GeneAware™ (version 4) Reproductive Carrier Screen

BAYLOR GENETICS

GeneAware is a reproductive carrier test that screens for disease-causing variants in over 400 genes through full gene sequencing, supplemented with copy number analysis and fragile X triplet repeat analysis

The risk for carrying certain genetic conditions varies from patient to patient based on several factors. Because of this diversity, we offer five different GeneAware panels to better meet the needs of your patients.

In all five panel options, individuals are screened for cystic fibrosis (CFTR) and spinal muscular atrophy (SMN1). In addition, females are screened for X-linked Duchenne and Becker muscular dystrophies (DMD) and Fragile X syndrome (FMR1).

ACMG AND ACOG	Disorders recommended for screening by the ACMG and ACOG
ASHKENAZI JEWISH	Disorders specific for individuals of Ashkenazi Jewish descent
BASIC	The most commonly requested disorders
COMPLETE	A selection of common disorders
EXPANDED	The most comprehensive screening

Gene	Disease Name
AAAS	Triple A syndrome
ABAT	GABA-transaminase deficiency
ABCA12	Congenital ichthyosis, ABCA12-related
ABCA3	Surfactant dysfunction, ABCA3-related
ABCA4	ABCA4-related disorders
ABCB11	Progressive familial intrahepatic cholestasis 2
ABCB4	Progressive familial intrahepatic cholestasis 3
ABCC2	Dubin-Johnson syndrome
ABCC6	Pseudoxanthoma elasticum
ABCC8	Familial hyperinsulinism, ABCC8-related
ABCD1	Adrenoleukodystrophy, X-linked
ACAD9	Mitochondrial complex I deficiency, ACAD9-related
ACADM	Medium chain acyl-CoA dehydrogenase deficiency

Gene	Disease Name
ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency
ACAT1	Beta-ketothiolase deficiency
ACOX1	Peroxisomal acyl-CoA oxidase deficiency
ACSF3	Combined malonic and methylmalonic aciduria
ADA	Adenosine deaminase deficiency
ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type
ADAMTSL4	ADAMTSL4-related eye disorders
ADGRG1	Bilateral frontoparietal polymicrogyria
ADGRV1	Usher syndrome, type 2C
AFF2	Fragile XE syndrome
AGA	Aspartylglycosaminuria
AGL	Glycogen storage disease, type III
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Primary hyperoxaluria, type I

Gene	Disease Name
AHI1	Joubert syndrome 3
AIFM1	Combined oxidative phosphorylation deficiency 6
AIPL1	Leber congenital amaurosis 4
AIRE	Autoimmune polyglandular syndrome, type 1
ALDH3A2	Sjogren-Larsson syndrome
ALDH7A1	Pyridoxine-dependent epilepsy
ALDOB	Hereditary fructose intolerance
ALG1	Congenital disorder of glycosylation, type Ik
ALG13	Developmental and epileptic encephalopathy 36
ALG6	Congenital disorder of glycosylation, type Ic
ALMS1	Alstrom syndrome
ALPL	Hypophosphatasia
AMN	Imerslund-Gräsbeck syndrome 2
AMT	Glycine encephalopathy, AMT-related
ANO10	Autosomal recessive spinocerebellar ataxia, type 10
AP1S1	MEDNIK syndrome
AP3B1	Hermansky-Pudlak syndrome, type 2
AQP2	Nephrogenic diabetes insipidus
AR	Androgen insensitivity syndrome
ARG1	Argininemia
ARL13B	Joubert syndrome 8
ARL6	Bardet-Biedl syndrome 3
ARSA	Metachromatic leukodystrophy, ARSA-related
ARSB	Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome
ARSL	X-linked chondrodysplasia punctata 1
ARX	X-linked developmental disorders, ARX-related

Gene	Disease Name
ASAH1	Farber lipogranulomatosis
ASL	Argininosuccinic aciduria
ASNS	Asparagine synthetase deficiency
ASPA	Canavan disease
ASS1	Citrullinemia, type I
ATM	Ataxia-telangiectasia
ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related
ATP7A	Menkes disease
ATP7B	Wilson disease
ATP8B1	Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1
ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked
AVPR2	AVPR2-related disorders
B9D1	Joubert syndrome 27
B9D2	Joubert syndrome 34
BBS1	Bardet-Biedl syndrome 1
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
BCHE	Pseudocholinesterase deficiency
BCKDHA	Maple syrup urine disease, type 1A
BCKDHB	Maple syrup urine disease, type 1B
BCS1L	GRACILE syndrome
BLM	Bloom syndrome
BLOC1S3	Hermansky-Pudlak syndrome, type 8

Gene	Disease Name
BLOC1S6	Hermansky-Pudlak syndrome, type 9
BMP1	Osteogenesis imperfecta, type XIII
BRIP1	Fanconi anemia, complementation group J
BSND	Bartter syndrome, type 4A
BTD	Biotinidase deficiency
BTK	X-linked agammaglobulinemia
C2CD3	Orofaciodigital syndrome, type XIV
CAD	Developmental and epileptic encephalopathy 50
CANT1	Desbuquois dysplasia, type I
CAPN3	Limb-girdle muscular dystrophy, type 2A
CASQ2	Catecholaminergic polymorphic ventricular tachycardia, type 2
CBS	Homocystinuria, CBS-related
CC2D1A	Autosomal recessive intellectual developmental disorder, type 3
CC2D2A	Joubert syndrome 9
CCDC103	Primary ciliary dyskinesia, CCDC103-related
CCDC39	Primary ciliary dyskinesia, CCDC39-related
CCDC88C	Congenital hydrocephalus 1
CCN6	Progressive pseudorheumatoid dysplasia
CD3D	Severe combined immunodeficiency, CD3D-related
CD3E	Severe combined immunodeficiency, CD3E-related
CD40	Hyper-IgM syndrome, type 3
CD40LG	X-Linked hyper IgM syndrome
CD59	CD59-mediated hemolytic anemia
CDH23	Usher syndrome, type 1D
CEP104	Joubert syndrome 25
CEP120	Joubert syndrome 31
CEP152	CEP152-related disorders

Gene	Disease Name
CEP290	Leber congenital amaurosis, CEP290-related / CEP290-related conditions
CEP41	Joubert syndrome 15
CERKL	Retinitis pigmentosa 26
CFTR	Cystic fibrosis
CHAT	Congenital myasthenic syndrome, CHAT-related
CHM	Choroideremia, X-linked
CHRNE	Congenital myasthenic syndrome, CHRNE-related
CHRNG	Multiple pterygium syndrome, lethal type
CIB2	Usher syndrome, type 1J
CIITA	Bare lymphocyte syndrome, type II
CLCN1	Myotonia congenita
CLCN5	Dent disease
CLN3	Neuronal ceroid lipofuscinosis, CLN3-related
CLN5	Neuronal ceroid lipofuscinosis, CLN5-related
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related
CLN8	Neuronal ceroid lipofuscinosis, CLN8-related
CLRN1	Usher syndrome, type 3A
CNGB3	Achromatopsia, CNGB3-related
COL11A2	COL11A2-related disorders
COL17A1	Junctional epidermolysis bullosa, COL17A1-related
COL27A1	Steel syndrome
COL4A3	Alport syndrome, COL4A3-related
COL4A4	Alport syndrome, COL4A4-related
COL4A5	Alport syndrome, COL4A5-related, X-linked
COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related
COLQ	Congenital myasthenic syndrome, COLQ-related

Gene	Disease Name
COX15	Mitochondrial complex IV deficiency, nuclear type 6
CPLANE1	Joubert syndrome 17
CPS1	Carbamoyl phosphate synthetase I deficiency
CPT1A	Carnitine palmitoyltransferase I deficiency
CPT2	Carnitine palmitoyltransferase II deficiency
CRB1	CRB1-related retinal dystrophies
CRPPA	Muscular dystrophy-dystroglycanopathy, type A, 7
CRTAP	Osteogenesis imperfecta, type VII
CSPP1	Joubert syndrome 21
CTNS	Cystinosis
CTSA	Galactosialidosis
CTSC	CTSC-related disorders
CTSD	Neuronal ceroid lipofuscinosis, CTSD-related
CTSK	Pycnodysostosis
CYBA	Chronic granulomatous disease 4
CYBB	Chronic granulomatous disease, X-linked
CYP11A1	Congenital adrenal insufficiency, CYP11A1-related
CYP11B1	Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency
CYP11B2	Corticosterone methyloxidase deficiency
CYP17A1	Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency
CYP19A1	Aromatase deficiency
CYP1B1	Primary congenital glaucoma
CYP21A2	Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency
CYP27A1	Cerebrotendinous xanthomatosis
CYP27B1	Vitamin D-dependent rickets, type 1A

Gene	Disease Name
CYP7B1	CYP7B1-related disorders
DBT	Maple syrup urine disease, type 2
DCAF17	Woodhouse-Sakati syndrome
DCLRE1C	Omenn syndrome
DCX	DCX-related disorders
DDX11	Warsaw breakage syndrome
DGAT1	DGAT1 deficiency
DGUOK	Deoxyguanosine kinase deficiency / Mitochondrial DNA depletion syndrome 3
DHCR7	Smith-Lemli-Opitz syndrome
DHDDS	Retinitis pigmentosa 59
DIS3L2	Perlman syndrome
DKC1	Dyskeratosis congenita, X-linked
DLD	Dihydrolipoamide dehydrogenase deficiency
DLL3	Spondylocostal dysostosis 1
DMD	Duchenne/Becker muscular dystrophy, X-linked
DNAH11	Primary ciliary dyskinesia, DNAH11-related
DNAH5	Primary ciliary dyskinesia, DNAH5-related
DNAI1	Primary ciliary dyskinesia, DNAI1-related
DNAI2	Primary ciliary dyskinesia, DNAI2-related
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1
DOK7	Congenital myasthenic syndrome, DOK7-related
DPYD	Dihydropyrimidine dehydrogenase deficiency
DUOX2	Thyroid dyshormonogenesis 6
DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly
DYSF	Limb-girdle muscular dystrophy, type 2B
EDA	Hypohidrotic ectodermal dysplasia, X-linked
EIF2AK3	Wolcott-Rallison syndrome

Gene	Disease Name
EIF2B1	Leukoencephalopathy with vanishing white matter 1
EIF2B2	Leukoencephalopathy with vanishing white matter 2
EIF2B3	Leukoencephalopathy with vanishing white matter 3
EIF2B4	Leukoencephalopathy with vanishing white matter 4
EIF2B5	Leukoencephalopathy with vanishing white matter 5
ELP1	Familial dysautonomia
EMD	Emery-Dreifuss muscular dystrophy, X-linked
EPG5	EPG5-related disorder
ERCC2	ERCC2-related conditions
ERCC6	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B
ERCC8	Cockayne syndrome, type A
ESCO2	Roberts-SC phocomelia syndrome
ETFA	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA
ETFB	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB
ETFDH	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC
ETHE1	Ethylmalonic encephalopathy
EVC	Ellis-van Creveld syndrome
EVC2	Ellis-van Creveld syndrome
EXOSC3	Pontocerebellar hypoplasia, type 1B
EYS	Retinitis pigmentosa 25
F11	Factor XI deficiency / Hemophilia C
F2	Factor II deficiency / Prothrombin deficiency
F5	Factor V deficiency
F8	Factory VIII deficiency / Hemophilia A
F9	Factor IX deficiency / Hemophilia B

Gene	Disease Name
FAH	Tyrosinemia, type I
FAM161A	Retinitis pigmentosa 28
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
FBP1	Fructose-1,6-bisphosphatase deficiency
FBXO7	Parkinson disease 15
FH	Fumarase deficiency
FHL1	FHL1-related disorders
FKBP10	Osteogenesis imperfecta, type XI
FKRP	Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5
FKTN	Fukuyama congenital muscular dystrophy
FMO3	Trimethylaminuria
FMR1	Fragile X syndrome
FOXN1	Severe combined immunodeficiency, FOXN1-related
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
FOXRED1	Mitochondrial complex I deficiency, nuclear type 19
FRAS1	Fraser syndrome, type 1
FREM2	Fraser syndrome, type 2
FUCA1	Fucosidosis

Gene	Disease Name
FXN	Friedreich ataxia
G6PC1	Glycogen storage disease, type Ia
G6PC3	G6PC3 deficiency
G6PD	Glucose-6-phosphate dehydrogenase deficiency
GAA	Glycogen storage disease, type II / Pompe disease
GALC	Krabbe disease
GALE	Galactosemia, type III / Galactose epimerase deficiency
GALK1	Galactosemia, type II / Galactokinase deficiency
GALNS	Mucopolysaccharidosis, type IVA / Morquio syndrome
GALNT3	Hyperphosphatemic familial tumoral calcinosis
GALT	Galactosemia
GAMT	Guanidinoacetate methyltransferase deficiency
GATM	Arginine:glycine amidinotransferase deficiency
GBA	Gaucher disease
GBE1	Glycogen storage disease, type IV / Adult polyglucosan body disease
GCDH	Glutaric acidemia, type I
GCH1	GCH1-related disorders
GDF5	GDF5-related disorders
GFM1	Combined oxidative phosphorylation deficiency 1
GHR	Laron syndrome
GJB1	Charcot-Marie-Tooth disease, type 1X
GJB2	Nonsyndromic hearing loss and deafness (DFNB) 1
GLA	Fabry disease, X-linked
GLB1	GLB1-related disorders

Gene	Disease Name
GLDC	Glycine encephalopathy / Nonketotic hyperglycinemia
GLE1	Lethal congenital contracture syndrome 1
GNE	GNE myopathy
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis II and mucolipidosis III alpha/beta
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D
GORAB	Geroderma osteodysplastica
GP1BA	Bernard-Soulier syndrome, type A
GP9	Bernard-Soulier syndrome, type C
GRHPR	Primary hyperoxaluria, type II
GRIP1	Fraser syndrome, type 3
GSS	Glutathione synthetase deficiency
GUCY2D	Leber congenital amaurosis 1
GUSB	Mucopolysaccharidosis, type VII / Sly syndrome
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency
HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
HADHB	Mitochondrial trifunctional protein deficiency, HADHB-related
HAMP	Hereditary hemochromatosis, type 2B
HAX1	Congenital neutropenia, HAX1-related
HBA1/HBA2	Alpha-thalassemia
HBB	Beta hemoglobinopathies
HCFC1	Methylmalonic aciduria and homocystinuria, cblX type
HEXA	Tay-Sachs disease

Gene	Disease Name
HEXB	Sandhoff disease
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C
HJV	Hereditary hemochromatosis, type 2
HLCS	Holocarboxylase synthetase deficiency
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency
HMOX1	Heme oxygenase 1 deficiency
HOGA1	Primary hyperoxaluria, type III
HPD	Tyrosinemia, type III
HPRT1	HPRT1-related disorders
HPS1	Hermansky-Pudlak syndrome, type 1
HPS3	Hermansky-Pudlak syndrome, type 3
HPS4	Hermansky-Pudlak syndrome, type 4
HPS5	Hermansky-Pudlak syndrome, type 5
HPS6	Hermansky-Pudlak syndrome, type 6
HSD17B10	HSD10 disease
HSD17B3	17-beta-hydroxysteroid dehydrogenase deficiency, type III
HSD17B4	D-bifunctional protein deficiency
HSD3B2	3-beta-hydroxysteroid dehydrogenase deficiency, type II
HYAL1	Mucopolysaccharidosis, type IX / Hyaluronidase deficiency
HYLS1	Hydrolethalus syndrome
IDS	Mucopolysaccharidosis, type II / Hunter syndrome
IDUA	Mucopolysaccharidosis, type I / Hurler syndrome
IGHMBP2	IGHMBP2-related disorders
IKBKB	Severe combined immunodeficiency, IKBKB-related

Gene	Disease Name
IL2RG	Severe combined immunodeficiency, X-linked
IL7R	Severe combined immunodeficiency, IL7R-related
INPP5E	Joubert syndrome 1
INVS	Nephronophthisis 2
IQCB1	Senior-Loken syndrome 5
ITGA6	Junctional epidermolysis bullosa, ITGA6-related
ITGB3	ITGB3-related disorders
ITGB4	Junctional epidermolysis bullosa, ITGB4-related
IVD	Isovaleric acidemia
JAK3	Severe combined immunodeficiency, JAK3-related
KCNJ1	Bartter syndrome, type 2
KCNJ11	Familial hyperinsulinism, KCNJ11-related
L1CAM	L1 syndrome
LAMA2	LAMA2 muscular dystrophy
LAMA3	Junctional epidermolysis bullosa, LAMA3-related
LAMB3	Junctional epidermolysis bullosa, LAMB3-related
LAMC2	Junctional epidermolysis bullosa, LAMC2-related
LARGE1	Muscular dystrophy-dystroglycanopathy, type A, 6
LCA5	Leber congenital amaurosis 5
LDLR	Familial hypercholesterolemia, LDLR-related
LDLRAP1	Familial hypercholesterolemia, LDLRAP1-related
LHX3	Combined pituitary hormone deficiency, type 3
LIFR	Stuve-Wiedemann syndrome
LIG4	LIG4 syndrome
LIPA	Lysosomal acid lipase deficiency

Gene	Disease Name
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LOXHD1	Nonsyndromic hearing loss and deafness (DFNB) 77
LPL	Lipoprotein lipase deficiency
LRAT	LRAT-related disorders
LRP2	Donnai-Barrow syndrome
LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type
LYST	Chediak-Higashi syndrome
MAK	Retinitis pigmentosa 62
MAN2B1	Alpha-mannosidosis
MANBA	Beta-mannosidosis
MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency
MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency
MCEE	Methylmalonyl-CoA epimerase deficiency
MCOLN1	Mucolipidosis IV
MCPH1	Autosomal recessive primary microcephaly 1
MECR	MECR-related neurologic disorder
MED17	Infantile cerebral and cerebellar atrophy
MEFV	Familial Mediterranean fever
MESP2	Spondylothoracic dysostosis and spondylocostal dysostosis 2
MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related
MID1	X-linked Opitz G/BBB syndrome
MKKS	Bardet-Biedl syndrome 6
MKS1	MKS1-related disorders
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts

Gene	Disease Name
MLYCD	Malonyl-CoA decarboxylase deficiency
MMAA	Methylmalonic aciduria, MMAA-related
MMAB	Methylmalonic aciduria, MMAB-related
MMACHC	Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency
MMADHC	Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency
MMUT	Methylmalonic aciduria, MMUT-related
MOCS1	Molybdenum cofactor deficiency of complementation group A
MOCS2	Molybdenum cofactor deficiency of complementation group B
MPI	Congenital disorder of glycosylation, type Ib
MPL	Congenital amegakaryocytic thrombocytopenia
MPV17	MPV17-related mitochondrial DNA (mtDNA) maintenance defect
MRE11	Ataxia-telangiectasia-like disorder 1
MTHFR	Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency
MTM1	X-linked myotubular myopathy
MTR	Homocystinuria-megaloblastic anemia, cblG type
MTRR	Homocystinuria, type cblE
MTTP	Abetalipoproteinemia
MUSK	Congenital myasthenic syndrome, MUSK-related
MVK	Mevalonic aciduria / Hyper-IgD syndrome
MYO15A	Nonsyndromic hearing loss and deafness (DFNB) 3
MYO7A	Usher syndrome, type 1B
NAGA	Schindler disease
NAGLU	Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B
NAGS	N-acetylglutamate synthase deficiency
NBN	Nijmegen breakage syndrome

Gene	Disease Name
NCF2	Chronic granulomatous disease 2
NDRG1	Charcot-Marie-Tooth disease, type 4D
NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10
NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16
NDUFAF6	Mitochondrial complex I deficiency, nuclear type 17
NDUFS4	Mitochondrial complex I deficiency, nuclear type 1
NDUFS6	Mitochondrial complex I deficiency, nuclear type 9
NDUFS7	Mitochondrial complex I deficiency, nuclear type 3
NDUFV1	Mitochondrial complex I deficiency, nuclear type 4
NEB	Nemaline myopathy 2
NEU1	Sialidosis
NGLY1	Congenital disorder of deglycosylation, type 1
NPC1	Niemann-Pick disease, type C1
NPC2	Niemann-Pick disease, type C2
NPHP1	NPHP1 nephronophthisis-related ciliopathies
NPHP3	NPHP3 nephronophthisis-related ciliopathies
NPHP4	NPHP4 nephronophthisis-related ciliopathies
NPHS1	Steroid resistant nephrotic syndrome, type 1
NPHS2	Steroid-resistant nephrotic syndrome, type 2
NR0B1	X-linked congenital adrenal hypoplasia
NR2E3	Enhanced S-cone syndrome
NSMCE3	Lung disease, immunodeficiency, and chromosome breakage syndrome (LICS)
NTRK1	Congenital insensitivity to pain with anhidrosis

Gene	Disease Name
OAT	Ornithine aminotransferase deficiency
OCA2	Oculocutaneous albinism, type II
OCRL	Lowe syndrome, X-linked
OPA3	3-methylglutaconic aciduria, type III / Costeff syndrome
OSTM1	Osteopetrosis, OSTM1-related
OTC	Ornithine transcarbamylase deficiency, X-linked
OTOA	Nonsyndromic hearing loss and deafness (DFNB) 22
OTOF	Nonsyndromic hearing loss and deafness (DFNB) 9
P3H1	Osteogenesis imperfecta, type VIII
PAH	Phenylalanine hydroxylase deficiency
PANK2	Pantothenate kinase-associated neurodegeneration
PC	Pyruvate carboxylase deficiency
PCBD1	Pterin-4 alpha-carbinolamine dehydratase (PCD) deficiency
PCCA	Propionic acidemia, PCCA-related
PCCB	Propionic acidemia, PCCB-related
PCDH15	Usher syndrome, type 1F
PCNT	Microcephalic osteodysplastic primordial dwarfism, type II
PDHA1	Pyruvate dehydrogenase E1-alpha deficiency
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PEPD	Prolidase deficiency
PET100	Mitochondrial complex IV deficiency, nuclear type 12
PEX1	Zellweger spectrum disorders, PEX1-related
PEX10	Zellweger spectrum disorders, PEX10-related
PEX12	Zellweger spectrum disorders, PEX12-related
PEX13	Zellweger spectrum disorder, PEX13-related
PEX16	Zellweger spectrum disorder, PEX16-related

Gene	Disease Name
PEX2	Zellweger spectrum disorders, PEX2-related
PEX26	Zellweger spectrum disorders, PEX26-related
PEX5	Zellweger spectrum disorder, PEX5-related
PEX6	Zellweger spectrum disorders, PEX6-related
PEX7	Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease, type VII
PGM3	PGM3-congenital disorder of glycosylation / Immunodeficiency 23
PHGDH	Phosphoglycerate dehydrogenase deficiency
PHKB	Glycogen storage disease, type IXb
PHKG2	Glycogen storage disease, type IXc
PHYH	Refsum disease
PIGN	PIGN-related disorders
PJVK	Nonsyndromic hearing loss and deafness (DFNB) 59
PKHD1	Autosomal recessive polycystic kidney disease
PLA2G6	PLA2G6-associated neurodegeneration
PLCE1	Steroid-resistant nephrotic syndrome, type 3
PLEKHG5	PLEKHG5-related disorders
PLOD1	PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome
PLP1	PLP1-related disorders
PMM2	Congenital disorder of glycosylation, type Ia
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POLG	POLG-related disorders
POLH	Xeroderma pigmentosum, variant type (XP-V)
POMGNT1	Muscular dystrophy-dystroglycanopathy, type A, 3
POMT1	Muscular dystrophy-dystroglycanopathy, type A, 1
POMT2	Muscular dystrophy-dystroglycanopathy, type A, 2
POR	Cytochrome P450 oxidoreductase deficiency
POU1F1	Combined or isolated pituitary hormone deficiency, type 1

Gene	Disease Name
PPT1	Neuronal ceroid lipofuscinosis, PPT1-related
PRCD	Retinitis pigmentosa 36
PRDM5	Brittle cornea syndrome 2
PRF1	Familial hemophagocytic lymphohistiocytosis 2
PROP1	Combined pituitary hormone deficiency, type 2
PRPS1	PRPS1-related disorders
PSAP	Metachromatic leukodystrophy due to saposin B deficiency
PTPRC	Severe combined immunodeficiency, PTPRC-related
PTS	6-pyruvoyl-tetrahydropterin synthase deficiency
PUS1	Myopathy, lactic acidosis, and sideroblastic anemia
PYGM	Glycogen storage disease, type V
QDPR	Dihydropteridine reductase (DHPR) deficiency
RAB23	Carpenter syndrome
RAG1	Severe combined immunodeficiency, RAG1-related
RAG2	Severe combined immunodeficiency, RAG2-related
RAPSN	Congenital myasthenic syndrome, RAPSN-related
RARS2	Pontocerebellar hypoplasia, type 6
RDH12	Leber congenital amaurosis 13
RLBP1	RLBP1-related retinopathies
RMRP	Cartilage-hair hypoplasia
RNASEH2A	Aicardi-Goutieres syndrome 4
RNASEH2B	Aicardi-Goutieres syndrome 2
RNASEH2C	Aicardi-Goutieres syndrome 3
RP2	Retinitis pigmentosa 2
RPE65	Leber congenital amaurosis 2
RPGR	Retinitis pigmentosa 3
RPGRIP1L	Ciliopathies, RPGRIP1L-related
RS1	Juvenile retinoschisis, X-linked
RTEL1	Dyskeratosis congenita, RTEL1-related
RXYLT1	Muscular dystrophy-dystroglycanopathy, type A, 10

Gene	Disease Name
RYR1	RYR1-related disorders
SACS	Spastic ataxia, Charlevoix-Saguenay type
SAMD9	Normophosphatemic familial tumoral calcinosis
SAMHD1	Aicardi-Goutieres syndrome 5
SBDS	Shwachman-Diamond syndrome
SCARB2	Action myoclonus renal failure syndrome
SCO2	Mitochondrial complex IV deficiency, nuclear type 2
SEC23B	Congenital dyserythropoietic anemia, type II
SEPSECS	Progressive cerebello-cerebral atrophy
SERPINA1	Alpha-1 antitrypsin deficiency
SGCA	Limb-girdle muscular dystrophy, type 3
SGCB	Limb-girdle muscular dystrophy, type 4
SGCD	Limb-girdle muscular dystrophy, type 6
SGCG	Limb-girdle muscular dystrophy, type 5
SGSH	Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A
SKIC2	Trichohepatoenteric syndrome 2
SLC12A1	Bartter syndrome, type 1
SLC12A3	Gitelman syndrome
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy
SLC17A5	Free sialic acid storage disorders
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome
SLC19A3	Biotin-thiamine-responsive basal ganglia disease
SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrin deficiency / Citrullinemia, type II
SLC25A15	Ornithine translocase deficiency
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC26A2	Skeletal dysplasia, SLC26A2-related
SLC26A3	Congenital secretory chloride diarrhea 1
SLC26A4	Pendred syndrome
SLC27A4	Ichthyosis prematurity syndrome
SLC35A3	Arthrogryposis, mental retardation, and seizures

Gene	Disease Name
SLC37A4	Glycogen storage disease, type Ib / IIw
SLC38A8	Foveal hypoplasia 2
SLC39A4	Acrodermatitis enteropathica
SLC45A2	Oculocutaneous albinism, type IV
SLC4A11	Corneal dystrophy and perceptive deafness syndrome
SLC4A4	Renal tubular acidosis
SLC5A5	Thyroid dyshormonogenesis 1
SLC6A8	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome
SLC7A7	Lysinuric protein intolerance
SMARCAL1	Schimke immunoosseous dysplasia
SMN1	Spinal muscular atrophy
SMPD1	Niemann-Pick disease, types A/B
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma (CEDNIK) syndrome
SPG11	SPG11-related disorders
SPR	Sepiapterin reductase deficiency
SRD5A2	5-alpha-reductase deficiency
ST3GAL5	Amish infantile epilepsy syndrome
STAR	Lipoid congenital adrenal hyperplasia
STX11	Familial hemophagocytic lymphohistiocytosis 4
STXBP2	Familial hemophagocytic lymphohistiocytosis 5
SUMF1	Multiple sulfatase deficiency
SUOX	Isolated sulfite oxidase deficiency
SURF1	SURF1-related disorders
SYNE4	Nonsyndromic hearing loss and deafness (DFNB) 76
TAFAZZIN (formerly TAZ)	Barth syndrome
TANGO2	Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration
TAT	Tyrosinemia, type II
TBCD	Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum
TBCE	TBCE-related disorders

Gene	Disease Name
TCIRG1	Osteopetrosis, infantile malignant, TCIRG1-related
TCN2	Transcobalamin II deficiency
TECPR2	TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability
TERT	Dyskeratosis congenita spectrum disorders
TF	Atransferrinemia
TFR2	Hereditary hemochromatosis, type 3
TG	Thyroid dyshormonogenesis 3
TGM1	Autosomal recessive congenital ichthyosis
TH	Tyrosine hydroxylase deficiency
TJP2	Progressive familial intrahepatic cholestasis 4
TK2	TK2-related mitochondrial disorders
TMC1	Nonsyndromic hearing loss and deafness (DFNB) 7
TMEM216	Joubert syndrome 2
TMEM67	TMEM67-related disorders
TMPRSS3	Nonsyndromic hearing loss and deafness (DFNB) 8
TNXB	TNXB-related classical-like Ehlers-Danlos syndrome
TPO	Thyroid dyshormonogenesis 2A
TPP1	Neuronal ceroid lipofuscinosis, TPP1-related
TREX1	TREX1-related disorders
TRIM32	TRIM32-related disorders
TRIM37	Mulibrey nanism
TRMU	Acute infantile liver failure
TSEN2	Pontocerebellar hypoplasia, type 2B
TSEN54	Pontocerebellar hypoplasia, types 4 and 2A
TSFM	Combined oxidative phosphorylation deficiency 3

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Gene	Disease Name
TSHB	Congenital hypothyroidism, TSHB-related
TSHR	Congenital hypothyroidism, TSHR-related
TTC37	Trichohepatoenteric syndrome 1
TTC8	Bardet-Biedl syndrome 8
TTPA	Ataxia with isolated vitamin E deficiency
TULP1	TULP1-related disorders
TYMP	Mitochondrial DNA depletion syndrome 1, MNGIE type
TYR	Oculocutaneous albinism, type I
TYRP1	Oculocutaneous albinism, type III
UBA1	X-linked infantile spinal muscular atrophy
UBR1	Johanson-Blizzard syndrome
UNC13D	Familial hemophagocytic lymphohistiocytosis 3
UPB1	Beta-ureidopropionase deficiency
USH1C	Usher syndrome, type 1C
USH2A	Usher syndrome, type 2A
VDR	Vitamin D-resistant rickets, type 2A
VLDLR	VLDLR-associated cerebellar hypoplasia
VPS11	Hypomyelinating leukodystrophy 12
VPS13A	Choreoacanthocytosis
VPS13B	Cohen syndrome
VPS45	Severe congenital neutropenia 5
VPS53	Pontocerebellar hypoplasia, type 2E
VRK1	Pontocerebellar hypoplasia, type 1A
VSX2	Microphthalmia / Anophthalmia
WAS	Wiskott-Aldrich syndrome, X-linked
WNT10A	Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome
WRN	Werner syndrome
WWOX	WWOX deficiency
XPA	Xeroderma pigmentosum, group A
XPC	Xeroderma pigmentosum, group C
ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2
ZFYVE26	Spastic paraplegia, type 15
ZIC3	X-linked heterotaxy-1
ZNF469	Brittle cornea syndrome 1