YENİDOĞAN GENETİK TARAMA TESTİ GEN LİSTESİ

ACIBADEM LABGEN

Genes	Disease
AAAS	Glucocorticoid deficiency with achalasia
AARS2	Combined oxidative phosphorylation deficiency 8
AARS2	Leukoencephalopathy, progressive, with ovarian failure
ABAT	GABA-transaminase deficiency
ABCC8	Diabetes mellitus, permanent neonatal 3
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1
ABCC8	Diabetes mellitus, transient neonatal, 2
ABCC8	Leucine-induced hypoglycemia
ABCD1	Adrenoleukodystrophy
ABCD4	Methylmalonic acidemia with homocystinuria, type cblJ
ACAD8	Deficiency of isobutyryl-CoA dehydrogenase
ACAD9	Acyl-CoA dehydrogenase 9 deficiency
ACADM	Medium-chain acyl-coenzyme A dehydrogenase deficiency
ACADS	Deficiency of butyryl-CoA dehydrogenase
ACADSB	2-methylbutyrylglycinuria
ACADVL	Very long chain acyl-CoA dehydrogenase deficiency
ACAT1	Deficiency of acetyl-CoA acetyltransferase
ACOX1	Acyl-CoA oxidase deficiency
ACOX2	Bile acid synthesis defect, congenital, 6
ACSF3	Combined malonic and methylmalonic acidemia
ADA	Severe combined immunodeficiency, autosomal recessive
ADK	Adenosine kinase deficiency
ADNP	Helsmoortel-van der Aa syndrome
ADSL	Adenylosuccinase deficiency
AGA	Aspartylglucosaminuria
AGA	Congenital myasthenic syndrome 8
AGL	Glycogen storage disease type III

Genes	Disease
AGXT	Primary hyperoxaluria, type I
AGXT	Glycogen storage disease type III
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AKR1D1	Congenital bile acid synthesis defect 2
ALAD	Porphyria, acute hepatic
ALDH4A1	Hyperprolinemia, type II
ALDH5A1	Succinate-semialdehyde dehydrogenase deficiency
ALDH7A1	Pyridoxine-dependent epilepsy
ALDOA	Glycogen storage disease XII
ALDOB	Hereditary fructosuria
ALG1	ALG1-CDG
ALG11	Congenital disorder of glycosylation, type Ip
ALG12	ALG12-congenital disorder of glycosylation
ALG13	Developmental and epileptic encephalopathy 36
ALG14	Intellectual developmental disorder with epilepsy, behavioral abnormalities
ALG14	Congenital myasthenic syndrome 15
ALG14	Myopathy, epilepsy, and progressive cerebral atrophy
ALG2	Congenital disorder of glycosylation, type Ii
ALG2	Myasthenic syndrome, congenital, 14, with tubular aggregates
ALG3	ALG3-CDG
ALG6	Congenital disorder of glycosylation type 1C
ALG8	ALG8 congenital disorder of glycosylation
ALG9	Congenital disorder of glycosylation, type Il
ALG9	Gillessen-Kaesbach-Nishimura syndrome
ALPL	Adult hypophosphatasia
ALPL	Childhood hypophosphatasia
AMACR	Alpha-methylacyl-CoA racemase deficiency
AMACR	Bile acid synthesis defect, congenital, 4
AMT	Non-ketotic hyperglycinemia
AP1S1	MEDNIK syndrome
ARG1	Arginase deficiency
ARSA	Metachromatic leukodystrophy

Genes	Disease
ARSB	Mucopolysaccharidosis type 6
ASAH1	Farber lipogranulomatosis
ASAH1	Spinal muscular atrophy with progressive myoclonic epilepsy
ASL	Argininosuccinate lyase deficiency
ASS1	Citrullinemia type I
ATP6V1B1	Renal tubular acidosis with progressive nerve deafness
ATP7A	Menkes kinky-hair syndrome
ATP7A	Cutis laxa, X-linked
ATP7B	Wilson disease
ATP7B	Wilson disease
AUH	3-methylglutaconic aciduria type 1
AVPR2	Diabetes insipidus, nephrogenic, X-linked
AVPR2	Nephrogenic syndrome of inappropriate antidiuresis
BCAT2	?Hypervalinemia or hyperleucine-isoleucinemia
BCKDHA	Maple syrup urine disease
BCKDHB	Maple syrup urine disease
BCKDK	Branched-chain keto acid dehydrogenase kinase deficiency
BTD	Biotinidase deficiency
BTK	X-linked agammaglobulinemia
BTK	X-linked agammaglobulinemia with growth hormone deficiency
CA5A	Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency
CAD	Developmental and epileptic encephalopathy 50
CASR	Epilepsy, idiopathic generalized, susceptibility to, 8
CASR	Familial hypocalciuric hypercalcemia 1
CASR	Autosomal dominant hypocalcemia 1
CASR	Neonatal severe primary hyperparathyroidism
CBS	Classic homocystinuria
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect
CD3D	Immunodeficiency 19
CD3E	Immunodeficiency 18
CDH23	Autosomal recessive nonsyndromic hearing loss 12
CDH23	Usher syndrome type 1D

Genes	Disease
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
CLPB	-methylglutaconic aciduria, type VIIA, autosomal dominant
CLPB	3-methylglutaconic aciduria, type VIIB, autosomal recessive
CLPB	Neutropenia, severe congenital, 9, autosomal dominant
COQ5	?Coenzyme Q10 deficiency, primary, 9
COQ8A	Autosomal recessive ataxia due to ubiquinone deficiency
CP	Deficiency of ferroxidase
CPS1	Pulmonary hypertension, neonatal, susceptibility to
CPS1	Congenital hyperammonemia, type I
CPT1A	Carnitine palmitoyl transferase 1A deficiency
CPT2	Carnitine palmitoyl transferase II deficiency, severe infantile form
CPT2	Carnitine palmitoyl transferase II deficiency, neonatal form
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
CTH	Cystathioninuria
CTNS	Nephropathic cystinosis
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
CYBC1	Chronic granulomatous disease 5, autosomal recessive
CYP11B1	Glucocorticoid-remediable aldosteronism
CYP11B1	Deficiency of steroid 11-beta-monooxygenase
CYP11B2	Corticosterone methyloxidase type 2 deficiency
CYP17A1	Deficiency of steroid 17-alpha-monooxygenase
CYP27A1	Cholestanol storage disease
CYP27B1	Vitamin D-dependent rickets, type 1A
DBT	Maple syrup urine disease

Genes	Disease
DCLRE1C	Severe combined immunodeficiency due to DCLRE1C deficiency
DCLRE1C	Histiocytic medullary reticulosis
DDC	Deficiency of aromatic-L-amino-acid decarboxylase
DHCR7	Smith-Lemli-Opitz syndrome
DHFR	Megaloblastic anemia due to dihydrofolate reductase deficiency
DLAT	Pyruvate dehydrogenase E2 deficiency
DLD	Pyruvate dehydrogenase E3 deficiency
DMD	Becker muscular dystrophy
DMD	Duchenne muscular dystrophy
DMD	Dilated cardiomyopathy 3B
DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient
DOCK8	Combined immunodeficiency due to DOCK8 deficiency
DUOX2	Thyroid dyshormonogenesis 6
DUOXA2	Thyroid dyshormonogenesis 5
ECHS1	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
ELANE	Neutropenia, severe congenital, 1, autosomal dominant
ETFA	Multiple acyl-CoA dehydrogenase deficiency
ETFB	Multiple acyl-CoA dehydrogenase deficiency
ETFDH	Multiple acyl-CoA dehydrogenase deficiency
ETHE1	Ethylmalonic encephalopathy
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
FARSB	Rajab interstitial lung disease with brain calcifications 1
FBP1	Fructose-biphosphatase deficiency
FECH	Protoporphyria, erythropoietic, 1
FGFR3	Severe achondroplasia-developmental delay-acanthosis nigricans syndrome
FGFR3	Hypochondroplasia
FGFR3	Crouzon syndrome-acanthosis nigricans syndrome
FGFR3	Thanatophoric dysplasia type 1
FGFR3	Muenke syndrome

Genes	Disease
FOLR1	Neurodegeneration due to cerebral folate transport deficiency
FUCA1	Fucosidosis
G6PC1	Glycogen storage disease due to glucose-6-phosphatase deficiency type IA
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
GALM	Galactosemia IV
GALNS	Mucopolysaccharidosis, MPS-IV-A
GALT	Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
GAMT	Deficiency of guanidinoacetate methyltransferase
GATM	Cerebral creatine deficiency syndrome 3
GATM	Fanconi renotubular syndrome 1
GBA1	Gaucher disease type I
GBA1	Gaucher disease type II
GBA1	Gaucher disease perinatal lethal
GCDH	Glutaric aciduria, type 1
GCH1	Dystonia 5
GCH1	GTP cyclohydrolase I deficiency with hyperphenylalaninemia
GJB2	Autosomal recessive nonsyndromic hearing loss 1A
GJB2	Autosomal dominant nonsyndromic hearing loss 3A
GJB2	Knuckle pads, deafness AND leukonychia syndrome
GJB2	Palmoplantar keratoderma-deafness syndrome
GJB6	Deafness, autosomal dominant 3B
GJB6	Deafness, autosomal recessive 1B
GJB6	Deafness, digenic GJB2/GJB6
GJB6	Ectodermal dysplasia 2, Clouston type
GLA	Fabry disease
GLB1	Infantile GM1 gangliosidosis
GLDC	Non-ketotic hyperglycinemia
GLUD1	Hyperinsulinism-hyperammonemia syndrome

Genes	Disease
GLUL	Glutamine deficiency, congenital
GNMT	Glycine N-methyltransferase deficiency
GNS	Mucopolysaccharidosis, MPS-III-D
GOT2	Developmental and epileptic encephalopathy, 82
GRHPR	Primary hyperoxaluria, type II
GRIN1	Developmental and epileptic encephalopathy 101
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal dominant
GRIN1	Neurodevelopmental disorder with or without hyperkinetic movements and seizures, autosomal recessive
GRIN2A	Landau-Kleffner syndrome
GRIN2B	Developmental and epileptic encephalopathy 27
GRIN2B	Intellectual developmental disorder, autosomal dominant 6, with or without seizures
GRIN2D	Developmental and epileptic encephalopathy 46
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
HADHB	Mitochondrial trifunctional protein deficiency
HAX1	Kostmann syndrome
HBB	beta Thalassemia
HBB	Hb SS disease
HCFC1	Methylmalonic acidemia with homocystinuria, type cblX
HEXA	Tay-Sachs disease
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, MPS-III-C
HIBCH	3-hydroxyisobutryl-CoA hydrolase deficiency
HLCS	Holocarboxylase synthetase deficiency
HMGCL	Deficiency of hydroxymethylglutaryl-CoA lyase
HMGCS2	HMG-CoA synthase-2 deficiency
HOGA1	Hyperoxaluria, primary, type III

Genes	Disease
HPD	Hawkinsinuria
HPD	Tyrosinemia type III
HSD17B10	HSD10 mitochondrial disease
HSD3B2	3 beta-Hydroxysteroid dehydrogenase deficiency
HSD3B7	Congenital bile acid synthesis defect 1
IARS1	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy
IDS	Mucopolysaccharidosis, MPS-II
IDUA	Hurler syndrome
IDUA	Mucopolysaccharidosis, MPS-I-H/S
IDUA	Mucopolysaccharidosis, MPS-I-S
IGF1	Insulin-like growth factor I deficiency
IGSF1	X-linked central congenital hypothyroidism with late-onset testicular enlargement
IL2RG	X-linked severe combined immunodeficiency
IL2RG	Combined immunodeficiency, X-linked
IL7R	Immunodeficiency 104
IRS4	Hypothyroidism, congenital, nongoitrous, 9
IVD	Isovaleryl-CoA dehydrogenase deficiency
IYD	Thyroid dyshormonogenesis 4
JAG1	Alagille syndrome due to a JAG1 point mutation
JAK3	T-B+ severe combined immunodeficiency due to JAK3 deficiency
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
KCNQ2	Seizures, benign familial neonatal, 1
KCNQ2	Developmental and epileptic encephalopathy, 7
LARS1	?Infantile liver failure syndrome 1
LHX3	Non-acquired combined pituitary hormone deficiency with spine abnormalities
LIPA	Lysosomal acid lipase deficiency
LMBRD1	Methylmalonic aciduria and homocystinuria type cblF
MAN2B1	Deficiency of alpha-mannosidase
MARS1	?Trichothiodystrophy 9, nonphotosensitive

Genes	Disease
MARS1	Charcot-Marie-Tooth disease, axonal, type 2U
MARS1	Interstitial lung and liver disease
MARS1	Spastic paraplegia 70, autosomal recessive
MAT1A	Hepatic methionine adenosyltransferase deficiency
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
MFSD8	Neuronal ceroid lipofuscinosis 7
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
MOCS1	Sulfite oxidase deficiency due to molybdenum cofactor deficiency type A
MPI	MPI-CDG
MT-CO1	Mitochondrial disorders
MT-CO3	Mitochondrial disorders
MT-CPO2	Mitochondrial disorders
MTHFR	Homocystinuria due to methylene tetrahydrofolate reductase deficiency
MTHFR	Thrombophilia due to thrombin defect
MTHFR	Neural tube defects, folate-sensitive
MTHFS	Mitochondrial disorders
MT-ND1	Mitochondrial disorders
MT-ND4	Mitochondrial disorders
MT-ND5	Mitochondrial disorders
MT-ND6	Mitochondrial disorders
MTR	Methylcobalamin deficiency type cblG
MTR	Neural tube defects, folate-sensitive
MTRR	Methylcobalamin deficiency type cblE
MTRR	Neural tube defects, folate-sensitive
MT-TF	Mitochondrial disorders

Genes	Disease
MT-TH	Mitochondrial disorders
MT-TL1	Mitochondrial disorders
MT-TQ	Mitochondrial disorders
MT-TS1	Mitochondrial disorders
MT-TS2	Mitochondrial disorders
MT-TW	Mitochondrial disorders
MVK	Hyperimmunoglobulin D with periodic fever
MVK	Mevalonic aciduria
MVK	Hyperimmunoglobulin D with periodic fever
MVK	Porokeratosis 3, disseminated superficial actinic type
MVK	Mevalonic aciduria
MYO7A	Usher syndrome type 1
NAGLU	Mucopolysaccharidosis, MPS-III-B
NAGS	Hyperammonemia, type III
NAXE	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy
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
NFE2L2	Immunodeficiency, developmental delay, and hypohomocysteinemia
NKX2-5	Atrial septal defect 7, with or without AV conduction defects
NKX2-5	Conotruncal heart malformations, variable
NKX2-5	Hypoplastic left heart syndrome 2
NKX2-5	Hypothyroidism, congenital nongoitrous, 5
NKX2-5	Tetralogy of Fallot
NKX2-5	Ventricular septal defect 3
NLRP3	CINCA syndrome
NLRP3	Deafness, autosomal dominant 34, with or without inflammation
NLRP3	Familial cold inflammatory syndrome 1
NLRP3	Keratoendothelitis fugax hereditaria
NLRP3	Muckle-Wells syndrome
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-Pick disease, type C2

Genes	Disease
NR0B1	Congenital adrenal hypoplasia, X-linked
NR0B1	Congenital adrenal hypoplasia, X-linked
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia
OPA3	3-Methylglutaconic aciduria type 3
OTC	Ornithine carbamoyltransferase deficiency
PAH	Phenylketonuria
PAX8	Hypothyroidism, congenital, nongoitrous, 2
PCBD1	Pterin-4 alpha-carbinolamine dehydratase 1 deficiency
PCCA	Propionic acidemia
PCCB	Propionic acidemia
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PDHX	Pyruvate dehydrogenase E3-binding protein deficiency
PDP1	Pyruvate dehydrogenase phosphatase deficiency
PHEX	Familial X-linked hypophosphatemic vitamin D refractory rickets
PHGDH	Neu-Laxova syndrome 1
PHGDH	PHGDH deficiency
PIGA	Multiple congenital anomalies-hypotonia-seizures syndrome 2
PIGA	Neurodevelopmental disorder with epilepsy and hemochromatosis
PIGM	Glycosylphosphatidylinositol deficiency
PIGO	Hyperphosphatasia with impaired intellectual development syndrome 2
PLPBP	Epilepsy, early-onset, vitamin B6-dependent
PMM2	PMM2-CDG
PNPO	Pyridoxal phosphate-responsive seizures
POU1F1	Pituitary hormone deficiency, combined, 1
PPM1K	?Maple syrup urine disease, mild variant
PRDX1	Cobalamin C disease
PRODH	Hyperprolinemia, type I
PROP1	Pituitary hormone deficiency, combined, 2
PROSC	Epilepsy, early-onset, 1, vitamin B6-dependent
PRPS1	Arts syndrome
PRPS1	Charcot-Marie-Tooth disease, X-linked recessive, 5

Genes	Disease
PRPS1	Deafness, X-linked 1
PRPS1	Gout, PRPS-related
PRPS1	Phosphoribosylpyrophosphate synthetase superactivity
PSAT1	PSAT deficiency
PSAT1	Neu-Laxova syndrome 2
PSPH	Deficiency of phosphoserine phosphatase
PTS	6-Pyruvoyl-tetrahydrobiopterin synthase deficiency
PYGL	Glycogen storage disease, type VI
QDPR	Dihydropteridine reductase deficiency
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
RFX5	Bare lymphocyte syndrome, type II, complementation group C
RFX5	Bare lymphocyte syndrome, type II, complementation group E
RFXANK	MHC class II deficiency
RFXAP	Bare lymphocyte syndrome, type II, complementation group D
RPE65	Leber congenital amaurosis 2
RPE65	Retinitis pigmentosa 20
RPE65	Retinitis pigmentosa 87 with choroidal involvement
SARS1	Neurodevelopmental disorder with microcephaly, ataxia, and seizures
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
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

Genes	Disease
SCNN1B	Bronchiectasis with or without elevated sweat chloride 1
SECISBP2	Thyroid hormone metabolism, abnormal, 1
SERPING1	Angioedema, hereditary, 1 and 2
SERPING1	Complement component 4, partial deficiency of
SGSH	Mucopolysaccharidosis, MPS-III-A
SLC18A2	Parkinsonism-dystonia, infantile, 2
SLC19A3	Biotin-responsive basal ganglia disease
SLC22A5	Renal carnitine transport defect
SLC25A13	Citrullinemia type II
SLC25A13	Citrullinemia, type II, adult-onset
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
SLC25A19	Microcephaly, Amish type
SLC25A19	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type)
SLC2A1	Childhood onset GLUT1 deficiency syndrome 2
SLC2A1	Dystonia 9
SLC2A1	Hereditary cryohydrocytosis with reduced stomatin
SLC2A1	Epilepsy, idiopathic generalized, susceptibility to, 12
SLC34A3	Autosomal recessive hypophosphatemic bone disease
SLC35A2	SLC35A2-CDG
SLC35C1	Congenital disorder of glycosylation, type IIc
SLC37A4	Phosphate transport defect
SLC37A4	Glucose-6-phosphate transport defect
SLC37A4	Phosphate transport defect
SLC39A4	Hereditary acrodermatitis enteropathica
SLC39A8	SLC39A8-CDG
SLC3A1	Cystinuria
SLC46A1	Congenital defect of folate absorption
SLC5A5	Familial thyroid dyshormonogenesis 1
SLC5A6	Peripheral motor neuropathy, childhood-onset, biotin-responsive
SLC5A6	Sodium-dependent multivitamin transporter deficiency
SLC6A8	Creatine transporter deficiency
SLC7A7	Lysinuric protein intolerance

Genes	Disease
SLC7A9	Cystinuria
SMN1	Werdnig-Hoffmann disease
SMN1	Spinal muscular atrophy, type II
SMN1	Kugelberg-Welander disease
SMN1	Spinal muscular atrophy, type IV
SMPD1	Niemann-Pick disease, type A
SMPD1	Niemann-Pick disease, type B
SPR	Dopa-responsive dystonia due to sepiapterin reductase deficiency
TAT	Tyrosinemia type II
TBL1X	Hypothyroidism, congenital, nongoitrous, 8
TCN2	Transcobalamin II deficiency
TG	Autoimmune thyroid disease, susceptibility to, 3
THRA	Congenital nongoitrous hypothryoidism 6
TIMM50	3-methylglutaconic aciduria, type IX
TMIE	Autosomal recessive nonsyndromic hearing loss 6
TMLHE	{Autism, susceptibility to, X-linked 6}
TMPRSS3	Autosomal recessive nonsyndromic hearing loss 8
TPK1	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)
TPO	Deficiency of iodide peroxidase
TPP1	Neuronal ceroid lipofuscinosis 2
TPRN	Deafness, autosomal recessive 79
TRHR	Hypothyroidism, congenital, nongoitrous, 7
TRIOBP	Autosomal recessive nonsyndromic hearing loss 28
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
TSHB	Secondary hypothyroidism
TSHR	Hypothyroidism due to TSH receptor mutations
TTPA	Familial isolated deficiency of vitamin E

Genes	Disease
TTR	Amyloidogenic transthyretin amyloidosis
UGT1A1	Crigler-Najjar syndrome type 1
USH1C	Usher syndrome type 1
USH1G	Usher syndrome type 1G
USH2A	Usher syndrome type 2A
VDR	Vitamin D-dependent rickets type II with alopecia
WHRN	Deafness, autosomal recessive 31
WHRN	Usher syndrome, type 2D