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
Alpha-thalassemia	*HBA1/ HBA2*
Beta hemoglobinopathies	*HBB*
Bloom syndrome	*BLM*
Canavan disease	*ASPA*
Cystic fibrosis	*CFTR*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Fanconi anemia, Group C	*FANCC*

DISEASE	GENE
Familial dysautonomia	*ELP1* (AKA: *IKBKAP)*
Familial hyperinsulinism, ABCC8-related	*ABCC8*
Fragile X syndrome[c]	*FMR1*
Fukuyama congenital muscular dystrophy (Walker-Warburg)	*FKTN*
Gaucher disease	*GBA*
Glycogen storage disease, type Ia	*G6PC* (AKA: *G6PC1)*

DISEASE	GENE
Joubert syndrome 2	*TMEM216*
Maple syrup urine disease, type 1B	*BCKDHB*
Mucolipidosis, type IV	*MCOLN1*
Nemaline myopathy 2	*NEB*
Niemann-Pick disease, types A/B	*SMPD1*
Spinal muscular atrophy	*SMN1*
Tay-Sachs disease	*HEXA*
Usher syndrome, type 1F	*PCDH15*
Usher syndrome, type 3A	*CLRN1*

[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.

Quest Diagnostics®

QHerit 112 diseases[b] 113 genes

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

DISEASE	GENE
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*
Achromatopsia, CNGB3-related	*CNGB3*
Adrenoleukodystrophy, X-linked[c]	*ABCD1*
Aicardi-Goutieres syndrome 2	*RNASEH2B*
Alpha-thalassemia	*HBA1/ HBA2*
Argininosuccinic aciduria	*ASL*
Aspartylglycosaminuria	*AGA*
Atransferrinemia	*TF*
Autoimmune polyglandular syndrome, type 1	*AIRE*
Autosomal recessive polycystic kidney disease	*PKHD1*
Autosomal recessive primary microcephaly 1	*MCPH1*
Autosomal recessive spinocerebellar ataxia, type 10	*ANO10*
Bardet-Biedl syndrome 1	*BBS1*
Bardet-Biedl syndrome 2	*BBS2*
Beta hemoglobinopathies	*HBB*
Beta-ketothiolase deficiency	*ACAT1*
Biotinidase deficiency	*BTD*
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*
Bloom syndrome	*BLM*
Canavan disease	*ASPA*
Carnitine palmitoyltransferase II deficiency	*CPT2*
Cerebrotendinous xanthomatosis	*CYP27A1*
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	*MMACHC*
Congenital adrenal insufficiency, CYP11A1-related	*CYP11A1*
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	*CYP21A2*
Congenital disorder of glycosylation, type Ia	*PMM2*
Congenital hydrocephalus 1	*CCDC88C*
Congenital myasthenic syndrome, CHRNE-related	*CHRNE*
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome[c]	*SLC6A8*
Cystic fibrosis	*CFTR*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Donnai-Barrow syndrome	*LRP2*
Duchenne/Becker muscular dystrophy, X-linked[c]	*DMD*
Dystrophic epidermolysis bullosa, COL7A1-related	*COL7A1*
Ellis-van Creveld syndrome	*EVC2*
ERCC2-related conditions	*ERCC2*
Fabry disease, X-linked[c]	*GLA*
Factor IX deficiency / Hemophilia B[c]	*F9*
Factory VIII deficiency / Hemophilia A[c]	*F8*
Familial dysautonomia	*ELP1*
Familial hemophagocytic lymphohistiocytosis 2	*PRF1*
Familial hyperinsulinism, ABCC8-related	*ABCC8*
Fanconi anemia, complementation group C	*FANCC*
Fragile X syndrome	*FMR1*
Fraser syndrome, type 3	*GRIP1*
Fragile X syndrome[c]	*AFF2*
Friedreich ataxia	*FXN*
Fukuyama congenital muscular dystrophy	*FKTN*
Galactosemia	*GALT*
Gaucher disease	*GBA*
Glycogen storage disease, type Ia	*G6PC1*
Glycogen storage disease, type Ib / IIw	*SLC37A4*
Glycogen storage disease, type II / Pompe disease	*GAA*
Glycogen storage disease, type IV / Adult polyglucosan body disease	*GBE1*
Hereditary fructose intolerance	*ALDOB*
Hermansky-Pudlak syndrome, type 1	*HPS1*
Hermansky-Pudlak syndrome, type 3	*HPS3*
Homocystinuria, CBS-related	*CBS*
Hypophosphatasia	*ALPL*
Joubert syndrome 2	*TMEM216*
Joubert syndrome 3	*AHI1*
Joubert syndrome 9	*CC2D2A*
Juvenile retinoschisis, X-linked[c]	*RS1*
L1 syndrome[c]	*L1CAM*
Leber congenital amaurosis, CEP290-related / CEP290-related conditions	*CEP290*
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*
Maple syrup urine disease, type 1B	*BCKDHB*
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*
Metachromatic leukodystrophy, ARSA-related	*ARSA*
Methylmalonic aciduria, MMUT-related	*MMUT*
Mevalonic aciduria / Hyper-IgD syndrome	*MVK*
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*
Mucolipidosis IV	*MCOLN1*
Mucopolysaccharidosis, type I / Hurler syndrome	*IDUA*
Myotonia congenita	*CLCN1*
Nemaline myopathy 2	*NEB*
Niemann-Pick disease, types A/B	*SMPD1*
Nonsyndromic hearing loss and deafness (DFNB) 1	*GJB2*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 112 diseases[b] 113 genes (continued)

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

DISEASE	GENE
Oculocutaneous albinism, type I	*TYR*
Oculocutaneous albinism, type II	*OCA2*
Ornithine transcarbamylase deficiency, X-linked[c]	*OTC*
Pendred syndrome	*SLC26A4*
Phenylalanine hydroxylase deficiency	*PAH*
PLP1-related disorders[c]	*PLP1*
POLG-related disorders	*POLG*
Pontocerebellar hypoplasia, type 6	*RARS2*
Primary hyperoxaluria, type I	*AGXT*
Retinitis pigmentosa 3[c]	*RPGR*

DISEASE	GENE
Retinitis pigmentosa 59	*DHDDS*
Schindler disease	*NAGA*
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*
Skeletal dysplasia, SLC26A2-related	*SLC26A2*
Smith-Lemli-Opitz syndrome	*DHCR7*
Spinal muscular atrophy	*SMN1*
Steroid resistant nephrotic syndrome, type 1	*NPHS1*
Surfactant dysfunction, ABCA3-related	*ABCA3*
Tay-Sachs disease	*HEXA*
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*
Trimethylaminuria	*FMO3*

DISEASE	GENE
Tyrosinemia, type I	*FAH*
Usher syndrome, type 1F	*PCDH15*
Usher syndrome, type 2A	*USH2A*
Usher syndrome, type 3A	*CLRN1*
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*
Vitamin D-dependent rickets, type 1A	*CYP27B1*
Wilson disease	*ATP7B*
X-linked congenital adrenal hypoplasia[c]	*NR0B1*
X-linked developmental disorders, ARX-related[c]	*ARX*
X-linked Opitz G/BBB syndrome[c]	*MID1*
Xeroderma pigmentosum, group C[c]	*XPC*

QHerit 198 diseases[b] 199 genes

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

DISEASE	GENE
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*
Abetalipoproteinemia	*MTTP*
Achromatopsia, CNGB3-related	*CNGB3*
Adenosine deaminase deficiency	*ADA*
Adrenoleukodystrophy, X-linked[c]	*ABCD1*
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*
Aicardi-Goutieres syndrome 2	*RNASEH2B*
Alpha-mannosidosis	*MAN2B1*
Alpha-thalassemia	*HBA1/ HBA2*
Alport syndrome, COL4A3-related	*COL4A3*
Alport syndrome, COL4A4-related	*COL4A4*

DISEASE	GENE
Alport syndrome, COL4A5-related, X-linked[c]	*COL4A5*
Argininosuccinic aciduria	*ASL*
Arthrogryposis, mental retardation, and seizures	*SLC35A3*
Aspartylglycosaminuria	*AGA*
Ataxia-telangiectasia	*ATM*
Atransferrinemia	*TF*
Autoimmune polyglandular syndrome, type 1	*AIRE*
Autosomal recessive congenital ichthyosis	*TGM1*
Autosomal recessive polycystic kidney disease	*PKHD1*
Autosomal recessive primary microcephaly 1	*MCPH1*
Autosomal recessive spinocerebellar ataxia, type 10	*ANO10*
Bardet-Biedl syndrome 1	*BBS1*
Bardet-Biedl syndrome 2	*BBS2*
Bardet-Biedl syndrome 10	*BBS10*
Beta hemoglobinopathies	*HBB*
Beta-ketothiolase deficiency	*ACAT1*
Biotinidase deficiency	*BTD*

DISEASE	GENE
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*
Bloom syndrome	*BLM*
Canavan disease	*ASPA*
Carnitine deficiency, systemic primary	*SLC22A5*
Carnitine palmitoyltransferase II deficiency	*CPT2*
Cartilage-hair hypoplasia	*RMRP*
Cerebrotendinous xanthomatosis	*CYP27A1*
Citrullinemia, type I	*ASS1*
Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	*MMACHC*
Combined pituitary hormone deficiency, type 2	*PROP1*
Congenital adrenal insufficiency, CYP11A1-related	*CYP11A1*
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	*CYP21A2*
Congenital amegakaryocytic thrombocytopenia	*MPL*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 198 diseases[b] 199 genes (continued)

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

DISEASE	GENE
Congenital disorder of glycosylation, type Ia	*PMM2*
Congenital hydrocephalus 1	*CCDC88C*
Congenital myasthenic syndrome, CHRNE-related	*CHRNE*
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome[c]	*SLC6A8*
Cystic fibrosis	*CFTR*
Cystinosis	*CTNS*
D-bifunctional protein deficiency	*HSD17B4*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Donnai-Barrow syndrome	*LRP2*
Duchenne/Becker muscular dystrophy, X-linked[c]	*DMD*
Dyskeratosis congenita, RTEL1-related	*RTEL1*
Dystrophic epidermolysis bullosa, COL7A1-related	*COL7A1*
Ellis-van Creveld syndrome	*EVC2*
ERCC2-related conditions	*ERCC2*
Fabry disease, X-linked[c]	*GLA*
Factor IX deficiency / Hemophilia B[c]	*F9*
Factor XI deficiency / Hemophilia C	*F11*
Factory VIII deficiency / Hemophilia A[c]	*F8*
Familial dysautonomia	*ELP1*
Familial hemophagocytic lymphohistiocytosis 2	*PRF1*
Familial hyperinsulinism, ABCC8-related	*ABCC8*
Familial hyperinsulinism, KCNJ11-related	*KCNJ11*
Familial Mediterranean fever	*MEFV*
Fanconi anemia, complementation group A	*FANCA*
Fanconi anemia, complementation group C	*FANCC*
Fragile X syndrome[c]	*FMR1*
Fragile XE syndrome[c]	*AFF2*
Fraser syndrome, type 3	*GRIP1*
Friedreich ataxia	*FXN*

DISEASE	GENE
Fukuyama congenital muscular dystrophy	*FKTN*
Galactosemia	*GALT*
Gaucher disease	*GBA*
GLB1-related disorders	*GLB1*
Glutaric acidemia, type I	*GCDH*
Glycine encephalopathy, AMT-related	*AMT*
Glycine encephalopathy / Nonketotic hyperglycinemia	*GLDC*
Glycogen storage disease, type Ia	*G6PC1*
Glycogen storage disease, type Ib / IIw	*SLC37A4*
Glycogen storage disease, type II / Pompe disease	*GAA*
Glycogen storage disease, type III	*AGL*
Glycogen storage disease, type IV / Adult polyglucosan body disease	*GBE1*
GNE myopathy	*GNE*
GRACILE syndrome	*BCS1L*
Hereditary fructose intolerance	*ALDOB*
Hermansky-Pudlak syndrome, type 1	*HPS1*
Hermansky-Pudlak syndrome, type 3	*HPS3*
Holocarboxylase synthetase deficiency	*HLCS*
Homocystinuria, CBS-related	*CBS*
Hydrolethalus syndrome	*HYLS1*
Hypophosphatasia	*ALPL*
Infantile cerebral and cerebellar atrophy	*MED17*
Isovaleric acidemia	*IVD*
Joubert syndrome 2	*TMEM216*
Joubert syndrome 3	*AHI1*
Joubert syndrome 9	*CC2D2A*
Junctional epidermolysis bullosa, LAMA3-related	*LAMA3*
Junctional epidermolysis bullosa, LAMB3-related	*LAMB3*
Junctional epidermolysis bullosa, LAMC2-related	*LAMC2*
Juvenile retinoschisis, X-linked[c]	*RS1*

DISEASE	GENE
Krabbe disease	*GALC*
L1 syndrome[c]	*L1CAM*
LAMA2 muscular dystrophy	*LAMA2*
Leber congenital amaurosis, CEP290-related / CEP290-related conditions	*CEP290*
Lethal congenital contracture syndrome 1	*GLE1*
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*
Limb-girdle muscular dystrophy, type 2A	*CAPN3*
Limb-girdle muscular dystrophy, type 3	*SGCA*
Limb-girdle muscular dystrophy, type 4	*SGCB*
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*
Lysinuric protein intolerance	*SLC7A7*
Maple syrup urine disease, type 1A	*BCKDHA*
Maple syrup urine disease, type 1B	*BCKDHB*
Maple syrup urine disease, type 2	*DBT*
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*
Menkes disease[c]	*ATP7A*
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*
Metachromatic leukodystrophy, ARSA-related	*ARSA*
Methylmalonic aciduria, MMAA-related	*MMAA*
Methylmalonic aciduria, MMAB-related	*MMAB*
Methylmalonic aciduria, MMUT-related	*MMUT*
Mevalonic aciduria / Hyper-IgD syndrome	*MVK*
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	*LRPPRC*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 198 diseases[b] 199 genes (continued)

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

DISEASE	GENE
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*
Mucolipidosis IV	*MCOLN1*
Mucopolysaccharidosis, type I / Hurler syndrome	*IDUA*
Mucopolysaccharidosis, type II / Hunter syndrome[c]	*IDS*
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	*SGSH*
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	*NAGLU*
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	*HGSNAT*
Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	*GNS*
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	*ARSB*
Myotonia congenita	*CLCN1*
Nemaline myopathy 2	*NEB*
Neuronal ceroid lipofuscinosis, CLN3-related	*CLN3*
Neuronal ceroid lipofuscinosis, CLN5-related	*CLN5*
Neuronal ceroid lipofuscinosis, CLN6-related	*CLN6*
Neuronal ceroid lipofuscinosis, CLN8-related	*CLN8*
Neuronal ceroid lipofuscinosis, PPT1-related	*PPT1*
Neuronal ceroid lipofuscinosis, TPP1-related	*TPP1*
Niemann-Pick disease, types A/B	*SMPD1*
Niemann-Pick disease, type C1	*NPC1*
Nijmegen breakage syndrome	*NBN*
Nonsyndromic hearing loss and deafness (DFNB) 1	*GJB2*
Nonsyndromic hearing loss and deafness (DFNB) 77	*LOXHD1*
Oculocutaneous albinism, type I	*TYR*
Oculocutaneous albinism, type II	*OCA2*
Ornithine transcarbamylase deficiency, X-linked[c]	*OTC*
Pendred syndrome	*SLC26A4*
Phenylalanine hydroxylase deficiency	*PAH*
PLP1-related disorders	*PLP1*
POLG-related disorders	*POLG*
Pontocerebellar hypoplasia, type 6	*RARS2*
Primary hyperoxaluria, type I	*AGXT*
Propionic acidemia, PCCA-related	*PCCA*
Propionic acidemia, PCCB-related	*PCCB*
Pyruvate carboxylase deficiency	*PC*
Retinitis pigmentosa 3[c]	*RPGR*
Retinitis pigmentosa 59	*DHDDS*
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*
Sandhott disease	*HEXB*
Schindler disease	*NAGA*
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*
Sjögren-Larsson syndrome	*ALDH3A2*
Skeletal dysplasia, SLC26A2-related	*SLC26A2*
Smith-Lemli-Opitz syndrome	*DHCR7*
Spastic ataxia, Charlevoix-Saguenay type	*SACS*
Spinal muscular atrophy	*SMN1*
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*
Steroid resistant nephrotic syndrome, type 1	*NPHS1*
Steroid-resistant nephrotic syndrome, type 2	*NPHS2*
Surfactant dysfunction, ABCA3-related	*ABCA3*
Tay-Sachs disease	*HEXA*
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*
Trimethylaminuria	*FMO3*
Tyrosine hydroxylase deficiency	*TH*
Tyrosinemia, type I	*FAH*
Tyrosinemia, type II	*TAT*
Usher syndrome, type 1B	*MYO7A*
Usher syndrome, type 1C	*USH1C*
Usher syndrome, type 1D	*CDH23*
Usher syndrome, type 1F	*PCDH15*
Usher syndrome, type 2A	*USH2A*
Usher syndrome, type 3A	*CLRN1*
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*
Vitamin D-dependent rickets, type 1A	*CYP27B1*
Wilson disease	*ATP7B*
X-linked congenital adrenal hypoplasia[c]	*NR0B1*
X-linked developmental disorders, ARX-related[c]	*ARX*
X-linked Opitz G/BBB syndrome[c]	*MID1*
Xeroderma pigmentosum, group C	*XPC*
Zellweger spectrum disorders, PEX1-related	*PEX1*
Zellweger spectrum disorders, PEX2-related	*PEX2*
Zellweger spectrum disorders, PEX6-related	*PEX6*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 445 diseases[b] 446 genes

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 445 diseases[b] 446 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 445 diseases[b] 446 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 445 diseases[b] 446 genes (continued)

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

DISEASE	GENE
Maple syrup urine disease, type 1B	*BCKDHB*
Maple syrup urine disease, type 2	*DBT*
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*
MEDNIK syndrome	*AP1S1*
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*
Menkes disease[c]	*ATP7A*
Metachromatic leukodystrophy, ARSA-related	*ARSA*
Metachromatic leukodystrophy due to saposin B deficiency	*PSAP*
Methylmalonic aciduria, MMAA-related	*MMAA*
Methylmalonic aciduria, MMAB-related	*MMAB*
Methylmalonic aciduria, MMUT-related	*MMUT*
Mevalonic aciduria / Hyper-IgD syndrome	*MVK*
Microphthalmia / Anophthalmia	*VSX2*
Mitochondrial complex I deficiency, ACAD9-related	*ACAD9*
Mitochondrial complex I deficiency, nuclear type 1	*NDUFS4*
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*
Mitochondrial complex I deficiency, nuclear type 17	*NDUFAF6*
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*
Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	*LRPPRC*
Mitochondrial complex IV deficiency, nuclear type 12	*PET100*
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*
Mitochondrial trifunctional protein deficiency, HADHB-related	*HADHB*
MKS1-related disorders	*MKS1*
Molybdenum cofactor deficiency of complementation group A	*MOCS1*
MPV17-related mitochondrial DNA (mtDNA) maintenance defect	*MPV17*
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*
Mucolipidosis III gamma	*GNPTG*
Mucolipidosis IV	*MCOLN1*
Mucopolysaccharidosis, type I / Hurler syndrome	*IDUA*
Mucopolysaccharidosis, type II / Hunter syndrome[c]	*IDS*
Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	*SGSH*
Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	*NAGLU*
Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	*HGSNAT*
Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	*GNS*
Mucopolysaccharidosis, type IVA / Morquio syndrome	*GALNS*
Mucopolysaccharidosis, type VI / Maroteaux-Lamy syndrome	*ARSB*
Mucopolysaccharidosis, type VII / Sly syndrome	*GUSB*
Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	*HYAL1*
Mulibrey nanism	*TRIM37*
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	*ETFA*
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB	*ETFB*
Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	*ETFDH*
Multiple pterygium syndrome, lethal type	*CHRNG*
Multiple sulfatase deficiency	*SUMF1*
Muscular dystrophy-dystroglycanopathy, type A, 1	*POMT1*
Muscular dystrophy-dystroglycanopathy, type A, 2	*POMT2*
Muscular dystrophy-dystroglycanopathy, type A, 3	*POMGNT1*
Muscular dystrophy-dystroglycanopathy, type A, 6	*LARGE1*
Muscular dystrophy-dystroglycanopathy, type A, 7	*CRPPA*
Myopathy, lactic acidosis, and sideroblastic anemia	*PUS1*
Myotonia congenita	*CLCN1*
N-acetylglutamate synthase deficiency	*NAGS*
Nemaline myopathy 2	*NEB*
Nephrogenic diabetes insipidus	*AQP2*
Nephronophthisis 2	*INVS*
Neuronal ceroid lipofuscinosis, CLN3-related	*CLN3*
Neuronal ceroid lipofuscinosis, CLN5-related	*CLN5*
Neuronal ceroid lipofuscinosis, CLN6-related	*CLN6*
Neuronal ceroid lipofuscinosis, CLN8-related	*CLN8*
Neuronal ceroid lipofuscinosis, CTSD-related	*CTSD*
Neuronal ceroid lipofuscinosis, MFSD8-related	*MFSD8*
Neuronal ceroid lipofuscinosis, PPT1-related	*PPT1*
Neuronal ceroid lipofuscinosis, TPP1-related	*TPP1*
Niemann-Pick disease, types A/B	*SMPD1*
Niemann-Pick disease, type C1	*NPC1*
Niemann-Pick disease, type C2	*NPC2*
Nijmegen breakage syndrome	*NBN*
Nonsyndromic hearing loss and deafness (DFNB) 1	*GJB2*
Nonsyndromic hearing loss and deafness (DFNB) 3	*MYO15A*
Nonsyndromic hearing loss and deafness (DFNB) 77	*LOXHD1*
NPHP1 nephronophthisis-related ciliopathies	*NPHP1*
NPHP3 nephronophthisis-related ciliopathies	*NPHP3*
NPHP4 nephronophthisis-related ciliopathies	*NPHP4*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 445 diseases[b] 446 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 445 diseases[b] 446 genes (continued)

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

DISEASE	GENE
Surfactant dysfunction, ABCA3-related	*ABCA3*
Tay-Sachs disease	*HEXA*
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*
Trichohepatoenteric syndrome 1	*TTC37*
Trimethylaminuria	*FMO3*
Triple A syndrome	*AAAS*
Tyrosine hydroxylase deficiency	*TH*
Tyrosinemia, type I	*FAH*
Tyrosinemia, type II	*TAT*
Usher syndrome, type 1B	*MYO7A*
Usher syndrome, type 1C	*USH1C*
Usher syndrome, type 1D	*CDH23*
Usher syndrome, type 1F	*PCDH15*
Usher syndrome, type 1J	*CIB2*
Usher syndrome, type 2A	*USH2A*
Usher syndrome, type 2C	*ADGRV1*
Usher syndrome, type 3A	*CLRN1*
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*
Vitamin D-dependent rickets, type 1A	*CYP27B1*
Werner syndrome	*WRN*
Wilson disease	*ATP7B*
Wiskott-Aldrich syndrome, X-linked[c]	*WAS*
Wolcott-Rallison syndrome	*EIF2AK3*
WWOX deficiency	*WWOX*
X-linked agammaglobulinemia[c]	*BTK*
X-linked chondrodysplasia punctata 1[c]	*ARSL*
X-linked congenital adrenal hypoplasia[c]	*NR0B1*
X-linked developmental disorders, ARX-related[c]	*ARX*
X-linked heterotaxy-1[c]	*ZIC3*
X-linked hyper IgM syndrome[c]	*CD40LG*
X-linked infantile spinal muscular atrophy[c]	*UBA1*
X-linked myotubular myopathy[c]	*MTM1*
X-linked Opitz G/BBB syndrome[c]	*MID1*
Xeroderma pigmentosum, group A	*XPA*
Xeroderma pigmentosum, group C	*XPC*
Zellweger spectrum disorders, PEX1-related	*PEX1*
Zellweger spectrum disorders, PEX2-related	*PEX2*
Zellweger spectrum disorders, PEX6-related	*PEX6*
Zellweger spectrum disorders, PEX10-related	*PEX10*
Zellweger spectrum disorders, PEX12-related	*PEX12*
Zellweger spectrum disorders, PEX26-related	*PEX26*

QHerit 611 diseases[b] 612 genes

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 611 diseases[b] 612 genes (continued)

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

DISEASE	GENE
Combined oxidative phosphorylation deficiency 1	*GFM1*
Combined oxidative phosphorylation deficiency 3	*TSFM*
Combined oxidative phosphorylation deficiency 6[c]	*AIFM1*
Combined pituitary hormone deficiency, type 2	*PROP1*
Combined pituitary hormone deficiency, type 3	*LHX3*
Congenital adrenal insufficiency, CYP11A1-related	*CYP11A1*
Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	*CYP11B1*
Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	*CYP17A1*
Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	*CYP21A2*
Congenital amegakaryocytic thrombocytopenia	*MPL*
Congenital disorder of deglycosylation, type 1	*NGLY1*
Congenital disorder of glycosylation, type Ia	*PMM2*
Congenital disorder of glycosylation, type Ib	*MPI*
Congenital disorder of glycosylation, type Ic	*ALG6*
Congenital disorder of glycosylation, type Ik	*ALG1*
Congenital dyserythropoietic anemia, type II	*SEC23B*
Congenital hydrocephalus	*CCDC88C*
Congenital hypothyroidism, TSHB-related	*TSHB*
Congenital hypothyroidism, TSHR-related	*TSHR*
Congenital ichthyosis, ABCA12-related	*ABCA12*
Congenital insensitivity to pain with anhidrosis	*NTRK1*
Congenital myasthenic syndrome, CHAT-related	*CHAT*
Congenital myasthenic syndrome, CHRNE-related	*CHRNE*
Congenital myasthenic syndrome, COLQ-related	*COLQ*
Congenital myasthenic syndrome, DOK7-related	*DOK7*
Congenital myasthenic syndrome, MUSK-related	*MUSK*
Congenital myasthenic syndrome, RAPSN-related	*RAPSN*
Congenital neutropenia, HAX1-related	*HAX1*
Congenital secretory chloride diarrhea 1	*SLC26A3*
Corneal dystrophy and perceptive deafness syndrome	*SLC4A11*
Corticosterone methyloxidase deficiency	*CYP11B2*
CRB1-related retinal dystrophies	*CRB1*
Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome[c]	*SLC6A8*
CTSC-related disorders	*CTSC*
CYP7B1-related disorders	*CYP7B1*
Cystic fibrosis	*CFTR*
Cystinosis	*CTNS*
Cytochrome P450 oxidoreductase deficiency	*POR*
D-bifunctional protein deficiency	*HSD17B4*
DCX-related disorders[c]	*DCX*
DGAT1 deficiency	*DGAT1*
Dent disease[c]	*CLCN5*
Developmental and epileptic encephalopathy 36	*ALG13*
Developmental and epileptic encephalopathy 50	*CAD*
Deoxyguanosine kinase deficiency / Mitochondrial DNA depletion syndrome 3	*DGUOK*
Desbuquois dysplasia, type I	*CANT1*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Dihydrolipoamide dehydrogenase deficiency	*DLD*
Dihydropteridine reductase (DHPR) deficiency	*QDPR*
Dihydropyrimidine dehydrogenase deficiency	*DPYD*
Donnai-Barrow syndrome	*LRP2*
Dubin-Johnson syndrome	*ABCC2*
Dubin-Johnson syndrome	*ABCC2*
Duchenne/Becker muscular dystrophy, X-linked[c]	*DMD*
Dyskeratosis congenita, RTEL1-related	*RTEL1*
Dyskeratosis congenita spectrum disorders	*TERT*
Dyskeratosis congenita, X-linked[c]	*DKC1*
Dystrophic epidermolysis bullosa, COL7A1-related	*COL7A1*
Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum	*TBCD*
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*
Ellis-van Creveld syndrome	*EVC*
Ellis-van Creveld syndrome	*EVC2*
Emery-Dreifuss muscular dystrophy, X-linked[c]	*EMD*
Enhanced S-cone syndrome	*NR2E3*
EPG5-related disorder	*EPG5*
ERCC2-related conditions	*ERCC2*
Ethylmalonic encephalopathy	*ETHE1*
Fabry disease, X-linked[c]	*GLA*
Factor II deficiency / Prothrombin deficiency	*F2*
Factor V deficiency	*F5*
Factor IX deficiency / Hemophilia B[c]	*F9*
Factor XI deficiency / Hemophilia C	*F11*
Factory VIII deficiency / Hemophilia A[c]	*F8*
Familial dysautonomia	*ELP1*
Familial hemophagocytic lymphohistiocytosis 2	*PRF1*
Familial hemophagocytic lymphohistiocytosis 3	*UNC13D*
Familial hemophagocytic lymphohistiocytosis 4	*STX11*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

Quest Diagnostics®

QHerit 611 diseases[b] 612 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

DISEASE	GENE
Hypophosphatasia	*ALPL*
Ichthyosis prematurity syndrome	*SLC27A4*
IGHMBP2-related disorders	*IGHMBP2*
Imerslund-Gräsbeck syndrome 2	*AMN*
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	*ZBTB24*
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked[c]	*FOXP3*
Infantile cerebral and cerebellar atrophy	*MED17*
Isolated sulfite oxidase deficiency	*SUOX*
Isovaleric acidemia	*IVD*
ITGB3-related disorders	*ITGB3*
Johanson-Blizzard syndrome	*UBR1*
Joubert syndrome 1	*INPP5E*
Joubert syndrome 2	*TMEM216*
Joubert syndrome 3	*AHI1*
Joubert syndrome 8	*ARL13B*
Joubert syndrome 9	*CC2D2A*
Joubert syndrome 15	*CEP41*
Joubert syndrome 17	*CPLANE1*
Joubert syndrome 21	*CSPP1*
Joubert syndrome 25	*CEP104*
Joubert syndrome 27	*B9D1*
Joubert syndrome 31	*CEP120*
Joubert syndrome 34	*B9D2*
Junctional epidermolysis bullosa, COL17A1-related	*COL17A1*
Junctional epidermolysis bullosa, ITGA6-related	*ITGA6*
Junctional epidermolysis bullosa, ITGB4-related	*ITGB4*
Junctional epidermolysis bullosa, LAMA3-related	*LAMA3*
Junctional epidermolysis bullosa, LAMB3-related	*LAMB3*
Junctional epidermolysis bullosa, LAMC2-related	*LAMC2*

DISEASE	GENE
Juvenile retinoschisis, X-linked[c]	*RS1*
Krabbe disease	*GALC*
L1 syndrome[c]	*L1CAM*
LAMA2 muscular dystrophy	*LAMA2*
Laron syndrome	*GHR*
Leber congenital amaurosis 1	*GUCY2D*
Leber congenital amaurosis 2	*RPE65*
Leber congenital amaurosis 4	*AIPL1*
Leber congenital amaurosis 5	*LCA5*
Leber congenital amaurosis 13	*RDH12*
Leber congenital amaurosis, CEP290-related / CEP290-related conditions	*CEP290*
Lethal congenital contracture syndrome 1	*GLE1*
Leukoencephalopathy with vanishing white matter 1	*EIF2B1*
Leukoencephalopathy with vanishing white matter 2	*EIF2B2*
Leukoencephalopathy with vanishing white matter 3	*EIF2B3*
Leukoencephalopathy with vanishing white matter 4	*EIF2B4*
Leukoencephalopathy with vanishing white matter 5	*EIF2B5*
LIG4 syndrome	*LIG4*
Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*
Limb-girdle muscular dystrophy, type 2A	*CAPN3*
Limb-girdle muscular dystrophy, type 2B	*DYSF*
Limb-girdle muscular dystrophy, type 3	*SGCA*
Limb-girdle muscular dystrophy, type 4	*SGCB*
Limb-girdle muscular dystrophy, type 5	*SGCG*
Limb-girdle muscular dystrophy, type 6	*SGCD*
Lipoid congenital adrenal hyperplasia	*STAR*
Lipoprotein lipase deficiency	*LPL*

DISEASE	GENE
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*
Lowe syndrome, X-linked[c]	*OCRL*
LRAT-related disorders	*LRAT*
Lung disease, immunodeficiency, and chromosome breakage syndrome (LICS)	*NSMCE3*
Lysinuric protein intolerance	*SLC7A7*
Lysosomal acid lipase deficiency	*LIPA*
Malonyl-CoA decarboxylase deficiency	*MLYCD*
Maple syrup urine disease, type 1A	*BCKDHA*
Maple syrup urine disease, type 1B	*BCKDHB*
Maple syrup urine disease, type 2	*DBT*
MECR-related neurologic disorder	*MECR*
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*
MEDNIK syndrome	*AP1S1*
Menkes disease[c]	*ATP7A*
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*
Metachromatic leukodystrophy, ARSA-related	*ARSA*
Metachromatic leukodystrophy due to saposin B deficiency	*PSAP*
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*
Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*
Methylmalonic aciduria, MMAA-related	*MMAA*
Methylmalonic aciduria, MMAB-related	*MMAB*
Methylmalonic aciduria, MMUT-related	*MMUT*
Methylmalonyl-CoA epimerase deficiency	*MCEE*
Mevalonic aciduria / Hyper-IgD syndrome	*MVK*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

DISEASE	GENE
Normophosphatemic familial tumoral calcinosis	*SAMD9*
NPHP1 nephronophthisis-related ciliopathies	*NPHP1*
NPHP3 nephronophthisis-related ciliopathies	*NPHP3*
NPHP4 nephronophthisis-related ciliopathies	*NPHP4*
Oculocutaneous albinism, type I	*TYR*
Oculocutaneous albinism, type II	*OCA2*
Oculocutaneous albinism, type IV	*SLC45A2*
Oculocutaneous albinism, type III	*TYRP1*
Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	*WNT10A*
Omenn syndrome	*DCLRE1C*
Orofaciodigital syndrome, type XIV	*C2CD3*
Ornithine aminotransferase deficiency	*OAT*
Ornithine transcarbamylase deficiency, X-linked[c]	*OTC*
Ornithine translocase deficiency	*SLC25A15*
Osteogenesis imperfecta, type VII	*CRTAP*
Osteogenesis imperfecta, type VIII	*P3H1*
Osteogenesis imperfecta, type XI	*FKBP10*
Osteogenesis imperfecta, type XIII	*BMP1*
Osteopetrosis, infantile malignant, TCIRG1-related	*TCIRG1*
Osteopetrosis, OSTM1-related	*OSTM1*
Parkinson disease 15	*FBXO7*
Pantothenate kinase-associated neurodegeneration	*PANK2*
Pendred syndrome	*SLC26A4*
Perlman syndrome	*DIS3L2*
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*
PGM3-congenital disorder of glycosylation / Immunodeficiency 23	*PGM3*
Phenylalanine hydroxylase deficiency	*PAH*
Phosphoglycerate dehydrogenase deficiency	*PHGDH*
PIGN-related disorders	*PIGN*
PLA2G6-associated neurodegeneration	*PLA2G6*
PLEKHG5-related disorders	*PLEKHG5*
PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome	*PLOD1*
PLP1-related disorders[c]	*PLP1*
POLG-related disorders	*POLG*
Pontocerebellar hypoplasia, type 1A	*VRK1*
Pontocerebellar hypoplasia, type 1B	*EXOSC3*
Pontocerebellar hypoplasia, type 2B	*TSEN2*
Pontocerebellar hypoplasia, type 2E	*VPS53*
Pontocerebellar hypoplasia, types 4 and 2A	*TSEN54*
Pontocerebellar hypoplasia, type 6	*RARS2*
Primary ciliary dyskinesia, CCDC39-related	*CCDC39*
Primary ciliary dyskinesia, CCDC103-related	*CCDC103*
Primary ciliary dyskinesia, DNAH5-related	*DNAH5*
Primary ciliary dyskinesia, DNAH11-related	*DNAH11*
Primary ciliary dyskinesia, DNAI1-related	*DNAI1*
Primary ciliary dyskinesia, DNAI2-related	*DNAI2*
Primary congenital glaucoma	*CYP1B1*
Primary hyperoxaluria, type I	*AGXT*
Primary hyperoxaluria, type II	*GRHPR*
Primary hyperoxaluria, type III	*HOGA1*
Progressive cerebello-cerebral atrophy	*SEPSECS*
Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	*ATP8B1*
Progressive familial intrahepatic cholestasis 2	*ABCB11*
Progressive familial intrahepatic cholestasis 3	*ABCB4*
Progressive familial intrahepatic cholestasis 4	*TJP2*
Progressive pseudorheumatoid dysplasia	*CCN6*
Prolidase deficiency	*PEPD*
Propionic acidemia, PCCA-related	*PCCA*
Propionic acidemia, PCCB-related	*PCCB*
PRPS1-related disorders[c]	*PRPS1*
Pseudocholinesterase deficiency	*BCHE*
Pseudoxanthoma elasticum	*ABCC6*
Pterin-4 alpha-carbinolamine dehydratase (PCD) deficiency	*PCBD1*
Pycnodysostosis	*CTSK*
Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*
Pyridoxine-dependent epilepsy	*ALDH7A1*
Pyruvate carboxylase deficiency	*PC*
Pyruvate dehydrogenase E1-alpha deficiency[c]	*PDHA1*
Pyruvate dehydrogenase E1-beta deficiency	*PDHB*
Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	*TANGO2*
Refsum disease	*PHYH*
Renal tubular acidosis	*SLC4A4*
Renal tubular acidosis and deafness, ATP6V1B1-related	*ATP6V1B1*
Retinitis pigmentosa 2	*RP2*
Retinitis pigmentosa 3[c]	*RPGR*
Retinitis pigmentosa 25	*EYS*
Retinitis pigmentosa 26	*CERKL*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

DISEASE	GENE
Retinitis pigmentosa 28	*FAM161A*
Retinitis pigmentosa 36	*PRCD*
Retinitis pigmentosa 59	*DHDDS*
Retinitis pigmentosa 62	*MAK*
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*
Rhizomelic chondrodysplasia punctata, type 2	*GNPAT*
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*
RLBP1-related retinopathies	*RLBP1*
Roberts-SC phocomelia syndrome	*ESCO2*
RYR1-related disorders	*RYR1*
Sandhott disease	*HEXB*
Schimke immunoosseous dysplasia	*SMARCAL1*
Schindler disease	*NAGA*
Senior-Loken syndrome 5	*IQCB1*
Sepiapterin reductase deficiency	*SPR*
Severe combined immunodeficiency, CD3D-related	*CD3D*
Severe combined immunodeficiency, CD3E-related	*CD3E*
Severe combined immunodeficiency, FOXN1-related	*FOXN1*
Severe combined immunodeficiency, IKBKB-related	*IKBKB*
Severe combined immunodeficiency, IL7R-related	*IL7R*
Severe combined immunodeficiency, JAK3-related	*JAK3*
Severe combined immunodeficiency, PTPRC-related	*PTPRC*
Severe combined immunodeficiency, RAG1-related	*RAG1*
Severe combined immunodeficiency, RAG2-related	*RAG2*
Severe combined immunodeficiency, X-linked[c]	*IL2RG*
Severe congenital neutropenia 5	*VPS45*
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*
Shwachman-Diamond syndrome	*SBDS*
Sialidosis	*NEU1*
Sjögren-Larsson syndrome	*ALDH3A2*
Skeletal dysplasia, SLC26A2-related	*SLC26A2*
Smith-Lemli-Opitz syndrome	*DHCR7*
Spastic ataxia, Charlevoix-Saguenay type	*SACS*
Spastic paraplegia, type 15	*ZFYVE26*
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	*SLC1A4*
SPG11-related disorders	*SPG11*
Spinal muscular atrophy	*SMN1*
Spondylocostal dysostosis 1	*DLL3*
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*
Steel syndrome	*COL27A1*
Steroid resistant nephrotic syndrome, type 1	*NPHS1*
Steroid-resistant nephrotic syndrome, type 2	*NPHS2*
Steroid-resistant nephrotic syndrome, type 3	*PLCE1*
Stuve-Wiedemann syndrome	*LIFR*
SURF1-related disorders	*SURF1*
Surfactant dysfunction, ABCA3-related	*ABCA3*
Tay-Sachs disease	*HEXA*
TBCE-related disorders	*TBCE*
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*
Thyroid dyshormonogenesis 1	*SLC5A5*
Thyroid dyshormonogenesis 2A	*TPO*
Thyroid dyshormonogenesis 3	*TG*
Thyroid dyshormonogenesis 6	*DUOX2*
TK2-related mitochondrial disorders	*TK2*
TMEM67-related disorders	*TMEM67*
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*
Transcobalamin II deficiency	*TCN2*
TREX1-related disorders	*TREX1*
Trichohepatoenteric syndrome 1	*TTC37*
Trichohepatoenteric syndrome 2	*SKIC2*
TRIM32-related disorders	*TRIM32*
Trimethylaminuria	*FMO3*
Triple A syndrome	*AAAS*
TULP1-related disorders	*TULP1*
Tyrosine hydroxylase deficiency	*TH*
Tyrosinemia, type I	*FAH*
Tyrosinemia, type II	*TAT*
Tyrosinemia, type III	*HPD*
Usher syndrome, type 1B	*MYO7A*
Usher syndrome, type 1C	*USH1C*
Usher syndrome, type 1D	*CDH23*
Usher syndrome, type 1F	*PCDH15*
Usher syndrome, type 1J	*CIB2*
Usher syndrome, type 2A	*USH2A*
Usher syndrome, type 2C	*ADGRV1*
Usher syndrome, type 3A	*CLRN1*
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*
Vitamin D-dependent rickets, type 1A	*CYP27B1*
Vitamin D-resistant rickets, type 2A	*VDR*
VLDLR-associated cerebellar hypoplasia	*VLDLR*
Warsaw breakage syndrome	*DDX11*
Werner syndrome	*WRN*
Wilson disease	*ATP7B*
Wiskott-Aldrich syndrome, X-linked[c]	*WAS*
Wolcott-Rallison syndrome	*EIF2AK3*
Woodhouse-Sakati syndrome	*DCAF17*

[b] Panel components for males do not include specified X-linked diseases.
[c] Designated X-linked disease.

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QHerit 611 diseases[b] 612 genes (continued)

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

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

[b] Panel components for males do not include specified X-linked diseases.
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Test codes may vary by location. Please contact your local laboratory for more information.

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