

Newborn Genetic Screening 737-Gene Panel

Detects pathogenic or likely pathogenic variants across 737 genes associated with 1,287 single-gene disorders

Disorders	Genes	Inheritance
Cholestasis, progressive familial intrahepatic 2	ABCB11	AR
Cholestasis, benign recurrent intrahepatic, 2		AR
Gallbladder disease 1	ABCB4	AD/AR
Cholestasis, progressive familial intrahepatic 3		AR
Cholestasis, intrahepatic, of pregnancy, 3	ABCD1	AD/AR
Adrenomyeloneuropathy, adult		XLR
Adrenoleukodystrophy	ABCD1	XLR
Sitosterolemia 2	ABCG5	AR
Sitosterolemia 1	ABCG8	AR
Isobutyryl-CoA dehydrogenase deficiency	ACAD8	AR
Acyl-CoA dehydrogenase, medium chain, deficiency of	ACADM	AR
Acyl-CoA dehydrogenase, short-chain, deficiency of	ACADS	AR
2-methylbutyrylglycinuria	ACADSB	AR
VLCAD deficiency	ACADVL	AR
Alpha-methylacetoacetic aciduria	ACAT1	AR
Spondyloenchondrodysplasia with immune dysregulation	ACP5	AR
Baraitser-Winter syndrome 1	ACTB	AD
Atrial septal defect 5	ACTC1	AD
Cardiomyopathy, hypertrophic, 11		AD
Cardiomyopathy, dilated, 1R		AD
Left ventricular noncompaction 4		AD
Baraitser-Winter syndrome 2	ACTG1	AD
Deafness, autosomal dominant 20/26		AD
Glomerulosclerosis, focal segmental, 1	ACTN4	AD
Telangiectasia, hereditary hemorrhagic, type 2	ACVRL1	AD
Severe combined immunodeficiency due to ADA deficiency	ADA	AR/SMo
Adenosine deaminase deficiency, partial		AR/SMo
Sneddon syndrome	ADA2	AR
Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome		AR
Thrombotic thrombocytopenic purpura, hereditary	ADAMTS13	AR

Disorders	Genes	Inheritance
Aicardi-Goutieres syndrome 6	ADAR	AR
Dyschromatosis symmetrica hereditaria		AD
Deafness, autosomal recessive 44	ADCY1	AR
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	AHCY	AR
Immunodeficiency with hyper-IgM, type 2	AICDA	AR
Cowchock syndrome	AIFM1	XLR
Deafness, X-linked 5		XLR
Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy		XLR
Combined oxidative phosphorylation deficiency 6		XLR
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	AIRE	AD/AR
Reticular dysgenesis	AK2	AR
Bile acid synthesis defect, congenital, 2	AKR1D1	AR
Porphyria, acute hepatic	ALAD	AR
Protoporphyrin, erythropoietic, X-linked	ALAS2	XL
Anemia, sideroblastic, 1		XLR
Sjogren-Larsson syndrome	ALDH3A2	AR
Fructose intolerance, hereditary	ALDOB	AR
Congenital disorder of glycosylation, type Ik	ALG1	AR
Alstrom syndrome	ALMS1	AR
Hypophosphatasia, adult	ALPL	AD/AR
Hypophosphatasia, childhood		AR
Odontohypophosphatasia		AD/AR
Hypophosphatasia, infantile		AR
Imerslund-Grasbeck syndrome 2	AMN	AR
Glycine encephalopathy 2	AMT	AR
Spherocytosis, type 1	ANK1	AD/AR
Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	ANOS1	XLR
Hermansky-Pudlak syndrome 2	AP3B1	AR
Gardner syndrome	APC	AD
Adenomatous polyposis coli		AD
Brain tumor-polyposis syndrome 2		AD
Gastric adenocarcinoma and proximal polyposis of the stomach		AD
Desmoid disease, hereditary		AD
Hypobetalipoproteinemia	APOB	AR
Hypercholesterolemia, familial, 2		AD
Diabetes insipidus, nephrogenic, 2	AQP2	AD/AR
Androgen insensitivity, partial, with or without breast cancer	AR	XLR
Spinal and bulbar muscular atrophy, X-linked 1		XLR
Hypospadias 1, X-linked		XLR
Androgen insensitivity		XLR

Disorders	Genes	Inheritance
Argininemia	<i>ARG1</i>	AR
Metachromatic leukodystrophy	<i>ARSA</i>	AR
Argininosuccinic aciduria	<i>ASL</i>	AR
Citrullinemia	<i>ASS1</i>	AR
Spastic paraplegia 3A, autosomal dominant	<i>ATL1</i>	AD
Neuropathy, hereditary sensory, type ID		AD
Ataxia-telangiectasia	<i>ATM</i>	AR
{Deafness, autosomal recessive 12, modifier of}	<i>ATP2B2</i>	AR
Deafness, autosomal dominant 82		AD
Distal renal tubular acidosis 3, with or without sensorineural hearing loss	<i>ATP6V0A4</i>	AR
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	<i>ATP6V1B1</i>	AR
Menkes disease	<i>ATP7A</i>	XLR
Neuronopathy, distal hereditary motor, X-linked		XLR
Occipital horn syndrome		XLR
Wilson disease	<i>ATP7B</i>	AR
Cholestasis, progressive familial intrahepatic 1	<i>ATP8B1</i>	AR
Cholestasis, benign recurrent intrahepatic		AR
Cholestasis, intrahepatic, of pregnancy, 1		AD
3-methylglutaconic aciduria, type I	<i>AUH</i>	AR
Nephrogenic syndrome of inappropriate antidiuresis	<i>AVPR2</i>	XLR
Diabetes insipidus, nephrogenic, 1		XLR
Immunodeficiency 43	<i>B2M</i>	AR
Myopathy, myofibrillar, 6	<i>BAG3</i>	AD
Cardiomyopathy, dilated, 1HH		AD
Maple syrup urine disease, type Ia	<i>BCKDHA</i>	AR
Maple syrup urine disease, type Ib	<i>BCKDHB</i>	AR
Immunodeficiency 37	<i>BCL10</i>	AR
Cataract 12, multiple types	<i>BFSP2</i>	AD
Bloom syndrome	<i>BLM</i>	AR
Agammaglobulinemia 4	<i>BLNK</i>	AR
Hermansky-Pudlak syndrome 8	<i>BLOC1S3</i>	AR
Hermansky-Pudlak syndrome 9	<i>BLOC1S6</i>	AR
Pulmonary venoocclusive disease 1	<i>BMPR2</i>	AD
Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated		AD
Pulmonary hypertension, familial primary, 1, with or without HHT		AD
LEOPARD syndrome 3	<i>BRAF</i>	AD
Noonan syndrome 7		AD
Cardiofaciocutaneous syndrome		AD
Bartter syndrome, type 4a	<i>BSND</i>	AR
Sensorineural deafness with mild renal dysfunction		AR

Disorders	Genes	Inheritance
Biotinidase deficiency	<i>BTBD</i>	AR
Isolated growth hormone deficiency, type III, with agammaglobulinemia	<i>BTK</i>	XLR
Agammaglobulinemia, X-linked 1		XLR
C1q deficiency 1	<i>C1QA</i>	AR
C1q deficiency 2	<i>C1QB</i>	AR
C1q deficiency 3	<i>C1QC</i>	AR
Ehlers-Danlos syndrome, periodontal type, 1	<i>C1R</i>	AD
C1s deficiency	<i>C1S</i>	.
Ehlers-Danlos syndrome, periodontal type, 2		AD
C2 deficiency	<i>C2</i>	AR
C3 deficiency	<i>C3</i>	AR
C4a deficiency	<i>C4A</i>	AR
C4B deficiency	<i>C4B</i>	.
C5 deficiency	<i>C5</i>	AR
C6 deficiency	<i>C6</i>	AR
C7 deficiency	<i>C7</i>	.
C8 deficiency, type I	<i>C8A</i>	AR
C8 deficiency, type II	<i>C8B</i>	AR
C9 deficiency	<i>C9</i>	.
Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	<i>CA2</i>	AR
Deafness, autosomal recessive 93	<i>CABP2</i>	AR
Ventricular tachycardia, catecholaminergic polymorphic, 4	<i>CALM1</i>	AD
Long QT syndrome 14		AD
Long QT syndrome 15	<i>CALM2</i>	AD
Long QT syndrome 16	<i>CALM3</i>	AD
B-cell expansion with NFKB and T-cell anergy	<i>CARD11</i>	AD
Immunodeficiency 11A		AR
Immunodeficiency 11B with atopic dermatitis		AD
Pityriasis rubra pilaris	<i>CARD14</i>	AD
Psoriasis 2		AD
Immunodeficiency 103, susceptibility to fungal infection	<i>CARD9</i>	AR
FG syndrome 4	<i>CASK</i>	XLR
Intellectual developmental disorder, with or without nystagmus		XLR
Intellectual developmental disorder and microcephaly with pontine and cerebellar hypoplasia		XL
Autoimmune lymphoproliferative syndrome, type II	<i>CASP10</i>	AD
Caspase 8 lymphadenopathy syndrome	<i>CASP8</i>	AR
Homocystinuria, B6-responsive and nonresponsive types	<i>CBS</i>	AR
Thrombosis, hyperhomocysteinemic		AR
Hennekam lymphangiectasia-lymphedema syndrome 1	<i>CCBE1</i>	AR

Disorders	Genes	Inheritance
Ciliary dyskinesia, primary, 14	<i>CCDC39</i>	AR
Ciliary dyskinesia, primary, 15	<i>CCDC40</i>	AR
Deafness, autosomal dominant 44	<i>CCDC50</i>	AD
Immunodeficiency, common variable, 3	<i>CD19</i>	AR
Immunodeficiency 25	<i>CD247</i>	AR
Lymphoproliferative syndrome 2	<i>CD27</i>	AR
Immunodeficiency 19, severe combined	<i>CD3D</i>	AR
Immunodeficiency 18	<i>CD3E</i>	AR
Immunodeficiency 18, SCID variant		AR
Immunodeficiency 17, CD3 gamma deficient	<i>CD3G</i>	AR
Immunodeficiency with hyper-IgM, type 3	<i>CD40</i>	AR
Immunodeficiency, X-linked, with hyper-IgM	<i>CD40LG</i>	XLR
Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	<i>CD55</i>	AR
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	<i>CD59</i>	AR
Agammaglobulinemia 3	<i>CD79A</i>	AR
Agammaglobulinemia 6	<i>CD79B</i>	AR
Immunodeficiency, common variable, 6	<i>CD81</i>	AR
Immunodeficiency 116	<i>CD8A</i>	AR
Dyserythropoietic anemia, congenital, type Ia	<i>CDAN1</i>	AR
Usher syndrome, type 1D	<i>CDH23</i>	AR/DR
Deafness, autosomal recessive 12		AR
Usher syndrome, type 1D/F digenic		AR/DR
Deafness, autosomal recessive 113	<i>CEACAM1</i> 6	AR
Deafness, autosomal dominant 4B		AD
Specific granule deficiency	<i>CEBPE</i>	AD/AR
Joubert syndrome 5	<i>CEP290</i>	AR
Leber congenital amaurosis 10		.
Meckel syndrome 4		AR
Senior-Loken syndrome 6		AR
Complement factor D deficiency	<i>CFD</i>	AR
Complement factor H deficiency	<i>CFH</i>	AD/AR
Basal laminar drusen		AD
Nephropathy due to CFHR5 deficiency	<i>CFHR5</i>	AD
Complement factor I deficiency	<i>CFI</i>	AR
Properdin deficiency, X-linked	<i>CFP</i>	XLR
{Bronchiectasis with or without elevated sweat chloride 1, modifier of}	<i>CFTR</i>	AD
Cystic fibrosis		AR
CHARGE syndrome	<i>CHD7</i>	AD
Hypogonadotropic hypogonadism 5 with or without anosmia		AD

Disorders	Genes	Inheritance
Myasthenic syndrome, congenital, 4B, fast-channel	<i>CHRNAE</i>	AR
Myasthenic syndrome, congenital, 4A, slow-channel		AD/AR
Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency		AR
Usher syndrome, type IJ	<i>CIB2</i>	AR
Deafness, autosomal recessive 48		AR
MHC class II deficiency 1	<i>CIITA</i>	AR
Myotonia congenita, dominant	<i>CLCN1</i>	AD
Myotonia congenita, recessive		AR
Myotonia levior		AD
Dent disease 1	<i>CLCN5</i>	XLR
Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis		XLR
Hypophosphatemic rickets		XLR
Nephrolithiasis, type I	<i>CLCNKB</i>	AR
Bartter syndrome, type 3		DR
Bartter syndrome, type 4b, digenic	<i>CLDN14</i>	AR
Deafness, autosomal recessive 29	<i>CLDN16</i>	AR
Hypomagnesemia 3, renal	<i>CLIC5</i>	AR
Deafness, autosomal recessive 103	<i>CLPB</i>	AD
3-methylglutaconic aciduria, type VIIA, autosomal dominant		AD
3-methylglutaconic aciduria, type VIIB, autosomal recessive		AR
Neutropenia, severe congenital, 9, autosomal dominant	<i>CLPP</i>	AD
Perrault syndrome 3	<i>CLRN1</i>	AR
Usher syndrome, type 3A		AR
Retinitis pigmentosa 61	<i>CNBP</i>	.
Myotonic dystrophy 2	<i>COCH</i>	AD
Deafness, autosomal recessive 110		AR
Deafness, autosomal dominant 9	<i>COL11A1</i>	AD
Marshall syndrome		AD
Stickler syndrome, type II		AD
Deafness, autosomal dominant 37		AR
Fibrochondrogenesis 1	<i>COL11A2</i>	AD
Otospondylomegapiphyseal dysplasia, autosomal dominant		AR
Otospondylomegapiphyseal dysplasia, autosomal recessive		AD
Deafness, autosomal dominant 13		AR
Deafness, autosomal recessive 53		AD/AR
Fibrochondrogenesis 2	<i>COL17A1</i>	AD
Epithelial recurrent erosion dystrophy		AR
Epidermolysis bullosa, junctional 4, intermediate		

Disorders	Genes	Inheritance
Caffey disease	COL1A1	AD
Osteogenesis imperfecta, type III		AD
Osteogenesis imperfecta, type II		AD
Osteogenesis imperfecta, type IV		AD
Osteogenesis imperfecta, type I		AD
Ehlers-Danlos syndrome, arthrochalasia type, 1		AD
Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1		AD
Osteogenesis imperfecta, type III	COL1A2	AD
Osteogenesis imperfecta, type II		AD
Osteogenesis imperfecta, type IV		AD
Ehlers-Danlos syndrome, arthrochalasia type, 2		AD
Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2		AD
Ehlers-Danlos syndrome, cardiac valvular type		AR
Czech dysplasia	COL2A1	AD
Kniest dysplasia		AD
Legg-Calve-Perthes disease		AD
Spondyloepiphyseal dysplasia, Stanescu type		AD
Stickler syndrome, type I		AD
SMED Strudwick type		AD
Platyspondylic skeletal dysplasia, Torrance type		AD
Stickler syndrome, type I, nonsyndromic ocular		AD
Avascular necrosis of the femoral head		AD
Osteoarthritis with mild chondrodysplasia		AD
Spondyloperipheral dysplasia		AD
Achondrogenesis, type II or hypochondrogenesis		AD
SED congenita		AD
Alport syndrome 3A, autosomal dominant	COL4A3	AD
Alport syndrome 3B, autosomal recessive		AR
Hematuria, benign familial, 2		AD
Alport syndrome 2, autosomal recessive	COL4A4	AR
Hematuria, familial benign, 1		AD
Alport syndrome 1, X-linked	COL4A5	XLD
Deafness, X-linked 6	COL4A6	XLR
Epiphyseal dysplasia, multiple, 6	COL9A1	AD
Stickler syndrome, type IV		AR
Stickler syndrome, type V	COL9A2	AR
Epiphyseal dysplasia, multiple, 2		AD
Stickler syndrome, type VI	COL9A3	AR
Epiphyseal dysplasia, multiple, 3, with or without myopathy		AD
Myasthenic syndrome, congenital, 5	COLQ	AR

Disorders	Genes	Inheritance
Epiphyseal dysplasia, multiple, 1	COMP	AD
Pseudoachondroplasia		AD
Carpal tunnel syndrome 2		AD
{Autoinflammation and autoimmunity, systemic, with immune dysregulation}	COPA	AD
Coenzyme Q10 deficiency, primary, 1	COQ2	AR
Coenzyme Q10 deficiency, primary, 6	COQ6	AR
Nephrotic syndrome, type 9	COQ8B	AR
Immunodeficiency 8	CORO1A	AR
Coproporphria	CPOX	AD/AR
Harderoporphyria		AR
Carbamoylphosphate synthetase I deficiency	CPS1	AR
CPT deficiency, hepatic, type IA	CPT1A	AR
CPT II deficiency, lethal neonatal	CPT2	AR
CPT II deficiency, myopathic, stress-induced		AD/AR
CPT II deficiency, infantile		AR
Leber congenital amaurosis 8	CRB1	AR
Pigmented paravenous chorioretinal atrophy		AD
Retinitis pigmentosa-12		AR
Osteogenesis imperfecta, type VII	CRTAP	AR
Cataract 9, multiple types	CRYAA	AD/AR
Myopathy, myofibrillar, 2A, adult-onset	CRYAB	AD
Cataract 16, multiple types		AD/AR
Myopathy, myofibrillar, 2B, infantile-onset		AR
Cardiomyopathy, dilated, 1II	CRYBA1	AD
Cataract 10, multiple types		AD
Cataract 17, multiple types	CRYBB1	AD/AR
Cataract 3, multiple types	CRYBB2	AD
Cataract 2, multiple types	CRYGC	AD
Cataract 4, multiple types	CRYGD	AD
Deafness, autosomal dominant 40	CRYM	AD
Surfactant metabolism dysfunction, pulmonary, 4	CSF2RA	PR
Neutropenia, severe congenital, 7, autosomal recessive	CSF3R	AR
Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation	CTLA4	AD
Immunodeficiency 24	CTPS1	AR
Haim-Munk syndrome	CTSC	AR
Papillon-Lefevre syndrome		AR
Periodontitis 1, juvenile		AR
Imerslund-Grasbeck syndrome 1	CUBN	AR
WHIM syndrome 1	CXCR4	AD
Myelokathexis, isolated		AD
Chronic granulomatous disease 4, autosomal recessive	CYBA	AR

Disorders	Genes	Inheritance
Chronic granulomatous disease, X-linked	CYBB	XLR
Immunodeficiency 34, mycobacteriosis, X-linked		XLR
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	CYP11B1	AR
Aldosteronism, glucocorticoid-remediable		AD
Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2	AR
Hypoaldosteronism, congenital, due to CMO I deficiency		AR
Anterior segment dysgenesis 6, multiple subtypes	CYP1B1	AR
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset		AR
Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	CYP21A2	AR
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency		AR
Cerebrotendinous xanthomatosis	CYP27A1	AR
Vitamin D-dependent rickets, type I	CYP27B1	AR
Maple syrup urine disease, type II	DBT	AR
Deafness, autosomal recessive 66	DCDC2	AR
Nephronophthisis 19		AR
Sclerosing cholangitis, neonatal		AR
Severe combined immunodeficiency, Athabaskan type	DCLRE1C	AR
Omenn syndrome		AR
Myopathy, myofibrillar, 1	DES	AD/AR
Cardiomyopathy, dilated, 1I		AD
Scapuloperoneal syndrome, neurogenic, Kaeser type		AD
Nephrotic syndrome, type 7	DGKE	AR
Deafness, autosomal dominant 64	DIABLO	AD
Seizures, cortical blindness, microcephaly syndrome	DIAPH1	AR
Deafness, autosomal dominant 1, with or without thrombocytopenia		AD
Dyskeratosis congenita, X-linked	DKC1	XLR
Becker muscular dystrophy	DMD	XLR
Duchenne muscular dystrophy		XLR
Cardiomyopathy, dilated, 3B		XL
Myotonic dystrophy 1	DMPK	AD
Ciliary dyskinesia, primary, 13	DNAAF1	AR
Ciliary dyskinesia, primary, 19	DNAAF11	AR
Ciliary dyskinesia, primary, 17	DNAAF19	AR
Ciliary dyskinesia, primary, 10	DNAAF2	AR
Ciliary dyskinesia, primary, 2	DNAAF3	AR
Ciliary dyskinesia, primary, 25	DNAAF4	AR
Ciliary dyskinesia, primary, 18	DNAAF5	AR
Ciliary dyskinesia, primary, 7, with or without situs inversus	DNAH11	AR

Disorders	Genes	Inheritance
Ciliary dyskinesia, primary, 3, with or without situs inversus	DNAH5	AR
Ciliary dyskinesia, primary, 1, with or without situs inversus	DNAI1	AR
Ciliary dyskinesia, primary, 9, with or without situs inversus	DNAI2	AR
Ciliary dyskinesia, primary, 16	DNAL1	AR
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	DNMT3B	AR
Facioscapulohumeral muscular dystrophy 4, digenic		DD
Immunodeficiency 40	DOCK2	AR
Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	DOCK8	AR
Fetal akinesia deformation sequence 3	DOK7	AR
Myasthenic syndrome, congenital, 10		AR
Ciliary dyskinesia, primary, 21	DRC1	AR
Ciliary dyskinesia, primary, 27	DRC2	AR
Arrhythmogenic right ventricular dysplasia 11	DSC2	AD/AR
Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair		AD/AR
Cardiomyopathy, dilated, 1BB	DSG2	AR
Arrhythmogenic right ventricular dysplasia 10		AD
Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis	DSP	AD
Cardiomyopathy, dilated, with woolly hair and keratoderma		AR
Keratosis palmoplantaris striata II		AD
Epidermolysis bullosa, lethal acantholytic		AR
Arrhythmogenic right ventricular dysplasia 8		AD
Dentinogenesis imperfecta, Shields type III	DSPP	AD
Dentinogenesis imperfecta, Shields type II		AD
Deafness, autosomal dominant 39, with dentinogenesis		AD
Dentin dysplasia, type II		AD
Hermansky-Pudlak syndrome 7	DTNBP1	AR
Thyroid dysmorphogenesis 6	DUOX2	AR
Short-rib thoracic dysplasia 3 with or without polydactyly	DYNC2H1	AR/DR
Waardenburg syndrome, type 4B	EDN3	AD/AR
Waardenburg syndrome, type 4A	EDNRB	AD/AR
Charcot-Marie-Tooth disease, type 1D	EGR2	AD
Dejerine-Sottas disease		AD/AR
Hypomyelinating neuropathy, congenital, 1		AD/AR
Leukoencephalopathy with vanishing white matter 1, with or without ovarian failure	EIF2B1	AR
Leukoencephalopathy with vanishing white matter 2, with or without ovarian failure	EIF2B2	AR
Leukoencephalopathy with vanishing white matter 3, with or without ovarian failure	EIF2B3	AR

Disorders	Genes	Inheritance
Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure	<i>EIF2B4</i>	AR
Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	<i>EIF2B5</i>	AR
Neutropenia, severe congenital 1, autosomal dominant	<i>ELANE</i>	AD
Neutropenia, cyclic		AD
Deafness, autosomal dominant 81	<i>ELMOD3</i>	AD
Deafness, autosomal recessive 88		AR
Cutis laxa, autosomal dominant	<i>ELN</i>	AD
Supravalvar aortic stenosis		AD
Telangiectasia, hereditary hemorrhagic, type 1	<i>ENG</i>	AD
Elliptocytosis-1	<i>EPB41</i>	AD/AR
Spherocytosis, type 5	<i>EPB42</i>	.
Vici syndrome	<i>EPG5</i>	AR
Deafness, autosomal recessive 102	<i>EPS8</i>	AR
Deafness, autosomal recessive 36	<i>ESPN</i>	AR
Deafness, neurosensory, without vestibular involvement, autosomal dominant		AR
Deafness, autosomal recessive 35	<i>ESRRB</i>	AR
Glutaric acidemia IIA	<i>ETFA</i>	AR
Glutaric acidemia IIB	<i>ETFB</i>	AR
Glutaric acidemia IIC	<i>ETFDH</i>	AR
Ethylmalonic encephalopathy	<i>ETHE1</i>	AR
Otofaciocervical syndrome	<i>EYA1</i>	AD
Branchiootic syndrome 1		AD
Branchiootorenal syndrome 1, with or without cataracts		AD
Anterior segment anomalies with or without cataract		AD
Thrombophilia 13, X-linked, due to factor VIII defect	<i>F8</i>	.
Hemophilia A		XLR
{Deep venous thrombosis, protection against}	<i>F9</i>	XLR
Thrombophilia 8, X-linked, due to factor IX defect		XLR
Hemophilia B		XLR
Immunodeficiency 90 with encephalopathy, functional hyposplenism, and hepatic dysfunction	<i>FADD</i>	AR
Tyrosinemia, type I	<i>FAH</i>	AR
Fanconi anemia, complementation group A	<i>FANCA</i>	AR
Fanconi anemia, complementation group C	<i>FANCC</i>	AR
Fanconi anemia, complementation group G	<i>FANCG</i>	AR
Autoimmune lymphoproliferative syndrome, type IA	<i>FAS</i>	AD
Autoimmune lymphoproliferative syndrome, type IB	<i>FASLG</i>	AD

Disorders	Genes	Inheritance
Acromicric dysplasia	<i>FBN1</i>	AD
Geleophysic dysplasia 2		AD
Marfan lipodystrophy syndrome		AD
Marfan syndrome		AD
MASS syndrome		AD
Weill-Marchesani syndrome 2, dominant		AD
Ectopia lentis, familial		AD
Stiff skin syndrome		AD
Immunodeficiency due to ficolin 3 deficiency	<i>FCN3</i>	AR
Protoporphyrin, erythropoietic, 1	<i>FECH</i>	AR
Hartsfield syndrome	<i>FGFR1</i>	AD
Jackson-Weiss syndrome		AD
Pfeiffer syndrome		AD
Hypogonadotropic hypogonadism 2 with or without anosmia		AD
Osteoglophonic dysplasia		AD
Encephalocraniocutaneous lipomatosis, somatic mosaic		.
Trigonocephaly 1	<i>FGFR2</i>	AD
Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis		AD
Apert syndrome		AD
Beare-Stevenson cutis gyrata syndrome		AD
Crouzon syndrome		AD
Jackson-Weiss syndrome		AD
LADD syndrome 1		AD
Pfeiffer syndrome		AD
Saethre-Chotzen syndrome		AD
Craniofacial-skeletal-dermatologic dysplasia		AD
Bent bone dysplasia syndrome		AD
CATSHL syndrome	<i>FGFR3</i>	AD/AR
Crouzon syndrome with acanthosis nigricans		AD
LADD syndrome 2		AD
Muenke syndrome		AD
Hypochondroplasia		AD
Achondroplasia		AD
SADDAN		AD
Thanatophoric dysplasia, type II		AD
Thanatophoric dysplasia, type I	<i>FLNC</i>	AD
Myopathy, myofibrillar, 5		AD
Cardiomyopathy, familial hypertrophic, 26		AD
Cardiomyopathy, familial restrictive 5		AD
Myopathy, distal, 4	<i>FOXI1</i>	AD
Enlarged vestibular aqueduct		AR

Disorders	Genes	Inheritance
T-cell immunodeficiency, congenital alopecia, and nail dystrophy	FOXN1	AR
T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant		AD
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	FOXP3	XLR
Nystagmus 1, congenital, X-linked	FRMD7	XL
Nystagmus, infantile periodic alternating, X-linked		XL
Glycogen storage disease Ia	G6PC1	AR
Dursun syndrome	G6PC3	AR
Neutropenia, severe congenital 4, autosomal recessive		AR
Anemia, congenital, nonspherocytic hemolytic, 1, G6PD deficient	G6PD	XL
Glycogen storage disease II	GAA	AR
Krabbe disease	GALC	AR
Galactose epimerase deficiency	GALE	AR
Galactokinase deficiency with cataracts	GALK1	AR
Galactosemia IV	GALM	AR
Galactosemia	GALT	AR
Emberger syndrome	GATA2	AD
Immunodeficiency 21		AD
Tetralogy of Fallot	GATA4	AD
Atrial septal defect 2		AD
Atrioventricular septal defect 4		AD
Ventricular septal defect 1		AD
Tetralogy of Fallot	GATA6	AD
Atrial septal defect 9		AD
Atrioventricular septal defect 5		AD
Pancreatic agenesis and congenital heart defects		AD
Persistent truncus arteriosus	GBA1	.
Gaucher disease, type IIIC		AR
Gaucher disease, type III		AR
Gaucher disease, type II		AR
Gaucher disease, type I		AR
Gaucher disease, perinatal lethal	GCDH	AR
Glutaricaciduria, type I		AR
Hyperphenylalaninemia, BH4-deficient, B	GCH1	AR
Dystonia, DOPA-responsive		AD/AR
Charcot-Marie-Tooth disease, type 4A	GDAP1	AR
Charcot-Marie-Tooth disease, recessive intermediate, A		AR
Charcot-Marie-Tooth disease, axonal, type 2K		AD/AR
Charcot-Marie-Tooth disease, axonal, with vocal cord paresis		AR
Congenital heart defects, multiple types, 6	GDF1	AD
Right atrial isomerism (Ivemark)		AR

Disorders	Genes	Inheritance
Neutropenia, severe congenital 2, autosomal dominant	GFI1	AD
Myasthenia, congenital, 12, with tubular aggregates	GFPT1	AR
Laron dwarfism	GHR	AR
Increased responsiveness to growth hormone		AD
Growth hormone insensitivity, partial		AD
Deafness, autosomal recessive 15	GIPC3	AR
Cataract 14, multiple types	GJA3	AD
Cataract 1, multiple types	GJA8	AD
Charcot-Marie-Tooth neuropathy, X-linked dominant, 1	GJB1	XLD
Bart-Pumphrey syndrome	GJB2	AD
Vohwinkel syndrome		AD
Deafness, autosomal recessive 1A		AR/DD
Deafness, autosomal dominant 3A		AD
Hystrix-like ichthyosis with deafness		AD
Keratitis-ichthyosis-deafness syndrome		AD
Keratoderma, palmoplantar, with deafness		AD
Deafness, autosomal dominant 2B, with or without peripheral neuropathy	GJB3	AD
Erythrokeratoderma variabilis et progressiva 1		AD/AR
Deafness, digenic, GJB2/GJB3	GJB6	AR/DD
Ectodermal dysplasia 2, Clouston type		AD
Deafness, autosomal recessive 1B		AR
Deafness, autosomal dominant 3B		AD
Deafness, digenic GJB2/GJB6	GLA	AR/DD
Fabry disease		XL
Fabry disease, cardiac variant	GLB1	XL
GM1-gangliosidosis, type III		AR
GM1-gangliosidosis, type II		AR
GM1-gangliosidosis, type I		AR
Mucopolysaccharidosis type IVB (Morquio)	GLDC	AR
Glycine encephalopathy1		AR
Simpson-Golabi-Behmel syndrome, type 1	GPC3	XLR
Wilms tumor, somatic		.
Ocular albinism, type I, Nettlehip-Falls type	GPR143	XL
Nystagmus 6, congenital, X-linked		XLR
Deafness, autosomal recessive 25	GRXCR1	AR
Deafness, autosomal dominant 5	GSDME	AD
Leber congenital amaurosis 1	GUCY2D	AR
Cone-rod dystrophy 6		AD/AR
Night blindness, congenital stationary, type 1I	HADH	AR
3-hydroxyacyl-CoA dehydrogenase deficiency		AR
Hyperinsulinemic hypoglycemia, familial, 4		AR

Disorders	Genes	Inheritance
HELLP syndrome, maternal, of pregnancy	HADHA	AR
Fatty liver, acute, of pregnancy		AR
Mitochondrial trifunctional protein deficiency 1		AR
LCHAD deficiency		AR
Mitochondrial trifunctional protein deficiency 2	HADHB	AR
Perrault syndrome 2	HARS2	AR
Neutropenia, severe congenital 3, autosomal recessive	HAX1	AR
Heinz body anemias, alpha-	HBA1	AD
Thalassemias, alpha-		.
Methemoglobinemia, alpha type		AD
Hemoglobin H disease, nondeletional		.
Erythrocytosis, familial, 7	HBA2	AD
Heinz body anemia		AD
Thalassemia, alpha-		.
Erythrocytosis, familial, 7		AD
Hemoglobin H disease, deletional and nondeletional	HBB	.
Heinz body anemia		AD
Thalassemia, beta		.
Delta-beta thalassemia		AD
Methemoglobinemia, beta type		AD
Erythrocytosis, familial, 6		AD
Sickle cell disease		AR
Thalassemia-beta, dominant inclusion-body		AD
Hereditary persistence of fetal hemoglobin		AD
Hemochromatosis, type 1	HFE	AR
Holocarboxylase synthetase deficiency	HLCS	AR
Porphyria, acute intermittent	HMBS	AD
Porphyria, acute intermittent, nonerythroid variant		AD
HMG-CoA lyase deficiency	HMGCL	AR
Deafness, autosomal dominant 68	HOMER2	AD
Hawkinsinuria	HPD	AD
Tyrosinemia, type III		AR
Hermansky-Pudlak syndrome 1	HPS1	AR
Hermansky-Pudlak syndrome 3	HPS3	AR
Hermansky-Pudlak syndrome 4	HPS4	AR
Hermansky-Pudlak syndrome 5	HPS5	AR
Hermansky-Pudlak syndrome 6	HPS6	AR
HSD10 mitochondrial disease	HSD17B10	XLD
D-bifunctional protein deficiency	HSD17B4	AR
Perrault syndrome 1		AR
Cataract 5, multiple types	HSF4	AD

Disorders	Genes	Inheritance
Charcot-Marie-Tooth disease, axonal, type 2F	HSPB1	AD
Neuronopathy, distal hereditary motor, autosomal dominant 3		AD
Neuronopathy, distal hereditary motor, autosomal dominant 2	HSPB8	AD
Charcot-Marie-Tooth disease, axonal, type 2L		AD
Ciliary dyskinesia, primary, 5	HYDIN	AR
Immunodeficiency, common variable, 1	ICOS	AR
Mucopolysaccharidosis II	IDS	XLR
Aicardi-Goutieres syndrome 7	IFIH1	AD
Singleton-Merten syndrome 1		AD
Immunodeficiency 95		AR
Immunodeficiency 27A, mycobacteriosis, AR	IFNGR1	AR
Immunodeficiency 27B, mycobacteriosis, AD		AD
Immunodeficiency 28, mycobacteriosis	IFNGR2	AR
Short-rib thoracic dysplasia 9 with or without polydactyly	IFT140	AR
Retinitis pigmentosa 80		AR
Agammaglobulinemia 1	IGHM	AR
Kappa light chain deficiency	IGKC	AR
Agammaglobulinemia 2	IGLL1	AR
Immunodeficiency 15A	IKBKB	AD
Immunodeficiency 15B		AR
Immunodeficiency 33	IKBKG	XLR
Autoinflammatory disease, systemic, X-linked		XL
Incontinentia pigmenti		XLD
Ectodermal dysplasia and immunodeficiency 1		XLR
Inflammatory bowel disease 28, early onset, autosomal recessive	IL10RA	AR
Inflammatory bowel disease 25, early onset, autosomal recessive	IL10RB	AR
Immunodeficiency 29, mycobacteriosis	IL12B	AR
Immunodeficiency 30	IL12RB1	AR
Immunodeficiency 51	IL17RA	AR
Candidiasis, familial, 9	IL17RC	AR
Interleukin 1 receptor antagonist deficiency	IL1RN	AR
Immunodeficiency 56	IL21R	AR
Immunodeficiency 41 with lymphoproliferation and autoimmunity	IL2RA	AR
Severe combined immunodeficiency, X-linked	IL2RG	XLR
Combined immunodeficiency, X-linked, moderate		XLR
Psoriasis 14, pustular	IL36RN	AR
Immunodeficiency 104, severe combined	IL7R	AR
Deafness, autosomal recessive 42	ILDR1	AR
Glomerulosclerosis, focal segmental, 5	INF2	.
Charcot-Marie-Tooth disease, dominant intermediate E		AD

Disorders	Genes	Inheritance
Nephronophthisis 2, infantile	<i>INVS</i>	AR
Immunodeficiency 67	<i>IRAK4</i>	AR
Immunodeficiency 32A, mycobacteriosis, autosomal dominant	<i>IRF8</i>	AD
Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive		AR
Immunodeficiency 38	<i>ISG15</i>	AR
Autoimmune disease, multisystem, with facial dysmorphism	<i>ITCH</i>	AR
Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome	<i>ITGA3</i>	AR
Leukocyte adhesion deficiency	<i>ITGB2</i>	AR
Epidermolysis bullosa, junctional 5B, with pyloric atresia	<i>ITGB4</i>	AR
Epidermolysis bullosa, junctional 5A, intermediate		AR
Lymphoproliferative syndrome 1	<i>ITK</i>	AR
Isovaleric acidemia	<i>IVD</i>	AR
Alagille syndrome 1	<i>JAG1</i>	AD
Tetralogy of Fallot		AD
Charcot-Marie-Tooth disease, axonal, type 2HH		AD
Neutropenia, severe congenital, 6, autosomal recessive	<i>JAGN1</i>	AR
Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type	<i>JAK3</i>	AR
Naxos disease	<i>JUP</i>	AR
Nephrotic syndrome, type 16	<i>KANK2</i>	AR
Palmoplantar keratoderma and woolly hair		AR
Charcot-Marie-Tooth disease, recessive intermediate, B	<i>KARS1</i>	AR
Deafness, autosomal recessive 89		AR
Deafness, congenital, and adult-onset progressive leukoencephalopathy		AR
Leukoencephalopathy, progressive, infantile-onset, with or without deafness		AR
Jervell and Lange-Nielsen syndrome 2	<i>KCNE1</i>	AR
Long QT syndrome 5		AD
Short QT syndrome 1	<i>KCNH2</i>	.
Long QT syndrome 2		AD
Bartter syndrome, type 2	<i>KCNJ1</i>	AR
SESAME syndrome	<i>KCNJ10</i>	AR
Enlarged vestibular aqueduct, digenic		AR
Jervell and Lange-Nielsen syndrome	<i>KCNQ1</i>	AR
Short QT syndrome 2		AD
Atrial fibrillation, familial, 3		AD
Long QT syndrome 1		AD
Deafness, autosomal dominant 2A	<i>KCNQ4</i>	AD
Charcot-Marie-Tooth disease, type 2A1	<i>KIF1B</i>	AD

Disorders	Genes	Inheritance
Noonan syndrome 3	<i>KRAS</i>	AD
RAS-associated autoimmune leukoproliferative disorder		AD
Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic		.
Cardiofaciocutaneous syndrome 2		AD
Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	<i>LAMA3</i>	AR
Epidermolysis bullosa, junctional 2B, severe		AR
Epidermolysis bullosa, junctional 2A, intermediate		AR
Pierson syndrome	<i>LAMB2</i>	AR
Nephrotic syndrome, type 5, with or without ocular abnormalities		AR
Epidermolysis bullosa, junctional 1B, severe	<i>LAMB3</i>	AR
Amelogenesis imperfecta, type 1A		AD
Epidermolysis bullosa, junctional 1A, intermediate		AR
Epidermolysis bullosa, junctional 3B, severe	<i>LAMC2</i>	AR
Epidermolysis bullosa, junctional 3A, intermediate		AR
Immunodeficiency due to defect in MAPBP-interacting protein	<i>LAMTOR2</i>	AR
Perrault syndrome 4	<i>LARS2</i>	AR
Hydrops, lactic acidosis, and sideroblastic anemia		AR
Immunodeficiency 52	<i>LAT</i>	AR
Hypercholesterolemia, familial, 1	<i>LDLR</i>	AD/AR
Deafness, autosomal recessive 67	<i>LHFPL5</i>	AR
Pituitary hormone deficiency, combined, 3	<i>LHX3</i>	AR
LIG4 syndrome	<i>LIG4</i>	AR
Wolman disease	<i>LIPA</i>	AR
Cholesteryl ester storage disease		AR
Charcot-Marie-Tooth disease, type 1C	<i>LITAF</i>	AD
Charcot-Marie-Tooth disease, type 2B1	<i>LMNA</i>	AR
Emery-Dreifuss muscular dystrophy 2, autosomal dominant		AD
Emery-Dreifuss muscular dystrophy 3, autosomal recessive		AR
Hutchinson-Gilford progeria		AD
Malouf syndrome		AD
Heart-hand syndrome, Slovenian type		AD
Lipodystrophy, familial partial, type 2		AD
Cardiomyopathy, dilated, 1A		AD
Mandibuloacral dysplasia		AR
Muscular dystrophy, congenital		AD
Restrictive dermopathy 2		AD
Focal segmental glomerulosclerosis 10	<i>LMX1B</i>	AD
Nail-patella syndrome		AD
Deafness, autosomal recessive 77	<i>LOXHD1</i>	AR

Disorders	Genes	Inheritance
Majeed syndrome	<i>LPIN2</i>	.
Immunodeficiency, common variable, 8, with autoimmunity	<i>LRBA</i>	AR
Deafness, autosomal recessive 63	<i>LRTOMT</i>	AR
Glaucoma 3, primary congenital, D	<i>LTBP2</i>	.
Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma		AR
Chediak-Higashi syndrome	<i>LYST</i>	AR
Ayme-Gripp syndrome	<i>MAF</i>	AD
Cataract 21, multiple types		AD
Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia	<i>MAGT1</i>	XLR
Congenital disorder of glycosylation, type Icc		XLR
Immunodeficiency 12	<i>MALT1</i>	AR
Deafness, autosomal recessive 49	<i>MARVELD2</i>	AR
MASP2 deficiency	<i>MASP2</i>	AR
Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency	<i>MAT1A</i>	AD/AR
Methionine adenosyltransferase deficiency, autosomal recessive		AD/AR
Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type	<i>MATN3</i>	AR
Epiphyseal dysplasia, multiple, 5		AD
3-Methylcrotonyl-CoA carboxylase 1 deficiency	<i>MCCC1</i>	AR
3-Methylcrotonyl-CoA carboxylase 2 deficiency	<i>MCCC2</i>	AR
Methylmalonyl-CoA epimerase deficiency	<i>MCEE</i>	AR
Immunodeficiency 54	<i>MCM4</i>	AR
Rett syndrome	<i>MECP2</i>	XLD
Rett syndrome, preserved speech variant		XLD
Rett syndrome, atypical		XLD
Encephalopathy, neonatal severe		XLR
Intellectual developmental disorder, X-linked syndromic 13		XLR
Intellectual developmental disorder, X-linked syndromic, Lubs type		XLR
Hardikar syndrome		XLD
Lujan-Fryns syndrome	<i>MED12</i>	XLR
Ohdo syndrome, X-linked		XLR
Opitz-Kaveggia syndrome		XLR
Neutrophilic dermatosis, acute febrile	<i>MEFV</i>	AD
Familial Mediterranean fever, AD		AD
Familial Mediterranean fever, AR		AR
Deafness, autosomal recessive 97	<i>MET</i>	AR

Disorders	Genes	Inheritance
Hereditary motor and sensory neuropathy VIA	<i>MFN2</i>	AD
Charcot-Marie-Tooth disease, axonal, type 2A2A		AD
Charcot-Marie-Tooth disease, axonal, type 2A2B		AR
Cataract 15, multiple types	<i>MIP</i>	AD
Deafness, autosomal dominant 50	<i>MIR96</i>	AD
COMMAD syndrome	<i>MITF</i>	AR
Tietz albinism-deafness syndrome		AD
Waardenburg syndrome, type 2A		AD
Malonyl-CoA decarboxylase deficiency	<i>MLYCD</i>	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	<i>MMAA</i>	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	<i>MMAB</i>	AR
Methylmalonic aciduria and homocystinuria, cblC type	<i>MMACHC</i>	AR
Methylmalonic aciduria, cblD type	<i>MMADHC</i>	AR
Methylmalonic aciduria and homocystinuria, cblD type		AR
Homocystinuria-megaloblastic anemia, cblD type		AR
Methylmalonic aciduria, mut(0) type	<i>MMUT</i>	AR
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	<i>MPV17</i>	AR
Charcot-Marie-Tooth disease, axonal, type 2EE		AR
Charcot-Marie-Tooth disease, type 1B	<i>MPZ</i>	AD
Charcot-Marie-Tooth disease, type 2I		AD
Charcot-Marie-Tooth disease, type 2J		AD
Dejerine-Sottas disease		AD/AR
Roussy-Levy syndrome		AD
Hypomyelinating neuropathy, congenital, 2		AD
Charcot-Marie-Tooth disease, dominant intermediate D		AD
Ataxia-telangiectasia-like disorder 1	<i>MRE11</i>	AR
Lynch syndrome 5	<i>MSH6</i>	AD
Mismatch repair cancer syndrome 3		AR
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	<i>MTHFD1</i>	AR
Homocystinuria due to MTHFR deficiency	<i>MTHFR</i>	AR
Charcot-Marie-Tooth disease, type 4B1	<i>MTMR2</i>	AR
Homocystinuria-megaloblastic anemia, cblG complementation type	<i>MTR</i>	AR
Homocystinuria-megaloblastic anemia, cbl E type	<i>MTRR</i>	AR
Adenomas, multiple colorectal	<i>MUTYH</i>	AR
Porokeratosis 3, multiple types	<i>MVK</i>	AD
Hyper-IgD syndrome		AR
Mevalonic aciduria		AR

Disorders	Genes	Inheritance
Cardiomyopathy, hypertrophic, 4	MYBPC3	AD/AR
Cardiomyopathy, dilated, 1MM		AD
Left ventricular noncompaction 10		AD
Macroglobulinemia, Waldenstrom, somatic	MYD88	.
Immunodeficiency 68		AR
Deafness, autosomal dominant 4A	MYH14	AD
Laing distal myopathy	MYH7	AD
Cardiomyopathy, hypertrophic, 1		AD/DD
Congenital myopathy 7A, myosin storage, autosomal dominant		AD
Congenital myopathy 7B, myosin storage, autosomal recessive		AR
Cardiomyopathy, dilated, 1S		AD
Left ventricular noncompaction 5		AD
Deafness, autosomal dominant 17	MYH9	AD
Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss		AD
Cardiomyopathy, hypertrophic, 10	MYL2	AD
Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy		AR
Cardiomyopathy, hypertrophic, 8	MYL3	AD/AR
Deafness, autosomal recessive 3	MYO15A	AR
Glomerulosclerosis, focal segmental, 6	MYO1E	AR
Deafness, autosomal recessive 30	MYO3A	AR
Griscelli syndrome, type 1	MYO5A	AR
Deafness, autosomal dominant 22	MYO6	AD
Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy		AD
Deafness, autosomal recessive 37		AR
Usher syndrome, type 1B	MYO7A	AR
Deafness, autosomal dominant 11		AD
Deafness, autosomal recessive 2		AR
Glaucoma 1A, primary open angle	MYOC	AD
2,4-dienoyl-CoA reductase deficiency	NADK2	AR
N-acetylglutamate synthase deficiency	NAGS	AR
Combined oxidative phosphorylation deficiency 24	NARS2	AR
Nijmegen breakage syndrome	NBN	AR
Leukemia, acute lymphoblastic		.
Aplastic anemia		.
Chronic granulomatous disease 1, autosomal recessive	NCF1	AR
Chronic granulomatous disease 2, autosomal recessive	NCF2	AR
Chronic granulomatous disease 3, autosomal recessive	NCF4	AR
Norrie disease	NDP	XLR
Exudative vitreoretinopathy 2, X-linked		XLD/XLR

Disorders	Genes	Inheritance
Charcot-Marie-Tooth disease, axonal, type 2CC	NEFH	AD
Charcot-Marie-Tooth disease, type 1F	NEFL	AD/AR
Charcot-Marie-Tooth disease, type 2E		AD
Charcot-Marie-Tooth disease, dominant intermediate G		AD
Short-rib thoracic dysplasia 6 with or without polydactyly	NEK1	AR/DR
Watson syndrome	NF1	AD
Neurofibromatosis, familial spinal		AD
Neurofibromatosis-Noonan syndrome		AD
Neurofibromatosis, type 1		AD
Leukemia, juvenile myelomonocytic		AD/SMu
Schwannomatosis, vestibular	NF2	AD
Immunodeficiency, common variable, 10	NFKB2	AD
Immunodeficiency 124, severe combined	NHEJ1	AR
Dyskeratosis congenita, autosomal recessive 2	NHP2	AR
Tetralogy of Fallot	NKX2-5	AD
Atrial septal defect 7, with or without AV conduction defects		AD
Conotruncal heart malformations, variable		.
Ventricular septal defect 3		AD
Hypothyroidism, congenital nongoitrous, 5		AD
Hypoplastic left heart syndrome 2	NLRC4	AD
Familial cold autoinflammatory syndrome 4		AD
Autoinflammation with infantile enterocolitis		AD
Familial cold autoinflammatory syndrome 2	NLRP12	AD
CINCA syndrome	NLRP3	AD
Muckle-Wells syndrome		AD
Deafness, autosomal dominant 34, with or without inflammation		AD
Familial cold inflammatory syndrome 1		AD
Keratoendothelitis fugax hereditaria		AD
Ciliary dyskinesia, primary, 6	NME8	AR
Blau syndrome	NOD2	AD
Dyskeratosis congenita, autosomal recessive 1	NOP10	AR
Alagille syndrome 2	NOTCH2	AD
Hajdu-Cheney syndrome		AD
Niemann-Pick disease, type C1	NPC1	AR
Niemann-Pick disease, type D		AR
Niemann-pick disease, type C2	NPC2	AR
Joubert syndrome 4	NPHP1	AR
Senior-Loken syndrome-1		AR
Nephronophthisis 1, juvenile		AR
Meckel syndrome 7	NPHP3	AR
Nephronophthisis 3		AR
Renal-hepatic-pancreatic dysplasia 1		AR

Disorders	Genes	Inheritance
Senior-Loken syndrome 4	NPHP4	AR
Nephronophthisis 4		AR
Nephrotic syndrome, type 1	NPHS1	AR
Nephrotic syndrome, type 2	NPHS2	AR
46XY sex reversal 2, dosage-sensitive	NR0B1	XL
Adrenal hypoplasia, congenital		XLR
46XX sex reversal 4	NR5A1	AD
46XY sex reversal 3		AD
Premature ovarian failure 7		AD
Adrenocortical insufficiency		AD
Noonan syndrome 6	NRAS	AD
Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic		.
Sotos syndrome	NSD1	AD
Albinism, oculocutaneous, type II	OCA2	AR
Albinism, brown oculocutaneous		AR
Dent disease 2	OCRL	XLR
Lowe syndrome		XLR
Ciliary dyskinesia, primary, 20	ODAD1	AR
Ciliary dyskinesia, primary, 23	ODAD2	AR
Retinitis pigmentosa 23	OFD1	XLR
Joubert syndrome 10		XLR
Simpson-Golabi-Behmel syndrome, type 2		XLR
Orofaciodigital syndrome I		XLD
Immunodeficiency 9	ORAI1	AR
Myopathy, tubular aggregate, 2		AD
Deafness, autosomal dominant 67	OSBPL2	AD
Ornithine transcarbamylase deficiency	OTC	XL
Deafness, autosomal recessive 22	OTOA	AR
Deafness, autosomal recessive 9		AR
Auditory neuropathy, autosomal recessive, 1	OTOF	AR
Deafness, autosomal recessive 18B		AR
Deafness, autosomal recessive 84B	OTOGL	AR
Deafness, autosomal dominant 41	P2RX2	AD
Osteogenesis imperfecta, type VIII	P3H1	AR
Phenylketonuria	PAH	AR
Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4	PARN	AD
Dyskeratosis congenita, autosomal recessive 6		AR
Papillorenal syndrome	PAX2	AD
Glomerulosclerosis, focal segmental, 7		AD
Waardenburg syndrome, type 1	PAX3	AD
Waardenburg syndrome, type 3		AD/AR
Craniofacial-deafness-hand syndrome		AD

Disorders	Genes	Inheritance
Cataract with late-onset corneal dystrophy	PAX6	AD
Anterior segment dysgenesis 5, multiple subtypes		AD
Keratitis		AD
Optic nerve hypoplasia		AD
Aniridia		AD
Foveal hypoplasia 1		AD
Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	PAX8	AD
Hyperphenylalaninemia, BH4-deficient, D	PCBD1	AR
Propionicacidemia	PCCA	AR
Propionicacidemia	PCCB	AR
Usher syndrome, type 1F	PCDH15	AR
Deafness, autosomal recessive 23		AR
Usher syndrome, type 1D/F digenic	PCSK9	AR/DR
Hypercholesterolemia, familial, 3		AD
Coenzyme Q10 deficiency, primary, 3	PDSS2	AR
{Retinal disease in Usher syndrome type IIA, modifier of}	PDZD7	AR
Usher syndrome, type IIC, GPR98/PDZD7 digenic		AR/DD
Deafness, autosomal recessive 57		AR
Heimler syndrome 1	PEX1	AR
Peroxisome biogenesis disorder 1A (Zellweger)		AR
Peroxisome biogenesis disorder 1B (NALD/IRD)		AR
Peroxisome biogenesis disorder 6A (Zellweger)	PEX10	AR
Peroxisome biogenesis disorder 6B		AR
Immunodeficiency 23	PGM3	AR
Hypophosphatemic rickets, X-linked dominant	PHEX	XLD
Glycogen storage disease IXc	PHKG2	AR
Multiple congenital anomalies-hypotonia-seizures syndrome 2	PIGA	XLR
Neurodevelopmental disorder with epilepsy and hemochromatosis		XLR
Multiple congenital anomalies-hypotonia-seizures syndrome 3	PIGT	AR
Immunodeficiency 14A, autosomal dominant	PIK3CD	AD
Immunodeficiency 14B, autosomal recessive		AR
Agammaglobulinemia 7, autosomal recessive	PIK3R1	AR
SHORT syndrome		AD
Immunodeficiency 36	PINK1	AD
Parkinson disease 6, early onset		AR
Cataract 11, multiple types		AD/AR
Anterior segment dysgenesis 1, multiple subtypes		AD
Cataract 11, syndromic, autosomal recessive	PITX3	AD/AR

Disorders	Genes	Inheritance
Deafness, autosomal recessive 59	<i>PJVK</i>	AR
Polycystic kidney disease 2	<i>PKD2</i>	AD
Polycystic kidney disease 4, with or without hepatic disease	<i>PKHD1</i>	AR
[Adenosine triphosphate, elevated, of erythrocytes]	<i>PKLR</i>	AD
Anemia, congenital, nonspherocytic hemolytic, 2, pyruvate kinase deficient		AR
Arrhythmogenic right ventricular dysplasia 9	<i>PKP2</i>	AD
Nephrotic syndrome, type 3	<i>PLCE1</i>	AR
Familial cold autoinflammatory syndrome 3	<i>PLCG2</i>	AD
Autoinflammation, antibody deficiency, and immune dysregulation syndrome		AD
Plasminogen deficiency, type I	<i>PLG</i>	AR
Angioedema, hereditary, 4		AD
Dysplasminogenemia		AR
Cardiomyopathy, hypertrophic, 18	<i>PLN</i>	AD
Cardiomyopathy, dilated, 1P		.
Congenital disorder of glycosylation, type Ia	<i>PMM2</i>	AR
Charcot-Marie-Tooth disease, type 1A	<i>PMP22</i>	AD
Charcot-Marie-Tooth disease, type 1E		AD
Dejerine-Sottas disease		AD/AR
Roussy-Levy syndrome		AD
Neuropathy, recurrent, with pressure palsies		AD
Lynch syndrome 4	<i>PMS2</i>	.
Mismatch repair cancer syndrome 4		AR
Immunodeficiency due to purine nucleoside phosphorylase deficiency	<i>PNP</i>	AR
Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration	<i>PNPT1</i>	AR
Spinocerebellar ataxia 25		AD
Combined oxidative phosphorylation deficiency 13		AR
Treacher Collins syndrome 3	<i>POLR1C</i>	AR
Leukodystrophy, hypomyelinating, 11		AR
Treacher Collins syndrome 2	<i>POLR1D</i>	AD/AR
Pituitary hormone deficiency, combined or isolated, 1	<i>POU1F1</i>	AD/AR
Deafness, X-linked 2	<i>POU3F4</i>	XLR
Deafness, autosomal dominant 15/52	<i>POU4F3</i>	AD
Variegate porphyria	<i>PPOX</i>	AD
Lymphoma, non-Hodgkin	<i>PRF1</i>	.
Hemophagocytic lymphohistiocytosis, familial, 2		AR
Aplastic anemia		.
Autoimmune lymphoproliferative syndrome, type III	<i>PRKCD</i>	AR
Immunodeficiency 26, with or without neurologic abnormalities	<i>PRKDC</i>	AR
Parkinson disease, juvenile, type 2	<i>PRKN</i>	AR

Disorders	Genes	Inheritance
Thrombophilia 3 due to protein C deficiency, autosomal dominant	<i>PROC</i>	AD
Thrombophilia 3 due to protein C deficiency, autosomal recessive		AR
Hypogonadotropic hypogonadism 4 with or without anosmia	<i>PROK2</i>	AD
Hypogonadotropic hypogonadism 3 with or without anosmia	<i>PROKR2</i>	AD
Pituitary hormone deficiency, combined, 2	<i>PROP1</i>	AR
Thrombophilia 5 due to protein S deficiency, autosomal dominant	<i>PROS1</i>	AD
Thrombophilia 5 due to protein S deficiency, autosomal recessive		AR
Arts syndrome	<i>PRPS1</i>	XLR
Charcot-Marie-Tooth disease, X-linked recessive, 5		XLR
Gout, PRPS-related		XLR
Deafness, X-linked 1		XL
Phosphoribosylpyrophosphate synthetase superactivity		XLR
Charcot-Marie-Tooth disease, type 4F	<i>PRX</i>	AR
Dejerine-Sottas disease		AD/AR
Metachromatic leukodystrophy due to SAP-b deficiency	<i>PSAP</i>	AR
Gaucher disease, atypical		.
Krabbe disease, atypical		AR
Combined SAP deficiency		AR
Proteasome-associated autoinflammatory syndrome 1 and digenic forms	<i>PSMB8</i>	AR
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	<i>PSTPIP1</i>	AD
LEOPARD syndrome 1	<i>PTPN11</i>	AD
Noonan syndrome 1		AD
Metachondromatosis		AD
Immunodeficiency 105, severe combined	<i>PTPRC</i>	AR
Nephrotic syndrome, type 6	<i>PTPRO</i>	AR
Deafness, autosomal dominant 73	<i>PTPRQ</i>	AD
Deafness, autosomal recessive 84A		AR
Hyperphenylalaninemia, BH4-deficient, A	<i>PTS</i>	AR
Hyperphenylalaninemia, BH4-deficient, C	<i>QDPR</i>	AR
Griscelli syndrome, type 2	<i>RAB27A</i>	AR
Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia	<i>RAC2</i>	AR
Immunodeficiency 73A with defective neutrophil chemotaxis and leukocytosis		AD
Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia		AD
LEOPARD syndrome 2	<i>RAF1</i>	AD
Noonan syndrome 5		AD
Cardiomyopathy, dilated, 1NN		AD

Disorders	Genes	Inheritance
Severe combined immunodeficiency, B cell-negative	RAG1	AR
Omenn syndrome		AR
Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity		.
Combined cellular and humoral immune defects with granulomas		AR
Severe combined immunodeficiency, B cell-negative	RAG2	AR
Omenn syndrome		AR
Combined cellular and humoral immune defects with granulomas		AR
Fetal akinesia deformation sequence 2	RAPSN	AR
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency		AR
Retinoblastoma, trilateral	RB1	AD/SMu
Retinoblastoma		AD/SMu
Polyglucosan body myopathy 1 with or without immunodeficiency	RBCK1	AR
Cardiomyopathy, dilated, 1DD	RBM20	AD
Deafness, autosomal recessive 24	RDX	AR
MHC class II deficiency 5	RFX5	AR
MHC class II deficiency 3		AR
MHC class II deficiency 2	RFXANK	AR
MHC class II deficiency 4	RFXAP	AR
Deafness, autosomal recessive 104	RIPOR2	AR
Deafness, autosomal dominant 21		AD
Anauxetic dysplasia 1	RMRP	AR
Metaphyseal dysplasia without hypotrichosis		AR
Cartilage-hair hypoplasia		AR
Aicardi-Goutieres syndrome 4	RNASEH2A	AR
Aicardi-Goutieres syndrome 2	RNASEH2B	AR
Aicardi-Goutieres syndrome 3	RNASEH2C	AR
RIDDLE syndrome	RNF168	AR
Immunodeficiency 42	RORC	AR
Leber congenital amaurosis 2	RPE65	AR
Retinitis pigmentosa 20		AR
Retinitis pigmentosa 87 with choroidal involvement		AD
Retinitis pigmentosa 3	RPGR	XL
Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness		XL
Cone-rod dystrophy, X-linked, 1		XLR
Macular degeneration, X-linked atrophic		XLR

Disorders	Genes	Inheritance
Diamond-Blackfan anemia 6	RPL5	AD
Diamond-Blackfan anemia 1	RPS19	AD
Asplenia, isolated congenital	RPSA	AD
Ciliary dyskinesia, primary, 24	RSPH1	AR
Ciliary dyskinesia, primary, 11	RSPH4A	AR
Ciliary dyskinesia, primary, 12	RSPH9	AR
Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3	RTEL1	AD
Dyskeratosis congenita, autosomal dominant 4		AD/AR
Dyskeratosis congenita, autosomal recessive 5		AD/AR
Townes-Brocks branchiootorenal-like syndrome	SALL1	AD
Townes-Brocks syndrome 1		AD
Aicardi-Goutieres syndrome 5	SAMHD1	AR
Shwachman-Diamond syndrome 1	SBDS	AR
Charcot-Marie-Tooth disease, type 4B2	SBF2	AR
Epilepsy, progressive myoclonic 4, with or without renal failure	SCARB2	AR
Dravet syndrome	SCN1A	AD
Developmental and epileptic encephalopathy 6B, non-Dravet		AD
Migraine, familial hemiplegic, 3		AD
Febrile seizures, familial, 3A		AD
Generalized epilepsy with febrile seizures plus, type 2		AD
Hypokalemic periodic paralysis, type 2	SCN4A	AD
Congenital myopathy 22A, classic		AR
Hyperkalemic periodic paralysis		AD
Paramyotonia congenita		AD
Myasthenic syndrome, congenital, 16		AR
Congenital myopathy 22B, severe fetal		AR
Myotonia congenita, atypical, acetazolamide-responsive		AD
Brugada syndrome 1	SCN5A	AD
Sick sinus syndrome 1		AR
Heart block, nonprogressive		AD
Atrial fibrillation, familial, 10		AD
Ventricular fibrillation, familial, 1		.
Heart block, progressive, type 1A		AD
Cardiomyopathy, dilated, 1E		AD
Long QT syndrome 3		AD
Liddle syndrome 1	SCNN1B	AD
Pseudohypoaldosteronism, type IB2, autosomal recessive		AR
Bronchiectasis with or without elevated sweat chloride 1		AD

Disorders	Genes	Inheritance
Liddle syndrome 2	SCNN1G	AD
Pseudohypoaldosteronism, type IB3, autosomal recessive		AR
Bronchiectasis with or without elevated sweat chloride 3		AD
Complement component 4, partial deficiency of	SERPING1	AD
Angioedema, hereditary, 1 and 2		AD/AR
Lymphoproliferative syndrome, X-linked, 1	SH2D1A	XLR
Cherubism	SH3BP2	AD
Deafness, autosomal dominant 23	SIX1	AD
Branchiootic syndrome 3		AD
Branchiootorenal syndrome 2	SIX5	.
Trichohepatoenteric syndrome 1	SKIC3	AR
Bartter syndrome, type 1	SLC12A1	AR
Gitelman syndrome	SLC12A3	AR
Allan-Herndon-Dudley syndrome	SLC16A2	XL
Deafness, autosomal dominant 25	SLC17A8	AD
Carnitine deficiency, systemic primary	SLC22A5	AR
Albinism, oculocutaneous, type VI	SLC24A5	AR
Citrullinemia, adult-onset type II	SLC25A13	AR
Citrullinemia, type II, neonatal-onset		AR
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15	AR
Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR
De la Chapelle dysplasia	SLC26A2	AR
Epiphyseal dysplasia, multiple, 4		AR
Atelosteogenesis, type II		AR
Diastrophic dysplasia		AR
Diastrophic dysplasia, broad bone-platyspondylic variant		AR
Achondrogenesis Ib	SLC26A4	AR
Pendred syndrome		AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	SLC26A4	AR
Histiocytosis-lymphadenopathy plus syndrome	SLC29A3	AR
Nephrolithiasis/osteoporosis, hypophosphatemic, 1	SLC34A1	AD
Hypercalcemia, infantile, 2		AR
Pulmonary alveolar microlithiasis	SLC34A2	AR
Congenital disorder of glycosylation, type IIc	SLC35C1	AR
Glycogen storage disease Ib	SLC37A4	AR
Glycogen storage disease Ic		AR
Congenital disorder of glycosylation, type IIw		AD
Cystinuria	SLC3A1	AD/AR
Albinism, oculocutaneous, type IV	SLC45A2	AR
Folate malabsorption, hereditary	SLC46A1	AR

Disorders	Genes	Inheritance
Ovalocytosis, SA type	SLC4A1	AD
Cryohydrocytosis		AD
Spherocytosis, type 4		AD
Distal renal tubular acidosis 1		AD
Distal renal tubular acidosis 4 with hemolytic anemia		AR
Proximal renal tubular acidosis-ocular anomaly syndrome	SLC4A4	AR
Thyroid dysmorphogenesis 1	SLC5A5	AR
Hartnup disorder	SLC6A19	AR
Lysinuric protein intolerance	SLC7A7	AR
Cystinuria	SLC7A9	AD/AR
Myhre syndrome	SMAD4	AD
Polyposis, juvenile intestinal		AD
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome		AD
Schimke immunosseous dysplasia	SMARCA1	AR
Spinal muscular atrophy-1	SMN1	AR
Spinal muscular atrophy-2		AR
Spinal muscular atrophy-3		AR
Spinal muscular atrophy-4		AR
Niemann-Pick disease, type A	SMPD1	AR
Niemann-Pick disease, type B		AR
Myopathy, distal, 7, adult-onset, X-linked	SMPX	XLR
Deafness, X-linked 4		XLD
Noonan syndrome 4	SOS1	AD
PCWH syndrome	SOX10	AD
Waardenburg syndrome, type 2E, with or without neurologic involvement		AD
Waardenburg syndrome, type 4C		AD
Hepatic venoocclusive disease with immunodeficiency	SP110	AR
Ciliary dyskinesia, primary, 28	SPAG1	AR
Netherton syndrome	SPINK5	AR
Spherocytosis, type 3	SPTA1	AR
Pyropoikilocytosis		AR
Elliptocytosis-2		AD
Spherocytosis, type 2	SPTB	AD
Elliptocytosis-3		AD/AR
Anemia, neonatal hemolytic, fatal or near-fatal		AD/AR
Pseudovaginal perineoscrotal hypospadias	SRD5A2	AR
46XX sex reversal 1	SRY	XLD
46XY sex reversal 1		YL

Disorders	Genes	Inheritance
Immunodeficiency 31A, mycobacteriosis, autosomal dominant	STAT1	AD
Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive		AR
Immunodeficiency 31C, chronic mucocutaneous candidiasis, autosomal dominant		AD
Pseudo-TORCH syndrome 3	STAT2	AR
Immunodeficiency 44		AR
Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections	STAT3	AD
Autoimmune disease, multisystem, infantile-onset, 1		AD
Growth hormone insensitivity with immune dysregulation 1, autosomal recessive	STAT5B	AR
Growth hormone insensitivity with immune dysregulation 2, autosomal dominant		AD
Stormorken syndrome	STIM1	AD
Immunodeficiency 10		AR
Myopathy, tubular aggregate, 1		AD
STING-associated vasculopathy, infantile-onset	STING1	AD
T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations	STK4	AR
Deafness, autosomal recessive 16	STRC	AR
Ichthyosis, X-linked	STS	XLR
Hemophagocytic lymphohistiocytosis, familial, 4	STX11	AR
Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease	STXBP2	AR
Barth syndrome	TAFAZZIN	XLR
MHC class I deficiency 1	TAP1	AR
MHC class I deficiency 2	TAP2	AR
MHC class I deficiency 3	TAPBP	AR
Tyrosinemia, type II	TAT	AR
DOORS syndrome	TBC1D24	AR
Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp		AR
Deafness, autosomal dominant 65		AD
Deafness, autosomal recessive 86		AR
Developmental and epileptic encephalopathy 16		AR
Myoclonic epilepsy, infantile, familial		AR
Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	TBK1	AD
DiGeorge syndrome	TBX1	AD
Tetralogy of Fallot		AD
Velocardiofacial syndrome		AD
Conotruncal anomaly face syndrome		.
Spondylocostal dysostosis 5	TBX6	AD/AR

Disorders	Genes	Inheritance
Agammaglobulinemia 8A, autosomal dominant	TCF3	AD
Agammaglobulinemia 8B, autosomal recessive		AR
Transcobalamin II deficiency	TCN2	AR
Treacher Collins syndrome 1	TCOF1	AD
Deafness, autosomal recessive 21	TECTA	AR
Deafness, autosomal dominant 8/12		AD
Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2	TERC	AD
Dyskeratosis congenita, autosomal dominant 1		AD
Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1	TERT	AD
Dyskeratosis congenita, autosomal dominant 2		AD/AR
Dyskeratosis congenita, autosomal recessive 4		AD/AR
Thyroid dysmorphogenesis 3	TG	AR
Thrombophilia 12 due to thrombomodulin defect	THBD	AD
Revesz syndrome	TNF2	AD
Dyskeratosis congenita, autosomal dominant 3		AD
Hypercholanemia, familial 1	TJP2	AR
Cholestasis, progressive familial intrahepatic 4		AR
Deafness, autosomal dominant 36	TMC1	AD
Deafness, autosomal recessive 7		AR
{Epidermodysplasia verruciformis, susceptibility to, 1}	TMC6	AR
{Epidermodysplasia verruciformis, susceptibility to, 2}	TMC8	AR
Emery-Dreifuss muscular dystrophy 7, AD	TMEM43	AD
Auditory neuropathy, autosomal dominant 3		AD
Arrhythmogenic right ventricular dysplasia 5		AD
Deafness, autosomal recessive 6	TMIE	AR
Deafness, autosomal recessive 8/10	TMPRSS3	AR
Immunoglobulin A deficiency 2	TNFRSF13B	.
Immunodeficiency, common variable, 2		AD/AR
Immunodeficiency, common variable, 4	TNFRSF13C	AR
Periodic fever, familial	TNFRSF1A	AD
Immunodeficiency 16	TNFRSF4	AR
Cardiomyopathy, hypertrophic, 13	TNNC1	AD
Cardiomyopathy, dilated, 1Z		AD
Cardiomyopathy, hypertrophic, 7	TNNI3	AD
Cardiomyopathy, familial restrictive, 1		AD
Cardiomyopathy, dilated, 1FF		.
Cardiomyopathy, hypertrophic, 2	TNNT2	AD
Cardiomyopathy, familial restrictive, 3		AD
Cardiomyopathy, dilated, 1D		AD
Left ventricular noncompaction 6		AD

Disorders	Genes	Inheritance
Cardiomyopathy, hypertrophic, 3	TPM1	AD
Cardiomyopathy, dilated, 1Y		AD
Left ventricular noncompaction 9		AD
Thyroid dysmorphogenesis 2A	TPO	AR
Deafness, autosomal recessive 79	TPRN	AR
Immunodeficiency 7, TCR-alpha/beta deficient	TRAC	AR
Cardiac arrhythmia syndrome, with or without skeletal muscle weakness	TRDN	AR
Aicardi-Goutieres syndrome 1, dominant and recessive	TRESX1	AD/AR
Chilblain lupus		AD
Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations		AD
Deafness, autosomal recessive 28	TRIOBP	AR
Retinitis pigmentosa and erythrocytic microcytosis	TRNT1	AR
Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay		AR
Hypomagnesemia 1, intestinal	TRPM6	AR
Tuberous sclerosis-1	TSC1	AD
Lymphangioleiomyomatosis		.
Tuberous sclerosis-2	TSC2	AD
Hypothyroidism, congenital, nongoitrous 4	TSHB	AR
Hyperthyroidism, nonautoimmune	TSHR	AD
Hyperthyroidism, familial gestational		AD
Hypothyroidism, congenital, nongoitrous, 1		AR
Deafness, autosomal recessive 98	TSPEAR	AR
Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type		AR
Tooth agenesis, selective, 10		AR
Short-rib thoracic dysplasia 4 with or without polydactyly	TTC21B	AR
Nephronophthisis 12		AD/AR
Gastrointestinal defects and immunodeficiency syndrome	TTC7A	AR
Tibial muscular dystrophy, tardive	TTN	AD
Myopathy, myofibrillar, 9, with early respiratory failure		AD
Cardiomyopathy, familial hypertrophic, 9		AD
Cardiomyopathy, dilated, 1G		AD
Congenital myopathy 5 with cardiomyopathy		AR
Muscular dystrophy, limb-girdle, autosomal recessive 10		AR
Immunodeficiency 35	TYK2	AR
Albinism, oculocutaneous, type IA	TYR	AR
Albinism, oculocutaneous, type IB		AR
Albinism, oculocutaneous, type III	TYRP1	AR
Spinal muscular atrophy, X-linked 2, infantile	UBA1	XLR

Disorders	Genes	Inheritance
Angelman syndrome	UBE3A	AD
Crigler-Najjar syndrome, type II	UGT1A1	AR
Crigler-Najjar syndrome, type I		AR
Hyperbilirubinemia, familial transient neonatal		AD/AR
Hemophagocytic lymphohistiocytosis, familial, 3	UNC13D	AR
Immunodeficiency with hyper IgM, type 5	UNG	AR
Porphyria cutanea tarda	UROD	AD/AR
Porphyria, hepatoerythropoietic		AD/AR
Porphyria, congenital erythropoietic	UROS	AR
Poikiloderma with neutropenia	USB1	AR
Usher syndrome, type 1C	USH1C	AR
Deafness, autosomal recessive 18A		AR
Usher syndrome, type 1G	USH1G	AR
Usher syndrome, type 2A	USH2A	AR
Retinitis pigmentosa 39		AR
Rickets, vitamin D-resistant, type IIA	VDR	AR
Cohen syndrome	VPS13B	AR
Neutropenia, severe congenital, 5, autosomal recessive	VPS45	AR
Wiskott-Aldrich syndrome	WAS	XLR
Thrombocytopenia, X-linked, intermittent		XLR
Neutropenia, severe congenital, X-linked		XLR
Thrombocytopenia, X-linked		XLR
Glaucoma 1, open angle, G	WDR36	.
Wolfram-like syndrome, autosomal dominant	WFS1	AD
Wolfram syndrome 1		AR
Deafness, autosomal dominant 6/14/38		AD
Usher syndrome, type 2D	WHRN	AR
Deafness, autosomal recessive 31		AR
Wiskott-Aldrich syndrome 2	WIPF1	AR
Denys-Drash syndrome	WT1	AD/SMu
Frasier syndrome		AD/SMu
Meacham syndrome		AD
Wilms tumor, type 1		AD/SMu
Nephrotic syndrome, type 4		AD
Lymphoproliferative syndrome, X-linked, 2	XIAP	XLR
Immunodeficiency 48	ZAP70	AR
Autoimmune disease, multisystem, infantile-onset, 2		AR
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	ZBTB24	AR
46XY sex reversal 9	ZFPM2	AD
Tetralogy of Fallot		AD
Diaphragmatic hernia 3		.

Disorders	Genes	Inheritance
Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24	AR
Restrictive dermopathy 1		AR
Ciliary dyskinesia, primary, 22	ZMYND10	AR

Note: AD: Autosomal dominant; AR: Autosomal recessive; XL: X-linked; XLD: X-linked dominant; XLR: X-linked recessive; YL: Y-linked; DD: Digenic dominant; DR: Digenic recessive; PR: Pseudoautosomal recessive; SMo: Somatic mosaicism; SMu: Somatic mutation

Conditions surrounded by brackets '{ }' indicate mutations that contribute to susceptibility to multifactorial disorders or to infection.

Conditions surrounded by brackets '[']' indicate nondiseases that mainly genetic variations that lead to apparently abnormal laboratory test values.