Gene	Disease
ABCB11	Cholestasis, progressive familial intrahepatic 2
ABCB11	Cholestasis, benign recurrent intrahepatic, 2
ABCC2	Dubin-Johnson syndrome
ABCC8	Diabetes, noninsulin dependent
ABCC8	Diabetes, permanent neonatal
ABCC8	Hyperinsulinemic hypoglycemia, familial, 1
ABCC8	Hypoglycemia, infantile
ABCD1	Adrenoleukodystrophy
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of
ACADVL	Acyl-CoA dehydrogenase, very long-chain, deficiency of
ACAT1	Alpha-methylacetoacetic aciduria
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACTA1	Myopathy congenital, with fiber-type disproportion
ACTA1	Nemaline myopathy 3, AD or recessive
ADA	Severe combined immunodeficiency due to ADA deficiency
ADAMTS2	Ehlers-Danlos syndrome
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease IIIa, IIb
AGXT	Hyperoxaluria, primary, type 1
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia
ALDH3A2	Sjogren-Larsson syndrome
ALDH4A1	Hyperprolinemia type II
ALDOB	Fructose intolerance, hereditary
ALG6	Congenital disorder of glycosylation, type Ic
ALPL	Hypophosphatasia
AMH	Persistent Mullerian duct syndrome, type I
AMHR2	Persistent Mullerian duct syndrome, type II
AMPD1	Myopathy due to myoadenylate deaminase deficiency
AMT	Glycine encephalopathy
AR	Androgen insensitivity
AR	Androgen insensitivity
AR	Hypospadias
AR	Spinal and bulbar muscular atrophy of Kennedy
ARG1	Argininemia
ARL13B	Joubert syndrome 8
ARSA	Metachromatic leukodystrophy
ASL	Argininosuccinic aciduria

ASPA	Canavan disease
ASS1	Citrullinemia
ATM	Ataxia-telangiectasia
ATP7A	Menkes disease
ATP7A	Occipital horn syndrome
ATP7A	Spinal muscular atrophy, distal, X-linked 3
ATP7B	Wilson disease
BBS1	Bardet-Biedl syndrome 1
BBS10	Bardet-Biedl syndrome 10
BBS12	Bardet-Biedl syndrome 12
BBS2	Bardet-Biedl syndrome 2
BBS2	Retinitis pigmentosa 74
BCHE	Postanesthesia apnea
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BCS1L	Bjornstad syndrome
BCS1L	GRACILE syndrome
BCS1L	Leigh syndrome
BCS1L	Mitochondrial complex III deficiency, nuclear type 1
BLM	Bloom syndrome
BRIP1	Fanconi anemia, complementation group J
BTD	biotinidase deficiency
CAPN3	Muscular dystrophy, limb-girdle, type 2A
CBS	Homocystinuria, B6-responsive and nonresponsive types
CDH23	Deafness
CDH23	Usher syndrome, type 1D/F digenic
CEP290	Bardet-Biedl syndrome 14
CEP290	Joubert syndrome 5
CEP290	Meckel Syndrome 4
CEP290	Senior-Loken syndrome 6
CFTR	Cystic fibrosis
CFTR	Congenital bilateral absence of vas deferens
CHM	Choroideremia
CHRNA1	Multiple pterygium syndrome, lethal type
CHRNA1	Myasthenic syndrome, congenital, 1B, fast-channel
CHRNA1	Myasthenic syndrome, congenital, 1A, slow-channel
CHRND	Multiple pterygium syndrome, lethal type
CHRND	Myasthenic syndrome, congenital, 3A, slow-channel
CHRND	Myasthenic syndrome, congenital, 3b, fast-channel
CHRND	Myasthenic syndrome, congenital, 3C, acetylcholine receptor deficiency
CHRNG	Escobar syndrome

CHRNG	Multiple pterygium syndrome, lethal type
CIITA	Bare lymphocyte syndrome, type II, complementation group A
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN5	Ceroid lipofuscinosis, neuronal, 5
CLN6	Ceroid lipofuscinosis, neuronal, 6