Gene	Pathology
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
AUH	3-methylglutaconic aciduria Type 1
B4GALT1	Congenital disorder of the 2D glycosylation type
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
BLM	Bloom's syndrome
BRWD3	Mentally retarded, X-linked 93
BTD	Biotinidase deficiency
BTK	Isolated growth hormone deficiency Type III
	X-linked agammaglobulinemia
C10ORF2	Infantile onset spinocerebellar ataxia
CA2	Osteopetrosis with renal tubular acidosis
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
CBS	Classical homocystinuria
CD19	Immunodeficiency, common variable, 3
CD3D	Immunodeficiency 19
CD3E	Immunodeficiency 18, SCID variant
CD3G	Immunodeficiency 17, gamma CD3 deficient
CD40LG	X-linked hyper-IgM syndrome
CDH23	DFNB12 autosomal non-syndromic deafness sensorimic type Dfnb12

Gene	Pathology
CDKL5	Epileptic encephalopathy, early infantile, 2
CEP290	Joubert syndrome with oculorenal defect 5
	Senior syndrome
CFP	Corrected dyne deficiency, linked to x
CFTR	Cystic fibrosis; mucoviscidosis
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
CHRNG	Escobar syndrome
	Multiple pterygium syndrome, lethal type
	Myasthenia gravis, neonatal transient
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
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
COG1	Congenital glycosylation disorder, type IIG
COG7	Congenital glycosylation disorder, type IIE
COG8	Congenital glycosylation disorder, type IIH
COL17A1	Generalized junctional epidermolysis bullosa, non-erlitz type
COL1A1	Caffey's disease
	Ehlers-Danlos syndrome, type I

Gene	Pathology
	Ehlers-Danlos syndrome, type VIIA
	Osteogenesis imperfecta, type I
	Osteogenesis imperfecta, type II
	Osteogenesis imperfecta, type III
	Osteogenesis imperfecta, type IV
COL1A2	Ehlers-Danlos syndrome, heart valve type
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
COQ2	Leigh syndrome with nephrotic syndrome
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
CRLF1	Cold-induced sweating syndrome
CRTAP	Osteogenesis imperfecta type VII
CSTB	Unverricht-Lundborg disease
CTNS	Cystinosis
CTSD	Adult neuronal ceroid lipofuscinosis 10
CTSK	Pycnodysostosis

Gene	Pathology
CUL4B	Mental retardation, syndromic, syndromic X (Cabeza type)
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
CYP17A1	17-alpha-hydroxylase/17,20 deficiency
CYP27A1	Cerebrotenndous xanthomatosis
CYP27B1	VITAMIN D-dependent RICKET, Type I
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
DGUOK	Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK 3 deficiency
DHCR24	Desmosterosis
DHCR7	Smith-Lemli-Opitz syndrome
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
DNAJC19	Dilated cardiomyopathy with ataxia
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

Gene	Pathology
DPYD	Dihydropyrimidine dehydrogenase deficiency
DSP	Lethal acantholytic epidermolysis bullosa
DYNC2H1	Short kibble thoracic dysplasia 3 with or without polydacty
EDA	Muscular dystrophy, club limbs, autosomal recessive 2
	Ectodermal dysplasia 1, hypohidrotic, X-linked
	Tooth agenesis, selective, X-linked 1
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
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
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
F8	Hemophilia a
F9	Hemophilia b

Gene	Pathology
FAH	Tyrosinemia Type 1
FAM126A	Hypomyelination - congenital cataract
FAM20C	Lethal osteosclerotic bone dysplasia
FANCC	Fanconi anemia complementation group C.
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)
FGD1	Aarskog-Scott syndrome
	Mental retardation, X-linked syndromic 16
FGD4	Charcot-Marie-tooth type 4h disease
FH	Fumaric aciduria
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
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)
FTSJ1	Mentally retarded, X-linked 9
FUCA1	Fucosidosis
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

Gene	Pathology
GALC	Krabbe disease
GALK1	Galactokinase deficiency with cataracts
GALT	Classic galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
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
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
GDI1	Mental retardation, X-linked 41
GFM1	Hepaenencephalopathy due to combined deficiency of type 1 oxidative phosphorylation
GJB2	Autosomal Recessive Non-Syndromic Sensory Deafness DFNB1A (GJB2 Gene)
GJC2	Pelizaeus-Merzbacher due to the GJC2 mutation
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
GNPTAB	Mucolipidosis type 2
	Mucolipidosis type 3
GNRHR	Fertile eunuch syndrome
	Hypogonadotropic hypogonadism 7 without anosmia
GPC3	Simpson-Golabi-Behmel Syndrome, Type 1
GPR98	Usher Syndrome Type 2C
GRIK2	Mental retardation, autosomal recessive, 6
GSS	Glutathione synthetase deficiency with 5-oxoprolinia

Gene	Pathology
GTF2H5	Trichothiodiophycy, complementation group A
GUSB	Mucopolysaccharidosis type 7
HADH	Long-chain 3-hydroxyl-CoA dehydrogenase deficiency
HADHA	Protein deficiency
HADHB	Protein deficiency
HAMP	Histidinemia
HAX1	Neutropenia, severe congenital 3, autosomal recessive
HBB	Beta-thalassemia
	Sickle cell anemia
HESX1	Combined pituitary hormone deficiencies, genetic forms
HEXA	Tay-Sachs disease
HEXB	Sandhoff's disease
HFE2	Hemochromatosis, type 2A
HGSNAT	Sanfilippo Syndrome Type C
HIBCH	Neurodegeneration due to hydroxyisobutyryl-CoA hydrolase deficiency
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaric aciduria
HPD	Tyrosinemia Type 3
HPRT1	Kelley-Seegmiller syndrome
	Lesch-Nyhan syndrome
HSD11B2	Apparent mineralocorticoid excess
HSD17B3	Pseudohermaphroditism, male, with gynecomastia
HSD17B4	Deficiency of bifunctional enzymes
	Perrault syndrome
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
HSPG2	Schwartz-Jampel syndrome
HUWE1	Mental retardation, X-linked syndrome, Turner type
ICOS	Immunodeficiency, common variable, 1
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

Gene	Pathology
IGHMBP2	Spinal muscular atrophy with respiratory distress
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
IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency
	Severe, X-linked T-B+ combined immunodeficiency
INSR	Leprechaunism
INVS	Nephronophtisis 2, infantile
IQCB1	Senior syndrome 5
ITGA6	Junctional epidermolysis bullosa - Pyloric atresia
ITGB4	Junctional epidermolysis bullosa with pyoric atresia
	Junctional epidermolysis bullosa, non-erlitz type
IVD	Isovaleric acidemia
JAK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency
KCNJ1	Prenatal bartter syndrome
KDM5C	Mental retardation, syndromic, syndromic, Claes-jensen type
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)
LARGE	Congenital muscular dystrophy type 1D

Gene	Pathology
	Muscle-eye-brain disease
LBR	Greenberg dysplasia
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
LHX3	Combined pituitary hormone deficiency with spinal abnormalities
LIFR	Stüve-Wiedemann syndrome
LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation
LMNA	Carcotta-maritime disease of axonal type 2B1
	Lethal restrictive dermopathy
	Mandibuloacral dysplasia with lipodystrophy type A
LRP2	Retinal dystrophy, severe with early onset
	Retinal dystrophy, severe with early onset
	Retinitis pigmentosa, juvenile
	Donnai-Barrow syndrome
LRPPRC	Leigh syndrome of the French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Mannosidosis, alpha-, types I and II
MBTPS2	Ichthyosis follicolaris - Alopecia - photophobia
MCOLN1	Mucolipidosis Type 4
MECP2	Severe neonatal-onset encephalopathy with microcephaly
MED12	X-Linked Intellectual Deficit with Marfanoid Habitus
MEFV	Familial Mediterranean fever
MFSD8	Nanophthalmos 2
	Nanophthalmos 2
	Late neuronal ceroid lipofuscinosis
MGAT2	Congenital glycosylation disorder type 2A
MID1	Opitz GbbB syndrome, type I
MKS1	McKusick-Kaufman syndrome
	McKusick-Kaufman syndrome
	Meckel Syndrome Type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts

Gene	Pathology
MMAA	Methylmalonic acidemia sensitive to vitamin B12 CBLA
MMAB	Methylmalonic acidemia sensitive to vitamin B12 CBLB
MMACHC	Methylmalonic acidemia with homocystinuria, CBLC type
	Methylmalonic acidemia with homocystinuria, CBLD type
MOCS1	Methylmalonic aciduria and homocystinuria, CBLD type
	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
MRPS16	Combined oxidative phosphorylation defect Type 2
MRPS22	Combined oxidative phosphorylation defect Type 5
MTM1	X-linked centronuclear myopathy
MUT	Metabolic syndrome, protection against
	Metabolic syndrome, protection against
	Methylmalonic acidemia unresponsive to vitamin B12 Mut-
MVK	Mevalonic aciduria
MYD88	Macroglobulinemia, Waldenstrom
	Pyogenic, recurrent bacterial infections due to MyD88 deficiency
MYO5A	Griscelli's disease Type 1
MYO7A	Autosomal non-syndromic deafness non-syndromic DFNB2

Gene	Pathology
	Usher Syndrome Type 1
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
NDUFA1	Mitochondrial complex I deficiency, nuclear type 12
	Mitochondrial complex I deficiency
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 12
	Leigh's syndrome
	Mitochondrial complex I deficiency
NDUFAF4	Mitochondrial complex I deficiency
NDUFS3	Leigh syndrome due to mitochondrial complex I deficiency
	Mitochondrial complex I deficiency
NDUFS4	Leigh's syndrome
	Mitochondrial complex I deficiency
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
NEU1	Sialidosis, type I
	Sialidosis, type II
NHEJ1	Severe immunodeficiency combined with microcephaly, growth retardation and sensitivity to ionizing radiation
NHLRC1	Epilepsy, Progressive Myoclonic 2b (Lafora)
NHS	Cataract 40, x-connected
	Nance-Horan syndrome
NLGN4X	Mental retardation, X-linked
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

Gene	Pathology
NPHP4	Senior syndrome
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR5A1	46xy Sex Reversion 3
	Adrenocortical insufficiency
NSD1	Beckwith-Wiedemann syndrome
	Sotos syndrome 1
NSUN2	Mental retardation, autosomal recessive 5
NTRK1	Hereditary sensory and autonomic neuropathy Type 4
NUP62	Infantile bilateral striatal necrosis
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
OXCT1	Succinyl COA: COA 3-oxoacid transferase deficiency
PAH	Phenylketonuria
PAK3	Mental retardation, 30/47
PANK2	Neurodegeneration associated with pantothenate kinase
PC	Pyruvate carboxylase deficiency
PCCA	Propionic acidemia (PCCA gene)
PCCB	Propionic Acidemia (PCCB Gene)
PCDH19	Epileptic encephalopathy, early infantile, 9
PDHA1	Leigh syndrome, X-linked
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
PEX1	Zellweger syndrome 1A
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)

Gene	Pathology
	Peroxisome biogenesis disorder 6b
PEX12	Neonatal adrenoleukodystrophy (PEX12 gene)
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)
	Peroxisome biogenesis disorder 11b
PEX26	Peroxisome biogenesis disorder 5b
	Peroxisome biogenesis disorder 5b
	Neonatal adrenoleukodystrophy (PEX26 gene)
	Zellweger syndrome 7A
PEX5	Neonatal adrenoleukodystrophy (PEX5 gene)
PEX7	Peroxisome biogenesis disorder 4A (Zellweger)
	Peroxisome biogenesis disorder 4b
	Rhizomelic chondrodysplasia punctata type 1
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
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

Gene	Pathology
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
PPT1	Adult neuronal ceroid lipofuscinosis
PQBP1	Renpenning syndrome
PRF1	Hemophagocytic lymphocytosis, familial, 2
PROP1	Macular dystrophy, retina, 2
	Combined pituitary hormone deficiencies, genetic forms
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
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

Gene	Pathology
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
RAB23	Carpenter's syndrome
RAB27A	Griscelli's disease type 2
RAB39B	Mentally retarded, linked to X 72
RAG1	Breast-ovarian cancer, familial, susceptibility to, 3
	Breast-ovarian cancer, familial, susceptibility to, 3
	Combined immunodeficiency with skin granulomas
	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
RELN	Lissencephaly syndrome, Norman-Roberts type
RFT1	Congenital disorder of glycosylation, type
RMRP	Anuxtic dysplasia
	Cartilage hair hypoplasia
	Metaphyseal dysplasia without hypotrichosis
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)
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
SAMHD1	Aicardi-Goutieres syndrome 5
	Chilblain Lupus 2
SBDS	Shwachman-Diamond syndrome
SC5DL	Lathosterolosis
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

Gene	Pathology
SCO2	Cardioencephalomyopathy, fatal in childhood, due to cytochrome 1 oxidase C deficiency
SEPN1	Stiff spine syndrome
SFTPB	Dysfunction of surfactant metabolism, pulmonary, 1
SFTPC	Dysfunction of surfactant metabolism, pulmonary, 2
SGSH	Mucopolysaccharidosis Type 3A (Sanfilippo syndrome type A)
SH2D1A	X-linked lymphoproliferative disease
SIL1	Marinesco-Sjögren syndrome
SLC12A1	Prenatal Bartter Syndrome Type 1
SLC12A6	Corpus callosum Agenesis - Neuronopathy
SLC16A2	Allan-Herndon-Dudley syndrome
SLC17A5	Free sialic acid retention disease, infantile form
SLC22A5	Carnitine deficiency, primary systemic
SLC25A15	Hyperornithinemia-hyperammemia-homocitrullinuria
SLC25A20	Carnitine-acylcarnitine deficiency
SLC25A22	Early childhood epileptic encephalopathy
SLC26A2	Achondrogenesis Type 1B
	Type II atelosteogenesis
	Diastrophic Dwanism
	Multiple epiphyseal dysplasia Type 4
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
SLC4A11	Congenital endothelial endothelial dystrophy Type II
	Corneal dystrophy - perceptual deafness
SLC6A8	X-linked creatine transporter deficiency
SLC9A6	Mental retardation, X-linked syndromic, Christianson Type
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
SMPD1	Niemann-Pick Disease Type A
	Niemann-Pick Disease Type B

Gene	Pathology
SMS	Mental trimming, X-linked type, Snyder-Robinson type
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
SRD5A2	Perineoscrotal pseudovaginal hypospadias
SRD5A3	Congenital disorder of glycosylation, IQ type
	Kahrizi syndrome
ST3GAL3	Epileptic encephalopathy, early infantile, 15
	Mental retardation, autosomal recessive 12
	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
STIM1	Immunodeficiency 10
	Myopathy, tubular aggregate, 1
	Storken syndrome
STRA6	Syndromic microphthalmia Type 9
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
SUOX	Sulphocysteinuria
SURF1	Leigh syndrome, due to Cox deficiency
SYP	Mentally retarded, X-linked 96
TAT	Tyrosinemia Type 2
TAZ	Barth syndrome
TBCE	Hypoparathyroidism - Intellectual disc - Dysmorphic syndrome
TCF4	Pitt-Hopkins syndrome
TCIRG1	Autosomal recessive malignant osteopetrosis 1
TGM1	Ichthyosis, congenital, autosomal recessive 1
TH	Autosomal recessive-sensitive dystonia
TIMM8A	Mohr-Tranebjaerg syndrome

Gene	Pathology
TK2	Mitochondrial DNA depletion syndrome, myopathic form
TMEM67	Coach Syndrome
	Joubert syndrome 6
TPP1	Neuronal ceroid lipofuscinosis 2
TRAPPC9	Mental retardation, autosomal recessive 13
TREX1	Aicardi-Goutières syndrome
TRIM37	Mulibrey dwarfism
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
TSPYL1	Sudden infant death with Testes syndrome dysgenesis
TTPA	Ataxia with vitamin E deficiency
TUBA1A	Lissencephaly 3
TUFM	Combined oxidative phosphorylation deficiency 4
TUSC3	Ntal delay, autosomal recessive 7
TYK2	Immunodeficiency 35
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
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
UNC13D	Hemophagocytic lymphothiocytosis, familial, 3
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
VDR	VITAMIN DEPENDENT RICKETS TYPE 2A
VIPAR	Arthrogryposis, renal dysfunction and cholestasis 2
VLDLR	Cerebellar ataxia - Intellectual disc - Dysevelibum syndrome

Gene	Pathology
VPS13B	Cohen Syndrome Type 1
VPS33B	Arthrogryposis - renal dysfunction - cholestasis
WAS	Wiskott-Aldrich syndrome
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
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
ZIC3	Congenital heart defects, nonsyndromic, 1, X-linked
	Heterotaxy, visceral, 1, X-linked
ZNF469	Brittle cornea syndrome
ZNF711	Mentally retarded, X-linked 97