NBSure™

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		AD/AR
Adrenomyeloneuropathy, adult	*ABCD1*	XLR
Adrenoleukodystrophy		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*
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
Protoporphyria, 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	*ARG1*	AR
Metachromatic leukodystrophy	*ARSA*	AR
Argininosuccinic aciduria	*ASL*	AR
Citrullinemia	*ASS1*	AR
Spastic paraplegia 3A, autosomal dominant	*ATL1*	AD
Neuropathy, hereditary sensory, type ID		AD
Ataxia-telangiectasia	*ATM*	AR
{Deafness, autosomal recessive 12, modifier of}	*ATP2B2*	AR
Deafness, autosomal dominant 82		AD
Distal renal tubular acidosis 3, with or without sensorineural hearing loss	*ATP6V0A4*	AR
Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	*ATP6V1B1*	AR
Menkes disease	*ATP7A*	XLR
Neuronopathy, distal hereditary motor, X-linked		XLR
Occipital horn syndrome		XLR
Wilson disease	*ATP7B*	AR
Cholestasis, progressive familial intrahepatic 1	*ATP8B1*	AR
Cholestasis, benign recurrent intrahepatic		AR
Cholestasis, intrahepatic, of pregnancy, 1		AD
3-methylglutaconic aciduria, type I	*AUH*	AR
Nephrogenic syndrome of inappropriate antidiuresis	*AVPR2*	XLR
Diabetes insipidus, nephrogenic, 1		XLR
Immunodeficiency 43	*B2M*	AR
Myopathy, myofibrillar, 6	*BAG3*	AD
Cardiomyopathy, dilated, 1HH		AD
Maple syrup urine disease, type Ia	*BCKDHA*	AR
Maple syrup urine disease, type Ib	*BCKDHB*	AR
Immunodeficiency 37	*BCL10*	AR
Cataract 12, multiple types	*BFSP2*	AD
Bloom syndrome	*BLM*	AR
Agammaglobulinemia 4	*BLNK*	AR
Hermansky-Pudlak syndrome 8	*BLOC1S3*	AR
Hermansky-Pudlak syndrome 9	*BLOC1S6*	AR
Pulmonary venoocclusive disease 1	*BMPR2*	AD
Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated		AD
Pulmonary hypertension, familial primary, 1, with or without HHT		AD
LEOPARD syndrome 3	*BRAF*	AD
Noonan syndrome 7		AD
Cardiofaciocutaneous syndrome		AD
Bartter syndrome, type 4a	*BSND*	AR
Sensorineural deafness with mild renal dysfunction		AR

Disorders	Genes	Inheritance
Biotinidase deficiency	*BTD*	AR
Isolated growth hormone deficiency, type III, with agammaglobulinemia	*BTK*	XLR
Agammaglobulinemia, X-linked 1		XLR
C1q deficiency 1	*C1QA*	AR
C1q deficiency 2	*C1QB*	AR
C1q deficiency 3	*C1QC*	AR
Ehlers-Danlos syndrome, periodontal type, 1	*C1R*	AD
C1s deficiency	*C1S*	.
Ehlers-Danlos syndrome, periodontal type, 2		AD
C2 deficiency	*C2*	AR
C3 deficiency	*C3*	AR
C4a deficiency	*C4A*	AR
C4B deficiency	*C4B*	.
C5 deficiency	*C5*	AR
C6 deficiency	*C6*	AR
C7 deficiency	*C7*	.
C8 deficiency, type I	*C8A*	AR
C8 deficiency, type II	*C8B*	AR
C9 deficiency	*C9*	.
Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	*CA2*	AR
Deafness, autosomal recessive 93	*CABP2*	AR
Ventricular tachycardia, catecholaminergic polymorphic, 4	*CALM1*	AD
Long QT syndrome 14		AD
Long QT syndrome 15	*CALM2*	AD
Long QT syndrome 16	*CALM3*	AD
B-cell expansion with NFKB and T-cell anergy	*CARD11*	AD
Immunodeficiency 11A		AR
Immunodeficiency 11B with atopic dermatitis		AD
Pityriasis rubra pilaris	*CARD14*	AD
Psoriasis 2		AD
Immunodeficiency 103, susceptibility to fungal infection	*CARD9*	AR
FG syndrome 4	*CASK*	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	*CASP10*	AD
Caspase 8 lymphadenopathy syndrome	*CASP8*	AR
Homocystinuria, B6-responsive and nonresponsive types	*CBS*	AR
Thrombosis, hyperhomocysteinemic		AR
Hennekam lymphangiectasia-lymphedema syndrome 1	*CCBE1*	AR

Disorders	Genes	Inheritance
Ciliary dyskinesia, primary, 14	*CCDC39*	AR
Ciliary dyskinesia, primary, 15	*CCDC40*	AR
Deafness, autosomal dominant 44	*CCDC50*	AD
Immunodeficiency, common variable, 3	*CD19*	AR
Immunodeficiency 25	*CD247*	AR
Lymphoproliferative syndrome 2	*CD27*	AR
Immunodeficiency 19, severe combined	*CD3D*	AR
Immunodeficiency 18	*CD3E*	AR
Immunodeficiency 18, SCID variant		AR
Immunodeficiency 17, CD3 gamma deficient	*CD3G*	AR
Immunodeficiency with hyper-IgM, type 3	*CD40*	AR
Immunodeficiency, X-linked, with hyper-IgM	*CD40LG*	XLR
Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy	*CD55*	AR
Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy	*CD59*	AR
Agammaglobulinemia 3	*CD79A*	AR
Agammaglobulinemia 6	*CD79B*	AR
Immunodeficiency, common variable, 6	*CD81*	AR
Immunodeficiency 116	*CD8A*	AR
Dyserythropoietic anemia, congenital, type Ia	*CDAN1*	AR
Usher syndrome, type 1D	*CDH23*	AR/DR
Deafness, autosomal recessive 12		AR
Usher syndrome, type 1D/F digenic		AR/DR
Deafness, autosomal recessive 113	*CEACAM16*	AR
Deafness, autosomal dominant 4B		AD
Specific granule deficiency	*CEBPE*	AD/AR
Joubert syndrome 5	*CEP290*	AR
Leber congenital amaurosis 10		.
Meckel syndrome 4		AR
Senior-Loken syndrome 6		AR
Complement factor D deficiency	*CFD*	AR
Complement factor H deficiency	*CFH*	AD/AR
Basal laminar drusen		AD
Nephropathy due to CFHR5 deficiency	*CFHR5*	AD
Complement factor I deficiency	*CFI*	AR
Properdin deficiency, X-linked	*CFP*	XLR
{Bronchiectasis with or without elevated sweat chloride 1, modifier of}	*CFTR*	AD
Cystic fibrosis		AR
CHARGE syndrome	*CHD7*	AD
Hypogonadotropic hypogonadism 5 with or without anosmia		AD

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

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
Coproporphyria	*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		AD
Cataract 10, multiple types	*CRYBA1*	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, Athabascan 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 dyshormonogenesis 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	*EIF2B4*	AR
Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	*EIF2B5*	AR
Neutropenia, severe congenital 1, autosomal dominant	*ELANE*	AD
Neutropenia, cyclic		AD
Deafness, autosomal dominant 81	*ELMOD3*	AD
Deafness, autosomal recessive 88		AR
Cutis laxa, autosomal dominant	*ELN*	AD
Supravalvar aortic stenosis		AD
Telangiectasia, hereditary hemorrhagic, type 1	*ENG*	AD
Elliptocytosis-1	*EPB41*	AD/AR
Spherocytosis, type 5	*EPB42*	.
Vici syndrome	*EPG5*	AR
Deafness, autosomal recessive 102	*EPS8*	AR
Deafness, autosomal recessive 36	*ESPN*	AR
Deafness, neurosensory, without vestibular involvement, autosomal dominant		AR
Deafness, autosomal recessive 35	*ESRRB*	AR
Glutaric acidemia IIA	*ETFA*	AR
Glutaric acidemia IIB	*ETFB*	AR
Glutaric acidemia IIC	*ETFDH*	AR
Ethylmalonic encephalopathy	*ETHE1*	AR
Otofaciocervical syndrome	*EYA1*	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	*F8*	.
Hemophilia A		XLR
{Deep venous thrombosis, protection against}	*F9*	XLR
Thrombophilia 8, X-linked, due to factor IX defect		XLR
Hemophilia B		XLR
Immunodeficiency 90 with encephalopathy, functional hyposplenia, and hepatic dysfunction	*FADD*	AR
Tyrosinemia, type I	*FAH*	AR
Fanconi anemia, complementation group A	*FANCA*	AR
Fanconi anemia, complementation group C	*FANCC*	AR
Fanconi anemia, complementation group G	*FANCG*	AR
Autoimmune lymphoproliferative syndrome, type IA	*FAS*	AD
Autoimmune lymphoproliferative syndrome, type IB	*FASLG*	AD

Disorders	Genes	Inheritance
Acromicric dysplasia	*FBN1*	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	*FCN3*	AR
Protoporphyria, erythropoietic, 1	*FECH*	AR
Hartsfield syndrome	*FGFR1*	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		AD
Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	*FGFR2*	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	*FGFR3*	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		AD
Myopathy, myofibrillar, 5	*FLNC*	AD
Cardiomyopathy, familial hypertrophic, 26		AD
Cardiomyopathy, familial restrictive 5		AD
Myopathy, distal, 4		AD
Enlarged vestibular aqueduct	*FOXI1*	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		.
Gaucher disease, type IIIC	*GBA1*	AR
Gaucher disease, type III		AR
Gaucher disease, type II		AR
Gaucher disease, type I		AR
Gaucher disease, perinatal lethal		AR
Glutaricaciduria, type I	*GCDH*	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
Erythrokeratodermia variabilis et progressiva 1		AD/AR
Deafness, digenic, GJB2/GJB3		AR/DD
Ectodermal dysplasia 2, Clouston type	*GJB6*	AD
Deafness, autosomal recessive 1B		AR
Deafness, autosomal dominant 3B		AD
Deafness, digenic GJB2/GJB6		AR/DD
Fabry disease	*GLA*	XL
Fabry disease, cardiac variant		XL
GM1-gangliosidosis, type III	*GLB1*	AR
GM1-gangliosidosis, type II		AR
GM1-gangliosidosis, type I		AR
Mucopolysaccharidosis type IVB (Morquio)		AR
Glycine encephalopathy1	*GLDC*	AR
Simpson-Golabi-Behmel syndrome, type 1	*GPC3*	XLR
Wilms tumor, somatic		.
Ocular albinism, type I, Nettleship-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		AR
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	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		AD
Heinz body anemia	*HBA2*	AD
Thalassemia, alpha-		.
Erythrocytosis, familial, 7		AD
Hemoglobin H disease, deletional and nondeletional		.
Heinz body anemia	*HBB*	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	*INVS*	AR
Immunodeficiency 67	*IRAK4*	AR
Immunodeficiency 32A, mycobacteriosis, autosomal dominant	*IRF8*	AD
Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive		AR
Immunodeficiency 38	*ISG15*	AR
Autoimmune disease, multisystem, with facial dysmorphism	*ITCH*	AR
Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome	*ITGA3*	AR
Leukocyte adhesion deficiency	*ITGB2*	AR
Epidermolysis bullosa, junctional 5B, with pyloric atresia	*ITGB4*	AR
Epidermolysis bullosa, junctional 5A, intermediate		AR
Lymphoproliferative syndrome 1	*ITK*	AR
Isovaleric acidemia	*IVD*	AR
Alagille syndrome 1	*JAG1*	AD
Tetralogy of Fallot		AD
Charcot-Marie-Tooth disease, axonal, type 2HH		AD
Neutropenia, severe congenital, 6, autosomal recessive	*JAGN1*	AR
Severe combined immunodeficiency, autosomal recessive, T-negative/B-positive type	*JAK3*	AR
Naxos disease	*JUP*	AR
Nephrotic syndrome, type 16	*KANK2*	AR
Palmoplantar keratoderma and woolly hair		AR
Charcot-Marie-Tooth disease, recessive intermediate, B	*KARS1*	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	*KCNE1*	AR
Long QT syndrome 5		AD
Short QT syndrome 1	*KCNH2*	.
Long QT syndrome 2		AD
Bartter syndrome, type 2	*KCNJ1*	AR
SESAME syndrome	*KCNJ10*	AR
Enlarged vestibular aqueduct, digenic		AR
Jervell and Lange-Nielsen syndrome	*KCNQ1*	AR
Short QT syndrome 2		AD
Atrial fibrillation, familial, 3		AD
Long QT syndrome 1		AD
Deafness, autosomal dominant 2A	*KCNQ4*	AD
Charcot-Marie-Tooth disease, type 2A1	*KIF1B*	AD

Disorders	Genes	Inheritance
Noonan syndrome 3	*KRAS*	AD
RAS-associated autoimmune leukoproliferative disorder		AD
Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic		.
Cardiofaciocutaneous syndrome 2		AD
Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	*LAMA3*	AR
Epidermolysis bullosa, junctional 2B, severe		AR
Epidermolysis bullosa, junctional 2A, intermediate		AR
Pierson syndrome	*LAMB2*	AR
Nephrotic syndrome, type 5, with or without ocular abnormalities		AR
Epidermolysis bullosa, junctional 1B, severe	*LAMB3*	AR
Amelogenesis imperfecta, type IA		AD
Epidermolysis bullosa, junctional 1A, intermediate		AR
Epidermolysis bullosa, junctional 3B, severe	*LAMC2*	AR
Epidermolysis bullosa, junctional 3A, intermediate		AR
Immunodeficiency due to defect in MAPBP-interacting protein	*LAMTOR2*	AR
Perrault syndrome 4	*LARS2*	AR
Hydrops, lactic acidosis, and sideroblastic anemia		AR
Immunodeficiency 52	*LAT*	AR
Hypercholesterolemia, familial, 1	*LDLR*	AD/AR
Deafness, autosomal recessive 67	*LHFPL5*	AR
Pituitary hormone deficiency, combined, 3	*LHX3*	AR
LIG4 syndrome	*LIG4*	AR
Wolman disease	*LIPA*	AR
Cholesteryl ester storage disease		AR
Charcot-Marie-Tooth disease, type 1C	*LITAF*	AD
Charcot-Marie-Tooth disease, type 2B1	*LMNA*	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	*LMX1B*	AD
Nail-patella syndrome		AD
Deafness, autosomal recessive 77	*LOXHD1*	AR

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

Disorders	Genes	Inheritance
Hereditary motor and sensory neuropathy VIA	*MFN2*	AD
Charcot-Marie-Tooth disease, axonal, type 2A2A		AD
Charcot-Marie-Tooth disease, axonal, type 2A2B		AR
Cataract 15, multiple types	*MIP*	AD
Deafness, autosomal dominant 50	*MIR96*	AD
COMMAD syndrome	*MITF*	AR
Tietz albinism-deafness syndrome		AD
Waardenburg syndrome, type 2A		AD
Malonyl-CoA decarboxylase deficiency	*MLYCD*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblA type	*MMAA*	AR
Methylmalonic aciduria, vitamin B12-responsive, cblB type	*MMAB*	AR
Methylmalonic aciduria and homocystinuria, cblC type	*MMACHC*	AR
Methylmalonic aciduria, cblD type	*MMADHC*	AR
Methylmalonic aciduria and homocystinuria, cblD type		AR
Homocystinuria-megaloblastic anemia, cblD type		AR
Methylmalonic aciduria, mut(0) type	*MMUT*	AR
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	*MPV17*	AR
Charcot-Marie-Tooth disease, axonal, type 2EE		AR
Charcot-Marie-Tooth disease, type 1B	*MPZ*	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	*MRE11*	AR
Lynch syndrome 5	*MSH6*	AD
Mismatch repair cancer syndrome 3		AR
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	*MTHFD1*	AR
Homocystinuria due to MTHFR deficiency	*MTHFR*	AR
Charcot-Marie-Tooth disease, type 4B1	*MTMR2*	AR
Homocystinuria-megaloblastic anemia, cblG complementation type	*MTR*	AR
Homocystinuria-megaloblastic anemia, cbl E type	*MTRR*	AR
Adenomas, multiple colorectal	*MUTYH*	AR
Porokeratosis 3, multiple types	*MVK*	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		AD
Familial cold autoinflammatory syndrome 4	*NLRC4*	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	*OTOF*	AR
Auditory neuropathy, autosomal recessive, 1		AR
Deafness, autosomal recessive 18B	*OTOG*	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		AR/DR
Hypercholesterolemia, familial, 3	*PCSK9*	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		AD
Parkinson disease 6, early onset	*PINK1*	AR
Cataract 11, multiple types	*PITX3*	AD/AR
Anterior segment dysgenesis 1, multiple subtypes		AD
Cataract 11, syndromic, autosomal recessive		AD/AR

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

Disorders	Genes	Inheritance
Thrombophilia 3 due to protein C deficiency, autosomal dominant	*PROC*	AD
Thrombophilia 3 due to protein C deficiency, autosomal recessive		AR
Hypogonadotropic hypogonadism 4 with or without anosmia	*PROK2*	AD
Hypogonadotropic hypogonadism 3 with or without anosmia	*PROKR2*	AD
Pituitary hormone deficiency, combined, 2	*PROP1*	AR
Thrombophilia 5 due to protein S deficiency, autosomal dominant	*PROS1*	AD
Thrombophilia 5 due to protein S deficiency, autosomal recessive		AR
Arts syndrome	*PRPS1*	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	*PRX*	AR
Dejerine-Sottas disease		AD/AR
Metachromatic leukodystrophy due to SAP-b deficiency	*PSAP*	AR
Gaucher disease, atypical		.
Krabbe disease, atypical		AR
Combined SAP deficiency		AR
Proteasome-associated autoinflammatory syndrome 1 and digenic forms	*PSMB8*	AR
Pyogenic sterile arthritis, pyoderma gangrenosum, and acne	*PSTPIP1*	AD
LEOPARD syndrome 1	*PTPN11*	AD
Noonan syndrome 1		AD
Metachondromatosis		AD
Immunodeficiency 105, severe combined	*PTPRC*	AR
Nephrotic syndrome, type 6	*PTPRO*	AR
Deafness, autosomal dominant 73	*PTPRQ*	AD
Deafness, autosomal recessive 84A		AR
Hyperphenylalaninemia, BH4-deficient, A	*PTS*	AR
Hyperphenylalaninemia, BH4-deficient, C	*QDPR*	AR
Griscelli syndrome, type 2	*RAB27A*	AR
Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia	*RAC2*	AR
Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis		AD
Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia		AD
LEOPARD syndrome 2	*RAF1*	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 IA		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	*SERPING 1*	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		AR
Pendred syndrome	*SLC26A4*	AR
Deafness, autosomal recessive 4, with enlarged vestibular aqueduct		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 dyshormonogenesis 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 immunoosseous dysplasia	*SMARCAL 1*	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 dyshormonogenesis 3	*TG*	AR
Thrombophilia 12 due to thrombomodulin defect	*THBD*	AD
Revesz syndrome	*TINF2*	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 dyshormonogenesis 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	*TREX1*	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.

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