TAŞIYICILIK GENETİK TARAMA TESTİ GEN LİSTESİ

ACIBADEM LABGEN

Genes	Disease
HBB	beta Thalassemia
XPC	Xeroderma pigmentosum, group C
TYR	Tyrosinase-negative oculocutaneous albinism
TYR	Oculocutaneous albinism type 1B
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency
PAH	Phenylketonuria
CFTR	Cystic fibrosis
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency
HEXA	Tay-Sachs disease
GJB2	Autosomal recessive nonsyndromic hearing loss 1A
GJB2	Autosomal dominant nonsyndromic hearing loss 3A
DHCR7	Smith-Lemli-Opitz syndrome
ATP7B	Wilson disease
ASPA	Spongy degeneration of central nervous system
ACADM	Medium-chain acyl-coenzyme A dehydrogenase deficiency
NPHS1	Finnish congenital nephrotic syndrome
PMM2	PMM2-CDG
FKTN	Dilated cardiomyopathy 1X
FKTN	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
SLC26A4	Autosomal recessive nonsyndromic hearing loss 4
SLC26A5	Pendred syndrome
ERCC2	Cerebrooculofacioskeletal syndrome 2
ERCC2	Trichothiodystrophy 1, photosensitive
DYNC2H1	Asphyxiating thoracic dystrophy 3
CEP290	Joubert syndrome 5
CEP290	Leber congenital amaurosis 10
GBE1	Glycogen storage disease, type IV

Genes	Disease
GBE1	Adult polyglucosan body disease
GAA	Glycogen storage disease, type II
CHRNE	Congenital myasthenic syndrome 4A
CHRNE	Congenital myasthenic syndrome 4B
G6PC1	Glycogen storage disease due to glucose-6-phosphatase deficiency type IA
OCA2	Tyrosinase-positive oculocutaneous albinism
COL7A1	Recessive dystrophic epidermolysis bullosa
ABCC8	Diabetes mellitus, permanent neonatal 3
ALDOB	Hereditary fructosuria
FANCC	Fanconi anemia complementation group C
GRIP1	Fraser syndrome 3
BCKDHB	Maple syrup urine disease
ANO10	Autosomal recessive spinocerebellar ataxia 10
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 1
NAGA	Alpha-N-acetylgalactosaminidase deficiency type 2
SMPD1	Niemann-Pick disease, type A
SMPD1	Niemann-Pick disease, type B
USH2A	Usher syndrome type 2A
MMUT	Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
CPT2	Carnitine palmitoyl transferase II deficiency, severe infantile form
CPT2	Carnitine palmitoyl transferase II deficiency, neonatal form
AHI1	Joubert syndrome 3
DHDDS	Retinitis pigmentosa 59
SLC19A3	Biotin-responsive basal ganglia disease
GALT	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
CYP11A1	Congenital adrenal insuffiency with 46, XY sex reversal OR 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
TF	Atransferrinemia
MMACHC	Cobalamin C disease
ABCA3	Interstitial lung disease due to ABCA3 deficiency
GBA1	Gaucher disease type I
GBA1	Gaucher disease type II

Genes	Disease
MCOLN1	Mucolipidosis type IV
GNPTAB	Mucolipidosis type II
GNPTAB	Pseudo-Hurler polydystrophy
AGA	Aspartylglucosaminuria
PCDH15	Autosomal recessive nonsyndromic hearing loss 23
PCDH15	Usher syndrome type 1F
FAH	Tyrosinemia type I
AIRE	Polyglandular autoimmune syndrome, type 1
BBS2	Bardet-Biedl syndrome 2
BBS2	Retinitis pigmentosa 74
CYP27A1	Cholestanol storage disease
CCDC88C	Hydrocephalus, nonsyndromic, autosomal recessive 1
FMO3	Trimethylaminuria
TMEM216	Joubert syndrome 2
TMEM216	Meckel syndrome, type 2
CNGB3	Achromatopsia 3
MCPH1	Primary microcephaly 1, recessive
SLC37A4	Glucose-6-phosphate transport defect
SLC37A4	Phosphate transport defect
PRF1	Familial hemophagocytic lymphohistiocytosis 2
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1
AGXT	Primary hyperoxaluria, type I
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency
ASL	Argininosuccinate lyase deficiency
EVC2	Ellis-van Creveld syndrome
ARSA	Metachromatic leukodystrophy
MVK	Hyperimmunoglobulin D with periodic fever
MVK	Mevalonic aciduria
PKHD1	Polycystic kidney disease 4
BTD	Biotinidase deficiency
ALPL	Adult hypophosphatasia
ALPL	Childhood hypophosphatasia

Genes	Disease
BBS1	Bardet-Biedl syndrome 1
CLCN1	Congenital myotonia, autosomal recessive form
CYP27B1	Vitamin D-dependent rickets, type 1A
POLG	Progressive sclerosing poliodystrophy
POLG	Mitochondrial DNA depletion syndrome 4b
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1
ACAT1	Deficiency of acetyl-CoA acetyltransferase
CC2D2A	Joubert syndrome 9
CC2D2A	Meckel syndrome, type 6
SLC26A2	Multiple epiphyseal dysplasia type 4
SLC26A2	Achondrogenesis, type IB
CBS	Classic homocystinuria
LRP2	Donnai-Barrow syndrome
IDUA	Hurler syndrome
IDUA	Mucopolysaccharidosis, MPS-I-H/S
FKRP	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A5
FKRP	Muscular dystrophy-dystroglycanopathy type B5
RNASEH2B	Aicardi-Goutieres syndrome 2
RARS2	Pontocerebellar hypoplasia type 6
HBA1	alpha Thalassemia
SMN1	Werdnig-Hoffmann disease
SMN1	Spinal muscular atrophy, type II
SMN1	Kugelberg-Welander disease
SMN1	Spinal muscular atrophy, type IV
HPS1	Hermansky-Pudlak syndrome 1
HPS3	Hermansky-Pudlak syndrome 3
ELP1	Familial dysautonomia
FXN	Friedreich ataxia 1
DLD	Pyruvate dehydrogenase E3 deficiency
NEB	Nemaline myopathy 2
CLRN1	Usher syndrome type 3A

Genes	Disease
BLM	Bloom syndrome
ABCD1	Adrenoleukodystrophy
AFF2	FRAXE
ARX	Developmental and epileptic encephalopathy, 1
DMD	Becker muscular dystrophy
DMD	Duchenne muscular dystrophy
F8	Hereditary factor VIII deficiency disease
F9	Hereditary factor IX deficiency disease
FMR1	Fragile X syndrome
GLA	Fabry disease
L1CAM	X-linked hydrocephalus syndrome
MID1	X-linked Opitz G/BBB syndrome
NR0B1	Congenital adrenal hypoplasia, X-linked
OTC	Ornithine carbamoyltransferase deficiency
PLP1	Hereditary spastic paraplegia 2
RPGR	Retinitis pigmentosa 3
RPGR	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness
RPGR	Macular degeneration, X-linked atrophic
RS1	Juvenile retinoschisis
SLC6A8	Creatine transporter deficiency
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SACS	Charlevoix-Saguenay spastic ataxia
HSD17B4	Bifunctional peroxisomal enzyme deficiency
MEFV	Familial Mediterranean fever
BCS1L	GRACILE syndrome
LAMB3	Junctional epidermolysis bullosa gravis of Herlitz
LRPPRC	Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type
SGCA	Autosomal recessive limb-girdle muscular dystrophy type 2D
SGCB	Autosomal recessive limb-girdle muscular dystrophy type 2E
CLN5	Neuronal ceroid lipofuscinosis 5
PPT1	Neuronal ceroid lipofuscinosis 1
NBN	Microcephaly, normal intelligence and immunodeficiency

Genes	Disease
GRHPR	Primary hyperoxaluria, type II
PKLR	Pyruvate kinase deficiency of red cells
PEX7	Rhizomelic chondrodysplasia punctata type 1
SLC17A5	Salla disease
ALDH3A2	Sjvðgren-Larsson syndrome
PEX1	Peroxisome biogenesis disorder 1A (Zellweger)
AAAS	Glucocorticoid deficiency with achalasia
AARS1	Charcot-Marie-Tooth disease axonal type 2N
ABCA12	Autosomal recessive congenital ichthyosis 4B
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
ABCC9	Hypertrichotic osteochondrodysplasia Cantu type
ABCC9	Dilated cardiomyopathy 1O
ABCC9	Atrial fibrillation, familial, 12
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
ACE	Renal tubular dysgenesis of genetic origin
ACOX1	Acyl-CoA oxidase deficiency
ACSF3	Combined malonic and methylmalonic acidemia
ACTA1	Actin accumulation myopathy
ACTA2	Aortic aneurysm, familial thoracic 6
ACTB	Dilated cardiomyopathy 1AA
ACTC1	Dilated cardiomyopathy 1R

Genes	Disease
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
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	Congenital myasthenic syndrome 8
AGL	Glycogen storage disease type III
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AIFM1	Charcot-Marie-Tooth disease X-linked recessive 4
AIP	Pituitary dependent hypercortisolism
AK2	Reticular dysgenesis
AKR1D1	Congenital bile acid synthesis defect 2
ALAS2	X-linked sideroblastic anemia 1
ALB	Analbuminemia
ALDH18A1	Cutis laxa, autosomal dominant 3
ALDH5A1	Succinate-semialdehyde dehydrogenase deficiency
ALDH7A1	Pyridoxine-dependent epilepsy
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

Genes	Disease
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
ALS2	Amyotrophic lateral sclerosis type 2, juvenile
ALX4	Parietal foramina 2
AMELX	Amelogenesis imperfecta type 1E
AMN	Imerslund-Grasbeck syndrome type 2
AMT	Non-ketotic hyperglycinemia
ANK1	Hereditary spherocytosis type 1
ANK2	Cardiac arrhythmia, ankyrin-B-related
ANKH	Craniometaphyseal dysplasia, autosomal dominant
ANKRD1	ANKRD1-related dilated cardiomyopathy
ANKRD26	Thrombocytopenia 2
ANO5	Autosomal recessive limb-girdle muscular dystrophy type 2L
ANTXR2	Hyaline fibromatosis syndrome
AP3B1	Hermansky-Pudlak syndrome 2
APC	Familial adenomatous polyposis 1
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
ARSB	Mucopolysaccharidosis type 6
ARX	X-linked lissencephaly with abnormal genitalia
ASS1	Citrullinemia type I

Genes	Disease
ATM	Ataxia-telangiectasia syndrome
ATP1A2	Migraine, familial hemiplegic, 2
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
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
BBS4	Bardet-Biedl syndrome 4
BBS5	Bardet-Biedl syndrome 5
BBS7	Bardet-Biedl syndrome 7
BBS9	Bardet-Biedl syndrome 9
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
BLNK	Agammaglobulinemia 4, autosomal recessive
BMPR1A	Juvenile polyposis syndrome
BMPR2	Pulmonary hypertension, primary, 1
BRAF	Cardiofaciocutaneous syndrome 1
BRCA2	Fanconi anemia complementation group D1

Genes	Disease
BRCA2	Medulloblastoma
BRCA2	Wilms tumor 1
BRIP1	Fanconi anemia complementation group J
BSCL2	Congenital generalized lipodystrophy type 2
BSND	Bartter disease type 4a
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
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
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
CASR	Familial hypocalciuric hypercalcemia 1
CASR	Autosomal dominant hypocalcemia 1

Genes	Disease
CASR	Neonatal severe primary hyperparathyroidism
CAV3	Rippling muscle disease 2
CBL	CBL-related disorder
CBLIF	Hereditary intrinsic factor deficiency
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
CDH1	Hereditary diffuse gastric adenocarcinoma
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
CDKN2A	Melanoma-pancreatic cancer syndrome
CDSN	Hypotrichosis 2
CEP152	Seckel 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
CFI	Atypical hemolytic-uremic syndrome with I factor anomaly
CFI	Factor I deficiency
CFL2	Nemaline myopathy 7
CFP	Properdin deficiency, X-linked

Genes	Disease
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	Congenital myasthenic syndrome 3B
CHRND	Congenital myasthenic syndrome 3C
CHRNE	Congenital myasthenic syndrome 4C
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
CLDN14	Autosomal recessive nonsyndromic hearing loss 29
CLDN19	Renal hypomagnesemia 5 with ocular involvement
CLN3	Neuronal ceroid lipofuscinosis 3
CLN6	Ceroid lipofuscinosis, neuronal, 6A
CLN8	Neuronal ceroid lipofuscinosis 8
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

Genes	Disease
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
COL5A1	Ehlers-Danlos syndrome, classic type, 1
COL5A2	Ehlers-Danlos syndrome, classic type, 2
COL6A1	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
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
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

Genes	Disease
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
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
CYP11B1	Glucocorticoid-remediable aldosteronism
CYP11B1	Deficiency of steroid 11-beta-monooxygenase
CYP11B2	Corticosterone methyloxidase type 2 deficiency
CYP17A1	Deficiency of steroid 17-alpha-monooxygenase
CYP4F22	Autosomal recessive congenital ichthyosis 5
D2HGDH	D-2-hydroxyglutaric aciduria 1
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
DES	Dilated cardiomyopathy 1I
GSDME	Autosomal dominant nonsyndromic hearing loss 5
PJVK	Autosomal recessive nonsyndromic hearing loss 59
DGAT1	Congenital diarrhea 7 with exudative enteropathy

Genes	Disease
DGUOK	Mitochondrial DNA depletion syndrome 3
DKC1	Dyskeratosis congenita, X-linked
DLL3	Spondylocostal dysostosis 1, autosomal recessive
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
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
DSP	Lethal acantholytic epidermolysis bullosa
DSP	Arrhythmogenic right ventricular dysplasia 8
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

Genes	Disease
EFTUD2	Mandibulofacial dysostosis-microcephaly syndrome
EGR2	Charcot-Marie-Tooth disease type 1D
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
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
ETHE1	Ethylmalonic encephalopathy
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	Congenital prothrombin deficiency

Genes	Disease
F9	Thrombophilia, X-linked, due to factor 9 defect
HYCC1	Hypomyelination and Congenital Cataract
FAM161A	Retinitis pigmentosa 28
FAM20C	Lethal osteosclerotic bone dysplasia
CCNQ	Syndactyly-telecanthus-anogenital and renal malformations syndrome
FANCA	Fanconi anemia complementation group A
FANCB	Fanconi anemia complementation group B
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
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
FGFR3	Muenke syndrome
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
FLAD1	Myopathy with abnormal lipid metabolism
FLCN	Multiple fibrofolliculomas
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
G6PC3	Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency
G6PD	Anemia, nonspherocytic hemolytic, due to G6PD deficiency
GALC	Galactosylceramide beta-galactosidase deficiency
GALE	UDPglucose-4-epimerase deficiency
GALK1	Deficiency of galactokinase
GALNS	Mucopolysaccharidosis, MPS-IV-A
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

Genes	Disease
GATA4	Atrioventricular septal defect 4
GATA6	Conotruncal heart malformations
GATA6	Pancreatic hypoplasia-diabetes-congenital heart disease syndrome
GATA6	Atrioventricular septal defect 5
GATA6	Atrial septal defect 9
GBA1	Gaucher disease perinatal lethal
GCDH	Glutaric aciduria, type 1
GCH1	Dystonia 5
GCH1	GTP cyclohydrolase I deficiency with hyperphenylalaninemia
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 12
GIPC3	Autosomal recessive nonsyndromic hearing loss 15
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
GLB1	Infantile GM1 gangliosidosis
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

Genes	Disease
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
GRIN2A	Landau-Kleffner syndrome
GSS	Glutathione synthetase deficiency without 5-oxoprolinuria
GUSB	Mucopolysaccharidosis type 7
GYS2	Glycogen storage disorder due to hepatic glycogen synthase deficiency
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
HAX1	Kostmann syndrome
HBA1	Hemoglobin H disease
HBA1	Heinz body anemia
HBA1	Methemoglobinemia, alpha type
HBA1	Erythrocytosis, familial, 7
HBB	Hb SS disease
HBG2	Cyanosis, transient neonatal
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX
HDAC8	Cornelia de Lange syndrome 5
HESX1	Septo-optic dysplasia sequence
HEXB	Sandhoff disease
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, MPS-III-C
HINT1	Autosomal recessive axonal neuropathy with neuromyotonia
HK1	Retinitis pigmentosa 79

Genes	Disease
HK1	Neurodevelopmental disorder with visual defects and brain anomalies
HK1	Hemolytic anemia due to hexokinase deficiency
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
HPS4	Hermansky-Pudlak syndrome 4
HPS5	Hermansky-Pudlak syndrome 5
HRAS	Costello syndrome
HSD17B10	HSD10 mitochondrial disease
HSD17B3	Testosterone 17-beta-dehydrogenase 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	Mucopolysaccharidosis, MPS-I-S
IER3IP1	Microcephaly, epilepsy, and diabetes syndrome 1
IGHM	Autosomal recessive agammaglobulinemia 1
IGHMBP2	Autosomal recessive distal spinal muscular atrophy 1
IGLL1	Agammaglobulinemia 2, autosomal recessive
IGSF1	X-linked central congenital hypothyroidism with late-onset testicular enlargement
IKBKG	Incontinentia pigmenti syndrome

Genes	Disease
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
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

Genes	Disease
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
KRT16	Pachyonychia congenita 1
KRT17	Pachyonychia congenita 2
KRT5	Epidermolysis bullosa simplex with mottled pigmentation
KRT6A	Pachyonychia congenita 3
LAMA2	Merosin deficient congenital muscular dystrophy
LAMB2	Pierson syndrome

Genes	Disease
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-HuV´t anomaly
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	StVºve-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
LOXHD1	Autosomal recessive nonsyndromic hearing loss 77
LRP4	Sclerosteosis 2
LRP4	Cenani-Lenz syndactyly syndrome
LRP4	Congenital myasthenic syndrome 17
LRP5	Osteoporosis with pseudoglioma
LRP5	Autosomal dominant osteopetrosis 1
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	ChV©diak-Higashi syndrome
MAD2L2	Fanconi anemia complementation group V

Genes	Disease
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
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
MCFD2	Factor 5 and Factor VIII, combined deficiency of, 2
MCPH1	Microcephaly 1, primary, autosomal recessive
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
MMADHC	Methylmalonic aciduria and homocystinuria type cblD
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

Genes	Disease
MPL	Congenital amegakaryocytic thrombocytopenia
MPV17	Navajo neurohepatopathy
MPZ	Charcot-Marie-Tooth disease type 2J
MRAP	Glucocorticoid deficiency 2
MSX2	Parietal foramina 1
MTHFR	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
MTHFR	Neural tube defects, folate-sensitive
MTM1	Severe X-linked myotubular myopathy
MTR	Methylcobalamin deficiency type cblG
MTRR	Methylcobalamin deficiency type cblE
MTTP	Abetalipoproteinaemia
MUSK	Congenital myasthenic syndrome 9
MUSK	Fetal akinesia deformation sequence 1
MUTYH	MUTYH-related attenuated familial adenomatous polyposis
MVK	Porokeratosis 3, disseminated superficial actinic type
MYBPC3	Hypertrophic cardiomyopathy 4
MYCN	Feingold syndrome type 1
MYH11	Aortic aneurysm, familial thoracic 4
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
MYL3	Hypertrophic cardiomyopathy 8
MYLK	Aortic aneurysm, familial thoracic 7
MYO15A	Autosomal recessive nonsyndromic hearing loss 3

Genes	Disease
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
NAGLU	Mucopolysaccharidosis, MPS-III-B
NAGS	Hyperammonemia, type III
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
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
NIPAL4	Autosomal recessive congenital ichthyosis 6
NIPBL	Cornelia de Lange syndrome 1
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

Genes	Disease
NR3C2	Pseudohyperaldosteronism type 2
NR3C2	Autosomal dominant pseudohypoaldosteronism type 1
NR5A1	Premature ovarian failure 7
NR5A1	46,XY sex reversal 3
NR5A1	Spermatogenic failure 8
NR5A1	46,XX sex reversal 4
NSD1	Sotos syndrome 1
NSDHL	Child syndrome
NTRK1	Hereditary insensitivity to pain with anhidrosis
OBSL1	3M syndrome 2
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
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
PAK3	Intellectual disability, X-linked 30
PANK2	Pigmentary pallidal degeneration
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
PCNT	Microcephalic osteodysplastic primordial dwarfism type II
PCSK9	Hypercholesterolemia, autosomal dominant, 3
PDE4D	Acrodysostosis 2 with or without hormone resistance

Genes	Disease
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
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
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

Genes	Disease
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
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type 1
PLP1	Pelizaeus-Merzbacher disease
PLPBP	Epilepsy, early-onset, vitamin B6-dependent
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
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
PQBP1	Renpenning syndrome
PREPL	Myasthenic syndrome, congenital, 22
PRKAG2	Wolff-Parkinson-White pattern
PRKAG2	Hypertrophic cardiomyopathy 6
PRKAR1A	Carney complex, type 1
PRKDC	Severe combined immunodeficiency due to DNA-PKcs deficiency
PROC	Thrombophilia due to protein C deficiency, autosomal dominant

Genes	Disease
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
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
RAB27A	Griscelli syndrome type 2
RAB3GAP1	Warburg micro syndrome 1
RAB7A	Charcot-Marie-Tooth disease type 2B
RAF1	Noonan syndrome 5
RAG1	Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive
RAG1	Combined immunodeficiency with skin granulomas
RAI1	Smith-Magenis syndrome
RAI1	Potocki-Lupski syndrome
RAPSN	Congenital myasthenic syndrome 11

Genes	Disease
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
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
RMRP	Anauxetic dysplasia 1
RNASEH2A	Aicardi-Goutieres syndrome 4
RNASEH2C	Aicardi-Goutieres syndrome 3
ROR2	Autosomal recessive Robinow syndrome
ROR2	Brachydactyly type B1
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

Genes	Disease
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
RRM2B	Mitochondrial DNA depletion syndrome 8a
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
SALL1	Townes-Brocks syndrome 1
SAMHD1	Aicardi-Goutieres syndrome 5
SBDS	Shwachman-Diamond syndrome 1
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

Genes	Disease
SCN4A	Hyperkalemic periodic paralysis
SCN4A	Potassium-aggravated myotonia
SCN4A	Congenital myasthenic syndrome 16
SCN5A	Long QT syndrome
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	Liddle syndrome 1
SCNN1G	Bronchiectasis with or without elevated sweat chloride 3
SCNN1G	Liddle syndrome 2
SDHAF2	Paragangliomas 2
SDHB	Paragangliomas 4
SDHB	Pheochromocytoma
SDHC	Paragangliomas 3
SDHD	Paragangliomas 1
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
SETX	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2
SFTPB	Neonatal acute respiratory distress due to SP-B deficiency
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-Sjʋðgren syndrome
SIX3	Holoprosencephaly 2
SKI	Shprintzen-Goldberg syndrome
SLC12A1	Bartter disease type 1
SLC12A3	Familial hypokalemia-hypomagnesemia
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
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

Genes	Disease
SLC25A38	Sideroblastic anemia 2
SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2
SLC26A3	Congenital secretory diarrhea, chloride type
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
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
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
SLC7A7	Lysinuric protein intolerance
SLC9A6	Christianson syndrome
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive, 2

Genes	Disease
SLX4	Fanconi anemia complementation group P
SMAD3	Aneurysm-osteoarthritis syndrome
SMARCAL1	Schimke immuno-osseous dysplasia
SMC1A	Congenital muscular hypertrophy-cerebral syndrome
SMPX	Myopathy, distal, 7, adult-onset, X-linked
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
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
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

Genes	Disease
SURF1	Cytochrome-c oxidase deficiency disease
TAT	Tyrosinemia type II
TAFAZZIN	3-Methylglutaconic aciduria type 2
TBC1D24	DOORS syndrome
TBX1	DiGeorge syndrome
TBX19	Congenital isolated adrenocorticotropic hormone deficiency
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
TFAP2A	Branchiooculofacial syndrome
TFAP2B	Char syndrome
TFG	Hereditary motor and sensory neuropathy, Okinawa type
TG	Autoimmune thyroid disease, susceptibility to, 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

Genes	Disease
TMEM43	Arrhythmogenic right ventricular dysplasia 5
TMEM67	Joubert syndrome 6
TMEM67	Meckel syndrome, type 3
TMIE	Autosomal recessive nonsyndromic hearing loss 6
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
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
TRMU	Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
TRPM4	Progressive familial heart block type IB

Genes	Disease
TSC1	Tuberous sclerosis 1
TSC1	Lymphangiomyomatosis
TSC1	Isolated focal cortical dysplasia type II
TSC2	Tuberous sclerosis 2
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
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
USH1G	Usher syndrome type 1G
VAMP1	Spastic ataxia 1
VAMP1	Myasthenic syndrome, congenital, 25, presynaptic
VCAN	Wagner syndrome
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
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2
VLDLR	Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1

Genes	Disease
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
XPA	Xeroderma pigmentosum group A
ZAP70	Combined immunodeficiency due to ZAP70 deficiency
ZEB2	Mowat-Wilson syndrome
ZIC2	Holoprosencephaly 5
ZIC3	Heterotaxy, visceral, 1, X-linked
ZMPSTE24	Lethal tight skin contracture syndrome
ZNF469	Brittle cornea syndrome 1