Gene	Pathology
ABCB11	Cholestasis, recurrent benign intrahepatic, 2
	Cholestasis, progressive familial intrahepatic 2
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
ACAD9	Acyl coa dehydrogenase deficiency 9
ACADM	Medium-chain acyl-COA dehydrogenase deficiency
ACADS	Acyl coa dehydrogenase, short chain, deficiency
ACADVL	Deficiency of very long chain acyl-CoA dehydrogenase
ACAT1	Ketoacidosis due to beta-ketoolase deficiency
ACOX1	Peroxisomal acyl-coa oxidase deficiency
ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease due to deficiency of glycogen debranching enzymes
AGPS	Rhizomelic chondrodysplasia punctata type 3
AGXT	Hyperoxaluria, primary, type 1
AIRE	Autoimmune polyedocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia
ALDH3A2	Sjogren-Larsson syndrome
ALG6	Congenital disorder of the IC glycosylation type
ALMS1	Alström syndrome
ALPL	Infantile onset hypophosphatasia
ALPL	Infantile hypophosphatasia
AMT	Glycine encephalopathy
AR	Complete androgen insensitivity syndrome
	Kennedy's disease
	Partial androgen insensitivity syndrome
	Hypospadias 1, X-linked
	Prostate cancer, susceptibility to
ARG1	Argininemia

Gene	Pathology
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis type 6
ARSE	Brachytelephalangic chondrodysplasia punctata
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASS1	Citrullinemia Type I.
ATM	Ataxia-telangiectasia
	Lymphoma, non-Hodgkin B-cell, somatic
	Lymphoma, mantle cell, somatic
	T-cell prolymphocytic leukemia, somatic
	Breast cancer, susceptibility to
ATP6V1B1	Renal tubular acidosis with deafness
ATP7A	Menkes disease
	Occipital horn syndrome
	X-linked distal spinal muscular atrophy
ATP7B	Wilson's disease
ATRX	Alpha-thalassemia myelodysplasia syndrome, somatic
	Alpha-thalassemia/mental retardation syndrome
	Intellectual disability syndrome and X-linked hypotonic facies
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)
BCS1L	Björnstad syndrome
	Gracilis syndrome
	Isolated coq-cytochrome C deficiency
	Leigh's syndrome
BLM	Bloom's syndrome

Gene	Pathology
BSND	Bartter Syndrome, Type 4A
	Sensory deafness with mild renal dysfunction
BTD	Biotinidase deficiency
BTK	Isolated growth hormone deficiency Type III
	X-linked agammaglobulinemia
CAPN3	Muscular dystrophy, club limbs, autosomal dominant 4
	Muscular dystrophy, club of limbs, autosomal recessive 1
CBS	Classical homocystinuria
	Thrombosis, hyperhomocysteinemia
CC2D2A	Coach Syndrome
	Joubert syndrome 9
	Meckel syndrome 6
CD40LG	X-linked hyper-IgM syndrome
CDH23	Autosomal recessive deafness 12
	Usher syndrome, type 1D
	Usher syndrome, digenic type 1D
	Pituitary adenoma 5, multiple types
CEP290	Joubert syndrome with oculorenal defect 5
	Joubert syndrome
	Leber congenital amaurosis 10
	Meckel's syndrome
	Senior-Loken syndrome
CERKL	Retinitis pigmentosa 26
CFTR	Cystic fibrosis
	Congenital bilateral absence of the vas deferens
	Elevation of sweat chloride without cystic fibrosis
	Bronchiectasis with or without elevated sweat chloride, modifier of
	Hypertrypsinemia, neonatal
	Hereditary pancreatitis
CHM	Choroiremia
CHRNE	Myostenic syndrome, congenital, 4a, slow canal
	Myostenic syndrome, congenital, 4b, fast canal
	Myostenic syndrome, congenital, 4c, associated with acetylcholine receptor deficiency

Gene	Pathology
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
	Retinitis pigmentosa 61
CNGB3	Achromatopsia 3
	Macular degeneration, juvenile
COL17A1	Generalized junctional epidermolysis bullosa, non-erlitz type
	Epidermolysis bullosa, junctional, localized variant
	Recurrent erosion epithelial dystrophy
COL4A3	Alport syndrome 3, autosomal dominant
	Alport syndrome 2, autosomal recessive
	Familial benign hematuria
COL4A4	Autosomal Recessive Alport Syndrome (COL4A4 Gene)
	Familial benign hematuria
COL4A5	Alport syndrome, X-linked
COL7A1	Dystrophic epidermolysis bullosa pruriginosa
	Dystrophic epidermolysis bullosa, AD
	Dystrophic epidermolysis bullosa, AR
	Epidermolysis bullosa, pretibial
	Toenail dystrophy, isolated
	Reverse EBD
	EBD, Bart type
	EBD, localized variant
	Transient thrombosis of the newborn
CPS1	Carbamoyl phosphate synthetase deficiency
	Pulmonary hypertension, neonatal, susceptibility to
	Venoocclusive disease after bone marrow transplant
CPT1A	Carnitine palmitoyltransferase 1a deficiency
CPT2	Carnitine palmitoyl Transferase II deficiency, infantile form
	Carnitine palmitoyl transferase II deficiency, neonatal form

Gene	Pathology
	Carnitine palmitoyl transferase II deficiency, myopathic, stress-induced
	Acute infection-induced encephalopathy, 4, susceptibility to
CRB1	Leber congenital Amaurosis 8
	Pigmented paravenous chorioretinal atrophy
	Retinitis pigmentosa-12
CRTAP	Osteogenesis imperfecta type VII
CSTB	Unverricht-Lundborg disease
CTNS	Cystinosis, atypical nephropathic
	Cystinosis, juvenile or adolescent nephropathic with late onset
	Cystinosis, nephropathic
	Non-nephropathic ocular cystinosis
CTSD	Adult neuronal ceroid lipofuscinosis 10
CTSK	Pycnodysostosis
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
	Low-renin hypertension, susceptibility to
CYP17A1	7,20-lyase deficiency, isolated
	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
DBT	Classic maple syrup urine disease
DCLRE1C	Omenn syndrome
	Severe combined immunodeficiency due to DCLRE1c deficiency
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

Gene	Pathology
DLD	Dihydrolipoamide dehydrogenase deficiency
DMD	Becker muscular dystrophy
	Dilated cardiomyopathy, 3B
	Duchenne muscular dystrophy
DNAH5	Ciliary dyskinesia, primary, 3, with or without situs inversus
DNAI1	Ciliary dyskinesia, primary, 1, with or without situs inversus
DNAI2	Ciliary dyskinesia, primary, 9, with or without situs inversus
DOK7	Deformation sequence of Fetal Akinesia
	Myasthenia, girle of the limbs, family members
DPYD	Dihydropyrimidine dehydrogenase deficiency
	Congenital myasthenic syndrome, 10
DYSF	Miyoshi muscular dystrophy 1
	Muscular dystrophy, club limbs, autosomal recessive 2
	Myopathy, distal, with anterior tibial onset
EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked
EDAR	Ectodermal dysplasia 10a, hypohidrotic/hair/nail type, autosomal dominant
	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive
	Hair morphology 1, hair thickness
EIF2AK3	Wolcott-Rallison syndrome
EIF2B1	Leukoencephalopathy with disappearance of the white matter
EIF2B2	Leukoencephalopathy with disappearance of the white matter
	Ovarioleukodystrophy
EIF2B3	Leukoencephalopathy with disappearance of the white matter
EIF2B4	Leukoencephalopathy with disappearance of the white matter
	Ovarioleukodystrophy
EIF2B5	Leukoencephalopathy with escaping white matter
	Ovarioleukodystrophy
EMD	Muscular dystrophy 1, X-linked.
ERCC6	Cockayne syndrome type B
	Cerebro-oculo-facio-skeletal syndrome 1
	De Sanctis-Cacchione syndrome
	Premature ovarian failure 1
	UV sensitivity syndrome 1
	Lung cancer, susceptibility to

Gene	Pathology
	Macular degeneration, age-related, susceptibility to, 5
ERCC8	Cockayne syndrome Type A
	UV sensitivity syndrome 2
ESCO2	Roberts syndrome
	Phocomelia syndrome SC
ETFA	Glutaric Acidemia Type 2 (ETFA Gene)
ETFB	Glutaric acidemia type 2 (ETB gene)
ETFDH	Glutaric acidemia type 2 (ETFDH gene)
ETHE1	Ethylmalonic encephalopathy
EVC2	Ellis-Van Creveld syndrome
	Weyers acrodental dysostosis
EVC2	Ellis-Van Creveld syndrome
EYS	Retinitis pigmentosa 25
F11	Factor XI deficiency, autosomal dominant
	Factor XI Deficiency, autosomal recessive
F8	Hemophilia a
F9	Hemophilia b
	Thrombophilia, X-linked, due to a defect in factor IX
	Deep vein thrombosis, protection from
	Sensitivity to warfarin
FAH	Tyrosinemia Type 1
FANCA	Fanconi anemia, complementation group A
FANCC	Fanconi anemia complementation group C.
FANCG	Fanconi Anemia, Complementation Group G
FH	Fumaric aciduria
	Leiomyomatosis and renal cell cancer
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and ocular anomalies), type A, 5
	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5
FKTN	Dilated cardiomyopathy, 1X
	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4

Gene	Pathology
	Muscular dystrophy-dystroglycanopathy (limb-arm), type C, 4
FMR1	Fragile X syndrome
	Fragile X tremor/ataxia syndrome
	Premature ovarian failure 1
G6PC	Glycogen storage disease due to glucose-6-phosphatase type 1A deficiency
G6PD	Favism
	Hemolytic anemia due to G6PD deficiency
GAA	Glycogen storage disease due to acid maltase deficiency
GALC	Krabbe disease
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis IVA
GALT	Classic galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GBA	Perinatal lethal gaucher disease
	Type 1 Gaucher disease
	Type 2 Gaucher disease
	Type 3 Gaucher disease
	Gaucher disease Type 3C
	Dementia with Lewy bodies, susceptibility to
	Late-onset Parkinson's disease, susceptibility to
GBE1	Glycogen storage disease, type IV
	Polyglucose body disease, adult form
GCDH	Glutaryl-CoA dehydrogenase deficiency
GFM1	Hepaenencephalopathy due to combined deficiency of type 1 oxidative phosphorylation
GJB1	Carcotta-maritime neuropathy, X-linked dominant, 1
GJB2	Bart-Pumphrey syndrome
	Autosomal dominant deafness 3A
	Autosomal recessive deafness 1A
	Keratitis-ichthyosis-deafness syndrome
	Hystrix type ichthyosis with deafness
	Palmoplantar keratoderma with deafness
	Vohwinkel syndrome
GJB6	Deafness, autosomal dominant 3B

Gene	Pathology
	Deafness, autosomal recessive 1B
	Deafness, digenic GJB2/GJB6
	Ectodermal dysplasia 2, Clouston type
GLA	Fabry disease
	Fabry disease, cardiac variant
GLB1	GM1 Gangliosidosis Type 1
	GM1 Gangliosidosis Type 2
	GM1 Gangliosidosis Type 3
	Mucopolysaccharidosis Type 4B
GLDC	Glycine encephalopathy
GLE1	Congenital arthrogryposis with anterior horn cell disease
	Lethal congenital contracture syndrome Type 1
GNE	Nonaka myopathy
	Sialiaria
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis type 2
	Mucolipidosis type 3
GNS	Mucopolysaccharidosis type IIID
GP9	Bernard-Soulier syndrome, type C
GRHPR	Hyperoxaluria, primary, type II
GUSB	Mucopolysaccharidosis type 7
HADHA	Fatty liver, acute, of pregnancy
	HELLP syndrome, maternal, of pregnancy
	LCHAD deficiency
	Deficiency of trifunctional proteins
HADHB	Deficiency of trifunctional proteins
HAX1	Neutropenia, severe congenital 3, autosomal recessive
HBA1	Erythrocytosis, 7
	Heinz body anemias, alpha
	Non-deleterious hemoglobin H disease
	Methemoglobinemia, alpha type
	Alpha-thalassemia
HBA2	Erythrocytosis 7
	Heinz body anemia

Gene	Pathology
	Hemoglobin H disease, deletional and non-deletional
	Alpha-thalassemia
HBB	Beta-thalassemia
	Delta-beta thalassemia
	Erythrocytosis 6
	Heinz body anemia
	Hereditary persistence of fetal hemoglobin
	Metamoglobinemia, beta type
	Sickle cell anemia
	Beta-thalassemia
	Beta-thalassemia, inclusion body dominant
HEXA	GM2-gangliosidosis, different forms
	Tay-Sachs disease
	Hexosaminidase A pseudodeficiency
HEXB	Sandhoff disease, infantile, juvenile and adult forms
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
HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo C)
	Retinitis pigmentosa 73
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaric aciduria
HOGA1	Hyperoxaluria, primary, type III
HPS1	Hermansky-pudlak syndrome 1
HPS3	Hermansky-pudlak syndrome 3
HSD17B4	D-bifunctional protein deficiency
	Perrault syndrome
HSD3B2	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
HYLS1	Hydroletus syndrome

Gene	Pathology
IDS	Mucopolysaccharidosis type 2
IDUA	Mucopolysaccharidosis IH
	Mucopolysaccharidosis ih/s
	Mucopolysaccharidosis is
IKBKAP	Familial dysautonomy
IL2RG	T-B+ severe combined immunodeficiency due to gamma chain deficiency
	Severe, X-linked T-B+ combined immunodeficiency
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7
IVD	Isovaleric acidemia
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
L1CAM	Partial agenesis of the corpus callosum
	CRASH syndrome
	Hydrocephalus due to stenosis of the aqueduct
	Hydrocephalus with idiopathic congenital intestinal pseudoobstruction
	Hydrocephalus with Hirschsprung's disease
	MASA syndrome
LAMA2	Congenital muscular dystrophy type 1a
	Bullosa junctional epidermolysis, Herlitz type (Lama3 gene)
LAMA3	Bullosa junctional epidermolysis, Herlitz type
	Epidermolysis bullosa, benign generalized atrophic
	Laryngo-monocutaneous syndrome
LAMB3	Amelogenesis imperfecta, type 1A
	Junctional epidermolysis bullosa, Herlitz type
	Junctional epidermolysis bullosa, non-Herlitz type
LAMC2	Junctional epidermolysis bullosa, Herlitz type
	Junctional epidermolysis bullosa, non-Herlitz type
LCA5	Leber congenital Amaurosis 5
LHCGR	Leydig cell adenoma, somatic, with precocious puberty

Gene	Pathology
	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
LIPA	Cholesteria ester storage disease
	Wolman's disease
LIPH	Hypotrichosis 7
	Woolly hair, autosomal recessive 2 with or without hypotrichosis
LOXHD1	Deafness, autosomal recessive 77
LPL	Combined, familial hyperlipidemia
	High-density lipoprotein Lipoprotel QTL level 11
	Lipoprotein lipase deficiency
LRPPRC	Leigh syndrome of the French-Canadian type
LYST	Chediak-Higashi syndrome
MAN2B1	Mannosidosis, alpha-, types I and II
MCCC1	3-methylcrotonic-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonic-CoA carboxylase 2 deficiency
MCOLN1	Mucolipidosis Type 4
MECP2	Severe neonatal-onset encephalopathy
	X-linked syndromic mental retardation, Lubs type
	Syndromic X-linked mental retardation, 13
	Rett syndrome
	Rett syndrome, preserved vocal variant
	Rett syndrome, atypical
	Susceptibility to autism, X-linked 3
MED17	Microcephaly, progressive postnatal, with seizures and cerebral atrophy
MEFV	Familial Mediterranean fever, AD
	Familial Mediterranean fever, AR
MFSD8	Macular dystrophy with central cone involvement
	Neuronal ceroid lipofuscinosis, 7
MKS1	Bardet-Biedl syndrome 13
	Joubert syndrome 28

Gene	Pathology
	Meckel Syndrome Type 1
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts
MMAA	Methylmalonic acidemia sensitive to vitamin B12 CBLA
MMAB	Methylmalonic acidemia sensitive to vitamin B12 CBLB
MMACHC	Methylmalonic acidemia with homocystinuria, CBLC type
MMADHC	Homocystinuria, CBLD type, variant 1
MPI	Congenital glycosylation disorder type 1b
MPL	Thrombocythemia 2
	Myelofibrosis with myeloid metaplasia, somatic
	Thrombocytopenia, congenital amegakaryocytosis
MPV17	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)
MTHFR	Homocystinuria due to MTHFR deficiency
	Neural tube defects, predisposition to
	Schizophrenia, predisposition to
	Thromboembolism, predisposition to
	Vascular diseases, predisposition to
MTM1	X-linked centronuclear myopathy
MTTP	Abetalipoproteinemia
	Metabolic syndrome, protection against
MUSK	Fetal akinesia deformation sequence
	Myasthenic syndrome, congenital, 9, associated with
MUT	Methylmalonic aciduria, mut(0) type
MYO7A	Deafness, autosomal dominant 11
	Deafness, autosomal recessive 2
	Usher Syndrome Type 1B
NAGLU	Charcot-Marie-Tooth disease, axonal, type 2V
	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	Hyperammonemia due to n-acetylglutamate synthetase deficiency
NBN	Aplastic anemia
	Acute lymphoblastic leukemia
	Nijmegen rupture syndrome
NDUFS6	Complex I, mitochondrial respiratory chain, deficiency
NEB	Nemaline Myopathy 2
NPC1	Niemann-Pick Disease Type C1

Gene	Pathology
	Niemann-Pick Type D disease
NPC2	Niemann-Pick disease Type C2
NPHP1	Joubert syndrome 4
	Nephronophthisis 1, juvenile
	Senior-Loken syndrome
NPHS1	Nephrotic syndrome, type 1
NPHS2	Nephrotic syndrome, type 2
NR2E3	S-Cone syndrome improved
	Retinitis pigmentosa 37
NTRK1	Insensitivity to pain, congenital, with anhidrosis
	Familial medullary thyroid carcinoma
OCRL	Dental disease 2
	Lowe's syndrome
OPA3	3-methylglutaconic aciduria, type 3
	Optic atrophy 3 with cataract
OTC	Ornithine transcarbamylase deficiency
PAH	Phenylketonuria
	Hyperphenylalaninemia, mild non-PKU
PANK2	Neurodegeneration with accumulation of iron in the brain 1
	HARP syndrome
PC	Pyruvate carboxylase deficiency
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
PDHA1	Leigh syndrome, X-linked
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PEX1	Heimler syndrome 1
	Peroxisome biogenesis disorder 1A (Zellweger)
	Peroxisome biogenesis disorder 1B (NALD/IRD)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)
	Peroxisome biogenesis disorder 6B
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)

Gene	Pathology
	Peroxisome biogenesis disorder 3B
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)
	Peroxisome biogenesis disorder 5B
PEX26	Peroxisome biogenesis disorder 7A (Zellweger)
	Peroxisome biogenesis disorder 7B
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)
	Peroxisome biogenesis disorder 4B
	Heimler syndrome 2
PEX7	Peroxisome biogenesis disorder 9B
	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease VII
PHGDH	Neu-Laxova syndrome 1
	Phosphoglycerate dehydrogenase deficiency
PKHD1	Autosomal recessive polycystic kidney disease
PLA2G6	Infantile neuroaxonal dystrophy 2A
	Neurodegeneration with iron accumulation in the brain 2B
	Parkinson's disease 14, autosomal recessive
PMM2	Congenital glycosylation disorder type 1A
POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type)
	Mitochondrial DNA depletion syndrome 4B (MNGIE type)
	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)
	Progressive external ophthalmoplegia, autosomal dominant 1
	Progressive external ophthalmoplegia, autosomal recessive 1
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3
	Retinitis pigmentosa 76
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and ocular anomalies), type A, 1
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1
POMT2	Muscular dystrophy-dystroglycanopathy (congenital with brain and ocular anomalies), type A, 2

Gene	Pathology
	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2
	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2
PPT1	Adult neuronal ceroid lipofuscinosis
PROP1	Pituitary hormone deficiency, combined, 2
PRPS1	Arts Syndrome
	Charcot-Marie-Tooth disease, X-linked, recessive, 5
	Deafness, X-linked 1
	Gout, related to PRPS
	Superactivity of phosphoribosylpyrophosphate synthetase
PSAP	Combined prosaposin deficiency
	Gaucher disease, atypical
	Krabber's disease, atypical
	Superactivity of phosphoribosylpyrophosphate synthetase
PTS	Hyperphenylalaninemia, with BH4 deficiency, a
PUS1	Myopathy, lactic acidosis and sideroblastic anemia 1
PYGM	Glycogen storage disease due to muscle glycogen phosphorylase deficiency
RAB23	Carpenter's syndrome
RAG1	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity
	Cellular and humoral immune defects combined with granulomas
	severe combined immunodeficiency, B cell negative
	Omenn syndrome
RAG2	Cellular and humoral immune defects combined with granulomas
	severe combined immunodeficiency, B cell negative
	Omenn syndrome
RAPSN	Fetal akinesia deformation sequence
	Congenital myasthenic syndrome, 11, associated with acetylcholine receptor deficiency
RARS2	Pontocerebellar Hypoplasia, Type 6
RDH12	Leber congenital Amaurosis 13
RMRP	Anuxtic dysplasia
	Cartilage hair hypoplasia
	Metaphyseal dysplasia without hypotrichosis
RPE65	Leber congenital Amaurosis 2

Gene	Pathology
	Retinitis pigmentosa 20
RPGRIP1L	Joubert syndrome, Type 7
	Meckel syndrome, Type 5
	COACH syndrome
RS1	Retinoschisis
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay
SAMHD1	Aicardi-Goutieres syndrome 5
	Lupus pernio (Chilblain Lupus) 2
SBDS	Shwachman-Diamond syndrome
	Aplastic anemia, predisposition to
SEPSECS	Pontocerebellar Hypoplasia Type 2D
SERPINA1	Emphysema due to AAT deficiency
	Emphysema-cirrhosis, due to AAT deficiency
	Hemorrhagic diathesis due to Pittsburgh antithrombin
	Chronic obstructive pulmonary disease, susceptibility to
SGCA	Muscular dystrophy, club limbs, autosomal recessive 3
SGCB	Muscular dystrophy, club limbs, autosomal recessive 4
SGCG	Muscular dystrophy, club limbs, autosomal recessive 5
SGSH	Mucopolysaccharidosis Type 3A (Sanfilippo syndrome type A)
SLC12A3	Gitelman syndrome
SLC12A6	Corpus callosum Agenesis - Neuronopathy
SLC17A5	Free sialic acid retention disease, infantile form
	Salla's disease
SLC22A5	Carnitine deficiency, primary systemic
SLC25A13	Citrullinemia, adult-onset type II
	Citrullinemia, type II, neonatal onset
SLC25A15	Hyperornithinemia-hyperammemia-homocitrullinuria
SLC25A20	Carnitine-acylcarnitine deficiency
SLC26A2	Achondrogenesis Type 1B
	Type II atelosteogenesis
	Diastrophic dysplasia
	Diastrophic dysplasia, broad-platyspondyl bone variant
	De la Chapelle dysplasia
	Multiple epiphyseal dysplasia, 4

Gene	Pathology
SLC26A4	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct
	Pendred syndrome
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
SLC4A11	Fuchs endothelial corneal dystrophy, 4
	Corneal endothelial dystrophy and perceptual deafness
	Corneal endothelial dystrophy, autosomal recessive
SLC6A8	Cerebral creatine deficiency syndrome 1
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 Type A disease
STAR	Congenital lipoid adrenal hyperplasia
SUMF1	Multiple sulfatase deficiency
TAT	Tyrosinemia Type 2
TCIRG1	Autosomal recessive malignant osteopetrosis 1
TFR2	Hemochromatosis, type 3
TGM1	Ichthyosis, congenital, autosomal recessive 1
TH	Autosomal recessive-sensitive dystonia
TMEM216	Joubert syndrome 2
	Meckel syndrome 2
TPP1	Neuronal ceroid lipofuscinosis 2
	Autosomal recessive spinocerebellar ataxia 7
TREX1	Aicardi-Goutières 1 syndrome, dominant and recessive
	Lupus pernio (Chilblain Lupus)
	Retinal vasculopathy with cerebral leukodystrophy
	Systemic lupus erythematosus, susceptibility to
TRIM37	Mulibrey dwarfism
TSEN2	Pontocerebellar hypoplasia type 2B
TSEN34	Pontocerebellar hypoplasia type 2C
TSEN54	Pontocerebellar hypoplasia type 2A
	Pontocerebellar hypoplasia type 4

Gene	Pathology
	Pontocerebellar hypoplasia type 5
TSFM	Fatal mitochondrial disease due to combined deficiency of oxidative phosphorylation 3
TSHB	Isolated thyroid-stimulating hormone deficiency
TTC37	Trichoheparaenteric syndrome 1
TTPA	Ataxia with vitamin E deficiency
TYMP	Mitochondrial DNA depletion syndrome 1 (MNGIE type)
TYR	Albinism, oculocutaneous, type 1A
	Albinism, oculocutaneous, type 1B
	Waardenburg syndrome/digenic albinism
	Skin/hair/eye pigmentation 3, blue/green eyes
	Skin/hair/eye pigmentation 3, light/dark/freckled skin
	Melanoma, cutaneous malignancy, susceptibility to, 8
UGT1A1	Bilirubin, serum level of, QTL1
	Crigler-Najjar syndrome, type I
	Crigler-Najjar syndrome, type II
	Gilbert's syndrome
	Hyperbilirubinemia, transient familial neonatal
USH1C	Deafness, autosomal recessive 18A
	Usher Syndrome Type 1C
USH2A	Usher Syndrome Type 2A
	Retinitis pigmentosa 39
VPS13A	Choreoacanthosis
VPS13B	Cohen Syndrome Type 1
VRK1	Pontocerebellar Hypoplasia Type 1A
WAS	Severe, X-linked congenital neutropenia
	Thrombocytopenia, X-linked
	X-linked intermittent thrombocytopenia
	Wiskott-Aldrich syndrome
WNT10A	Odontoioncodermal dysplasia
	Schopf-Schulz-Passarge syndrome
	Tooth agenesis, selective, 4
XPA	Xeroderma Pigmentosum Complementation Group A
XPC	Xeroderma Pigmentosum, group C

Gene	Pathology
ZFYVE26	Spastic paraplegia 15, autosomal recessive