YENİDOĞAN GENETİK TARAMA TESTİ GEN LİSTESİ

ACIBADEM LABGEN

Genes	Disease	OMIM 2
AAAS	Glucocorticoid deficiency with achalasia	
AARS1	Charcot-Marie-Tooth disease axonal type 2N	Developmental and epileptic encephalopathy 29
ABCA12	Autosomal recessive congenital ichthyosis 4B	
ABCA3	Interstitial lung disease due to ABCA3 deficiency	
ABCA4	Severe early-childhood-onset retinal dystrophy	
ABCB11	Progressive familial intrahepatic cholestasis type 2	
ABCB4	Progressive familial intrahepatic cholestasis type 3	
ABCC6	Pseudoxanthoma elasticum, forme fruste	
ABCC6	Arterial calcification, generalized, of infancy, 2	
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1	
ABCC8	Diabetes mellitus, transient neonatal, 2	
ABCC8	Leucine-induced hypoglycemia	
ABCC8	Diabetes mellitus, permanent neonatal 3	
ABCC9	Hypertrichotic osteochondrodysplasia Cantu type	Intellectual disability and myopathy syndrome
ABCC9	Dilated cardiomyopathy 1O	
ABCC9	Atrial fibrillation, familial, 12	
ABCD1	Adrenoleukodystrophy	
ABCD4	Methylmalonic acidemia with homocystinuria, type cblJ	
ABCG5	Sitosterolemia 2	
ACAD8	Deficiency of isobutyryl-CoA dehydrogenase	
ACAD9	Acyl-CoA dehydrogenase 9 deficiency	
ACADS	Deficiency of butyryl-CoA dehydrogenase	
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency	
ACAT1	Deficiency of acetyl-CoA acetyltransferase	
ACE	Renal tubular dysgenesis of genetic origin	
ACOX1	Acyl-CoA oxidase deficiency	Mitchell syndrome
ACSF3	Combined malonic and methylmalonic acidemia	

Genes	Disease	OMIM 2
ACTA1	Actin accumulation myopathy	
ACTA2	Aortic aneurysm, familial thoracic 6	
ACTB	Baraitser-winter syndrome 1	
ACTC1	Dilated cardiomyopathy 1R	
ACTC1	Hypertrophic cardiomyopathy 11	
ACTG1	Autosomal dominant nonsyndromic hearing loss 20	
ACTG1	Baraitser-winter syndrome 2	
ACTG2	Megacystis-microcolon-intestinal hypoperistalsis syndrome 5	
ACTN1	Platelet-type bleeding disorder 15	
ACTN2	Dilated cardiomyopathy 1AA	
ACTN4	Focal segmental glomerulosclerosis 1	
ACVR1	Progressive myositis ossificans	
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	
ADA	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-negative, due to adenosine deaminase deficiency	
ADAMTS13	Upshaw-Schulman syndrome	
ADAMTSL2	Geleophysic dysplasia 1	
ADAR	Aicardi-Goutieres syndrome 6	
ADAR	Symmetrical dyschromatosis of extremities	
ADK	Adenosine kinase deficiency	
AGA	Aspartylglucosaminuria	
AGL	Glycogen storage disease type III	
AGRN	Congenital myasthenic syndrome 8	
AGXT	Primary hyperoxaluria type I	
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	
AHI1	Joubert syndrome 3	
AIFM1	Charcot-Marie-Tooth disease X-linked recessive 4	Combined oxidative phosphorylation deficiency 6
AIP	Pituitary dependent hypercortisolism	

Genes	Disease	OMIM 2
AIRE	Polyglandular autoimmune syndrome, type 1	
AK2	Reticular dysgenesis	
AKR1D1	Congenital bile acid synthesis defect 2	
ALAS2	X-linked sideroblastic anemia 1	
ALB	Analbuminemia	
ALDH18A1	Cutis laxa, autosomal dominant 3	
ALDH3A2	Sjogren-Larsson syndrome	
ALDH5A1	Succinate-semialdehyde dehydrogenase deficiency	
ALDH7A1	Pyridoxine-dependent epilepsy	
ALDOB	Hereditary fructosuria	
ALG1	ALG1-CDG	
ALG12	ALG12-congenital disorder of glycosylation	
ALG14	Intellectual developmental disorder with epilepsy, behavioral abnormalities, and coarse facies	
ALG14	Congenital myasthenic syndrome 15	
ALG14	Myopathy, epilepsy, and progressive cerebral atrophy	
ALG3	ALG3-CDG	
ALG6	Congenital disorder of glycosylation type 1C	
ALG8	ALG8 congenital disorder of glycosylation	
ALMS1	Alstrom syndrome	
ALOX12B	Autosomal recessive congenital ichthyosis 2	
ALOXE3	Autosomal recessive congenital ichthyosis 3	
ALPL	Hypophosphatasia, infantile	
ALS2	Amyotrophic lateral sclerosis type 2, juvenile	
ALX4	Parietal foramina 2	Frontonasal dysplasia 2
AMELX	Amelogenesis imperfecta type 1E	
AMN	Imerslund-Grasbeck syndrome type 2	
AMT	Non-ketotic hyperglycinemia	Glycine encephalopathy
ANK1	Hereditary spherocytosis type 1	
ANK2	Cardiac arrhythmia, ankyrin-B-related	Long QT Syndrome 4
ANKH	Craniometaphyseal dysplasia, autosomal dominant	
ANKRD1	ANKRD1-related dilated cardiomyopathy	

Genes	Disease	OMIM 2
ANKRD26	Thrombocytopenia 2	
ANO10	Autosomal recessive spinocerebellar ataxia 10	
ANO5	Autosomal recessive limb-girdle muscular dystrophy type 2L	
ANTXR2	Hyaline fibromatosis syndrome	
AP3B1	Hermansky-Pudlak syndrome 2	
APOB	Hypercholesterolemia, autosomal dominant, type B	
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	
AQP2	Diabetes insipidus, nephrogenic, autosomal	
AR	Partial androgen insensitivity syndrome	
ARFGEF2	Periventricular heterotopia with microcephaly, autosomal recessive	
ARG1	Arginase deficiency	
ARID1B	Coffin-Siris syndrome 1	
ODAD2	Primary ciliary dyskinesia 23	
ARPC1B	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease	
ARSA	Metachromatic leukodystrophy	
ARSB	Mucopolysaccharidosis type 6	
ARX	X-linked lissencephaly with abnormal genitalia	
ASL	Argininosuccinate lyase deficiency	
ASPA	Spongy degeneration of central nervous system	
ASS1	Citrullinemia type I	
ATM	Ataxia-telangiectasia syndrome	
ATP1A2	Migraine, familial hemiplegic, 2	Fetal akinesia, respiratory insufficiency, microcephaly, polymicrogyria, and dysmorphic facies
ATP2A1	Brody myopathy	
ATP6V0A2	Cutis laxa with osteodystrophy	
ATP6V0A4	Renal tubular acidosis, distal, 3, with or without sensorineural hearing loss	
ATP6V1B1	Renal tubular acidosis with progressive nerve deafness	
ATP7A	Menkes kinky-hair syndrome	
ATP7A	Cutis laxa, X-linked	
ATP7B	Wilson disease	

Genes	Disease	OMIM 2
ATP8B1	Progressive familial intrahepatic cholestasis type 1	
ATRX	Alpha thalassemia-X-linked intellectual disability syndrome	
AUH	3-methylglutaconic aciduria type 1	
AVPR2	Diabetes insipidus, nephrogenic, X-linked	
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis	
B3GLCT	Peters plus syndrome	
BAAT	Bile acid conjugation defect 1	
BAG3	Primary dilated cardiomyopathy	
BBS1	Bardet-Biedl syndrome	
BBS10	Bardet-Biedl syndrome 10	
BBS12	Bardet-Biedl syndrome 12	
BBS2	Bardet-Biedl syndrome 2	
BBS4	Bardet-Biedl syndrome 4	
BBS5	Bardet-Biedl syndrome 5	
BBS7	Bardet-Biedl syndrome 7	
BBS9	Bardet-Biedl syndrome 9	
BCKDHA	Maple syrup urine disease	
BCKDHB	Maple syrup urine disease	
BCKDK	Branched-chain keto acid dehydrogenase kinase deficiency	
BCS1L	Mitochondrial complex III deficiency nuclear type 1	
BICD2	Spinal muscular atrophy, lower extremity-predominant, 2b, prenatal onset, autosomal dominant	
BIN1	Myopathy, centronuclear, 2	
BLM	Bloom syndrome	
BLNK	Agammaglobulinemia 4, autosomal recessive	
BMPR1A	Juvenile polyposis syndrome	
BMPR2	Pulmonary hypertension, primary, 1	
BRAF	Cardiofaciocutaneous syndrome 1	
BRCA2	Fanconi anemia complementation group D1	
BRCA2	Medulloblastoma	
BRCA2	Wilms tumor 1	
BRIP1	Fanconi anemia complementation group J	

Genes	Disease	OMIM 2
BSCL2	Congenital generalized lipodystrophy type 2	
BSND	Bartter disease type 4a	
BTD	Biotinidase deficiency	
BTK	X-linked agammaglobulinemia	
BTK	X-linked agammaglobulinemia with growth hormone deficiency	
TWNK	Infantile onset spinocerebellar ataxia	
C2	Complement component 2 deficiency	
C3	Atypical hemolytic-uremic syndrome with C3 anomaly	
C3	Complement component 3 deficiency	
C5	Complement component 5 deficiency	
C6	Complement component 6 deficiency	
C7	Complement component 7 deficiency	
C8A	Type I complement component 8 deficiency	
C8B	Type II complement component 8 deficiency	
C9	Complement component 9 deficiency	
CA2	Osteopetrosis with renal tubular acidosis	
CA5A	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency	
CACNA1A	Episodic ataxia type 2	Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures
CACNA1C	Brugada syndrome 3	
CACNA1C	Long qt syndrome 8	
CACNA1C	Timothy syndrome	
CACNA1D	Aldosterone-producing adenoma with seizures and neurological abnormalities	
CACNA1D	Sinoatrial node dysfunction and deafness	
CACNA1F	Congenital stationary night blindness 2A	Cone-rod dystrophy, X-linked, 3
CAPN3	Autosomal recessive limb-girdle muscular dystrophy type 2A	
CASK	Syndromic X-linked intellectual disability Najm type	
CASQ2	Catecholaminergic polymorphic ventricular tachycardia 2	
CASR	Epilepsy, idiopathic generalized, susceptibility to, 8	

Genes	Disease	OMIM 2
CASR	Familial hypocalciuric hypercalcemia 1	
CASR	Autosomal dominant hypocalcemia 1	
CASR	Neonatal severe primary hyperparathyroidism	
CAV3	Rippling muscle disease 2	
CBL	CBL-related disorder	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
CBLIF	Hereditary intrinsic factor deficiency	
CBS	Classic homocystinuria	
CC2D2A	Joubert syndrome 9	
CCDC39	Primary ciliary dyskinesia 14	
CCDC40	Primary ciliary dyskinesia 15	
CD3D	Immunodeficiency 19	
CD3E	Immunodeficiency 18	
CD40LG	Hyper-IgM syndrome type 1	
CD46	Atypical hemolytic-uremic syndrome with MCP/CD46 anomaly	
CD79A	Agammaglobulinemia 3, autosomal recessive	
CD79B	Agammaglobulinemia 6, autosomal recessive	
CDAN1	Congenital dyserythropoietic anemia, type I	
CDH23	Autosomal recessive nonsyndromic hearing loss 12	
CDH23	Usher syndrome type 1D	
CDKL5	Developmental and epileptic encephalopathy, 2	
CDKN1C	Beckwith-Wiedemann syndrome	
CDKN1C	IMAGe syndrome	
CDSN	Hypotrichosis 2	
CEP152	Seckel syndrome 5	
CEP290	Joubert syndrome 5	
CFB	Atypical hemolytic-uremic syndrome with B factor anomaly	
CFB	Complement factor b deficiency	
CFD	Recurrent Neisseria infections due to factor D deficiency	
CFH	Basal laminar drusen	
CFH	Hemolytic uremic syndrome, atypical, susceptibility to, 1	
CFH	Factor H deficiency	

Genes	Disease	OMIM 2
CFHR1	Hemolytic uremic syndrome, atypical, susceptibility to, 1	
CFI	Atypical hemolytic-uremic syndrome with I factor anomaly	
CFI	Factor I deficiency	
CFL2	Nemaline myopathy 7	
CFP	Properdin deficiency, X-linked	
CFTR	Cystic fibrosis	
CFTR	Hereditary pancreatitis	
CFTR	Bronchiectasis with or without elevated sweat chloride 1	
CFTR	Congenital bilateral aplasia of vas deferens from CFTR mutation	
CHAT	Familial infantile myasthenia	
CHD2	Developmental and epileptic encephalopathy 94	
CHD7	CHARGE association	
CHKB	Megaconial type congenital muscular dystrophy	
CHM	Choroideremia	
CHRNA1	Congenital myasthenic syndrome 1A	
CHRNA1	Myasthenic syndrome, congenital, 1B, fast-channel	
CHRNA1	Lethal multiple pterygium syndrome	
CHRNB1	Congenital myasthenic syndrome 2A	
CHRNB1	Congenital myasthenic syndrome 2C	
CHRND	Congenital myasthenic syndrome 3A	
CHRND	Lethal multiple pterygium syndrome	
CHRND	Congenital myasthenic syndrome 3B	
CHRND	Congenital myasthenic syndrome 3C	
CHRNE	Congenital myasthenic syndrome 4A	
CHRNE	Congenital myasthenic syndrome 4C	
CHRNE	Congenital myasthenic syndrome 4B	
CHRNG	Autosomal recessive multiple pterygium syndrome	
CLCN5	Dent disease type 1	
CLCN7	Autosomal dominant osteopetrosis 2	
CLCN7	Hypopigmentation, organomegaly, and delayed myelination and development	
CLCN7	Autosomal recessive osteopetrosis 4	

Genes	Disease	OMIM 2
CLDN14	Autosomal recessive nonsyndromic hearing loss 29	
CLDN19	Renal hypomagnesemia 5 with ocular involvement	
CLN3	Neuronal ceroid lipofuscinosis 3	
CLN5	Neuronal ceroid lipofuscinosis 5	
CLN6	Ceroid lipofuscinosis, neuronal, 6A	
CLN8	Neuronal ceroid lipofuscinosis 8	
CLRN1	Usher syndrome type 3A	
CNGB3	Achromatopsia 3	
COCH	Autosomal dominant nonsyndromic hearing loss 9	
COL11A1	Stickler syndrome type 2	
COL11A2	Otospondylomegaepiphyseal dysplasia, autosomal dominant	
COL13A1	Congenital myasthenic syndrome 19	
COL17A1	Epidermolysis bullosa, junctional 4, intermediate	
COL1A1	Osteogenesis imperfecta type I	
COL1A2	Osteogenesis imperfecta type III	
COL2A1	Stickler syndrome type 1	
COL3A1	Polymicrogyria with or without vascular-type ehlers-danlos syndrome	
COL4A3	Alport syndrome	
COL4A4	Autosomal recessive Alport syndrome	
COL4A5	Alport syndrome	
COL5A1	Ehlers-Danlos syndrome, classic type, 1	
COL5A2	Ehlers-Danlos syndrome, classic type, 2	
COL6A1	Ullrich congenital muscular dystrophy 1	
COL6A2	Ullrich congenital muscular dystrophy 1	
COL6A3	Ullrich congenital muscular dystrophy 1	
COL7A1	Epidermolysis bullosa dystrophica	
COLQ	Congenital myasthenic syndrome 5	
COQ2	Coenzyme Q10 deficiency, primary, 1	
COQ4	Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome	

Genes	Disease	OMIM 2
COQ6	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	
COQ7	Primary coenzyme Q10 deficiency 8	
COQ8A	Autosomal recessive ataxia due to ubiquinone deficiency	
COQ8B	Nephrotic syndrome, type 9	
COQ9	Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome	
CP	Deficiency of ferroxidase	
CPOX	Hereditary coproporphyria	
CPOX	Harderoporphyria	
CPS1	Pulmonary hypertension, neonatal, susceptibility to	
CPS1	Congenital hyperammonemia, type I	
CPT1A	Carnitine palmitoyl transferase 1A deficiency	
CPT2	Carnitine palmitoyltransferase II deficiency	
CPT2	Carnitine palmitoyl transferase II deficiency, myopathic form	
CPT2	Encephalopathy, acute, infection-induced, susceptibility to, 4	
CPT2	Carnitine palmitoyl transferase II deficiency, neonatal form	
CREBBP	Rubinstein-Taybi syndrome due to CREBBP mutations	
CRLF1	Cold-induced sweating syndrome 1	
CRTAP	Osteogenesis imperfecta type 7	
CRYAB	Fatal infantile hypertonic myofibrillar myopathy	
CRYAB	Dilated cardiomyopathy 1II	
CSF2RA	Surfactant metabolism dysfunction, pulmonary, 4	
CSF3R	Hereditary neutrophilia	
CSF3R	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency	
CSRP3	Hypertrophic cardiomyopathy 12	
CSTB	Unverricht-Lundborg syndrome	
CTC1	Cerebroretinal microangiopathy with calcifications and cysts 1	
CTNS	Nephropathic cystinosis	
CTPS1	Severe combined immunodeficiency due to CTPS1 deficiency	
CTSD	Neuronal ceroid lipofuscinosis 10	

Genes	Disease	OMIM 2
CTSK	Pyknodysostosis	
CUBN	Imerslund-Grasbeck syndrome type 1	
CUL7	3M syndrome 1	
CXCR4	WHIM syndrome 1	
CYBA	Granulomatous disease, chronic, autosomal recessive, cytochrome b-negative	
CYBB	Granulomatous disease, chronic, X-linked	
CYBB	X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency	
CYP11A1	Congenital adrenal insuffiency with 46, XY sex reversal OR 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	
CYP11B1	Glucocorticoid-remediable aldosteronism	
CYP11B1	Deficiency of steroid 11-beta-monooxygenase	
CYP11B2	Corticosterone methyloxidase type 2 deficiency	
CYP17A1	Deficiency of steroid 17-alpha-monooxygenase	
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	
CYP27A1	Cholestanol storage disease	
CYP27B1	Vitamin D-dependent rickets, type 1A	
CYP4F22	Autosomal recessive congenital ichthyosis 5	
D2HGDH	D-2-hydroxyglutaric aciduria 1	
DBT	Maple syrup urine disease	
DCLRE1C	Severe combined immunodeficiency due to DCLRE1C deficiency	
DCLRE1C	Histiocytic medullary reticulosis	
DCX	Lissencephaly type 1 due to doublecortin gene mutation	
DDB2	Xeroderma pigmentosum, group E	
DDC	Deficiency of aromatic-L-amino-acid decarboxylase	
DES	Desmin-related myofibrillar myopathy	Scapuloperoneal syndrome, neurogenic, Kaeser type
DES	Dilated cardiomyopathy 1I	
GSDME	Autosomal dominant nonsyndromic hearing loss 5	
PJVK	Autosomal recessive nonsyndromic hearing loss 59	

Genes	Disease	OMIM 2
DGAT1	Congenital diarrhea 7 with exudative enteropathy	
DGUOK	Mitochondrial DNA depletion syndrome 3	
DHCR7	Smith-Lemli-Opitz syndrome	
DKC1	Dyskeratosis congenita, X-linked	
DLD	Pyruvate dehydrogenase E3 deficiency	
DLL3	Spondylocostal dysostosis 1, autosomal recessive	
DMD	Duchenne muscular dystrophy	
DMD	Becker muscular dystrophy	
DMD	Dilated cardiomyopathy 3B	
DMP1	Hypophosphatemic rickets, autosomal recessive, 1	
DMPK	Steinert myotonic dystrophy syndrome	
DNAAF1	Primary ciliary dyskinesia 13	
DNAH11	Primary ciliary dyskinesia 7	
DNAH5	Primary ciliary dyskinesia 3	
DNAI1	Kartagener syndrome	
DNAJB6	Autosomal dominant limb-girdle muscular dystrophy type 1D (DNAJB6)	
DNM2	Autosomal dominant centronuclear myopathy	Lethal congenital contracture syndrome 5
DNM2	Charcot-Marie-Tooth disease dominant intermediate B	
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	
DOCK8	Combined immunodeficiency due to DOCK8 deficiency	
DOK7	Congenital myasthenic syndrome 10	
DOK7	Fetal akinesia deformation sequence 3	
DPAGT1	Congenital myasthenic syndrome 13	
DSC2	Arrhythmogenic right ventricular cardiomyopathy	
DSG2	Arrhythmogenic right ventricular cardiomyopathy	
DSP	Lethal acantholytic epidermolysis bullosa	

Genes	Disease	OMIM 2
DSP	Arrhythmogenic right ventricular dysplasia 8	Cardiomyopathy, dilated, with woolly hair and keratoderma+Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis+Epidermolysis bullosa, lethal acantholytic+Skin fragility-woolly hair syndrome+
DSP	Keratosis palmoplantaris striata 2	
DSP	Cardiomyopathy, dilated, with woolly hair, keratoderma, and tooth agenesis	
DSP	Skin fragility-woolly hair-palmoplantar keratoderma syndrome	
DUOX2	Thyroid dyshormonogenesis 6	
DYSF	Autosomal recessive limb-girdle muscular dystrophy type 2B	
DYSF	Miyoshi muscular dystrophy 1	
EDA	Hypohidrotic X-linked ectodermal dysplasia	
EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	
EDARADD	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant	
EFHC1	Myoclonic epilepsy, juvenile, susceptibility to, 1	
EFL1	Shwachman-Diamond syndrome 2	
EFTUD2	Mandibulofacial dysostosis-microcephaly syndrome	
EGR2	Charcot-Marie-Tooth disease type 1D	Hypomyelinating neuropathy, congenital, 1
EIF2AK3	Wolcott-Rallison dysplasia	
ELANE	Neutropenia, severe congenital, 1, autosomal dominant	
ELN	Supravalvar aortic stenosis	
EMD	X-linked Emery-Dreifuss muscular dystrophy	
ENG	Telangiectasia, hereditary hemorrhagic, type 1	
ENPP1	Arterial calcification, generalized, of infancy, 1	
ENPP1	Hypopigmentation-punctate palmoplantar keratoderma syndrome	
ENPP1	Hypophosphatemic rickets, autosomal recessive, 2	
EPM2A	Lafora disease	
ERCC2	Xeroderma pigmentosum, group D	
ERCC4	Xeroderma pigmentosum, group F	

Genes	Disease	OMIM 2
ERCC4	XFE progeroid syndrome	
ERCC4	Fanconi anemia complementation group Q	
ERCC5	Xeroderma pigmentosum, group G	
ERCC6	Cockayne syndrome type 2	
ERCC8	Cockayne syndrome type 1	
ESCO2	Roberts-SC phocomelia syndrome	
ESRRB	Autosomal recessive nonsyndromic hearing loss 35	
ETFA	Multiple acyl-CoA dehydrogenase deficiency	
ETFB	Multiple acyl-CoA dehydrogenase deficiency	
ETFDH	Multiple acyl-CoA dehydrogenase deficiency	
ETHE1	Ethylmalonic encephalopathy	
EVC	Ellis-van Creveld syndrome	
EVC2	Ellis-van Creveld syndrome	
EXT1	Exostoses, multiple, type 1	
EXT2	Exostoses, multiple, type 2	
EYA1	Branchiootorenal syndrome 1	
EYA4	Autosomal dominant nonsyndromic hearing loss 10	
EZH2	Weaver syndrome	
F11	Hereditary factor XI deficiency disease	
F13A1	Thrombophilia due to thrombin defect	
F13A1	Factor XIII, A subunit, deficiency of	
F13B	Factor XIII, b subunit, deficiency of	
F2	Thrombophilia due to thrombin defect	
F2	Congenital prothrombin deficiency	
F8	Hereditary factor VIII deficiency disease	
F9	Hereditary factor IX deficiency disease	
F9	Thrombophilia, X-linked, due to factor 9 defect	
FAH	Tyrosinemia type I	
FAM126A	Hypomyelination and Congenital Cataract	
FAM161A	Retinitis pigmentosa 28	
FAM20C	Lethal osteosclerotic bone dysplasia	

Genes	Disease	OMIM 2
CCNQ	Syndactyly-telecanthus-anogenital and renal malformations syndrome	
FANCA	Fanconi anemia complementation group A	
FANCB	Fanconi anemia complementation group B	
FANCC	Fanconi anemia complementation group C	
FANCD2	Fanconi anemia complementation group D2	
FANCE	Fanconi anemia complementation group E	
FANCF	Fanconi anemia complementation group F	
FANCG	Fanconi anemia complementation group G	
FANCI	Fanconi anemia complementation group I	
FANCL	Fanconi anemia complementation group L	
FAS	Autoimmune lymphoproliferative syndrome type 1	
FBLN5	Cutis laxa, autosomal dominant 2	
FBN1	Marfan syndrome	
FBN2	Congenital contractural arachnodactyly	
FBP1	Fructose-biphosphatase deficiency	
FERMT3	Leukocyte adhesion deficiency 3	
FGA	Congenital afibrinogenemia	
FGB	Congenital afibrinogenemia	
FGD1	Aarskog syndrome	
FGD4	Charcot-Marie-Tooth disease type 4H	
FGF3	Deafness with labyrinthine aplasia, microtia, and microdontia	
FGFR2	Pfeiffer syndrome	
FGFR2	Acrocephalosyndactyly type I	
FGFR2	Crouzon syndrome	
FGFR2	Beare-Stevenson cutis gyrata syndrome	
FGFR2	Jackson-Weiss syndrome	
FGFR3	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome	
FGFR3	Hypochondroplasia	
FGFR3	Crouzon syndrome-acanthosis nigricans syndrome	
FGFR3	Thanatophoric dysplasia type 1	

Genes	Disease	OMIM 2
FGFR3	Muenke syndrome	
FGG	Congenital afibrinogenemia	
FH	Fumarase deficiency	
FHL1	X-linked scapuloperoneal muscular dystrophy	
FKRP	Autosomal recessive limb-girdle muscular dystrophy type 2I	
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A1	
FKTN	Autosomal recessive limb-girdle muscular dystrophy type 2M	
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A1	
FLAD1	Myopathy with abnormal lipid metabolism	
FLNA	Oto-palato-digital syndrome, type I	
FOXC1	Axenfeld-Rieger syndrome type 3	
FOXC2	Distichiasis-lymphedema syndrome	
FOXF1	Alveolar capillary dysplasia with pulmonary venous misalignment	
FOXN1	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant	
FOXN1	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	
FOXP3	Insulin-dependent diabetes mellitus secretory diarrhea syndrome	
FRAS1	Fraser syndrome 1	
FTL	Neuroferritinopathy	
FUCA1	Fucosidosis	
FXN	Friedreich ataxia 1	
G6PC1	Glycogen storage disease due to glucose-6-phosphatase deficiency type IA	
G6PC3	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency	
G6PD	Anemia, nonspherocytic hemolytic, due to G6PD deficiency	
GAA	Glycogen storage disease, type II	
GALC	Galactosylceramide beta-galactosidase deficiency	
GALE	UDPglucose-4-epimerase deficiency	
GALK1	Deficiency of galactokinase	

Genes	Disease	OMIM 2
GALNS	Mucopolysaccharidosis, MPS-IV-A	
GALT	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase	
GAMT	Deficiency of guanidinoacetate methyltransferase	
GAN	Giant axonal neuropathy 1	
GATA1	Complete trisomy 21 syndrome	
GATA1	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia	
GATA4	Testicular anomalies with or without congenital heart disease	
GATA4	Tetralogy of Fallot	
GATA4	Atrial septal defect 2	
GATA4	Ventricular septal defect 1	
GATA4	Atrioventricular septal defect 4	
GATA6	Conotruncal heart malformations	
GATA6	Tetralogy of Fallot	
GATA6	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome	
GATA6	Atrioventricular septal defect 5	
GATA6	Atrial septal defect 9	
GBA1	Gaucher disease perinatal lethal	
GBE1	Glycogen storage disease, type IV	
GCDH	Glutaric aciduria, type 1	
GCH1	Dystonia 5	
GCH1	GTP cyclohydrolase I deficiency with hyperphenylalaninemia	Hyperphenylalaninemia, BH4-deficient, B
GCK	Maturity-onset diabetes of the young type 2	
GCK	Hyperinsulinism due to glucokinase deficiency	
GCK	Permanent neonatal diabetes mellitus 1	
GDAP1	Charcot-Marie-Tooth disease axonal type 2K	
GFAP	Alexander disease	
GFM1	Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1	
GFPT1	Congenital myasthenic syndrome 4C	
GFPT1	Congenital myasthenic syndrome 12	
GIPC3	Autosomal recessive nonsyndromic hearing loss 15	

Genes	Disease	OMIM 2
GJA1	Oculodentodigital dysplasia, autosomal recessive	
GJA5	Atrial fibrillation, familial, 11	
GJB1	Charcot-Marie-Tooth disease X-linked dominant 1	
GJB2	Knuckle pads, deafness AND leukonychia syndrome	
GJB2	Palmoplantar keratoderma-deafness syndrome	
GJC2	Hypomyelinating leukodystrophy 2	
GLA	Fabry disease	
GLB1	Infantile GM1 gangliosidosis	
GLDC	Non-ketotic hyperglycinemia	
GLI3	Greig cephalopolysyndactyly syndrome	
GLIS3	Neonatal diabetes mellitus with congenital hypothyroidism	
GLRA1	Hyperekplexia 1	
GLRB	Hyperekplexia 2	
GLUD1	Hyperinsulinism-hyperammonemia syndrome	
GNAS	Pseudohypoparathyroidism type 1B	
GNAS	Pseudopseudohypoparathyroidism	
GNE	GNE myopathy	
GNPTAB	Mucolipidosis type II	
GNPTG	Mucolipidosis type III gamma	
GNS	Mucopolysaccharidosis, MPS-III-D	
GOT2	Developmental and epileptic encephalopathy, 82	
GPC3	Simpson-Golabi-Behmel syndrome type 1	
GPD1L	Brugada syndrome 2	
GPR143	Ocular albinism, type I	
ADGRG1	Bilateral frontoparietal polymicrogyria	
ADGRV1	Usher syndrome type 2C	
GPSM2	Chudley-McCullough syndrome	
GRHPR	Primary hyperoxaluria, type II	
GRIN2A	Landau-Kleffner syndrome	
GSS	Glutathione synthetase deficiency without 5-oxoprolinuria	
GUSB	Mucopolysaccharidosis type 7	

Genes	Disease	OMIM 2
GYS2	Glycogen storage disorder due to hepatic glycogen synthase deficiency	
H19	Beckwith-Wiedemann syndrome	
HADH	Deficiency of 3-hydroxyacyl-CoA dehydrogenase	
HADH	Hyperinsulinemic hypoglycemia, familial, 4	
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	
HADHA	Mitochondrial trifunctional protein deficiency	LCHAD deficiency
HADHB	Mitochondrial trifunctional protein deficiency	
HAX1	Kostmann syndrome	
HBA1	alpha Thalassemia	
HBA1	Hemoglobin H disease	
HBA1	Heinz body anemia	
HBA1	Methemoglobinemia, alpha type	
HBA1	Erythrocytosis, familial, 7	
HBA2	alpha Thalassemia	
HBA2	Hemoglobin H disease	
HBA2	Heinz body anemia	
HBA2	Erythrocytosis, familial, 7	
HBB	Hb SS disease	Thalassemia, beta
HBG2	Cyanosis, transient neonatal	
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX	
HDAC8	Cornelia de Lange syndrome 5	
HESX1	Septo-optic dysplasia sequence	
HEXA	Tay-Sachs disease	
HEXB	Sandhoff disease	
HGD	Alkaptonuria	
HGSNAT	Mucopolysaccharidosis, MPS-III-C	
HINT1	Autosomal recessive axonal neuropathy with neuromyotonia	
HK1	Retinitis pigmentosa 79	
HK1	Neurodevelopmental disorder with visual defects and brain anomalies	
HK1	Hemolytic anemia due to hexokinase deficiency	

Genes	Disease	OMIM 2
HK1	Charcot-Marie-Tooth disease type 4G	
HLCS	Holocarboxylase synthetase deficiency	
HMGCL	Deficiency of hydroxymethylglutaryl-CoA lyase	
HNF1A	Hepatic adenomas, familial	
HNF1A	Diabetes mellitus type 1	
HNF1A	Type 1 diabetes mellitus 20	
HNF1A	Maturity-onset diabetes of the young type 3	
HNF4A	Maturity-onset diabetes of the young type 1	
HNF4A	Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young	
HPD	Hawkinsinuria	
HPD	Tyrosinemia type III	
HPRT1	Lesch-Nyhan syndrome	
HPS1	Hermansky-Pudlak syndrome 1	
HPS3	Hermansky-Pudlak syndrome 3	
HPS4	Hermansky-Pudlak syndrome 4	
HPS5	Hermansky-Pudlak syndrome 5	
HRAS	Costello syndrome	
HSD17B10	HSD10 mitochondrial disease	
HSD17B3	Testosterone 17-beta-dehydrogenase deficiency	
HSD17B4	Bifunctional peroxisomal enzyme deficiency	
HSD3B2	3 beta-Hydroxysteroid dehydrogenase deficiency	
HSD3B7	Congenital bile acid synthesis defect 1	
HSPB8	Charcot-Marie-Tooth disease axonal type 2L	
HSPG2	Schwartz-Jampel syndrome type 1	
HTRA1	CARASIL syndrome	
IDS	Mucopolysaccharidosis, MPS-II	
IDUA	Hurler syndrome	
IDUA	Mucopolysaccharidosis, MPS-I-S	
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome 1	
IGHM	Autosomal recessive agammaglobulinemia 1	
IGHMBP2	Autosomal recessive distal spinal muscular atrophy 1	

Genes	Disease	OMIM 2
IGLL1	Agammaglobulinemia 2, autosomal recessive	
IGSF1	X-linked central congenital hypothyroidism with late-onset testicular enlargement	
ELP1	Familial dysautonomia	
IKBKG	Incontinentia pigmenti syndrome	
IL10RA	Inflammatory bowel disease 28	
IL10RB	Inflammatory bowel disease 25	
IL2RB	Immunodeficiency 63 with lymphoproliferation and autoimmunity	
IL2RG	X-linked severe combined immunodeficiency	
IL2RG	Combined immunodeficiency, X-linked	
IL7R	Immunodeficiency 104	
ILDR1	Autosomal recessive nonsyndromic hearing loss 42	
INS	Type 1 diabetes mellitus 2	
INS	Maturity-onset diabetes of the young type 10	
INS	Hyperproinsulinemia	
INS	Diabetes mellitus, permanent neonatal 4	
INSR	Leprechaunism syndrome	
INVS	Infantile nephronophthisis	
IQCB1	Senior-Loken syndrome 5	
IRAK4	Immunodeficiency 67	
IRF6	Van der Woude syndrome 1	
CRPPA	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type a, 7	
ITGB2	Leukocyte adhesion deficiency 1	
ITGB4	Junctional epidermolysis bullosa with pyloric atresia	
IVD	Isovaleryl-CoA dehydrogenase deficiency	
JAG1	Alagille syndrome due to a JAG1 point mutation	
JAK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency	
JUP	Naxos disease	
JUP	Arrhythmogenic right ventricular dysplasia 12	
KANSL1	Koolen-de Vries syndrome	
KAT6B	Genitopatellar syndrome	

Genes	Disease	OMIM 2
KBTBD13	Nemaline myopathy 6	
KCNA1	Episodic ataxia type 1	
KCNA5	Atrial fibrillation, familial, 7	
KCNE1	Jervell and Lange-Nielsen syndrome 2	
KCNE1	Long QT syndrome 5	
KCNE2	Long QT syndrome 6	
KCNH2	Long QT syndrome 2	
KCNJ1	Bartter disease type 2	
KCNJ11	Hyperinsulinemic hypoglycemia, familial, 2	
KCNJ11	Diabetes mellitus, transient neonatal, 3	
KCNJ11	Maturity-onset diabetes of the young type 13	
KCNJ11	Diabetes mellitus, permanent neonatal 2	
KCNJ2	Andersen Tawil syndrome	
KCNQ1	Long QT syndrome 1	
KCNQ1	Atrial fibrillation, familial, 3	
KCNQ1	Short QT syndrome type 2	
KCNQ1	Jervell and Lange-Nielsen syndrome 1	
KCNQ2	Seizures, benign familial neonatal, 1	
KCNQ2	Developmental and epileptic encephalopathy, 7	
KCNQ4	Autosomal dominant nonsyndromic hearing loss 2A	
KCNT1	Developmental and epileptic encephalopathy, 14	
KCNT1	Autosomal dominant nocturnal frontal lobe epilepsy 5	
KCTD7	Progressive myoclonic epilepsy type 3	
KDM6A	Kabuki syndrome 2	
KDSR	Erythrokeratodermia variabilis et progressiva 4	
KIF21A	Congenital fibrosis of extraocular muscles type 1	
KIT	Partial albinism	
KLHL40	Nemaline myopathy 8	
KLHL41	Nemaline myopathy 9	
KMT2D	Kabuki syndrome 1	
KRAS	Noonan syndrome 3	
KRT14	Epidermolysis bullosa simplex, Koebner type	

Genes	Disease	OMIM 2
KRT16	Pachyonychia congenita 1	
KRT17	Pachyonychia congenita 2	
KRT5	Epidermolysis bullosa simplex with mottled pigmentation	
KRT6A	Pachyonychia congenita 3	
L1CAM	X-linked hydrocephalus syndrome	
LAMA2	Merosin deficient congenital muscular dystrophy	
LAMA3	Junctional epidermolysis bullosa gravis of Herlitz	
LAMB2	Pierson syndrome	
LAMB3	Junctional epidermolysis bullosa gravis of Herlitz	
LAMC2	Junctional epidermolysis bullosa gravis of Herlitz	
LAMP2	Danon disease	
LAMTOR2	Primary immunodeficiency syndrome due to p14 deficiency	
LARGE1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A6	
LBR	Pelger-Huet anomaly	Greenberg skeletal dysplasia
LDLR	Familial hypercholesterolemia	
LEPR	Obesity due to leptin receptor gene deficiency	
LHFPL5	Autosomal recessive nonsyndromic hearing loss 67	
LHX3	Non-acquired combined pituitary hormone deficiency with spine abnormalities	
LHX4	Short stature-pituitary and cerebellar defects-small sella turcica syndrome	
LIFR	Stuve-Wiedemann syndrome 1	
LIG4	DNA ligase IV deficiency	
LIPA	Lysosomal acid lipase deficiency	
LITAF	Charcot-Marie-Tooth disease type 1C	
LMBRD1	Methylmalonic aciduria and homocystinuria type cblF	
LMNA	Emery-Dreifuss muscular dystrophy 3, autosomal recessive	
LMNA	Charcot-Marie-Tooth disease type 2B1	
LMNA	Dilated cardiomyopathy 1A	
LMOD3	Nemaline myopathy 10	
LMX1B	Nail-patella syndrome	

Genes	Disease	OMIM 2
LOXHD1	Autosomal recessive nonsyndromic hearing loss 77	
LRP2	Donnai-Barrow syndrome	
LRP4	Sclerosteosis 2	
LRP4	Cenani-Lenz syndactyly syndrome	
LRP4	Congenital myasthenic syndrome 17	
LRP5	Osteoporosis with pseudoglioma	
LRP5	Autosomal dominant osteopetrosis 1	
LRPPRC	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type	
DNAAF11	Primary ciliary dyskinesia 19	
LRRC8A	Agammaglobulinemia 5, autosomal dominant	
LRSAM1	Charcot-Marie-Tooth disease axonal type 2P	
LRTOMT	Autosomal recessive nonsyndromic hearing loss 63	
LTBP4	Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies	
LYST	Chediak-Higashi syndrome	
MAD2L2	Fanconi anemia complementation group V	
MAFB	Multicentric carpo-tarsal osteolysis with or without nephropathy	
MAN2B1	Deficiency of alpha-mannosidase	
MAP2K1	Cardiofaciocutaneous syndrome 3	
MAP2K2	Cardiofaciocutaneous syndrome 4	
MARVELD2	Autosomal recessive nonsyndromic hearing loss 49	
MAT1A	Hepatic methionine adenosyltransferase deficiency	
MBTPS2	Keratosis follicularis spinulosa decalvans, X-linked	
MC2R	Glucocorticoid deficiency 1	
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency	
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency	
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	
MCFD2	Factor 5 and Factor VIII, combined deficiency of, 2	
MCOLN1	Mucolipidosis type IV	
MCPH1	Microcephaly 1, primary, autosomal recessive	

Genes	Disease	OMIM 2
MECP2	Rett syndrome	
MED12	Blepharophimosis - intellectual disability syndrome, MKB type	
MEFV	Familial Mediterranean fever, autosomal dominant	
MEGF10	MEGF10-Related Myopathy	
MEN1	Multiple endocrine neoplasia, type 1	
MFN2	Charcot-Marie-Tooth disease, axonal, autosomal recessive, type 2a2b	
MFSD8	Neuronal ceroid lipofuscinosis 7	
MGP	Keutel syndrome	
MITF	Waardenburg syndrome type 2A	
MKKS	Bardet-Biedl syndrome 6	
MKS1	Meckel syndrome, type 1	
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	
MLYCD	Deficiency of malonyl-CoA decarboxylase	
MMAA	Methylmalonic aciduria, cblA type	
MMAB	Methylmalonic aciduria, cblB type	
MMACHC	Cobalamin C disease	
MMADHC	Methylmalonic aciduria and homocystinuria type cblD	
MMUT	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency	
MNX1	Currarino triad	
MOCS1	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A	
MOCS2	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type B	
MPI	MPI-CDG	Congenital disorder of glycosylation, type Ib
MPL	Congenital amegakaryocytic thrombocytopenia	
MPV17	Navajo neurohepatopathy	
MPZ	Charcot-Marie-Tooth disease type 2J	
MRAP	Glucocorticoid deficiency 2	
MSX2	Parietal foramina 1	Craniosynostosis 2

Genes	Disease	OMIM 2
MTHFR	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	
MTHFR	Thrombophilia due to thrombin defect	
MTHFR	Neural tube defects, folate-sensitive	
MTM1	Severe X-linked myotubular myopathy	
MTR	Methylcobalamin deficiency type cblG	
MTR	Neural tube defects, folate-sensitive	
MTRR	Methylcobalamin deficiency type cblE	
MTRR	Neural tube defects, folate-sensitive	
MTTP	Abetalipoproteinaemia	
MUSK	Congenital myasthenic syndrome 9	
MUSK	Fetal akinesia deformation sequence 1	
MUTYH	MUTYH-related attenuated familial adenomatous polyposis	
MVK	Hyperimmunoglobulin D with periodic fever	
MVK	Porokeratosis 3, disseminated superficial actinic type	
MVK	Mevalonic aciduria	
MYBPC3	Hypertrophic cardiomyopathy 4	
MYCN	Feingold syndrome type 1	
MYH11	Aortic aneurysm, familial thoracic 4	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2
MYH14	Autosomal dominant nonsyndromic hearing loss 4A	
MYH2	Myopathy, proximal, and ophthalmoplegia	
MYH3	Arthrogryposis, distal, type 2B3	
MYH7	MYH7-related skeletal myopathy	
MYH7	Myopathy, myosin storage, autosomal recessive	
MYH7	Hypertrophic cardiomyopathy 1	
MYH7	Dilated cardiomyopathy 1S	
MYH7	Myosin storage myopathy	
MYH7	MYH7-related late-onset scapuloperoneal muscular dystrophy	
MYH9	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	
MYL2	Hypertrophic cardiomyopathy 10	

Genes	Disease	OMIM 2
MYL3	Hypertrophic cardiomyopathy 8	
MYLK	Aortic aneurysm, familial thoracic 7	
MYO15A	Autosomal recessive nonsyndromic hearing loss 3	
MYO3A	Autosomal recessive nonsyndromic hearing loss 30	
MYO6	Autosomal recessive nonsyndromic hearing loss 37	
MYO7A	Usher syndrome type 1	
MYO9A	Myasthenic syndrome, congenital, 24, presynaptic	
MYSM1	Bone marrow failure syndrome 4	
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 2	
NAGLU	Mucopolysaccharidosis, MPS-III-B	
NAGS	Hyperammonemia, type III	
NBN	Microcephaly, normal intelligence and immunodeficiency	
NBN	Aplastic anemia	
NBN	Acute lymphoid leukemia	
NCF1	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 1	
NCF2	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 2	
NCF4	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type 3	
NDP	Atrophia bulborum hereditaria	
NEB	Nemaline myopathy 2	
NEFL	Charcot-Marie-Tooth disease type 1F	
NEU1	Sialidosis type 2	
NEUROG3	Congenital malabsorptive diarrhea 4	
NF1	Neurofibromatosis, type 1	
NF2	Neurofibromatosis, type 2	
NGLY1	Congenital disorder of deglycosylation 1	
NHEJ1	Cernunnos-XLF deficiency	
NHLRC1	Lafora disease	
NIPAL4	Autosomal recessive congenital ichthyosis 6	
NIPBL	Cornelia de Lange syndrome 1	

Genes	Disease	OMIM 2
NKX2-1	Brain-lung-thyroid syndrome	
NNT	Glucocorticoid deficiency 4	
NOG	Proximal symphalangism 1A	
NOTCH2	Hajdu-Cheney syndrome	
NOTCH3	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 1	
NPC1	Niemann-Pick disease, type C1	
NPC2	Niemann-Pick disease, type C2	
NPHP1	Nephronophthisis 1	
NPHP3	Nephronophthisis 3	
NPHP4	Nephronophthisis 4	
NPHS1	Finnish congenital nephrotic syndrome	
NR0B1	Congenital adrenal hypoplasia, X-linked	
NR3C2	Pseudohyperaldosteronism type 2	
NR3C2	Autosomal dominant pseudohypoaldosteronism type 1	
NR5A1	Premature ovarian failure 7	Adrenocortical insufficiency
NR5A1	46,XY sex reversal 3	
NR5A1	Spermatogenic failure 8	Adrenocortical insufficiency
NR5A1	46,XX sex reversal 4	
NSD1	Sotos syndrome 1	
NSDHL	Child syndrome	
NTRK1	Hereditary insensitivity to pain with anhidrosis	
OBSL1	3M syndrome 2	
OCA2	Tyrosinase-positive oculocutaneous albinism	
OCRL	Lowe syndrome	
OFD1	Orofaciodigital syndrome I	
OPA1	Autosomal dominant optic atrophy classic form	
OPA3	3-Methylglutaconic aciduria type 3	
ORC1	Meier-Gorlin syndrome 1	
OSMR	Amyloidosis, primary localized cutaneous, 1	
OSTM1	Autosomal recessive osteopetrosis 5	
OTC	Ornithine carbamoyltransferase deficiency	

Genes	Disease	OMIM 2
OTOA	Autosomal recessive nonsyndromic hearing loss 22	
OTOF	Autosomal recessive nonsyndromic hearing loss 9	
OTOGL	Autosomal recessive nonsyndromic hearing loss 84B	
OXCT1	Succinyl-CoA acetoacetate transferase deficiency	
PAH	Phenylketonuria	
PAK3	Intellectual disability, X-linked 30	
PANK2	Pigmentary pallidal degeneration	Neurodegeneration with brain iron accumulation 1
PAX3	Waardenburg syndrome type 1	
PAX6	Aniridia 1	
PAX8	Hypothyroidism, congenital, nongoitrous, 2	
PC	Pyruvate carboxylase deficiency	
PCBD1	Pterin-4 alpha-carbinolamine dehydratase 1 deficiency	
PCCA	Propionic acidemia	
PCCB	Propionic acidemia	
PCDH15	Usher syndrome type 1D	
PCNT	Microcephalic osteodysplastic primordial dwarfism type II	
PCSK9	Hypercholesterolemia, autosomal dominant, 3	
PDE4D	Acrodysostosis 2 with or without hormone resistance	
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency	
PDSS1	Deafness-encephaloneuropathy-obesity-valvulopathy syndrome	
PDSS2	Coenzyme Q10 deficiency, primary, 3	
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)	
PEX10	Peroxisome biogenesis disorder 6A (Zellweger)	
PEX12	Peroxisome biogenesis disorder 3A (Zellweger)	
PEX13	Peroxisome biogenesis disorder 11A (Zellweger)	
PEX2	Peroxisome biogenesis disorder 5A (Zellweger)	
PEX26	Peroxisome biogenesis disorder 7A (Zellweger)	
PEX3	Peroxisome biogenesis disorder 10A (Zellweger)	
PEX5	Peroxisome biogenesis disorder 2A (Zellweger)	
PEX6	Peroxisome biogenesis disorder 4A (Zellweger)	
PEX7	Peroxisome biogenesis disorder 9B	

Genes	Disease	OMIM 2
PEX7	Rhizomelic chondrodysplasia punctata type 1	
PFKM	Glycogen storage disease, type VII	
PGM1	Congenital disorder of glycosylation	
PGM3	Immunodeficiency 23	
PHEX	Familial X-linked hypophosphatemic vitamin D refractory rickets	
PHF6	Borjeson-Forssman-Lehmann syndrome	
PHGDH	Neu-Laxova syndrome 1	
PHGDH	PHGDH deficiency	
PHKA2	Glycogen storage disease IXa1	
PHKB	Glycogen storage disease IXb	
PHKG2	Glycogen storage disease IXc	
PHOX2B	Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease	
PHYH	Phytanic acid storage disease	
PIEZO2	Arthrogryposis, distal, with impaired proprioception and touch	
PIK3R1	SHORT syndrome	
PIK3R1	Immunodeficiency 36	
PIK3R1	Agammaglobulinemia 7, autosomal recessive	
PINK1	Autosomal recessive early-onset Parkinson disease 6	
PKD1	Polycystic kidney disease, adult type	
PKD2	Polycystic kidney disease 2	
PKHD1	Polycystic kidney disease 4	
PKLR	Pyruvate kinase deficiency of red cells	
PKLR	Pyruvate kinase hyperactivity	
PKP2	Arrhythmogenic right ventricular dysplasia 9	
PLA2G6	Infantile neuroaxonal dystrophy	
PLCE1	Nephrotic syndrome, type 3	
PLEC	Epidermolysis bullosa simplex with nail dystrophy	
PLEC	Autosomal recessive limb-girdle muscular dystrophy type 2Q	
PLG	Plasminogen deficiency, type I	
PLN	Primary dilated cardiomyopathy	
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type 1	

Genes	Disease	OMIM 2
PLP1	Pelizaeus-Merzbacher disease	
PLP1	Hereditary spastic paraplegia 2	
PLPBP	Epilepsy, early-onset, vitamin B6-dependent	
PMM2	PMM2-CDG	
PMP22	Charcot-Marie-Tooth disease type 1E	
PNKD	Paroxysmal nonkinesigenic dyskinesia 1	
PNKP	Microcephaly, seizures, and developmental delay	
PNPO	Pyridoxal phosphate-responsive seizures	
POLH	Xeroderma pigmentosum variant type	
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A3	
POMGNT1	Autosomal recessive limb-girdle muscular dystrophy type 2O	
POMT1	Autosomal recessive limb-girdle muscular dystrophy type 2K	
POMT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A1	
POMT2	Autosomal recessive limb-girdle muscular dystrophy type 2N	
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	
POR	Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency	
PORCN	Focal dermal hypoplasia	
POU1F1	Pituitary hormone deficiency, combined, 1	
POU3F4	X-linked mixed hearing loss with perilymphatic gusher	
POU4F3	Autosomal dominant nonsyndromic hearing loss 15	
PPT1	Neuronal ceroid lipofuscinosis 1	
PQBP1	Renpenning syndrome	
PRDX1	Cobalamin C disease	
PREPL	Myasthenic syndrome, congenital, 22	
PRF1	Familial hemophagocytic lymphohistiocytosis 2	
PRF1	Aplastic anemia	
PRKAG2	Wolff-Parkinson-White pattern	Glycogen storage disease of heart, lethal congenital
PRKAG2	Hypertrophic cardiomyopathy 6	

Genes	Disease	OMIM 2
PRKAR1A	Carney complex, type 1	
PRKDC	Severe combined immunodeficiency due to DNA-PKcs deficiency	
PROC	Thrombophilia due to protein C deficiency, autosomal dominant	
PROKR2	Hypogonadotropic hypogonadism 3 with or without anosmia	
PROP1	Pituitary hormone deficiency, combined, 2	
PROS1	Thrombophilia due to protein S deficiency, autosomal dominant	
PRRT2	Episodic kinesigenic dyskinesia 1	
PRRT2	Infantile convulsions and choreoathetosis	
PRRT2	Seizures, benign familial infantile, 2	
PRX	Charcot-Marie-Tooth disease type 4F	
PSAP	Sphingolipid activator protein 1 deficiency	
PSAT1	PSAT deficiency	
PSAT1	Neu-Laxova syndrome 2	
PSPH	Deficiency of phosphoserine phosphatase	
PTCH1	Basal cell carcinoma, susceptibility to, 1	Basal cell nevus syndrome
PTEN	Cowden syndrome 1	
PTEN	Cowden syndrome 1	
PTEN	Cowden syndrome 1	
PTEN	Cowden syndrome 1	
PTEN	Cowden syndrome 1	
PTEN	Cowden syndrome 1	
PTF1A	Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome	
PTF1A	Pancreatic agenesis 2	
PTH1R	Metaphyseal chondrodysplasia, Jansen type	
PTPN11	Noonan syndrome 1	
CAVIN1	Congenital generalized lipodystrophy type 4	
PTS	6-Pyruvoyl-tetrahydrobiopterin synthase deficiency	
PYGL	Glycogen storage disease, type VI	
PYGM	Glycogen storage disease, type V	
QDPR	Dihydropteridine reductase deficiency	
RAB23	RAB23-related Carpenter syndrome	

Genes	Disease	OMIM 2
RAB27A	Griscelli syndrome type 2	
RAB3GAP1	Warburg micro syndrome 1	
RAB7A	Charcot-Marie-Tooth disease type 2B	
RAF1	Noonan syndrome 5	
RAG1	Histiocytic medullary reticulosis	
RAG1	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive	
RAG1	Combined immunodeficiency with skin granulomas	
RAG2	Histiocytic medullary reticulosis	
RAG2	Combined immunodeficiency with skin granulomas	
RAG2	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive	
RAI1	Smith-Magenis syndrome	
RAI1	Potocki-Lupski syndrome	
RAPSN	Congenital myasthenic syndrome 11	
RAPSN	Fetal akinesia deformation sequence 2	
RASA1	Capillary malformation-arteriovenous malformation 1	
RB1	Retinoblastoma	
RBM20	Dilated cardiomyopathy 1DD	
RBM8A	Radial aplasia-thrombocytopenia syndrome	
RECQL4	Baller-Gerold syndrome	
RECQL4	Rapadilino syndrome	
RECQL4	Rothmund-Thomson syndrome type 2	
REN	Renal tubular dysgenesis of genetic origin	
RET	Multiple endocrine neoplasia, type 2a	
RFWD3	Fanconi anemia, complementation group W	
RFX6	Hypoplastic pancreas-intestinal atresia-hypoplastic gallbalder syndrome	
RFXANK	MHC class II deficiency	
RMRP	Metaphyseal chondrodysplasia, McKusick type	
RMRP	Metaphyseal dysplasia without hypotrichosis	

Genes	Disease	OMIM 2
RMRP	Anauxetic dysplasia 1	
RNASEH2A	Aicardi-Goutieres syndrome 4	
RNASEH2B	Aicardi-Goutieres syndrome 2	
RNASEH2C	Aicardi-Goutieres syndrome 3	
ROR2	Autosomal recessive Robinow syndrome	
ROR2	Brachydactyly type B1	
RPGR	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness	
RPGRIP1L	Meckel syndrome, type 5	
RPGRIP1L	Joubert syndrome 7	
RPL11	Diamond-Blackfan anemia 7	
RPL15	Diamond-Blackfan anemia 12	
RPL18	Diamond-Blackfan anemia 18	
RPL26	Diamond-Blackfan anemia 11	
RPL27	Diamond-Blackfan anemia 16	
RPL35	Diamond-Blackfan anemia 19	
RPL35A	Diamond-Blackfan anemia 5	
RPL5	Diamond-Blackfan anemia 6	
RPS10	Diamond-Blackfan anemia 9	
RPS15A	Diamond-Blackfan anemia 20	
RPS17	Diamond-Blackfan anemia 4	
RPS19	Diamond-Blackfan anemia 1	
RPS24	Diamond-Blackfan anemia 3	
RPS26	Diamond-Blackfan anemia 10	
RPS27	Diamond-Blackfan anemia 17	
RPS28	Diamond-Blackfan anemia 15 with mandibulofacial dysostosis	
RPS29	Diamond-Blackfan anemia 13	
RPS6KA3	Coffin-Lowry syndrome	
RPS7	Diamond-Blackfan anemia 8	

Genes	Disease	OMIM 2
RRM2B	Mitochondrial DNA depletion syndrome 8a	
RS1	Juvenile retinoschisis	
RSPH4A	Primary ciliary dyskinesia 11	
RSPH9	Primary ciliary dyskinesia 12	
RUNX2	Cleidocranial dysostosis	
RYR1	Central core myopathy	
RYR1	Congenital multicore myopathy with external ophthalmoplegia	
RYR1	Malignant hyperthermia, susceptibility to, 1	
RYR2	Arrhythmogenic right ventricular dysplasia 2	
RYR2	Catecholaminergic polymorphic ventricular tachycardia 1	
SACS	Charlevoix-Saguenay spastic ataxia	
SALL1	Townes-Brocks syndrome 1	
SAMHD1	Aicardi-Goutieres syndrome 5	
SBDS	Shwachman-Diamond syndrome 1	
SBDS	Aplastic anemia	
SCN11A	Familial episodic pain syndrome with predominantly lower limb involvement	
SCN1A	Severe myoclonic epilepsy in infancy	
SCN1A	Generalized epilepsy with febrile seizures plus, type 2	
SCN1A	Developmental and epileptic encephalopathy, 6	
SCN1A	Migraine, familial hemiplegic, 3	
SCN2A	Seizures, benign familial infantile, 3	
SCN2A	Developmental and epileptic encephalopathy, 11	
SCN2A	Episodic ataxia, type 9	
SCN3A	Epilepsy, familial focal, with variable foci 4	
SCN3A	Developmental and epileptic encephalopathy, 62	
SCN4A	Hypokalemic periodic paralysis, type 2	
SCN4A	Paramyotonia congenita of Von Eulenburg	
SCN4A	Hyperkalemic periodic paralysis	
SCN4A	Potassium-aggravated myotonia	
SCN4A	Congenital myasthenic syndrome 16	
SCN5A	Long QT syndrome	

Genes	Disease	OMIM 2
SCN5A	Brugada syndrome 1	
SCN5A	Ventricular fibrillation, paroxysmal familial, type 1	
SCN5A	Progressive familial heart block, type 1A	
SCN5A	Dilated cardiomyopathy 1E	
SCN5A	Long QT syndrome 3	
SCN5A	Atrial fibrillation, familial, 10	
SCN5A	SUDDEN INFANT DEATH SYNDROME	
SCN5A	Sick sinus syndrome 1	
SCN8A	Cognitive impairment with or without cerebellar ataxia	
SCN8A	Developmental and epileptic encephalopathy, 13	
SCN8A	Seizures, benign familial infantile, 5	
SCN8A	Myoclonus, familial, 2	
SCNN1A	Autosomal recessive pseudohypoaldosteronism type 1	
SCNN1A	Bronchiectasis with or without elevated sweat chloride 2	
SCNN1A	Liddle syndrome 3	
SCNN1B	Autosomal recessive pseudohypoaldosteronism type 1	
SCNN1B	Liddle syndrome 1	
SCNN1B	Bronchiectasis with or without elevated sweat chloride 1	
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3	
SCNN1G	Liddle syndrome 2	
SCNN1G	Autosomal recessive pseudohypoaldosteronism type 1	
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	
SDHB	Paragangliomas 4	Mitochondrial complex II deficiency, nuclear type 4
SDHB	Pheochromocytoma	
SDHD	Paragangliomas 1	
SDHD	Pheochromocytoma	
SELENON	Eichsfeld type congenital muscular dystrophy	
SELENON	Congenital myopathy with fiber type disproportion	
SEPTIN9	Amyotrophic neuralgia	
SERPINA1	Alpha-1-antitrypsin deficiency	
SETBP1	Schinzel-Giedion syndrome	

Genes	Disease	OMIM 2
SETX	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2	
SFTPB	Neonatal acute respiratory distress due to SP-B deficiency	
SGCA	Autosomal recessive limb-girdle muscular dystrophy type 2D	
SGCB	Autosomal recessive limb-girdle muscular dystrophy type 2E	
SGCD	Autosomal recessive limb-girdle muscular dystrophy type 2F	
SGCG	Severe autosomal recessive muscular dystrophy of childhood - North African type	
SGPL1	Nephrotic syndrome 14	
SGSH	Mucopolysaccharidosis, MPS-III-A	
SH2D1A	X-linked lymphoproliferative disease due to SH2D1A deficiency	
SH3TC2	Charcot-Marie-Tooth disease type 4C	
SHANK3	Phelan-McDermid syndrome	
SHH	Holoprosencephaly 3	
SI	Sucrase-isomaltase deficiency	
SIL1	Marinesco-SjѴðgren syndrome	
SIX1	Branchiootorenal syndrome 1	
SIX3	Holoprosencephaly 2	
SKI	Shprintzen-Goldberg syndrome	
SLC12A1	Bartter disease type 1	
SLC12A3	Familial hypokalemia-hypomagnesemia	
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	
SLC13A5	Developmental and epileptic encephalopathy, 25	
SLC16A1	Metabolic myopathy due to lactate transporter defect	
SLC16A1	Exercise-induced hyperinsulinism	
SLC16A1	Ketoacidosis due to monocarboxylate transporter-1 deficiency	
SLC16A2	Allan-Herndon-Dudley syndrome	
SLC17A5	Sialic acid storage disease, severe infantile type	
SLC18A2	Parkinsonism-dystonia, infantile, 2	
SLC18A3	Congenital myasthenic syndrome 21	
SLC19A2	Megaloblastic anemia, thiamine-responsive, with diabetes mellitus and sensorineural deafness	

Genes	Disease	OMIM 2
SLC19A3	Biotin-responsive basal ganglia disease	
SLC22A5	Renal carnitine transport defect	
SLC25A1	D,L-2-hydroxyglutaric aciduria	
SLC25A1	Myasthenic syndrome, congenital, 23, presynaptic	
SLC25A13	Citrullinemia type II	
SLC25A13	Citrullinemia, type II, adult-onset	
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	
SLC25A20	Carnitine acylcarnitine translocase deficiency	
SLC25A38	Sideroblastic anemia 2	
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2	
SLC26A2	Achondrogenesis, type IB	
SLC26A3	Congenital secretory diarrhea, chloride type	
SLC26A4	Pendred syndrome	
SLC27A4	Ichthyosis prematurity syndrome	
SLC2A1	Childhood onset GLUT1 deficiency syndrome 2	
SLC2A1	Dystonia 9	
SLC2A1	Hereditary cryohydrocytosis with reduced stomatin	
SLC2A1	Epilepsy, idiopathic generalized, susceptibility to, 12	
SLC2A10	Arterial tortuosity syndrome	
SLC34A2	PULMONARY ALVEOLAR MICROLITHIASIS	
SLC34A3	Autosomal recessive hypophosphatemic bone disease	
SLC35A2	SLC35A2-CDG	
SLC35D1	Schneckenbecken dysplasia	
SLC37A4	Glucose-6-phosphate transport defect	
SLC37A4	Phosphate transport defect	
SLC39A4	Hereditary acrodermatitis enteropathica	
SLC39A8	SLC39A8-CDG	
SLC3A1	Cystinuria	
SLC45A2	Oculocutaneous albinism type 4	
SLC46A1	Congenital defect of folate absorption	
SLC4A1	Hereditary spherocytosis type 4	

Genes	Disease	OMIM 2
SLC4A11	Corneal dystrophy, Fuchs endothelial, 4	
SLC52A2	Brown-Vialetto-van Laere syndrome 2	
SLC52A3	Progressive bulbar palsy of childhood	
SLC52A3	Brown-Vialetto-van Laere syndrome 1	
SLC5A1	Congenital glucose-galactose malabsorption	
SLC5A2	Familial renal glucosuria	
SLC5A5	Familial thyroid dyshormonogenesis 1	
SLC5A7	Neuronopathy, distal hereditary motor, type 7A	
SLC5A7	Congenital myasthenic syndrome 20	
SLC6A5	Hyperekplexia 3	
SLC6A8	Creatine transporter deficiency	
SLC7A7	Lysinuric protein intolerance	
SLC7A9	Cystinuria	
SLC9A6	Christianson syndrome	
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, 2	
SLX4	Fanconi anemia complementation group P	
SMAD3	Aneurysm-osteoarthritis syndrome	
SMAD4	Juvenile polyposis syndrome	
SMAD4	Juvenile polyposis syndrome	
SMARCAL1	Schimke immuno-osseous dysplasia	
SMC1A	Congenital muscular hypertrophy-cerebral syndrome	
SMN1	Spinal muscular atrophy, type IV	
SMN1	Spinal muscular atrophy, type II	
SMPD1	Niemann-Pick disease, type A	
SMPD1	Niemann-Pick disease, type B	
SMPX	Myopathy, distal, 7, adult-onset, X-linked	Deafness, X-linked 4
SNAP25	Congenital myasthenic syndrome 18	
SNTA1	Long QT syndrome 12	
SOX10	Waardenburg syndrome type 2E	
SOX9	Camptomelic dysplasia	
SP110	Hepatic veno-occlusive disease-immunodeficiency syndrome	

Genes	Disease	OMIM 2
SPINK5	Netherton syndrome	
SPR	Dopa-responsive dystonia due to sepiapterin reductase deficiency	
SPRED1	Legius syndrome	
SPTA1	Elliptocytosis 2	
SPTB	Hereditary spherocytosis type 2	
SPTLC1	Hereditary sensory and autonomic neuropathy type 1	
SRCAP	Floating-Harbor syndrome	
SRP54	Neutropenia, severe congenital, 8, autosomal dominant	
STAR	Congenital lipoid adrenal hyperplasia due to STAR deficency	
STAT3	Hyper-IgE recurrent infection syndrome 1	
STAT5B	Growth hormone insensitivity syndrome with immune dysregulation 2, autosomal dominant	
STAT5B	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive	
STK11	Peutz-Jeghers syndrome	
STK11	Peutz-Jeghers syndrome	
STRA6	Matthew-Wood syndrome	
STRC	Autosomal recessive nonsyndromic hearing loss 16	
STS	X-linked ichthyosis with steryl-sulfatase deficiency	
STX11	Familial hemophagocytic lymphohistiocytosis 4	
STXBP1	Developmental and epileptic encephalopathy, 4	
STXBP2	Familial hemophagocytic lymphohistiocytosis 5	
SUCLA2	Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria	
SUCLG1	Mitochondrial DNA depletion syndrome 9	
SUOX	Sulfite oxidase deficiency	
SURF1	Cytochrome-c oxidase deficiency disease	
TAT	Tyrosinemia type II	
TAFAZZIN	3-Methylglutaconic aciduria type 2	
TBC1D24	DOORS syndrome	Developmental and epileptic encephalopathy 16
TBX1	DiGeorge syndrome	
TBX19	Congenital isolated adrenocorticotropic hormone deficiency	

Genes	Disease	OMIM 2
TBX5	Holt-Oram syndrome	
TCAP	Autosomal recessive limb-girdle muscular dystrophy type 2G	
TCF3	Agammaglobulinemia 8, autosomal dominant	
TCIRG1	Autosomal recessive osteopetrosis 1	
TCN2	Transcobalamin II deficiency	
TCOF1	Treacher Collins syndrome 1	
TECTA	Autosomal dominant nonsyndromic hearing loss 12	
TERC	Dyskeratosis congenita, autosomal dominant 1	
TERT	Dyskeratosis congenita, autosomal dominant 1	
TFAP2A	Branchiooculofacial syndrome	
TFAP2B	Char syndrome	
TFG	Hereditary motor and sensory neuropathy, Okinawa type	
TG	Autoimmune thyroid disease, susceptibility to, 3	Thyroid dyshormonogenesis 3
TGFBR1	Loeys-Dietz syndrome 1	
TGFBR2	Loeys-Dietz syndrome 2	
TGM1	Autosomal recessive congenital ichthyosis 1	
TGM5	Acral peeling skin syndrome	
TH	Autosomal recessive DOPA responsive dystonia	
THBD	Thrombomodulin-related bleeding disorder	
THBD	Atypical hemolytic-uremic syndrome with thrombomodulin anomaly	
THRA	Congenital nongoitrous hypothryoidism 6	
THRB	Thyroid hormone resistance, generalized, autosomal recessive	
TIMM8A	Deafness dystonia syndrome	
TINF2	Dyskeratosis congenita, autosomal dominant 3	
TK2	Mitochondrial DNA depletion syndrome, myopathic form	
TMC1	Autosomal dominant nonsyndromic hearing loss 36	
TMC1	Autosomal recessive nonsyndromic hearing loss 7	
TMEM43	Arrhythmogenic right ventricular dysplasia 5	
TMEM67	Joubert syndrome 6	
TMEM67	Meckel syndrome, type 3	
TMIE	Autosomal recessive nonsyndromic hearing loss 6	

Genes	Disease	OMIM 2
TMPRSS3	Autosomal recessive nonsyndromic hearing loss 8	
TNFRSF11A	Familial expansile osteolysis	
TNFRSF11A	Paget disease of bone 2, early-onset	
TNFRSF11A	Autosomal recessive osteopetrosis 7	
TNFRSF11B	Hyperphosphatasemia with bone disease	
TNFSF11	Autosomal recessive osteopetrosis 2	
TNNC1	Dilated cardiomyopathy 1Z	
TNNI2	Distal arthrogryposis type 2B1	
TNNI3	Dilated cardiomyopathy 2A	
TNNI3	Hypertrophic cardiomyopathy 7	
TNNT1	Nemaline myopathy 5	
TNNT2	Dilated cardiomyopathy 1D	
TNNT2	Cardiomyopathy, familial restrictive, 3	
TNNT3	Arthrogryposis, distal, type 2B2	
TP53	Li-Fraumeni syndrome	
TPM1	Hypertrophic cardiomyopathy 3	
TPM2	Nemaline myopathy 4	
TPM2	Arthrogryposis, distal, type 1A	
TPM3	Nemaline myopathy 1	
TPM3	Congenital myopathy with fiber type disproportion	
TPO	Deficiency of iodide peroxidase	
TPP1	Neuronal ceroid lipofuscinosis 2	
TRAPPC2	Spondyloepiphyseal dysplasia tarda, X-linked	
TREX1	Aicardi-Goutieres syndrome 1	
TRIM32	Sarcotubular myopathy	
TRIM37	Mulibrey nanism syndrome	
TRIOBP	Autosomal recessive nonsyndromic hearing loss 28	

Genes	Disease	OMIM 2
TRMU	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	
TRPM4	Progressive familial heart block type IB	
TSC1	Tuberous sclerosis 1	
TSC1	Lymphangiomyomatosis	
TSC1	Isolated focal cortical dysplasia type II	
TSC2	Tuberous sclerosis 2	
TSC2	Lymphangiomyomatosis	
TSC2	Isolated focal cortical dysplasia type II	
TSEN54	Pontocerebellar hypoplasia type 4	
TSHB	Secondary hypothyroidism	
TSHR	Hypothyroidism due to TSH receptor mutations	
TSR2	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis	
SKIC3	Trichohepatoenteric syndrome 1	
TTC7A	Gastrointestinal defects and immunodeficiency syndrome 1	
TTN	Myopathy, myofibrillar, 9, with early respiratory failure	
TTN	Dilated cardiomyopathy 1G	
TTPA	Familial isolated deficiency of vitamin E	
TTR	Amyloidogenic transthyretin amyloidosis	
TWIST1	Saethre-Chotzen syndrome	
TYMP	Mitochondrial DNA depletion syndrome 1	
TYR	Tyrosinase-negative oculocutaneous albinism	
UBE2T	Fanconi anemia complementation group T	
UBR1	Johanson-Blizzard syndrome	
UGT1A1	Crigler-Najjar syndrome type 1	
UMOD	Familial juvenile hyperuricemic nephropathy type 1	
UNC13D	Familial hemophagocytic lymphohistiocytosis 3	
UROD	Familial porphyria cutanea tarda	
UROS	Cutaneous porphyria	
USH1C	Usher syndrome type 1	
USH1G	Usher syndrome type 1G	
USH2A	Usher syndrome type 2A	

Genes	Disease	OMIM 2
VAMP1	Spastic ataxia 1	
VAMP1	Myasthenic syndrome, congenital, 25, presynaptic	
VCAN	Wagner syndrome	
VCL	Primary dilated cardiomyopathy	
VCP	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	
VDR	Vitamin D-dependent rickets type II with alopecia	
VHL	Von Hippel-Lindau syndrome	
VHL	Von Hippel-Lindau syndrome	
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2	
VLDLR	Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1	
VPS13A	Chorea-acanthocytosis	
VPS13B	Cohen syndrome	
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	
VPS45	Congenital neutropenia-myelofibrosis-nephromegaly syndrome	
VWF	von Willebrand disease type 1	
WAS	X-linked severe congenital neutropenia	
WAS	Wiskott-Aldrich syndrome	
WAS	Thrombocytopenia 1	
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	
WFS1	Wolfram-like syndrome	
WNT10A	Odonto-onycho-dermal dysplasia	
WRN	Werner syndrome	
WT1	Drash syndrome	
WT1	Frasier syndrome	
WT1	Wilms tumor 1	
XIAP	X-linked lymphoproliferative disease due to XIAP deficiency	
XPA	Xeroderma pigmentosum group A	
XPC	Xeroderma pigmentosum, group C	
ZAP70	Combined immunodeficiency due to ZAP70 deficiency	

Genes	Disease	OMIM 2
ZEB2	Mowat-Wilson syndrome	
ZIC2	Holoprosencephaly 5	
ZIC3	Heterotaxy, visceral, 1, X-linked	
ZMPSTE24	Lethal tight skin contracture syndrome	
ZNF469	Brittle cornea syndrome 1	