

QHerit[®] carrier screening

Right-sized solutions to help patients navigate their family planning journey

As part of our commitment to providing clinically relevant, right-sized solutions, our **QHerit[®] product portfolio** offers medically appropriate and comprehensive insights to help you understand your patient's genetic risks. Empowered with that insight, together you can make informed decisions about family planning.

An ideal panel for each patient^a

QHerit panel options					
Panel size:	22 diseases^b 24 genes	112 diseases^b 113 genes	198 diseases^b 199 genes	445 diseases^b 446 genes	611 diseases^b 612 genes
Screens for:	The most common diseases including cystic fibrosis, spinal muscular atrophy, fragile X, and Tay-Sachs	Recommended genes listed by ACMG tier 3, which includes the 24 genes in the smaller panel	Builds on the 113-gene panel with 86 additional genes	An additional 247 genes included plus the 199-gene panel	Our most comprehensive panel with 166 additional genes offering the greatest insights
Test code:	94372	Female: 14232 Male: 14227	Female: 14230 Male: 14231	Female: 13832 Male: 13831	Female: 14228 Male: 14229

QHerit 22 diseases^b 24 genes

Test code: 94372

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Alpha-thalassemia	<i>HBA1/HBA2</i>	Familial dysautonomia	<i>ELP1</i> (AKA: <i>IKBKAP</i>)	Joubert syndrome 2	<i>TMEM216</i>
Beta hemoglobinopathies	<i>HBB</i>	Familial hyperinsulinism, ABCC8-related	<i>ABCC8</i>	Maple syrup urine disease, type 1B	<i>BCKDHB</i>
Bloom syndrome	<i>BLM</i>	Fragile X syndrome ^c	<i>FMR1</i>	Mucopolidosis, type IV	<i>MCOLN1</i>
Canavan disease	<i>ASPA</i>	Fukuyama congenital muscular dystrophy (Walker-Warburg)	<i>FKTN</i>	Nemaline myopathy 2	<i>NEB</i>
Cystic fibrosis	<i>CFTR</i>	Gaucher disease	<i>GBA</i>	Niemann-Pick disease, types A/B	<i>SMPD1</i>
Dihydrofolate reductase deficiency	<i>DLD</i>	Glycogen storage disease, type Ia	<i>G6PC</i> (AKA: <i>G6PC1</i>)	Spinal muscular atrophy	<i>SMN1</i>
Fanconi anemia, Group C	<i>FANCC</i>			Tay-Sachs disease	<i>HEXA</i>
				Usher syndrome, type 1F	<i>PCDH15</i>
				Usher syndrome, type 3A	<i>CLRN1</i>

^a QHerit panels are screening tests. QHerit does not diagnose a disease or disorder.

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

Important information

QHerit[®] 22, QHerit 112, QHerit 198, QHerit 445, and QHerit 611 are carrier screening tests, and they screen for variations in genes linked to certain health disorders that can be passed from parents to children. QHerit 22 screens for 24 genes, QHerit 112 screens for 113 genes, QHerit 198 screens for 199 genes, QHerit 445 screens for 446 genes, and QHerit 611 screens for 612 genes. For a full list of genes that each panel in the QHerit family screens, visit QHerit.com. If the results from any panel in the QHerit family suggest that a patient may be a carrier of a gene variation that can cause a health disorder in her offspring, it is recommended that her reproductive partner be offered genetic screening, and that genetic counseling be provided. Pregnancy management decisions should not be based on the results of these screening tests alone. As with any test, there may be false positives or false negatives. The positive predictive value of the screening test varies by genetic variation, and may be lower for rare conditions. Each panel in the QHerit family is a laboratory-developed test that has been developed and validated pursuant to the Clinical Laboratory Improvements Amendments of 1988 (CLIA) and, as such, it has not been reviewed by FDA.



QHerit 112 diseases^b 113 genes

Test codes: Female (113 diseases): 14232 | Male (97 diseases): 14227

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
3-methylcrotonyl-CoA carboxylase 2 deficiency	<i>MCCC2</i>	Congenital myasthenic syndrome, CHRNE-related	<i>CHRNE</i>	Hereditary fructose intolerance	<i>ALDOB</i>
Achromatopsia, CNGB3-related	<i>CNGB3</i>	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome ^c	<i>SLC6A8</i>	Hermansky-Pudlak syndrome, type 1	<i>HPS1</i>
Adrenoleukodystrophy, X-linked ^c	<i>ABCD1</i>	Cystic fibrosis	<i>CFTR</i>	Hermansky-Pudlak syndrome, type 3	<i>HPS3</i>
Aicardi-Goutieres syndrome 2	<i>RNASEH2B</i>	Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Homocystinuria, CBS-related	<i>CBS</i>
Alpha-thalassemia	<i>HBA1/HBA2</i>	Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Hypophosphatasia	<i>ALPL</i>
Argininosuccinic aciduria	<i>ASL</i>	Donnai-Barrow syndrome	<i>LRP2</i>	Joubert syndrome 2	<i>TMEM216</i>
Aspartylglycosaminuria	<i>AGA</i>	Duchenne/Becker muscular dystrophy, X-linked ^c	<i>DMD</i>	Joubert syndrome 3	<i>AHI1</i>
Atransferrinemia	<i>TF</i>	Dystrophic epidermolysis bullosa, COL7A1-related	<i>COL7A1</i>	Joubert syndrome 9	<i>CC2D2A</i>
Autoimmune polyglandular syndrome, type 1	<i>AIRE</i>	Ellis-van Creveld syndrome	<i>EVC2</i>	Juvenile retinoschisis, X-linked ^c	<i>RS1</i>
Autosomal recessive polycystic kidney disease	<i>PKHD1</i>	ERCC2-related conditions	<i>ERCC2</i>	L1 syndrome ^c	<i>L1CAM</i>
Autosomal recessive primary microcephaly 1	<i>MCPH1</i>	Fabry disease, X-linked ^c	<i>GLA</i>	Leber congenital amaurosis, CEP290-related / CEP290-related conditions	<i>CEP290</i>
Autosomal recessive spinocerebellar ataxia, type 10	<i>ANO10</i>	Factor IX deficiency / Hemophilia B ^c	<i>F9</i>	Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	<i>FKRP</i>
Bardet-Biedl syndrome 1	<i>BBS1</i>	Factor VIII deficiency / Hemophilia A ^c	<i>F8</i>	Maple syrup urine disease, type 1B	<i>BCKDHB</i>
Bardet-Biedl syndrome 2	<i>BBS2</i>	Familial dysautonomia	<i>ELP1</i>	Medium chain acyl-CoA dehydrogenase deficiency	<i>ACADM</i>
Beta hemoglobinopathies	<i>HBB</i>	Familial hemophagocytic lymphohistiocytosis 2	<i>PRF1</i>	Megalencephalic leukoencephalopathy with subcortical cysts	<i>MLC1</i>
Beta-ketothiolase deficiency	<i>ACAT1</i>	Familial hyperinsulinism, ABCC8-related	<i>ABCC8</i>	Metachromatic leukodystrophy, ARSA-related	<i>ARSA</i>
Biotinidase deficiency	<i>BTD</i>	Fanconi anemia, complementation group C	<i>FANCC</i>	Methylmalonic aciduria, MMUT-related	<i>MMUT</i>
Biotin-thiamine-responsive basal ganglia disease	<i>SLC19A3</i>	Fragile X syndrome	<i>FMR1</i>	Mevalonic aciduria / Hyper-IgD syndrome	<i>MVK</i>
Bloom syndrome	<i>BLM</i>	Fraser syndrome, type 3	<i>GRIP1</i>	Mitochondrial complex IV deficiency, nuclear type 2	<i>SCO2</i>
Canavan disease	<i>ASPA</i>	Fragile X syndrome ^c	<i>AFF2</i>	Mucopolysaccharidosis, type I / Hurler syndrome	<i>IDUA</i>
Carnitine palmitoyltransferase II deficiency	<i>CPT2</i>	Friedreich ataxia	<i>FXN</i>	Myotonia congenita	<i>CLCN1</i>
Cerebrotendinous xanthomatosis	<i>CYP27A1</i>	Fukuyama congenital muscular dystrophy	<i>FKTN</i>	Nemaline myopathy 2	<i>NEB</i>
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	<i>MMACHC</i>	Galactosemia	<i>GALT</i>	Niemann-Pick disease, types A/B	<i>SMPD1</i>
Congenital adrenal insufficiency, CYP11A1-related	<i>CYP11A1</i>	Gaucher disease	<i>GBA</i>	Nonsyndromic hearing loss and deafness (DFNB) 1	<i>GJB2</i>
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	<i>CYP21A2</i>	Glycogen storage disease, type Ia	<i>G6PC1</i>		
Congenital disorder of glycosylation, type Ia	<i>PMM2</i>	Glycogen storage disease, type Ib / IIw	<i>SLC37A4</i>		
Congenital hydrocephalus 1	<i>CCDC88C</i>	Glycogen storage disease, type II / Pompe disease	<i>GAA</i>		
		Glycogen storage disease, type IV / Adult polyglucosan body disease	<i>GBE1</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 112 diseases^b 113 genes (continued)

Test codes: Female (113 diseases): 14232 | Male (97 diseases): 14227

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Oculocutaneous albinism, type I	<i>TYR</i>	Retinitis pigmentosa 59	<i>DHDDS</i>	Tyrosinemia, type I	<i>FAH</i>
Oculocutaneous albinism, type II	<i>OCA2</i>	Schindler disease	<i>NAGA</i>	Usher syndrome, type 1F	<i>PCDH15</i>
Ornithine transcarbamylase deficiency, X-linked ^c	<i>OTC</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	<i>DYNC2H1</i>	Usher syndrome, type 2A	<i>USH2A</i>
Pendred syndrome	<i>SLC26A4</i>	Skeletal dysplasia, SLC26A2-related	<i>SLC26A2</i>	Usher syndrome, type 3A	<i>CLRN1</i>
Phenylalanine hydroxylase deficiency	<i>PAH</i>	Smith-Lemli-Opitz syndrome	<i>DHCR7</i>	Very long-chain acyl-CoA dehydrogenase deficiency	<i>ACADVL</i>
PLP1-related disorders ^c	<i>PLP1</i>	Spinal muscular atrophy	<i>SMN1</i>	Vitamin D-dependent rickets, type 1A	<i>CYP27B1</i>
POLG-related disorders	<i>POLG</i>	Steroid resistant nephrotic syndrome, type 1	<i>NPHS1</i>	Wilson disease	<i>ATP7B</i>
Pontocerebellar hypoplasia, type 6	<i>RARS2</i>	Surfactant dysfunction, ABCA3-related	<i>ABCA3</i>	X-linked congenital adrenal hypoplasia ^c	<i>NR0B1</i>
Primary hyperoxaluria, type I	<i>AGXT</i>	Tay-Sachs disease	<i>HEXA</i>	X-linked developmental disorders, ARX-related ^c	<i>ARX</i>
Retinitis pigmentosa 3 ^c	<i>RPGR</i>	TNXB-related classical-like Ehlers-Danlos syndrome	<i>TNXB</i>	X-linked Opitz G/BBB syndrome ^c	<i>MID1</i>
		Trimethylaminuria	<i>FMO3</i>	Xeroderma pigmentosum, group C ^c	<i>XPC</i>

QHerit 198 diseases^b 199 genes

Test codes: Female (198 diseases): 14230 | Male (179 diseases): 14231

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	<i>HMGL</i>	Alport syndrome, COL4A5-related, X-linked ^c	<i>COL4A5</i>	Biotin-thiamine-responsive basal ganglia disease	<i>SLC19A3</i>
3-methylcrotonyl-CoA carboxylase 1 deficiency	<i>MCCC1</i>	Argininosuccinic aciduria	<i>ASL</i>	Bloom syndrome	<i>BLM</i>
3-methylcrotonyl-CoA carboxylase 2 deficiency	<i>MCCC2</i>	Arthrogryposis, mental retardation, and seizures	<i>SLC35A3</i>	Canavan disease	<i>ASPA</i>
6-pyruvoyl-tetrahydropterin synthase deficiency	<i>PTS</i>	Aspartylglycosaminuria	<i>AGA</i>	Carnitine deficiency, systemic primary	<i>SLC22A5</i>
Abetalipoproteinemia	<i>MTPP</i>	Ataxia-telangiectasia	<i>ATM</i>	Carnitine palmitoyltransferase II deficiency	<i>CPT2</i>
Achromatopsia, CNGB3-related	<i>CNGB3</i>	Atransferrinemia	<i>TF</i>	Cartilage-hair hypoplasia	<i>RMRP</i>
Adenosine deaminase deficiency	<i>ADA</i>	Autoimmune polyglandular syndrome, type 1	<i>AIRE</i>	Cerebrotendinous xanthomatosis	<i>CYP27A1</i>
Adrenoleukodystrophy, X-linked ^c	<i>ABCD1</i>	Autosomal recessive congenital ichthyosis	<i>TGM1</i>	Citrullinemia, type I	<i>ASS1</i>
Agenesis of the corpus callosum with peripheral neuropathy	<i>SLC12A6</i>	Autosomal recessive polycystic kidney disease	<i>PKHD1</i>	Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	<i>MMACHC</i>
Aicardi-Goutieres syndrome 2	<i>RNASEH2B</i>	Autosomal recessive primary microcephaly 1	<i>MCPH1</i>	Combined pituitary hormone deficiency, type 2	<i>PROP1</i>
Alpha-mannosidosis	<i>MAN2B1</i>	Autosomal recessive spinocerebellar ataxia, type 10	<i>ANO10</i>	Congenital adrenal insufficiency, CYP11A1-related	<i>CYP11A1</i>
Alpha-thalassemia	<i>HBA1/HBA2</i>	Bardet-Biedl syndrome 1	<i>BBS1</i>	Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	<i>CYP21A2</i>
Alport syndrome, COL4A3-related	<i>COL4A3</i>	Bardet-Biedl syndrome 2	<i>BBS2</i>	Congenital amegakaryocytic thrombocytopenia	<i>MPL</i>
Alport syndrome, COL4A4-related	<i>COL4A4</i>	Bardet-Biedl syndrome 10	<i>BBS10</i>		
		Beta hemoglobinopathies	<i>HBB</i>		
		Beta-ketothiolase deficiency	<i>ACAT1</i>		
		Biotinidase deficiency	<i>BTD</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 198 diseases^b 199 genes (continued)

Test codes: Female (198 diseases): 14230 | Male (179 diseases): 14231

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Congenital disorder of glycosylation, type Ia	<i>PMM2</i>	Fukuyama congenital muscular dystrophy	<i>FKTN</i>	Krabbe disease	<i>GALC</i>
Congenital hydrocephalus 1	<i>CCDC88C</i>	Galactosemia	<i>GALT</i>	L1 syndrome ^c	<i>L1CAM</i>
Congenital myasthenic syndrome, CHRNE-related	<i>CHRNE</i>	Gaucher disease	<i>GBA</i>	LAMA2 muscular dystrophy	<i>LAMA2</i>
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome ^c	<i>SLC6A8</i>	GLB1-related disorders	<i>GLB1</i>	Leber congenital amaurosis, CEP290-related / CEP290-related conditions	<i>CEP290</i>
Cystic fibrosis	<i>CFTR</i>	Glutaric acidemia, type I	<i>GCDH</i>	Lethal congenital contracture syndrome 1	<i>GLE1</i>
Cystinosis	<i>CTNS</i>	Glycine encephalopathy, AMT-related	<i>AMT</i>	Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	<i>FKRP</i>
D-bifunctional protein deficiency	<i>HSD17B4</i>	Glycine encephalopathy / Nonketotic hyperglycinemia	<i>GLDC</i>	Limb-girdle muscular dystrophy, type 2A	<i>CAPN3</i>
Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Glycogen storage disease, type Ia	<i>G6PC1</i>	Limb-girdle muscular dystrophy, type 3	<i>SGCA</i>
Donnai-Barrow syndrome	<i>LRP2</i>	Glycogen storage disease, type Ib / IIw	<i>SLC37A4</i>	Limb-girdle muscular dystrophy, type 4	<i>SGCB</i>
Duchenne/Becker muscular dystrophy, X-linked ^c	<i>DMD</i>	Glycogen storage disease, type II / Pompe disease	<i>GAA</i>	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	<i>HADHA</i>
Dyskeratosis congenita, RTEL1-related	<i>RTEL1</i>	Glycogen storage disease, type III	<i>AGL</i>	Lysinuric protein intolerance	<i>SLC7A7</i>
Dystrophic epidermolysis bullosa, COL7A1-related	<i>COL7A1</i>	Glycogen storage disease, type IV / Adult polyglucosan body disease	<i>GBE1</i>	Maple syrup urine disease, type 1A	<i>BCKDHA</i>
Ellis-van Creveld syndrome	<i>EVC2</i>	GNE myopathy	<i>GNE</i>	Maple syrup urine disease, type 1B	<i>BCKDHB</i>
ERCC2-related conditions	<i>ERCC2</i>	GRACILE syndrome	<i>BCS1L</i>	Maple syrup urine disease, type 2	<i>DBT</i>
Fabry disease, X-linked ^c	<i>GLA</i>	Hereditary fructose intolerance	<i>ALDOB</i>	Medium chain acyl-CoA dehydrogenase deficiency	<i>ACADM</i>
Factor IX deficiency / Hemophilia B ^c	<i>F9</i>	Hermansky-Pudlak syndrome, type 1	<i>HPS1</i>	Menkes disease ^c	<i>ATP7A</i>
Factor XI deficiency / Hemophilia C	<i>F11</i>	Hermansky-Pudlak syndrome, type 3	<i>HPS3</i>	Megalencephalic leukoencephalopathy with subcortical cysts	<i>MLC1</i>
Factory VIII deficiency / Hemophilia A ^c	<i>F8</i>	Holocarboxylase synthetase deficiency	<i>HLCS</i>	Metachromatic leukodystrophy, ARSA-related	<i>ARSA</i>
Familial dysautonomia	<i>ELP1</i>	Homocystinuria, CBS-related	<i>CBS</i>	Methylmalonic aciduria, MMAA-related	<i>MMAA</i>
Familial hemophagocytic lymphohistiocytosis 2	<i>PRF1</i>	Hydrolethalus syndrome	<i>HYLS1</i>	Methylmalonic aciduria, MMAB-related	<i>MMAB</i>
Familial hyperinsulinism, ABCC8-related	<i>ABCC8</i>	Hypophosphatasia	<i>ALPL</i>	Methylmalonic aciduria, MMUT-related	<i>MMUT</i>
Familial hyperinsulinism, KCNJ11-related	<i>KCNJ11</i>	Infantile cerebral and cerebellar atrophy	<i>MED17</i>	Mevalonic aciduria / Hyper-IgD syndrome	<i>MVK</i>
Familial Mediterranean fever	<i>MEFV</i>	Isovaleric acidemia	<i>IVD</i>	Mitochondrial complex IV deficiency, nuclear type 2	<i>SCO2</i>
Fanconi anemia, complementation group A	<i>FANCA</i>	Joubert syndrome 2	<i>TMEM216</i>	Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	<i>LRPPRC</i>
Fanconi anemia, complementation group C	<i>FANCC</i>	Joubert syndrome 3	<i>AHI1</i>		
Fragile X syndrome ^c	<i>FMR1</i>	Joubert syndrome 9	<i>CC2D2A</i>		
Fragile XE syndrome ^c	<i>AFF2</i>	Junctional epidermolysis bullosa, LAMA3-related	<i>LAMA3</i>		
Fraser syndrome, type 3	<i>GRIP1</i>	Junctional epidermolysis bullosa, LAMB3-related	<i>LAMB3</i>		
Friedreich ataxia	<i>FXN</i>	Junctional epidermolysis bullosa, LAMC2-related	<i>LAMC2</i>		
		Juvenile retinoschisis, X-linked ^c	<i>RS1</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 198 diseases^b 199 genes (continued)

Test codes: Female (198 diseases): 14230 | Male (179 diseases): 14231

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Mucopolipidosis II and mucopolipidosis III alpha/beta	<i>GNPTAB</i>	Oculocutaneous albinism, type II	<i>OCA2</i>	Tay-Sachs disease	<i>HEXA</i>
Mucopolipidosis IV	<i>MCOLN1</i>	Ornithine transcarbamylase deficiency, X-linked ^c	<i>OTC</i>	TNXB-related classical-like Ehlers-Danlos syndrome	<i>TNXB</i>
Mucopolysaccharidosis, type I / Hurler syndrome	<i>IDUA</i>	Pendred syndrome	<i>SLC26A4</i>	Trimethylaminuria	<i>FM03</i>
Mucopolysaccharidosis, type II / Hunter syndrome ^c	<i>IDS</i>	Phenylalanine hydroxylase deficiency	<i>PAH</i>	Tyrosine hydroxylase deficiency	<i>TH</i>
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	<i>SGSH</i>	PLP1-related disorders	<i>PLP1</i>	Tyrosinemia, type I	<i>FAH</i>
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	<i>NAGLU</i>	POLG-related disorders	<i>POLG</i>	Tyrosinemia, type II	<i>TAT</i>
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	<i>HGSNAT</i>	Pontocerebellar hypoplasia, type 6	<i>RARS2</i>	Usher syndrome, type 1B	<i>MYO7A</i>
Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	<i>GNS</i>	Primary hyperoxaluria, type I	<i>AGXT</i>	Usher syndrome, type 1C	<i>USH1C</i>
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	<i>ARSB</i>	Propionic acidemia, PCCA-related	<i>PCCA</i>	Usher syndrome, type 1D	<i>CDH23</i>
Myotonia congenita	<i>CLCN1</i>	Propionic acidemia, PCCB-related	<i>PCCB</i>	Usher syndrome, type 1F	<i>PCDH15</i>
Nemaline myopathy 2	<i>NEB</i>	Pyruvate carboxylase deficiency	<i>PC</i>	Usher syndrome, type 2A	<i>USH2A</i>
Neuronal ceroid lipofuscinosis, CLN3-related	<i>CLN3</i>	Retinitis pigmentosa 3 ^c	<i>RPGR</i>	Usher syndrome, type 3A	<i>CLRN1</i>
Neuronal ceroid lipofuscinosis, CLN5-related	<i>CLN5</i>	Retinitis pigmentosa 59	<i>DHDDS</i>	Very long-chain acyl-CoA dehydrogenase deficiency	<i>ACADVL</i>
Neuronal ceroid lipofuscinosis, CLN6-related	<i>CLN6</i>	Rhizomelic chondrodysplasia punctata, type 1	<i>PEX7</i>	Vitamin D-dependent rickets, type 1A	<i>CYP27B1</i>
Neuronal ceroid lipofuscinosis, CLN8-related	<i>CLN8</i>	Sandhott disease	<i>HEXB</i>	Wilson disease	<i>ATP7B</i>
Neuronal ceroid lipofuscinosis, PPT1-related	<i>PPT1</i>	Schindler disease	<i>NAGA</i>	X-linked congenital adrenal hypoplasia ^c	<i>NR0B1</i>
Neuronal ceroid lipofuscinosis, TPP1-related	<i>TPP1</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	<i>DYNC2H1</i>	X-linked developmental disorders, ARX-related ^c	<i>ARX</i>
Niemann-Pick disease, types A/B	<i>SMPD1</i>	Sjögren-Larsson syndrome	<i>ALDH3A2</i>	X-linked Opitz G/BBB syndrome ^c	<i>MID1</i>
Niemann-Pick disease, type C1	<i>NPC1</i>	Skeletal dysplasia, SLC26A2-related	<i>SLC26A2</i>	Xeroderma pigmentosum, group C	<i>XPC</i>
Nijmegen breakage syndrome	<i>NBN</i>	Smith-Lemli-Opitz syndrome	<i>DHCR7</i>	Zellweger spectrum disorders, PEX1-related	<i>PEX1</i>
Nonsyndromic hearing loss and deafness (DFNB) 1	<i>GJB2</i>	Spastic ataxia, Charlevoix-Saguenay type	<i>SACS</i>	Zellweger spectrum disorders, PEX2-related	<i>PEX2</i>
Nonsyndromic hearing loss and deafness (DFNB) 77	<i>LOXHD1</i>	Spinal muscular atrophy	<i>SMN1</i>	Zellweger spectrum disorders, PEX6-related	<i>PEX6</i>
Oculocutaneous albinism, type I	<i>TYR</i>	Spondylothoracic dysostosis and spondylocostal dysostosis 2	<i>MESP2</i>		
		Steroid resistant nephrotic syndrome, type 1	<i>NPHS1</i>		
		Steroid-resistant nephrotic syndrome, type 2	<i>NPHS2</i>		
		Surfactant dysfunction, ABCA3-related	<i>ABCA3</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
3-beta-hydroxysteroid dehydrogenase deficiency, type II	<i>HSD3B2</i>	Alport syndrome, COL4A3-related	<i>COL4A3</i>	Bernard-Soulier syndrome, type C	<i>GP9</i>
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	<i>HMGCL</i>	Alport syndrome, COL4A4-related	<i>COL4A4</i>	Beta hemoglobinopathies	<i>HBB</i>
3-hydroxyacyl-CoA dehydrogenase deficiency	<i>HADH</i>	Alport syndrome, COL4A5-related, X-linked ^c	<i>COL4A5</i>	Beta-ketothiolase deficiency	<i>ACAT1</i>
3-methylcrotonyl-CoA carboxylase 1 deficiency	<i>MCCC1</i>	Alstrom syndrome	<i>ALMS1</i>	Beta-ureidopropionase deficiency	<i>UPB1</i>
3-methylcrotonyl-CoA carboxylase 2 deficiency	<i>MCCC2</i>	Argininemia	<i>ARG1</i>	Bilateral frontoparietal polymicrogyria	<i>ADGRG1</i>
3-methylglutaconic aciduria, type III / Costett syndrome	<i>OPA3</i>	Argininosuccinic aciduria	<i>ASL</i>	Biotinidase deficiency	<i>BTB</i>
5-alpha-reductase deficiency	<i>SRD5A2</i>	Arthrogryposis, mental retardation, and seizures	<i>SLC35A3</i>	Biotin-thiamine-responsive basal ganglia disease	<i>SLC19A3</i>
6-pyruvoyl-tetrahydropterin synthase deficiency	<i>PTS</i>	Aromatase deficiency	<i>CYP19A1</i>	Bloom syndrome	<i>BLM</i>
17-beta-hydroxysteroid dehydrogenase deficiency, type III	<i>HSD17B3</i>	Asparagine synthetase deficiency	<i>ASNS</i>	Canavan disease	<i>ASPA</i>
Abetalipoproteinemia	<i>MTTP</i>	Aspartylglycosaminuria	<i>AGA</i>	Carbamoyl phosphate synthetase I deficiency	<i>CPS1</i>
Acrodermatitis enteropathica	<i>SLC39A4</i>	Ataxia-telangiectasia	<i>ATM</i>	Carnitine-acylcarnitine translocase deficiency	<i>SLC25A20</i>
Achromatopsia, CNGB3-related	<i>CNGB3</i>	Ataxia-telangiectasia-like disorder 1	<i>MRE11</i>	Carnitine deficiency, systemic primary	<i>SLC22A5</i>
Action myoclonus renal failure syndrome	<i>SCARB2</i>	Ataxia with isolated vitamin E deficiency	<i>TTPA</i>	Carnitine palmitoyltransferase I deficiency	<i>CPT1A</i>
Acute infantile liver failure	<i>TRMU</i>	Atransferrinemia	<i>TF</i>	Carnitine palmitoyltransferase II deficiency	<i>CPT2</i>
Adenosine deaminase deficiency	<i>ADA</i>	Autoimmune polyglandular syndrome, type 1	<i>AIRE</i>	Carpenter syndrome	<i>RAB23</i>
Adrenoleukodystrophy, X-linked ^c	<i>ABCD1</i>	Autosomal recessive congenital ichthyosis	<i>TGM1</i>	Cartilage-hair hypoplasia	<i>RMRP</i>
Agenesis of the corpus callosum with peripheral neuropathy	<i>SLC12A6</i>	Autosomal recessive polycystic kidney disease	<i>PKHD1</i>	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	<i>ERCC6</i>
Aicardi-Goutieres syndrome 2	<i>RNASEH2B</i>	Autosomal recessive primary microcephaly 1	<i>MCPH1</i>	Cerebrotendinous xanthomatosis	<i>CYP27A1</i>
Aicardi-Goutieres syndrome 3	<i>RNASEH2C</i>	Autosomal recessive spinocerebellar ataxia, type 10	<i>ANO10</i>	Charcot-Marie-Tooth disease, type 1X ^c	<i>GJB1</i>
Aicardi-Goutieres syndrome 4	<i>RNASEH2A</i>	Bardet-Biedl syndrome 1	<i>BBS1</i>	Charcot-Marie-Tooth disease, type 4D	<i>NDRG1</i>
Aicardi-Goutieres syndrome 5	<i>SAMHD1</i>	Bardet-Biedl syndrome 2	<i>BBS2</i>	Chediak-Higashi syndrome	<i>LYST</i>
Alpha-1 antitrypsin deficiency	<i>SERPINA1</i>	Bardet-Biedl syndrome 4	<i>BBS4</i>	Choreoacanthocytosis	<i>VPS13A</i>
Alpha-mannosidosis	<i>MAN2B1</i>	Bardet-Biedl syndrome 6	<i>MKKS</i>	Choroideremia, X-linked ^c	<i>CHM</i>
Alpha-thalassemia	<i>HBA1/ HBA2</i>	Bardet-Biedl syndrome 7	<i>BBS7</i>	Chronic granulomatous disease 4	<i>CYBA</i>
Alpha-thalassemia intellectual disability syndrome, X-linked ^c	<i>ATRX</i>	Bardet-Biedl syndrome 8	<i>TTC8</i>	Chronic granulomatous disease, X-linked ^c	<i>CYBB</i>
		Bardet-Biedl syndrome 9	<i>BBS9</i>	Ciliopathies, RPGRIP1 L-related	<i>RPGRIP1L</i>
		Bardet-Biedl syndrome 10	<i>BBS10</i>	Citrin deficiency / Citrullinemia, type II	<i>SLC25A13</i>
		Bardet-Biedl syndrome 12	<i>BBS12</i>	Citrullinemia, type I	<i>ASS1</i>
		Bare lymphocyte syndrome, type II	<i>CIITA</i>		
		Barth syndrome ^c	<i>TAFAZZIN</i>		
		Bartter syndrome, type 4A	<i>BSND</i>		
		Bernard-Soulier syndrome, type A	<i>GP1BA</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes (continued)

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Cockayne syndrome, type A	<i>ERCC8</i>	Congenital myasthenic syndrome, CHRNE-related	<i>CHRNE</i>	Ethylmalonic encephalopathy	<i>ETHE1</i>
Cohen syndrome	<i>VPS13B</i>	Congenital myasthenic syndrome, COLQ-related	<i>COLQ</i>	Fabry disease, X-linked ^c	<i>GLA</i>
Combined malonic and methylmalonic aciduria	<i>ACSF3</i>	Congenital myasthenic syndrome, DOK7-related	<i>DOK7</i>	Factor IX deficiency / Hemophilia B ^c	<i>F9</i>
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	<i>MMACHC</i>	Congenital myasthenic syndrome, RAPSN-related	<i>RAPSN</i>	Factor XI deficiency / Hemophilia C	<i>F11</i>
Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	<i>MMADHC</i>	Congenital neutropenia, HAX1-related	<i>HAX1</i>	Factor VIII deficiency / Hemophilia A ^c	<i>F8</i>
Combined oxidative phosphorylation deficiency 1	<i>GFM1</i>	Corneal dystrophy and perceptive deafness syndrome	<i>SLC4A11</i>	Familial dysautonomia	<i>ELP1</i>
Combined oxidative phosphorylation deficiency 3	<i>TSMF</i>	Corticosterone methyloxidase deficiency	<i>CYP11B2</i>	Familial hemophagocytic lymphohistiocytosis 2	<i>PRF1</i>
Combined oxidative phosphorylation deficiency 6 ^c	<i>AIFM1</i>	CRB1-related retinal dystrophies	<i>CRB1</i>	Familial hemophagocytic lymphohistiocytosis 4	<i>STX11</i>
Combined pituitary hormone deficiency, type 2	<i>PROP1</i>	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome ^c	<i>SLC6A8</i>	Familial hemophagocytic lymphohistiocytosis 5	<i>STXBP2</i>
Combined pituitary hormone deficiency, type 3	<i>LHX3</i>	Cystic fibrosis	<i>CFTR</i>	Familial hypercholesterolemia, LDLR-related	<i>LDLR</i>
Congenital adrenal insufficiency, CYP11A1-related	<i>CYP11A1</i>	Cystinosis	<i>CTNS</i>	Familial hypercholesterolemia, LDLRAP1-related	<i>LDLRAP1</i>
Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	<i>CYP11B1</i>	D-bifunctional protein deficiency	<i>HSD17B4</i>	Familial hyperinsulinism, ABCC8-related	<i>ABCC8</i>
Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	<i>CYP17A1</i>	DCX-related disorders ^c	<i>DCX</i>	Familial hyperinsulinism, KCNJ11-related	<i>KCNJ11</i>
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	<i>CYP21A2</i>	Dent disease ^c	<i>CLCN5</i>	Familial Mediterranean fever	<i>MEFV</i>
Congenital amegakaryocytic thrombocytopenia	<i>MPL</i>	Desbuquois dysplasia, type I	<i>CANT1</i>	Fanconi anemia, complementation group A	<i>FANCA</i>
Congenital disorder of glycosylation, type Ia	<i>PMM2</i>	Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Fanconi anemia, complementation group B ^c	<i>FANCB</i>
Congenital disorder of glycosylation, type Ib	<i>MPI</i>	Dihydropyrimidine dehydrogenase deficiency	<i>DPYD</i>	Fanconi anemia, complementation group C	<i>FANCC</i>
Congenital disorder of glycosylation, type Ic	<i>ALG6</i>	Donnai-Barrow syndrome	<i>LRP2</i>	Fanconi anemia, complementation group D2	<i>FANCD2</i>
Congenital hydrocephalus 1	<i>CCDC88C</i>	Duchenne/Becker muscular dystrophy, X-linked ^c	<i>DMD</i>	Fanconi anemia, complementation group E	<i>FANCE</i>
Congenital ichthyosis, ABCA12-related	<i>ABCA12</i>	Dyskeratosis congenita, RTEL1-related	<i>RTEL1</i>	Fanconi anemia, complementation group F	<i>FANCF</i>
Congenital insensitivity to pain with anhidrosis	<i>NTRK1</i>	Dyskeratosis congenita, X-linked ^c	<i>DKC1</i>	Fanconi anemia, complementation group G	<i>FANCG</i>
Congenital myasthenic syndrome, CHAT-related	<i>CHAT</i>	Dystrophic epidermolysis bullosa, COL7A1-related	<i>COL7A1</i>	Fanconi anemia, complementation group I	<i>FANCI</i>
		Ehlers-Danlos syndrome, dermatosparaxis type	<i>ADAMTS2</i>	Fanconi anemia, complementation group L	<i>FANCL</i>
		Ellis-van Creveld syndrome	<i>EVC</i>	Farber lipogranulomatosis	<i>ASAH1</i>
		Ellis-van Creveld syndrome	<i>EVC2</i>	Fragile X syndrome ^c	<i>FMR1</i>
		Emery-Dreifuss muscular dystrophy, X-linked ^c	<i>EMD</i>	Fragile XE syndrome ^c	<i>AFF2</i>
		Enhanced S-cone syndrome	<i>NR2E3</i>	Fraser syndrome, type 3	<i>GRIP1</i>
		ERCC2-related conditions	<i>ERCC2</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes (continued)

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Free sialic acid storage disorders	<i>SLC17A5</i>	Hermansky-Pudlak syndrome, type 1	<i>HPS1</i>	Junctional epidermolysis bullosa, LAMC2-related	<i>LAMC2</i>
Friedreich ataxia	<i>FXN</i>	Hermansky-Pudlak syndrome, type 2	<i>AP3B1</i>	Juvenile retinoschisis, X-linked ^c	<i>RS1</i>
Fukuyama congenital muscular dystrophy	<i>FKTN</i>	Hermansky-Pudlak syndrome, type 3	<i>HPS3</i>	Krabbe disease	<i>GALC</i>
Fumarase deficiency	<i>FH</i>	Hermansky-Pudlak syndrome, type 4	<i>HPS4</i>	L1 syndrome ^c	<i>L1CAM</i>
GABA-transaminase deficiency	<i>ABAT</i>	Holocarboxylase synthetase deficiency	<i>HLCS</i>	LAMA2 muscular dystrophy	<i>LAMA2</i>
Galactosemia	<i>GALT</i>	Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	<i>MTHFR</i>	Leber congenital amaurosis 2	<i>RPE65</i>
Galactosemia, type II / Galactokinase deficiency	<i>GALK1</i>	Homocystinuria, CBS-related	<i>CBS</i>	Leber congenital amaurosis 5	<i>LCA5</i>
Galactosialidosis	<i>CTSA</i>	Homocystinuria, type cbIE	<i>MTRR</i>	Leber congenital amaurosis 13	<i>RDH12</i>
Gaucher disease	<i>GBA</i>	HPRT1-related disorders ^c	<i>HPRT1</i>	Leber congenital amaurosis, CEP290-related / CEP290-related conditions	<i>CEP290</i>
Gitelman syndrome	<i>SLC12A3</i>	Hydrolethalus syndrome	<i>HYLS1</i>	Lethal congenital contracture syndrome 1	<i>GLE1</i>
GLB1-related disorders	<i>GLB1</i>	Hyperphosphatemic familial tumoral calcinosis	<i>GALNT3</i>	Leukoencephalopathy with vanishing white matter 5	<i>EIF2B5</i>
Glucose-6-phosphate dehydrogenase deficiency ^c	<i>G6PD</i>	Hypohidrotic ectodermal dysplasia, X-linked ^c	<i>EDA</i>	Limb-girdle muscular dystrophy, type 21 / Muscular dystrophy-dystroglycanopathy, type A, 5	<i>FKRP</i>
Glutaric acidemia, type I	<i>GCDH</i>	Hypophosphatasia	<i>ALPL</i>	Limb-girdle muscular dystrophy, type 2A	<i>CAPN3</i>
Glycine encephalopathy, AMT-related	<i>AMT</i>	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked ^c	<i>FOXP3</i>	Limb-girdle muscular dystrophy, type 2B	<i>DYSF</i>
Glycine encephalopathy / Nonketotic hyperglycinemia	<i>GLDC</i>	Infantile cerebral and cerebellar atrophy	<i>MED17</i>	Limb-girdle muscular dystrophy, type 3	<i>SGCA</i>
Glycogen storage disease, type Ia	<i>G6PC1</i>	Isovaleric acidemia	<i>IVD</i>	Limb-girdle muscular dystrophy, type 4	<i>SGCB</i>
Glycogen storage disease, type Ib / IIw	<i>SLC37A4</i>	Johanson-Blizzard syndrome	<i>UBR1</i>	Limb-girdle muscular dystrophy, type 5	<i>SGCG</i>
Glycogen storage disease, type II / Pompe disease	<i>GAA</i>	Joubert syndrome 1	<i>INPP5E</i>	Limb-girdle muscular dystrophy, type 6	<i>SGCD</i>
Glycogen storage disease, type III	<i>AGL</i>	Joubert syndrome 2	<i>TMEM216</i>	Lipoid congenital adrenal hyperplasia	<i>STAR</i>
Glycogen storage disease, type IV / Adult polyglucosan body disease	<i>GBE1</i>	Joubert syndrome 3	<i>AHI1</i>	Lipoprotein lipase deficiency	<i>LPL</i>
Glycogen storage disease, type V	<i>PYGM</i>	Joubert syndrome 8	<i>ARL13B</i>	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	<i>HADHA</i>
Glycogen storage disease, type VII	<i>PFKM</i>	Joubert syndrome 9	<i>CC2D2A</i>	Lowe syndrome, X-linked ^c	<i>OCRL</i>
GNE myopathy	<i>GNE</i>	Joubert syndrome 15	<i>CEP41</i>	Lysinuric protein intolerance	<i>SLC7A7</i>
GRACILE syndrome	<i>BCS1L</i>	Joubert syndrome 17	<i>CPLANE1</i>	Lysosomal acid lipase deficiency	<i>LIPA</i>
Guanidinoacetate methyltransferase deficiency	<i>GAMT</i>	Joubert syndrome 21	<i>CSPP1</i>	Malonyl-CoA decarboxylase deficiency	<i>MLYCD</i>
Hereditary fructose intolerance	<i>ALDOB</i>	Joubert syndrome 25	<i>CEP104</i>	Maple syrup urine disease, type 1A	<i>BCKDHA</i>
Hereditary hemochromatosis, type 2	<i>HJV</i>	Joubert syndrome 27	<i>B9D1</i>		
Hereditary hemochromatosis, type 3	<i>TFR2</i>	Joubert syndrome 31	<i>CEP120</i>		
		Joubert syndrome 34	<i>B9D2</i>		
		Junctional epidermolysis bullosa, LAMA3-related	<i>LAMA3</i>		
		Junctional epidermolysis bullosa, LAMB3-related	<i>LAMB3</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes (continued)

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Maple syrup urine disease, type 1B	<i>BCKDHB</i>	Molybdenum cofactor deficiency of complementation group A	<i>MOCS1</i>	Muscular dystrophy-dystroglycanopathy, type A, 6	<i>LARGE1</i>
Maple syrup urine disease, type 2	<i>DBT</i>	MPV17-related mitochondrial DNA (mtDNA) maintenance defect	<i>MPV17</i>	Muscular dystrophy-dystroglycanopathy, type A, 7	<i>CRPPA</i>
Medium chain acyl-CoA dehydrogenase deficiency	<i>ACADM</i>	Mucopolipidosis II and mucopolipidosis III alpha/beta	<i>GNPTAB</i>	Myopathy, lactic acidosis, and sideroblastic anemia	<i>PUS1</i>
MEDNIK syndrome	<i>AP1S1</i>	Mucopolipidosis III gamma	<i>GNPTG</i>	Myotonia congenita	<i>CLCN1</i>
Megalencephalic leukoencephalopathy with subcortical cysts	<i>MLC1</i>	Mucopolipidosis IV	<i>MCOLN1</i>	N-acetylglutamate synthase deficiency	<i>NAGS</i>
Menkes disease ^c	<i>ATP7A</i>	Mucopolysaccharidosis, type I / Hurler syndrome	<i>IDUA</i>	Nemaline myopathy 2	<i>NEB</i>
Metachromatic leukodystrophy, ARSA-related	<i>ARSA</i>	Mucopolysaccharidosis, type II / Hunter syndrome ^c	<i>IDS</i>	Nephrogenic diabetes insipidus	<i>AQP2</i>
Metachromatic leukodystrophy due to saposin B deficiency	<i>PSAP</i>	Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	<i>SGSH</i>	Nephronophthisis 2	<i>INVS</i>
Methylmalonic aciduria, MMAA-related	<i>MMAA</i>	Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	<i>NAGLU</i>	Neuronal ceroid lipofuscinosis, CLN3-related	<i>CLN3</i>
Methylmalonic aciduria, MMAB-related	<i>MMAB</i>	Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	<i>HGSNAT</i>	Neuronal ceroid lipofuscinosis, CLN5-related	<i>CLN5</i>
Methylmalonic aciduria, MMUT-related	<i>MMUT</i>	Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	<i>GNS</i>	Neuronal ceroid lipofuscinosis, CLN6-related	<i>CLN6</i>
Mevalonic aciduria / Hyper-IgD syndrome	<i>MVK</i>	Mucopolysaccharidosis, type IIV / Morquio syndrome	<i>GALNS</i>	Neuronal ceroid lipofuscinosis, CLN8-related	<i>CLN8</i>
Microphthalmia / Anophthalmia	<i>VSX2</i>	Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	<i>ARSB</i>	Neuronal ceroid lipofuscinosis, CTSD-related	<i>CTSD</i>
Mitochondrial complex I deficiency, ACAD9-related	<i>ACAD9</i>	Mucopolysaccharidosis, type VII / Sly syndrome	<i>GUSB</i>	Neuronal ceroid lipofuscinosis, MFSD8-related	<i>MFSD8</i>
Mitochondrial complex I deficiency, nuclear type 1	<i>NDUFS4</i>	Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	<i>HYAL1</i>	Neuronal ceroid lipofuscinosis, PPT1-related	<i>PPT1</i>
Mitochondrial complex I deficiency, nuclear type 9	<i>NDUFS6</i>	Mulibrey nanism	<i>TRIM37</i>	Neuronal ceroid lipofuscinosis, TPP1-related	<i>TPP1</i>
Mitochondrial complex I deficiency, nuclear type 16	<i>NDUFAF5</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	<i>ETFA</i>	Niemann-Pick disease, types A/B	<i>SMPD1</i>
Mitochondrial complex I deficiency, nuclear type 17	<i>NDUFAF6</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB	<i>ETFB</i>	Niemann-Pick disease, type C1	<i>NPC1</i>
Mitochondrial complex IV deficiency, nuclear type 2	<i>SCO2</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	<i>ETFDH</i>	Niemann-Pick disease, type C2	<i>NPC2</i>
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	<i>LRPPRC</i>	Multiple pterygium syndrome, lethal type	<i>CHRNA3</i>	Nijmegen breakage syndrome	<i>NBN</i>
Mitochondrial complex IV deficiency, nuclear type 12	<i>PET100</i>	Multiple sulfatase deficiency	<i>SUMF1</i>	Nonsyndromic hearing loss and deafness (DFNB) 1	<i>GJB2</i>
Mitochondrial DNA depletion syndrome 1, MNGIE type	<i>TYMP</i>	Muscular dystrophy-dystroglycanopathy, type A, 1	<i>POMT1</i>	Nonsyndromic hearing loss and deafness (DFNB) 3	<i>MYO15A</i>
Mitochondrial trifunctional protein deficiency, HADHB-related	<i>HADHB</i>	Muscular dystrophy-dystroglycanopathy, type A, 2	<i>POMT2</i>	Nonsyndromic hearing loss and deafness (DFNB) 77	<i>LOXHD1</i>
MKS1-related disorders	<i>MKS1</i>	Muscular dystrophy-dystroglycanopathy, type A, 3	<i>POMGNT1</i>	NPHP1 nephronophthisis-related ciliopathies	<i>NPHP1</i>

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes (continued)

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Oculocutaneous albinism, type I	<i>TYR</i>	Primary hyperoxaluria, type I	<i>AGXT</i>	Rhizomelic chondrodysplasia punctata, type 2	<i>GNPAT</i>
Oculocutaneous albinism, type II	<i>OCA2</i>	Primary hyperoxaluria, type II	<i>GRHPR</i>	Rhizomelic chondrodysplasia punctata, type 3	<i>AGPS</i>
Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	<i>WNT10A</i>	Primary hyperoxaluria, type III	<i>HOGA1</i>	Roberts-SC phocomelia syndrome	<i>ESCO2</i>
Omenn syndrome	<i>DCLRE1C</i>	Progressive cerebello-cerebral atrophy	<i>SEPSECS</i>	Sandhott disease	<i>HEXB</i>
Orofacioidigital syndrome, type XIV	<i>C2CD3</i>	Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	<i>ATP8B1</i>	Schimke immunosseous dysplasia	<i>SMARCAL1</i>
Ornithine aminotransferase deficiency	<i>OAT</i>	Progressive familial intrahepatic cholestasis 2	<i>ABCB11</i>	Schindler disease	<i>NAGA</i>
Ornithine transcarbamylase deficiency, X-linked ^c	<i>OTC</i>	Progressive familial intrahepatic cholestasis 4	<i>TJP2</i>	Senior-Loken syndrome 5	<i>IQCB1</i>
Ornithine translocase deficiency	<i>SLC25A15</i>	Prolidase deficiency	<i>PEPD</i>	Severe combined immunodeficiency, RAG1-related	<i>RAG1</i>
Osteopetrosis, infantile malignant, TCIRG1-related	<i>TCIRG1</i>	Propionic acidemia, PCCA-related	<i>PCCA</i>	Severe combined immunodeficiency, RAG2-related	<i>RAG2</i>
Pendred syndrome	<i>SLC26A4</i>	Propionic acidemia, PCCB-related	<i>PCCB</i>	Severe combined immunodeficiency, X-linked ^c	<i>IL2RG</i>
Perlman syndrome	<i>DIS3L2</i>	PRPS1-related disorders ^c	<i>PRPS1</i>	Severe congenital neutropenia 5	<i>VPS45</i>
Peroxisomal acyl-CoA oxidase deficiency	<i>ACOX1</i>	Pseudocholinesterase deficiency	<i>BCHE</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	<i>DYNC2H1</i>
Phenylalanine hydroxylase deficiency	<i>PAH</i>	Pseudoxanthoma elasticum	<i>ABCC6</i>	Shwachman-Diamond syndrome	<i>SBDS</i>
Phosphoglycerate dehydrogenase deficiency	<i>PHGDH</i>	Pycnodysostosis	<i>CTSK</i>	Sialidosis	<i>NEU1</i>
PLA2G6-associated neurodegeneration	<i>PLA2G6</i>	Pyridoxine-dependent epilepsy	<i>ALDH7A1</i>	Sjögren-Larsson syndrome	<i>ALDH3A2</i>
PLP1-related disorders ^c	<i>PLP1</i>	Pyruvate carboxylase deficiency	<i>PC</i>	Skeletal dysplasia, SLC26A2-related	<i>SLC26A2</i>
POLG-related disorders	<i>POLG</i>	Pyruvate dehydrogenase E1-alpha deficiency ^c	<i>PDHA1</i>	Smith-Lemli-Opitz syndrome	<i>DHCR7</i>
Pontocerebellar hypoplasia, type 1A	<i>VRK1</i>	Pyruvate dehydrogenase E1-beta deficiency	<i>PDHB</i>	Spastic ataxia, Charlevoix-Saguenay type	<i>SACS</i>
Pontocerebellar hypoplasia, type 1B	<i>EXOSC3</i>	Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	<i>TANGO2</i>	Spastic paraplegia, type 15	<i>ZFYVE26</i>
Pontocerebellar hypoplasia, type 2B	<i>TSEN2</i>	Refsum disease	<i>PHYH</i>	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	<i>SLC1A4</i>
Pontocerebellar hypoplasia, type 2E	<i>VPS53</i>	Renal tubular acidosis	<i>SLC4A4</i>	Spinal muscular atrophy	<i>SMN1</i>
Pontocerebellar hypoplasia, types 4 and 2A	<i>TSEN54</i>	Renal tubular acidosis and deafness, ATP6V1B1-related	<i>ATP6V1B1</i>	Spondylothoracic dysostosis and spondylocostal dysostosis 2	<i>MESP2</i>
Pontocerebellar hypoplasia, type 6	<i>RARS2</i>	Retinitis pigmentosa 3 ^c	<i>RPGR</i>	Steel syndrome	<i>COL27A1</i>
Primary ciliary dyskinesia, DNAH5-related	<i>DNAH5</i>	Retinitis pigmentosa 25	<i>EYS</i>	Steroid resistant nephrotic syndrome, type 1	<i>NPHS1</i>
Primary ciliary dyskinesia, DNAI1-related	<i>DNAI1</i>	Retinitis pigmentosa 26	<i>CERKL</i>	Steroid-resistant nephrotic syndrome, type 2	<i>NPHS2</i>
Primary ciliary dyskinesia, DNAI2-related	<i>DNAI2</i>	Retinitis pigmentosa 28	<i>FAM161A</i>	Steroid-resistant nephrotic syndrome, type 3	<i>PLCE1</i>
Primary congenital glaucoma	<i>CYP1B1</i>	Retinitis pigmentosa 59	<i>DHDDS</i>	Stuve-Wiedemann syndrome	<i>LIFR</i>
		Rhizomelic chondrodysplasia punctata, type 1	<i>PEX7</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 445 diseases^b 446 genes (continued)

Test codes: Female (445 diseases): 13832 | Male (400 diseases): 13831

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Surfactant dysfunction, ABCA3-related	<i>ABCA3</i>	Usher syndrome, type 2A	<i>USH2A</i>	X-linked hyper IgM syndrome ^c	<i>CD40LG</i>
Tay-Sachs disease	<i>HEXA</i>	Usher syndrome, type 2C	<i>ADGRV1</i>	X-linked infantile spinal muscular atrophy ^c	<i>UBA1</i>
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	<i>TECPR2</i>	Usher syndrome, type 3A	<i>CLRN1</i>	X-linked myotubular myopathy ^c	<i>MTM1</i>
TNXB-related classical-like Ehlers-Danlos syndrome	<i>TNXB</i>	Very long-chain acyl-CoA dehydrogenase deficiency	<i>ACADVL</i>	X-linked Opitz G/BBB syndrome ^c	<i>MID1</i>
Trichohepatoenteric syndrome 1	<i>TTC37</i>	Vitamin D-dependent rickets, type 1A	<i>CYP27B1</i>	Xeroderma pigmentosum, group A	<i>XPA</i>
Trimethylaminuria	<i>FMO3</i>	Werner syndrome	<i>WRN</i>	Xeroderma pigmentosum, group C	<i>XPC</i>
Triple A syndrome	<i>AAAS</i>	Wilson disease	<i>ATP7B</i>	Zellweger spectrum disorders, PEX1-related	<i>PEX1</i>
Tyrosine hydroxylase deficiency	<i>TH</i>	Wiskott-Aldrich syndrome, X-linked ^c	<i>WAS</i>	Zellweger spectrum disorders, PEX2-related	<i>PEX2</i>
Tyrosinemia, type I	<i>FAH</i>	Wolcott-Rallison syndrome	<i>EIF2AK3</i>	Zellweger spectrum disorders, PEX6-related	<i>PEX6</i>
Tyrosinemia, type II	<i>TAT</i>	WWOX deficiency	<i>WWOX</i>	Zellweger spectrum disorders, PEX10-related	<i>PEX10</i>
Usher syndrome, type 1B	<i>MYO7A</i>	X-linked agammaglobulinemia ^c	<i>BTK</i>	Zellweger spectrum disorders, PEX12-related	<i>PEX12</i>
Usher syndrome, type 1C	<i>USH1C</i>	X-linked chondrodysplasia punctata 1 ^c	<i>ARSL</i>	Zellweger spectrum disorders, PEX26-related	<i>PEX26</i>
Usher syndrome, type 1D	<i>CDH23</i>	X-linked congenital adrenal hypoplasia ^c	<i>NROB1</i>		
Usher syndrome, type 1F	<i>PCDH15</i>	X-linked developmental disorders, ARX-related ^c	<i>ARX</i>		
Usher syndrome, type 1J	<i>CIB2</i>	X-linked heterotaxy-1 ^c	<i>ZIC3</i>		

QHerit 611 diseases^b 612 genes

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
3-beta-hydroxysteroid dehydrogenase deficiency, type II	<i>HSD3B2</i>	17-beta-hydroxysteroid dehydrogenase deficiency, type III	<i>HSD17B3</i>	Agnesis of the corpus callosum with peripheral neuropathy	<i>SLC12A6</i>
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	<i>HMGCL</i>	ABCA4-related disorders	<i>ABCA4</i>	Aicardi-Goutieres syndrome 2	<i>RNASEH2B</i>
3-hydroxyacyl-CoA dehydrogenase deficiency	<i>HADH</i>	Abetalipoproteinemia	<i>MTTP</i>	Aicardi-Goutieres syndrome 3	<i>RNASEH2C</i>
3-methylcrotonyl-CoA carboxylase 1 deficiency	<i>MCCC1</i>	Acrodermatitis enteropathica	<i>SLC39A4</i>	Aicardi-Goutieres syndrome 4	<i>RNASEH2A</i>
3-methylcrotonyl-CoA carboxylase 2 deficiency	<i>MCCC2</i>	Achromatopsia, CNGB3-related	<i>CNGB3</i>	Aicardi-Goutieres syndrome 5	<i>SAMHD1</i>
3-methylglutaconic aciduria, type III / Costett syndrome	<i>OPA3</i>	Action myoclonus renal failure syndrome	<i>SCARB2</i>	Alpha-1 antitrypsin deficiency	<i>SERPINA1</i>
5-alpha-reductase deficiency	<i>SRD5A2</i>	Acute infantile liver failure	<i>TRMU</i>	Alpha-mannosidosis	<i>MAN2B1</i>
6-pyruvoyl-tetrahydropterin synthase deficiency	<i>PTS</i>	ADAMTSL4-related eye disorders	<i>ADAMTSL4</i>	Alpha-thalassemia	<i>HBA1/HBA2</i>
		Adenosine deaminase deficiency	<i>ADA</i>	Alpha-thalassemia intellectual disability syndrome, X-linked ^c	<i>ATRX</i>
		Adrenoleukodystrophy, X-linked ^c	<i>ABCD1</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Alport syndrome, COL4A3-related	COL4A3	Bardet-Biedl syndrome 5	BBS5	Catecholaminergic polymorphic ventricular tachycardia, type 2	CASQ2
Alport syndrome, COL4A4-related	COL4A4	Bardet-Biedl syndrome 6	MKKS	CD59-mediated hemolytic anemia	CD59
Alport syndrome, COL4A5-related, X-linked ^c	COL4A5	Bardet-Biedl syndrome 7	BBS7	CEP152-related disorders	CEP152
Alkaptonuria	HGD	Bardet-Biedl syndrome 8	TTC8	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma (CEDNIK) syndrome	SNAP29
Alstrom syndrome	ALMS1	Bardet-Biedl syndrome 9	BBS9	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	ERCC6
Amish infantile epilepsy syndrome	ST3GAL5	Bardet-Biedl syndrome 10	BBS10	Cerebrotendinous xanthomatosis	CYP27A1
Androgen insensitivity syndrome	AR	Bardet-Biedl syndrome 12	BBS12	Charcot-Marie-Tooth disease, type 1X ^c	GJB1
Argininemia	ARG1	Bare lymphocyte syndrome, type II	CIITA	Charcot-Marie-Tooth disease, type 4D	NDRG1
Arginine: glycine amidinotransferase deficiency	GATM	Barth syndrome ^c	TFAZZIN	Chediak-Higashi syndrome	LYST
Argininosuccinic aciduria	ASL	Bartter syndrome, type 1	SLC12A1	Choreoacanthocytosis	VPS13A
Arthrogryposis, mental retardation, and seizures	SLC35A3	Bartter syndrome, type 2	KCNJ1	Choroideremia, X-linked ^c	CHM
Aromatase deficiency	CYP19A1	Bartter syndrome, type 4A	BSND	Chronic granulomatous disease 2	NCF2
Asparagine synthetase deficiency	ASNS	Bernard-Soulier syndrome, type A	GP1BA	Chronic granulomatous disease 4	CYBA
Aspartylglycosaminuria	AGA	Bernard-Soulier syndrome, type C	GP9	Chronic granulomatous disease, X-linked ^c	CYBB
Ataxia-telangiectasia	ATM	Beta hemoglobinopathies	HBB	Ciliopathies, RPGRIP1L-related	RPGRIP1L
Ataxia-telangiectasia-like disorder 1	MRE11	Beta-ketothiolase deficiency	ACAT1	Citrin deficiency / Citrullinemia, type II	SLC25A13
Ataxia with isolated vitamin E deficiency	TTPA	Beta-mannosidosis	MANBA	Citrullinemia, type I	ASS1
Atransferrinemia	TF	Beta-ureidopropionase deficiency	UPB1	Cockayne syndrome, type A	ERCC8
Autoimmune polyglandular syndrome, type 1	AIRE	Bilateral frontoparietal polymicrogyria	ADGRG1	Cohen syndrome	VPS13B
Autosomal recessive congenital ichthyosis	TGM1	Biotinidase deficiency	BTD	COL11A2-related disorders	COL11A2
Autosomal recessive intellectual developmental disorder, type 3	CC2D1A	Biotin-thiamine-responsive basal ganglia disease	SLC19A3	Combined malonic and methylmalonic aciduria	ACSF3
Autosomal recessive polycystic kidney disease	PKHD1	Bloom syndrome	BLM	Combined methylmalonic aciduria and homocystinuria, cbIC type / Cobalamin C deficiency	MMACHC
Autosomal recessive primary microcephaly 1	MCPH1	Brittle cornea syndrome 1	ZNF469	Combined methylmalonic aciduria and homocystinuria, cbID type / Cobalamin D deficiency	MMADHC
Autosomal recessive spinocerebellar ataxia, type 10	ANO10	Brittle cornea syndrome 2	PRDM5	Combined or isolated pituitary hormone deficiency, type 1	POU1F1
AVPR2-related disorders	AVPR2	Canavan disease	ASPA		
Bardet-Biedl syndrome 1	BBS1	Carbamoyl phosphate synthetase I deficiency	CPS1		
Bardet-Biedl syndrome 2	BBS2	Carnitine-acylcarnitine translocase deficiency	SLC25A20		
Bardet-Biedl syndrome 3	ARL6	Carnitine deficiency, systemic primary	SLC22A5		
Bardet-Biedl syndrome 4	BBS4	Carnitine palmitoyltransferase I deficiency	CPT1A		
		Carnitine palmitoyltransferase II deficiency	CPT2		
		Carpenter syndrome	RAB23		
		Cartilage-hair hypoplasia	RMRP		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Combined oxidative phosphorylation deficiency 1	<i>GFM1</i>	Congenital myasthenic syndrome, COLQ-related	<i>COLQ</i>	Dihydropyrimidine dehydrogenase deficiency	<i>DPYD</i>
Combined oxidative phosphorylation deficiency 3	<i>TSMF</i>	Congenital myasthenic syndrome, DOK7-related	<i>DOK7</i>	Donnai-Barrow syndrome	<i>LRP2</i>
Combined oxidative phosphorylation deficiency 6 ^c	<i>AIFM1</i>	Congenital myasthenic syndrome, MUSK-related	<i>MUSK</i>	Dubin-Johnson syndrome	<i>ABCC2</i>
Combined pituitary hormone deficiency, type 2	<i>PROT1</i>	Congenital myasthenic syndrome, RAPSN-related	<i>RAPSN</i>	Dubin-Johnson syndrome	<i>ABCC2</i>
Combined pituitary hormone deficiency, type 3	<i>LHX3</i>	Congenital neutropenia, HAX1-related	<i>HAX1</i>	Duchenne/Becker muscular dystrophy, X-linked ^c	<i>DMD</i>
Congenital adrenal insufficiency, CYP11A1-related	<i>CYP11A1</i>	Congenital secretory chloride diarrhea 1	<i>SLC26A3</i>	Dyskeratosis congenita, RTEL1-related	<i>RTEL1</i>
Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	<i>CYP11B1</i>	Corneal dystrophy and perceptive deafness syndrome	<i>SLC4A11</i>	Dyskeratosis congenita spectrum disorders	<i>TERT</i>
Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	<i>CYP17A1</i>	Corticosterone methyloxidase deficiency	<i>CYP11B2</i>	Dyskeratosis congenita, X-linked ^c	<i>DKC1</i>
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	<i>CYP21A2</i>	CRB1-related retinal dystrophies	<i>CRB1</i>	Dystrophic epidermolysis bullosa, COL7A1-related	<i>COL7A1</i>
Congenital amegakaryocytic thrombocytopenia	<i>MPL</i>	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome ^c	<i>SLC6A8</i>	Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum	<i>TBCD</i>
Congenital disorder of deglycosylation, type 1	<i>NGLY1</i>	CTSC-related disorders	<i>CTSC</i>	Ehlers-Danlos syndrome, dermatosparaxis type	<i>ADAMTS2</i>
Congenital disorder of glycosylation, type Ia	<i>PMM2</i>	CYP7B1-related disorders	<i>CYP7B1</i>	Ellis-van Creveld syndrome	<i>EVC</i>
Congenital disorder of glycosylation, type Ib	<i>MPI</i>	Cystic fibrosis	<i>CFTR</i>	Ellis-van Creveld syndrome	<i>EVC2</i>
Congenital disorder of glycosylation, type Ic	<i>ALG6</i>	Cystinosis	<i>CTNS</i>	Emery-Dreifuss muscular dystrophy, X-linked ^c	<i>EMD</i>
Congenital disorder of glycosylation, type Ik	<i>ALG1</i>	Cytochrome P450 oxidoreductase deficiency	<i>POR</i>	Enhanced S-cone syndrome	<i>NR2E3</i>
Congenital dyserythropoietic anemia, type II	<i>SEC23B</i>	D-bifunctional protein deficiency	<i>HSD17B4</i>	EPG5-related disorder	<i>EPG5</i>
Congenital hydrocephalus	<i>CCDC88C</i>	DCX-related disorders ^c	<i>DCX</i>	ERCC2-related conditions	<i>ERCC2</i>
Congenital hypothyroidism, TSHB-related	<i>TSHB</i>	DGAT1 deficiency	<i>DGAT1</i>	Ethylmalonic encephalopathy	<i>ETHE1</i>
Congenital hypothyroidism, TSHR-related	<i>TSHR</i>	Dent disease ^c	<i>CLCN5</i>	Fabry disease, X-linked ^c	<i>GLA</i>
Congenital ichthyosis, ABCA12-related	<i>ABCA12</i>	Developmental and epileptic encephalopathy 36	<i>ALG13</i>	Factor II deficiency / Prothrombin deficiency	<i>F2</i>
Congenital insensitivity to pain with anhidrosis	<i>NTRK1</i>	Developmental and epileptic encephalopathy 50	<i>CAD</i>	Factor V deficiency	<i>F5</i>
Congenital myasthenic syndrome, CHAT-related	<i>CHAT</i>	Deoxyguanosine kinase deficiency / Mitochondrial DNA depletion syndrome 3	<i>DGUOK</i>	Factor IX deficiency / Hemophilia B ^c	<i>F9</i>
Congenital myasthenic syndrome, CHRNE-related	<i>CHRNE</i>	Desbuquois dysplasia, type I	<i>CANT1</i>	Factor XI deficiency / Hemophilia C	<i>F11</i>
		Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Factor VIII deficiency / Hemophilia A ^c	<i>F8</i>
		Dihydroliipoamide dehydrogenase deficiency	<i>DLD</i>	Familial dysautonomia	<i>ELP1</i>
		Dihydropteridine reductase (DHPR) deficiency	<i>QDPR</i>	Familial hemophagocytic lymphohistiocytosis 2	<i>PRF1</i>
				Familial hemophagocytic lymphohistiocytosis 3	<i>UNC13D</i>
				Familial hemophagocytic lymphohistiocytosis 4	<i>STX11</i>

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Familial hemophagocytic lymphohistiocytosis 5	<i>STXBP2</i>	Fukuyama congenital muscular dystrophy	<i>FKTN</i>	GRACILE syndrome	<i>BCS1L</i>
Familial hypercholesterolemia, LDLR-related	<i>LDLR</i>	Fumarase deficiency	<i>FH</i>	Guanidinoacetate methyltransferase deficiency	<i>GAMT</i>
Familial hypercholesterolemia, LDLRAP1-related	<i>LDLRAP1</i>	G6PC3 deficiency	<i>G6PC3</i>	Heme oxygenase 1 deficiency	<i>HMOX1</i>
Familial hyperinsulinism, ABCC8-related	<i>ABCC8</i>	GABA-transaminase deficiency	<i>ABAT</i>	Hereditary fructose intolerance	<i>ALDOB</i>
Familial hyperinsulinism, KCNJ11-related	<i>KCNJ11</i>	Galactosemia	<i>GALT</i>	Hereditary hemochromatosis, type 2	<i>HJV</i>
Familial Mediterranean fever	<i>MEFV</i>	Galactosemia, type II / Galactokinase deficiency	<i>GALK1</i>	Hereditary hemochromatosis, type 2B	<i>HAMP</i>
Fanconi anemia, complementation group A	<i>FANCA</i>	Galactosemia, type III / Galactose epimerase deficiency	<i>GALE</i>	Hereditary hemochromatosis, type 3	<i>TFR2</i>
Fanconi anemia, complementation group B ^c	<i>FANCB</i>	Galactosialidosis	<i>CTSA</i>	Hermansky-Pudlak syndrome, type 1	<i>HPS1</i>
Fanconi anemia, complementation group C	<i>FANCC</i>	Gaucher disease	<i>GBA</i>	Hermansky-Pudlak syndrome, type 2	<i>AP3B1</i>
Fanconi anemia, complementation group D2	<i>FANCD2</i>	GCH1-related disorders	<i>GCH1</i>	Hermansky-Pudlak syndrome, type 3	<i>HPS3</i>
Fanconi anemia, complementation group E	<i>FANCE</i>	GDF5-related disorders	<i>GDF5</i>	Hermansky-Pudlak syndrome, type 4	<i>HPS4</i>
Fanconi anemia, complementation group F	<i>FANCF</i>	Geroderma osteodysplastica	<i>GORAB</i>	Hermansky-Pudlak syndrome, type 5	<i>HPS5</i>
Fanconi anemia, complementation group G	<i>FANCG</i>	Gitelman syndrome	<i>SLC12A3</i>	Hermansky-Pudlak syndrome, type 6	<i>HPS6</i>
Fanconi anemia, complementation group I	<i>FANCI</i>	GLB1-related disorders	<i>GLB1</i>	Hermansky-Pudlak syndrome, type 8	<i>BLOC1S3</i>
Fanconi anemia, complementation group J	<i>BRIP1</i>	Glucose-6-phosphate dehydrogenase deficiency ^c	<i>G6PD</i>	Hermansky-Pudlak syndrome, type 9	<i>BLOC1S6</i>
Fanconi anemia, complementation group L	<i>FANCL</i>	Glutaric acidemia, type I	<i>GCDH</i>	Holocarboxylase synthetase deficiency	<i>HLCS</i>
Farber lipogranulomatosis	<i>ASAH1</i>	Glutathione synthetase deficiency	<i>GSS</i>	Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	<i>MTHFR</i>
FHL1-related disorders	<i>FHL1</i>	Glycine encephalopathy, AMT-related	<i>AMT</i>	Homocystinuria, CBS-related	<i>CBS</i>
Foveal hypoplasia 2	<i>SLC38A8</i>	Glycine encephalopathy / Nonketotic hyperglycinemia	<i>GLDC</i>	Homocystinuria-megaloblastic anemia, cblG type	<i>MTR</i>
Fragile X syndrome ^c	<i>FMR1</i>	Glycogen storage disease, type Ia	<i>G6PC1</i>	Homocystinuria, type cblE	<i>MTRR</i>
Fragile XE syndrome ^c	<i>AFF2</i>	Glycogen storage disease, type Ib / IIw	<i>SLC37A4</i>	HPRT1-related disorders ^c	<i>HPRT1</i>
Fraser syndrome, type 1	<i>FRAS1</i>	Glycogen storage disease, type II / Pompe disease	<i>GAA</i>	HSD10 disease	<i>HSD17B10</i>
Fraser syndrome, type 2	<i>FREM2</i>	Glycogen storage disease, type III	<i>AGL</i>	Hydrolethalus syndrome	<i>HYLS1</i>
Fraser syndrome, type 3	<i>GRIP1</i>	Glycogen storage disease, type IV / Adult polyglucosan body disease	<i>GBE1</i>	Hyper-IgM syndrome, type 3	<i>CD40</i>
Free sialic acid storage disorders	<i>SLC17A5</i>	Glycogen storage disease, type V	<i>PYGM</i>	Hyperphosphatemic familial tumoral calcinosis	<i>GALNT3</i>
Friedreich ataxia	<i>FXN</i>	Glycogen storage disease, type VII	<i>PFKM</i>	Hypohidrotic ectodermal dysplasia, X-linked ^c	<i>EDA</i>
Fructose-1,6-bisphosphatase deficiency	<i>FBP1</i>	Glycogen storage disease, type IXb	<i>PHKB</i>	Hypomyelinating leukodystrophy 12	<i>VPS11</i>
Fucosidosis	<i>FUCA1</i>	Glycogen storage disease, type IXc	<i>PHKG2</i>		
		GNE myopathy	<i>GNE</i>		

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Hypophosphatasia	<i>ALPL</i>	Juvenile retinoschisis, X-linked ^c	<i>RS1</i>	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	<i>HADHA</i>
Ichthyosis prematurity syndrome	<i>SLC27A4</i>	Krabbe disease	<i>GALC</i>	Lowe syndrome, X-linked ^c	<i>OCRL</i>
IGHMBP2-related disorders	<i>IGHMBP2</i>	L1 syndrome ^c	<i>L1CAM</i>	LRAT-related disorders	<i>LRAT</i>
Imerslund-Gräsbeck syndrome 2	<i>AMN</i>	LAMA2 muscular dystrophy	<i>LAMA2</i>	Lung disease, immunodeficiency, and chromosome breakage syndrome (LICS)	<i>NSMCE3</i>
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	<i>DNMT3B</i>	Laron syndrome	<i>GHR</i>	Lysinuric protein intolerance	<i>SLC7A7</i>
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	<i>ZBTB24</i>	Leber congenital amaurosis 1	<i>GUCY2D</i>	Lysosomal acid lipase deficiency	<i>LIPA</i>
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked ^c	<i>FOXP3</i>	Leber congenital amaurosis 2	<i>RPE65</i>	Malonyl-CoA decarboxylase deficiency	<i>MLYCD</i>
Infantile cerebral and cerebellar atrophy	<i>MED17</i>	Leber congenital amaurosis 4	<i>AIPL1</i>	Maple syrup urine disease, type 1A	<i>BCKDHA</i>
Isolated sulfite oxidase deficiency	<i>SUOX</i>	Leber congenital amaurosis 5	<i>LCA5</i>	Maple syrup urine disease, type 1B	<i>BCKDHB</i>
Isovaleric acidemia	<i>IVD</i>	Leber congenital amaurosis 13	<i>RDH12</i>	Maple syrup urine disease, type 2	<i>DBT</i>
ITGB3-related disorders	<i>ITGB3</i>	Leber congenital amaurosis, CEP290-related / CEP290-related conditions	<i>CEP290</i>	MECR-related neurologic disorder	<i>MECR</i>
Johanson-Blizzard syndrome	<i>UBR1</i>	Lethal congenital contracture syndrome 1	<i>GLE1</i>	Medium chain acyl-CoA dehydrogenase deficiency	<i>ACADM</i>
Joubert syndrome 1	<i>INPP5E</i>	Leukoencephalopathy with vanishing white matter 1	<i>EIF2B1</i>	MEDNIK syndrome	<i>AP1S1</i>
Joubert syndrome 2	<i>TMEM216</i>	Leukoencephalopathy with vanishing white matter 2	<i>EIF2B2</i>	Menkes disease ^c	<i>ATP7A</i>
Joubert syndrome 3	<i>AHI1</i>	Leukoencephalopathy with vanishing white matter 3	<i>EIF2B3</i>	Megalencephalic leukoencephalopathy with subcortical cysts	<i>MLC1</i>
Joubert syndrome 8	<i>ARL13B</i>	Leukoencephalopathy with vanishing white matter 4	<i>EIF2B4</i>	Metachromatic leukodystrophy, ARSA-related	<i>ARSA</i>
Joubert syndrome 9	<i>CC2D2A</i>	Leukoencephalopathy with vanishing white matter 5	<i>EIF2B5</i>	Metachromatic leukodystrophy due to saposin B deficiency	<i>PSAP</i>
Joubert syndrome 15	<i>CEP41</i>	LIG4 syndrome	<i>LIG4</i>	Methylmalonic aciduria and homocystinuria, cbIF type	<i>LMBRD1</i>
Joubert syndrome 17	<i>CPLANE1</i>	Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	<i>FKRP</i>	Methylmalonic aciduria and homocystinuria, cbIX type	<i>HCFC1</i>
Joubert syndrome 21	<i>CSPP1</i>	Limb-girdle muscular dystrophy, type 2A	<i>CAPN3</i>	Methylmalonic aciduria, MMAA-related	<i>MMAA</i>
Joubert syndrome 25	<i>CEP104</i>	Limb-girdle muscular dystrophy, type 2B	<i>DYSF</i>	Methylmalonic aciduria, MMAB-related	<i>MMAB</i>
Joubert syndrome 27	<i>B9D1</i>	Limb-girdle muscular dystrophy, type 3	<i>SGCA</i>	Methylmalonic aciduria, MMUT-related	<i>MMUT</i>
Joubert syndrome 31	<i>CEP120</i>	Limb-girdle muscular dystrophy, type 4	<i>SGCB</i>	Methylmalonyl-CoA epimerase deficiency	<i>MCEE</i>
Joubert syndrome 34	<i>B9D2</i>	Limb-girdle muscular dystrophy, type 5	<i>SGCG</i>	Mevalonic aciduria / Hyper-IgD syndrome	<i>MVK</i>
Junctional epidermolysis bullosa, COL17A1-related	<i>COL17A1</i>	Limb-girdle muscular dystrophy, type 6	<i>SGCD</i>		
Junctional epidermolysis bullosa, ITGA6-related	<i>ITGA6</i>	Lipoid congenital adrenal hyperplasia	<i>STAR</i>		
Junctional epidermolysis bullosa, ITGB4-related	<i>ITGB4</i>	Lipoprotein lipase deficiency	<i>LPL</i>		
Junctional epidermolysis bullosa, LAMA3-related	<i>LAMA3</i>				
Junctional epidermolysis bullosa, LAMB3-related	<i>LAMB3</i>				
Junctional epidermolysis bullosa, LAMC2-related	<i>LAMC2</i>				

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Microcephalic osteodysplastic primordial dwarfism, type II	<i>PCNT</i>	Mucopolipidosis IV	<i>MCOLN1</i>	Myotonia congenita	<i>CLCN1</i>
Microphthalmia / Anophthalmia	<i>VSX2</i>	Mucopolysaccharidosis, type I / Hurler syndrome	<i>IDUA</i>	N-acetylglutamate synthase deficiency	<i>NAGS</i>
Mitochondrial complex I deficiency, ACAD9-related	<i>ACAD9</i>	Mucopolysaccharidosis, type II / Hunter syndrome ^c	<i>IDS</i>	Nemaline myopathy 2	<i>NEB</i>
Mitochondrial complex I deficiency, nuclear type 1	<i>NDUFS4</i>	Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	<i>SGSH</i>	Nephrogenic diabetes insipidus	<i>AQP2</i>
Mitochondrial complex I deficiency, nuclear type 3	<i>NDUFS7</i>	Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	<i>NAGLU</i>	Nephronophthisis 2	<i>INVS</i>
Mitochondrial complex I deficiency, nuclear type 4	<i>NDUFV1</i>	Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	<i>HGSNAT</i>	Neuronal ceroid lipofuscinosis, CLN3-related	<i>CLN3</i>
Mitochondrial complex I deficiency, nuclear type 9	<i>NDUFS6</i>	Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	<i>GNS</i>	Neuronal ceroid lipofuscinosis, CLN5-related	<i>CLN5</i>
Mitochondrial complex I deficiency, nuclear type 10	<i>NDUFAF2</i>	Mucopolysaccharidosis, type IVA / Morquio syndrome	<i>GALNS</i>	Neuronal ceroid lipofuscinosis, CLN6-related	<i>CLN6</i>
Mitochondrial complex I deficiency, nuclear type 16	<i>NDUFAF5</i>	Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	<i>ARSB</i>	Neuronal ceroid lipofuscinosis, CLN8-related	<i>CLN8</i>
Mitochondrial complex I deficiency, nuclear type 17	<i>NDUFAF6</i>	Mucopolysaccharidosis, type VII / Sly syndrome	<i>GUSB</i>	Neuronal ceroid lipofuscinosis, CTSD-related	<i>CTSD</i>
Mitochondrial complex I deficiency, nuclear type 19	<i>FOXRED1</i>	Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	<i>HYAL1</i>	Neuronal ceroid lipofuscinosis, MFSD8-related	<i>MFSD8</i>
Mitochondrial complex IV deficiency, nuclear type 2	<i>SCO2</i>	Mulibrey nanism	<i>TRIM37</i>	Neuronal ceroid lipofuscinosis, PPT1-related	<i>PPT1</i>
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	<i>LRPPRC</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	<i>ETFA</i>	Neuronal ceroid lipofuscinosis, TPP1-related	<i>TPP1</i>
Mitochondrial complex IV deficiency, nuclear type 6	<i>COX15</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB	<i>ETFB</i>	Niemann-Pick disease, types A/B	<i>SMPD1</i>
Mitochondrial complex IV deficiency, nuclear type 12	<i>PET100</i>	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	<i>ETFDH</i>	Niemann-Pick disease, type C1	<i>NPC1</i>
Mitochondrial DNA depletion syndrome 1, MNGIE type	<i>TYMP</i>	Multiple pterygium syndrome, lethal type	<i>CHRNA3</i>	Niemann-Pick disease, type C2	<i>NPC2</i>
Mitochondrial trifunctional protein deficiency, HADHB-related	<i>HADHB</i>	Multiple sulfatase deficiency	<i>SUMF1</i>	Nijmegen breakage syndrome	<i>NBN</i>
MKS1-related disorders	<i>MKS1</i>	Muscular dystrophy-dystroglycanopathy, type A, 1	<i>POMT1</i>	Nonsyndromic hearing loss and deafness (DFNB) 1	<i>GJB2</i>
Molybdenum cofactor deficiency of complementation group A	<i>MOCS1</i>	Muscular dystrophy-dystroglycanopathy, type A, 2	<i>POMT2</i>	Nonsyndromic hearing loss and deafness (DFNB) 3	<i>MYO15A</i>
Molybdenum cofactor deficiency of complementation group B	<i>MOCS2</i>	Muscular dystrophy-dystroglycanopathy, type A, 3	<i>POMGNT1</i>	Nonsyndromic hearing loss and deafness (DFNB) 7	<i>TMC1</i>
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	<i>MPV17</i>	Muscular dystrophy-dystroglycanopathy, type A, 6	<i>LARGE1</i>	Nonsyndromic hearing loss and deafness (DFNB) 8	<i>TMPRSS3</i>
Mucopolipidosis II and mucopolipidosis III alpha/beta	<i>GNPTAB</i>	Muscular dystrophy-dystroglycanopathy, type A, 7	<i>CRPPA</i>	Nonsyndromic hearing loss and deafness (DFNB) 9	<i>OTOF</i>
Mucopolipidosis III gamma	<i>GNPTG</i>	Muscular dystrophy-dystroglycanopathy, type A, 10	<i>RXYLT1</i>	Nonsyndromic hearing loss and deafness (DFNB) 22	<i>OTOA</i>
		Myopathy, lactic acidosis, and sideroblastic anemia	<i>PUS1</i>	Nonsyndromic hearing loss and deafness (DFNB) 59	<i>PJVK</i>
				Nonsyndromic hearing loss and deafness (DFNB) 76	<i>SYNE4</i>
				Nonsyndromic hearing loss and deafness (DFNB) 77	<i>LOXHD1</i>

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Normophosphatemic familial tumoral calcinosis	<i>SAMD9</i>	PGM3-congenital disorder of glycosylation / Immunodeficiency 23	<i>PGM3</i>	Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	<i>ATP8B1</i>
NPHP1 nephronophthisis-related ciliopathies	<i>NPHP1</i>	Phenylalanine hydroxylase deficiency	<i>PAH</i>	Progressive familial intrahepatic cholestasis 2	<i>ABCB11</i>
NPHP3 nephronophthisis-related ciliopathies	<i>NPHP3</i>	Phosphoglycerate dehydrogenase deficiency	<i>PHGDH</i>	Progressive familial intrahepatic cholestasis 3	<i>ABCB4</i>
NPHP4 nephronophthisis-related ciliopathies	<i>NPHP4</i>	PIGN-related disorders	<i>PIGN</i>	Progressive familial intrahepatic cholestasis 4	<i>TJP2</i>
Oculocutaneous albinism, type I	<i>TYR</i>	PLA2G6-associated neurodegeneration	<i>PLA2G6</i>	Progressive pseudorheumatoid dysplasia	<i>CCN6</i>
Oculocutaneous albinism, type II	<i>OCA2</i>	PLEKHG5-related disorders	<i>PLEKHG5</i>	Prolidase deficiency	<i>PEPD</i>
Oculocutaneous albinism, type IV	<i>SLC45A2</i>	PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome	<i>PLOD1</i>	Propionic acidemia, PCCA-related	<i>PCCA</i>
Oculocutaneous albinism, type III	<i>TYRP1</i>	PLP1-related disorders ^c	<i>PLP1</i>	Propionic acidemia, PCCB-related	<i>PCCB</i>
Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	<i>WNT10A</i>	POLG-related disorders	<i>POLG</i>	PRPS1-related disorders ^c	<i>PRPS1</i>
Omenn syndrome	<i>DCLRE1C</i>	Pontocerebellar hypoplasia, type 1A	<i>VRK1</i>	Pseudocholinesterase deficiency	<i>BCHE</i>
Orofaciodigital syndrome, type XIV	<i>C2CD3</i>	Pontocerebellar hypoplasia, type 1B	<i>EXOSC3</i>	Pseudoxanthoma elasticum	<i>ABCC6</i>
Ornithine aminotransferase deficiency	<i>OAT</i>	Pontocerebellar hypoplasia, type 2B	<i>TSEN2</i>	Pterin-4 alpha-carbinolamine dehydratase (PCD) deficiency	<i>PCBD1</i>
Ornithine transcarbamylase deficiency, X-linked ^c	<i>OTC</i>	Pontocerebellar hypoplasia, type 2E	<i>VPS53</i>	Pycnodysostosis	<i>CTSK</i>
Ornithine translocase deficiency	<i>SLC25A15</i>	Pontocerebellar hypoplasia, types 4 and 2A	<i>TSEN54</i>	Pyridoxamine 5'-phosphate oxidase deficiency	<i>PNPO</i>
Osteogenesis imperfecta, type VII	<i>CRTAP</i>	Pontocerebellar hypoplasia, type 6	<i>RARS2</i>	Pyridoxine-dependent epilepsy	<i>ALDH7A1</i>
Osteogenesis imperfecta, type VIII	<i>P3H1</i>	Primary ciliary dyskinesia, CCDC39-related	<i>CCDC39</i>	Pyruvate carboxylase deficiency	<i>PC</i>
Osteogenesis imperfecta, type XI	<i>FKBP10</i>	Primary ciliary dyskinesia, CCDC103-related	<i>CCDC103</i>	Pyruvate dehydrogenase E1-alpha deficiency ^c	<i>PDHA1</i>
Osteogenesis imperfecta, type XIII	<i>BMP1</i>	Primary ciliary dyskinesia, DNAH5-related	<i>DNAH5</i>	Pyruvate dehydrogenase E1-beta deficiency	<i>PDHB</i>
Osteopetrosis, infantile malignant, TCIRG1-related	<i>TCIRG1</i>	Primary ciliary dyskinesia, DNAH11-related	<i>DNAH11</i>	Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	<i>TANGO2</i>
Osteopetrosis, OSTM1-related	<i>OSTM1</i>	Primary ciliary dyskinesia, DNAI1-related	<i>DNAI1</i>	Refsum disease	<i>PHYH</i>
Parkinson disease 15	<i>FBX07</i>	Primary ciliary dyskinesia, DNAI2-related	<i>DNAI2</i>	Renal tubular acidosis	<i>SLC4A4</i>
Pantothenate kinase-associated neurodegeneration	<i>PANK2</i>	Primary congenital glaucoma	<i>CYP1B1</i>	Renal tubular acidosis and deafness, ATP6V1B1-related	<i>ATP6V1B1</i>
Pendred syndrome	<i>SLC26A4</i>	Primary hyperoxaluria, type I	<i>AGXT</i>	Retinitis pigmentosa 2	<i>RP2</i>
Perlman syndrome	<i>DIS3L2</i>	Primary hyperoxaluria, type II	<i>GRHPR</i>	Retinitis pigmentosa 3 ^c	<i>RPGR</i>
Peroxisomal acyl-CoA oxidase deficiency	<i>ACOX1</i>	Primary hyperoxaluria, type III	<i>HOGA1</i>	Retinitis pigmentosa 25	<i>EYS</i>
		Progressive cerebello-cerebral atrophy	<i>SEPSECS</i>	Retinitis pigmentosa 26	<i>CERKL</i>

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
Retinitis pigmentosa 28	<i>FAM161A</i>	Severe congenital neutropenia 5	<i>VPS45</i>	Thyroid dys-hormonogenesis 6	<i>DUOX2</i>
Retinitis pigmentosa 36	<i>PRCD</i>	Short-rib thoracic dysplasia 3 with or without polydactyly	<i>DYNC2H1</i>	TK2-related mitochondrial disorders	<i>TK2</i>
Retinitis pigmentosa 59	<i>DHDDS</i>	Shwachman-Diamond syndrome	<i>SBDS</i>	TMEM67-related disorders	<i>TMEM67</i>
Retinitis pigmentosa 62	<i>MAK</i>	Sialidosis	<i>NEU1</i>	TNXB-related classical-like Ehlers-Danlos syndrome	<i>TNXB</i>
Rhizomelic chondrodysplasia punctata, type 1	<i>PEX7</i>	Sjögren-Larsson syndrome	<i>ALDH3A2</i>	Transcobalamin II deficiency	<i>TCN2</i>
Rhizomelic chondrodysplasia punctata, type 2	<i>GNPAT</i>	Skeletal dysplasia, SLC26A2-related	<i>SLC26A2</i>	TREX1-related disorders	<i>TREX1</i>
Rhizomelic chondrodysplasia punctata, type 3	<i>AGPS</i>	Smith-Lemli-Opitz syndrome	<i>DHCR7</i>	Trichohepatoenteric syndrome 1	<i>TTC37</i>
RLBP1-related retinopathies	<i>RLBP1</i>	Spastic ataxia, Charlevoix-Saguenay type	<i>SACS</i>	Trichohepatoenteric syndrome 2	<i>SKIC2</i>
Roberts-SC phocomelia syndrome	<i>ESCO2</i>	Spastic paraplegia, type 15	<i>ZFYVE26</i>	TRIM32-related disorders	<i>TRIM32</i>
RYR1-related disorders	<i>RYR1</i>	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	<i>SLC1A4</i>	Trimethylaminuria	<i>FM03</i>
Sandhott disease	<i>HEXB</i>	SPG11-related disorders	<i>SPG11</i>	Triple A syndrome	<i>AAAS</i>
Schimke immunosseous dysplasia	<i>SMARCAL1</i>	Spinal muscular atrophy	<i>SMN1</i>	TULP1-related disorders	<i>TULP1</i>
Schindler disease	<i>NAGA</i>	Spondylocostal dysostosis 1	<i>DLL3</i>	Tyrosine hydroxylase deficiency	<i>TH</i>
Senior-Loken syndrome 5	<i>IQCB1</i>	Spondylothoracic dysostosis and spondylocostal dysostosis 2	<i>MESP2</i>	Tyrosinemia, type I	<i>FAH</i>
Sepiapterin reductase deficiency	<i>SPR</i>	Steel syndrome	<i>COL27A1</i>	Tyrosinemia, type II	<i>TAT</i>
Severe combined immunodeficiency, CD3D-related	<i>CD3D</i>	Steroid resistant nephrotic syndrome, type 1	<i>NPHS1</i>	Tyrosinemia, type III	<i>HPD</i>
Severe combined immunodeficiency, CD3E-related	<i>CD3E</i>	Steroid-resistant nephrotic syndrome, type 2	<i>NPHS2</i>	Usher syndrome, type 1B	<i>MYO7A</i>
Severe combined immunodeficiency, FOXP1-related	<i>FOXP1</i>	Steroid-resistant nephrotic syndrome, type 3	<i>PLCE1</i>	Usher syndrome, type 1C	<i>USH1C</i>
Severe combined immunodeficiency, IKBKB-related	<i>IKBKB</i>	Stuve-Wiedemann syndrome	<i>LIFR</i>	Usher syndrome, type 1D	<i>CDH23</i>
Severe combined immunodeficiency, IL7R-related	<i>IL7R</i>	SURF1-related disorders	<i>SURF1</i>	Usher syndrome, type 1F	<i>PCDH15</i>
Severe combined immunodeficiency, JAK3-related	<i>JAK3</i>	Surfactant dysfunction, ABCA3-related	<i>ABCA3</i>	Usher syndrome, type 1J	<i>CIB2</i>
Severe combined immunodeficiency, PTPRC-related	<i>PTPRC</i>	Tay-Sachs disease	<i>HEXA</i>	Usher syndrome, type 2A	<i>USH2A</i>
Severe combined immunodeficiency, RAG1-related	<i>RAG1</i>	TBCE-related disorders	<i>TBCE</i>	Usher syndrome, type 2C	<i>ADGRV1</i>
Severe combined immunodeficiency, RAG2-related	<i>RAG2</i>	TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	<i>TECPR2</i>	Usher syndrome, type 3A	<i>CLRN1</i>
Severe combined immunodeficiency, X-linked ^c	<i>IL2RG</i>	Thiamine-responsive megaloblastic anemia syndrome	<i>SLC19A2</i>	Very long-chain acyl-CoA dehydrogenase deficiency	<i>ACADVL</i>
		Thyroid dys-hormonogenesis 1	<i>SLC5A5</i>	Vitamin D-dependent rickets, type 1A	<i>CYP27B1</i>
		Thyroid dys-hormonogenesis 2A	<i>TPO</i>	Vitamin D-resistant rickets, type 2A	<i>VDR</i>
		Thyroid dys-hormonogenesis 3	<i>TG</i>	VLDLR-associated cerebellar hypoplasia	<i>VLDLR</i>
				Warsaw breakage syndrome	<i>DDX11</i>
				Werner syndrome	<i>WRN</i>
				Wilson disease	<i>ATP7B</i>
				Wiskott-Aldrich syndrome, X-linked ^c	<i>WAS</i>
				Wolcott-Rallison syndrome	<i>EIF2AK3</i>
				Woodhouse-Sakati syndrome	<i>DCAF17</i>

^b Panel components for males do not include specified X-linked diseases.

^c Designated X-linked disease.

QHerit 611 diseases^b 612 genes (continued)

Test codes: Female (611 diseases): 14228 | Male (559 diseases): 14229

DISEASE	GENE	DISEASE	GENE	DISEASE	GENE
WWOX deficiency	<i>WWOX</i>	X-linked Opitz G/BBB syndrome ^c	<i>MID1</i>	Zellweger spectrum disorders, PEX10-related	<i>PEX10</i>
X-linked agammaglobulinemia ^c	<i>BTK</i>	Xeroderma pigmentosum, group A	<i>XPA</i>	Zellweger spectrum disorders, PEX12-related	<i>PEX12</i>
X-linked chondrodysplasia punctata 1 ^c	<i>ARSL</i>	Xeroderma pigmentosum, group C	<i>XPC</i>	Zellweger spectrum disorder, PEX13-related	<i>PEX13</i>
X-linked congenital adrenal hypoplasia ^c	<i>NR0B1</i>	Xeroderma pigmentosum, variant type (XP-V)	<i>POLH</i>	Zellweger spectrum disorder, PEX16-related	<i>PEX16</i>
X-linked developmental disorders, ARX-related ^c	<i>ARX</i>	Zellweger spectrum disorders, PEX1-related	<i>PEX1</i>	Zellweger spectrum disorders, PEX26-related	<i>PEX26</i>
X-linked heterotaxy-1 ^c	<i>ZIC3</i>	Zellweger spectrum disorders, PEX2-related	<i>PEX2</i>		
X-linked hyper IgM syndrome ^c	<i>CD40LG</i>	Zellweger spectrum disorder, PEX5-related	<i>PEX5</i>		
X-linked infantile spinal muscular atrophy ^c	<i>UBA1</i>	Zellweger spectrum disorders, PEX6-related	<i>PEX6</i>		
X-linked myotubular myopathy ^c	<i>MTM1</i>				

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^c Designated X-linked disease.



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Test codes may vary by location. Please contact your local laboratory for more information.

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