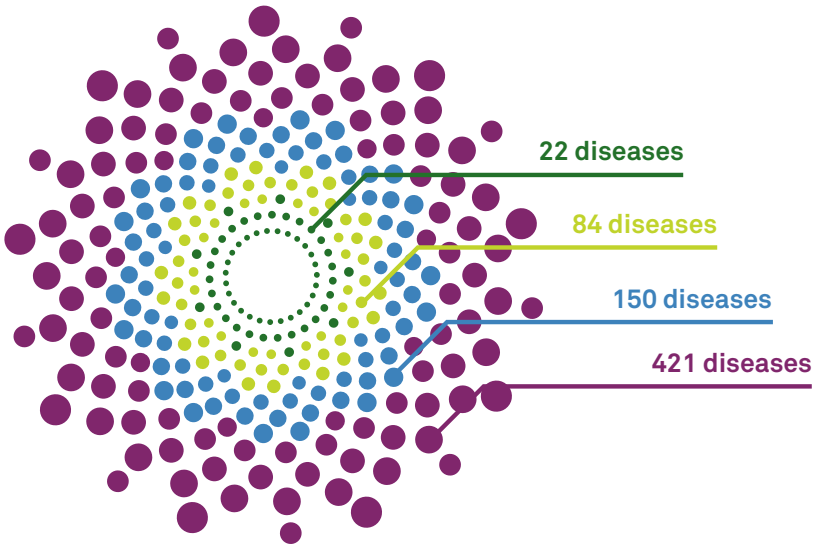
Quest Advanced® Women's Health

QHerit® carrier screening

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

As part of our commitment to provide clinically relevant, right-sized solutions, our **QHerit® product portfolio** provides medically appropriate and comprehensive insights to help you understand your patient's genetic risks so that together you can make informed decisions for their family planning.



An ideal panel for each patient

Our QHerit panels are designed with the American College of Obstetricians and Gynecologists (ACOG) guidelines, and the American College of Medical Genetics and Genomics (ACMG) practice resource in mind.

QHerit panel options[a]				
Panel size:	**22 diseases**[b] 24 genes	**84 diseases**[b] 85 genes	**150 diseases**[b] 150 genes	**421 diseases**[b] 421 genes
Screens for:	**The most common diseases,** including cystic fibrosis (CF), spinal muscular atrophy (SMA), fragile X, and Tay-Sachs	A mid-size panel that expands upon the most common diseases	Expands upon our mid-sized panel with medically guided screening	Our **most comprehensive panel,** offering the greatest insights
Test code:	94372	Female: 39867 Male: 39988[b]	Female: 39866 Male: 39987[b]	Female: 12593 Male: 12594[b]

[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.

Genetic testing should be accessible and affordable

Your patients won't be surprised by out-of-pocket costs when they choose QHerit.

Quest Diagnostics® is in-network with most health plans nationwide. If a patient is enrolled with an in-network health plan, patient responsibility is limited to $300 when not covered for QHerit.

QHerit No Surprise program determines prior authorization requirements and patient coverage. If Quest estimates that the patient will receive a bill of over $300, Quest will notify you and/or your patient. If you and/or your patient are not notified, the patient will owe no more than $300.

QHerit Supplemental Financial Assistance program is available for both insured and uninsured patients who qualify.

[c] Based on Quest Diagnostics 2021 fiscal year national claims analysis.

80%

of insured patients pay less than $25[c]

Relevant results, confident care*

QHerit carrier screens provide the insights you and your patients need to optimize family planning and care decisions.

Clinically relevant

Screens for ACOG guideline-recommended carefully selected set of diseases

- 4 testing options—up to 421 genes—empower you to select a medically appropriate panel
- Identifies genetic diseases regardless of ethnic group

Actionable information

Screens only for diseases that have actionable medical recommendations

- Next-generation sequencing helps to ensure accuracy across a greater number of genes*
- 1:1 consultations with board-certified genetic counselors are available to support your test selection and results interpretation

Patient-focused

Provides medically appropriate and accessible information

- Appropriate for all women and couples, regardless of ethnicity
- In-network coverage with a majority of insurance carriers
- Up-front patient coverage estimates and financial assistance available

*See important information on back page.

Consultation available on genetic test selection and results interpretation: 1.866.GENE.INFO (1.866.436.3463)

Diseases tested by QHerit® carrier screen panels

421 diseases[b] (421 genes)
Test code, female: 12593 Test code, male: 12594

150 diseases[b] (150 genes)
Test code, female: 39866 Test code, male: 39987

84 diseases[b] (85 genes)
Test code, female: 39867 Test code, male: 39988

22 diseases[b] (24 genes)
Test code: 94372

This panel analyzes 24 genes associated with 22 diseases. Includes the most common conditions across all ethnicities: CF, SMA, fragile X, and Tay-Sachs.

- Alpha-thalassemia *(HBA1/HBA2)*
- Beta hemoglobinopathies *(HBB)*
- Bloom syndrome *(BLM)*
- Canavan disease *(ASPA)*
- Cystic fibrosis *(CFTR)*
- Dihydrolipoamide dehydrogenase deficiency *(DLD)*
- Familial dysautonomia *(ELP1* (AKA: *IKBKAP))*
- Familial hyperinsulinism, ABCC8-related *(ABCC8)*
- Fanconi anemia, Group C *(FANCC)*
- Fragile X syndrome[d] *(FMR1)*
- Gaucher disease *(GBA)*
- Glycogen storage disease, type Ia *(G6PC* (AKA: *G6PC1))*
- 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)*
- Fukuyama congenital muscular dystrophy (Walker-Warburg) *(FKTN)*

This panel analyzes 85 genes associated with 84 diseases, including all genes in the 24-gene panel.

- Abetalipoproteinemia *(MTTP)*
- Adrenoleukodystrophy, X-linked[d] *(ABCD1)*
- Argininosuccinic aciduria *(ASL)*
- Ataxia-telangiectasia *(ATM)*
- Autosomal recessive polycystic kidney disease *(PKHD1)*
- Bardet-Biedl syndrome 1 *(BBS1)*
- Bardet-Biedl syndrome 2 *(BBS2)*
- Biotinidase deficiency *(BTD)*
- Carnitine palmitoyltransferase II deficiency *(CPT2)*
- Cerebrotendinous xanthomatosis *(CYP27A1)*
- Citrullinemia, type I *(ASS1)*
- Combined pituitary hormone deficiency, type 2 *(PROP1)*
- Congenital amegakaryocytic thrombocytopenia *(MPL)*
- Congenital disorder of glycosylation, type Ia *(PMM2)*
- Cystinosis *(CTNS)*
- D-bifunctional protein deficiency *(HSD17B4)*
- Factor XI deficiency / Hemophilia C *(F11)*
- Familial Mediterranean fever *(MEFV)*
- Galactosemia *(GALT)*
- Glutaric acidemia, type I *(GCDH)*
- Glycogen storage disease, type II / Pompe disease *(GAA)*
- Glycogen storage disease, type III *(AGL)*
- Glycogen storage disease, type IV / Adult polyglucosan body disease *(GBE1)*
- GRACILE syndrome *(BCS1L)*
- Hereditary fructose intolerance *(ALDOB)*
- Hermansky-Pudlak syndrome, type 1 *(HPS1)*
- Hermansky-Pudlak syndrome, type 3 *(HPS3)*
- Hypophosphatasia *(ALPL)*
- Krabbe disease *(GALC)*
- Limb-girdle muscular dystrophy, type 2A *(CAPN3)*
- Limb-girdle muscular dystrophy, type 3 *(SGCA)*
- Long chain 3-hydroxyacyl-coa dehydrogenase deficiency *(HADHA)*
- Maple syrup urine disease, type 1A *(BCKDHA)*
- Medium chain acyl-CoA dehydrogenase deficiency *(ACADM)*
- Metachromatic leukodystrophy, ARSA-related *(ARSA)*
- Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency *(MMACHC)*
- Mucolipidosis II and mucolipidosis III alpha/beta *(GNPTAB)*
- Mucopolysaccharidosis, type I / Hurler syndrome *(IDUA)*
- Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A *(SGSH)*
- Steroid resistant nephrotic syndrome, type 1 *(NPHS1)*
- Neuronal ceroid lipofuscinosis, CLN3-related *(CLN3)*
- Neuronal ceroid lipofuscinosis, CLN5-related *(CLN5)*
- Neuronal ceroid lipofuscinosis, CLN8-related *(CLN8)*
- Neuronal ceroid lipofuscinosis, PPT1-related *(PPT1)*
- Neuronal ceroid lipofuscinosis, TPP1-related *(TPP1)*
- Nijmegen breakage syndrome *(NBN)*
- Nonsyndromic hearing loss and deafness (DFNB) 1 *(GJB2)*
- Ornithine transcarbamylase deficiency, X-linked[d] *(OTC)*
- Pendred syndrome *(SLC26A4)*
- Phenylalanine hydroxylase deficiency *(PAH)*
- Primary hyperoxaluria, type I *(AGXT)*
- Propionic acidemia, PCCA-related *(PCCA)*
- Propionic acidemia, PCCB-related *(PCCB)*
- Sjögren-Larsson syndrome *(ALDH3A2)*
- Skeletal dysplasias, SLC26A2-related *(SLC26A2)*
- Smith-Lemli-Opitz syndrome *(DHCR7)*
- Carnitine deficiency, systemic primary *(SLC22A5)*
- Tyrosinemia, type I *(FAH)*
- Usher syndrome, type 2A *(USH2A)*
- Very long-chain acyl-CoA dehydrogenase deficiency *(ACADVL)*
- Wilson disease *(ATP7B)*
- Zellweger spectrum disorders, PEX2-related *(PEX2)*

This panel analyzes 150 genes asso
including all genes in the 85-gene p

- Beta-ketothiolase deficiency *(ACAT1)*
- 3-methylcrotonyl-CoA carboxylase 1 deficiency *(MCCC1)*
- 3-methylcrotonyl-CoA carboxylase 2 deficiency *(MCCC2)*
- 6-pyruvoyl-tetrahydropterin synthase deficiency *(PTS)*
- Adenosine deaminase deficiency *(ADA)*
- Alpha-mannosidosis *(MAN2B1)*
- Alport syndrome, COL4A3-related *(COL4A3)*
- Alport syndrome, COL4A4-related *(COL4A4)*
- Alport syndrome, COL4A5-related, X-linked[d] *(COL4A5)*
- Agenesis of the corpus callosum with peripheral neuropathy *(SLC12A6)*
- Arthrogryposis, mental retardation, and seizures *(SLC35A3)*
- Aspartylglycosaminuria *(AGA)*
- Menkes disease[d] *(ATP7A)*
- Autoimmune polyglandular syndrome, type 1 *(AIRE)*
- Spastic ataxia, Charlevoix-Saguenay type *(SACS)*
- Bardet-Biedl syndrome 10 *(BBS10)*
- Cartilage-hair hypoplasia *(RMRP)*
- LAMA2 muscular dystrophy *(LAMA2)*
- Nonsyndromic hearing loss and deafness (DFNB) 77 *(LOXHD1)*
- Dyskeratosis congenita, RTEL1-related *(RTEL1)*
- Familial hyperinsulinism, KCNJ11-related *(KCNJ11)*
- Fanconi anemia, Group A *(FANCA)*
- Glycine encephalopathy, AMT-related *(AMT)*
- Glycine encephalopathy / Nonketotic hyperglycinemia *(GLDC)*
- Glycogen storage disease, type Ib / IIw *(SLC37A4)*
- GLB1-related disorders *(GLB1)*
- 3-hydroxy-3-methylglutaryl-coA lyase deficiency *(HMGCL)*
- Holocarboxylase synthetase deficiency *(HLCS)*
- Homocystinuria, CBS-related *(CBS)*
- Hydrolethalus syndrome *(HYLS1)*
- GNE myopathy *(GNE)*
- Infantile cerebral and cerebellar atrophy *(MED17)*
- Isovaleric acidemia *(IVD)*
- Junctional epidermolysis bullosa, LAMA3-related *(LAMA3)*

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

While we offer a comprehensive testing menu, some patients may have an interest in screening for a specific disorder, such as cystic fibrosis. For these patients, Quest Diagnostics offers single-gene screening. Consultation available on genetic test selection and results interpretation 1.866.GENE.INFO (1.866.436.3463).

Please note that Quest offers a variety of single-gene and gene panel testing. For the genetic panel noted in this document, there may be single-gene tests or smaller panels that may be applicable for your patient. Refer to the Quest Diagnostics Test Directory for further information: https://TestDirectory.QuestDiagnostics.com

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- Junctional epidermolysis bullosa, LAMB3-related *(LAMB3)*
- Junctional epidermolysis bullosa, LAMC2-related *(LAMC2)*
- Autosomal recessive congenital ichthyosis 1 *(TGM1)*
- CEP290-related conditions *(CEP290)*
- Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type *(LRPPRC)*
- Lethal congenital contracture syndrome 1 *(GLE1)*
- Limb-girdle muscular dystrophy, type 4 *(SGCB)*
- Lysinuric protein intolerance *(SLC7A7)*
- Maple syrup urine disease, type 2 *(DBT)*
- Methylmalonic aciduria, MMAA-related *(MMAA)*
- Methylmalonic aciduria, MMAB-related *(MMAB)*
- Methylmalonic aciduria, MUT-related *(MMUT* (AKA: *MUT))*
- Mucopolysaccharidosis, type II / Hunter syndrome[d] *(IDS)*
- 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)*
- Neuronal ceroid lipofuscinosis, CLN6-related *(CLN6)*
- Niemann-Pick disease, type C1 *(NPC1)*
- Pyruvate carboxylase deficiency *(PC)*
- Retinitis pigmentosa 59 *(DHDDS)*
- Rhizomelic chondrodysplasia punctata, type 1 *(PEX7)*
- Sandhoff disease *(HEXB)*
- Spondylothoracic dysostosis and spondylocostal dysostosis 2 *(MESP2)*
- Steroid-resistant nephrotic syndrome, type 2 *(NPHS2)*
- Tyrosine hydroxylase deficiency *(TH)*
- Tyrosinemia, type II *(TAT)*
- Usher syndrome, type 1B *(MYO7A)*
- Usher syndrome, type 1C *(USH1C)*
- Usher syndrome, type 1D *(CDH23)*
- Zellweger spectrum disorders, PEX1-related *(PEX1)*
- Zellweger spectrum disorders, PEX6-related *(PEX6)*

This panel analyzes 421 genes associated with 421 diseases, including all genes in the 150-gene panel.

- 17-beta-hydroxysteroid dehydrogenase deficiency, type III *(HSD17B3)*
- 3-beta-hydroxysteroid dehydrogenase deficiency, type II *(HSD3B2)*
- 3-hydroxyacyl-CoA dehydrogenase deficiency *(HADH)*
- 3-methylglutaconic aciduria, type III / Costeff syndrome *(OPA3)*
- Phosphoglycerate dehydrogenase deficiency *(PHGDH)*
- Achromatopsia, CNGB3-related *(CNGB3)*
- Acrodermatitis enteropathica *(SLC39A4)*
- Action myoclonus renal failure syndrome *(SCARB2)*
- Acute infantile liver failure *(TRMU)*
- Peroxisomal acyl-CoA oxidase deficiency *(ACOX1)*
- Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency *(CYP11B1)*
- X-linked agammaglobulinemia[d] *(BTK)*
- 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-thalassemia intellectual disability syndrome, X-linked[d] *(ATRX)*
- Alstrom Syndrome *(ALMS1)*
- Amish infantile epilepsy syndrome *(ST3GAL5)*
- Argininemia *(ARG1)*
- Aromatase deficiency *(CYP19A1)*
- PRPS1-related disorders[d] *(PRPS1)*
- Asparagine synthetase deficiency *(ASNS)*
- Ataxia with isolated vitamin E deficiency *(TTPA)*
- Ataxia-telangiectasia-like disorder 1 *(MRE11)*
- 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 12 *(BBS12)*
- Bare lymphocyte syndrome, type II *(CIITA)*
- Barth syndrome[d] *(TAFAZZIN* (AKA: *TAZ))*
- Bartter syndrome, type 4A *(BSND)*
- Bernard-Soulier syndrome, type A *(GP1BA)*
- Bernard-Soulier syndrome, type C *(GP9)*
- Beta-ureidopropionase deficiency *(UPB1)*
- Bilateral frontoparietal polymicrogyria *(ADGRG1)*
- Carbamoyl phosphate synthetase I deficiency *(CPS1)*
- Carnitine palmitoyltransferase I deficiency *(CPT1A)*
- Carnitine-acylcarnitine translocase deficiency *(SLC25A20)*
- Carpenter syndrome *(RAB23)*
- Neuronal ceroid lipofuscinosis, CTSD-related *(CTSD)*
- Charcot-Marie-Tooth disease, type 1X[d] *(GJB1)*
- Charcot-Marie-Tooth disease, type 4D *(NDRG1)*
- Chediak-Higashi syndrome *(LYST)*
- Progressive familial intrahepatic cholestasis 1 (PFIC1) and benign familial intrahepatic cholestasis 1 (BRIC1) *(ATP8B1)*
- Cholestasis, progressive familial intrahepatic 4 *(TJP2)*
- Lysosomal acid lipase deficiency *(LIPA)*
- Choreoacanthocytosis *(VPS13A)*
- Choroideremia, X-linked[d] *(CHM)*
- Chronic granulomatous disease 4 *(CYBA)*
- Chronic granulomatous disease, X-linked[d] *(CYBB)*
- Ciliopathies, RPGRIP1L-related *(RPGRIP1L)*
- Citrin deficiency / Citrullinemia, type II *(SLC25A13)*
- Cockayne syndrome, type A *(ERCC8)*
- Cohen syndrome *(VPS13B)*
- Combined malonic and methylmalonic aciduria *(ACSF3)*
- Combined oxidative phosphorylation deficiency 3 *(TSFM)*
- Combined oxidative phosphorylation deficiency 1 *(GFM1)*
- Combined oxidative phosphorylation deficiency 6[d] *(AIFM1)*
- Combined pituitary hormone deficiency, type 3 *(LHX3)*
- Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency *(CYP17A1)*
- Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency *(CYP21A2)*
- Congenital disorder of glycosylation, type Ib *(MPI)*
- Congenital disorder of glycosylation, type Ic *(ALG6)*
- Congenital ichthyosis, ABCA12-related *(ABCA12)*
- Congenital insensitivity to pain with anhidrosis *(NTRK1)*
- Lipoid congenital adrenal hyperplasia *(STAR)*
- Congenital muscular dystrophy-dystroglycanopathy 1 *(POMT1)*
- Congenital myasthenic syndrome, CHAT-related *(CHAT)*
- Congenital myasthenic syndrome, CHRNE-related *(CHRNE)*
- Congenital myasthenic syndrome, DOK7-related *(DOK7)*
- Congenital myasthenic syndrome, RAPSN-related *(RAPSN)*
- Congenital neutropenia, HAX1-related *(HAX1)*
- Severe congenital neutropenia 5 *(VPS45)*
- Corneal dystrophy and perceptive deafness syndrome *(SLC4A11)*
- Corticosterone methyloxidase deficiency *(CYP11B2)*
- L1 syndrome[d] *(L1CAM)*
- CRB1-related retinal dystrophies *(CRB1)*
- Creatine Transporter Defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome[d] *(SLC6A8)*
- Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B *(ERCC6)*
- Usher syndrome, type 1J *(CIB2)*
- Dent disease[d] *(CLCN5)*
- Desbuquois dysplasia, type I *(CANT1)*
- Spastic tetraplegia, thin corpus callosum, and progressive microcephaly *(SLC1A4)*
- Dihydropyrimidine dehydrogenase deficiency *(DPYD)*
- Duchenne/Becker muscular dystrophy, X-linked[d] *(DMD)*
- Dyskeratosis congenita, X-linked[d] *(DKC1)*
- Dystrophic epidermolysis bullosa, COL7A1-related *(COL7A1)*
- Ehlers-Danlos syndrome, dermatosparaxis type *(ADAMTS2)*
- Ellis-van Creveld syndrome *(EVC2)*
- Ellis-van Creveld syndrome *(EVC)*
- Emery-Dreifuss muscular dystrophy, X-linked[d] *(EMD)*
- Enhanced S-cone syndrome *(NR2E3)*
- Ethylmalonic encephalopathy *(ETHE1)*
- Fabry disease, X-linked[d] *(GLA)*
- Familial hemophagocytic lymphohistiocytosis 2 *(PRF1)*
- Familial hemophagocytic lymphohistiocytosis 4 *(STX11)*
- Familial hemophagocytic lymphohistiocytosis 5 *(STXBP2)*
- Familial hypercholesterolemia, LDLRAP1-related *(LDLRAP1)*
- Familial hypercholesterolemia, LDLR-related *(LDLR)*
- Fanconi anemia, Group B[d] *(FANCB)*
- Fanconi anemia, Group D2 *(FANCD2)*
- Fanconi anemia, Group E *(FANCE)*
- Fanconi anemia, Group F *(FANCF)*
- Fanconi anemia, Group G *(FANCG)*
- Fanconi anemia, Group I *(FANCI)*
- Fanconi anemia, Group L *(FANCL)*
- Farber lipogranulomatosis *(ASAH1)*
- Fumarate hydratase deficiency *(FH)*
- GABA-transaminase deficiency *(ABAT)*
- Galactosemia, type II / Galactokinase deficiency *(GALK1)*
- Galactosialidosis *(CTSA)*
- Gitelman syndrome *(SLC12A3)*
- Glucose-6-phosphate dehydrogenase deficiency[d] *(G6PD)*
- Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA *(ETFA)*
- Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB *(ETFDH)*
- Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC *(ETFB)*
- Glycogen storage disease, type V *(PYGM)*
- Glycogen storage disease, type VII *(PFKM)*
- Guanidinoacetate methyltransferase deficiency *(GAMT)*
- Factor IX deficiency / Hemophilia B[d] *(F9)*

MPV17-related mitochondrial DNA (mtDNA) maintenance defect *(MPV17)*

Hereditary hemochromatosis, type 2 *(HJV)*

Hereditary hemochromatosis, type 3 *(TFR2)*

TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability *(TECPR2)*

Hermansky-Pudlak syndrome, type 2 *(AP3B1)*

Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency *(MTHFR)*

Homocystinuria, type cblE *(MTRR)*

HPRT1-related disorders[d] *(HPRT1)*

Hermansky-Pudlak syndrome, type 4 *(HPS4)*

Hyperphosphatemic familial tumoral calcinosis *(GALNT3)*

Hypohidrotic ectodermal dysplasia, X-linked[d] *(EDA)*

Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked[d] *(FOXP3)*

PLA2G6-associated neurodegeneration *(PLA2G6)*

X-linked infantile spinal muscular atrophy[d] *(UBA1)*

Johanson-Blizzard syndrome *(UBR1)*

Joubert syndrome 1 *(INPP5E)*

Joubert syndrome 15 *(CEP41)*

Joubert syndrome 21 *(CSPP1)*

Joubert syndrome 25 *(CEP104)*

Joubert syndrome 27 *(B9D1)*

Joubert syndrome 3 *(AHI1)*

Joubert syndrome 31 *(CEP120)*

Joubert syndrome 34 *(B9D2)*

Joubert syndrome 8 *(ARL13B)*

Nephronophthisis 2 *(INVS)*

NPHP1 nephronophthisis-related ciliopathies *(NPHP1)*

Juvenile retinoschisis, X-linked[d] *(RS1)*

Leber congenital amaurosis 5 *(LCA5)*

Leber congenital amaurosis 2 *(RPE65)*

Leber congenital amaurosis 13 *(RDH12)*

Leukoencephalopathy with vanishing white matter *(EIF2B5)*

Limb-girdle muscular dystrophy, type 2B *(DYSF)*

Limb-girdle muscular dystrophy, type 5 *(SGCG)*

Limb-girdle muscular dystrophy, type 6 *(SGCD)*

Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy 5 *(FKRP)*

Lipoprotein lipase deficiency *(LPL)*

X-linked developmental disorders, ARX-related[d] *(ARX)*

Lowe syndrome, X-linked[d] *(OCRL)*

Malonyl-CoA decarboxylase deficiency *(MLYCD)*

Joubert syndrome 9 *(CC2D2A)*

MEDNIK syndrome *(AP1S1)*

Megalencephalic leukoencephalopathy with subcortical cysts *(MLC1)*

Metachromatic leukodystrophy due to saposin B deficiency *(PSAP)*

Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency *(MMADHC)*

Micropthalmia / Anopthalmia *(VSX2)*

Mitochondrial complex I deficiency, nuclear type 16 *(NDUFAF5)*

Mitochondrial complex I deficiency, nuclear type 9 *(NDUFS6)*

Mitochondrial complex I deficiency, nuclear type 1 *(NDUFS4)*

Mitochondrial complex I deficiency, nuclear type 17 *(NDUFAF6)*

Mitochondrial complex IV deficiency, nuclear type 12 *(PET100)*

Myopathy, lactic acidosis, and sideroblastic anemia *(PUS1)*

Mitochondrial trifunctional protein deficiency, HADHB-related *(HADHB)*

MKS1-related disorders *(MKS1)*

Molybdenum cofactor deficiency of complementation group A *(MOCS1)*

Mucolipidosis III gamma *(GNPTG)*

Mucopolysaccharidosis, type IVA / Morquio syndrome *(GALNS)*

Mucopolysaccharidosis, type VII / Sly syndrome *(GUSB)*

Mucopolysaccharidosis, type IX / Hyaluronidase deficiency *(HYAL1)*

Mulibrey nanism *(TRIM37)*

Multiple pterygium syndrome, lethal type *(CHRNG)*

Multiple sulfatase deficiency *(SUMF1)*

Muscular dystrophy-dystroglycanopathy 3 *(POMGNT1)*

Muscular dystrophy-dystroglycanopathy 7 *(CRPPA)*

Muscular dystrophy-dystroglycanopathy 6 *(LARGE1)*

Muscular dystrophy-dystroglycanopathy 2 *(POMT2)*

Congenital myasthenic syndrome, COLQ-related *(COLQ)*

Mitochondrial neurogastrointestinal encephalopathy *(TYMP)*

X-linked myotubular myopathy[d] *(MTM1)*

Nephrogenic diabetes insipidus *(AQP2)*

Steroid-resistant nephrotic syndrome, type 3 *(PLCE1)*

Neuronal ceroid lipofuscinosis, MFSD8-related *(MFSD8)*

Niemann-Pick disease, type C2 *(NPC2)*

N-acetylglutamate synthase deficiency *(NAGS)*

Nonsyndromic hearing loss and deafness (DFNB) 3 *(MYO15A)*

Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome *(WNT10A)*

Omenn syndrome *(DCLRE1C)*

Severe combined immunodeficiency, RAG2-related *(RAG2)*

Ornithine aminotransferase deficiency *(OAT)*

Ornithine translocase deficiency *(SLC25A15)*

Joubert syndrome 17 *(CPLANE1)*

Orofaciodigital syndrome XIV *(C2CD3)*

Osteopetrosis, infantile malignant, TCIRG1-related *(TCIRG1)*

Perlman syndrome *(DIS3L2)*

Zellweger spectrum disorders, PEX12-related *(PEX12)*

POLG-related disorders *(POLG)*

Pontocerebellar hypoplasia, type 1B *(EXOSC3)*

Pontocerebellar hypoplasia, type 2B *(TSEN2)*

Pontocerebellar hypoplasia, type 4 and 2A *(TSEN54)*

Pontocerebellar hypoplasia, type 6 *(RARS2)*

Pontocerebellar hypoplasia, type 2E *(VPS53)*

Pontocerebellar hypoplasia, type 1A *(VRK1)*

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 II *(GRHPR)*

Primary hyperoxaluria, type III *(HOGA1)*

Progressive cerebello-cerebral atrophy *(SEPSECS)*

Progressive familial intrahepatic cholestastasis, type 2 *(ABCB11)*

Prolidase deficiency *(PEPD)*

Pseudocholinesterase deficiency *(BCHE)*

Pseudoxanthoma elasticum *(ABCC6)*

Pycnodysostosis *(CTSK)*

Pyridoxine-dependent epilepsy *(ALDH7A1)*

Pyruvate dehydrogenase E1-alpha deficiency[d] *(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)*

NPHP3 nephronophthisis-related ciliopathies *(NPHP3)*

Retinitis pigmentosa 25 *(EYS)*

Retinitis pigmentosa 26 *(CERKL)*

Retinitis pigmentosa 28 *(FAM161A)*

Rhizomelic chondrodysplasia punctata, type 2 *(GNPAT)*

Rhizomelic chondrodysplasia punctata, type 3 *(AGPS)*

Mitochondrial complex I deficiency, ACAD9-related *(ACAD9)*

Roberts-SC phocomelia syndrome *(ESCO2)*

Free sialic acid storage disorders *(SLC17A5)*

Schimke immunoosseous dysplasia *(SMARCAL1)*

NPHP4 nephronophthisis-related ciliopathies *(NPHP4)*

Senior-Loken syndrome 5 *(IQCB1)*

Severe combined immunodeficiency, X-linked[d] *(IL2RG)*

Shwachman-Diamond syndrome *(SBDS)*

Sialidosis *(NEU1)*

Spastic paraplegia, type 15 *(ZFYVE26)*

WWOX deficiency *(WWOX)*

Steel syndrome *(COL27A1)*

Stuve-Wiedemann syndrome *(LIFR)*

Severe combined immunodeficiency, RAG1-related *(RAG1)*

Trichohepatoenteric syndrome 1 *(TTC37)*

ERCC2-related conditions *(ERCC2)*

Triple A syndrome *(AAAS)*

Usher syndrome, type 2C *(ADGRV1)*

Vitamin D-dependent rickets, type 1A *(CYP27B1)*

Werner syndrome *(WRN)*

Wiskott-Aldrich syndrome, X-linked[d] *(WAS)*

Wolcott-Rallison syndrome *(EIF2AK3)*

Xeroderma pigmentosum, group A *(XPA)*

Xeroderma pigmentosum, group C *(XPC)*

X-linked chondrodysplasia punctata 1[d] *(ARSL)*

X-linked congenital adrenal hypoplasia[d] *(NR0B1)*

X-linked heterotaxy-1[d] *(ZIC3)*

X-Linked Hyper IgM Syndrome[d] *(CD40LG)*

DCX-related disorders[d] *(DCX)*

Zellweger spectrum disorders, PEX26-related *(PEX26)*

Zellweger spectrum disorders, PEX10-related *(PEX10)*

A single-source laboratory solution for family planning, from preconception through delivery

At Quest Advanced® Women's Health, our team is committed to helping you do more to support your patients along the reproductive journey.

3,500+ tests

including a portfolio of solutions throughout the reproductive journey: fertility testing, carrier screening, prenatal screening and diagnostic insights, and lab tests for each trimester

In-network for >90% of lives nationwide[e]

and preferred lab network status with major health plans including Aetna®, Horizon®, and UnitedHealthcare®

650+ MDs PhDs

and dozens of genetic counselors to help with test selection and results interpretation, making testing more actionable

40+ years of experience

in genetic testing, so you can be confident in our solutions and services to help meet your patient's needs

[e]This is directional data. It is based on 2020 HealthLeaders membership data of private third-party payers at the Managed Care Organization (MCO) level, as well as Quest internal data. Information is believed to be accurate as of January 1, 2020; however, it is subject to change. Does not include Kaiser Permanente access data.

Quest Advanced Women's Health

Delivering care for all stages of a woman's life requires testing that you can rely on for the insights you need to make informed health decisions. Quest Advanced Women's Health makes testing more actionable and accessible to support you, your patients, and your patients' families.*

Helpful resources are available at QHerit.com. Contact a genetic counselor at 1.866.GENE.INFO (1.866.436.3463) or GeneInfo@QuestDiagnostics.com

Important Information

* QHerit, QHerit Plus, QHerit Extended, QHerit 421 and QHerit 381 are carrier "screening" tests, and they screen for variations in genes linked to certain health disorders, which can be passed from parents to children. QHerit screens 24 genes; QHerit Plus screens 85 genes, QHerit Extended screens 150 genes, QHerit 421 screens 421 genes and QHerit 381 screens 381 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.

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

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