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. 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 Liaison.

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	Autosomal Recessive	General Population	1 in 116	99%	1 in 11,501	•	•	•	•
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	Autosomal Recessive	African American	1 in 68	99%	1 in 6,701	•	•	•	•
Surfactant dysfunction, *ABCA3*-related	*ABCA3*	Autosomal Recessive	Caucasian	1 in 28	99%	1 in 2,701	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	Autosomal Recessive	General Population	1 in 112	99%	1 in 11,101	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	Autosomal Recessive	Ashkenazi Jewish	1 in 52	99%	1 in 5,101	•	•	•	•
Familial hyperinsulinism, *ABCC8*-related	*ABCC8*	Autosomal Recessive	Finnish	1 in 29	99%	1 in 2,801	•	•	•	•
Adrenoleukodystrophy, X-linked	*ABCD1*	X-Linked	General Population	1 in 10,500	99%	1 in 1,049,901	•	•	•	•
Adrenoleukodystrophy, X-linked	*ABCD1*	X-Linked	Sephardic Jewish	1 in 10,500	99%	1 in 1,049,901	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	Autosomal Recessive	Asian	1 in 178	99%	1 in 17,701	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	Autosomal Recessive	Caucasian	1 in 64	99%	1 in 6,301	•	•	•	•
Medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	Autosomal Recessive	General Population	1 in 35	99%	1 in 3,401	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	Autosomal Recessive	Asian	1 in 194	99%	1 in 19,301	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	Autosomal Recessive	Caucasian	1 in 88	99%	1 in 8,701	•	•	•	•
Very long-chain acyl-CoA dehydrogenase deficiency	*ACADVL*	Autosomal Recessive	General Population	1 in 86	99%	1 in 8,501	•	•	•	•
Beta-ketothiolase deficiency	*ACAT1*	Autosomal Recessive	Caucasian	1 in 354	99%	1 in 35,301	•	•	•	•
Beta-ketothiolase deficiency	*ACAT1*	Autosomal Recessive	General Population	1 in 347	99%	1 in 34,601	•	•	•	•
Beta-ketothiolase deficiency	*ACAT1*	Autosomal Recessive	Asian	1 in 289	99%	1 in 28,801	•	•	•	•
Fragile XE syndrome	*AFF2*	X-Linked	General Population	1 in 18,750	99%	1 in 1,874,901	•	•	•	•
Aspartylglycosaminuria	*AGA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Aspartylglycosaminuria	*AGA*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Aspartylglycosaminuria	*AGA*	Autosomal Recessive	Finnish	1 in 36	99%	1 in 3,501	•	•	•	•
Primary hyperoxaluria, type 1	*AGXT*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Joubert syndrome 3	*AHI1*	Autosomal Recessive	General Population	1 in 176	99%	1 in 17,501	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Autosomal Recessive	General Population	1 in 354	99%	1 in 35,301	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Autosomal Recessive	Finnish	1 in 79	99%	1 in 7,801	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Autosomal Recessive	Sardinian	1 in 60	99%	1 in 5,901	•	•	•	•
Autoimmune polyglandular syndrome, type 1	*AIRE*	Autosomal Recessive	Sephardic Jewish–Iranian	1 in 27	99%	1 in 2,601	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Autosomal Recessive	Hispanic	<1 in 500	99%	<1 in 49,901	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Autosomal Recessive	African American	1 in 406	99%	1 in 40,501	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Autosomal Recessive	Caucasian	1 in 80	99%	1 in 7,901	•	•	•	•
Hereditary fructose intolerance	*ALDOB*	Autosomal Recessive	General Population	1 in 55	99%	1 in 5,401	•	•	•	•
Hypophosphatasia	*ALPL*	Autosomal Recessive	Northern European Caucasian	1 in 274	99%	1 in 27,301	•	•	•	•

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Hypophosphatasia	*ALPL*	Autosomal Recessive	East Asian	1 in 203	99%	1 in 20,201	•	•	•	•
Hypophosphatasia	*ALPL*	Autosomal Recessive	Asian	1 in 203	99%	1 in 20,201	•	•	•	•
Hypophosphatasia	*ALPL*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Hypophosphatasia	*ALPL*	Autosomal Recessive	Mennonite	1 in 25	99%	1 in 2,401	•	•	•	•
Autosomal recessive spinocerebellar ataxia, type 10	*ANO10*	Autosomal Recessive	General Population	1 in 93	99%	1 in 9,201	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Autosomal Recessive	General Population	1 in 100	99%	1 in 9,901	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Autosomal Recessive	Sephardic Jewish–Yemenite	1 in 46	99%	1 in 4,501	•	•	•	•
Metachromatic leukodystrophy, *ARSA*-related	*ARSA*	Autosomal Recessive	Navajo	1 in 25	99%	1 in 2,401	•	•	•	•
X-linked developmental disorders, *ARX*-related	*ARX*	X-Linked	General Population	<1 in 750,000	50%	<1 in 1,499,999	•	•	•	•
Argininosuccinic aciduria	*ASL*	Autosomal Recessive	Finnish	1 in 190	99%	1 in 18,901	•	•	•	•
Argininosuccinic aciduria	*ASL*	Autosomal Recessive	General Population	1 in 133	99%	1 in 13,201	•	•	•	•
Canavan disease	*ASPA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Canavan disease	*ASPA*	Autosomal Recessive	Ashkenazi Jewish	1 in 60	99%	1 in 5,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Southern European Caucasian	1 in 250	99%	1 in 24,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Northern European Caucasian	1 in 90	99%	1 in 8,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	General Population	1 in 90	99%	1 in 8,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Caucasian	1 in 90	99%	1 in 8,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Ashkenazi Jewish	1 in 70	99%	1 in 6,901	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Sephardic Jewish–North African, Iraqi, Yemenite, Iranian, Bukharan	1 in 65	99%	1 in 6,401	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Sardinian	1 in 42	99%	1 in 4,101	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	East Asian	1 in 27	99%	1 in 2,601	•	•	•	•
Wilson disease	*ATP7B*	Autosomal Recessive	Canary Islands	1 in 25	99%	1 in 2,401	•	•	•	•
Bardet-Biedl syndrome 1	*BBS1*	Autosomal Recessive	General Population	1 in 265	99%	1 in 26,401	•	•	•	•
Bardet-Biedl syndrome 1	*BBS1*	Autosomal Recessive	Faroese	1 in 30	99%	1 in 2,901	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	Autosomal Recessive	Ashkenazi Jewish	1 in 135	99%	1 in 13,401	•	•	•	•
Bardet-Biedl syndrome 2	*BBS2*	Autosomal Recessive	Hutterite	1 in 22	99%	1 in 2,101	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Autosomal Recessive	Caucasian	1 in 433	99%	1 in 43,201	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Autosomal Recessive	General Population	1 in 364	99%	1 in 36,301	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Autosomal Recessive	Asian	1 in 163	99%	1 in 16,201	•	•	•	•
Maple syrup urine disease, type 1B	*BCKDHB*	Autosomal Recessive	Ashkenazi Jewish	1 in 97	99%	1 in 9,601	•	•	•	•
Bloom syndrome	*BLM*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Bloom syndrome	*BLM*	Autosomal Recessive	Ashkenazi Jewish	1 in 140	99%	1 in 13,901	•	•	•	•
Biotinidase deficiency	*BTD*	Autosomal Recessive	General Population	1 in 120	99%	1 in 11,901	•	•	•	•
Biotinidase deficiency	*BTD*	Autosomal Recessive	Hispanic	1 in 30	99%	1 in 2,901	•	•	•	•
Biotinidase deficiency	*BTD*	Autosomal Recessive	Caucasian	1 in 12	99%	1 in 1,101	•	•	•	•
Homocystinuria, *CBS*-related	*CBS*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301	•	•	•	•
Homocystinuria, *CBS*-related	*CBS*	Autosomal Recessive	Caucasian	1 in 52	99%	1 in 5,101	•	•	•	•

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Homocystinuria, *CBS*-related	*CBS*	Autosomal Recessive	Qatari	1 in 21	99%	1 in 2,001	•	•	•	•
Joubert syndrome 9	*CC2D2A*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Congenital hydrocephalus 1	*CCDC88C*	Autosomal Recessive	General Population	1 in 137	99%	1 in 13,601	•	•	•	•
Leber congenital amaurosis, *CEP290*-related/*CEP290*-related conditions	*CEP290*	Autosomal Recessive	General Population	1 in 185	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	South Asian	1 in 90	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	Southeast Asian	1 in 90	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	East Asian	1 in 90	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	African American	1 in 61	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	Hispanic	1 in 46	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	Northern European Caucasian	1 in 25	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	Ashkenazi Jewish	1 in 25	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	Caucasian	1 in 25	99%	99%	•	•	•	•
Cystic fibrosis	*CFTR*	Autosomal Recessive	General Population	1 in 25	99%	99%	•	•	•	•
Congenital myasthenic syndrome, *CHRNE*-related	*CHRNE*	Autosomal Recessive	General Population	<1 in 500	99%	99%	•	•	•	•
Congenital myasthenic syndrome, *CHRNE*-related	*CHRNE*	Autosomal Recessive	Caucasian	1 in 383	99%	99%	•	•	•	•
Myotonia congenita	*CLCN1*	Autosomal Recessive	General Population	1 in 158	99%	99%	•	•	•	•
Myotonia congenita	*CLCN1*	Autosomal Recessive	Finnish	1 in 59	99%	99%	•	•	•	•
Myotonia congenita	*CLCN1*	Autosomal Recessive	Norwegian	1 in 53	99%	99%	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	Autosomal Recessive	General Population	<1 in 500	99%	99%	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	Autosomal Recessive	Ashkenazi Jewish	1 in 120	99%	99%	•	•	•	•
Usher syndrome, type 3A	*CLRN1*	Autosomal Recessive	Finnish	1 in 70	99%	99%	•	•	•	•
Achromatopsia, *CNGB3*-related	*CNGB3*	Autosomal Recessive	General Population	1 in 98	99%	99%	•	•	•	•
Achromatopsia, *CNGB3*-related	*CNGB3*	Autosomal Recessive	Caucasian	1 in 91	99%	99%	•	•	•	•
Dystrophic epidermolysis bullosa, *COL7A1*-related	*COL7A1*	Autosomal Recessive	General Population	1 in 370	99%	99%	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Autosomal Recessive	General Population	<1 in 500	99%	99%	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Autosomal Recessive	Asian	<1 in 500	99%	99%	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Autosomal Recessive	African American	1 in 308	99%	99%	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Autosomal Recessive	Caucasian	1 in 200	99%	99%	•	•	•	•
Carnitine palmitoyltransferase II deficiency	*CPT2*	Autosomal Recessive	Ashkenazi Jewish	1 in 51	99%	99%	•	•	•	•
Congenital adrenal insufficiency, *CYP11A1*-related	*CYP11A1*	Autosomal Recessive	General Population	1 in 114	99%	99%	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	Autosomal Recessive	Caucasian	1 in 67	98%	98%	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	Autosomal Recessive	General Population	1 in 61	98%	98%	•	•	•	•
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	*CYP21A2*	Autosomal Recessive	Ashkenazi Jewish	1 in 40	98%	98%	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	Autosomal Recessive	Southern European Caucasian	<1 in 500	99%	99%	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	Autosomal Recessive	General Population	1 in 115	99%	99%	•	•	•	•
Cerebrotendinous xanthomatosis	*CYP27A1*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 5	99%	99%	•	•	•	•
Vitamin D-dependent rickets, type 1A	*CYP27B1*	Autosomal Recessive	General Population	1 in 22	99%	99%	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	Asian	<1 in 500	99%	99%	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	General Population	1 in 100	99%	99%	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	African American	1 in 93	99%	99%	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	Northern European Caucasian	1 in 50	99%	99%	•	•	•	•

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Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	Caucasian	1 in 50	99%	99%	•	•	•	•
Smith-Lemli-Opitz syndrome	*DHCR7*	Autosomal Recessive	Ashkenazi Jewish	1 in 36	99%	99%	•	•	•	•
Retinitis pigmentosa 59	*DHDDS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Retinitis pigmentosa 59	*DHDDS*	Autosomal Recessive	Ashkenazi Jewish	1 in 117	99%	1 in 11,601	•	•	•	•
Dihydrolipoamide dehydrogenase deficiency	*DLD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Dihydrolipoamide dehydrogenase deficiency	*DLD*	Autosomal Recessive	Ashkenazi Jewish	1 in 94	99%	1 in 9,301	•	•	•	•
Duchenne/Becker muscular dystrophy, X-linked	*DMD*	X-Linked	General Population	1 in 4200	99%	1 in 419,901	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	Autosomal Recessive	East Asian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	Autosomal Recessive	Japanese	1 in 297	99%	1 in 29,601	•	•	•	•
Short-rib thoracic dysplasia 3 with or without polydactyly	*DYNC2H1*	Autosomal Recessive	General Population	1 in 67	99%	1 in 6,601	•	•	•	•
Familial dysautonomia	*ELP1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Familial dysautonomia	*ELP1*	Autosomal Recessive	Ashkenazi Jewish	1 in 34	99%	1 in 3,301	•	•	•	•
ERCC2-related conditions	*ERCC2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Ellis-van Creveld syndrome	*EVC2*	Autosomal Recessive	General Population	1 in 122	99%	1 in 12,101	•	•	•	•
Factor VIII deficiency/Hemophilia A	*F8*	X-Linked	General Population	1 in 2000	95%	1 in 39,981	•	•	•	•
Factor IX deficiency/Hemophilia B	*F9*	X-Linked	General Population	1 in 10,000	99%	1 in 999,901	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	African American	1 in 478	99%	1 in 47,701	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	Caucasian	1 in 333	99%	1 in 33,201	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	General Population	1 in 100	99%	1 in 9,901	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	French Canadian–Québec	1 in 66	99%	1 in 6,501	•	•	•	•
Tyrosinemia, type I	*FAH*	Autosomal Recessive	French Canadian–Saguenay-Lac-Saint-Jean	1 in 25	99%	1 in 2,401	•	•	•	•
Fanconi anemia, complementation group C	*FANCC*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Fanconi anemia, complementation group C	*FANCC*	Autosomal Recessive	Ashkenazi Jewish	1 in 98	99%	1 in 9,701	•	•	•	•
Limb-girdle muscular dystrophy, type 2I/ Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Limb-girdle muscular dystrophy, type 2I/ Muscular dystrophy-dystroglycanopathy, type A, 5	*FKRP*	Autosomal Recessive	Norwegian	1 in 116	99%	1 in 11,501	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	Autosomal Recessive	Japanese	1 in 188	99%	1 in 18,701	•	•	•	•
Fukuyama congenital muscular dystrophy	*FKTN*	Autosomal Recessive	Ashkenazi Jewish	1 in 150	99%	1 in 14,901	•	•	•	•
Trimethylaminuria	*FMO3*	Autosomal Recessive	General Population	1 in 139	99%	1 in 13,801	•	•	•	•
Trimethylaminuria	*FMO3*	Autosomal Recessive	Northern European Caucasian	1 in 100	99%	1 in 9,901	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	Hispanic	<1 in 500	99%	<1 in 49,901	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	African American	1 in 251	99%	1 in 25,001	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	Caucasian	1 in 178	99%	1 in 17,701	•	•	•	•
Fragile X syndrome	*FMR1*	Autosomal Recessive	Ashkenazi Jewish	1 in 58	99%	1 in 5,701	•	•	•	•
Friedreich ataxia	*FXN*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901	•	•	•	•
Friedreich ataxia	*FXN*	Autosomal Recessive	Caucasian	1 in 80	99%	1 in 7,901	•	•	•	•

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Glycogen storage disease, type Ia	*G6PC1*	Autosomal Recessive	Asian	1 in 192	99%	1 in 19,101	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Autosomal Recessive	General Population	1 in 177	99%	1 in 17,601	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Autosomal Recessive	Caucasian	1 in 177	99%	1 in 17,601	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Autosomal Recessive	Ashkenazi Jewish	1 in 71	99%	1 in 7,001	•	•	•	•
Glycogen storage disease, type Ia	*G6PC1*	Autosomal Recessive	Ashkenazi Jewish	1 in 71	99%	1 in 7,001	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Autosomal Recessive	General Population	1 in 132	99%	1 in 13,101	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Autosomal Recessive	Asian	1 in 112	99%	1 in 11,101	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Autosomal Recessive	Caucasian	1 in 100	99%	1 in 9,901	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Autosomal Recessive	African American	1 in 70	99%	1 in 6,901	•	•	•	•
Glycogen storage disease, type II/Pompe disease	*GAA*	Autosomal Recessive	Ashkenazi Jewish	1 in 58	99%	1 in 5,701	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	East Asian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	Hispanic	1 in 305	99%	1 in 30,401	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	Ashkenazi Jewish	1 in 172	99%	1 in 17,101	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	General Population	1 in 110	99%	1 in 10,901	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	Northern European Caucasian	1 in 108	99%	1 in 10,701	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	Caucasian	1 in 108	99%	1 in 10,701	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	African American	1 in 78	99%	1 in 7,701	•	•	•	•
Galactosemia	*GALT*	Autosomal Recessive	Irish Travellers	1 in 11	99%	1 in 1,001	•	•	•	•
Gaucher disease	*GBA*	Autosomal Recessive	Caucasian	1 in 164	95%	1 in 3,261	•	•	•	•
Gaucher disease	*GBA*	Autosomal Recessive	General Population	1 in 153	95%	1 in 3,041	•	•	•	•
Gaucher disease	*GBA*	Autosomal Recessive	Ashkenazi Jewish	1 in 18	95%	1 in 341	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	Autosomal Recessive	General Population	1 in 387	99%	1 in 38,601	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	Autosomal Recessive	Caucasian	1 in 144	99%	1 in 14,301	•	•	•	•
Glycogen storage disease, type IV/Adult polyglucosan body disease	*GBE1*	Autosomal Recessive	Ashkenazi Jewish	1 in 68	99%	1 in 6,701	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Autosomal Recessive	General Population	1 in 42	99%	1 in 4,101	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Autosomal Recessive	Caucasian	1 in 30	99%	1 in 2,901	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Autosomal Recessive	Ashkenazi Jewish	1 in 21	99%	1 in 2,001	•	•	•	•
Nonsyndromic hearing loss and deafness 1	*GJB2*	Autosomal Recessive	East Asian	1 in 10	99%	1 in 901	•	•	•	•
Fabry disease, X-linked	*GLA*	X-Linked	General Population	1 in 1,500	99%	1 in 149,901	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	Autosomal Recessive	Asian	1 in 389	99%	1 in 38,801	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	Autosomal Recessive	Caucasian	1 in 225	99%	1 in 22,401	•	•	•	•
Mucolipidosis II and mucolipidosis III alpha/beta	*GNPTAB*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Fraser syndrome, type 3	*GRIP1*	Autosomal Recessive	General Population	1 in 83	99%	1 in 8,201	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	Caucasian	<1 in 500	95%	<1 in 9,981	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	African American	1 in 30	95%	1 in 581	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	General Population	1 in 25	95%	1 in 481	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	Asian	1 in 20	95%	1 in 381	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	East Asian	1 in 16	95%	1 in 301	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	Southeast Asian	1 in 7	95%	1 in 121	•	•	•	•
Alpha-thalassemia	*HBA1/HBA2*	Autosomal Recessive	South Asian	1 in 2	95%	1 in 21	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Caucasian	1 in 373	99%	1 in 37,201	•	•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	General Population	1 in 129	99%	1 in 12,801	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Hispanic	1 in 83	99%	1 in 8,201	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	East Asian	1 in 78	99%	1 in 7,701	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Southern European Caucasian	1 in 59	99%	1 in 5,801	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Asian	1 in 54	99%	1 in 5,301	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	South Asian	1 in 32	99%	1 in 3,101	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Southeast Asian	1 in 30	99%	1 in 2,901	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Mediterranean	1 in 28	99%	1 in 2,701	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	African American	1 in 10	99%	1 in 901	•	•	•	•
Beta hemoglobinopathies	*HBB*	Autosomal Recessive	Middle Eastern	1 in 5	99%	1 in 401	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	African American	1 in 271	99%	1 in 27,001	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	Caucasian	1 in 182	99%	1 in 18,101	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	Asian	1 in 126	99%	1 in 12,501	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	Sephardic Jewish–Moroccan, Iraqi	1 in 125	99%	1 in 12,401	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	French Canadian–Other	1 in 73	99%	1 in 7,201	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	Irish	1 in 41	99%	1 in 4,001	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	Ashkenazi Jewish	1 in 27	99%	1 in 2,601	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	French Canadian	1 in 13	99%	1 in 1,201	•	•	•	•
Tay-Sachs disease	*HEXA*	Autosomal Recessive	French Canadian–Gaspésie	1 in 13	99%	1 in 1,201	•	•	•	•
Hermansky-Pudlak syndrome, type 1	*HPS1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Hermansky-Pudlak syndrome, type 1	*HPS1*	Autosomal Recessive	Puerto Rican	1 in 59	99%	1 in 5,801	•	•	•	•
Hermansky-Pudlak syndrome, type 3	*HPS3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Hermansky-Pudlak syndrome, type 3	*HPS3*	Autosomal Recessive	Ashkenazi Jewish	1 in 235	99%	1 in 23,401	•	•	•	•
Mucopolysaccharidosis, type I/Hurler syndrome	*IDUA*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Mucopolysaccharidosis, type I/Hurler syndrome	*IDUA*	Autosomal Recessive	Northern European Caucasian	1 in 145	99%	1 in 14,401	•	•	•	•
L1 syndrome	*L1CAM*	X-Linked	General Population	1 in 22,500	99%	1 in 2,249,901	•	•	•	•
Donnai-Barrow syndrome	*LRP2*	Autosomal Recessive	General Population	1 in 213	99%	1 in 21,201	•	•	•	•
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	Autosomal Recessive	General Population	1 in 120	99%	1 in 11,901	•	•	•	•
3-methylcrotonyl-CoA carboxylase 2 deficiency	*MCCC2*	Autosomal Recessive	Caucasian	1 in 112	99%	1 in 11,101	•	•	•	•
Mucolipidosis IV	*MCOLN1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Mucolipidosis IV	*MCOLN1*	Autosomal Recessive	Ashkenazi Jewish	1 in 89	99%	1 in 8,801	•	•	•	•
Autosomal recessive primary microcephaly 1	*MCPH1*	Autosomal Recessive	Northern European Caucasian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Autosomal recessive primary microcephaly 1	*MCPH1*	Autosomal Recessive	General Population	1 in 146	99%	1 in 14,501	•	•	•	•
X-linked Opitz G/BBB syndrome	*MID1*	X-Linked	General Population	1 in 37,500	99%	1 in 3,749,901	•	•	•	•
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	Autosomal Recessive	Libyan Jewish	1 in 40	99%	1 in 3,901	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Autosomal Recessive	General Population	1 in 138	99%	1 in 13,701	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Autosomal Recessive	Caucasian	1 in 138	99%	1 in 13,701	•	•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Autosomal Recessive	Asian	1 in 113	99%	1 in 11,201	•	•	•	•
Combined methylmalonic aciduria and homocystinuria, cblC type/Cobalamin C deficiency	*MMACHC*	Autosomal Recessive	East Asian	1 in 112	99%	1 in 11,101	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Autosomal Recessive	General Population	1 in 383	99%	1 in 38,201	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Autosomal Recessive	Hispanic	1 in 383	99%	1 in 38,201	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Autosomal Recessive	Caucasian	1 in 224	99%	1 in 22,301	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Autosomal Recessive	African American	1 in 177	99%	1 in 17,601	•	•	•	•
Methylmalonic aciduria, *MMUT*-related	*MMUT*	Autosomal Recessive	Asian	1 in 53	99%	1 in 5,201	•	•	•	•
Mevalonic aciduria/Hyper-IgD syndrome	*MVK*	Autosomal Recessive	General Population	1 in 167	99%	1 in 16,601	•	•	•	•
Schindler disease	*NAGA*	Autosomal Recessive	General Population	1 in 94	99%	1 in 9,301	•	•	•	•
Nemaline myopathy 2	*NEB*	Autosomal Recessive	General Population	1 in 224	95%	1 in 4,461	•	•	•	•
Nemaline myopathy 2	*NEB*	Autosomal Recessive	Ashkenazi Jewish	1 in 168	95%	1 in 3,341	•	•	•	•
Nemaline myopathy 2	*NEB*	Autosomal Recessive	Finnish	1 in 112	95%	1 in 2,221	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	Autosomal Recessive	General Population	1 in 325	99%	1 in 32,401	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	Autosomal Recessive	Finnish	1 in 45	99%	1 in 4,401	•	•	•	•
Steroid resistant nephrotic syndrome, type 1	*NPHS1*	Autosomal Recessive	Groffdale Conference Mennonite	1 in 12	99%	1 in 1,101	•	•	•	•
X-linked congenital adrenal hypoplasia	*NR0B1*	X-Linked	General Population	1 in 52,500	99%	1 in 5,249,901	•	•	•	•
Oculocutaneous albinism, type II	*OCA2*	Autosomal Recessive	General Population	1 in 76	99%	1 in 7,501	•	•	•	•
Oculocutaneous albinism, type II	*OCA2*	Autosomal Recessive	African American	1 in 50	99%	1 in 4,901	•	•	•	•
Oculocutaneous albinism, type II	*OCA2*	Autosomal Recessive	Navajo	1 in 22	99%	1 in 2,101	•	•	•	•
Ornithine transcarbamylase deficiency, X-linked	*OTC*	X-Linked	General Population	1 in 30,000	99%	1 in 2,999,901	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Finnish	1 in 225	99%	1 in 22,401	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Ashkenazi Jewish	1 in 225	99%	1 in 22,401	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Hispanic American	1 in 163	99%	1 in 16,201	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	African American	1 in 143	99%	1 in 14,201	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Asian	1 in 78	99%	1 in 7,701	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	General Population	1 in 65	99%	1 in 6,401	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Caucasian	1 in 50	99%	1 in 4,901	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Southern European Caucasian	1 in 40	99%	1 in 3,901	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Irish	1 in 34	99%	1 in 3,301	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Turkish	1 in 32	99%	1 in 3,101	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Sicilian	1 in 26	99%	1 in 2,501	•	•	•	•
Phenylalanine hydroxylase deficiency	*PAH*	Autosomal Recessive	Sephardic Jewish–Iranian, Bukharan, Kavkazi, Tunisian, Moroccan	1 in 18	99%	1 in 1,701	•	•	•	•
Usher syndrome, type 1F	*PCDH15*	Autosomal Recessive	General Population	1 in 395	99%	1 in 39,401	•	•	•	•
Usher syndrome, type 1F	*PCDH15*	Autosomal Recessive	Ashkenazi Jewish	1 in 78	99%	1 in 7,701	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Autosomal Recessive	General Population	1 in 144	99%	1 in 14,301	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Autosomal Recessive	Ashkenazi Jewish	1 in 106	99%	1 in 10,501	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Autosomal Recessive	Caucasian	1 in 100	99%	1 in 9,901	•	•	•	•
Autosomal recessive polycystic kidney disease	*PKHD1*	Autosomal Recessive	South African Afrikaner	1 in 52	99%	1 in 5,101	•	•	•	•
PLP1-related disorders	*PLP1*	X-Linked	Northern European Caucasian	1 in 57,700	99%	1 in 5,769,901	•	•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
PLP1-related disorders	*PLP1*	X-Linked	General Population	1 in 37,500	99%	1 in 3,749,901	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Autosomal Recessive	Asian	1 in 449	99%	1 in 44,801	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Autosomal Recessive	General Population	1 in 124	99%	1 in 12,301	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Autosomal Recessive	Ashkenazi Jewish	1 in 61	99%	1 in 6,001	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Autosomal Recessive	Northern European Caucasian	1 in 60	99%	1 in 5,901	•	•	•	•
Congenital disorder of glycosylation, type Ia	*PMM2*	Autosomal Recessive	Caucasian	1 in 42	99%	1 in 4,101	•	•	•	•
POLG-related disorders	*POLG*	Autosomal Recessive	General Population	1 in 50	99%	1 in 4,901	•	•	•	•
Familial hemophagocytic lymphohistiocytosis 2	*PRF1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Pontocerebellar hypoplasia, type 6	*RARS2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Pontocerebellar hypoplasia, type 6	*RARS2*	Autosomal Recessive	Sephardic Jewish–Iraqi, Syrian, Tunisian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Aicardi-Goutières syndrome 2	*RNASEH2B*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Retinitis pigmentosa 3	*RPGR*	X-Linked	General Population	1 in 20,000	99%	1 in 1,999,901	•	•	•	•
Juvenile retinoschisis, X-linked	*RS1*	X-Linked	General Population	1 in 2,500	99%	1 in 249,901	•	•	•	•
Mitochondrial complex IV deficiency, nuclear type 2	*SCO2*	Autosomal Recessive	General Population	1 in 149	99%	1 in 14,801	•	•	•	•
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*	Autosomal Recessive	Middle Eastern	<1 in 500	99%	<1 in 49,901	•	•	•	•
Biotin-thiamine-responsive basal ganglia disease	*SLC19A3*	Autosomal Recessive	General Population	1 in 109	99%	1 in 10,801	•	•	•	•
Skeletal dysplasia, *SLC26A2*-related	*SLC26A2*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701	•	•	•	•
Skeletal dysplasia, *SLC26A2*-related	*SLC26A2*	Autosomal Recessive	Finnish	1 in 50	99%	1 in 4,901	•	•	•	•
Pendred syndrome	*SLC26A4*	Autosomal Recessive	Caucasian	1 in 88	99%	1 in 8,701	•	•	•	•
Pendred syndrome	*SLC26A4*	Autosomal Recessive	General Population	1 in 80	99%	1 in 7,901	•	•	•	•
Pendred syndrome	*SLC26A4*	Autosomal Recessive	African American	1 in 76	99%	1 in 7,501	•	•	•	•
Pendred syndrome	*SLC26A4*	Autosomal Recessive	Asian	1 in 74	99%	1 in 7,301	•	•	•	•
Pendred syndrome	*SLC26A4*	Autosomal Recessive	Northern European Caucasian	1 in 60	99%	1 in 5,901	•	•	•	•
Glycogen storage disease, type Ib/IIw	*SLC37A4*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901	•	•	•	•
Glycogen storage disease, type Ib/IIw	*SLC37A4*	Autosomal Recessive	General Population	1 in 354	99%	1 in 35,301	•	•	•	•
Creatine transporter defect, *SLC6A8*-related, X-linked/Cerebral creatine deficiency syndrome	*SLC6A8*	X-Linked	General Population	1 in 20,600	99%	1 in 2,059,901	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Hispanic	1 in 117	93%	1 in 11,601	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	African American	1 in 72	93%	1 in 7,101	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Hispanic American	1 in 68	93%	1 in 6,701	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Ashkenazi Jewish	1 in 67	93%	1 in 6,601	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	East Asian	1 in 59	93%	1 in 5,801	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	South Asian	1 in 59	93%	1 in 5,801	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Southeast Asian	1 in 59	93%	1 in 5,801	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Asian	1 in 59	93%	1 in 5,801	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	General Population	1 in 54	93%	1 in 5,301	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Northern European Caucasian	1 in 47	93%	1 in 4,601	•	•	•	•
Spinal muscular atrophy	*SMN1*	Autosomal Recessive	Caucasian	1 in 47	93%	1 in 4,601	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	Autosomal Recessive	Caucasian	1 in 244	99%	1 in 24,301	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	Autosomal Recessive	General Population	1 in 196	99%	1 in 19,501	•	•	•	•
Niemann-Pick disease, types A/B	*SMPD1*	Autosomal Recessive	Ashkenazi Jewish	1 in 115	99%	1 in 11,401	•	•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Atransferrinemia	*TF*	Autosomal Recessive	General Population	1 in 116	99%	1 in 11,501	•	•	•	•
Joubert syndrome 2	*TMEM216*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Joubert syndrome 2	*TMEM216*	Autosomal Recessive	Ashkenazi Jewish	1 in 110	99%	1 in 10,901	•	•	•	•
TNXB-related classical-like Ehlers-Danlos syndrome	*TNXB*	Autosomal Recessive	General Population	1 in 28	95%	1 in 541	•	•	•	•
Oculocutaneous albinism, type I	*TYR*	Autosomal Recessive	General Population	1 in 20	99%	1 in 1,901	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	French Canadian	1 in 207	99%	1 in 20,601	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	General Population	1 in 126	99%	1 in 12,501	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	East Asian	1 in 113	99%	1 in 11,201	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	Northern European Caucasian	1 in 113	99%	1 in 11,201	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	Caucasian	1 in 73	99%	1 in 7,201	•	•	•	•
Usher syndrome, type 2A	*USH2A*	Autosomal Recessive	Sephardic Jewish–Iraqi, Iranian	1 in 36	99%	1 in 3,501	•	•	•	•
Xeroderma pigmentosum, group C	*XPC*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901	•	•	•	•
Adenosine deaminase deficiency	*ADA*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301		•	•	•
Glycogen storage disease, type III	*AGL*	Autosomal Recessive	General Population	1 in 159	99%	1 in 15,801		•	•	•
Glycogen storage disease, type III	*AGL*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 37	99%	1 in 3,601		•	•	•
Glycogen storage disease, type III	*AGL*	Autosomal Recessive	Faroese	1 in 28	99%	1 in 2,701		•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	Autosomal Recessive	Northern European Caucasian	1 in 223	99%	1 in 22,201		•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	Autosomal Recessive	General Population	1 in 223	99%	1 in 22,201		•	•	•
Sjögren-Larsson syndrome	*ALDH3A2*	Autosomal Recessive	Swedish	1 in 204	99%	1 in 20,301		•	•	•
Glycine encephalopathy, *AMT*-related	*AMT*	Autosomal Recessive	Caucasian	1 in 271	99%	1 in 27,001		•	•	•
Glycine encephalopathy, *AMT*-related	*AMT*	Autosomal Recessive	General Population	1 in 262	99%	1 in 26,101		•	•	•
Mucopolysaccharidosis, type VI/ Maroteaux-Lamy syndrome	*ARSB*	Autosomal Recessive	Asian	1 in 423	99%	1 in 42,201		•	•	•
Mucopolysaccharidosis, type VI/ Maroteaux-Lamy syndrome	*ARSB*	Autosomal Recessive	General Population	1 in 291	99%	1 in 29,001		•	•	•
Mucopolysaccharidosis, type VI/ Maroteaux-Lamy syndrome	*ARSB*	Autosomal Recessive	Caucasian	1 in 273	99%	1 in 27,201		•	•	•
Citrullinemia, type I	*ASS1*	Autosomal Recessive	Caucasian	1 in 195	99%	1 in 19,401		•	•	•
Citrullinemia, type I	*ASS1*	Autosomal Recessive	Asian	1 in 123	99%	1 in 12,201		•	•	•
Citrullinemia, type I	*ASS1*	Autosomal Recessive	General Population	1 in 119	99%	1 in 11,801		•	•	•
Ataxia-telangiectasia	*ATM*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901		•	•	•
Ataxia-telangiectasia	*ATM*	Autosomal Recessive	General Population	1 in 100	99%	1 in 9,901		•	•	•
Ataxia-telangiectasia	*ATM*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 81	99%	1 in 8,001		•	•	•
Menkes disease	*ATP7A*	X-Linked	General Population	1 in 26,000	99%	1 in 2,599,901		•	•	•
Bardet-Biedl syndrome 10	*BBS10*	Autosomal Recessive	General Population	1 in 447	99%	1 in 44,601		•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Autosomal Recessive	General Population	1 in 321	99%	1 in 32,001		•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Autosomal Recessive	Caucasian	1 in 320	99%	1 in 31,901		•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Autosomal Recessive	Portuguese Roma	1 in 71	99%	1 in 7,001		•	•	•
Maple syrup urine disease, type 1A	*BCKDHA*	Autosomal Recessive	Mennonite	1 in 10	99%	1 in 901		•	•	•
GRACILE syndrome	*BCS1L*	Autosomal Recessive	Caucasian	1 in 407	99%	1 in 40,601		•	•	•
GRACILE syndrome	*BCS1L*	Autosomal Recessive	General Population	1 in 111	99%	1 in 11,001		•	•	•
GRACILE syndrome	*BCS1L*	Autosomal Recessive	Finnish	1 in 108	99%	1 in 10,701		•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	Hispanic	1 in 260	99%	1 in 25,901		•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	East Asian	1 in 232	99%	1 in 23,101		•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	Northern European Caucasian	1 in 103	99%	1 in 10,201		•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	Caucasian	1 in 103	99%	1 in 10,201		•	•	•
Limb-girdle muscular dystrophy, type 2A	*CAPN3*	Autosomal Recessive	Amish	1 in 50	99%	1 in 4,901		•	•	•
Usher syndrome, type 1D	*CDH23*	Autosomal Recessive	General Population	1 in 202	99%	1 in 20,101		•	•	•
Neuronal ceroid lipofuscinosis, *CLN3*-related	*CLN3*	Autosomal Recessive	Caucasian	1 in 188	99%	1 in 18,701		•	•	•
Neuronal ceroid lipofuscinosis, *CLN3*-related	*CLN3*	Autosomal Recessive	General Population	1 in 145	99%	1 in 14,401		•	•	•
Neuronal ceroid lipofuscinosis, *CLN5*-related	*CLN5*	Autosomal Recessive	General Population	1 in 317	99%	1 in 31,601		•	•	•
Neuronal ceroid lipofuscinosis, *CLN5*-related	*CLN5*	Autosomal Recessive	Finnish	1 in 289	99%	1 in 28,801		•	•	•
Neuronal ceroid lipofuscinosis, *CLN6*-related	*CLN6*	Autosomal Recessive	General Population	1 in 261	99%	1 in 26,001		•	•	•
Neuronal ceroid lipofuscinosis, *CLN8*-related	*CLN8*	Autosomal Recessive	General Population	1 in 349	99%	1 in 34,801		•	•	•
Neuronal ceroid lipofuscinosis, *CLN8*-related	*CLN8*	Autosomal Recessive	Finnish	1 in 135	99%	1 in 13,401		•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	Autosomal Recessive	General Population	1 in 323	99%	1 in 32,201		•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	Autosomal Recessive	Caucasian	1 in 284	99%	1 in 28,301		•	•	•
Alport syndrome, *COL4A3*-related	*COL4A3*	Autosomal Recessive	Ashkenazi Jewish	1 in 189	99%	1 in 18,801		•	•	•
Alport syndrome, *COL4A4*-related	*COL4A4*	Autosomal Recessive	General Population	1 in 353	99%	1 in 35,201		•	•	•
Alport syndrome, *COL4A5*-related, X-linked	*COL4A5*	X-Linked	General Population	1 in 47,000	99%	1 in 4,699,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	African American	<1 in 500	99%	<1 in 49,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	Hispanic	<1 in 500	99%	<1 in 49,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	Caucasian	1 in 220	99%	1 in 21,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 100	99%	1 in 9,901		•	•	•
Cystinosis	*CTNS*	Autosomal Recessive	French Canadian–Saguenay-Lac-Saint-Jean	1 in 39	99%	1 in 3,801		•	•	•
Maple syrup urine disease, type 2	*DBT*	Autosomal Recessive	General Population	1 in 321	99%	1 in 32,001		•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Autosomal Recessive	Asian	1 in 163	99%	1 in 16,201		•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Autosomal Recessive	Caucasian	1 in 101	99%	1 in 10,001		•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Autosomal Recessive	General Population	1 in 92	99%	1 in 9,101		•	•	•
Factor XI deficiency/Hemophilia C	*F11*	Autosomal Recessive	Ashkenazi Jewish	1 in 11	99%	1 in 1,001		•	•	•
Fanconi anemia, complementation group A	*FANCA*	Autosomal Recessive	General Population	1 in 345	99%	1 in 34,401		•	•	•
Fanconi anemia, complementation group A	*FANCA*	Autosomal Recessive	Sephardic Jewish–Moroccan, Tunisian	1 in 133	99%	1 in 13,201		•	•	•
Fanconi anemia, complementation group A	*FANCA*	Autosomal Recessive	Spanish Roma	1 in 64	99%	1 in 6,301		•	•	•
Krabbe disease	*GALC*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901		•	•	•
Krabbe disease	*GALC*	Autosomal Recessive	General Population	1 in 150	99%	1 in 14,901		•	•	•
Krabbe disease	*GALC*	Autosomal Recessive	Druze–Northern Israel	1 in 6	99%	1 in 501		•	•	•
Krabbe disease	*GALC*	Autosomal Recessive	Muslim Arab–Jerusalem	1 in 6	99%	1 in 501		•	•	•
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	Caucasian	1 in 172	99%	1 in 17,101		•	•	•
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	General Population	1 in 112	99%	1 in 11,101		•	•	•
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	African American	1 in 36	99%	1 in 3,501		•	•	•
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	Lumbee Native American	1 in 16	99%	1 in 1,501		•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	Lancaster County Amish	1 in 9	99%	1 in 801		•	•	•
Glutaric acidemia, type I	*GCDH*	Autosomal Recessive	Oji-Cree FirstNation–North Manitoba	1 in 8	99%	1 in 701		•	•	•
GLB1-related disorders	*GLB1*	Autosomal Recessive	Caucasian	1 in 278	99%	1 in 27,701		•	•	•
GLB1-related disorders	*GLB1*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701		•	•	•
GLB1-related disorders	*GLB1*	Autosomal Recessive	South Brazil	1 in 65	99%	1 in 6,401		•	•	•
GLB1-related disorders	*GLB1*	Autosomal Recessive	Roma	1 in 50	99%	1 in 4,901		•	•	•
GLB1-related disorders	*GLB1*	Autosomal Recessive	Maltese	1 in 30	99%	1 in 2,901		•	•	•
Glycine encephalopathy/Nonketotic hyperglycinemia	*GLDC*	Autosomal Recessive	Caucasian	1 in 140	99%	1 in 13,901		•	•	•
Glycine encephalopathy/Nonketotic hyperglycinemia	*GLDC*	Autosomal Recessive	General Population	1 in 135	99%	1 in 13,401		•	•	•
Lethal congenital contracture syndrome 1	*GLE1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Lethal congenital contracture syndrome 1	*GLE1*	Autosomal Recessive	Finnish	1 in 100	99%	1 in 9,901		•	•	•
GNE myopathy	*GNE*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901		•	•	•
GNE myopathy	*GNE*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901		•	•	•
GNE myopathy	*GNE*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
GNE myopathy	*GNE*	Autosomal Recessive	Asian	1 in 58	99%	1 in 5,701		•	•	•
GNE myopathy	*GNE*	Autosomal Recessive	Sephardic Jewish–Iranian, Syrian	1 in 12	99%	1 in 1,101		•	•	•
Mucopolysaccharidosis, type IIID/ Sanfilippo syndrome D	*GNS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	Autosomal Recessive	Caucasian	1 in 254	99%	1 in 25,301		•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	Autosomal Recessive	Finnish	1 in 240	99%	1 in 23,901		•	•	•
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	*HADHA*	Autosomal Recessive	General Population	1 in 138	99%	1 in 13,701		•	•	•
Sandhoff disease	*HEXB*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901		•	•	•
Sandhoff disease	*HEXB*	Autosomal Recessive	General Population	1 in 278	99%	1 in 27,701		•	•	•
Sandhoff disease	*HEXB*	Autosomal Recessive	Caucasian	1 in 235	99%	1 in 23,401		•	•	•
Sandhoff disease	*HEXB*	Autosomal Recessive	Argentinian Creole	1 in 64	99%	1 in 6,301		•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901		•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	Autosomal Recessive	General Population	1 in 482	99%	1 in 48,101		•	•	•
Mucopolysaccharidosis, type IIIC/ Sanfilippo syndrome C	*HGSNAT*	Autosomal Recessive	Caucasian	1 in 259	99%	1 in 25,801		•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901		•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Autosomal Recessive	Asian	1 in 158	99%	1 in 15,701		•	•	•
Holocarboxylase synthetase deficiency	*HLCS*	Autosomal Recessive	Faroese	1 in 20	99%	1 in 1,901		•	•	•
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	*HMGCL*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
D-bifunctional protein deficiency	*HSD17B4*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701		•	•	•
Hydrolethalus syndrome	*HYLS1*	Autosomal Recessive	General Population	1 in 455	99%	1 in 45,401		•	•	•
Hydrolethalus syndrome	*HYLS1*	Autosomal Recessive	Finnish	1 in 50	99%	1 in 4,901		•	•	•
Mucopolysaccharidosis, type II/Hunter syndrome	*IDS*	X-Linked	General Population	1 in 60,000	90%	1 in 599,991		•	•	•
Isovaleric acidemia	*IVD*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901		•	•	•
Isovaleric acidemia	*IVD*	Autosomal Recessive	Caucasian	1 in 144	99%	1 in 14,301		•	•	•
Isovaleric acidemia	*IVD*	Autosomal Recessive	Asian	1 in 75	99%	1 in 7,401		•	•	•
Familial hyperinsulinism, *KCNJ11*-related	*KCNJ11*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
LAMA2 muscular dystrophy	*LAMA2*	Autosomal Recessive	General Population	1 in 87	99%	1 in 8,601		•	•	•
Junctional epidermolysis bullosa, *LAMA3*-related	*LAMA3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Junctional epidermolysis bullosa, *LAMB3*-related	*LAMB3*	Autosomal Recessive	General Population	1 in 407	99%	1 in 40,601		•	•	•
Junctional epidermolysis bullosa, *LAMC2*-related	*LAMC2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Nonsyndromic hearing loss and deafness 77	*LOXHD1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Nonsyndromic hearing loss and deafness 77	*LOXHD1*	Autosomal Recessive	Ashkenazi Jewish	1 in 180	99%	1 in 17,901		•	•	•
Mitochondrial complex IV deficiency, nuclear type 5/Leigh syndrome, French-Canadian type	*LRPPRC*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Mitochondrial complex IV deficiency, nuclear type 5/Leigh syndrome, French-Canadian type	*LRPPRC*	Autosomal Recessive	French Canadian–Saguenay-Lac-Saint-Jean	1 in 23	99%	1 in 2,201		•	•	•
Alpha-mannosidosis	*MAN2B1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Alpha-mannosidosis	*MAN2B1*	Autosomal Recessive	Caucasian	1 in 485	99%	1 in 48,401		•	•	•
Alpha-mannosidosis	*MAN2B1*	Autosomal Recessive	Northern European Caucasian	1 in 354	99%	1 in 35,301		•	•	•
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	Autosomal Recessive	General Population	1 in 147	99%	1 in 14,601		•	•	•
3-methylcrotonyl-CoA carboxylase 1 deficiency	*MCCC1*	Autosomal Recessive	Caucasian	1 in 137	99%	1 in 13,601		•	•	•
Infantile cerebral and cerebellar atrophy	*MED17*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Infantile cerebral and cerebellar atrophy	*MED17*	Autosomal Recessive	Sephardic Jewish–Bukharan, Kurdish	1 in 20	99%	1 in 1,901		•	•	•
Familial Mediterranean fever	*MEFV*	Autosomal Recessive	General Population	1 in 115	99%	1 in 11,401		•	•	•
Familial Mediterranean fever	*MEFV*	Autosomal Recessive	Sephardic Jewish	1 in 14	99%	1 in 1,301		•	•	•
Familial Mediterranean fever	*MEFV*	Autosomal Recessive	Ashkenazi Jewish	1 in 13	99%	1 in 1,201		•	•	•
Familial Mediterranean fever	*MEFV*	Autosomal Recessive	Armenian	1 in 5	99%	1 in 401		•	•	•
Familial Mediterranean fever	*MEFV*	Autosomal Recessive	Turkish	1 in 5	99%	1 in 401		•	•	•
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301		•	•	•
Spondylothoracic dysostosis and spondylocostal dysostosis 2	*MESP2*	Autosomal Recessive	Puerto Rican	1 in 55	99%	1 in 5,401		•	•	•
Methylmalonic aciduria, *MMAA*-related	*MMAA*	Autosomal Recessive	General Population	1 in 316	99%	1 in 31,501		•	•	•
Methylmalonic aciduria, *MMAA*-related	*MMAA*	Autosomal Recessive	Caucasian	1 in 316	99%	1 in 31,501		•	•	•
Methylmalonic aciduria, *MMAB*-related	*MMAB*	Autosomal Recessive	Caucasian	1 in 456	99%	1 in 45,501		•	•	•
Methylmalonic aciduria, *MMAB*-related	*MMAB*	Autosomal Recessive	General Population	1 in 456	99%	1 in 45,501		•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	Autosomal Recessive	General Population	1 in 415	99%	1 in 41,401		•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	Autosomal Recessive	Caucasian	1 in 266	99%	1 in 26,501		•	•	•
Congenital amegakaryocytic thrombocytopenia	*MPL*	Autosomal Recessive	Ashkenazi Jewish	1 in 57	99%	1 in 5,601		•	•	•
Abetalipoproteinemia	*MTTP*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901		•	•	•
Abetalipoproteinemia	*MTTP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Abetalipoproteinemia	*MTTP*	Autosomal Recessive	Ashkenazi Jewish	1 in 186	99%	1 in 18,501		•	•	•
Usher syndrome, type 1B	*MYO7A*	Autosomal Recessive	African American	<1 in 500	99%	<1 in 49,901		•	•	•
Usher syndrome, type 1B	*MYO7A*	Autosomal Recessive	General Population	1 in 206	99%	1 in 20,501		•	•	•
Usher syndrome, type 1B	*MYO7A*	Autosomal Recessive	Caucasian	1 in 145	99%	1 in 14,401		•	•	•
Usher syndrome, type 1B	*MYO7A*	Autosomal Recessive	Japanese	1 in 123	99%	1 in 12,201		•	•	•
Usher syndrome, type 1B	*MYO7A*	Autosomal Recessive	Asian	1 in 62	99%	1 in 6,101		•	•	•
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	Autosomal Recessive	Caucasian	1 in 346	99%	1 in 34,501		•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Mucopolysaccharidosis, type IIIB/ Sanfilippo syndrome B	*NAGLU*	Autosomal Recessive	Asian	1 in 298	99%	1 in 29,701		•	•	•
Nijmegen breakage syndrome	*NBN*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Nijmegen breakage syndrome	*NBN*	Autosomal Recessive	Caucasian	1 in 155	99%	1 in 15,401		•	•	•
Niemann-Pick disease, type C1	*NPC1*	Autosomal Recessive	Asian	1 in 404	99%	1 in 40,301		•	•	•
Niemann-Pick disease, type C1	*NPC1*	Autosomal Recessive	General Population	1 in 282	99%	1 in 28,101		•	•	•
Niemann-Pick disease, type C1	*NPC1*	Autosomal Recessive	Caucasian	1 in 185	99%	1 in 18,401		•	•	•
Steroid-resistant nephrotic syndrome, type 2	*NPHS2*	Autosomal Recessive	General Population	1 in 377	99%	1 in 37,601		•	•	•
Pyruvate carboxylase deficiency	*PC*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901		•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	Autosomal Recessive	Caucasian	1 in 380	99%	1 in 37,901		•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301		•	•	•
Propionic acidemia, *PCCA*-related	*PCCA*	Autosomal Recessive	Asian	1 in 162	99%	1 in 16,101		•	•	•
Propionic acidemia, *PCCB*-related	*PCCB*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301		•	•	•
Propionic acidemia, *PCCB*-related	*PCCB*	Autosomal Recessive	Caucasian	1 in 202	99%	1 in 20,101		•	•	•
Propionic acidemia, *PCCB*-related	*PCCB*	Autosomal Recessive	Asian	1 in 145	99%	1 in 14,401		•	•	•
Zellweger spectrum disorder, *PEX1*-related	*PEX1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Zellweger spectrum disorder, *PEX1*-related	*PEX1*	Autosomal Recessive	Caucasian	1 in 147	99%	1 in 14,601		•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901		•	•	•
Zellweger spectrum disorder, *PEX2*-related	*PEX2*	Autosomal Recessive	Ashkenazi Jewish	1 in 227	99%	1 in 22,601		•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	Autosomal Recessive	General Population	1 in 280	99%	1 in 27,901		•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	Autosomal Recessive	French Canadian	1 in 55	99%	1 in 5,401		•	•	•
Zellweger spectrum disorder, *PEX6*-related	*PEX6*	Autosomal Recessive	Sephardic Jewish–Yemenite	1 in 18	99%	1 in 1,701		•	•	•
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Rhizomelic chondrodysplasia punctata, type 1	*PEX7*	Autosomal Recessive	Caucasian	1 in 158	99%	1 in 15,701		•	•	•
Neuronal ceroid lipofuscinosis, *PPT1*-related	*PPT1*	Autosomal Recessive	General Population	1 in 368	99%	1 in 36,701		•	•	•
Neuronal ceroid lipofuscinosis, *PPT1*-related	*PPT1*	Autosomal Recessive	Finnish	1 in 70	99%	1 in 6,901		•	•	•
Combined pituitary hormone deficiency, type 2	*PROP1*	Autosomal Recessive	General Population	1 in 141	99%	1 in 14,001		•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	Autosomal Recessive	East Asian	1 in 158	99%	1 in 15,701		•	•	•
6-pyruvoyl-tetrahydropterin synthase deficiency	*PTS*	Autosomal Recessive	Asian	1 in 122	99%	1 in 12,101		•	•	•
Cartilage-hair hypoplasia	*RMRP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Cartilage-hair hypoplasia	*RMRP*	Autosomal Recessive	Finnish	1 in 76	99%	1 in 7,501		•	•	•
Cartilage-hair hypoplasia	*RMRP*	Autosomal Recessive	Amish	1 in 19	99%	1 in 1,801		•	•	•
Dyskeratosis congenita, *RTEL1*-related	*RTEL1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Dyskeratosis congenita, *RTEL1*-related	*RTEL1*	Autosomal Recessive	Ashkenazi Jewish	1 in 165	99%	1 in 16,401		•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	Autosomal Recessive	Caucasian	1 in 450	99%	1 in 44,901		•	•	•
Spastic ataxia, Charlevoix-Saguenay type	*SACS*	Autosomal Recessive	French Canadian–Charlevoix-Saguenay	1 in 21	99%	1 in 2,001		•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Autosomal Recessive	Caucasian	1 in 290	99%	1 in 28,901		•	•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Limb-girdle muscular dystrophy, type 3	*SGCA*	Autosomal Recessive	Northern European Caucasian	1 in 160	99%	1 in 15,901		•	•	•
Limb-girdle muscular dystrophy, type 3	*SGCA*	Autosomal Recessive	Finnish	1 in 150	99%	1 in 14,901		•	•	•
Limb-girdle muscular dystrophy, type 4	*SGCB*	Autosomal Recessive	Northern European Caucasian	<1 in 500	99%	<1 in 49,901		•	•	•
Limb-girdle muscular dystrophy, type 4	*SGCB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Limb-girdle muscular dystrophy, type 4	*SGCB*	Autosomal Recessive	Caucasian	1 in 406	99%	1 in 40,501		•	•	•
Mucopolysaccharidosis, type IIIA/ Sanfilippo syndrome A	*SGSH*	Autosomal Recessive	General Population	1 in 415	99%	1 in 41,401		•	•	•
Mucopolysaccharidosis, type IIIA/ Sanfilippo syndrome A	*SGSH*	Autosomal Recessive	Caucasian	1 in 253	99%	1 in 25,201		•	•	•
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Agenesis of the corpus callosum with peripheral neuropathy	*SLC12A6*	Autosomal Recessive	French Canadian	1 in 23	99%	1 in 2,201		•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Autosomal Recessive	General Population	1 in 200	99%	1 in 19,901		•	å•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Autosomal Recessive	Caucasian	1 in 110	99%	1 in 10,901		•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Autosomal Recessive	East Asian	1 in 100	99%	1 in 9,901		•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Autosomal Recessive	Asian	1 in 100	99%	1 in 9,901		•	•	•
Carnitine deficiency, systemic primary	*SLC22A5*	Autosomal Recessive	Faroese	1 in 20	99%	1 in 1,901		•	•	•
Arthrogryposis, intellectual disability, and seizures	*SLC35A3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Arthrogryposis, intellectual disability, and seizures	*SLC35A3*	Autosomal Recessive	Ashkenazi Jewish	1 in 453	99%	1 in 45,201		•	•	•
Lysinuric protein intolerance	*SLC7A7*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Lysinuric protein intolerance	*SLC7A7*	Autosomal Recessive	Finnish	1 in 122	99%	1 in 12,101		•	•	•
Lysinuric protein intolerance	*SLC7A7*	Autosomal Recessive	Japanese	1 in 119	99%	1 in 11,801		•	•	•
Tyrosinemia, type II	*TAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	Autosomal Recessive	General Population	1 in 301	99%	1 in 30,001		•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	Autosomal Recessive	Caucasian	1 in 253	99%	1 in 25,201		•	•	•
Autosomal recessive congenital ichthyosis	*TGM1*	Autosomal Recessive	Norwegian	1 in 151	99%	1 in 15,001		•	•	•
Tyrosine hydroxylase deficiency	*TH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901		•	•	•
Tyrosine hydroxylase deficiency	*TH*	Autosomal Recessive	Asian	1 in 416	99%	1 in 41,501		•	•	•
Tyrosine hydroxylase deficiency	*TH*	Autosomal Recessive	Caucasian	1 in 224	99%	1 in 22,301		•	•	•
Neuronal ceroid lipofuscinosis, *TPP1*-related	*TPP1*	Autosomal Recessive	General Population	1 in 314	99%	1 in 31,301		•	•	•
Neuronal ceroid lipofuscinosis, *TPP1*-related	*TPP1*	Autosomal Recessive	Newfoundland	1 in 59	99%	1 in 5,801		•	•	•
Usher syndrome, type 1C	*USH1C*	Autosomal Recessive	General Population	1 in 353	99%	1 in 35,201		•	•	•
Usher syndrome, type 1C	*USH1C*	Autosomal Recessive	French Canadian, Acadian	1 in 227	99%	1 in 22,601		•	•	•
Usher syndrome, type 1C	*USH1C*	Autosomal Recessive	Acadian	1 in 41	99%	1 in 4,001		•	•	•
Triple A syndrome	*AAAS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
GABA-transaminase deficiency	*ABAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Congenital ichthyosis, *ABCA12*-related	*ABCA12*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Progressive familial intrahepatic cholestasis 2	*ABCB11*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701			•	•
Pseudoxanthoma elasticum	*ABCC6*	Autosomal Recessive	General Population	1 in 79	90%	1 in 781			•	•
Mitochondrial complex I deficiency, *ACAD9*-related	*ACAD9*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Peroxisomal acyl-CoA oxidase deficiency	*ACOX1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Combined malonic and methylmalonic aciduria	*ACSF3*	Autosomal Recessive	General Population	1 in 86	99%	1 in 8,501			•	•
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Ehlers-Danlos syndrome, dermatosparaxis type	*ADAMTS2*	Autosomal Recessive	Ashkenazi Jewish	1 in 248	99%	1 in 24,701			•	•
Bilateral frontoparietal polymicrogyria	*ADGRG1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Usher syndrome, type 2C	*ADGRV1*	Autosomal Recessive	General Population	1 in 176	99%	1 in 17,501			•	•
Rhizomelic chondrodysplasia punctata, type 3	*AGPS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Combined oxidative phosphorylation deficiency 6	*AIFM1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pyridoxine-dependent epilepsy	*ALDH7A1*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701			•	•
Congenital disorder of glycosylation, type Ic	*ALG6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Alström syndrome	*ALMS1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
MEDNIK syndrome	*AP1S1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Hermansky-Pudlak syndrome, type 2	*AP3B1*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701			•	•
Nephrogenic diabetes insipidus	*AQP2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Argininemia	*ARG1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Joubert syndrome 8	*ARL13B*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
X-linked chondrodysplasia punctata 1	*ARSL*	X-Linked	General Population	1 in 375,000	99%	1 in 37,499,901			•	•
Farber lipogranulomatosis	*ASAH1*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901			•	•
Asparagine synthetase deficiency	*ASNS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Asparagine synthetase deficiency	*ASNS*	Autosomal Recessive	Sephardic Jewish–Iranian	1 in 80	99%	1 in 7,901			•	•
Renal tubular acidosis and deafness, *ATP6V1B1*-related	*ATP6V1B1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Renal tubular acidosis and deafness, *ATP6V1B1*-related	*ATP6V1B1*	Autosomal Recessive	Sephardic Jewish–Syrian	1 in 140	99%	1 in 13,901			•	•
Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	*ATP8B1*	Autosomal Recessive	General Population	1 in 53	99%	1 in 5,201			•	•
Alpha-thalassemia intellectual disability syndrome, X-linked	*ATRX*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Joubert syndrome 27	*B9D1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Joubert syndrome 34	*B9D2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bardet-Biedl syndrome 12	*BBS12*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bardet-Biedl syndrome 4	*BBS4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bardet-Biedl syndrome 7	*BBS7*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bardet-Biedl syndrome 9	*BBS9*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pseudocholinesterase deficiency	*BCHE*	Autosomal Recessive	General Population	1 in 53	99%	1 in 5,201			•	•
Bartter syndrome, type 4A	*BSND*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
X-linked agammaglobulinemia	*BTK*	X-Linked	General Population	1 in 250,000	99%	1 in 24,999,901			•	•
Orofaciodigital syndrome, type XIV	*C2CD3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Desbuquois dysplasia, type I	*CANT1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
X-linked hyper IgM syndrome	*CD40LG*	X-Linked	General Population	1 in 500,000	99%	1 in 49,999,901			•	•
Joubert syndrome 25	*CEP104*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Joubert syndrome 31	*CEP120*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Joubert syndrome 15	*CEP41*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Retinitis pigmentosa 26	*CERKL*	Autosomal Recessive	General Population	1 in 137	99%	1 in 13,601			•	•
Retinitis pigmentosa 26	*CERKL*	Autosomal Recessive	Sephardic Jewish–Yemenite	1 in 24	99%	1 in 2,301			•	•
Congenital myasthenic syndrome, *CHAT*-related	*CHAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Choroideremia, X-linked	*CHM*	X-Linked	General Population	1 in 25,000	99%	1 in 2,499,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Multiple pterygium syndrome, lethal type	*CHRNG*	Autosomal Recessive	General Population	1 in 50	99%	1 in 4,901			•	•
Usher syndrome, type 1J	*CIB2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bare lymphocyte syndrome, type II	*CIITA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Dent disease	*CLCN5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Steel syndrome	*COL27A1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Congenital myasthenic syndrome, *COLQ*-related	*COLQ*	Autosomal Recessive	General Population	1 in 430	99%	1 in 42,901			•	•
Joubert syndrome 17	*CPLANE1*	Autosomal Recessive	General Population	1 in 423	99%	1 in 42,201			•	•
Carbamoyl phosphate synthetase I deficiency	*CPS1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Carbamoyl phosphate synthetase I deficiency	*CPS1*	Autosomal Recessive	Asian	1 in 447	99%	1 in 44,601			•	•
Carbamoyl phosphate synthetase I deficiency	*CPS1*	Autosomal Recessive	Caucasian	1 in 284	99%	1 in 28,301			•	•
Carnitine palmitoyltransferase I deficiency	*CPT1A*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Carnitine palmitoyltransferase I deficiency	*CPT1A*	Autosomal Recessive	Hutterite	1 in 16	99%	1 in 1,501			•	•
CRB1-related retinal dystrophies	*CRB1*	Autosomal Recessive	General Population	1 in 112	99%	1 in 11,101			•	•
Muscular dystrophy-dystroglycanopathy, type A, 7	*CRPPA*	Autosomal Recessive	General Population	1 in 371	99%	1 in 37,001			•	•
Joubert syndrome 21	*CSPP1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Galactosialidosis	*CTSA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Neuronal ceroid lipofuscinosis, *CTSD*-related	*CTSD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pycnodysostosis	*CTSK*	Autosomal Recessive	General Population	1 in 439	99%	1 in 43,801			•	•
Chronic granulomatous disease 4	*CYBA*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901			•	•
Chronic granulomatous disease 4	*CYBA*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 13	99%	1 in 1,201			•	•
Chronic granulomatous disease, X-linked	*CYBB*	X-Linked	General Population	1 in 150,000	99%	1 in 14,999,901			•	•
Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	*CYP11B1*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701			•	•
Corticosterone methyloxidase deficiency	*CYP11B2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Corticosterone methyloxidase deficiency	*CYP11B2*	Autosomal Recessive	Sephardic Jewish–Iranian	1 in 30	99%	1 in 2,901			•	•
Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	*CYP17A1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Aromatase deficiency	*CYP19A1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Primary congenital glaucoma	*CYP1B1*	Autosomal Recessive	General Population	1 in 74	99%	1 in 7,301			•	•
Omenn syndrome	*DCLRE1C*	Autosomal Recessive	Northern European Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Omenn syndrome	*DCLRE1C*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Omenn syndrome	*DCLRE1C*	Autosomal Recessive	Navajo, Apache	1 in 10	99%	1 in 901			•	•
DCX-related disorders	*DCX*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Perlman syndrome	*DIS3L2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Dyskeratosis congenita, X-linked	*DKC1*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Primary ciliary dyskinesia, *DNAH5*-related	*DNAH5*	Autosomal Recessive	Ashkenazi Jewish	1 in 174	99%	1 in 17,301			•	•
Primary ciliary dyskinesia, *DNAH5*-related	*DNAH5*	Autosomal Recessive	General Population	1 in 120	99%	1 in 11,901			•	•
Primary ciliary dyskinesia, *DNAI1*-related	*DNAI1*	Autosomal Recessive	Ashkenazi Jewish	1 in 352	99%	1 in 35,101			•	•
Primary ciliary dyskinesia, *DNAI1*-related	*DNAI1*	Autosomal Recessive	General Population	1 in 182	99%	1 in 18,101			•	•
Primary ciliary dyskinesia, *DNAI2*-related	*DNAI2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Primary ciliary dyskinesia, *DNAI2*-related	*DNAI2*	Autosomal Recessive	Ashkenazi Jewish	1 in 200	99%	1 in 19,901			•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Autosomal Recessive	General Population	1 in 454	99%	1 in 45,301			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Autosomal Recessive	French Canadian	1 in 353	99%	1 in 35,201			•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Autosomal Recessive	Northern European Caucasian	1 in 290	99%	1 in 28,901			•	•
Congenital myasthenic syndrome, *DOK7*-related	*DOK7*	Autosomal Recessive	Caucasian	1 in 290	99%	1 in 28,901			•	•
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	Autosomal Recessive	East Asian	1 in 50	99%	1 in 4,901			•	•
Dihydropyrimidine dehydrogenase deficiency	*DPYD*	Autosomal Recessive	General Population	1 in 20	99%	1 in 1,901			•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	Autosomal Recessive	General Population	1 in 311	99%	1 in 31,001			•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	Autosomal Recessive	Caucasian	1 in 158	99%	1 in 15,701			•	•
Limb-girdle muscular dystrophy, type 2B	*DYSF*	Autosomal Recessive	Sephardic Jewish–Libyan, Moroccan, Tunisian, Bulgarian	1 in 14	99%	1 in 1,301			•	•
Hypohidrotic ectodermal dysplasia, X-linked	*EDA*	X-Linked	General Population	1 in 3800	99%	1 in 379,901			•	•
Wolcott-Rallison syndrome	*EIF2AK3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Leukoencephalopathy with vanishing white matter 5	*EIF2B5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Emery-Dreifuss muscular dystrophy, X-linked	*EMD*	X-Linked	General Population	1 in 250,000	99%	1 in 24,999,901			•	•
Cerebrooculofacioskeletal syndrome 1/ Cockayne syndrome, type B	*ERCC6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Cockayne syndrome, type A	*ERCC8*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Roberts-SC phocomelia syndrome	*ESCO2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIA	*ETFA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIB	*ETFB*	Autosomal Recessive	General Population	1 in 408	99%	1 in 40,701			•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIC	*ETFDH*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901			•	•
Multiple acyl-CoA dehydrogenase deficiency/Glutaric aciduria, type IIC	*ETFDH*	Autosomal Recessive	Asian	1 in 87	99%	1 in 8,601			•	•
Ethylmalonic encephalopathy	*ETHE1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Ellis-van Creveld syndrome	*EVC*	Autosomal Recessive	General Population	1 in 345	99%	1 in 34,401			•	•
Ellis-van Creveld syndrome	*EVC*	Autosomal Recessive	Lancaster County Amish	1 in 12	99%	1 in 1,101			•	•
Pontocerebellar hypoplasia, type 1B	*EXOSC3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Retinitis pigmentosa 25	*EYS*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901			•	•
Retinitis pigmentosa 25	*EYS*	Autosomal Recessive	General Population	1 in 129	99%	1 in 12,801			•	•
Retinitis pigmentosa 25	*EYS*	Autosomal Recessive	Caucasian	1 in 53	99%	1 in 5,201			•	•
Retinitis pigmentosa 25	*EYS*	Autosomal Recessive	Sephardic Jewish–Moroccan	1 in 42	99%	1 in 4,101			•	•
Retinitis pigmentosa 28	*FAM161A*	Autosomal Recessive	General Population	1 in 289	99%	1 in 28,801			•	•
Retinitis pigmentosa 28	*FAM161A*	Autosomal Recessive	Ashkenazi Jewish	1 in 214	99%	1 in 21,301			•	•
Retinitis pigmentosa 28	*FAM161A*	Autosomal Recessive	Sephardic Jewish–Libyan, Moroccan, Tunisian, Bulgarian	1 in 41	99%	1 in 4,001			•	•
Fanconi anemia, complementation group B	*FANCB*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Fanconi anemia, complementation group D2	*FANCD2*	Autosomal Recessive	General Population	<1 in 500	98%	<1 in 24,951			•	•
Fanconi anemia, complementation group E	*FANCE*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Fanconi anemia, complementation group F	*FANCF*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Fanconi anemia, complementation group G	*FANCG*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Fanconi anemia, complementation group G	*FANCG*	Autosomal Recessive	African American	1 in 100	99%	1 in 9,901			•	•
Fanconi anemia, complementation group I	*FANCI*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901			•	•
Fanconi anemia, complementation group L	*FANCL*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901			•	•
Fumarase deficiency	*FH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	*FOXP3*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	Autosomal Recessive	General Population	1 in 30	99%	1 in 2,901			•	•
Glucose-6-phosphate dehydrogenase deficiency	*G6PD*	Autosomal Recessive	African American	1 in 5	99%	1 in 401			•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901			•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	Autosomal Recessive	General Population	1 in 122	99%	1 in 12,101			•	•
Galactosemia, type II/Galactokinase deficiency	*GALK1*	Autosomal Recessive	Roma	1 in 47	99%	1 in 4,601			•	•
Mucopolysaccharidosis, type IVA/Morquio syndrome	*GALNS*	Autosomal Recessive	General Population	1 in 307	99%	1 in 30,601			•	•
Hyperphosphatemic familial tumoral calcinosis	*GALNT3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Guanidinoacetate methyltransferase deficiency	*GAMT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Guanidinoacetate methyltransferase deficiency	*GAMT*	Autosomal Recessive	Portuguese	1 in 125	99%	1 in 12,401			•	•
Combined oxidative phosphorylation deficiency 1	*GFM1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Charcot-Marie-Tooth disease, type 1X	*GJB1*	X-Linked	General Population	1 in 7000	99%	1 in 699,901			•	•
Rhizomelic chondrodysplasia punctata, type 2	*GNPAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mucolipidosis III gamma	*GNPTG*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mucolipidosis III gamma	*GNPTG*	Autosomal Recessive	Caucasian	1 in 273	99%	1 in 27,201			•	•
Bernard-Soulier syndrome, type A	*GP1BA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bernard-Soulier syndrome, type C	*GP9*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Primary hyperoxaluria, type II	*GRHPR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mucopolysaccharidosis, type VII/Sly syndrome	*GUSB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
3-hydroxyacyl-CoA dehydrogenase deficiency	*HADH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial trifunctional protein deficiency, *HADHB*-related	*HADHB*	Autosomal Recessive	General Population	1 in 146	99%	1 in 14,501			•	•
Congenital neutropenia, *HAX1*-related	*HAX1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Hereditary hemochromatosis, type 2	*HJV*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Hereditary hemochromatosis, type 2	*HJV*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Primary hyperoxaluria, type III	*HOGA1*	Autosomal Recessive	General Population	1 in 309	99%	1 in 30,801			•	•
HPRT1-related disorders	*HPRT1*	X-Linked	General Population	1 in 285,000	99%	1 in 28,499,901			•	•
Hermansky-Pudlak syndrome, type 4	*HPS4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
17-beta-hydroxysteroid dehydrogenase deficiency, type III	*HSD17B3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
3-beta-hydroxysteroid dehydrogenase deficiency, type II	*HSD3B2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mucopolysaccharidosis, type IX/ Hyaluronidase deficiency	*HYAL1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Severe combined immunodeficiency, X-linked	*IL2RG*	X-Linked	General Population	1 in 69,000	99%	1 in 6,899,901			•	•
Joubert syndrome 1	*INPP5E*	Autosomal Recessive	General Population	1 in 264	99%	1 in 26,301			•	•
Nephronophthisis 2	*INVS*	Autosomal Recessive	General Population	1 in 373	99%	1 in 37,201			•	•
Senior-Løken syndrome 5	*IQCB1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Muscular dystrophy-dystroglycanopathy, type A, 6	*LARGE1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Leber congenital amaurosis 5	*LCA5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	French Canadian	1 in 267	99%	1 in 26,601			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	Caucasian	1 in 200	99%	1 in 19,901			•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	Finnish	1 in 143	99%	1 in 14,201			•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	South African Afrikaner	1 in 70	99%	1 in 6,901			•	•
Familial hypercholesterolemia, *LDLR*-related	*LDLR*	Autosomal Recessive	Ashkenazi Jewish	1 in 69	99%	1 in 6,801			•	•
Familial hypercholesterolemia, *LDLRAP1*-related	*LDLRAP1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Familial hypercholesterolemia, *LDLRAP1*-related	*LDLRAP1*	Autosomal Recessive	Sardinian	1 in 143	99%	1 in 14,201			•	•
Combined pituitary hormone deficiency, type 3	*LHX3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Stüve-Wiedemann syndrome	*LIFR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Lysosomal acid lipase deficiency	*LIPA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Lysosomal acid lipase deficiency	*LIPA*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901			•	•
Lysosomal acid lipase deficiency	*LIPA*	Autosomal Recessive	Sephardic Jewish–Iranian	1 in 26	99%	1 in 2,501			•	•
Lipoprotein lipase deficiency	*LPL*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Lipoprotein lipase deficiency	*LPL*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Lipoprotein lipase deficiency	*LPL*	Autosomal Recessive	Asian	1 in 189	99%	1 in 18,801			•	•
Lipoprotein lipase deficiency	*LPL*	Autosomal Recessive	French Canadian–Other	1 in 139	99%	1 in 13,801			•	•
Lipoprotein lipase deficiency	*LPL*	Autosomal Recessive	French Canadian–Saguenay-Lac-Saint-Jean	1 in 46	99%	1 in 4,501			•	•
Chédiak-Higashi syndrome	*LYST*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Neuronal ceroid lipofuscinosis, *MFSD8*-related	*MFSD8*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Bardet-Biedl syndrome 6	*MKKS*	Autosomal Recessive	General Population	1 in 219	99%	1 in 21,801			•	•
MKS1-related disorders	*MKS1*	Autosomal Recessive	General Population	1 in 260	99%	1 in 25,901			•	•
MKS1-related disorders	*MKS1*	Autosomal Recessive	Caucasian	1 in 260	99%	1 in 25,901			•	•
MKS1-related disorders	*MKS1*	Autosomal Recessive	Finnish	1 in 47	99%	1 in 4,601			•	•
Malonyl-CoA decarboxylase deficiency	*MLYCD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Combined methylmalonic aciduria and homocystinuria, cblD type/Cobalamin D deficiency	*MMADHC*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Combined methylmalonic aciduria and homocystinuria, cblD type/Cobalamin D deficiency	*MMADHC*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Molybdenum cofactor deficiency of complementation group A	*MOCS1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Congenital disorder of glycosylation, type Ib	*MPI*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
MPV17-related mitochondrial DNA maintenance defect	*MPV17*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
MPV17-related mitochondrial DNA maintenance defect	*MPV17*	Autosomal Recessive	Navajo	1 in 20	99%	1 in 1,901			•	•
Ataxia-telangiectasia-like disorder 1	*MRE11*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Homocystinuria caused by methylenetetrahydrofolate reductase deficiency	*MTHFR*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701			•	•
Homocystinuria caused by methylenetetrahydrofolate reductase deficiency	*MTHFR*	Autosomal Recessive	Sephardic Jewish–Bukharan	1 in 39	99%	1 in 3,801			•	•
X-linked myotubular myopathy	*MTM1*	X-Linked	General Population	1 in 38,000	99%	1 in 3,799,901			•	•
Homocystinuria, type cblE	*MTRR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Homocystinuria, type cblE	*MTRR*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Nonsyndromic hearing loss and deafness 3	*MYO15A*	Autosomal Recessive	General Population	1 in 117	99%	1 in 11,601			•	•
N-acetylglutamate synthase deficiency	*NAGS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Charcot-Marie-Tooth disease, type 4D	*NDRG1*	Autosomal Recessive	Roma	1 in 22	99%	1 in 2,101			•	•
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial complex I deficiency, nuclear type 16	*NDUFAF5*	Autosomal Recessive	Ashkenazi Jewish	1 in 290	99%	1 in 28,901			•	•
Mitochondrial complex I deficiency, nuclear type 17	*NDUFAF6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial complex I deficiency, nuclear type 1	*NDUFS4*	Autosomal Recessive	General Population	1 in 423	99%	1 in 42,201			•	•
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial complex I deficiency, nuclear type 9	*NDUFS6*	Autosomal Recessive	Caucasus Jewish	1 in 24	99%	1 in 2,301			•	•
Sialidosis	*NEU1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Niemann-Pick disease, type C2	*NPC2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
NPHP1 nephronophthisis-related ciliopathies	*NPHP1*	Autosomal Recessive	General Population	1 in 202	99%	1 in 20,101			•	•
NPHP3 nephronophthisis-related ciliopathies	*NPHP3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
NPHP4 nephronophthisis-related ciliopathies	*NPHP4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Enhanced S-cone syndrome	*NR2E3*	Autosomal Recessive	General Population	1 in 204	99%	1 in 20,301			•	•
Enhanced S-cone syndrome	*NR2E3*	Autosomal Recessive	Ashkenazi Jewish	1 in 100	99%	1 in 9,901			•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	Autosomal Recessive	Sephardic Jewish–Moroccan	<1 in 500	99%	<1 in 49,901			•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Congenital insensitivity to pain with anhidrosis	*NTRK1*	Autosomal Recessive	Asian	1 in 387	99%	1 in 38,601			•	•
Ornithine aminotransferase deficiency	*OAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Ornithine aminotransferase deficiency	*OAT*	Autosomal Recessive	Sephardic Jewish–Iraqi, Syrian	1 in 177	99%	1 in 17,601			•	•
Ornithine aminotransferase deficiency	*OAT*	Autosomal Recessive	Finnish	1 in 147	99%	1 in 14,601			•	•
Lowe syndrome, X-linked	*OCRL*	X-Linked	General Population	1 in 375,000	99%	1 in 37,499,901			•	•
3-methylglutaconic aciduria, type III/ Costeff syndrome	*OPA3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
3-methylglutaconic aciduria, type III/ Costeff syndrome	*OPA3*	Autosomal Recessive	Sephardic Jewish–Iraqi	1 in 13	99%	1 in 1,201			•	•
Pyruvate dehydrogenase E1-alpha deficiency	*PDHA1*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Pyruvate dehydrogenase E1-beta deficiency	*PDHB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Prolidase deficiency	*PEPD*	Autosomal Recessive	General Population	1 in 242	99%	1 in 24,101			•	•
Mitochondrial complex IV deficiency, nuclear type 12	*PET100*	Autosomal Recessive	General Population	1 in 452	99%	1 in 45,101			•	•
Zellweger spectrum disorder, *PEX10*-related	*PEX10*	Autosomal Recessive	Asian	<1 in 500	99%	<1 in 49,901			•	•
Zellweger spectrum disorder, *PEX10*-related	*PEX10*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Zellweger spectrum disorder, *PEX12*-related	*PEX12*	Autosomal Recessive	General Population	1 in 406	99%	1 in 40,501			•	•
Zellweger spectrum disorder, *PEX26*-related	*PEX26*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Glycogen storage disease, type VII	*PFKM*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Glycogen storage disease, type VII	*PFKM*	Autosomal Recessive	Ashkenazi Jewish	1 in 250	99%	1 in 24,901			•	•
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Phosphoglycerate dehydrogenase deficiency	*PHGDH*	Autosomal Recessive	Ashkenazi Jewish	1 in 453	99%	1 in 45,201			•	•
Refsum disease	*PHYH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
PLA2G6-associated neurodegeneration	*PLA2G6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Steroid-resistant nephrotic syndrome, type 3	*PLCE1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Muscular dystrophy-dystroglycanopathy, type A, 3	*POMGNT1*	Autosomal Recessive	General Population	1 in 462	99%	1 in 46,101			•	•
Muscular dystrophy-dystroglycanopathy, type A, 3	*POMGNT1*	Autosomal Recessive	Finnish	1 in 111	99%	1 in 11,001			•	•
Muscular dystrophy-dystroglycanopathy, type A, 1	*POMT1*	Autosomal Recessive	General Population	1 in 275	99%	1 in 27,401			•	•
Muscular dystrophy-dystroglycanopathy, type A, 2	*POMT2*	Autosomal Recessive	General Population	1 in 465	99%	1 in 46,401			•	•
PRPS1-related disorders	*PRPS1*	X-Linked	General Population	1 in 500,000	99%	1 in 49,999,901			•	•
Metachromatic leukodystrophy due to saposin B deficiency	*PSAP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Myopathy, lactic acidosis, and sideroblastic anemia	*PUS1*	Autosomal Recessive	Sephardic Jewish–Iranian	<1 in 500	99%	<1 in 49,901			•	•
Myopathy, lactic acidosis, and sideroblastic anemia	*PUS1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Glycogen storage disease, type V	*PYGM*	Autosomal Recessive	Caucasian	1 in 191	99%	1 in 19,001			•	•
Glycogen storage disease, type V	*PYGM*	Autosomal Recessive	General Population	1 in 191	99%	1 in 19,001			•	•
Glycogen storage disease, type V	*PYGM*	Autosomal Recessive	Sephardic Jewish–Kurdish	1 in 84	99%	1 in 8,301			•	•
Carpenter syndrome	*RAB23*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Carpenter syndrome	*RAB23*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Severe combined immunodeficiency, *RAG1*-related	*RAG1*	Autosomal Recessive	General Population	1 in 245	99%	1 in 24,401			•	•
Severe combined immunodeficiency, *RAG2*-related	*RAG2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Severe combined immunodeficiency, *RAG2*-related	*RAG2*	Autosomal Recessive	Sephardic Jewish–Iraqi	<1 in 500	99%	<1 in 49,901			•	•
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	Autosomal Recessive	Sephardic Jewish–Iraqi, Iranian	<1 in 500	99%	<1 in 49,901			•	•
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	Autosomal Recessive	General Population	1 in 252	99%	1 in 25,101			•	•
Congenital myasthenic syndrome, *RAPSN*-related	*RAPSN*	Autosomal Recessive	Caucasian	1 in 176	99%	1 in 17,501			•	•
Leber congenital amaurosis 13	*RDH12*	Autosomal Recessive	General Population	1 in 456	99%	1 in 45,501			•	•
Aicardi-Goutières syndrome 4	*RNASEH2A*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Aicardi-Goutières syndrome 3	*RNASEH2C*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Leber congenital amaurosis 2	*RPE65*	Autosomal Recessive	General Population	1 in 228	99%	1 in 22,701			•	•
Leber congenital amaurosis 2	*RPE65*	Autosomal Recessive	Sephardic Jewish–North African	1 in 90	99%	1 in 8,901			•	•
Ciliopathies, *RPGRIP1L*-related	*RPGRIP1L*	Autosomal Recessive	General Population	1 in 259	99%	1 in 25,801			•	•
Aicardi-Goutières syndrome 5	*SAMHD1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Shwachman-Diamond syndrome	*SBDS*	Autosomal Recessive	General Population	1 in 145	99%	1 in 14,401			•	•
Action myoclonus renal failure syndrome	*SCARB2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Progressive cerebello-cerebral atrophy	*SEPSECS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Progressive cerebello-cerebral atrophy	*SEPSECS*	Autosomal Recessive	Sephardic Jewish–Moroccan, Iraqi	1 in 41	99%	1 in 4,001			•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	Autosomal Recessive	General Population	1 in 38	99%	1 in 3,701			•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	Autosomal Recessive	Ashkenazi Jewish	1 in 24	99%	1 in 2,301			•	•
Alpha-1 antitrypsin deficiency	*SERPINA1*	Autosomal Recessive	Northern European Caucasian	1 in 15	99%	1 in 1,401			•	•
Limb-girdle muscular dystrophy, type 6	*SGCD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Autosomal Recessive	Northern European Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Autosomal Recessive	General Population	1 in 354	99%	1 in 35,301			•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Autosomal Recessive	Moroccan	1 in 250	99%	1 in 24,901			•	•
Limb-girdle muscular dystrophy, type 5	*SGCG*	Autosomal Recessive	Roma	1 in 96	99%	1 in 9,501			•	•
Gitelman syndrome	*SLC12A3*	Autosomal Recessive	General Population	1 in 100	99%	1 in 9,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Free sialic acid storage disorders	*SLC17A5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Free sialic acid storage disorders	*SLC17A5*	Autosomal Recessive	Canadian Inuit	1 in 129	99%	1 in 12,801			•	•
Free sialic acid storage disorders	*SLC17A5*	Autosomal Recessive	Swedish	1 in 125	99%	1 in 12,401			•	•
Free sialic acid storage disorders	*SLC17A5*	Autosomal Recessive	Finnish	1 in 100	99%	1 in 9,901			•	•
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	*SLC1A4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Autosomal Recessive	Asian	1 in 123	99%	1 in 12,201			•	•
Citrin deficiency/Citrullinemia, type II	*SLC25A13*	Autosomal Recessive	East Asian	1 in 65	99%	1 in 6,401			•	•
Ornithine translocase deficiency	*SLC25A15*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Ornithine translocase deficiency	*SLC25A15*	Autosomal Recessive	French Canadian	1 in 20	99%	1 in 1,901			•	•
Ornithine translocase deficiency	*SLC25A15*	Autosomal Recessive	Métis Nation–Saskatchewan	1 in 19	99%	1 in 1,801			•	•
Carnitine-acylcarnitine translocase deficiency	*SLC25A20*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Acrodermatitis enteropathica	*SLC39A4*	Autosomal Recessive	General Population	1 in 354	99%	1 in 35,301			•	•
Corneal dystrophy and perceptive deafnesssyndrome	*SLC4A11*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Renal tubular acidosis	*SLC4A4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Schimke immunoosseous dysplasia	*SMARCAL1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
5-alpha-reductase deficiency	*SRD5A2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Lipoid congenital adrenal hyperplasia	*STAR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Lipoid congenital adrenal hyperplasia	*STAR*	Autosomal Recessive	East Asian	1 in 177	99%	1 in 17,601			•	•
Familial hemophagocytic lymphohistiocytosis 4	*STX11*	Autosomal Recessive	General Population	1 in 354	99%	1 in 35,301			•	•
Familial hemophagocytic l ymphohistiocytosis 5	*STXBP2*	Autosomal Recessive	General Population	1 in 296	99%	1 in 29,501			•	•
Multiple sulfatase deficiency	*SUMF1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Multiple sulfatase deficiency	*SUMF1*	Autosomal Recessive	Ashkenazi Jewish	1 in 279	99%	1 in 27,801			•	•
Barth syndrome	*TAFAZZIN*	X-Linked	General Population	1 in 225,000	99%	1 in 22,499,901			•	•
Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	*TANGO2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Autosomal Recessive	Ashkenazi Jewish	1 in 350	99%	1 in 34,901			•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Autosomal Recessive	General Population	1 in 316	99%	1 in 31,501			•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Autosomal Recessive	Costa Rican	1 in 86	99%	1 in 8,501			•	•
Osteopetrosis, infantile malignant, *TCIRG1*-related	*TCIRG1*	Autosomal Recessive	Chuvashiya	1 in 60	99%	1 in 5,901			•	•
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	*TECPR2*	Autosomal Recessive	Sephardic Jewish–Bukharan	1 in 27	99%	1 in 2,601			•	•
Hereditary hemochromatosis, type 3	*TFR2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Progressive familial intrahepatic cholestasis 4	*TJP2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mulibrey nanism	*TRIM37*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Acute infantile liver failure	*TRMU*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Acute infantile liver failure	*TRMU*	Autosomal Recessive	Sephardic Jewish–Yemenite	1 in 34	99%	1 in 3,301			•	•
Pontocerebellar hypoplasia, type 2B	*TSEN2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Pontocerebellar hypoplasia, types 4 and 2A	*TSEN54*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Combined oxidative phosphorylation deficiency 3	*TSFM*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Combined oxidative phosphorylation deficiency 3	*TSFM*	Autosomal Recessive	Finnish	1 in 80	99%	1 in 7,901			•	•
Trichohepatoenteric syndrome 1	*TTC37*	Autosomal Recessive	General Population	1 in 381	99%	1 in 38,001			•	•
Bardet-Biedl syndrome 8	*TTC8*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Ataxia with isolated vitamin E deficiency	*TTPA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Ataxia with isolated vitamin E deficiency	*TTPA*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	Autosomal Recessive	Caucasian	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Mitochondrial DNA depletion syndrome 1, MNGIE type	*TYMP*	Autosomal Recessive	Sephardic Jewish–Iranian	1 in 158	99%	1 in 15,701			•	•
X-linked infantile spinal muscular atrophy	*UBA1*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
Johanson-Blizzard syndrome	*UBR1*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901			•	•
Beta-ureidopropionase deficiency	*UPB1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Choreoacanthocytosis	*VPS13A*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Choreoacanthocytosis	*VPS13A*	Autosomal Recessive	Ashkenazi Jewish	<1 in 500	99%	<1 in 49,901			•	•
Cohen syndrome	*VPS13B*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Severe congenital neutropenia 5	*VPS45*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pontocerebellar hypoplasia, type 2E	*VPS53*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pontocerebellar hypoplasia, type 1A	*VRK1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Pontocerebellar hypoplasia, type 1A	*VRK1*	Autosomal Recessive	Ashkenazi Jewish	1 in 225	99%	1 in 22,401			•	•
Microphthalmia/Anophthalmia	*VSX2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Microphthalmia/Anophthalmia	*VSX2*	Autosomal Recessive	Sephardic Jewish–Iranian, Syrian	1 in 145	99%	1 in 14,401			•	•
Wiskott-Aldrich syndrome, X-linked	*WAS*	X-Linked	General Population	1 in 187,500	99%	1 in 18,749,901			•	•
Odonto-onycho-dermal dysplasia/ Schopf-Schulz-Passarge syndrome	*WNT10A*	Autosomal Recessive	General Population	1 in 305	99%	1 in 30,401			•	•
Werner syndrome	*WRN*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301			•	•
WWOX deficiency	*WWOX*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Xeroderma pigmentosum, group A	*XPA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
Spastic paraplegia, type 15	*ZFYVE26*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901			•	•
X-linked heterotaxy-1	*ZIC3*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901			•	•
ABCA4-related disorders	*ABCA4*	Autosomal Recessive	General Population	1 in 45	99%	1 in 4,401				•
Progressive familial intrahepatic cholestasis 3	*ABCB4*	Autosomal Recessive	General Population	1 in 194	99%	1 in 19,301				•
Dubin-Johnson syndrome	*ABCC2*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701				•
Dubin-Johnson syndrome	*ABCC2*	Autosomal Recessive	Sephardic Jews–Iranian, Moroccan	1 in 27	99%	1 in 2,601				•
ADAMTSL4-related eye disorders	*ADAMTSL4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
ADAMTSL4-related eye disorders	*ADAMTSL4*	Autosomal Recessive	Bukharan Jewish	1 in 48	99%	1 in 4,701				•
ADAMTSL4-related eye disorders	*ADAMTSL4*	Autosomal Recessive	Norwegian	1 in 63	99%	1 in 6,201				•
Leber congenital amaurosis 4	*AIPL1*	Autosomal Recessive	General Population	1 in 387	99%	1 in 38,601				•
Congenital disorder of glycosylation, type Ik	*ALG1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Developmental and epileptic encephalopathy 36	*ALG13*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901				•
Imerslund-Gräsbeck syndrome 2	*AMN*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Androgen insensitivity syndrome	*AR*	X-Linked	General Population	1 in 5000	99%	1 in 499,901				•
Bardet-Biedl syndrome 3	*ARL6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Bardet-Biedl syndrome 3	*ARL6*	Autosomal Recessive	Newfoundland	1 in 289	99%	1 in 28,801				•
AVPR2-related disorders	*AVPR2*	X-Linked	General Population	1 in 63,000	99%	1 in 6,299,901				•
Bardet-Biedl syndrome 5	*BBS5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hermansky-Pudlak syndrome, type 8	*BLOC1S3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hermansky-Pudlak syndrome, type 9	*BLOC1S6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Osteogenesis imperfecta, type XIII	*BMP1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Fanconi anemia, complementation group J	*BRIP1*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901				•
Developmental and epileptic encephalopathy 50	*CAD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Developmental and epileptic encephalopathy 50	*CAD*	Autosomal Recessive	Finnish	1 in 212	99%	1 in 21,101				•
Developmental and epileptic encephalopathy 50	*CAD*	Autosomal Recessive	Ashkenazi Jewish	1 in 458	99%	1 in 45,701				•
Catecholaminergic polymorphic ventricular tachycardia, type 2	*CASQ2*	Autosomal Recessive	General Population	1 in 224	99%	1 in 22,301				•
Autosomal recessive intellectual developmental disorder, type 3	*CC2D1A*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Autosomal recessive intellectual developmental disorder, type 3	*CC2D1A*	Autosomal Recessive	Ashkenazi Jewish	1 in 445	99%	1 in 44,401				•
Primary ciliary dyskinesia, *CCDC103*-related	*CCDC103*	Autosomal Recessive	General Population	1 in 217	99%	1 in 21,601				•
Primary ciliary dyskinesia, *CCDC39*-related	*CCDC39*	Autosomal Recessive	General Population	1 in 144	99%	1 in 14,301				•
Progressive pseudorheumatoid dysplasia	*CCN6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *CD3D*-related	*CD3D*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *CD3E*-related	*CD3E*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hyper-IgM syndrome, type 3	*CD40*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
CD59-mediated hemolytic anemia	*CD59*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
CD59-mediated hemolytic anemia	*CD59*	Autosomal Recessive	Sephardic Jews–North African	1 in 66	99%	1 in 6,501				•
CEP152-related disorders	*CEP152*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
COL11A2-related disorders	*COL11A2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Junctional epidermolysis bullosa, *COL17A1*-related	*COL17A1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Mitochondrial complex IV deficiency, nuclear type 6	*COX15*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Osteogenesis imperfecta, type VII	*CRTAP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
CTSC-related disorders	*CTSC*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901				•
CYP7B1-related disorders	*CYP7B1*	Autosomal Recessive	General Population	1 in 338	99%	1 in 33,701				•
Woodhouse-Sakati syndrome	*DCAF17*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Warsaw breakage syndrome	*DDX11*	Autosomal Recessive	General Population	<1 in 500	15%	1 in 588				•
DGAT1 deficiency	*DGAT1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Deoxyguanosine kinase deficiency/ Mitochondrial DNA depletion syndrome 3	*DGUOK*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Spondylocostal dysostosis 1	*DLL3*	Autosomal Recessive	General Population	1 in 289	99%	1 in 28,801				•
Spondylocostal dysostosis 1	*DLL3*	Autosomal Recessive	Northern European Caucasian	1 in 350	99%	1 in 34,901				•
Primary ciliary dyskinesia, *DNAH11*-related	*DNAH11*	Autosomal Recessive	General Population	1 in 144	99%	1 in 14,301				•
Immunodeficiency-centromeric instability-facial anomalies syndrome 1	*DNMT3B*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Thyroid dyshormonogenesis 6	*DUOX2*	Autosomal Recessive	General Population	1 in 55	95%	1 in 1,081				•
Leukoencephalopathy with vanishing white matter 1	*EIF2B1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Leukoencephalopathy with vanishing white matter 2	*EIF2B2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Leukoencephalopathy with vanishing white matter 3	*EIF2B3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Leukoencephalopathy with vanishing white matter 4	*EIF2B4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
EPG5-related disorder	*EPG5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Factor II deficiency/Prothrombin deficiency	*F2*	Autosomal Recessive	General Population	1 in 33	99%	1 in 3,201				•
Factor V deficiency	*F5*	Autosomal Recessive	General Population	1 in 12	99%	1 in 1,101				•
Fructose-1,6-bisphosphatase deficiency	*FBP1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Fructose-1,6-bisphosphatase deficiency	*FBP1*	Autosomal Recessive	Caucasian	1 in 296	99%	1 in 29,501				•
Parkinson disease 15	*FBXO7*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
FHL1-related disorders	*FHL1*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901				•
Osteogenesis imperfecta, type XI	*FKBP10*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *FOXN1*-related	*FOXN1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Mitochondrial complex I deficiency, nuclear type 19	*FOXRED1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Fraser syndrome, type 1	*FRAS1*	Autosomal Recessive	General Population	1 in 316	99%	1 in 31,501				•
Fraser syndrome, type 2	*FREM2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Fucosidosis	*FUCA1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
G6PC3 deficiency	*G6PC3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Galactosemia, type III/Galactose epimerase deficiency	*GALE*	Autosomal Recessive	General Population	1 in 132	99%	1 in 13,101				•
Galactosemia, type III/Galactose epimerase deficiency	*GALE*	Autosomal Recessive	African American	1 in 41	99%	1 in 4,001				•
Arginine:glycine amidinotransferase deficiency	*GATM*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
GCH1-related disorders	*GCH1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
GDF5-related disorders	*GDF5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Laron syndrome	*GHR*	Autosomal Recessive	General Population	1 in 167	99%	1 in 16,601				•
Geroderma osteodysplastica	*GORAB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Glutathione synthetase deficiency	*GSS*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Leber congenital amaurosis 1	*GUCY2D*	Autosomal Recessive	General Population	1 in 200	99%	1 in 19,901				•
Hereditary hemochromatosis, type 2B	*HAMP*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Methylmalonic aciduria and homocystinuria, cblX type	*HCFC1*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901				•
Alkaptonuria	*HGD*	Autosomal Recessive	General Population	1 in 250	99%	1 in 24,901				•
Heme oxygenase 1 deficiency	*HMOX1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Tyrosinemia, type III	*HPD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hermansky-Pudlak syndrome, type 5	*HPS5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hermansky-Pudlak syndrome, type 6	*HPS6*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
HSD10 disease	*HSD17B10*	X-Linked	General Population	<1 in 750,000	99%	<1 in 74,999,901				•
IGHMBP2-related disorders	*IGHMBP2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *IKBKB*-related	*IKBKB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *IL7R*-related	*IL7R*	Autosomal Recessive	General Population	1 in 388	99%	1 in 38,701				•
Junctional epidermolysis bullosa, *ITGA6*-related	*ITGA6*	Autosomal Recessive	General Population	1 in 159	99%	1 in 15,801				•
ITGB3-related disorders	*ITGB3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Junctional epidermolysis bullosa, *ITGB4*-related	*ITGB4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Severe combined immunodeficiency, *JAK3*-related	*JAK3*	Autosomal Recessive	General Population	1 in 299	99%	1 in 29,801				•
Bartter syndrome, type 2	*KCNJ1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
LIG4 syndrome	*LIG4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Methylmalonic aciduria and homocystinuria, cblF type	*LMBRD1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
LRAT-related disorders	*LRAT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Retinitis pigmentosa 62	*MAK*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Retinitis pigmentosa 62	*MAK*	Autosomal Recessive	Ashkenazi Jewish	1 in 55	99%	1 in 5,401				•
Beta-mannosidosis	*MANBA*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Methylmalonyl-CoA epimerase deficiency	*MCEE*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
MECR-related neurologic disorder	*MECR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Molybdenum cofactor deficiency of complementation group B	*MOCS2*	Autosomal Recessive	General Population	1 in 236	99%	1 in 23,501				•
Homocystinuria-megaloblastic anemia, cblG type	*MTR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Congenital myasthenic syndrome, *MUSK*-related	*MUSK*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Chronic granulomatous disease 2	*NCF2*	Autosomal Recessive	General Population	1 in 500	99%	<1 in 49,901				•
Mitochondrial complex I deficiency, nuclear type 10	*NDUFAF2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Mitochondrial complex I deficiency, nuclear type 3	*NDUFS7*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Mitochondrial complex I deficiency, nuclear type 4	*NDUFV1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Congenital disorder of deglycosylation, type 1	*NGLY1*	Autosomal Recessive	General Population	1 in 274	99%	1 in 27,301				•
Lung disease, immunodeficiency, and chromosome breakage syndrome	*NSMCE3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Osteopetrosis, *OSTM1*-related	*OSTM1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Nonsyndromic hearing loss and deafness 22	*OTOA*	Autosomal Recessive	General Population	<1 in 500	88%	1 in 4,159				•
Nonsyndromic hearing loss and deafness 9	*OTOF*	Autosomal Recessive	General Population	1 in 96	99%	1 in 9,501				•
Osteogenesis imperfecta, type VIII	*P3H1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Osteogenesis imperfecta, type VIII	*P3H1*	Autosomal Recessive	African American	1 in 240	99%	1 in 23,901				•
Pantothenate kinase-associated neurodegeneration	*PANK2*	Autosomal Recessive	General Population	1 in 289	99%	1 in 28,801				•
Pterin-4 alpha-carbinolamine dehydratase deficiency	*PCBD1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Microcephalic osteodysplastic primordial dwarfism, type II	*PCNT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Microcephalic osteodysplastic primordial dwarfism, type II	*PCNT*	Autosomal Recessive	Druze–Northern Israel	1 in 30	99%	1 in 2,901				•
Zellweger spectrum disorder, *PEX13*-related	*PEX13*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Zellweger spectrum disorder, *PEX16*-related	*PEX16*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Zellweger spectrum disorder, *PEX5*-related	*PEX5*	Autosomal Recessive	General Population	1 in 408	99%	1 in 40,701				•
PGM3-congenital disorder of glycosylation/ Immunodeficiency 23	*PGM3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Glycogen storage disease, type IXb	*PHKB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Glycogen storage disease, type IXc	*PHKG2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
PIGN-related disorders	*PIGN*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Nonsyndromic hearing loss and deafness 59	*PJVK*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
PLEKHG5-related disorders	*PLEKHG5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome	*PLOD1*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701				•
Pyridoxamine 5'-phosphate oxidase deficiency	*PNPO*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Xeroderma pigmentosum, variant type	*POLH*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Xeroderma pigmentosum, variant type	*POLH*	Autosomal Recessive	Japanese	1 in 167	99%	1 in 16,601				•
Cytochrome P450 oxidoreductase deficiency	*POR*	Autosomal Recessive	General Population	1 in 370	99%	1 in 36,901				•
Combined or isolated pituitary hormone deficiency, type 1	*POU1F1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Retinitis pigmentosa 36	*PRCD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Brittle cornea syndrome 2	*PRDM5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Severe combined immunodeficiency, *PTPRC*-related	*PTPRC*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Dihydropteridine reductase deficiency	*QDPR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
RLBP1-related retinopathies	*RLBP1*	Autosomal Recessive	General Population	1 in 296	99%	1 in 29,501				•
Retinitis pigmentosa 2	*RP2*	X-LinkedL-LinkedL	General Population	1 in 58,000	99%	1 in 5,799,901				•
Muscular dystrophy-dystroglycanopathy, type A, 10	*RXYLT1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
RYR1-related disorders	*RYR1*	Autosomal Recessive	General Population	1 in 150	99%	1 in 14,901				•
Normophosphatemic familial tumoral calcinosis	*SAMD9*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Congenital dyserythropoietic anemia, type II	*SEC23B*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Trichohepatoenteric syndrome 2	*SKIC2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Bartter syndrome, type 1	*SLC12A1*	Autosomal Recessive	General Population	1 in 360	99%	1 in 35,901				•
Thiamine-responsive megaloblastic anemia syndrome	*SLC19A2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Congenital secretory chloride diarrhea 1	*SLC26A3*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Ichthyosis prematurity syndrome	*SLC27A4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Ichthyosis prematurity syndrome	*SLC27A4*	Autosomal Recessive	Norwegian	1 in 50	99%	1 in 4,901				•
Ichthyosis prematurity syndrome	*SLC27A4*	Autosomal Recessive	Swedish	1 in 50	99%	1 in 4,901				•
Foveal hypoplasia 2	*SLC38A8*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Oculocutaneous albinism, type IV	*SLC45A2*	Autosomal Recessive	General Population	1 in 158	99%	1 in 15,701				•
Oculocutaneous albinism, type IV	*SLC45A2*	Autosomal Recessive	Japanese	1 in 146	99%	1 in 14,501				•
Thyroid dyshormonogenesis 1	*SLC5A5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	*SNAP29*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
SPG11-related disorders	*SPG11*	Autosomal Recessive	General Population	1 in 141	99%	1 in 14,001				•
Sepiapterin reductase deficiency	*SPR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Amish infantile epilepsy syndrome	*ST3GAL5*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Isolated sulfite oxidase deficiency	*SUOX*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
SURF1-related disorders	*SURF1*	Autosomal Recessive	General Population	1 in 316	99%	1 in 31,501				•
Nonsyndromic hearing loss and deafness 76	*SYNE4*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum	*TBCD*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
TBCE-related disorders	*TBCE*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Transcobalamin II deficiency	*TCN2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Dyskeratosis congenita spectrum disorders	*TERT*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Thyroid dyshormonogenesis 3	*TG*	Autosomal Recessive	General Population	1 in 217	99%	1 in 21,601				•
TK2-related mitochondrial disorders	*TK2*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Nonsyndromic hearing loss and deafness 7	*TMC1*	Autosomal Recessive	General Population	1 in 96	99%	1 in 9,501				•
TMEM67-related disorders	*TMEM67*	Autosomal Recessive	General Population	1 in 216	99%	1 in 21,501				•

Genetic condition	Gene	Inheritance pattern	Ethnicity*	Carrier frequency*	Detection rate*	Residual risk*	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
Nonsyndromic hearing loss and deafness 8	*TMPRSS3*	Autosomal Recessive	General Population	1 in 96	99%	1 in 9,501				•
Thyroid dyshormonogenesis 2A	*TPO*	Autosomal Recessive	General Population	1 in 227	99%	1 in 22,601				•
TREX1-related disorders	*TREX1*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
TRIM32-related disorders	*TRIM32*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
TRIM32-related disorders	*TRIM32*	Autosomal Recessive	Hutterite	1 in 7	99%	1 in 601				•
Congenital hypothyroidism, *TSHB*-related	*TSHB*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Congenital hypothyroidism, *TSHR*-related	*TSHR*	Autosomal Recessive	General Population	1 in 322	99%	1 in 32,101				•
Congenital hypothyroidism, *TSHR*-related	*TSHR*	Autosomal Recessive	Japanese	1 in 172	99%	1 in 17,101				•
TULP1-related disorders	*TULP1*	Autosomal Recessive	General Population	1 in 186	99%	1 in 18,501				•
Oculocutaneous albinism, type III	*TYRP1*	Autosomal Recessive	General Population	1 in 488	99%	1 in 48,701				•
Familial hemophagocytic lymphohistiocytosis 3	*UNC13D*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Familial hemophagocytic lymphohistiocytosis 3	*UNC13D*	Autosomal Recessive	Swedish	1 in 187	99%	1 in 18,601				•
Vitamin D-resistant rickets, type 2A	*VDR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
VLDLR-associated cerebellar hypoplasia	*VLDLR*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
VLDLR-associated cerebellar hypoplasia	*VLDLR*	Autosomal Recessive	Hutterite	1 in 15	99%	1 in 1,401				•
Hypomyelinating leukodystrophy 12	*VPS11*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Hypomyelinating leukodystrophy 12	*VPS11*	Autosomal Recessive	Ashkenazi Jewish	1 in 160	99%	1 in 15,901				•
Immunodeficiency-centromeric instability-facial anomalies syndrome 2	*ZBTB24*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•
Brittle cornea syndrome 1	*ZNF469*	Autosomal Recessive	General Population	<1 in 500	99%	<1 in 49,901				•

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

Important Information:

QHerit, QHerit 112, QHerit 198, QHerit 445, and QHerit 611 are carrier "screening" tests, and they screen for variations in genes linked to certain health disorders, which can be passed from parents to children.

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

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