QHerit® Gene Crosswalk

Quest Diagnostics offers a range of QHerit® carrier screening panel options to help you select a medically appropriate panel based on your patient's needs. Below is detailed information for each panel, including genes and associated genetic conditions. All conditions follow an autosomal recessive pattern of inheritance, except for those noted as X-linked in the condition name. Consultation is available on genetic test selection and results interpretation. Call 1.866.GENE.INFO (1.866.436.3463) Monday through Friday 8:30 AM to 8:00 PM ET to speak with a Genomic Science Specialist.

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Cystic fibrosis	*CFTR*	South Asian	1 in 90	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	Southeast Asian	1 in 90	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	East Asian	1 in 90	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	African American	1 in 61	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	Hispanic	1 in 46	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	Northern European Caucasian	1 in 25	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	Ashkenazi Jewish	1 in 25	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	Caucasian	1 in 25	99%	99%	•	•	•	•	•	•	•
Cystic fibrosis	*CFTR*	General Population	1 in 25	99%	99%	•	•	•	•	•	•	•
Duchenne/Becker muscular dystrophy, X-linked	*DMD*	General Population	1 in 4200	99%	1 in 419,901	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	Asian	<1 in 500	99%	<1 in 49,901	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	Hispanic	<1 in 500	99%	<1 in 49,901	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	African American	1 in 251	99%	1 in 25,001	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	General Population	1 in 250	99%	1 in 24,901	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	Caucasian	1 in 178	99%	1 in 17,701	•	•	•	•	•	•	•
Fragile X syndrome	*FMR1*	Ashkenazi Jewish	1 in 58	99%	1 in 5,701	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Caucasian	<1 in 500	95%	<1 in 9,981	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	African American	1 in 30	95%	1 in 581	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	General Population	1 in 25	95%	1 in 481	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Asian	1 in 20	95%	1 in 381	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	East Asian	1 in 16	95%	1 in 301	•	•	•	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Southeast Asian	1 in 7	95%	1 in 121	•	•	•	•	•	•	•

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Alpha-thalassemia	*HBA1/HBA2*	South Asian	1 in 2	95%	1 in 21	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Caucasian	1 in 373	99%	1 in 37,201	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	General Population	1 in 129	99%	1 in 12,801	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Hispanic	1 in 83	99%	1 in 8,201	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	East Asian	1 in 78	99%	1 in 7,701	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Southern European Caucasian	1 in 59	99%	1 in 5,801	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Asian	1 in 54	99%	1 in 5,301	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	South Asian	1 in 32	99%	1 in 3,101	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Southeast Asian	1 in 30	99%	1 in 2,901	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Mediterranean	1 in 28	99%	1 in 2,701	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	African American	1 in 10	99%	1 in 901	•	•	•	•	•	•	•
Beta hemoglobinopathies	*HBB*	Middle Eastern	1 in 5	99%	1 in 401	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Hispanic	1 in 117	93%	1 in 11,601	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	African American	1 in 72	93%	1 in 7,101	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Hispanic American	1 in 68	93%	1 in 6,701	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Ashkenazi Jewish	1 in 67	93%	1 in 6,601	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	East Asian	1 in 59	93%	1 in 5,801	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	South Asian	1 in 59	93%	1 in 5,801	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Southeast Asian	1 in 59	93%	1 in 5,801	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Asian	1 in 59	93%	1 in 5,801	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	General Population	1 in 54	93%	1 in 5,301	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Northern European Caucasian	1 in 47	93%	1 in 4,601	•	•	•	•	•	•	•
Spinal muscular atrophy	*SMN1*	Caucasian	1 in 47	93%	1 in 4,601	•	•	•	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	General Population	1 in 112	99%	1 in 11,101		•	•	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	Ashkenazi Jewish	1 in 52	99%	1 in 5,101		•	•	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	Finnish	1 in 29	99%	1 in 2,801		•	•	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	Asian	1 in 178	99%	1 in 17,701		•	•	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	Caucasian	1 in 64	99%	1 in 6,301		•	•	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	General Population	1 in 35	99%	1 in 3,401		•	•	•	•	•	•
Canavan disease	*ASPA*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•

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Canavan disease	*ASPA*	Ashkenazi Jewish	1 in 60	99%	1 in 5,901		•	•	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Caucasian	1 in 433	99%	1 in 43,201		•	•	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	General Population	1 in 364	99%	1 in 36,301		•	•	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Asian	1 in 163	99%	1 in 16,201		•	•	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Ashkenazi Jewish	1 in 97	99%	1 in 9,601		•	•	•	•	•	•
Bloom syndrome	*BLM*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Bloom syndrome	*BLM*	Ashkenazi Jewish	1 in 140	99%	1 in 13,901		•	•	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	General Population	<1 in 500	99%	99%		•	•	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	Ashkenazi Jewish	1 in 120	99%	99%		•	•	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	Finnish	1 in 70	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Asian	<1 in 500	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	General Population	1 in 100	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	African American	1 in 93	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Northern European Caucasian	1 in 50	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Caucasian	1 in 50	99%	99%		•	•	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Ashkenazi Jewish	1 in 36	99%	99%		•	•	•	•	•	•
Dihydrolipoamide dehydrogenase deficiency	*DLD*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Dihydrolipoamide dehydrogenase deficiency	*DLD*	Ashkenazi Jewish	1 in 94	99%	1 in 9,301		•	•	•	•	•	•
Familial dysautonomia	*ELP1*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Familial dysautonomia	*ELP1*	Ashkenazi Jewish	1 in 34	99%	1 in 3,301		•	•	•	•	•	•
Fanconi anemia, complementation group C	*FANCC*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Fanconi anemia, complementation group C	*FANCC*	Ashkenazi Jewish	1 in 98	99%	1 in 9,701		•	•	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	Japanese	1 in 188	99%	1 in 18,701		•	•	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	Ashkenazi Jewish	1 in 150	99%	1 in 14,901		•	•	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Asian	1 in 192	99%	1 in 19,101		•	•	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	General Population	1 in 177	99%	1 in 17,601		•	•	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Caucasian	1 in 177	99%	1 in 17,601		•	•	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Ashkenazi Jewish	1 in 71	99%	1 in 7,001		•	•	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Ashkenazi Jewish	1 in 71	99%	1 in 7,001		•	•	•	•	•	•

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Galactosemia	*GALT*	East Asian	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Galactosemia	*GALT*	Hispanic	1 in 305	99%	1 in 30,401		•	•	•	•	•	•
Galactosemia	*GALT*	Ashkenazi Jewish	1 in 172	99%	1 in 17,101		•	•	•	•	•	•
Galactosemia	*GALT*	General Population	1 in 110	99%	1 in 10,901		•	•	•	•	•	•
Galactosemia	*GALT*	Northern European Caucasian	1 in 108	99%	1 in 10,701		•	•	•	•	•	•
Galactosemia	*GALT*	Caucasian	1 in 108	99%	1 in 10,701		•	•	•	•	•	•
Galactosemia	*GALT*	African American	1 in 78	99%	1 in 7,701		•	•	•	•	•	•
Galactosemia	*GALT*	Irish Travellers	1 in 11	99%	1 in 1,001		•	•	•	•	•	•
Gaucher disease	*GBA*	Caucasian	1 in 164	95%	1 in 3,261		•	•	•	•	•	•
Gaucher disease	*GBA*	General Population	1 in 153	95%	1 in 3,041		•	•	•	•	•	•
Gaucher disease	*GBA*	Ashkenazi Jewish	1 in 18	95%	1 in 341		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	African American	1 in 271	99%	1 in 27,001		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	General Population	1 in 250	99%	1 in 24,901		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	Caucasian	1 in 182	99%	1 in 18,101		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	Asian	1 in 126	99%	1 in 12,501		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	Sephardic Jewish–Moroccan, Iraqi	1 in 125	99%	1 in 12,401		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	French Canadian–Other	1 in 73	99%	1 in 7,201		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	Irish	1 in 41	99%	1 in 4,001		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	Ashkenazi Jewish	1 in 27	99%	1 in 2,601		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	French Canadian	1 in 13	99%	1 in 1,201		•	•	•	•	•	•
Tay-Sachs disease	*HEXA*	French Canadian–Gaspésie	1 in 13	99%	1 in 1,201		•	•	•	•	•	•
Mucolipidosis IV	*MCOLN1*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Mucolipidosis IV	*MCOLN1*	Ashkenazi Jewish	1 in 89	99%	1 in 8,801		•	•	•	•	•	•
Nemaline myopathy 2	*NEB*	General Population	1 in 224	95%	1 in 4,461		•	•	•	•	•	•
Nemaline myopathy 2	*NEB*	Ashkenazi Jewish	1 in 168	95%	1 in 3,341		•	•	•	•	•	•
Nemaline myopathy 2	*NEB*	Finnish	1 in 112	95%	1 in 2,221		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Finnish	1 in 225	99%	1 in 22,401		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Ashkenazi Jewish	1 in 225	99%	1 in 22,401		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Hispanic American	1 in 163	99%	1 in 16,201		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	African American	1 in 143	99%	1 in 14,201		•	•	•	•	•	•

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Phenylalanine hydroxylase deficiency	*PAH*	Asian	1 in 78	99%	1 in 7,701		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	General Population	1 in 65	99%	1 in 6,401		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Caucasian	1 in 50	99%	1 in 4,901		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Southern European Caucasian	1 in 40	99%	1 in 3,901		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Irish	1 in 34	99%	1 in 3,301		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Turkish	1 in 32	99%	1 in 3,101		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Sicilian	1 in 26	99%	1 in 2,501		•	•	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Sephardic Jewish–Iranian, Bukharan, Kavkazi, Tunisian, Moroccan	1 in 18	99%	1 in 1,701		•	•	•	•	•	•
Usher syndrome, type 1F	*PCDH15*	General Population	1 in 395	99%	1 in 39,401		•	•	•	•	•	•
Usher syndrome, type 1F	*PCDH15*	Ashkenazi Jewish	1 in 78	99%	1 in 7,701		•	•	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	General Population	1 in 144	99%	1 in 14,301		•	•	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Ashkenazi Jewish	1 in 106	99%	1 in 10,501		•	•	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Caucasian	1 in 100	99%	1 in 9,901		•	•	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	South African Afrikaner	1 in 52	99%	1 in 5,101		•	•	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	Caucasian	1 in 244	99%	1 in 24,301		•	•	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	General Population	1 in 196	99%	1 in 19,501		•	•	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	Ashkenazi Jewish	1 in 115	99%	1 in 11,401		•	•	•	•	•	•
Joubert syndrome 2	*TMEM216*	General Population	<1 in 500	99%	<1 in 49,901		•	•	•	•	•	•
Joubert syndrome 2	*TMEM216*	Ashkenazi Jewish	1 in 110	99%	1 in 10,901		•	•	•	•	•	•
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	General Population	1 in 116	99%	1 in 11,501			•	•	•	•	•
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	African American	1 in 68	99%	1 in 6,701			•	•	•	•	•
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	Caucasian	1 in 28	99%	1 in 2,701			•	•	•	•	•
Adrenoleukodystrophy, X-linked	*ABCD1*	General Population	1 in 10,500	99%	1 in 1,049,901			•	•	•	•	•
Adrenoleukodystrophy, X-linked	*ABCD1*	Sephardic Jewish	1 in 10,500	99%	1 in 1,049,901			•	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	Asian	1 in 194	99%	1 in 19,301			•	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	Caucasian	1 in 88	99%	1 in 8,701			•	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	General Population	1 in 86	99%	1 in 8,501			•	•	•	•	•
Beta-ketothiolase deficiency	*ACAT1*	Caucasian	1 in 354	99%	1 in 35,301			•	•	•	•	•
Beta-ketothiolase deficiency	*ACAT1*	General Population	1 in 347	99%	1 in 34,601			•	•	•	•	•

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Beta-ketothiolase deficiency	*ACAT1*	Asian	1 in 289	99%	1 in 28,801			•	•	•	•	•
Fragile XE syndrome, X-linked	*AFF2*	General Population	1 in 18,750	99%	1 in 1,874,901			•	•	•	•	•
Aspartylglycosaminuria	*AGA*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Aspartylglycosaminuria	*AGA*	Caucasian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Aspartylglycosaminuria	*AGA*	Finnish	1 in 36	99%	1 in 3,501			•	•	•	•	•
Primary hyperoxaluria, type 1	*AGXT*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Joubert syndrome 3	*AHI1*	General Population	1 in 176	99%	1 in 17,501			•	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	General Population	1 in 354	99%	1 in 35,301			•	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Finnish	1 in 79	99%	1 in 7,801			•	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Sardinian	1 in 60	99%	1 in 5,901			•	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Sephardic Jewish–Iranian	1 in 27	99%	1 in 2,601			•	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Hispanic	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	African American	1 in 406	99%	1 in 40,501			•	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Caucasian	1 in 80	99%	1 in 7,901			•	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	General Population	1 in 55	99%	1 in 5,401			•	•	•	•	•
Hypophosphatasia	*ALPL*	Northern European Caucasian	1 in 274	99%	1 in 27,301			•	•	•	•	•
Hypophosphatasia	*ALPL*	East Asian	1 in 203	99%	1 in 20,201			•	•	•	•	•
Hypophosphatasia	*ALPL*	Asian	1 in 203	99%	1 in 20,201			•	•	•	•	•
Hypophosphatasia	*ALPL*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Hypophosphatasia	*ALPL*	Mennonite	1 in 25	99%	1 in 2,401			•	•	•	•	•
Autosomal recessive spinocerebellar ataxia, type 10	*ANO10*	General Population	1 in 93	99%	1 in 9,201			•	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	General Population	1 in 100	99%	1 in 9,901			•	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Sephardic Jewish–Yemenite	1 in 46	99%	1 in 4,501			•	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Navajo	1 in 25	99%	1 in 2,401			•	•	•	•	•
X-linked developmental disorders, *ARX*-related	*ARX*	General Population	<1 in 750,000	50%	<1 in 1,499,999			•	•	•	•	•
Argininosuccinic aciduria	*ASL*	Finnish	1 in 190	99%	1 in 18,901			•	•	•	•	•
Argininosuccinic aciduria	*ASL*	General Population	1 in 133	99%	1 in 13,201			•	•	•	•	•
Wilson disease	*ATP7B*	Southern European Caucasian	1 in 250	99%	1 in 24,901			•	•	•	•	•
Wilson disease	*ATP7B*	Northern European Caucasian	1 in 90	99%	1 in 8,901			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Wilson disease	*ATP7B*	General Population	1 in 90	99%	1 in 8,901			•	•	•	•	•
Wilson disease	*ATP7B*	Caucasian	1 in 90	99%	1 in 8,901			•	•	•	•	•
Wilson disease	*ATP7B*	Ashkenazi Jewish	1 in 70	99%	1 in 6,901			•	•	•	•	•
Wilson disease	*ATP7B*	Sephardic Jewish–North African, Iraqi, Yemenite, Iranian, Bukharan	1 in 65	99%	1 in 6,401			•	•	•	•	•
Wilson disease	*ATP7B*	Sardinian	1 in 42	99%	1 in 4,101			•	•	•	•	•
Wilson disease	*ATP7B*	East Asian	1 in 27	99%	1 in 2,601			•	•	•	•	•
Wilson disease	*ATP7B*	Canary Islands	1 in 25	99%	1 in 2,401			•	•	•	•	•
Bardet-Biedl syndrome 1	*BBS1*	General Population	1 in 265	99%	1 in 26,401			•	•	•	•	•
Bardet-Biedl syndrome 1	*BBS1*	Faroese	1 in 30	99%	1 in 2,901			•	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	Ashkenazi Jewish	1 in 135	99%	1 in 13,401			•	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	Hutterite	1 in 22	99%	1 in 2,101			•	•	•	•	•
Biotinidase deficiency	*BTD*	General Population	1 in 120	99%	1 in 11,901			•	•	•	•	•
Biotinidase deficiency	*BTD*	Hispanic	1 in 30	99%	1 in 2,901			•	•	•	•	•
Biotinidase deficiency	*BTD*	Caucasian	1 in 12	99%	1 in 1,101			•	•	•	•	•
Homocystinuria, *CBS*-related	*CBS*	General Population	1 in 224	99%	1 in 22,301			•	•	•	•	•
Homocystinuria, *CBS*-related	*CBS*	Caucasian	1 in 52	99%	1 in 5,101			•	•	•	•	•
Homocystinuria, *CBS*-related	*CBS*	Qatari	1 in 21	99%	1 in 2,001			•	•	•	•	•
Joubert syndrome 9	*CC2D2A*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Congenital hydrocephalus 1	*CCDC88C*	General Population	1 in 137	99%	1 in 13,601			•	•	•	•	•
Leber congenital amaurosis, *CEP290*-related/*CEP290*-related conditions	*CEP290*	General Population	1 in 185	99%	99%			•	•	•	•	•
Congenital myasthenic syndrome, *CHRNE*-related	*CHRNE*	General Population	<1 in 500	99%	99%			•	•	•	•	•
Congenital myasthenic syndrome, *CHRNE*-related	*CHRNE*	Caucasian	1 in 383	99%	99%			•	•	•	•	•
Myotonia congenita	*CLCN1*	General Population	1 in 158	99%	99%			•	•	•	•	•
Myotonia congenita	*CLCN1*	Finnish	1 in 59	99%	99%			•	•	•	•	•
Myotonia congenita	*CLCN1*	Norwegian	1 in 53	99%	99%			•	•	•	•	•
Achromatopsia, *CNGB3*-related	*CNGB3*	General Population	1 in 98	99%	99%			•	•	•	•	•
Achromatopsia, *CNGB3*-related	*CNGB3*	Caucasian	1 in 91	99%	99%			•	•	•	•	•
Dystrophic epidermolysis bullosa, *COL7A1*-related	*COL7A1*	General Population	1 in 370	99%	99%			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Carnitine palmitoyltransferase II deficiency	*CPT2*	General Population	<1 in 500	99%	99%			•	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Asian	<1 in 500	99%	99%			•	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	African American	1 in 308	99%	99%			•	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Caucasian	1 in 200	99%	99%			•	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Ashkenazi Jewish	1 in 51	99%	99%			•	•	•	•	•
Congenital adrenal insufficiency, *CYP11A1*-related	*CYP11A1*	General Population	1 in 114	99%	99%			•	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	Caucasian	1 in 67	98%	98%			•	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	General Population	1 in 61	98%	98%			•	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	Ashkenazi Jewish	1 in 40	98%	98%			•	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	Southern European Caucasian	<1 in 500	99%	99%			•	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	General Population	1 in 115	99%	99%			•	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	Sephardic Jewish–Moroccan	1 in 5	99%	99%			•	•	•	•	•
Vitamin D-dependent rickets, type 1A	*CYP27B1*	General Population	1 in 22	99%	99%			•	•	•	•	•
Retinitis pigmentosa 59	*DHDDS*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Retinitis pigmentosa 59	*DHDDS*	Ashkenazi Jewish	1 in 117	99%	1 in 11,601			•	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	East Asian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	Japanese	1 in 297	99%	1 in 29,601			•	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	General Population	1 in 67	99%	1 in 6,601			•	•	•	•	•
ERCC2-related conditions	*ERCC2*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Ellis-van Creveld syndrome	*EVC2*	General Population	1 in 122	99%	1 in 12,101			•	•	•	•	•
Factor VIII deficiency/Hemophilia A, X-linked	*F8*	General Population	1 in 2000	95%	1 in 39,981			•	•	•	•	•
Factor IX deficiency/Hemophilia B, X-linked	*F9*	General Population	1 in 10,000	99%	1 in 999,901			•	•	•	•	•
Tyrosinemia, type I	*FAH*	Asian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Tyrosinemia, type I	*FAH*	African American	1 in 478	99%	1 in 47,701			•	•	•	•	•
Tyrosinemia, type I	*FAH*	Caucasian	1 in 333	99%	1 in 33,201			•	•	•	•	•
Tyrosinemia, type I	*FAH*	Ashkenazi Jewish	1 in 150	99%	1 in 14,901			•	•	•	•	•
Tyrosinemia, type I	*FAH*	General Population	1 in 100	99%	1 in 9,901			•	•	•	•	•
Tyrosinemia, type I	*FAH*	French Canadian–Québec	1 in 66	99%	1 in 6,501			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Tyrosinemia, type I	*FAH*	French Canadian–Saguenay-Lac-Saint-Jean	1 in 25	99%	1 in 2,401			•	•	•	•	•
Limb-girdle muscular dystrophy, type 2I/Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Limb-girdle muscular dystrophy, type 2I/Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*	Norwegian	1 in 116	99%	1 in 11,501			•	•	•	•	•
Trimethylaminuria	*FMO3*	General Population	1 in 139	99%	1 in 13,801			•	•	•	•	•
Trimethylaminuria	*FMO3*	Northern European Caucasian	1 in 100	99%	1 in 9,901			•	•	•	•	•
Friedreich ataxia	*FXN*	General Population	1 in 250	99%	1 in 24,901			•	•	•	•	•
Friedreich ataxia	*FXN*	Caucasian	1 in 80	99%	1 in 7,901			•	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	General Population	1 in 132	99%	1 in 13,101			•	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Asian	1 in 112	99%	1 in 11,101			•	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Caucasian	1 in 100	99%	1 in 9,901			•	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	African American	1 in 70	99%	1 in 6,901			•	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Ashkenazi Jewish	1 in 58	99%	1 in 5,701			•	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	General Population	1 in 387	99%	1 in 38,601			•	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	Caucasian	1 in 144	99%	1 in 14,301			•	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	Ashkenazi Jewish	1 in 68	99%	1 in 6,701			•	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	General Population	1 in 42	99%	1 in 4,101			•	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Caucasian	1 in 30	99%	1 in 2,901			•	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Ashkenazi Jewish	1 in 21	99%	1 in 2,001			•	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	East Asian	1 in 10	99%	1 in 901			•	•	•	•	•
Fabry disease, X-linked	*GLA*	General Population	1 in 1,500	99%	1 in 149,901			•	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	Asian	1 in 389	99%	1 in 38,801			•	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	Caucasian	1 in 225	99%	1 in 22,401			•	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Fraser syndrome, type 3	*GRIP1*	General Population	1 in 83	99%	1 in 8,201			•	•	•	•	•
Hermansky-Pudlak syndrome, type 1	*HPS1*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Hermansky-Pudlak syndrome, type 1	*HPS1*	Puerto Rican	1 in 59	99%	1 in 5,801			•	•	•	•	•
Hermansky-Pudlak syndrome, type 3	*HPS3*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Hermansky-Pudlak syndrome, type 3	*HPS3*	Ashkenazi Jewish	1 in 235	99%	1 in 23,401			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Mucopolysaccharidosis, type I/Hurler syndrome	*IDUA*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Mucopolysaccharidosis, type I/Hurler syndrome	*IDUA*	Northern European Caucasian	1 in 145	99%	1 in 14,401			•	•	•	•	•
L1 syndrome, X-linked	*L1CAM*	General Population	1 in 22,500	99%	1 in 2,249,901			•	•	•	•	•
Donnai-Barrow syndrome	*LRP2*	General Population	1 in 213	99%	1 in 21,201			•	•	•	•	•
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	General Population	1 in 120	99%	1 in 11,901			•	•	•	•	•
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	Caucasian	1 in 112	99%	1 in 11,101			•	•	•	•	•
Autosomal recessive primary microcephaly 1	*MCPH1*	Northern European Caucasian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Autosomal recessive primary microcephaly 1	*MCPH1*	General Population	1 in 146	99%	1 in 14,501			•	•	•	•	•
X-linked Opitz G/BBB syndrome	*MID1*	General Population	1 in 37,500	99%	1 in 3,749,901			•	•	•	•	•
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	Libyan Jewish	1 in 40	99%	1 in 3,901			•	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	General Population	1 in 138	99%	1 in 13,701			•	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Caucasian	1 in 138	99%	1 in 13,701			•	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Asian	1 in 113	99%	1 in 11,201			•	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	East Asian	1 in 112	99%	1 in 11,101			•	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	General Population	1 in 383	99%	1 in 38,201			•	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Hispanic	1 in 383	99%	1 in 38,201			•	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Caucasian	1 in 224	99%	1 in 22,301			•	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	African American	1 in 177	99%	1 in 17,601			•	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Asian	1 in 53	99%	1 in 5,201			•	•	•	•	•
Mevalonic aciduria/Hyper-IgD syndrome	*MVK*	General Population	1 in 167	99%	1 in 16,601			•	•	•	•	•
Schindler disease	*NAGA*	General Population	1 in 94	99%	1 in 9,301			•	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	General Population	1 in 325	99%	1 in 32,401			•	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	Finnish	1 in 45	99%	1 in 4,401			•	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	Groffdale Conference Mennonite	1 in 12	99%	1 in 1,101			•	•	•	•	•
X-linked congenital adrenal hypoplasia	*NR0B1*	General Population	1 in 52,500	99%	1 in 5,249,901			•	•	•	•	•
Oculocutaneous albinism, type II	*OCA2*	General Population	1 in 76	99%	1 in 7,501			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Oculocutaneous albinism, type II	*OCA2*	African American	1 in 50	99%	1 in 4,901			•	•	•	•	•
Oculocutaneous albinism, type II	*OCA2*	Navajo	1 in 22	99%	1 in 2,101			•	•	•	•	•
Ornithine transcarbamylase deficiency, X-linked	*OTC*	General Population	1 in 30,000	99%	1 in 2,999,901			•	•	•	•	•
PLP1-related disorders, X-linked	*PLP1*	Northern European Caucasian	1 in 57,700	99%	1 in 5,769,901			•	•	•	•	•
PLP1-related disorders, X-linked	*PLP1*	General Population	1 in 37,500	99%	1 in 3,749,901			•	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Asian	1 in 449	99%	1 in 44,801			•	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	General Population	1 in 124	99%	1 in 12,301			•	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Ashkenazi Jewish	1 in 61	99%	1 in 6,001			•	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Northern European Caucasian	1 in 60	99%	1 in 5,901			•	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Caucasian	1 in 42	99%	1 in 4,101			•	•	•	•	•
POLG-related disorders	*POLG*	General Population	1 in 50	99%	1 in 4,901			•	•	•	•	•
Familial hemophagocytic lymphohistiocytosis 2	*PRF1*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Pontocerebellar hypoplasia, type 6	*RARS2*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Pontocerebellar hypoplasia, type 6	*RARS2*	Sephardic Jewish–Iraqi, Syrian, Tunisian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Aicardi-Goutières syndrome 2	*RNASEH2B*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Retinitis pigmentosa 3, X-linked	*RPGR*	General Population	1 in 20,000	99%	1 in 1,999,901			•	•	•	•	•
Juvenile retinoschisis, X-linked	*RS1*	General Population	1 in 2,500	99%	1 in 249,901			•	•	•	•	•
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	General Population	1 in 149	99%	1 in 14,801			•	•	•	•	•
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*	Middle Eastern	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*	General Population	1 in 109	99%	1 in 10,801			•	•	•	•	•
Skeletal dysplasia, *SLC26A2*-related	*SLC26A2*	General Population	1 in 158	99%	1 in 15,701			•	•	•	•	•
Skeletal dysplasia, *SLC26A2*-related	*SLC26A2*	Finnish	1 in 50	99%	1 in 4,901			•	•	•	•	•
Pendred syndrome	*SLC26A4*	Caucasian	1 in 88	99%	1 in 8,701			•	•	•	•	•
Pendred syndrome	*SLC26A4*	General Population	1 in 80	99%	1 in 7,901			•	•	•	•	•
Pendred syndrome	*SLC26A4*	African American	1 in 76	99%	1 in 7,501			•	•	•	•	•
Pendred syndrome	*SLC26A4*	Asian	1 in 74	99%	1 in 7,301			•	•	•	•	•
Pendred syndrome	*SLC26A4*	Northern European Caucasian	1 in 60	99%	1 in 5,901			•	•	•	•	•
Glycogen storage disease, type Ib/IIw	*SLC37A4*	Caucasian	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Glycogen storage disease, type Ib/IIw	*SLC37A4*	General Population	1 in 354	99%	1 in 35,301			•	•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Creatine transporter defect, *SLC6A8*-related, X-linked/ Cerebral creatine deficiency syndrome	*SLC6A8*	General Population	1 in 20,600	99%	1 in 2,059,901			•	•	•	•	•
Atransferrinemia	*TF*	General Population	1 in 116	99%	1 in 11,501			•	•	•	•	•
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*	General Population	1 in 28	95%	1 in 541			•	•	•	•	•
Oculocutaneous albinism, type I	*TYR*	General Population	1 in 20	99%	1 in 1,901			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	French Canadian	1 in 207	99%	1 in 20,601			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	General Population	1 in 126	99%	1 in 12,501			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	East Asian	1 in 113	99%	1 in 11,201			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Northern European Caucasian	1 in 113	99%	1 in 11,201			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Caucasian	1 in 73	99%	1 in 7,201			•	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Sephardic Jewish–Iraqi, Iranian	1 in 36	99%	1 in 3,501			•	•	•	•	•
Xeroderma pigmentosum, group C	*XPC*	General Population	<1 in 500	99%	<1 in 49,901			•	•	•	•	•
Adenosine deaminase deficiency	*ADA*	General Population	1 in 224	99%	1 in 22,301				•	•	•	•
Glycogen storage disease, type III	*AGL*	General Population	1 in 159	99%	1 in 15,801				•	•	•	•
Glycogen storage disease, type III	*AGL*	Sephardic Jewish–Moroccan	1 in 37	99%	1 in 3,601				•	•	•	•
Glycogen storage disease, type III	*AGL*	Faroese	1 in 28	99%	1 in 2,701				•	•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	Northern European Caucasian	1 in 223	99%	1 in 22,201				•	•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	General Population	1 in 223	99%	1 in 22,201				•	•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	Swedish	1 in 204	99%	1 in 20,301				•	•	•	•
Glycine encephalopathy, *AMT*-related	*AMT*	Caucasian	1 in 271	99%	1 in 27,001				•	•	•	•
Glycine encephalopathy, *AMT*-related	*AMT*	General Population	1 in 262	99%	1 in 26,101				•	•	•	•
Mucopolysaccharidosis, type VI/Maroteaux-Lamy syndrome	*ARSB*	Asian	1 in 423	99%	1 in 42,201				•	•	•	•
Mucopolysaccharidosis, type VI/Maroteaux-Lamy syndrome	*ARSB*	General Population	1 in 291	99%	1 in 29,001				•	•	•	•
Mucopolysaccharidosis, type VI/Maroteaux-Lamy syndrome	*ARSB*	Caucasian	1 in 273	99%	1 in 27,201				•	•	•	•
Citrullinemia, type I	*ASS1*	Caucasian	1 in 195	99%	1 in 19,401				•	•	•	•
Citrullinemia, type I	*ASS1*	Asian	1 in 123	99%	1 in 12,201				•	•	•	•
Citrullinemia, type I	*ASS1*	General Population	1 in 119	99%	1 in 11,801				•	•	•	•
Ataxia-telangiectasia	*ATM*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901				•	•	•	•
Ataxia-telangiectasia	*ATM*	General Population	1 in 100	99%	1 in 9,901				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Ataxia-telangiectasia	*ATM*	Sephardic Jewish–Moroccan	1 in 81	99%	1 in 8,001				•	•	•	•
Menkes disease, X-linked	*ATP7A*	General Population	1 in 26,000	99%	1 in 2,599,901				•	•	•	•
Bardet-Biedl syndrome 10	*BBS10*	General Population	1 in 447	99%	1 in 44,601				•	•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	General Population	1 in 321	99%	1 in 32,001				•	•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Caucasian	1 in 320	99%	1 in 31,901				•	•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Portuguese Roma	1 in 71	99%	1 in 7,001				•	•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Mennonite	1 in 10	99%	1 in 901				•	•	•	•
GRACILE syndrome	*BCS1L*	Caucasian	1 in 407	99%	1 in 40,601				•	•	•	•
GRACILE syndrome	*BCS1L*	General Population	1 in 111	99%	1 in 11,001				•	•	•	•
GRACILE syndrome	*BCS1L*	Finnish	1 in 108	99%	1 in 10,701				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Hispanic	1 in 260	99%	1 in 25,901				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	East Asian	1 in 232	99%	1 in 23,101				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Northern European Caucasian	1 in 103	99%	1 in 10,201				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Caucasian	1 in 103	99%	1 in 10,201				•	•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Amish	1 in 50	99%	1 in 4,901				•	•	•	•
Usher syndrome, type 1D	*CDH23*	General Population	1 in 202	99%	1 in 20,101				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN3*-related	*CLN3*	Caucasian	1 in 188	99%	1 in 18,701				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN3*-related	*CLN3*	General Population	1 in 145	99%	1 in 14,401				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN5*-related	*CLN5*	General Population	1 in 317	99%	1 in 31,601				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN5*-related	*CLN5*	Finnish	1 in 289	99%	1 in 28,801				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN6*-related	*CLN6*	General Population	1 in 261	99%	1 in 26,001				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN8*-related	*CLN8*	General Population	1 in 349	99%	1 in 34,801				•	•	•	•
Neuronal ceroid lipofuscinosis, *CLN8*-related	*CLN8*	Finnish	1 in 135	99%	1 in 13,401				•	•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	General Population	1 in 323	99%	1 in 32,201				•	•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	Caucasian	1 in 284	99%	1 in 28,301				•	•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	Ashkenazi Jewish	1 in 189	99%	1 in 18,801				•	•	•	•
Alport syndrome, *COL4A4*-related	*COL4A4*	General Population	1 in 353	99%	1 in 35,201				•	•	•	•
Alport syndrome, *COL4A5*-related, X-linked	*COL4A5*	General Population	1 in 47,000	99%	1 in 4,699,901				•	•	•	•
Cystinosis	*CTNS*	African American	<1 in 500	99%	<1 in 49,901				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Cystinosis	*CTNS*	Asian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Cystinosis	*CTNS*	Hispanic	<1 in 500	99%	<1 in 49,901				•	•	•	•
Cystinosis	*CTNS*	Caucasian	1 in 220	99%	1 in 21,901				•	•	•	•
Cystinosis	*CTNS*	General Population	1 in 158	99%	1 in 15,701				•	•	•	•
Cystinosis	*CTNS*	Sephardic Jewish–Moroccan	1 in 100	99%	1 in 9,901				•	•	•	•
Cystinosis	*CTNS*	French Canadian–Saguenay-Lac-Saint-Jean	1 in 39	99%	1 in 3,801				•	•	•	•
Maple syrup urine disease, type 2	*DBT*	General Population	1 in 321	99%	1 in 32,001				•	•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Asian	1 in 163	99%	1 in 16,201				•	•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Caucasian	1 in 101	99%	1 in 10,001				•	•	•	•
Factor XI deficiency/Hemophilia C	*F11*	General Population	1 in 92	99%	1 in 9,101				•	•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Ashkenazi Jewish	1 in 11	99%	1 in 1,001				•	•	•	•
Fanconi anemia, complementation group A	*FANCA*	General Population	1 in 345	99%	1 in 34,401				•	•	•	•
Fanconi anemia, complementation group A	*FANCA*	Sephardic Jewish–Moroccan, Tunisian	1 in 133	99%	1 in 13,201				•	•	•	•
Fanconi anemia, complementation group A	*FANCA*	Spanish Roma	1 in 64	99%	1 in 6,301				•	•	•	•
Krabbe disease	*GALC*	Asian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Krabbe disease	*GALC*	General Population	1 in 150	99%	1 in 14,901				•	•	•	•
Krabbe disease	*GALC*	Druze–Northern Israel	1 in 6	99%	1 in 501				•	•	•	•
Krabbe disease	*GALC*	Muslim Arab–Jerusalem	1 in 6	99%	1 in 501				•	•	•	•
Glutaric acidemia, type I	*GCDH*	Caucasian	1 in 172	99%	1 in 17,101				•	•	•	•
Glutaric acidemia, type I	*GCDH*	General Population	1 in 112	99%	1 in 11,101				•	•	•	•
Glutaric acidemia, type I	*GCDH*	African American	1 in 36	99%	1 in 3,501				•	•	•	•
Glutaric acidemia, type I	*GCDH*	Lumbee Native American	1 in 16	99%	1 in 1,501				•	•	•	•
Glutaric acidemia, type I	*GCDH*	Lancaster County Amish	1 in 9	99%	1 in 801				•	•	•	•
Glutaric acidemia, type I	*GCDH*	Oji-Cree FirstNation–North Manitoba	1 in 8	99%	1 in 701				•	•	•	•
GLB1-related disorders	*GLB1*	Caucasian	1 in 278	99%	1 in 27,701				•	•	•	•
GLB1-related disorders	*GLB1*	General Population	1 in 158	99%	1 in 15,701				•	•	•	•
GLB1-related disorders	*GLB1*	South Brazil	1 in 65	99%	1 in 6,401				•	•	•	•
GLB1-related disorders	*GLB1*	Roma	1 in 50	99%	1 in 4,901				•	•	•	•
GLB1-related disorders	*GLB1*	Maltese	1 in 30	99%	1 in 2,901				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Glycine encephalopathy/Nonketotic hyperglycinemia	*GLDC*	Caucasian	1 in 140	99%	1 in 13,901				•	•	•	•
Glycine encephalopathy/Nonketotic hyperglycinemia	*GLDC*	General Population	1 in 135	99%	1 in 13,401				•	•	•	•
Lethal congenital contracture syndrome 1	*GLE1*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Lethal congenital contracture syndrome 1	*GLE1*	Finnish	1 in 100	99%	1 in 9,901				•	•	•	•
GNE myopathy	*GNE*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901				•	•	•	•
GNE myopathy	*GNE*	Caucasian	<1 in 500	99%	<1 in 49,901				•	•	•	•
GNE myopathy	*GNE*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
GNE myopathy	*GNE*	Asian	1 in 58	99%	1 in 5,701				•	•	•	•
GNE myopathy	*GNE*	Sephardic Jewish–Iranian, Syrian	1 in 12	99%	1 in 1,101				•	•	•	•
Mucopolysaccharidosis, type IIID/ Sanfilippo syndrome D	*GNS*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	Caucasian	1 in 254	99%	1 in 25,301				•	•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	Finnish	1 in 240	99%	1 in 23,901				•	•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	General Population	1 in 138	99%	1 in 13,701				•	•	•	•
Sandhoff disease	*HEXB*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901				•	•	•	•
Sandhoff disease	*HEXB*	General Population	1 in 278	99%	1 in 27,701				•	•	•	•
Sandhoff disease	*HEXB*	Caucasian	1 in 235	99%	1 in 23,401				•	•	•	•
Sandhoff disease	*HEXB*	Argentinian Creole	1 in 64	99%	1 in 6,301				•	•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	Asian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	General Population	1 in 482	99%	1 in 48,101				•	•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	Caucasian	1 in 259	99%	1 in 25,801				•	•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Caucasian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Asian	1 in 158	99%	1 in 15,701				•	•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Faroese	1 in 20	99%	1 in 1,901				•	•	•	•
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
D-bifunctional protein deficiency	*HSD17B4*	General Population	1 in 158	99%	1 in 15,701				•	•	•	•
Hydrolethalus syndrome	*HYLS1*	General Population	1 in 455	99%	1 in 45,401				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Hydrolethalus syndrome	*HYLS1*	Finnish	1 in 50	99%	1 in 4,901				•	•	•	•
Mucopolysaccharidosis, type II/Hunter syndrome, X-linked	*IDS*	General Population	1 in 60,000	90%	1 in 599,991				•	•	•	•
Isovaleric acidemia	*IVD*	General Population	1 in 250	99%	1 in 24,901				•	•	•	•
Isovaleric acidemia	*IVD*	Caucasian	1 in 144	99%	1 in 14,301				•	•	•	•
Isovaleric acidemia	*IVD*	Asian	1 in 75	99%	1 in 7,401				•	•	•	•
Familial hyperinsulinism, *KCNJ11*-related	*KCNJ11*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
LAMA2 muscular dystrophy	*LAMA2*	General Population	1 in 87	99%	1 in 8,601				•	•	•	•
Junctional epidermolysis bullosa, *LAMA3*-related	*LAMA3*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Junctional epidermolysis bullosa, *LAMB3*-related	*LAMB3*	General Population	1 in 407	99%	1 in 40,601				•	•	•	•
Junctional epidermolysis bullosa, *LAMC2*-related	*LAMC2*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Nonsyndromic hearing loss and deafness 77	*LOXHD1*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Nonsyndromic hearing loss and deafness 77	*LOXHD1*	Ashkenazi Jewish	1 in 180	99%	1 in 17,901				•	•	•	•
Mitochondrial complex IV deficiency, nuclear type 5/ Leigh syndrome, French-Canadian type	*LRPPRC*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Mitochondrial complex IV deficiency, nuclear type 5/ Leigh syndrome, French-Canadian type	*LRPPRC*	French Canadian–Saguenay-Lac-Saint-Jean	1 in 23	99%	1 in 2,201				•	•	•	•
Alpha-mannosidosis	*MAN2B1*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Alpha-mannosidosis	*MAN2B1*	Caucasian	1 in 485	99%	1 in 48,401				•	•	•	•
Alpha-mannosidosis	*MAN2B1*	Northern European Caucasian	1 in 354	99%	1 in 35,301				•	•	•	•
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	General Population	1 in 147	99%	1 in 14,601				•	•	•	•
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	Caucasian	1 in 137	99%	1 in 13,601				•	•	•	•
Infantile cerebral and cerebellar atrophy	*MED17*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Infantile cerebral and cerebellar atrophy	*MED17*	Sephardic Jewish–Bukharan, Kurdish	1 in 20	99%	1 in 1,901				•	•	•	•
Familial Mediterranean fever	*MEFV*	General Population	1 in 115	99%	1 in 11,401				•	•	•	•
Familial Mediterranean fever	*MEFV*	Sephardic Jewish	1 in 14	99%	1 in 1,301				•	•	•	•
Familial Mediterranean fever	*MEFV*	Ashkenazi Jewish	1 in 13	99%	1 in 1,201				•	•	•	•
Familial Mediterranean fever	*MEFV*	Armenian	1 in 5	99%	1 in 401				•	•	•	•
Familial Mediterranean fever	*MEFV*	Turkish	1 in 5	99%	1 in 401				•	•	•	•
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*	General Population	1 in 224	99%	1 in 22,301				•	•	•	•
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*	Puerto Rican	1 in 55	99%	1 in 5,401				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Methylmalonic aciduria, *MMAA*-related	*MMAA*	General Population	1 in 316	99%	1 in 31,501				•	•	•	•
Methylmalonic aciduria, *MMAA*-related	*MMAA*	Caucasian	1 in 316	99%	1 in 31,501				•	•	•	•
Methylmalonic aciduria, *MMAB*-related	*MMAB*	Caucasian	1 in 456	99%	1 in 45,501				•	•	•	•
Methylmalonic aciduria, *MMAB*-related	*MMAB*	General Population	1 in 456	99%	1 in 45,501				•	•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	General Population	1 in 415	99%	1 in 41,401				•	•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	Caucasian	1 in 266	99%	1 in 26,501				•	•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	Ashkenazi Jewish	1 in 57	99%	1 in 5,601				•	•	•	•
Abetalipoproteinemia	*MTTP*	Caucasian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Abetalipoproteinemia	*MTTP*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Abetalipoproteinemia	*MTTP*	Ashkenazi Jewish	1 in 186	99%	1 in 18,501				•	•	•	•
Usher syndrome, type 1B	*MYO7A*	African American	<1 in 500	99%	<1 in 49,901				•	•	•	•
Usher syndrome, type 1B	*MYO7A*	General Population	1 in 206	99%	1 in 20,501				•	•	•	•
Usher syndrome, type 1B	*MYO7A*	Caucasian	1 in 145	99%	1 in 14,401				•	•	•	•
Usher syndrome, type 1B	*MYO7A*	Japanese	1 in 123	99%	1 in 12,201				•	•	•	•
Usher syndrome, type 1B	*MYO7A*	Asian	1 in 62	99%	1 in 6,101				•	•	•	•
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	Caucasian	1 in 346	99%	1 in 34,501				•	•	•	•
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	Asian	1 in 298	99%	1 in 29,701				•	•	•	•
Nijmegen breakage syndrome	*NBN*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Nijmegen breakage syndrome	*NBN*	Caucasian	1 in 155	99%	1 in 15,401				•	•	•	•
Niemann-Pick disease, type C1	*NPC1*	Asian	1 in 404	99%	1 in 40,301				•	•	•	•
Niemann-Pick disease, type C1	*NPC1*	General Population	1 in 282	99%	1 in 28,101				•	•	•	•
Niemann-Pick disease, type C1	*NPC1*	Caucasian	1 in 185	99%	1 in 18,401				•	•	•	•
Steroid-resistant nephrotic syndrome, type 2	*NPHS2*	General Population	1 in 377	99%	1 in 37,601				•	•	•	•
Pyruvate carboxylase deficiency	*PC*	General Population	1 in 250	99%	1 in 24,901				•	•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	Caucasian	1 in 380	99%	1 in 37,901				•	•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	General Population	1 in 224	99%	1 in 22,301				•	•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	Asian	1 in 162	99%	1 in 16,101				•	•	•	•
Propionic acidemia, *PCCB*-related	*PCCB*	General Population	1 in 224	99%	1 in 22,301				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Propionic acidemia, *PCCB*-related	*PCCB*	Caucasian	1 in 202	99%	1 in 20,101				•	•	•	•
Propionic acidemia, *PCCB*-related	*PCCB*	Asian	1 in 145	99%	1 in 14,401				•	•	•	•
Zellweger spectrum disorder, *PEX1*-related	*PEX1*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Zellweger spectrum disorder, *PEX1*-related	*PEX1*	Caucasian	1 in 147	99%	1 in 14,601				•	•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	Caucasian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	Ashkenazi Jewish	1 in 227	99%	1 in 22,601				•	•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	General Population	1 in 280	99%	1 in 27,901				•	•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	French Canadian	1 in 55	99%	1 in 5,401				•	•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	Sephardic Jewish–Yemenite	1 in 18	99%	1 in 1,701				•	•	•	•
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	Caucasian	1 in 158	99%	1 in 15,701				•	•	•	•
Neuronal ceroid lipofuscinosis, *PPT1*-related	*PPT1*	General Population	1 in 368	99%	1 in 36,701				•	•	•	•
Neuronal ceroid lipofuscinosis, *PPT1*-related	*PPT1*	Finnish	1 in 70	99%	1 in 6,901				•	•	•	•
Combined pituitary hormone deficiency, type 2	*PROP1*	General Population	1 in 141	99%	1 in 14,001				•	•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	East Asian	1 in 158	99%	1 in 15,701				•	•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	Asian	1 in 122	99%	1 in 12,101				•	•	•	•
Cartilage-hair hypoplasia	*RMRP*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Cartilage-hair hypoplasia	*RMRP*	Finnish	1 in 76	99%	1 in 7,501				•	•	•	•
Cartilage-hair hypoplasia	*RMRP*	Amish	1 in 19	99%	1 in 1,801				•	•	•	•
Dyskeratosis congenita, *RTEL1*-related	*RTEL1*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Dyskeratosis congenita, *RTEL1*-related	*RTEL1*	Ashkenazi Jewish	1 in 165	99%	1 in 16,401				•	•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	Caucasian	1 in 450	99%	1 in 44,901				•	•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	French Canadian–Charlevoix-Saguenay	1 in 21	99%	1 in 2,001				•	•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Caucasian	1 in 290	99%	1 in 28,901				•	•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Northern European Caucasian	1 in 160	99%	1 in 15,901				•	•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Finnish	1 in 150	99%	1 in 14,901				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Limb-girdle muscular dystrophy, type 4	*SGCB*	Northern European Caucasian	<1 in 500	99%	<1 in 49,901				•	•	•	•
Limb-girdle muscular dystrophy, type 4	*SGCB*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Limb-girdle muscular dystrophy, type 4	*SGCB*	Caucasian	1 in 406	99%	1 in 40,501				•	•	•	•
Mucopolysaccharidosis, type IIIA/ Sanfilippo syndrome A	*SGSH*	General Population	1 in 415	99%	1 in 41,401				•	•	•	•
Mucopolysaccharidosis, type IIIA/ Sanfilippo syndrome A	*SGSH*	Caucasian	1 in 253	99%	1 in 25,201				•	•	•	•
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	French Canadian	1 in 23	99%	1 in 2,201				•	•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	General Population	1 in 200	99%	1 in 19,901				•	å•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Caucasian	1 in 110	99%	1 in 10,901				•	•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	East Asian	1 in 100	99%	1 in 9,901				•	•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Asian	1 in 100	99%	1 in 9,901				•	•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Faroese	1 in 20	99%	1 in 1,901				•	•	•	•
Arthrogryposis, intellectual disability, and seizures	*SLC35A3*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Arthrogryposis, intellectual disability, and seizures	*SLC35A3*	Ashkenazi Jewish	1 in 453	99%	1 in 45,201				•	•	•	•
Lysinuric protein intolerance	*SLC7A7*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Lysinuric protein intolerance	*SLC7A7*	Finnish	1 in 122	99%	1 in 12,101				•	•	•	•
Lysinuric protein intolerance	*SLC7A7*	Japanese	1 in 119	99%	1 in 11,801				•	•	•	•
Tyrosinemia, type II	*TAT*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	General Population	1 in 301	99%	1 in 30,001				•	•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	Caucasian	1 in 253	99%	1 in 25,201				•	•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	Norwegian	1 in 151	99%	1 in 15,001				•	•	•	•
Tyrosine hydroxylase deficiency	*TH*	General Population	<1 in 500	99%	<1 in 49,901				•	•	•	•
Tyrosine hydroxylase deficiency	*TH*	Asian	1 in 416	99%	1 in 41,501				•	•	•	•
Tyrosine hydroxylase deficiency	*TH*	Caucasian	1 in 224	99%	1 in 22,301				•	•	•	•
Neuronal ceroid lipofuscinosis, *TPP1*-related	*TPP1*	General Population	1 in 314	99%	1 in 31,301				•	•	•	•
Neuronal ceroid lipofuscinosis, *TPP1*-related	*TPP1*	Newfoundland	1 in 59	99%	1 in 5,801				•	•	•	•
Usher syndrome, type 1C	*USH1C*	General Population	1 in 353	99%	1 in 35,201				•	•	•	•
Usher syndrome, type 1C	*USH1C*	French Canadian, Acadian	1 in 227	99%	1 in 22,601				•	•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Usher syndrome, type 1C	*USH1C*	Acadian	1 in 41	99%	1 in 4,001				•	•	•	•
Triple A syndrome	*AAAS*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
GABA-transaminase deficiency	*ABAT*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital ichthyosis, *ABCA12*-related	*ABCA12*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Progressive familial intrahepatic cholestasis 2	*ABCB11*	General Population	1 in 158	99%	1 in 15,701					•	•	•
Pseudoxanthoma elasticum	*ABCC6*	General Population	1 in 79	90%	1 in 781					•	•	•
Mitochondrial complex I deficiency, *ACAD9*-related	*ACAD9*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Combined malonic and methylmalonic aciduria	*ACSF3*	General Population	1 in 86	99%	1 in 8,501					•	•	•
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*	Ashkenazi Jewish	1 in 248	99%	1 in 24,701					•	•	•
Bilateral frontoparietal polymicrogyria	*ADGRG1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Usher syndrome, type 2C	*ADGRV1*	General Population	1 in 176	99%	1 in 17,501					•	•	•
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Combined oxidative phosphorylation deficiency 6	*AIFM1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pyridoxine-dependent epilepsy	*ALDH7A1*	General Population	1 in 158	99%	1 in 15,701					•	•	•
Congenital disorder of glycosylation, type Ic	*ALG6*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Alström syndrome	*ALMS1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
MEDNIK syndrome	*AP1S1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Hermansky-Pudlak syndrome, type 2	*AP3B1*	General Population	1 in 158	99%	1 in 15,701					•	•	•
Nephrogenic diabetes insipidus	*AQP2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Argininemia	*ARG1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Joubert syndrome 8	*ARL13B*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
X-linked chondrodysplasia punctata 1	*ARSL*	General Population	1 in 375,000	99%	1 in 37,499,901					•	•	•
Farber lipogranulomatosis	*ASAH1*	General Population	1 in 500	99%	<1 in 49,901					•	•	•
Asparagine synthetase deficiency	*ASNS*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Asparagine synthetase deficiency	*ASNS*	Sephardic Jewish–Iranian	1 in 80	99%	1 in 7,901					•	•	•
Renal tubular acidosis and deafness, *ATP6V1B1*-related	*ATP6V1B1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Renal tubular acidosis and deafness, *ATP6V1B1*-related	*ATP6V1B1*	Sephardic Jewish–Syrian	1 in 140	99%	1 in 13,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	*ATP8B1*	General Population	1 in 53	99%	1 in 5,201					•	•	•
Alpha-thalassemia intellectual disability syndrome, X-linked	*ATRX*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Joubert syndrome 27	*B9D1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Joubert syndrome 34	*B9D2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bardet-Biedl syndrome 12	*BBS12*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bardet-Biedl syndrome 4	*BBS4*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bardet-Biedl syndrome 7	*BBS7*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bardet-Biedl syndrome 9	*BBS9*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pseudocholinesterase deficiency	*BCHE*	General Population	1 in 53	99%	1 in 5,201					•	•	•
Bartter syndrome, type 4A	*BSND*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
X-linked agammaglobulinemia	*BTK*	General Population	1 in 250,000	99%	1 in 24,999,901					•	•	•
Orofaciodigital syndrome, type XIV	*C2CD3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Desbuquois dysplasia, type I	*CANT1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
X-linked hyper IgM syndrome	*CD40LG*	General Population	1 in 500,000	99%	1 in 49,999,901					•	•	•
Joubert syndrome 25	*CEP104*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Joubert syndrome 31	*CEP120*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Joubert syndrome 15	*CEP41*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Retinitis pigmentosa 26	*CERKL*	General Population	1 in 137	99%	1 in 13,601					•	•	•
Retinitis pigmentosa 26	*CERKL*	Sephardic Jewish–Yemenite	1 in 24	99%	1 in 2,301					•	•	•
Congenital myasthenic syndrome, *CHAT*-related	*CHAT*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Choroideremia, X-linked	*CHM*	General Population	1 in 25,000	99%	1 in 2,499,901					•	•	•
Multiple pterygium syndrome, lethal type	*CHRNG*	General Population	1 in 50	99%	1 in 4,901					•	•	•
Usher syndrome, type 1J	*CIB2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bare lymphocyte syndrome, type II	*CIITA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Dent disease	*CLCN5*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Steel syndrome	*COL27A1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital myasthenic syndrome, *COLQ*-related	*COLQ*	General Population	1 in 430	99%	1 in 42,901					•	•	•
Joubert syndrome 17	*CPLANE1*	General Population	1 in 423	99%	1 in 42,201					•	•	•
Carbamoyl phosphate synthetase I deficiency	*CPS1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Carbamoyl phosphate synthetase I deficiency	*CPS1*	Asian	1 in 447	99%	1 in 44,601					•	•	•
Carbamoyl phosphate synthetase I deficiency	*CPS1*	Caucasian	1 in 284	99%	1 in 28,301					•	•	•
Carnitine palmitoyltransferase I deficiency	*CPT1A*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Carnitine palmitoyltransferase I deficiency	*CPT1A*	Hutterite	1 in 16	99%	1 in 1,501					•	•	•
CRB1-related retinal dystrophies	*CRB1*	General Population	1 in 112	99%	1 in 11,101					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 7	*CRPPA*	General Population	1 in 371	99%	1 in 37,001					•	•	•
Joubert syndrome 21	*CSPP1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Galactosialidosis	*CTSA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Neuronal ceroid lipofuscinosis, *CTSD*-related	*CTSD*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pycnodysostosis	*CTSK*	General Population	1 in 439	99%	1 in 43,801					•	•	•
Chronic granulomatous disease 4	*CYBA*	General Population	1 in 500	99%	<1 in 49,901					•	•	•
Chronic granulomatous disease 4	*CYBA*	Sephardic Jewish–Moroccan	1 in 13	99%	1 in 1,201					•	•	•
Chronic granulomatous disease, X-linked	*CYBB*	General Population	1 in 150,000	99%	1 in 14,999,901					•	•	•
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	*CYP11B1*	General Population	1 in 158	99%	1 in 15,701					•	•	•
Corticosterone methyloxidase deficiency	*CYP11B2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Corticosterone methyloxidase deficiency	*CYP11B2*	Sephardic Jewish–Iranian	1 in 30	99%	1 in 2,901					•	•	•
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	*CYP17A1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Aromatase deficiency	*CYP19A1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Primary congenital glaucoma	*CYP1B1*	General Population	1 in 74	99%	1 in 7,301					•	•	•
Omenn syndrome	*DCLRE1C*	Northern European Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Omenn syndrome	*DCLRE1C*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Omenn syndrome	*DCLRE1C*	Navajo, Apache	1 in 10	99%	1 in 901					•	•	•
DCX-related disorders, X-linked	*DCX*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Perlman syndrome	*DIS3L2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Dyskeratosis congenita, X-linked	*DKC1*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Primary ciliary dyskinesia, *DNAH5*-related	*DNAH5*	Ashkenazi Jewish	1 in 174	99%	1 in 17,301					•	•	•
Primary ciliary dyskinesia, *DNAH5*-related	*DNAH5*	General Population	1 in 120	99%	1 in 11,901					•	•	•
Primary ciliary dyskinesia, *DNAI1*-related	*DNAI1*	Ashkenazi Jewish	1 in 352	99%	1 in 35,101					•	•	•
Primary ciliary dyskinesia, *DNAI1*-related	*DNAI1*	General Population	1 in 182	99%	1 in 18,101					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Primary ciliary dyskinesia, *DNAI2*-related	*DNAI2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Primary ciliary dyskinesia, *DNAI2*-related	*DNAI2*	Ashkenazi Jewish	1 in 200	99%	1 in 19,901					•	•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	General Population	1 in 454	99%	1 in 45,301					•	•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	French Canadian	1 in 353	99%	1 in 35,201					•	•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Northern European Caucasian	1 in 290	99%	1 in 28,901					•	•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Caucasian	1 in 290	99%	1 in 28,901					•	•	•
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	East Asian	1 in 50	99%	1 in 4,901					•	•	•
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	General Population	1 in 20	99%	1 in 1,901					•	•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	General Population	1 in 311	99%	1 in 31,001					•	•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	Caucasian	1 in 158	99%	1 in 15,701					•	•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	Sephardic Jewish–Libyan, Moroccan, Tunisian, Bulgarian	1 in 14	99%	1 in 1,301					•	•	•
Hypohidrotic ectodermal dysplasia, X-linked	*EDA*	General Population	1 in 3800	99%	1 in 379,901					•	•	•
Wolcott-Rallison syndrome	*EIF2AK3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Leukoencephalopathy with vanishing white matter 5	*EIF2B5*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Emery-Dreifuss muscular dystrophy, X-linked	*EMD*	General Population	1 in 250,000	99%	1 in 24,999,901					•	•	•
Cerebrooculofacioskeletal syndrome 1/Cockayne syndrome, type B	*ERCC6*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Cockayne syndrome, type A	*ERCC8*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Roberts-SC phocomelia syndrome	*ESCO2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIA	*ETFA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIB	*ETFB*	General Population	1 in 408	99%	1 in 40,701					•	•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIC	*ETFDH*	General Population	1 in 250	99%	1 in 24,901					•	•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIC	*ETFDH*	Asian	1 in 87	99%	1 in 8,601					•	•	•
Ethylmalonic encephalopathy	*ETHE1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ellis-van Creveld syndrome	*EVC*	General Population	1 in 345	99%	1 in 34,401					•	•	•
Ellis-van Creveld syndrome	*EVC*	Lancaster County Amish	1 in 12	99%	1 in 1,101					•	•	•
Pontocerebellar hypoplasia, type 1B	*EXOSC3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Retinitis pigmentosa 25	*EYS*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901					•	•	•
Retinitis pigmentosa 25	*EYS*	General Population	1 in 129	99%	1 in 12,801					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Retinitis pigmentosa 25	*EYS*	Caucasian	1 in 53	99%	1 in 5,201					•	•	•
Retinitis pigmentosa 25	*EYS*	Sephardic Jewish–Moroccan	1 in 42	99%	1 in 4,101					•	•	•
Retinitis pigmentosa 28	*FAM161A*	General Population	1 in 289	99%	1 in 28,801					•	•	•
Retinitis pigmentosa 28	*FAM161A*	Ashkenazi Jewish	1 in 214	99%	1 in 21,301					•	•	•
Retinitis pigmentosa 28	*FAM161A*	Sephardic Jewish–Libyan, Moroccan, Tunisian, Bulgarian	1 in 41	99%	1 in 4,001					•	•	•
Fanconi anemia, complementation group B, X-linked	*FANCB*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Fanconi anemia, complementation group D2	*FANCD2*	General Population	<1 in 500	98%	<1 in 24,951					•	•	•
Fanconi anemia, complementation group E	*FANCE*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Fanconi anemia, complementation group F	*FANCF*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Fanconi anemia, complementation group G	*FANCG*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Fanconi anemia, complementation group G	*FANCG*	African American	1 in 100	99%	1 in 9,901					•	•	•
Fanconi anemia, complementation group I	*FANCI*	General Population	1 in 500	99%	<1 in 49,901					•	•	•
Fanconi anemia, complementation group L	*FANCL*	General Population	1 in 500	99%	<1 in 49,901					•	•	•
Fumarase deficiency	*FH*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	General Population	1 in 30	99%	1 in 2,901					•	•	•
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	African American	1 in 5	99%	1 in 401					•	•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	Asian	<1 in 500	99%	<1 in 49,901					•	•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	General Population	1 in 122	99%	1 in 12,101					•	•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	Roma	1 in 47	99%	1 in 4,601					•	•	•
Mucopolysaccharidosis, type IVA/Morquio syndrome	*GALNS*	General Population	1 in 307	99%	1 in 30,601					•	•	•
Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Guanidinoacetate methyltransferase deficiency	*GAMT*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Guanidinoacetate methyltransferase deficiency	*GAMT*	Portuguese	1 in 125	99%	1 in 12,401					•	•	•
Combined oxidative phosphorylation deficiency 1	*GFM1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Charcot-Marie-Tooth disease, type 1X, X-linked	*GJB1*	General Population	1 in 7000	99%	1 in 699,901					•	•	•
Rhizomelic chondrodysplasia punctata, type 2	*GNPAT*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mucolipidosis III gamma	*GNPTG*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mucolipidosis III gamma	*GNPTG*	Caucasian	1 in 273	99%	1 in 27,201					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Bernard-Soulier syndrome, type A	*GP1BA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bernard-Soulier syndrome, type C	*GP9*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Primary hyperoxaluria, type II	*GRHPR*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mucopolysaccharidosis, type VII/Sly syndrome	*GUSB*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial trifunctional protein deficiency, *HADHB*-related	*HADHB*	General Population	1 in 146	99%	1 in 14,501					•	•	•
Congenital neutropenia, *HAX1*-related	*HAX1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Hereditary hemochromatosis, type 2	*HJV*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Hereditary hemochromatosis, type 2	*HJV*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Primary hyperoxaluria, type III	*HOGA1*	General Population	1 in 309	99%	1 in 30,801					•	•	•
HPRT1-related disorders, X-linked	*HPRT1*	General Population	1 in 285,000	99%	1 in 28,499,901					•	•	•
Hermansky-Pudlak syndrome, type 4	*HPS4*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
17-beta-hydroxysteroid dehydrogenase deficiency, type III	*HSD17B3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
3-beta-hydroxysteroid dehydrogenase deficiency, type II	*HSD3B2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mucopolysaccharidosis, type IX/Hyaluronidase deficiency	*HYAL1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Severe combined immunodeficiency, X-linked	*IL2RG*	General Population	1 in 69,000	99%	1 in 6,899,901					•	•	•
Joubert syndrome 1	*INPP5E*	General Population	1 in 264	99%	1 in 26,301					•	•	•
Nephronophthisis 2	*INVS*	General Population	1 in 373	99%	1 in 37,201					•	•	•
Senior-Løken syndrome 5	*IQCB1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 6	*LARGE1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Leber congenital amaurosis 5	*LCA5*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	French Canadian	1 in 267	99%	1 in 26,601					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Caucasian	1 in 200	99%	1 in 19,901					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Finnish	1 in 143	99%	1 in 14,201					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	South African Afrikaner	1 in 70	99%	1 in 6,901					•	•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Ashkenazi Jewish	1 in 69	99%	1 in 6,801					•	•	•
Familial hypercholesterolemia, *LDLRAP1*-related	*LDLRAP1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Familial hypercholesterolemia, *LDLRAP1*-related	*LDLRAP1*	Sardinian	1 in 143	99%	1 in 14,201					•	•	•
Combined pituitary hormone deficiency, type 3	*LHX3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Stüve-Wiedemann syndrome	*LIFR*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Lysosomal acid lipase deficiency	*LIPA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Lysosomal acid lipase deficiency	*LIPA*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901					•	•	•
Lysosomal acid lipase deficiency	*LIPA*	Sephardic Jewish–Iranian	1 in 26	99%	1 in 2,501					•	•	•
Lipoprotein lipase deficiency	*LPL*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Lipoprotein lipase deficiency	*LPL*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Lipoprotein lipase deficiency	*LPL*	Asian	1 in 189	99%	1 in 18,801					•	•	•
Lipoprotein lipase deficiency	*LPL*	French Canadian–Other	1 in 139	99%	1 in 13,801					•	•	•
Lipoprotein lipase deficiency	*LPL*	French Canadian–Saguenay-Lac-Saint-Jean	1 in 46	99%	1 in 4,501					•	•	•
Chédiak-Higashi syndrome	*LYST*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Neuronal ceroid lipofuscinosis, *MFSD8*-related	*MFSD8*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Bardet-Biedl syndrome 6	*MKKS*	General Population	1 in 219	99%	1 in 21,801					•	•	•
MKS1-related disorders	*MKS1*	General Population	1 in 260	99%	1 in 25,901					•	•	•
MKS1-related disorders	*MKS1*	Caucasian	1 in 260	99%	1 in 25,901					•	•	•
MKS1-related disorders	*MKS1*	Finnish	1 in 47	99%	1 in 4,601					•	•	•
Malonyl-CoA decarboxylase deficiency	*MLYCD*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Combined methylmalonic aciduria and homocystinuria, cblD type/Cobalamin D deficiency	*MMADHC*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Combined methylmalonic aciduria and homocystinuria, cblD type/Cobalamin D deficiency	*MMADHC*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Molybdenum cofactor deficiency of complementation group A	*MOCS1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital disorder of glycosylation, type Ib	*MPI*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
MPV17-related mitochondrial DNA maintenance defect	*MPV17*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
MPV17-related mitochondrial DNA maintenance defect	*MPV17*	Navajo	1 in 20	99%	1 in 1,901					•	•	•
Ataxia-telangiectasia-like disorder 1	*MRE11*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Homocystinuria caused by methylenetetrahydrofolate reductase deficiency	*MTHFR*	General Population	1 in 158	99%	1 in 15,701					•	•	•
Homocystinuria caused by methylenetetrahydrofolate reductase deficiency	*MTHFR*	Sephardic Jewish–Bukharan	1 in 39	99%	1 in 3,801					•	•	•

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X-linked myotubular myopathy	*MTM1*	General Population	1 in 38,000	99%	1 in 3,799,901					•	•	•
Homocystinuria, type cblE	*MTRR*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Homocystinuria, type cblE	*MTRR*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Nonsyndromic hearing loss and deafness 3	*MYO15A*	General Population	1 in 117	99%	1 in 11,601					•	•	•
N-acetylglutamate synthase deficiency	*NAGS*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	Roma	1 in 22	99%	1 in 2,101					•	•	•
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*	Ashkenazi Jewish	1 in 290	99%	1 in 28,901					•	•	•
Mitochondrial complex I deficiency, nuclear type 17	*NDUFAF6*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial complex I deficiency, nuclear type 1	*NDUFS4*	General Population	1 in 423	99%	1 in 42,201					•	•	•
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*	Caucasus Jewish	1 in 24	99%	1 in 2,301					•	•	•
Sialidosis	*NEU1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Niemann-Pick disease, type C2	*NPC2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
NPHP1 nephronophthisis-related ciliopathies	*NPHP1*	General Population	1 in 202	99%	1 in 20,101					•	•	•
NPHP3 nephronophthisis-related ciliopathies	*NPHP3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
NPHP4 nephronophthisis-related ciliopathies	*NPHP4*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Enhanced S-cone syndrome	*NR2E3*	General Population	1 in 204	99%	1 in 20,301					•	•	•
Enhanced S-cone syndrome	*NR2E3*	Ashkenazi Jewish	1 in 100	99%	1 in 9,901					•	•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	Sephardic Jewish–Moroccan	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	Asian	1 in 387	99%	1 in 38,601					•	•	•
Ornithine aminotransferase deficiency	*OAT*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ornithine aminotransferase deficiency	*OAT*	Sephardic Jewish–Iraqi, Syrian	1 in 177	99%	1 in 17,601					•	•	•
Ornithine aminotransferase deficiency	*OAT*	Finnish	1 in 147	99%	1 in 14,601					•	•	•
Lowe syndrome, X-linked	*OCRL*	General Population	1 in 375,000	99%	1 in 37,499,901					•	•	•
3-methylglutaconic aciduria, type III/Costeff syndrome	*OPA3*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
3-methylglutaconic aciduria, type III/Costeff syndrome	*OPA3*	Sephardic Jewish–Iraqi	1 in 13	99%	1 in 1,201					•	•	•
Pyruvate dehydrogenase E1-alpha deficiency, X-linked	*PDHA1*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•

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Pyruvate dehydrogenase E1-beta deficiency	*PDHB*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Prolidase deficiency	*PEPD*	General Population	1 in 242	99%	1 in 24,101					•	•	•
Mitochondrial complex IV deficiency, nuclear type 12	*PET100*	General Population	1 in 452	99%	1 in 45,101					•	•	•
Zellweger spectrum disorder, *PEX10*-related	*PEX10*	Asian	<1 in 500	99%	<1 in 49,901					•	•	•
Zellweger spectrum disorder, *PEX10*-related	*PEX10*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Zellweger spectrum disorder, *PEX12*-related	*PEX12*	General Population	1 in 406	99%	1 in 40,501					•	•	•
Zellweger spectrum disorder, *PEX26*-related	*PEX26*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Glycogen storage disease, type VII	*PFKM*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Glycogen storage disease, type VII	*PFKM*	Ashkenazi Jewish	1 in 250	99%	1 in 24,901					•	•	•
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	Ashkenazi Jewish	1 in 453	99%	1 in 45,201					•	•	•
Refsum disease	*PHYH*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
PLA2G6-associated neurodegeneration	*PLA2G6*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Steroid-resistant nephrotic syndrome, type 3	*PLCE1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 3	*POMGNT1*	General Population	1 in 462	99%	1 in 46,101					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 3	*POMGNT1*	Finnish	1 in 111	99%	1 in 11,001					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 1	*POMT1*	General Population	1 in 275	99%	1 in 27,401					•	•	•
Muscular dystrophy-dystroglycanopathy, type A, 2	*POMT2*	General Population	1 in 465	99%	1 in 46,401					•	•	•
PRPS1-related disorders, X-linked	*PRPS1*	General Population	1 in 500,000	99%	1 in 49,999,901					•	•	•
Metachromatic leukodystrophy due to saposin B deficiency	*PSAP*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Myopathy, lactic acidosis, and sideroblastic anemia	*PUS1*	Sephardic Jewish–Iranian	<1 in 500	99%	<1 in 49,901					•	•	•
Myopathy, lactic acidosis, and sideroblastic anemia	*PUS1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Glycogen storage disease, type V	*PYGM*	Caucasian	1 in 191	99%	1 in 19,001					•	•	•
Glycogen storage disease, type V	*PYGM*	General Population	1 in 191	99%	1 in 19,001					•	•	•
Glycogen storage disease, type V	*PYGM*	Sephardic Jewish–Kurdish	1 in 84	99%	1 in 8,301					•	•	•
Carpenter syndrome	*RAB23*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Carpenter syndrome	*RAB23*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Severe combined immunodeficiency, *RAG1*-related	*RAG1*	General Population	1 in 245	99%	1 in 24,401					•	•	•
Severe combined immunodeficiency, *RAG2*-related	*RAG2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Severe combined immunodeficiency, *RAG2*-related	*RAG2*	Sephardic Jewish–Iraqi	<1 in 500	99%	<1 in 49,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	Sephardic Jewish–Iraqi, Iranian	<1 in 500	99%	<1 in 49,901					•	•	•
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	General Population	1 in 252	99%	1 in 25,101					•	•	•
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	Caucasian	1 in 176	99%	1 in 17,501					•	•	•
Leber congenital amaurosis 13	*RDH12*	General Population	1 in 456	99%	1 in 45,501					•	•	•
Aicardi-Goutières syndrome 4	*RNASEH2A*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Aicardi-Goutières syndrome 3	*RNASEH2C*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Leber congenital amaurosis 2	*RPE65*	General Population	1 in 228	99%	1 in 22,701					•	•	•
Leber congenital amaurosis 2	*RPE65*	Sephardic Jewish–North African	1 in 90	99%	1 in 8,901					•	•	•
Ciliopathies, *RPGRIP1L*-related	*RPGRIP1L*	General Population	1 in 259	99%	1 in 25,801					•	•	•
Aicardi-Goutières syndrome 5	*SAMHD1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Shwachman-Diamond syndrome	*SBDS*	General Population	1 in 145	99%	1 in 14,401					•	•	•
Action myoclonus renal failure syndrome	*SCARB2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Progressive cerebello-cerebral atrophy	*SEPSECS*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Progressive cerebello-cerebral atrophy	*SEPSECS*	Sephardic Jewish–Moroccan, Iraqi	1 in 41	99%	1 in 4,001					•	•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	General Population	1 in 38	99%	1 in 3,701					•	•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	Ashkenazi Jewish	1 in 24	99%	1 in 2,301					•	•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	Northern European Caucasian	1 in 15	99%	1 in 1,401					•	•	•
Limb-girdle muscular dystrophy, type 6	*SGCD*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Northern European Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	General Population	1 in 354	99%	1 in 35,301					•	•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Moroccan	1 in 250	99%	1 in 24,901					•	•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Roma	1 in 96	99%	1 in 9,501					•	•	•
Gitelman syndrome	*SLC12A3*	General Population	1 in 100	99%	1 in 9,901					•	•	•
Free sialic acid storage disorders	*SLC17A5*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Free sialic acid storage disorders	*SLC17A5*	Canadian Inuit	1 in 129	99%	1 in 12,801					•	•	•
Free sialic acid storage disorders	*SLC17A5*	Swedish	1 in 125	99%	1 in 12,401					•	•	•
Free sialic acid storage disorders	*SLC17A5*	Finnish	1 in 100	99%	1 in 9,901					•	•	•
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	*SLC1A4*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Asian	1 in 123	99%	1 in 12,201					•	•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	East Asian	1 in 65	99%	1 in 6,401					•	•	•
Ornithine translocase deficiency	*SLC25A15*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ornithine translocase deficiency	*SLC25A15*	French Canadian	1 in 20	99%	1 in 1,901					•	•	•
Ornithine translocase deficiency	*SLC25A15*	Métis Nation–Saskatchewan	1 in 19	99%	1 in 1,801					•	•	•
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Acrodermatitis enteropathica	*SLC39A4*	General Population	1 in 354	99%	1 in 35,301					•	•	•
Corneal dystrophy and perceptive deafnesssyndrome	*SLC4A11*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Renal tubular acidosis	*SLC4A4*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Schimke immunoosseous dysplasia	*SMARCAL1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
5-alpha-reductase deficiency	*SRD5A2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Lipoid congenital adrenal hyperplasia	*STAR*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Lipoid congenital adrenal hyperplasia	*STAR*	East Asian	1 in 177	99%	1 in 17,601					•	•	•
Familial hemophagocytic lymphohistiocytosis 4	*STX11*	General Population	1 in 354	99%	1 in 35,301					•	•	•
Familial hemophagocytic lymphohistiocytosis 5	*STXBP2*	General Population	1 in 296	99%	1 in 29,501					•	•	•
Multiple sulfatase deficiency	*SUMF1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Multiple sulfatase deficiency	*SUMF1*	Ashkenazi Jewish	1 in 279	99%	1 in 27,801					•	•	•
Barth syndrome, X-linked	*TAFAZZIN*	General Population	1 in 225,000	99%	1 in 22,499,901					•	•	•
Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	*TANGO2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Ashkenazi Jewish	1 in 350	99%	1 in 34,901					•	•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	General Population	1 in 316	99%	1 in 31,501					•	•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Costa Rican	1 in 86	99%	1 in 8,501					•	•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Chuvashiya	1 in 60	99%	1 in 5,901					•	•	•
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*	Sephardic Jewish–Bukharan	1 in 27	99%	1 in 2,601					•	•	•
Hereditary hemochromatosis, type 3	*TFR2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Progressive familial intrahepatic cholestasis 4	*TJP2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mulibrey nanism	*TRIM37*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Acute infantile liver failure	*TRMU*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Acute infantile liver failure	*TRMU*	Sephardic Jewish–Yemenite	1 in 34	99%	1 in 3,301					•	•	•
Pontocerebellar hypoplasia, type 2B	*TSEN2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pontocerebellar hypoplasia, types 4 and 2A	*TSEN54*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Combined oxidative phosphorylation deficiency 3	*TSFM*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Combined oxidative phosphorylation deficiency 3	*TSFM*	Finnish	1 in 80	99%	1 in 7,901					•	•	•
Trichohepatoenteric syndrome 1	*TTC37*	General Population	1 in 381	99%	1 in 38,001					•	•	•
Bardet-Biedl syndrome 8	*TTC8*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ataxia with isolated vitamin E deficiency	*TTPA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Ataxia with isolated vitamin E deficiency	*TTPA*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	Caucasian	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	Sephardic Jewish–Iranian	1 in 158	99%	1 in 15,701					•	•	•
X-linked infantile spinal muscular atrophy	*UBA1*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
Johanson-Blizzard syndrome	*UBR1*	General Population	1 in 250	99%	1 in 24,901					•	•	•
Beta-ureidopropionase deficiency	*UPB1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Choreoacanthocytosis	*VPS13A*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Choreoacanthocytosis	*VPS13A*	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901					•	•	•
Cohen syndrome	*VPS13B*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Severe congenital neutropenia 5	*VPS45*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pontocerebellar hypoplasia, type 2E	*VPS53*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pontocerebellar hypoplasia, type 1A	*VRK1*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Pontocerebellar hypoplasia, type 1A	*VRK1*	Ashkenazi Jewish	1 in 225	99%	1 in 22,401					•	•	•
Microphthalmia/Anophthalmia	*VSX2*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Microphthalmia/Anophthalmia	*VSX2*	Sephardic Jewish–Iranian, Syrian	1 in 145	99%	1 in 14,401					•	•	•
Wiskott-Aldrich syndrome, X-linked	*WAS*	General Population	1 in 187,500	99%	1 in 18,749,901					•	•	•
Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge syndrome	*WNT10A*	General Population	1 in 305	99%	1 in 30,401					•	•	•

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Werner syndrome	*WRN*	General Population	1 in 224	99%	1 in 22,301					•	•	•
WWOX deficiency	*WWOX*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Xeroderma pigmentosum, group A	*XPA*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
Spastic paraplegia, type 15	*ZFYVE26*	General Population	<1 in 500	99%	<1 in 49,901					•	•	•
X-linked heterotaxy-1	*ZIC3*	General Population	<1 in 750,000	99%	<1 in 74,999,901					•	•	•
ABCA4-related disorders	*ABCA4*	General Population	1 in 45	99%	1 in 4,401						•	•
Progressive familial intrahepatic cholestasis 3	*ABCB4*	General Population	1 in 194	99%	1 in 19,301						•	•
Dubin-Johnson syndrome	*ABCC2*	General Population	1 in 158	99%	1 in 15,701						•	•
Dubin-Johnson syndrome	*ABCC2*	Sephardic Jews–Iranian, Moroccan	1 in 27	99%	1 in 2,601						•	•
ADAMTSL4-related eye disorders	*ADAMTSL4*	General Population	<1 in 500	99%	<1 in 49,901						•	•
ADAMTSL4-related eye disorders	*ADAMTSL4*	Bukharan Jewish	1 in 48	99%	1 in 4,701						•	•
ADAMTSL4-related eye disorders	*ADAMTSL4*	Norwegian	1 in 63	99%	1 in 6,201						•	•
Leber congenital amaurosis 4	*AIPL1*	General Population	1 in 387	99%	1 in 38,601						•	•
Congenital disorder of glycosylation, type Ik	*ALG1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Developmental and epileptic encephalopathy 36, X-linked	*ALG13*	General Population	<1 in 750,000	99%	<1 in 74,999,901						•	•
Imerslund-Gräsbeck syndrome 2	*AMN*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Androgen insensitivity syndrome, X-linked	*AR*	General Population	1 in 5000	99%	1 in 499,901						•	•
Bardet-Biedl syndrome 3	*ARL6*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Bardet-Biedl syndrome 3	*ARL6*	Newfoundland	1 in 289	99%	1 in 28,801						•	•
AVPR2-related disorders, X-linked	*AVPR2*	General Population	1 in 63,000	99%	1 in 6,299,901						•	•
Bardet-Biedl syndrome 5	*BBS5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Hermansky-Pudlak syndrome, type 8	*BLOC1S3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Hermansky-Pudlak syndrome, type 9	*BLOC1S6*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Osteogenesis imperfecta, type XIII	*BMP1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Fanconi anemia, complementation group J	*BRIP1*	General Population	1 in 500	99%	<1 in 49,901						•	•
Developmental and epileptic encephalopathy 50	*CAD*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Developmental and epileptic encephalopathy 50	*CAD*	Finnish	1 in 212	99%	1 in 21,101						•	•
Developmental and epileptic encephalopathy 50	*CAD*	Ashkenazi Jewish	1 in 458	99%	1 in 45,701						•	•
Catecholaminergic polymorphic ventricular tachycardia, type 2	*CASQ2*	General Population	1 in 224	99%	1 in 22,301						•	•

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Autosomal recessive intellectual developmental disorder, type 3	*CC2D1A*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Autosomal recessive intellectual developmental disorder, type 3	*CC2D1A*	Ashkenazi Jewish	1 in 445	99%	1 in 44,401						•	•
Primary ciliary dyskinesia, *CCDC103*-related	*CCDC103*	General Population	1 in 217	99%	1 in 21,601						•	•
Primary ciliary dyskinesia, *CCDC39*-related	*CCDC39*	General Population	1 in 144	99%	1 in 14,301						•	•
Progressive pseudorheumatoid dysplasia	*CCN6*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *CD3D*-related	*CD3D*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *CD3E*-related	*CD3E*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Hyper-IgM syndrome, type 3	*CD40*	General Population	<1 in 500	99%	<1 in 49,901						•	•
CD59-mediated hemolytic anemia	*CD59*	General Population	<1 in 500	99%	<1 in 49,901						•	•
CD59-mediated hemolytic anemia	*CD59*	Sephardic Jews–North African	1 in 66	99%	1 in 6,501						•	•
CEP152-related disorders	*CEP152*	General Population	<1 in 500	99%	<1 in 49,901						•	•
COL11A2-related disorders	*COL11A2*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Junctional epidermolysis bullosa, *COL17A1*-related	*COL17A1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Mitochondrial complex IV deficiency, nuclear type 6	*COX15*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Osteogenesis imperfecta, type VII	*CRTAP*	General Population	<1 in 500	99%	<1 in 49,901						•	•
CTSC-related disorders	*CTSC*	General Population	1 in 250	99%	1 in 24,901						•	•
CYP7B1-related disorders	*CYP7B1*	General Population	1 in 338	99%	1 in 33,701						•	•
Woodhouse-Sakati syndrome	*DCAF17*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Warsaw breakage syndrome	*DDX11*	General Population	<1 in 500	15%	1 in 588						•	•
DGAT1 deficiency	*DGAT1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Deoxyguanosine kinase deficiency/Mitochondrial DNA depletion syndrome 3	*DGUOK*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Spondylocostal dysostosis 1	*DLL3*	General Population	1 in 289	99%	1 in 28,801						•	•
Spondylocostal dysostosis 1	*DLL3*	Northern European Caucasian	1 in 350	99%	1 in 34,901						•	•
Primary ciliary dyskinesia, *DNAH11*-related	*DNAH11*	General Population	1 in 144	99%	1 in 14,301						•	•
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Thyroid dyshormonogenesis 6	*DUOX2*	General Population	1 in 55	95%	1 in 1,081						•	•
Leukoencephalopathy with vanishing white matter 1	*EIF2B1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Leukoencephalopathy with vanishing white matter 2	*EIF2B2*	General Population	<1 in 500	99%	<1 in 49,901						•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Leukoencephalopathy with vanishing white matter 3	*EIF2B3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Leukoencephalopathy with vanishing white matter 4	*EIF2B4*	General Population	<1 in 500	99%	<1 in 49,901						•	•
EPG5-related disorder	*EPG5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Factor II deficiency/Prothrombin deficiency	*F2*	General Population	1 in 33	99%	1 in 3,201						•	•
Factor V deficiency	*F5*	General Population	1 in 12	99%	1 in 1,101						•	•
Fructose-1,6-bisphosphatase deficiency	*FBP1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Fructose-1,6-bisphosphatase deficiency	*FBP1*	Caucasian	1 in 296	99%	1 in 29,501						•	•
Parkinson disease 15	*FBXO7*	General Population	<1 in 500	99%	<1 in 49,901						•	•
FHL1-related disorders, X-linked	*FHL1*	General Population	<1 in 750,000	99%	<1 in 74,999,901						•	•
Osteogenesis imperfecta, type XI	*FKBP10*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *FOXN1*-related	*FOXN1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Mitochondrial complex I deficiency, nuclear type 19	*FOXRED1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Fraser syndrome, type 1	*FRAS1*	General Population	1 in 316	99%	1 in 31,501						•	•
Fraser syndrome, type 2	*FREM2*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Fucosidosis	*FUCA1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
G6PC3 deficiency	*G6PC3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Galactosemia, type III/Galactose epimerase deficiency	*GALE*	General Population	1 in 132	99%	1 in 13,101						•	•
Galactosemia, type III/Galactose epimerase deficiency	*GALE*	African American	1 in 41	99%	1 in 4,001						•	•
Arginine:glycine amidinotransferase deficiency	*GATM*	General Population	<1 in 500	99%	<1 in 49,901						•	•
GCH1-related disorders	*GCH1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
GDF5-related disorders	*GDF5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Laron syndrome	*GHR*	General Population	1 in 167	99%	1 in 16,601						•	•
Geroderma osteodysplastica	*GORAB*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Glutathione synthetase deficiency	*GSS*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Leber congenital amaurosis 1	*GUCY2D*	General Population	1 in 200	99%	1 in 19,901						•	•
Hereditary hemochromatosis, type 2B	*HAMP*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Methylmalonic aciduria and homocystinuria, cblX type, X-linked	*HCFC1*	General Population	<1 in 750,000	99%	<1 in 74,999,901						•	•
Alkaptonuria	*HGD*	General Population	1 in 250	99%	1 in 24,901						•	•
Heme oxygenase 1 deficiency	*HMOX1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Tyrosinemia, type III	*HPD*	General Population	<1 in 500	99%	<1 in 49,901						•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Hermansky-Pudlak syndrome, type 5	*HPS5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Hermansky-Pudlak syndrome, type 6	*HPS6*	General Population	<1 in 500	99%	<1 in 49,901						•	•
HSD10 disease, X-linked	*HSD17B10*	General Population	<1 in 750,000	99%	<1 in 74,999,901						•	•
IGHMBP2-related disorders	*IGHMBP2*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *IKBKB*-related	*IKBKB*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *IL7R*-related	*IL7R*	General Population	1 in 388	99%	1 in 38,701						•	•
Junctional epidermolysis bullosa, *ITGA6*-related	*ITGA6*	General Population	1 in 159	99%	1 in 15,801						•	•
ITGB3-related disorders	*ITGB3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Junctional epidermolysis bullosa, *ITGB4*-related	*ITGB4*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *JAK3*-related	*JAK3*	General Population	1 in 299	99%	1 in 29,801						•	•
Bartter syndrome, type 2	*KCNJ1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
LIG4 syndrome	*LIG4*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
LRAT-related disorders	*LRAT*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Retinitis pigmentosa 62	*MAK*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Retinitis pigmentosa 62	*MAK*	Ashkenazi Jewish	1 in 55	99%	1 in 5,401						•	•
Beta-mannosidosis	*MANBA*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Methylmalonyl-CoA epimerase deficiency	*MCEE*	General Population	<1 in 500	99%	<1 in 49,901						•	•
MECR-related neurologic disorder	*MECR*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Molybdenum cofactor deficiency of complementation group B	*MOCS2*	General Population	1 in 236	99%	1 in 23,501						•	•
Homocystinuria-megaloblastic anemia, cblG type	*MTR*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Congenital myasthenic syndrome, *MUSK*-related	*MUSK*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Chronic granulomatous disease 2	*NCF2*	General Population	1 in 500	99%	<1 in 49,901						•	•
Mitochondrial complex I deficiency, nuclear type 10	*NDUFAF2*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Mitochondrial complex I deficiency, nuclear type 3	*NDUFS7*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Mitochondrial complex I deficiency, nuclear type 4	*NDUFV1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Congenital disorder of deglycosylation, type 1	*NGLY1*	General Population	1 in 274	99%	1 in 27,301						•	•
Lung disease, immunodeficiency, and chromosome breakage syndrome	*NSMCE3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Osteopetrosis, *OSTM1*-related	*OSTM1*	General Population	<1 in 500	99%	<1 in 49,901						•	•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Nonsyndromic hearing loss and deafness 22	*OTOA*	General Population	<1 in 500	88%	1 in 4,159						•	•
Nonsyndromic hearing loss and deafness 9	*OTOF*	General Population	1 in 96	99%	1 in 9,501						•	•
Osteogenesis imperfecta, type VIII	*P3H1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Osteogenesis imperfecta, type VIII	*P3H1*	African American	1 in 240	99%	1 in 23,901						•	•
Pantothenate kinase-associated neurodegeneration	*PANK2*	General Population	1 in 289	99%	1 in 28,801						•	•
Pterin-4 alpha-carbinolamine dehydratase deficiency	*PCBD1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Microcephalic osteodysplastic primordial dwarfism, type II	*PCNT*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Microcephalic osteodysplastic primordial dwarfism, type II	*PCNT*	Druze–Northern Israel	1 in 30	99%	1 in 2,901						•	•
Zellweger spectrum disorder, *PEX13*-related	*PEX13*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Zellweger spectrum disorder, *PEX16*-related	*PEX16*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Zellweger spectrum disorder, *PEX5*-related	*PEX5*	General Population	1 in 408	99%	1 in 40,701						•	•
PGM3-congenital disorder of glycosylation/ Immunodeficiency 23	*PGM3*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Glycogen storage disease, type IXb	*PHKB*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Glycogen storage disease, type IXc	*PHKG2*	General Population	<1 in 500	99%	<1 in 49,901						•	•
PIGN-related disorders	*PIGN*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Nonsyndromic hearing loss and deafness 59	*PJVK*	General Population	<1 in 500	99%	<1 in 49,901						•	•
PLEKHG5-related disorders	*PLEKHG5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome	*PLOD1*	General Population	1 in 158	99%	1 in 15,701						•	•
Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Xeroderma pigmentosum, variant type	*POLH*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Xeroderma pigmentosum, variant type	*POLH*	Japanese	1 in 167	99%	1 in 16,601						•	•
Cytochrome P450 oxidoreductase deficiency	*POR*	General Population	1 in 370	99%	1 in 36,901						•	•
Combined or isolated pituitary hormone deficiency, type 1	*POU1F1*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Retinitis pigmentosa 36	*PRCD*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Brittle cornea syndrome 2	*PRDM5*	General Population	<1 in 500	99%	<1 in 49,901						•	•
Severe combined immunodeficiency, *PTPRC*-related	*PTPRC*	General Population	<1 in 500	99%	<1 in 49,901							•
Dihydropteridine reductase deficiency	*QDPR*	General Population	<1 in 500	99%	<1 in 49,901							•
RLBP1-related retinopathies	*RLBP1*	General Population	1 in 296	99%	1 in 29,501							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Retinitis pigmentosa 2, X-linked	*RP2*	General Population	1 in 58,000	99%	1 in 5,799,901							•
Muscular dystrophy-dystroglycanopathy, type A, 10	*RXYLT1*	General Population	<1 in 500	99%	<1 in 49,901							•
RYR1-related disorders	*RYR1*	General Population	1 in 150	99%	1 in 14,901							•
Normophosphatemic familial tumoral calcinosis	*SAMD9*	General Population	<1 in 500	99%	<1 in 49,901							•
Congenital dyserythropoietic anemia, type II	*SEC23B*	General Population	<1 in 500	99%	<1 in 49,901							•
Trichohepatoenteric syndrome 2	*SKIC2*	General Population	<1 in 500	99%	<1 in 49,901							•
Bartter syndrome, type 1	*SLC12A1*	General Population	1 in 360	99%	1 in 35,901							•
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*	General Population	<1 in 500	99%	<1 in 49,901							•
Congenital secretory chloride diarrhea 1	*SLC26A3*	General Population	<1 in 500	99%	<1 in 49,901							•
Ichthyosis prematurity syndrome	*SLC27A4*	General Population	<1 in 500	99%	<1 in 49,901							•
Ichthyosis prematurity syndrome	*SLC27A4*	Norwegian	1 in 50	99%	1 in 4,901							•
Ichthyosis prematurity syndrome	*SLC27A4*	Swedish	1 in 50	99%	1 in 4,901							•
Foveal hypoplasia 2	*SLC38A8*	General Population	<1 in 500	99%	<1 in 49,901							•
Oculocutaneous albinism, type IV	*SLC45A2*	General Population	1 in 158	99%	1 in 15,701							•
Oculocutaneous albinism, type IV	*SLC45A2*	Japanese	1 in 146	99%	1 in 14,501							•
Thyroid dyshormonogenesis 1	*SLC5A5*	General Population	<1 in 500	99%	<1 in 49,901							•
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*	General Population	<1 in 500	99%	<1 in 49,901							•
SPG11-related disorders	*SPG11*	General Population	1 in 141	99%	1 in 14,001							•
Sepiapterin reductase deficiency	*SPR*	General Population	<1 in 500	99%	<1 in 49,901							•
Amish infantile epilepsy syndrome	*ST3GAL5*	General Population	<1 in 500	99%	<1 in 49,901							•
Isolated sulfite oxidase deficiency	*SUOX*	General Population	<1 in 500	99%	<1 in 49,901							•
SURF1-related disorders	*SURF1*	General Population	1 in 316	99%	1 in 31,501							•
Nonsyndromic hearing loss and deafness 76	*SYNE4*	General Population	<1 in 500	99%	<1 in 49,901							•
Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum	*TBCD*	General Population	<1 in 500	99%	<1 in 49,901							•
TBCE-related disorders	*TBCE*	General Population	<1 in 500	99%	<1 in 49,901							•
Transcobalamin II deficiency	*TCN2*	General Population	<1 in 500	99%	<1 in 49,901							•
Dyskeratosis congenita spectrum disorders	*TERT*	General Population	<1 in 500	99%	<1 in 49,901							•
Thyroid dyshormonogenesis 3	*TG*	General Population	1 in 217	99%	1 in 21,601							•
TK2-related mitochondrial disorders	*TK2*	General Population	<1 in 500	99%	<1 in 49,901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Nonsyndromic hearing loss and deafness 7	*TMC1*	General Population	1 in 96	99%	1 in 9,501							•
TMEM67-related disorders	*TMEM67*	General Population	1 in 216	99%	1 in 21,501							•
Nonsyndromic hearing loss and deafness 8	*TMPRSS3*	General Population	1 in 96	99%	1 in 9,501							•
Thyroid dyshormonogenesis 2A	*TPO*	General Population	1 in 227	99%	1 in 22,601							•
TREX1-related disorders	*TREX1*	General Population	<1 in 500	99%	<1 in 49,901							•
TRIM32-related disorders	*TRIM32*	General Population	<1 in 500	99%	<1 in 49,901							•
TRIM32-related disorders	*TRIM32*	Hutterite	1 in 7	99%	1 in 601							•
Congenital hypothyroidism, *TSHB*-related	*TSHB*	General Population	<1 in 500	99%	<1 in 49,901							•
Congenital hypothyroidism, *TSHR*-related	*TSHR*	General Population	1 in 322	99%	1 in 32,101							•
Congenital hypothyroidism, *TSHR*-related	*TSHR*	Japanese	1 in 172	99%	1 in 17,101							•
TULP1-related disorders	*TULP1*	General Population	1 in 186	99%	1 in 18,501							•
Oculocutaneous albinism, type III	*TYRP1*	General Population	1 in 488	99%	1 in 48,701							•
Familial hemophagocytic lymphohistiocytosis 3	*UNC13D*	General Population	<1 in 500	99%	<1 in 49,901							•
Familial hemophagocytic lymphohistiocytosis 3	*UNC13D*	Swedish	1 in 187	99%	1 in 18,601							•
Vitamin D-resistant rickets, type 2A	*VDR*	General Population	<1 in 500	99%	<1 in 49,901							•
VLDLR-associated cerebellar hypoplasia	*VLDLR*	General Population	<1 in 500	99%	<1 in 49,901							•
VLDLR-associated cerebellar hypoplasia	*VLDLR*	Hutterite	1 in 15	99%	1 in 1,401							•
Hypomyelinating leukodystrophy 12	*VPS11*	General Population	<1 in 500	99%	<1 in 49,901							•
Hypomyelinating leukodystrophy 12	*VPS11*	Ashkenazi Jewish	1 in 160	99%	1 in 15,901							•
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	*ZBTB24*	General Population	<1 in 500	99%	<1 in 49,901							•
Brittle cornea syndrome 1	*ZNF469*	General Population	<1 in 500	99%	<1 in 49,901							•
Methylmalonic aciduria and homocystinuria type cblJ	*ABCD4*	General Population	<1 in 500	99%	<1 in 49901							•
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*	African American	1 in 36	99%	1 in 3501							•
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*	Ashkenazi Jewish	1 in 15	99%	1 in 1401							•
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*	Caucasian	1 in 14	99%	1 in 1301							•
Short chain acyl-CoA dehydrogenase deficiency	*ACADS*	General Population	1 in 94	99%	1 in 9301							•
Short/branched chain acyl-CoA dehydrogenase deficiency	*ACADSB*	Hmong	1 in 8	99%	1 in 701							•
Short/branched chain acyl-CoA dehydrogenase deficiency	*ACADSB*	General Population	<1 in 500	99%	<1 in 49901							•
Adenosine kinase deficiency	*ADK*	General Population	<1 in 500	99%	<1 in 49901							•

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Berardinelli-Siep congenital lipodystrophy	*AGPAT2*	General Population	<1 in 500	99%	<1 in 49901							•
S-adenosylhomocysteine hydrolase deficiency	*AHCY*	General Population	<1 in 500	99%	<1 in 49901							•
Hyper IgM syndrome type 2	*AICDA*	General Population	<1 in 500	99%	<1 in 49901							•
Hypomyelinating leukodystrophy 3	*AIMP1*	General Population	<1 in 500	99%	<1 in 49901							•
Reticular dysgenesis	*AK2*	General Population	<1 in 500	99%	<1 in 49901							•
Congenital bile acid synthesis defect 2	*AKR1D1*	General Population	<1 in 500	99%	<1 in 49901							•
Hyperprolinemia type II	*ALDH4A1*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *ALOX12B*-related	*ALOX12B*	General Population	1 in 488	99%	1 in 48701							•
Autosomal recessive congenital ichthyosis, *ALOXE3*-related	*ALOXE3*	General Population	<1 in 500	99%	<1 in 49901							•
Persistent Müllerian duct syndrome, *AMH*-related	*AMH*	General Population	<1 in 500	99%	<1 in 49901							•
Persistent Müllerian duct syndrome, *AMHR2*-related	*AMHR2*	General Population	<1 in 500	99%	<1 in 49901							•
Pontocerebellar hypoplasia, *AMPD2*-related	*AMPD2*	General Population	<1 in 500	99%	<1 in 49901							•
ANO5-related anoctaminopathy	*ANO5*	Finnish	1 in 112	99%	1 in 11101							•
ANO5-related anoctaminopathy	*ANO5*	General Population	1 in 228	99%	1 in 22701							•
Hyaline fibromatosis syndrome	*ANTXR2*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *AP1S2*-related (X-linked)	*AP1S2*	General Population	<1 in 750000	99%	<1 in 74999901							•
Hermansky-Pudlak syndrome, *AP3D1*-related	*AP3D1*	General Population	<1 in 500	99%	<1 in 49901							•
Kufor-Rakeb syndrome	*ATP13A2*	General Population	<1 in 500	99%	<1 in 49901							•
Cutis laxa, *ATP6V0A2*-related	*ATP6V0A2*	General Population	<1 in 500	99%	<1 in 49901							•
Hereditary distal renal tubular acidosis, *ATP6V0A4*-related	*ATP6V0A4*	General Population	<1 in 500	99%	<1 in 49901							•
Cutis laxa, *ATP6V1E1*-related	*ATP6V1E1*	General Population	<1 in 500	99%	<1 in 49901							•
Diaphanospondylodysostosis	*BMPER*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *BRWD3*-related (X-linked)	*BRWD3*	General Population	<1 in 750000	99%	<1 in 74999901							•
Mitochondrial membrane protein-associated neurodegeneration	*C19orf12*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *CASP14*-related	*CASP14*	General Population	<1 in 500	99%	<1 in 49901							•
Neonatal primary hyperparathyrodism	*CASR*	General Population	<1 in 500	99%	<1 in 49901							•
Congenital generalized lipodystrophy type 4	*CAVIN1*	General Population	<1 in 500	99%	<1 in 49901							•
3M syndrome 3	*CCDC8*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Severe combined immunodeficiency, *CD247*-related	*CD247*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency, *CD3G*-related	*CD3G*	General Population	<1 in 500	99%	<1 in 49901							•
Immunodeficiency, *CD8A*-related	*CD8A*	General Population	<1 in 500	99%	<1 in 49901							•
Congenital dyserythropoietic anemia type I, *CDAN1*-related	*CDAN1*	General Population	<1 in 500	99%	<1 in 49901							•
Immunodeficiency-centromeric instability-facial anomalies syndrome 3	*CDCA7*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *CERS3*-related	*CERS3*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *CFAP418*-related	*CFAP418*	General Population	<1 in 500	99%	<1 in 49901							•
Pontocerebellar hypoplasia, *CHMP1A*-related	*CHMP1A*	General Population	<1 in 500	99%	<1 in 49901							•
Macular corneal dystrophy	*CHST6*	General Population	1 in 53	99%	1 in 5201							•
Cold-induced sweating syndrome 2	*CLCF1*	General Population	<1 in 500	99%	<1 in 49901							•
Bartter syndrome type 3	*CLCNKB*	General Population	<1 in 500	99%	<1 in 49901							•
Pontocerebellar hypoplasia, *CLP1*-related	*CLP1*	General Population	<1 in 500	99%	<1 in 49901							•
Retinitis pigmentosa 49	*CNGA1*	General Population	1 in 433	99%	1 in 43201							•
Retinal dystrophy, *CNGA3*-related	*CNGA3*	Caucasian	1 in 158	99%	1 in 15701							•
Retinal dystrophy, *CNGA3*-related	*CNGA3*	General Population	1 in 173	99%	1 in 17201							•
Retinitis pigmentosa 45	*CNGB1*	General Population	<1 in 500	99%	<1 in 49901							•
Cortical dysplasia-focal epilepsy syndrome	*CNTNAP2*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex IV deficiency nuclear type 17	*COA8*	General Population	<1 in 500	99%	<1 in 49901							•
COASY-related conditions	*COASY*	General Population	<1 in 500	99%	<1 in 49901							•
COQ4-related conditions	*COQ4*	General Population	<1 in 500	99%	<1 in 49901							•
Severe combined immunodeficiency, *CORO1A*-related	*CORO1A*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex IV deficiency nuclear type 3	*COX10*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex IV deficiency nuclear type 11	*COX20*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex IV deficiency nuclear type 7	*COX6B1*	General Population	<1 in 500	99%	<1 in 49901							•
Aceruloplasminemia	*CP*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *CRADD*-related	*CRADD*	Finnish	1 in 169	99%	1 in 16801							•
Intellectual disability, *CRADD*-related	*CRADD*	Mennonite	1 in 58	99%	1 in 5701							•
Intellectual disability, *CRADD*-related	*CRADD*	General Population	<1 in 500	99%	<1 in 49901							•
Cold-induced sweating syndrome 1	*CRLF1*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Dyskeratosis congenita and related telomere biology disorders, *CTC1*-related	*CTC1*	General Population	<1 in 500	99%	<1 in 49901							•
CLN13 disease	*CTSF*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *CUL4B*-related (X-linked)	*CUL4B*	General Population	1 in 26700	99%	1 in 2669901							•
3M syndrome 1	*CUL7*	General Population	<1 in 500	99%	<1 in 49901							•
Metaphyseal chondrodysplasia-retinitis pigmentosa syndrome	*CWC27*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *CYP4F22*-related	*CYP4F22*	General Population	<1 in 500	99%	<1 in 49901							•
Xeroderma pigmentosum group E	*DDB2*	General Population	<1 in 500	99%	<1 in 49901							•
Aromatic L-amino acid decarboxylase deficiency	*DDC*	General Population	<1 in 500	99%	<1 in 49901							•
Spondylometaepiphyseal dysplasia-short limb-abnormal calcification syndrome	*DDR2*	General Population	<1 in 500	99%	<1 in 49901							•
Desmosterolosis	*DHCR24*	General Population	<1 in 500	99%	<1 in 49901							•
Pyruvate dehydrogenase complex deficiency, *DLAT*-related	*DLAT*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *DLG3*-related (X-linked)	*DLG3*	General Population	<1 in 750000	99%	<1 in 74999901							•
Primary ciliary dyskinesia, *DNAL1*-related	*DNAL1*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency, *DOCK8*-related	*DOCK8*	General Population	<1 in 500	99%	<1 in 49901							•
DOLK-congenital disorder of glycosylation	*DOLK*	General Population	<1 in 500	99%	<1 in 49901							•
Hermansky-Pudlak syndrome, *DTNBP1*-related	*DTNBP1*	General Population	<1 in 500	99%	<1 in 49901							•
Thyroid dyshormonogenesis, *DUOXA2*-related	*DUOXA2*	General Population	<1 in 500	99%	<1 in 49901							•
Cutis laxa, *EFEMP2*-related	*EFEMP2*	General Population	<1 in 500	99%	<1 in 49901							•
Hereditary spherocytosis, *EPB42*-related	*EPB42*	Japanese	1 in 33	99%	1 in 3201							•
Hereditary spherocytosis, *EPB42*-related	*EPB42*	General Population	1 in 71	99%	1 in 7001							•
Chronic intestinal pseudoobstruction, *ERBB3*-related	*ERBB3*	General Population	<1 in 500	99%	<1 in 49901							•
ERCC3-related conditions	*ERCC3*	General Population	<1 in 500	99%	<1 in 49901							•
ERCC4-related conditions	*ERCC4*	General Population	<1 in 500	99%	<1 in 49901							•
ERCC5-related conditions	*ERCC5*	General Population	<1 in 500	99%	<1 in 49901							•
Factor VII deficiency	*F7*	General Population	1 in 274	99%	1 in 27301							•
Fatty acid hydroxylase-associated neurodegeneration	*FA2H*	General Population	<1 in 500	99%	<1 in 49901							•
FBXL4-related mitochondrial DNA depletion syndrome, encephalomyopathic form	*FBXL4*	General Population	<1 in 500	99%	<1 in 49901							•
Aarskog-Scott syndrome (X-linked)	*FGD1*	General Population	1 in 33300	99%	1 in 3329901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Cerebral folate transport deficiency	*FOLR1*	General Population	<1 in 500	99%	<1 in 49901							•
Formiminoglutamic aciduria	*FTCD*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *FTSJ1*-related (X-linked)	*FTSJ1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Charcot-Marie-Tooth disease, *GDAP1*-related	*GDAP1*	General Population	1 in 124	99%	1 in 12301							•
Congenital myasthenic syndrome, *GFPT1*-related	*GFPT1*	General Population	<1 in 500	99%	<1 in 49901							•
Isolated growth hormone deficiency type IV	*GHRHR*	General Population	<1 in 500	99%	<1 in 49901							•
Isolated GnRH deficiency, *GNRHR*-related	*GNRHR*	General Population	1 in 112	99%	1 in 11101							•
Molybdenum cofactor deficiency type C	*GPHN*	General Population	<1 in 500	99%	<1 in 49901							•
Foveal hypoplasia, *GPR143*-related (X-linked)	*GPR143*	General Population	1 in 30000	99%	1 in 2999901							•
Glycogen storage disease type 0	*GYS2*	General Population	<1 in 500	99%	<1 in 49901							•
Immunodeficiency-centromeric instability-facial anomalies syndrome 4	*HELLS*	General Population	<1 in 500	99%	<1 in 49901							•
Charcot-Marie-Tooth disease, neuromyotonia and axonal neuropathy	*HINT1*	General Population	<1 in 500	99%	<1 in 49901							•
HMGCS2 deficiency	*HMGCS2*	General Population	<1 in 500	99%	<1 in 49901							•
HSD3B7 deficiency	*HSD3B7*	General Population	<1 in 500	99%	<1 in 49901							•
Hypomyelinating leukodystrophy 5	*HYCC1*	General Population	<1 in 500	99%	<1 in 49901							•
Retinitis pigmentosa 46	*IDH3B*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *IFT140*-related	*IFT140*	General Population	<1 in 500	99%	<1 in 49901							•
IGSF1 deficiency (X-linked)	*IGSF1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Intellectual disability, *IL1RAPL1*-related (X-linked)	*IL1RAPL1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Combined immunodeficiency, *IL2RA*-related	*IL2RA*	General Population	<1 in 500	99%	<1 in 49901							•
Glanzmann thrombasthenia, *ITGA2B*-related	*ITGA2B*	General Population	<1 in 500	99%	<1 in 49901							•
Developmental and epileptic encephalopathy 35	*ITPA*	General Population	<1 in 500	99%	<1 in 49901							•
Thyroid dyshormonogenesis, *IYD*-related	*IYD*	General Population	<1 in 500	99%	<1 in 49901							•
Progressive myoclonic epilepsy type 3	*KCTD7*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *KDM5C*-related (X-linked)	*KDM5C*	General Population	1 in 28600	99%	1 in 2859901							•
Primary microcephaly type 20	*KIF14*	General Population	<1 in 500	99%	<1 in 49901							•
Danon disease (X-linked)	*LAMP2*	General Population	1 in 16700	99%	1 in 1669901							•
Infantile liver failure syndrome type 1	*LARS1*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency, *LCK*-related	*LCK*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Leydig cell hypoplasia	*LHCGR*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *LIPN*-related	*LIPN*	General Population	<1 in 500	99%	<1 in 49901							•
Combined factor V and factor VIII deficiency, *LMAN1*-related	*LMAN1*	General Population	1 in 189	99%	1 in 18801							•
Autosomal recessive woolly hair/hypotrichosis, *LPAR6*-related	*LPAR6*	General Population	<1 in 500	99%	<1 in 49901							•
Cutis laxa, *LTBP4*-related	*LTBP4*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency, *MALT1*-related	*MALT1*	General Population	<1 in 500	99%	<1 in 49901							•
Methionine adenosyltransferase I/III deficiency	*MAT1A*	General Population	<1 in 500	99%	<1 in 49901							•
Carpenter syndrome, *MEGF8*-related	*MEGF8*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency and megaloblastic anemia, *MTHFD1*-related	*MTHFD1*	General Population	<1 in 500	99%	<1 in 49901							•
Charcot-Marie-Tooth disease type 4B1	*MTMR2*	General Population	<1 in 500	99%	<1 in 49901							•
NBAS-related conditions	*NBAS*	General Population	<1 in 500	99%	<1 in 49901							•
Gray platelet syndrome	*NBEAL2*	General Population	<1 in 500	99%	<1 in 49901							•
Chronic granulomatous disease, *NCF4*-related	*NCF4*	General Population	<1 in 500	99%	<1 in 49901							•
Retinal dystrophy, *NDP*-related (X-linked)	*NDP*	General Population	<1 in 750000	99%	<1 in 74999901							•
Mitochondrial complex I deficiency nuclear type 14	*NDUFA11*	General Population	<1 in 500	99%	<1 in 49901							•
Severe combined immunodeficiency, *NHEJ1*-related	*NHEJ1*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *NIPAL4*-related	*NIPAL4*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *NONO*-related (X-linked)	*NONO*	General Population	<1 in 750000	99%	<1 in 74999901							•
3M syndrome 2	*OBSL1*	General Population	<1 in 500	99%	<1 in 49901							•
Primary ciliary dyskinesia, *ODAD3*-related	*ODAD3*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *OPHN1*-related (X-linked)	*OPHN1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Intellectual disability, *PAK3*-related (X-linked)	*PAK3*	General Population	<1 in 750000	99%	<1 in 74999901							•
Retinitis pigmentosa 43	*PDE6A*	General Population	1 in 274	99%	1 in 27301							•
Pyruvate dehydrogenase complex deficiency, *PDHX*-related	*PDHX*	General Population	<1 in 500	99%	<1 in 49901							•
Pyruvate dehydrogenase complex deficiency, *PDP1*-related	*PDP1*	General Population	<1 in 500	99%	<1 in 49901							•
Peroxisome biogenesis disorder-Zellweger spectrum disorder, *PEX11B*-related	*PEX11B*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Peroxisome biogenesis disorder-Zellweger spectrum disorder, *PEX14*-related	*PEX14*	General Population	<1 in 500	99%	<1 in 49901							•
Peroxisome biogenesis disorder-Zellweger spectrum disorder, *PEX19*-related	*PEX19*	General Population	<1 in 500	99%	<1 in 49901							•
Peroxisome biogenesis disorder-Zellweger spectrum disorder, *PEX3*-related	*PEX3*	General Population	<1 in 500	99%	<1 in 49901							•
PGK1 deficiency (X-linked)	*PGK1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Intellectual disability, *PHF8*-related (X-linked)	*PHF8*	General Population	<1 in 750000	99%	<1 in 74999901							•
Glycogen storage disease type IXd (X-linked)	*PHKA1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Glycogen storage disease type IXa (X-linked)	*PHKA2*	General Population	1 in 266000	99%	1 in 26599901							•
Bruck syndrome, *PLOD2*-related	*PLOD2*	General Population	<1 in 500	99%	<1 in 49901							•
Purine nucleoside phosphorylase deficiency	*PNP*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *PNPLA1*-related	*PNPLA1*	General Population	<1 in 500	99%	<1 in 49901							•
SOFT syndrome	*POC1A*	General Population	<1 in 500	99%	<1 in 49901							•
POLR1C-related conditions	*POLR1C*	General Population	<1 in 500	99%	<1 in 49901							•
Nonsyndromic hearing loss, *POU3F4*-related (X-linked)	*POU3F4*	General Population	1 in 60000	99%	1 in 5999901							•
Osteogenesis imperfecta type IX	*PPIB*	General Population	<1 in 500	99%	<1 in 49901							•
Renpenning syndrome (X-linked)	*PQBP1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Progressive myoclonic epilepsy type 1B	*PRICKLE1*	General Population	<1 in 500	99%	<1 in 49901							•
Severe combined immunodeficiency, *PRKDC*-related	*PRKDC*	General Population	<1 in 500	99%	<1 in 49901							•
Cutis laxa, *PYCR1*-related	*PYCR1*	General Population	<1 in 500	99%	<1 in 49901							•
Glycogen storage disease type VI	*PYGL*	Mennonite	1 in 16	99%	1 in 1501							•
Glycogen storage disease type VI	*PYGL*	General Population	1 in 127	99%	1 in 12601							•
Microphthalmia/anophthalmia, *RAX*-related	*RAX*	General Population	<1 in 500	99%	<1 in 49901							•
Retinal dystrophy, *RD3*-related	*RD3*	General Population	<1 in 500	99%	<1 in 49901							•
Retinal dystrophy, *RDH5*-related	*RDH5*	General Population	<1 in 500	99%	<1 in 49901							•
Bare lymphocyte syndrome type II, *RFX5*-related	*RFX5*	General Population	<1 in 500	99%	<1 in 49901							•
Bare lymphocyte syndrome type II, *RFXANK*-related	*RFXANK*	General Population	<1 in 500	99%	<1 in 49901							•
Bare lymphocyte syndrome type II, *RFXAP*-related	*RFXAP*	General Population	<1 in 500	99%	<1 in 49901							•
Rh deficiency syndrome	*RHAG*	General Population	<1 in 500	99%	<1 in 49901							•
Kohlschutter-Tonz syndrome	*ROGDI*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Retinal dystrophy, *RPGRIP1*-related	*RPGRIP1*	General Population	1 in 406	99%	1 in 40501							•
Intellectual disability, *RPS6KA3*-related (X-linked)	*RPS6KA3*	General Population	1 in 30000	99%	1 in 2999901							•
Primary ciliary dyskinesia, *RSPH9*-related	*RSPH9*	General Population	<1 in 500	99%	<1 in 49901							•
Retinal dystrophy, *SAG*-related	*SAG*	General Population	<1 in 500	99%	<1 in 49901							•
HUPRA syndrome	*SARS2*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex IV deficiency nuclear type 4	*SCO1*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *SDCCAG8*-related	*SDCCAG8*	General Population	<1 in 500	99%	<1 in 49901							•
Autosomal recessive congenital ichthyosis, *SDR9C7*-related	*SDR9C7*	General Population	<1 in 500	99%	<1 in 49901							•
Rigid spine syndrome, *SELENON*-related	*SELENON*	General Population	<1 in 500	99%	<1 in 49901							•
Osteogenesis imperfecta type VI	*SERPINF1*	General Population	<1 in 500	99%	<1 in 49901							•
Congenital arthrogryposis, *PIP5K1C*-related	*PIP5K1C*	General Population	<1 in 500	99%	<1 in 49901							•
Charcot-Marie-Tooth disease type 4C	*SH3TC2*	General Population	1 in 166	99%	1 in 16501							•
Allan-Herndon-Dudley syndrome (X-linked)	*SLC16A2*	General Population	1 in 93300	99%	1 in 9329901							•
Arterial tortuosity syndrome	*SLC2A10*	General Population	<1 in 500	99%	<1 in 49901							•
Fanconi-Bickel syndrome	*SLC2A2*	General Population	<1 in 500	99%	<1 in 49901							•
Hereditary hypophosphatemic rickets with hypercalciuria	*SLC34A3*	General Population	<1 in 500	99%	<1 in 49901							•
Cystinuria type A	*SLC3A1*	General Population	1 in 62	99%	1 in 6101							•
Hereditary folate malabsorption	*SLC46A1*	General Population	<1 in 500	99%	<1 in 49901							•
Hereditary folate malabsorption	*SLC46A1*	Puerto Rican	1 in 16	99%	1 in 1501							•
SLC4A1-related conditions	*SLC4A1*	General Population	<1 in 500	99%	<1 in 49901							•
Hartnup disease	*SLC6A19*	General Population	1 in 87	99%	1 in 8601							•
Cystinuria type B	*SLC7A9*	Libyan Jewish	1 in 26	99%	1 in 2501							•
Cystinuria type B	*SLC7A9*	General Population	1 in 57	99%	1 in 5601							•
Malignant infantile osteopetrosis, *SNX10*-related	*SNX10*	General Population	<1 in 500	99%	<1 in 49901							•
Hepatic veno-occlusive disease with immunodeficiency	*SP110*	Lebanese	1 in 25	99%	1 in 2401							•
Hepatic veno-occlusive disease with immunodeficiency	*SP110*	General Population	<1 in 500	99%	<1 in 49901							•
Retinal dystrophy, *SPATA7*-related	*SPATA7*	General Population	<1 in 500	99%	<1 in 49901							•
Spastic paraplegia 21	*SPG21*	General Population	<1 in 500	99%	<1 in 49901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Spastic paraplegia 7	*SPG7*	General Population	1 in 337	99%	1 in 33601							•
Netherton syndrome	*SPINK5*	General Population	1 in 224	99%	1 in 22301	•						•
Combined immunodeficiency, *STK4*-related	*STK4*	General Population	<1 in 500	99%	<1 in 49901							•
SUCLA2-related mitochondrial DNA depletion syndrome, encephalomyopathic form	*SUCLA2*	Faroese	1 in 33	99%	1 in 3201							•
SUCLA2-related mitochondrial DNA depletion syndrome, encephalomyopathic form	*SUCLA2*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *SYN1*-related (X-linked)	*SYN1*	General Population	<1 in 750000	99%	<1 in 74999901							•
Congenital isolated adrenocorticotropic hormone deficiency	*TBX19*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *TCTN1*-related	*TCTN1*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *TCTN2*-related	*TCTN2*	Ethiopian Jewish	1 in 42	99%	1 in 4101							•
Ciliopathies, *TCTN2*-related	*TCTN2*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *TCTN2*-related	*TCTN2*	Yemenite Jewish	1 in 77	99%	1 in 7601							•
Ciliopathies, *TCTN3*-related	*TCTN3*	General Population	<1 in 500	99%	<1 in 49901							•
THOC2-related conditions (X-linked)	*THOC2*	General Population	<1 in 750000	99%	<1 in 74999901							•
Ciliopathies, *TMEM138*-related	*TMEM138*	General Population	<1 in 500	99%	<1 in 49901							•
Ciliopathies, *TMEM231*-related	*TMEM231*	General Population	<1 in 500	99%	<1 in 24951							•
Joubert syndrome, *TMEM237*-related	*TMEM237*	General Population	<1 in 500	99%	<1 in 49901							•
Osteogenesis imperfecta type XIV	*TMEM38B*	General Population	<1 in 500	99%	<1 in 49901							•
Mitochondrial complex V deficiency nuclear type 2	*TMEM70*	General Population	<1 in 500	99%	<1 in 49901							•
Malignant infantile osteopetrosis, *TNFSF11*-related	*TNFSF11*	General Population	<1 in 500	99%	<1 in 49901							•
Limb-girdle muscular dystrophy R19	*TRAPPC11*	General Population	<1 in 500	99%	<1 in 49901							•
Catecholaminergic polymorphic ventricular tachycardia, *TRDN*-related	*TRDN*	General Population	<1 in 500	99%	<1 in 49901							•
Isolated congenital hypothyroidism, *TRHR*-related	*TRHR*	General Population	<1 in 500	99%	<1 in 49901							•
Hypomagnesemia with secondary hypocalcemia	*TRPM6*	General Population	<1 in 500	99%	<1 in 49901							•
Pontocerebellar hypoplasia, *TSEN34*-related	*TSEN34*	General Population	<1 in 500	99%	<1 in 49901							•
Gastrointestinal defects and immunodeficiency syndrome 1	*TTC7A*	General Population	<1 in 500	99%	<1 in 49901							•
Crigler-Najjar syndrome	*UGT1A1*	General Population	1 in 158	99%	1 in 15701							•
Hyper IgM syndrome type 5	*UNG*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *UPF3B*-related (X-linked)	*UPF3B*	General Population	<1 in 750000	99%	<1 in 74999901							•

Genetic condition	Gene	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	QHerit 6 **Female** 6 conditions 7 genes Test code 14011 **Male** 4 conditions 5 genes Test code 14010	QHerit 28 **Female** 28 conditions 29 genes Test code 18294 **Male** 26 conditions 27 genes Test code 18293	QHerit 112 **Female** 112 conditions 113 genes Test code 14232 **Male** 96 conditions 97 genes Test code 14227	QHerit 198 **Female** 198 conditions 199 genes Test code 14230 **Male** 179 conditions 180 genes Test code 14231	QHerit 445 **Female** 445 conditions 446 genes Test code 13832 **Male** 400 conditions 401 genes Test code 13831	QHerit 611 **Female** 611 conditions 612 genes Test code 14228 **Male** 559 conditions 560 genes Test code 14229	QHerit 833 **Female** 833 conditions 834 genes Test code 22861 **Male** 753 conditions 754 genes Test code 22862
Usher syndrome type 1G	*USH1G*	General Population	<1 in 500	99%	<1 in 49901							•
Usher syndrome type 2D	*WHRN*	General Population	1 in 363	99%	1 in 36201							•
Osteogenesis imperfecta type XV	*WNT1*	General Population	<1 in 500	99%	<1 in 49901							•
Combined immunodeficiency, *ZAP70*-related	*ZAP70*	Mennonite	1 in 10	99%	1 in 901							•
Combined immunodeficiency, *ZAP70*-related	*ZAP70*	General Population	<1 in 500	99%	<1 in 49901							•
Intellectual disability, *ZDHHC9*-related (X-linked)	*ZDHHC9*	General Population	<1 in 750000	99%	<1 in 74999901							•
Intellectual disability, *ZNF711*-related (X-linked)	*ZNF711*	General Population	<1 in 750000	99%	<1 in 74999901							•

* Data related to ethnicity, carrier frequency, detection rate, and residual risk are reported by Baylor Genetics®.

Important Information

QHerit® 6, QHerit 28, QHerit 112, QHerit 198, QHerit 445, QHerit 611, and QHerit 833 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 6 screens 7 genes; QHerit 28 screens 29 genes; QHerit 112 screens 113 genes; QHerit 198 screens 199 genes; QHerit 445 screens 446 genes; QHerit 611 screens 612 genes; and QHerit 883 screens 834 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 the FDA.

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