/gjb6
	Ectodermal dysplasia 2, Clouston type
GJC2	Pelizaeus-Merzbacher due to the GJC2 mutation
GK	Glycerol kinase deficiency
GLA	Fabry disease
GLB1	GM1 Gangliosidosis Type 1
	GM1 Gangliosidosis Type 2
	GM1 Gangliosidosis Type 3
	Mucopolysaccharidosis Type 4B
GLDC	Glycine encephalopathy
GLE1	Lethal congenital contracture syndrome Type 1
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism
GM2A	GM2 gyniosidosis, AB variant
GNAS	ACTH-independent macronodular adrenal hyperplasia
	McCune-Albright syndrome, somatic, mosaic

Gene	Pathology
	Bony heteroplasia, progressive
	Pituitary adenoma 3, multiple types, somatic
	Pseudohypoparathyroidism ia
	Pseudohypoparathyroidism ib
	Pseudohypoparathyroidism IC
	Pseudopohypoparathyroidism
GNE	Nonaka myopathy
	Sialiaria
GNMT	Glycine N-methyltransferase deficiency
GNPTAB	Mucolipidosis type 2
	Mucolipidosis type 3
GNPTG	Mucolipidosis III Gamma
GNRHR	Fertile eunuch syndrome
	Hypogonadotropic hypogonadism 7 without anosmia
GNS	Mucopolysaccharidosis type IIID
GORAB	Geroderma osteodysplasticum
GP1BA	Bernard-Soulier syndrome, type A1 (recessive)
	Bernard-Soulier syndrome, type A2 (dominant)
	Non-arteritic anterior ischemic optic neuropathy, susceptibility to
	Von Willebrand's disease, platelet type
GP1BB	Bernard-Soulier syndrome, type B
	Giant platelet disorder, isolated
GP9	Bernard-Soulier syndrome, type C
GPC3	Simpson-Golabi-Behmel Syndrome, Type 1
GPR143	Nystagmus 6, congenital, X-linked
	Ocular albinism, type I, clear type
GPR179	Night blindness, congenital stationary (complete), 1E, autosomal recessive
GPR98	Usher Syndrome Type 2C
GRHPR	Hyperoxaluria, primary, type II
GRIA3	Mentally retarded, X-linked 94
GRIK2	Mental retardation, autosomal recessive, 6
GRM6	Night blindness, congenital stationary (complete), 1b, autosomal recessive
GRXCR1	Deafness, autosomal recessive 25
GSS	Glutathione synthetase deficiency with 5-oxoprolinia
GTF2H5	Trichothiodiophycy, complementation group A
GUCY2D	Choroidal dystrophy, central areolar 1

Gene	Pathology
	Cone-rod dystrophy 6
	Congenital Leber Amaurosis 1
GUSB	Mucopolysaccharidosis type 7
HADH	Long-chain 3-hydroxyl-CoA dehydrogenase deficiency
HADHA	Protein deficiency
HADHB	Protein deficiency
HAL	Histidinemia
HAMP	Histidinemia
HAX1	Neutropenia, severe congenital 3, autosomal recessive
HBA1	Alpha-thalassemia
HBA2	Erythrocytosis 7
	Heinz body anemia
	Hemoglobin H disease, deletional and non-elective
	Thalassemia, alpha-
HBB	Beta-thalassemia
	Sickle cell anemia
HCCS	Linear skin defects with multiple congenital anomalies 1
HESX1	Combined pituitary hormone deficiencies, genetic forms
HEXA	Tay-Sachs disease
HEXB	Sandhoff's disease
HFE	Alzheimer's disease, susceptibility to
	Hemochromatosis
	Microvascular complications of diabetes 7
	Porphyria cutaneatata, susceptibility to
	Variegated porphyria, susceptibility to
	Transferrin Serum level QTL2
HFE2	Hemochromatosis, type 2A
HGD	Alkaptonuria
HGF	Deafness, autosomal recessive 39
HGSNAT	Sanfilippo Syndrome Type C
HIBCH	Neurodegeneration due to hydroxyisobutyryl-CoA hydrolase deficiency
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaric aciduria
HMOX1	Heme oxygenase-1 deficiency
	Pulmonary disease, chronic obstructive, susceptibility to
HOGA1	Hyperoxaluria, primary, type III

Gene	Pathology
HP	Anhaptoglobinemia
	Hypohaptoglobinemia
HPD	Tyrosinemia Type 3
HPRT1	Kelley-Seegmiller syndrome
	Lesch-Nyhan syndrome
HPS1	Hermansky-pudlak syndrome 1
HPS3	Hermansky-pudlak syndrome 3
HSD11B2	Apparent mineralocorticoid excess
HSD17B1 0	17 beta-hydroxysteroid dehydrogenase x
HSD17B3	Pseudohermaphroditism, male, with gynecomastia
HSD17B4	Deficiency of bifunctional enzymes
	Perrault syndrome
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
HSPD1	Leucodystrophy, hypomyelinating, 4
	Spastic paraplegia 13, autosomal dominant
HSPG2	Schwartz-Jampel syndrome
HTRA1	Carasil syndrome
	Cerebral arteriopathy, autosomal dominant, with subcortical infarction and leukoencephalopathy, type 2
	Age-related macular degeneration 7
	Age-related macular degeneration, neovascular type
HUWE1	Mental retardation, X-linked syndrome, Turner type
HYAL1	Mucopolysaccharidosis type IX
HYLS1	Hydroletus syndrome
ICOS	Immunodeficiency, common variable, 1
IDH3B	Retinitis pigmentosa 46
IDS	Mucopolysaccharidosis type 2
IDUA	Mucopolysaccharidosis IH
	Mucopolysaccharidosis ih/s
	Mucopolysaccharidosis is
IFNGR1	Immunodeficiency 27a, mycobacteriosis, AR
IFNGR2	Immunodeficiency 28, mycobacteriosis
IFT80	Jeune's syndrome
IGBP1	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia
IGF1	Growth retardation with deafness and mental retardation due to IGF1 deficiency
IGHMBP2	Spinal muscular atrophy with respiratory distress

Gene	Pathology
IKBKAP	Familial dysautonomy
IKBKG	Ectodermal dysplasia, hypohidrotic, with immune deficiency
	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency
	Immunodeficiency 33
	INCONNINTENIA pigments, type II
IL12B	Immunodeficiency 29, mycobacteriosis
IL12RB1	Immunodeficiency 30
IL1RAPL1	Mental retardation, 21/34
IL1RN	Interleukin 1 receptor antagonist deficiency
IL2RA	Diabetes, mellitus, insulin-dependent, susceptibility to, 10
	Immunodeficiency 41 with lymphoproliferation and autoimmunity
IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency
	Severe, X-linked T-B+ combined immunodeficiency
IMPDH1	Leber congenital Amaurosis 11
	Retinitis pigmentosa 10
IMPG2	Macular dystrophy, vitelliform, 5
	Retinitis pigmentosa 56
INPP5E	Joubert syndrome 1
	Mental retardation, truncial obesity, retinal dystrophy and micropenis
INSR	Leprechaunism
INVS	Nephronophtisis 2, infantile
IQCB1	Senior syndrome 5
IQSEC2	Mental retardation, 1/78
ISCU	Myopathy with lactic acidosis, hereditary
ITGA6	Junctional epidermolysis bullosa - Pyloric atresia
ITGB4	Junctional epidermolysis bullosa with pyoric atresia
	Junctional epidermolysis bullosa, non-erlitz type
IVD	Isovaleric acidemia
IYD	Thyroid dyshormonogenesis 4
JAK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency
KCNJ1	Prenatal bartter syndrome
KCNJ11	Diabetes mellitus, transient neonatal, 3
	Diabetes mellitus, type 2, susceptibility to
	Diabetes, permanent neonatal, with or without neurological features
	Familial hyperinsulinemic hypoglycemia 2
	Maturity-onset diabetes in young people, type 13

Gene	Pathology
KCNJ13	Leber congenital Amaurosis 16
	Snowflake vitreoretinal degeneration
KCNV2	Retinal cone dystrophy 3b
KDM5C	Mental retardation, syndromic, syndromic, Claes-jensen type
KIAA2022	Mentally retarded, linked to X 98
KIF7	Acrocallosal syndrome
	al-Gazali-Bakalinova syndrome
	Hydroletus syndrome 2
	Joubert syndrome 12
L1CAM	Corpus callosum hypoplasia-retardation-thumb adductus-spasticity-hydroephalus syndrome
	Masa syndrome
LAMA2	Congenital muscular dystrophy type 1a
LAMA3	Bullosa junctional epidermolysis, Herlitz type (Lama3 gene)
	Junctional epidermolysis Bullosa, Herlitz type (Gene Lamb3)
	Junctional epidermolysis bullosa, non-erlitz type (Lama3 gene)
LAMB2	Nephrotic syndrome, type 5, with or without ocular abnormalities
	Pierson's syndrome
LAMB3	Junctional epidermolysis bullosa, non-erlitz type (Lamb3 gene)
LAMC2	Junctional epidermolysis bullosa, Herlitz type (Lamc2 gene)
	Junctional epidermolysis bullosa, non-erlitz type (Lamc2 gene)
LAMP2	Danon's disease
LARGE	Congenital muscular dystrophy type 1D
	Muscle-eye-brain disease
LBR	Greenberg dysplasia
LCA5	Leber congenital Amaurosis 5
LDHA	Glycogen storage disease XI
LDLR	Hypercholesterolemia, familial, 1
	LDL cholesterol level QTL2
LDLRAP1	Hypercholesterolemia, familial, 4
LEPRE1	Osteogenesis imperfecta type 8
LHCGR	Leydig cell adenoma, somatic, with precocious puberty
	Leydig cell hypoplasia with hypergonadotropic hypogonadism
	Leydig cell hypoplasia with pseudohermaphroditism
	Luteinizing hormone resistance, female
	Precocious puberty, male

Gene	Pathology
LHFPL5	Deafness, autosomal recessive 67
LHX3	Combined pituitary hormone deficiency with spinal abnormalities
LIFR	Stüve-Wiedemann syndrome
LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation
LIPA	Cholesteria ester storage disease
	Wolman's disease
LIPH	Hypotrichosis 7
	Woolly hair, autosomal recessive 2 with or without hypotrichosis
LMBRD1	Methylmalonic aciduria and homocystinuria, CBLF type
LMNA	Carcotta-maritime disease of axonal type 2B1
	Lethal restrictive dermopathy
	Mandibuloacral dysplasia with lipodystrophy type A
LOXHD1	Deafness, autosomal recessive 77
LPL	Combined, familial hyperlipidemia
	High-density lipoprotein Lipoprotel QTL level 11
	Lipoprotein lipase deficiency
LRAT	Leber congenital Amaurosis 14
LRP2	Retinal dystrophy, severe with early onset
	Retinal dystrophy, severe with early onset
	Retinitis pigmentosa, juvenile
	Donnai-Barrow syndrome
LRP5	Variability of bone mineral density 1
	Exudative vitreoretinopathy 4
	Hyperostosis, endosteal
	Osteopetrosis, autosomal dominant 1
	Osteoporosis
	Osteoporosis-pseudoglioma syndrome
	Osteosclerosis
	Polycystic liver disease 4 with or without renal cysts
	Van Buchem's disease, type 2
LRPPRC	Leigh syndrome of the French-Canadian type
LRTOMT	Deafness, autosomal recessive 63
LYST	Chediak-Higashi syndrome
MAGT1	Immunodeficiency, X-linked, with magnesium deficiency, Epstein-Barr virus infection, and malignancy
MAK	Retinitis pigmentosa 62

Gene	Pathology
MAN2B1	Mannosidosis, alpha-, types I and II
MARVELD2	Deafness, autosomal recessive 49
MAT1A	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency
	Methionine adenosyltransferase deficiency, autosomal recessive
MATN3	Epiphyseal dysplasia, multiple, 5
	Susceptibility to osteoarthritis 2
	Spondyloepimetaphysal dysplasia
MBTPS2	Ichthyosis follicolaris - Alopecia - photophobia
MCCC1	3-methylcrotonic-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonic-CoA carboxylase 2 deficiency
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis Type 4
MCPH1	Microcephaly 1, primary, autosomal recessive
MECP2	Severe neonatal-onset encephalopathy with microcephaly
MED12	X-Linked Intellectual Deficit with Marfanoid Habitus
MED17	Microcephaly, progressive postnatal, with seizures and cerebral atrophy
MED25	Basel-Vanagait-Smirin-Yosef syndrome
	Charcot-Marie-Tooth Disease, Type 2B2
MEFV	Familial Mediterranean fever
MERTK	Retinitis pigmentosa 38
MESP2	Spondylocostal dysostosis 2, autosomal recessive
MFRP	Microphthalmia, isolated 5
MFSD8	Nanophthalmos 2
	Nanophthalmos 2
	Late neuronal ceroid lipofuscinosis
MGAT2	Congenital glycosylation disorder type 2A
MID1	Opitz GbbB syndrome, type I
MKKS	Bardet-Biedl syndrome 6
MKS1	McKusick-Kaufman syndrome
	McKusick-Kaufman syndrome
	Meckel Syndrome Type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MLYCD	Malonyl Coa decarboxylase deficiency
MMAA	Methylmalonic acidemia sensitive to vitamin B12 CBLA
MMAB	Methylmalonic acidemia sensitive to vitamin B12 CBLB

Gene	Pathology
MMACHC	Methylmalonic acidemia with homocystinuria, CBLC type
	Methylmalonic acidemia with homocystinuria, CBLD type
MMADHC	Homocystinuria, CBLD type, variant 1
MOCS1	Methylmalonic aciduria and homocystinuria, CBLD type
MOCS1	Sulfidase deficiency due to molybdenum cofactor type A deficiency (MOCS1 gene)
MOCS2	Methylmalonic aciduria and homocystinuria, CBLD type
	Methylmalonic aciduria, CBLD type, variant 2
	Sulfidase deficiency due to molybdenum cofactor type A deficiency (MOCS2 gene)
MOGS	Congenital glycosylation disorder, type IIB
MPDU1	Congenital glycosylation disorder, type se
MPI	Congenital glycosylation disorder type 1b
MPL	Thrombocythemia 2
	Thrombocytopenia, congenital ammegakaryocytic
MPV17	Methylmalonic aciduria, CBLD type, variant 2
	Methylmalonic aciduria, CBLD type, variant 2
	Navajo neurohepatopathy
MPZ	Charcot-Marie-Tooth Disease, Type 1B
	Charcot-Marie-Tooth Disease, Type 2i
	Charcot-Marie-Tooth Disease, Type 2J
	Dejerine-Sottas disease
	Neuropathy, congenital hypomyelination
	Roussy-Levy syndrome
MRE11	Disorder similar to that of ataxia-telangiectasia 1
MRPS16	Combined oxidative phosphorylation defect Type 2
MRPS22	Combined oxidative phosphorylation defect Type 5
MTHFR	Homocystinuria due to MTHFR deficiency
MTM1	X-linked centronuclear myopathy
MTMR2	Charcot-Marie-Tooth Disease, Type 4B1
MTR	Homocystinuria-megaloblastic anemia, CBLG complementation type
	Neural tube defects, sensitive to folate, susceptibility to
MTRR	Homocystinuria-megaloblastic anemia, CBL E type
	Neural tube defects, sensitive to folate, susceptibility to
MTTP	Abetalipoproteinemia
MUT	Metabolic syndrome, protection against
	Metabolic syndrome, protection against
	Methylmalonic acidemia unresponsive to vitamin B12 Mut-

Gene	Pathology
MVK	Mevalonic aciduria
MYD88	Macroglobulinemia, Waldenstrom
	Pyogenic, recurrent bacterial infections due to MyD88 deficiency
MYO15A	Deafness, autosomal recessive 3
MYO3A	Deafness, autosomal recessive 30
MYO5A	Griscelli's disease Type 1
MYO6	Deafness, autosomal dominant 22
MYO7A	Autosomal non-syndromic deafness non-syndromic DFNB2
	Usher Syndrome Type 1
NAGA	Kanzaki disease
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	Hyperammonemia due to n-acetylglutamate synthetase deficiency
NBN	Aplastic anemia
	Nijmegen rupture syndrome
NDP	Vitreoretinopathy 2, X-linked
	Norrie's disease
NDRG1	Charcot-Marie-Tooth Disease, Type 4D
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12
	Mitochondrial complex I deficiency
NDUFA7	Mitochondrial complex I deficiency, nuclear type 12
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 12
	Leigh's syndrome
	Mitochondrial complex I deficiency
NDUFAF4	Mitochondrial complex I deficiency
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency
	Mitochondrial complex I deficiency
NDUFS4	Leigh's syndrome
	Mitochondrial complex I deficiency
NDUFS5	NADH-Ubiquinone oxidoreductase FE-S Protein 5
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency
NDUFS7	Leigh's syndrome
NDUFS8	Leigh syndrome due to mitochondrial complex I deficiency
NDUFV1	Mitochondrial complex I deficiency
NEB	Nemaline Myopathy 2
NEFL	Charcot-Marie-Tooth disease, intermediate dominant G

Gene	Pathology
NEU1	Sialidosis, type I
	Sialidosis, type II
NEUROG3	Congenital malabsorbent diarrhea due to paucity of enteroendocrine cells
NHEJ1	Severe immunodeficiency combined with microcephaly, growth retardation and sensitivity to ionizing radiation
NHLRC1	Epilepsy, Progressive Myoclonic 2b (Lafora)
NHP2	Congenital dyskatosis, autosomal recessive 2
NHS	Cataract 40, x-connected
	Nance-Horan syndrome
NKX2-1	Chorea, hereditary benign
	Choreoathetosis, hypothyroidism and neonatal respiratory distress
	Thyroid cancer, non-medium, 1
NKX2-5	Atrial septal defect 7, with or without AV conduction defects
	Conotruncal heart malformations, variable
	Hypoplastic left heart syndrome 2
	Hypothyroidism, congenital non-mongoitro, 5
	Tetralogy of Fallot
	Ventricular septal defect 3
NLGN3	Susceptibility to Asperger's syndrome, linked to X 1
	Susceptibility to autism, linked to X 1
NLGN4X	Mental retardation, X-linked
NLRP7	Mole hydatidiform, recurrent, 1
NMNAT1	Leber congenital Amaurosis 9
NOP10	Congenital dyskatosis, autosomal recessive 1
NPC1	Niemann-Pick Disease Type C1
NPC2	Niemann-Pick Disease Type C2
NPHP1	Joubert syndrome 4
NPHP3	Renal-hepatic dysplasia-pancreasia
	High Level Syndrome 1
NPHP4	Senior syndrome
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR0B1	46xy Sex Reversal 2, Sensitive dosage
NR2E3	S-Cone syndrome improved
	Retinitis pigmentosa 37
NR5A1	46xy Sex Reversion 3

Gene	Pathology
	Adrenocortical insufficiency
NSD1	Beckwith-Wiedemann syndrome
	Sotos syndrome 1
NSDHL	Baby syndrome
	CK syndrome
NSUN2	Mental retardation, autosomal recessive 5
NTRK1	Hereditary sensory and autonomic neuropathy Type 4
NUP62	Infantile bilateral striatal necrosis
NXF5	Nuclear RNA export factor 5
NYX	Night blindness, congenital stationary (complete), 1A, X-linked
OAT	Gyrated atrophy of choroid and retina with or without ornithinemia
OCA2	Albinism, Brown Oculocutaneous
	Albinism, oculocutaneous, type II
	Skin/hair/eye pigmentation 1, blonde/brown hair
	Skin/hair/eye pigmentation 1, blue/non-blue eyes
OCRL	Disease in dent 2
	Oculocerebrorenal syndrome
OFD1	Simpson-Golabi-Behmel Type 2 Syndrome
OPA3	3-methylglutaconic aciduria type 3
OPHN1	Mentally retarded, X-linked, with cerebellar hypoplasia and distinctive facial appearance
ORAI1	Immunodeficiency 9
	Myopathy, tubular aggregate, 2
OSTM1	Osteopetrosis, autosomal recessive 5
OTC	Ornithine transcarbamylase deficiency
OTOA	Deafness, autosomal recessive 22
OTOF	Auditory neuropathy, autosomal recessive, 1
OXCT1	Succinyl COA: COA 3-oxoacid transferase deficiency
PAH	Phenylketonuria
PAK3	Mental retardation, 30/47
PALB2	Fanconi Anemia, Complementation Group N
PANK2	Neurodegeneration associated with pantothenate kinase
PAX3	Craniofacial hand-deafness syndrome
	Rhabdomyosarcoma 2, alveolar
	Waardenburg syndrome, Type 1
	Waardenburg syndrome, Type 3
PAX6	Aniridia

Gene	Pathology
	Anterior segment dysgenesis 5, multiple subtypes
	Cataract with late-onset corneal dystrophy
	Coloboma of the optic nerve
	Coloboma, ocular
	Foveal hypoplasia 1
	Keratitis
	Morning glory disk anomaly
	Optic nerve hypoplasia
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia
PC	Pyruvate carboxylase deficiency
PCBD1	Hyperphenylalaninemia, with BH4 deficiency, D
PCCA	Propionic acidemia (PCCA gene)
PCCB	Propionic Acidemia (PCCB Gene)
PCDH15	Deafness, autosomal recessive 23
	Usher Syndrome, Type 1D/F Digenic
	Usher Syndrome, Type 1F
PCDH19	Epileptic encephalopathy, early infantile, 9
PDE6A	Retinitis pigmentosa 43
PDE6B	Night blindness, congenital stationary, autosomal dominant 2
	Retinitis pigmentosa-40
PDE6C	Cone dystrophy 4
PDE6G	Retinitis pigmentosa 57
PDHA1	Leigh syndrome, X-linked
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Lactic acidemia due to PDX1 deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PDSS1	Flow rates - Encephaloneuropathy - Obesity - Valvular disease
PDSS2	Leigh syndrome with nephrotic syndrome
PDX1	Diabetes mellitus, type II, susceptibility to
	Mody, type IV
	Pancreatic agenesis 1
PDZD7	Deafness, autosomal recessive 57
	Retinal disease in Usher syndrome type IIa, modifier of
	Usher Syndrome, Type IIC, GPR98/PDZD7 Digenic
PEPD	Prolidase deficiency
PEX1	Zellweger syndrome 1A

Gene	Pathology
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)
	Peroxisome biogenesis disorder 6b
PEX12	Neonatal adrenoleukodystrophy (PEX12 gene)
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)
	Peroxisome biogenesis disorder 11b
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)
PEX26	Peroxisome biogenesis disorder 5b
	Peroxisome biogenesis disorder 5b
	Neonatal adrenoleukodystrophy (PEX26 gene)
	Zellweger syndrome 7A
PEX5	Neonatal adrenoleukodystrophy (PEX5 gene)
PEX6	Heimler syndrome 2
PEX7	Peroxisome biogenesis disorder 4A (Zellweger)
	Peroxisome biogenesis disorder 4b
	Rhizomelic chondrodysplasia punctata type 1
PFKM	Glycogen storage disease VII
1 PGK	Phosphoglycerate kinase deficiency
PGM1	Congenital disorder of glycosylation, digitalis
PHF8	Mental retardation syndrome, X-linked type, Siderius type
PHGDH	Neu-Laxova syndrome 1
	Phosphoglycerate dehydrogenase deficiency
PHKG2	Cirrhosis due to hepatic phosphorylase kinase deficiency
	Glycogen storage disease IXC
PHYH	Reference disease
PKHD1	Autosomal recessive polycystic kidney disease
PKLR	Hemolytic anemia due to cellular pyruvate kinase deficiency
PLA2G6	Infantile neuroaxonal dystrophy 2A
	Infantile neuroaxonal dystrophy 2B
PLCE1	Nephrotic syndrome, Tupe 3
PLDN	Hermansky-pudlak syndrome 9
PLEC	Epidermolysis bullosa simplex with muscular dystrophy
	EpiderMolysis Bullosa simplex with pyloric atresia
	Belt dystrophy of the limb with epidermolysis bullosa simplex
PLEKHG5	Autosomal distal spinal muscular atrophy type 4
PLG	Type 1 plasminogen deficiency
PLOD1	Ehlers-Danlos Syndrome Type 6

Gene	Pathology
PLP1	Spastic paraplegia type 2, X-linked
PMM2	Congenital glycosylation disorder type 1A
PMP22	Charcot-Marie-Tooth Disease, Type 1A
	Charcot-Marie-Tooth Disease, Type 1E
	Dejerine-Sottas disease
	Roussy-Levy syndrome
PNPO	Pyridoxal seizures of phosphate sensitivity
POLG	Alpers syndrome
	Autosomal recessive progressive external ophthalmoplegia
	Mitochondrial neurogastrointestinal encephalomyopathy
	Sensory ataxic neuropathy - Dysarthria - Ophthalmoparesis
POLR1C	Leucodystrophy, hypomyelinating, 11
POMGNT1	Treacher Collins Syndrome 3
	Treacher Collins Syndrome 3
	Autosomal recessive type C muscle dystrophy
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg Syndrome (Gene Pomgnt1)
POMT1	Autosomal recessive type C muscle dystrophy
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg syndrome (POMT1 gene)
POMT2	Autosomal recessive type C muscle dystrophy
	Congenital muscular dystrophy with cerebellar involvement
	Walker-Warburg syndrome (POMT2 gene)
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
POU1F1	Combined pituitary hormone deficiencies, genetic forms
POU3F4	Deafness, X link 2
PPT1	Adult neuronal ceroid lipofuscinosis
PQBP1	Renpenning syndrome
PRCD	Retinitis pigmentosa 36
PRF1	Hemophagocytic lymphocytosis, familial, 2
PRKRA	Dystonia 16
PRODH	Hyperprolinemia, type I
	Schizophrenia, susceptibility to, 4
PROM1	Cone-rod dystrophy 12
PROP1	Macular dystrophy, retina, 2
	Combined pituitary hormone deficiencies, genetic forms

Gene	Pathology
PRPS1	Retinitis pigmentosa 41
	Lethal ataxia with deafness and optic atrophy
	Carcotta-Marco-Mars disease
PRSS12	Macular dystrophy, retina, 2
	Retinitis pigmentosa 41
	Stargardt's disease 4
	Stargardt's disease 4
	Mental retardation, autosomal recessive 1
PRX	Charcot-Marie-tooth disease type 4f
PSAP	Encephalopathy due to prosaposin deficiency
	Krabbe disease
	Metachromatic leukodystrophy
PSAT1	Phosphoserine aminotransferase deficiency
PTEN	Neu-Laxova syndrome 2
	Neu-Laxova syndrome 2
	Bannayan-Riley-Ruvalcaba syndrome
	Cowden syndrome 1
	Lhermitte-Duclos syndrome
	Macrocephaly/autism syndrome
PTH1R	Chondrodysplasia, blomstrand type
	Eiken syndrome
	Failure of tooth eruption, primary
	Metaphyseal chondrodysplasia, Murk Jansen type
PTS	Hyperphenylalaninemia, with BH4 deficiency, a
PUS1	Myopathy, lactic acidosis and sideroblastic anemia 1
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
QDPR	Hyperphenylalaninemia, with BH4 deficiency, C
RAB23	Carpenter's syndrome
RAB27A	Griscelli's disease type 2
RAB39B	Mentally retarded, linked to X 72
RAB3GAP1	Micro Syndrome
RAB3GAP2	Cataract - Intellectual deficit - Hypogonadism
RAD51C	Fanconi Anemia, Complementation Group O
RAG1	Breast-ovarian cancer, familial, susceptibility to, 3
	Breast-ovarian cancer, familial, susceptibility to, 3
	Combined immunodeficiency with skin granulomas

Gene	Pathology
	Omenn Syndrome (Rag1 Gene)
	Severe combined immunodeficiency due to complete Rag1/2 deficiency
RAG2	Combined immunodeficiency with skin granulomas
	Omenn Syndrome (Rag2 Gene)
	Severe combined immunodeficiency due to complete Rag1/2 deficiency
RAPSN	Deformation sequence of Fetal Akinesia
RARS2	Pontocerebellar Hypoplasia, Type 6
RAX	Microphthalmia, isolated 3
RDH12	Leber congenital Amaurosis 13
RDX	Deafness, autosomal recessive 24
RELN	Lissencephaly syndrome, Norman-Roberts type
REN	Hyperproreninemia
	Hyperuricemic nephropathy, juvenile familial 2
	Renal tubular dysgenesis
RFT1	Congenital disorder of glycosylation, type
RGR	Retinitis pigmentosa
RHO	Night blindness, congenital stationary, autosomal dominant 1
	Retinitis pigmentosa 4, autosomal dominant or recessive
	Retinitis punctata albescens
RLBP1	Retinal dystrophy of both
	Fundus albipunctatus
	Newfoundland Cone Dystrophy
	Retinitis punctata albescens
RMRP	Anuxtic dysplasia
	Cartilage hair hypoplasia
	Metaphyseal dysplasia without hypotrichosis
RNASEH2A	Aicardi-Goutieres syndrome 4
RNASEH2B	Aicardi-Goutieres syndrome 2
RNASEH2C	Aicardi-Goutieres syndrome 3
RP2	Retinitis pigmentosa 2
RPE65	Leber congenital Amaurosis 2
	Retinitis pigmentosa 20
RPGR	Cone-rod dystrophy, x-linked, 1
	X-linked macular degeneration, atrophic
	Retinitis pigmentosa 3
	Retinitis pigmentosa, X-linked and sinorespiratory infections, with or without deafness

Gene	Pathology
RPGRIP1L	Joubert syndrome with liver defect
	Meckel syndrome, Type 5
RPL10	Autism, susceptibility to, linked to x 5
RPS6KA3	Coffin-lowry syndrome
	Mentally retarded, X-linked 19
RRM2B	Mitochondrial DNA depletion syndrome 8A (encephalomyopathy type with renal tubulopathy)
	Mitochondrial DNA depletion syndrome 8B (MNGIE type)
RS1	Retinoschisis
RYR1	Central core disease
	King-Denborough syndrome
	Malignant susceptibility to hyperthermia 1
	Minicore myopathy with external ophthalmoplegia
	Neuromuscular disease, congenital, with uniform fiber type 1
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
SAG	Oguchi disease-1
	Retinitis pigmentosa 47
SAMD9	Mirage syndrome
	Tumor, familial, normophosphatemic calcinosis
SAMHD1	Aicardi-Goutieres syndrome 5
	Chilblain Lupus 2
SBDS	Shwachman-Diamond syndrome
SBF2	Charcot-Marie-Tooth Disease, Type 4B2
SC5DL	Lathosterolosis
SCN2A	Epileptic encephalopathy, early childhood, 11
	Convulsions, Benign Familial Infantile, 3
SCNN1A	Pseudohypoaldosteronism type 1, autosomal recessive (SCNN1A gene)
SCNN1B	Pseudohypoaldosteronism type 1, autosomal recessive (SCNN1B gene)
SCNN1G	Pseudohypoaldosteronism type 1, autosomal recessive (SCNN1G gene)
SCO1	Mitochondrial complex IV deficiency
SCO2	Cardioencephalomyopathy, fatal in childhood, due to cytochrome 1 oxidase C deficiency
SEMA4A	Cone-rod dystrophy 10
	Retinitis pigmentosa 35
SEPN1	Stiff spine syndrome
SEPSECS	Pontocerebellar Hypoplasia Type 2D
SERPINA1	Emphysema due to AAT deficiency

Gene	Pathology
	Emphysema-cirrhosis, due to AAT deficiency
	Hemorrhagic diathesis due to antithrombin Pittsburgh
	Pulmonary disease, chronic obstructive, susceptibility to
SETX	Amyotrophic lateral sclerosis 4, juvenile
	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2
SFTPB	Dysfunction of surfactant metabolism, pulmonary, 1
SFTPC	Dysfunction of surfactant metabolism, pulmonary, 2
SGCA	Muscular dystrophy, club limbs, autosomal recessive 3
SGCB	Muscular dystrophy, club limbs, autosomal recessive 4
SGCD	Cardiomyopathy, dilated, 1L
	Muscular dystrophy, club limbs, autosomal recessive 6
SGCG	Muscular dystrophy, club limbs, autosomal recessive 5
SGSH	Mucopolysaccharidosis Type 3A (Sanfilippo syndrome type A)
SH2D1A	X-linked lymphoproliferative disease
SH3TC2	Charcot-Marie-Tooth Disease, Type 4C
	Median nerve mononeuropathy, mild
SHROOM4	Stocco dos santos mental retardation syndrome
SIL1	Marinesco-Sjögren syndrome
SIX6	Optic disc anomalies with retinal and/or macular dystrophy
SLC12A1	Prenatal Bartter Syndrome Type 1
SLC12A3	Gitelman syndrome
SLC12A6	Corpus callosum Agenesis - Neuronopathy
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Free sialic acid retention disease, infantile form
SLC19A2	Sensitive megaloblastic anemia syndrome
SLC22A5	Carnitine deficiency, primary systemic
SLC24A1	Night blindness, congenital stationary (complete), 1D, autosomal recessive
SLC25A13	Citrullinemia, adult-onset type II
	Citrullinemia, type II, neonatal onset
SLC25A15	Hyperornithinemia-hyperammemia-homocitrullinuria
SLC25A20	Carnitine-acylcarnitine deficiency
SLC25A22	Early infantile epileptic encephalopathy
SLC26A2	Achondrogenesis Type 1B
SLC26A2	Type II atelosteogenesis
	Diastrophic Dwanism
	Multiple epiphyseal dysplasia Type 4

Gene	Pathology
SLC26A3	Diarrhea 1, secretory chloride, congenital
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
	Pendred syndrome
SLC26A5	Deafness, autosomal recessive 61
SLC35A1	Congenital glycosylation disorder type 2F
SLC35C1	Congenital glycosylation disorder type 2c
SLC35D1	Schneckenbecken dysplasia
SLC37A4	Glycogen storage disease due to glucose-6-phosphatase type B deficiency
	Glycogen storage disease due to glucose-6-phosphatase type C deficiency
SLC39A4	Acrodermatitis enteropathica
SLC3A1	Cystinuria
SLC45A2	Albinism, oculocutaneous, type IV
	Skin/hair/eye pigmentation 5, black/non-black hair
	Skin/hair/eye pigmentation 5, dark/light skin
	Skin/hair/eye pigmentation 5, dark/light eyes
SLC46A1	Folate malabsorption, hereditary
SLC4A11	Congenital endothelial endothelial dystrophy Type II
	Corneal dystrophy - perceptual deafness
SLC5A5	Folate malabsorption, hereditary
SLC6A19	Hartnup's disorder
	Hyperglycinuria
	Iminoglycinuria, digenic
SLC6A8	X-linked creatine transporter deficiency
SLC7A7	Intolerance to lysinuric protein
SLC7A9	Cystinuria
SLC9A6	Mental retardation, X-linked syndromic, Christianson Type
SLX4	Fanconi Anemia, Complementation Group P
SMARCA	Fanconi Anemia, Complementation Group P
SMN1	Proximal spinal muscular atrophy Type 1
	Proximal spinal muscular atrophy Type 2
	Proximal spinal muscular atrophy Type 3
	Proximal spinal muscular atrophy Type 4
SMN2	Spinal muscular atrophy, type III, modifier of
SMPD1	Niemann-Pick Disease Type A
	Niemann-Pick Disease Type B
SMS	Mental trimming, X-linked type, Snyder-Robinson type

Gene	Pathology
SNAI2	Piebaldism
	Waardenburg syndrome, type 2D
SNAP29	Cerebral dysgenesis-Neuropathy-ichthyosis-palmoplantar Kiratoderma Syndrome
SOX3	Mental retardation, X-linked, with isolated growth hormone deficiency
	Panhypopituitarism, X-linked
SP110	Hepatic veneocclusive disease with immunodeficiency
SPG11	Amyotrophic lateral sclerosis 5, juvenile
	Charcot-Marie-Tooth disease, axonal, type 2x
	Spastic paraplegia 11, autosomal recessive
SPG20	Troyer syndrome
SPG7	Spastic paraplegia 7, autosomal recessive
SRD5A2	Perineoscrotal pseudovaginal hypospadias
SRD5A3	Congenital disorder of glycosylation, IQ type
	Kahrizi syndrome
SRPX2	Rolandic epilepsy, mental retardation and speech dyspraxia
ST3GAL3	Epileptic encephalopathy, early infantile, 15
	Mental retardation, autosomal recessive 12
ST3GAL5	Amish childhood epilepsy syndrome
STAR	Congenital lipoid adrenal hyperplasia
STAT1	Immunodeficiency 31a, mycobacteriosis, autosomal dominant
	Immunodeficiency 31b, mycobacterial and viral infections, autosomal recessive
	Immunodeficiency 31c, autosomal dominant
STIL	Microcephaly 7, primary, autosomal recessive
STIM1	Immunodeficiency 10
	Myopathy, tubular aggregate, 1
	Storken syndrome
STRA6	Syndromic microphthalmia Type 9
STRC	Deafness, autosomal recessive 16
STX11	Hemophagocytic lymphothiocytosis, familial, 4
STXBP2	Hemophagocytic lymphothiocytosis, familial, 5
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathy with or without methylmalonic aciduria)
SUCLG1	Fatal infantile lactic acidosis with methylmalonic aciduria
SUMF1	Multiple sulfatase deficiency
SUOX	Sulphocysteinuria
SURF1	Leigh syndrome, due to Cox deficiency

Gene	Pathology
SYN1	Epilepsy, X-linked, with variable learning difficulties and behavioral disorders
SYP	Mentally retarded, X-linked 96
TAF1	Dystonia-parkinsonism, x-linked
	Mental retardation, syndromic 33, syndromic X
TAT	Tyrosinemia Type 2
TAZ	Barth syndrome
TBCE	Hypoparathyroidism - Intellectual disc - Dysmorphic syndrome
TCAP	Cardiomyopathy, hypertrophic, 25
	Muscular dystrophy, club limbs, autosomal recessive 7
TCF4	Pitt-Hopkins syndrome
TCIRG1	Autosomal recessive malignant osteopetrosis 1
TCN2	Transcobalamin II deficiency
TECTA	Deafness, autosomal dominant 8/12
	Deafness, autosomal recessive 21
TERT	Congenital dyskatosis, autosomal dominant 2
	Congenital dyskatosis, autosomal recessive 4
	Leukemia, acute myeloid
	Melanoma, cutaneous malignancy, 9
	Pulmonary fibrosis and/or bone marrow failure, related to telomeres, 1
TFR2	Hemochromatosis, type 3
TG	Autoimmune thyroid disease, susceptibility to, 3
	Thyroid dyshormonogenesis 3
TGM1	Ichthyosis, congenital, autosomal recessive 1
TH	Autosomal recessive-sensitive dystonia
THRA	Hypothyroidism, congenital, non-mongoitro, 6
THRB	Resistance to thyroid hormone
	Resistance to thyroid hormone, autosomal recessive
	Resistance to thyroid hormone, selective pituitary
TIMM8A	Mohr-Tranebjaerg syndrome
TK2	Mitochondrial DNA depletion syndrome, myopathic form
TLR3	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 2
	HIV1 infection, resistance to
TMC1	Deafness, autosomal dominant 36
	Deafness, autosomal recessive 7
TMEM216	Joubert syndrome 2
	Meckel syndrome 2

Gene	Pathology
TMEM67	Coach Syndrome
	Joubert syndrome 6
TMIE	Deafness, autosomal recessive 6
TMPRSS3	Deafness, autosomal recessive 8/10
TNFRSF1 1B	Paget disease, juvenile
TNNT1	Nemaline Myopaty 5, Amish type
TPO	Thyroid dyshormonogenesis 2A
TPP1	Neuronal ceroid lipofuscinosis 2
TPRN	Deafness, autosomal recessive 79
TRAPPC9	Mental retardation, autosomal recessive 13
TRDN	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness
TREX1	Aicardi-Goutières syndrome
TRIM32	Bardet-Biedl syndrome 11
	Muscular dystrophy, club limbs, autosomal recessive 8
TRIM37	Mulibrey dwarfism
TRIOBP	Deafness, autosomal recessive 28
TRMU	Deafness, mitochondrial, modifier of
	Liver failure, transient infantile
TSEN54	Pontocerebellar Hypoplasia Type 2A
	Pontocerebellar Hypoplasia Type 4
TSFM	Fatal mitochondrial disease due to combined deficiency of oxidative phosphorylation 3
TSHB	Isolated thyroid-stimulating hormone deficiency
TSHR	Hyperthyroidism, familial gestational
	Hyperthyroidism, non-autoimmune
	Hypothyroidism, congenital, non-mongoitro, 1
	Thyroid adenoma, hyperfunction, somatic
	Thyroid carcinoma with thyrotoxicosis
TSPAN7	Mentally retarded, linked to X 58
TSPYL1	Sudden infant death with Testes syndrome dysgenesis
TTC37	Trichoheparaenteric syndrome 1
TTN	Cardiomyopathy, dilated, 1g
	Cardiomyopathy, familial hypertrophic, 9
	Muscular dystrophy, club limbs, autosomal recessive 10
	Myopathy, myofibrillar, 9, with early respiratory failure
	Salih myopathy

Gene	Pathology
	Tibial muscular dystrophy, late
TTPA	Ataxia with vitamin E deficiency
TUBA1A	Lissencephaly 3
TUFM	Combined oxidative phosphorylation deficiency 4
TULP1	Leber congenital Amaurosis 15
	Retinitis pigmentosa 14
TUSC3	Ntal delay, autosomal recessive 7
TYK2	Immunodeficiency 35
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
TYR	Albinism, oculocutaneous, type IA
	Albinism, oculocutaneous, type Ib
	Melanoma, cutaneous malignancy, susceptibility to, 8
	Skin/hair/eye pigmentation 3, blue/green eyes
	Skin/hair/eye pigmentation 3, light/dark/freckled skin
	Waardenburg syndrome/albinism, digenic
TYRP1	Albinism, oculocutaneous, type III
	Skin/hair/eye pigmentation, variation in, 11 (Melanesian blonde hair)
UBA1	X-linked spinal muscular atrophy Type 2
UBE2A	Mental retardation, X-linked syndromic, birth type
UBE3A	Angelman syndrome
UBR1	Johanson-B-B-Bizzard syndrome
UGT1A1	Bilirubin, serum level of, qtl1
	Crigler-Najjar syndrome, type I
	Crigler-Najjar syndrome, type II
	Gilbert's syndrome
	Hyperbilirubinemia, transient familial neonatal
UNC13D	Hemophagocytic lymphothiocytosis, familial, 3
UNC93B1	Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 1
UPF3B	Mentally retarded, X-linked, syndromic 14
UQCRB	Mitochondrial respiratory chain deficiency III
UQCRQ	Mitochondrial respiratory chain deficiency III
UROS	Porphyria, congenital erythropoietic
USH1C	Autosomal DFNB18 Non-syndromic recessive of DFNB18 sensory deafness
	Usher Syndrome Type 1C
USH1G	Usher Syndrome Type 1G
USH2A	Usher Syndrome Type 2A

Gene	Pathology
USP9X	Mentally retarded, linked to X 99
	Mental retardation, X-linked 99, syndromic, female limited
VDR	VITAMIN DEPENDENT RICKETS TYPE 2A
VIPAR	Arthrogryposis, renal dysfunction and cholestasis 2
VLDLR	Cerebellar ataxia - Intellectual disc - Dysevelibum syndrome
VPS13A	Choreoacanthosis
VPS13B	Cohen Syndrome Type 1
VPS33B	Arthrogryposis - renal dysfunction - cholestasis
VRK1	Pontocerebellar Hypoplasia Type 1A
VSX2	Microphthalmia with Coloboma 3
	Microphthalmia, isolated 2
VWF	Von Willebrand's disease, type 1
	Von Willebrand disease, types 2a, 2b, 2m and 2n
	Von Wilbrand's disease, type 3
WAS	Wiskott-Aldrich syndrome
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations
WFS1	Wolfram syndrome 1
WHRN	Deafness, autosomal recessive 31
	Usher Syndrome, Type 2D
WISP3	Arthropathy, progressive pseudorheumatoid, of childhood
	Late spondyloepiphyseal dysplasia with progressive arthropathy
WNT10A	X-linked severe congenital neutropenia
WNT3	Tetra-Amelia, autosomal recessive
WNT7A	Aplasia/hypoplasia of limbs and pelvis
	Fibular hypoplasia or aplasia - Femoral bowing - oligodactyl
WRN	Werner syndrome
XIAP	Lymphoproliferative syndrome, X-linked, 2
XPA	Xeroderma Pigmentosum Complementation Group A
XPC	Xeroderma Pigmentosum, group C
ZDHHC9	Mental retardation, X-linked syndromic, Raymond type
ZEB2	Mowat-Wilson syndrome
ZFYVE26	Spastic paraplegia 15, autosomal recessive
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked
	Heterotaxy, visceral, 1, X-linked
ZMPSTE24	Lethal restrictive dermopathy
	Mandibuloacral dysplasia with lipodystrophy type B

Gene	Pathology
ZNF469	Brittle cornea syndrome
ZNF711	Mentally retarded, X-linked 97